MedLadder

HIGH-YIELD REVIEW · 28 CHAPTERS

Pediatrics

Concise, organised notes with tables, mnemonics & illustrations — plus a chapter-wise MCQ bank with answers and explanations.

NEET PG | INI-CET | FMGE | NExT

28
CHAPTERS · ALL TOPICS

4,232
MCQs WITH ANSWERS

Recall
PEARLS · TRAPS · MNEMONICS



MedLadder Pediatrics

High-Yield Review & MCQ Bank · Edition 2026

Growth · Development · Nutrition · Neonatology · Genetics · Metabolism · Immunology · Infections · Immunization · PALS · Systemic Pediatrics

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How to use this book

This book is built for **recall under exam pressure**. Every chapter follows the same three-step ladder:

1. **Chapter opener** – a one-page map of the subtopics so you know the territory before you start.
2. **High-yield notes** – short sentences, comparison tables, flow charts and labelled illustrations. Nothing is padded; if a line is here, it has been asked or is the logic behind something that is asked.
3. **Practice MCQs** – the MedLadder question bank for that chapter, grouped by subtopic, with the correct option in **green** and a short explanation under each question.

The colour-coded boxes

HIGH-YIELD PEARL
Facts that are asked again and again. Read these on every revision.

MNEMONIC
Memory hooks for lists you must reproduce in the exam hall.

EXAM TRAP
The look-alike option examiners use to catch you. Know why the wrong answer is wrong.

MANAGEMENT / DRUG OF CHOICE
First-line treatment, drug of choice and key doses.

PREVIOUS-YEAR PATTERN
How the topic has been framed in NEET PG / INI-CET / FMGE questions.

A 3-pass revision plan

Pass	What to read	Time
1st	Full notes + all MCQs of a chapter; mark every MCQ you got wrong	2–4 h per chapter
2nd	Notes (bold text, tables, boxes) + only marked MCQs	45–60 min per chapter
3rd (last 2 weeks)	Pearls, traps, tables and the Rapid Revision appendices	15 min per chapter

Conventions

- **MC** = most common · **MCC** = most common cause · **DOC** = drug of choice · **IOC** = investigation of choice · **TOC** = treatment of choice · **c/i** = contraindication.
- Doses are for orientation in MCQs; always confirm against current institutional protocols in practice.
- Guideline basis: WHO Child Growth Standards, IAP 2023 growth & immunisation recommendations, India's Universal Immunization Programme, NNF / AHA-2020 neonatal & paediatric resuscitation, IMNCI / WHO SAM protocols, and standard texts (Nelson, Ghai).

Online question practice, timed tests and progress analytics: **medladder.top**

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01

CHAPTER 01

Normal and Abnormal Growth



WHAT THIS CHAPTER COVERS

- Anthropometry & Measurements
- Growth Charts & WHO Standards
- Weight, Height & Growth Velocity
- Head Circumference & Skull
- Dentition & Teeth Eruption
- Puberty & Sexual Maturation
- Short Stature & Growth Disorders
- Bone Age & Skeletal Development
- Dysmorphology & Syndromes

81 MCQs **9** subtopics

1.1 Age Terminology & Anthropometry

Term	Age
Embryo	First 8 weeks of intrauterine life
Fetus	9th week till birth
Neonate	First 28 days of life (early neonatal = first 7 days)
Infant	First year of life
Toddler	1–3 years
Preschool	3–6 years
School age	6–10 years (to puberty)
Adolescent (WHO)	10–19 years · Youth 15–24 y · Young people 10–24 y

Weight

Age	Expected weight	Rule
Birth	3 kg (Indian avg 2.8–3 kg)	Physiological loss up to **10%** in 1st week; regained by **10–14 days**
5 months	**2 × birth weight** (6 kg)	Gain 25–30 g/day in first 3 months
1 year	**3 × birth weight** (9–10 kg)	
2 years	**4 × birth weight** (12 kg)	
3 years	5 × (14–15 kg)	
5 years	6 × (18 kg)	
7 years	7 × (21 kg)	
10 years	10 × (30 kg)	

WEIGHT FORMULAE (IAP / WEECH)

- 3–12 months: **(age in months + 9) / 2**
- 1–6 years: **(age in years × 2) + 8**
- 7–12 years: **(age in years × 7 − 5) / 2**

Length / Height

Age	Length	Comment
Birth	**50 cm**	
3 months	60 cm	+10 cm
9 months	70 cm	
1 year	**75 cm** (1.5 × birth)	+25 cm in 1st year (maximum postnatal velocity)
2 years	87–90 cm	+12 cm in 2nd year
4 years	**100 cm** (2 × birth)	
12 years	150 cm (3 × birth)	

- Height 2–12 years ≈ **(age in years × 6) + 77 cm**.
- Velocity after 2 years until puberty: **5–6 cm/year**; < 4 cm/year = pathological (think hormone deficiency).

- < 2 years → **recumbent length** (infantometer); ≥ 2 years → **standing height** (stadiometer). Standing height is ~**0.7 cm** less than recumbent length.
- Height at 2 years ≈ **half of adult height**.
- Upper segment : lower segment (US:LS) – **1.7 at birth**, 1.3 at 3 yrs, **1.0 at 7–10 years**, < 1 in adults.

US:LS RATIO

Increased US:LS (short limbs) → achondroplasia, rickets, hypothyroidism (infantile proportions). **Decreased** US:LS (long limbs) → Marfan, Klinefelter, homocystinuria, eunuchoidism, spondyloepiphyseal dysplasia (short trunk).

Head circumference (HC)

Age	HC	Monthly gain
Birth	**33–35 cm**	2 cm/month × first 3 m
3 months	40 cm	1 cm/month 3–6 m
1 year	**45–46 cm**	0.5 cm/month 6–12 m
2 years	48 cm	
12 years	52 cm (adult ≈ 54–56)	

- HC measured across the **occipital protuberance and supraorbital ridges** (maximum occipito-frontal circumference) with non-stretchable tape.
- **Microcephaly**: HC < −3 SD (some use < −2 SD). **Macrocephaly**: HC > +2 SD.
- Brain attains ~**25%** of adult size at birth, 75% at 2 years, 90% by 5 years.

Chest circumference & other parameters

Age	Relation
Birth	HC > chest circumference by ~3 cm
9 months–1 year	**HC = chest circumference**
> 1 year	Chest > HC

- Chest circumference is measured at the level of the **nipples** in mid-inspiration.
- **MUAC** (mid-upper arm circumference): **age-independent between 6 months and 5 years**; increases only ~1 cm (from 15 to 16 cm) in 1–5 yrs.
- MUAC cut-offs (WHO): **< 11.5 cm = severe acute malnutrition**, 11.5–12.5 = moderate, > 12.5 normal. Measured on **left arm**, midpoint between acromion and olecranon, with **Shakir's tape** (red < 11.5, yellow 11.5–12.5, green > 12.5).
- Skinfold thickness – **triceps** (most commonly used) and subscapular; measured with **Harpenden calipers**.
- **BMI** = weight (kg)/height (m)^2; used from **2 years** onwards (WHO charts give BMI-for-age from birth). Obesity in children is defined by **BMI-for-age percentiles** (IAP: adult equivalent 23 = overweight, 27 = obese).

1.2 Growth Charts

0–5 years	5–18 years	Interpretation
WHO Child Growth Standards 2006 (MGRS: Brazil, Ghana, India, Norway, Oman, USA) – prescriptive	IAP 2015 growth charts (height, weight, BMI) – Indian reference	Normal: 3rd–97th percentile or −2 to +2 Z-score. < −3 SD = severe

Choosing the right growth chart (IAP 2015 recommendations)

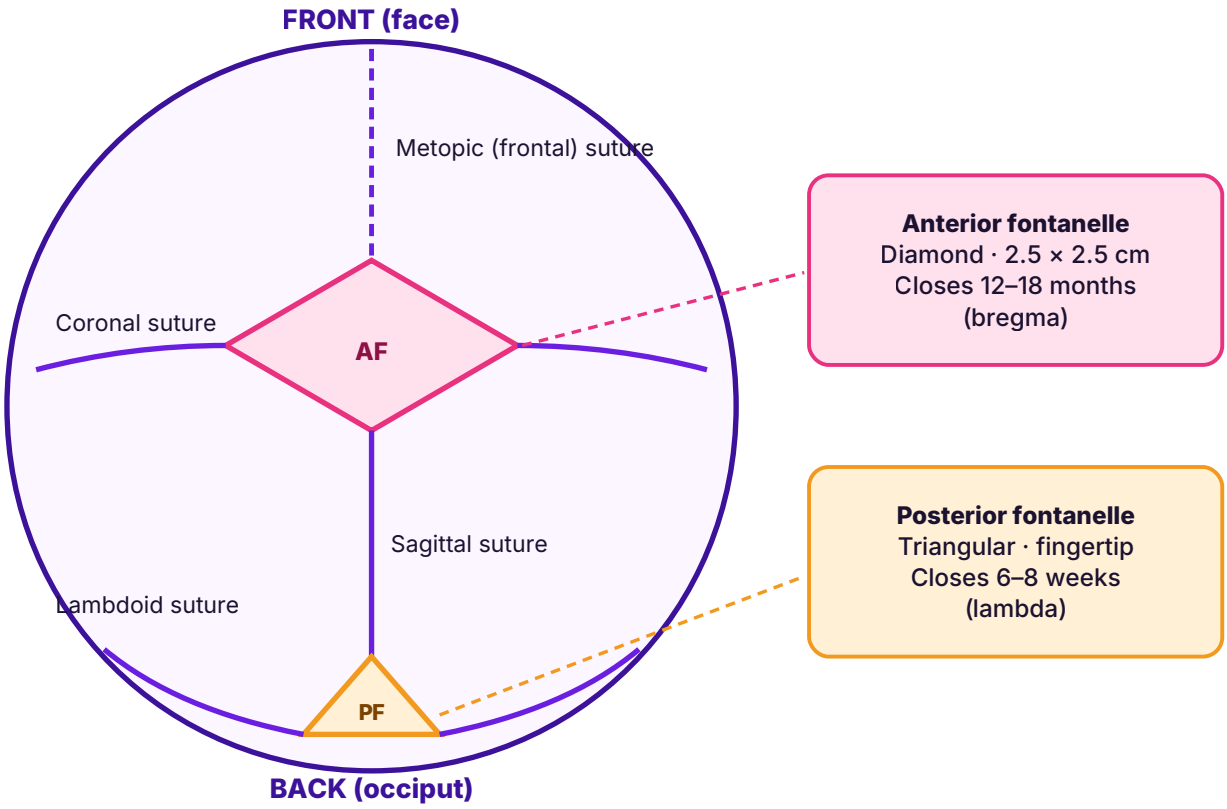
Chart	Population / basis	Key points
WHO Child Growth Standards (2006)	MGRS – 6 countries: **Brazil, Ghana, India, Norway, Oman, USA**	**Prescriptive standard** (how children **should** grow); exclusively/ predominantly breastfed, non-smoking mothers; used for **0–5 years**
WHO Reference 2007	Reanalysis of NCHS 1977 data	5–19 years
CDC 2000	US children (reference)	Descriptive
IAP 2015 charts	Indian children (affluent)	Recommended for **5–18 years** (height, weight, BMI)
Agarwal charts (1992)	Indian children	Older Indian reference

- WHO standards available for: weight-for-age, length/height-for-age, weight-for-length/height, BMI-for-age, HC-for-age, MUAC-for-age, triceps & subscapular skinfold-for-age, motor milestone windows.
- **Z-score** (SD score) = (observed – median)/SD. Normal **−2 to +2**.
- Percentile charts: normal between **3rd and 97th** percentile. 50th = median.
- **Growth monitoring chart in India** (MCP card / ICDS): weight-for-age with colour bands.

WHO DEFINITIONS OF UNDERNUTRITION (Z-SCORES)

- **Underweight** = weight-for-age < −2 SD (composite indicator; acute + chronic)
- **Stunting** = height-for-age < −2 SD → **chronic** malnutrition
- **Wasting** = weight-for-height < −2 SD → **acute** malnutrition
- Severe = < −3 SD for each.

1.3 Fontanelles & Skull



Fontanelles of the neonatal skull

Fontanelle	Shape	Size at birth	Closure
Anterior (bregma)	Diamond / rhomboid	~2.5 × 2.5 cm (up to 4 cm)	**12–18 months** (range 9–24 m)
Posterior (lambda)	Triangular	Fingertip	**6–8 weeks** (by 2–3 months)

Fontanelle	Shape	Size at birth	Closure
Sphenoidal (anterolateral)			6 months
Mastoid (posterolateral)			6–18 months

- **Third fontanelle** (between anterior and posterior, on sagittal suture) – seen in **Down syndrome**, congenital hypothyroidism, congenital rubella.

Large / delayed closure of AF	Early closure of AF
Hypothyroidism, **rickets**, Down syndrome, achondroplasia, hydrocephalus, osteogenesis imperfecta, **cleidocranial dysostosis**, IUGR/prematurity, malnutrition, trisomy 13/18, Apert, hypophosphatasia	**Microcephaly**, craniosynostosis, hyperthyroidism, hypervitaminosis D, hyperparathyroidism

- **Bulging AF**: raised ICT – meningitis, hydrocephalus, hypervitaminosis A, **tetracycline**, lead poisoning, pseudotumor cerebri. Normal crying can make it bulge transiently.
- **Depressed AF**: dehydration.

Craniosynostosis

Suture fused	Skull shape
Sagittal (most common)	**Scaphocephaly / dolichocephaly** (long boat-shaped)
Coronal (bilateral)	**Brachycephaly** (short, broad) — Apert, Crouzon
Coronal (unilateral)	Anterior plagiocephaly
Metopic	**Trigonocephaly**
Lambdoid (unilateral)	Posterior plagiocephaly (rare; positional plagiocephaly far commoner)
Multiple (coronal + lambdoid + sagittal)	**Kleeblattschädel / clover-leaf skull** (Pfeiffer, thanatophoric dysplasia)
All sutures	Oxycephaly / turricephaly (tower skull)

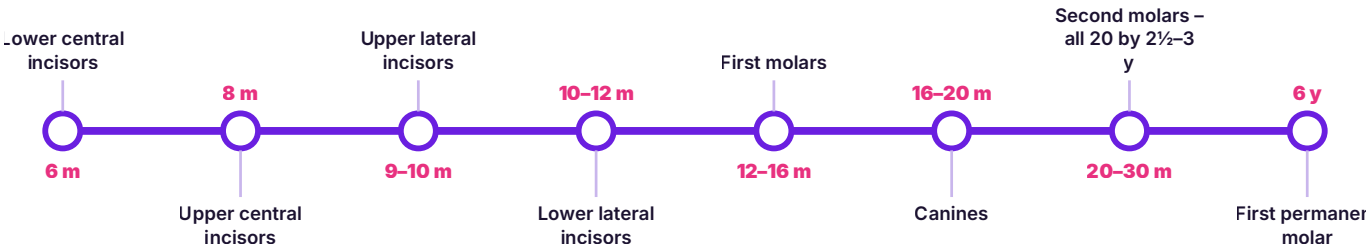
FGFR MUTATIONS

FGFR1 – Pfeiffer · **FGFR2** – Crouzon, Apert (also Pfeiffer) · **FGFR3** – achondroplasia, hypochondroplasia, thanatophoric dysplasia, Muenke craniosynostosis.

Microcephaly & Macrocephaly

Microcephaly – causes	Macrocephaly – causes
Primary (genetic): trisomies 13, 18, 21, Cri-du-chat, Cornelia de Lange, Rubinstein-Taybi, Seckel, Smith-Lemli-Opitz	**Benign familial megalencephaly** (MCC megalencephaly – check parents' HC)
Secondary: TORCH (CMV – periventricular calcification; **Zika**), fetal alcohol, maternal PKU, HIE, meningitis	Hydrocephalus, subdural collection
Acquired after birth: **Rett syndrome** (deceleration of head growth at 5–48 months), HIE	Storage disorders: Canavan, Alexander, **Tay–Sachs**, MPS, glutaric aciduria type 1; Sotos; achondroplasia; NF-1

1.4 Dentition



Eruption sequence of primary (deciduous) teeth

- **Primary teeth: 20** (no premolars) – eruption begins at **6 months** (lower central incisor first), all erupt by **2.5–3 years**. Formula for number of milk teeth ≈ age (months) − 6 (till 26 months).
- **Delayed dentition**: no tooth by **13 months**. MCC = **idiopathic/familial**; others – hypothyroidism, hypopituitarism, rickets, Down syndrome, cleidocranial dysostosis, malnutrition.
- **Permanent teeth: 32** – first is **first molar at 6 years** ("6-year molar"), followed by central incisors (6–7 y), lateral incisors (7–8 y), first premolars (10–11 y), canines (11–12 y), second premolars, **second molar (12 y)**, third molar 17–25 y.
- **Natal teeth** (present at birth) / **neonatal teeth** (erupt in first 30 days) – usually lower central incisors; associated with **Ellis–van Creveld**, Hallermann–Streiff, Pierre Robin, Soto, pachyonychia congenita; **Riga–Fede disease** = ulceration of tongue from natal teeth.
- Discoloured teeth: **tetracycline** (yellow-brown), **fluorosis** (mottled chalky white/brown pits), kernicterus/ hyperbilirubinemia (green), erythropoietic porphyria (red-brown; fluoresces), dentinogenesis imperfecta (opalescent – with OI).
- **Hutchinson's teeth** (notched, peg-shaped upper central incisors) & **mulberry molars** – congenital syphilis.
- **Early childhood caries** (bottle caries): upper incisors first, **lower incisors spared** (protected by tongue).

1.5 Puberty & Sexual Maturity Rating (Tanner)

	Girls	Boys
First sign	**Thelarche (breast bud, SMR 2)** at 8–13 y	**Testicular enlargement ≥ 4 mL / 2.5 cm** at 9–14 y
Sequence	Thelarche → pubarche → **peak height velocity (SMR 2–3)** → **menarche (SMR 4)**	Testis → pubic hair → penis length → **PHV (SMR 4)** → voice change, facial hair
Menarche / spermarche	Menarche ~**2–2.5 yrs after thelarche**, mean 12.5 y	Spermarche SMR 3
Peak height velocity	~8–9 cm/yr, **2 years earlier than boys**	~9.5–10 cm/yr
Gynecomastia	—	Physiological pubertal gynecomastia in up to 65% (SMR 2–4), resolves in 1–2 y

- Orchidometer = **Prader**; ≥ 4 mL = puberty has started; adult volume 15–25 mL.
- **Precocious puberty**: girls < **8 years**, boys < **9 years**. **Delayed puberty**: no breast development by **13 y** in girls; no testicular enlargement by **14 y** in boys.

1.6 Bone Age

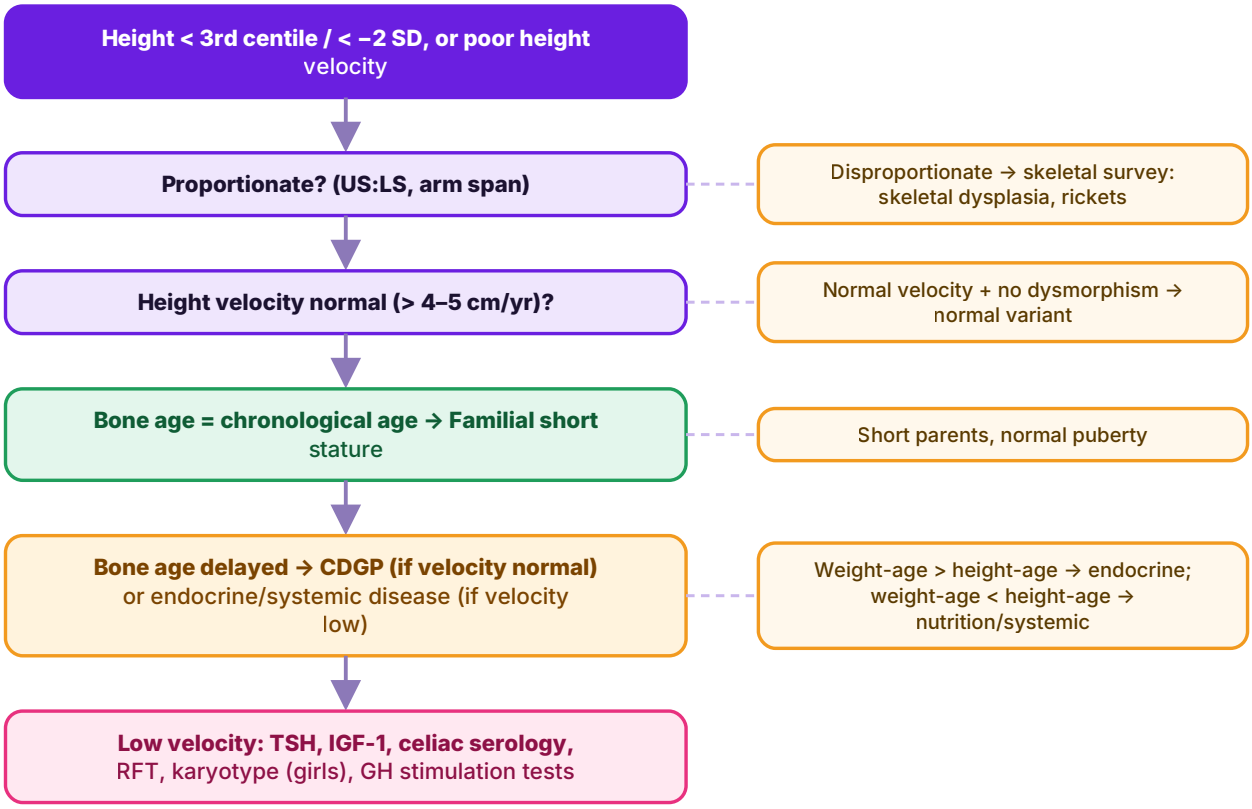
Age	X-ray used
Newborn / infants (< 1 y)	**Knee & foot** (distal femur epiphysis appears at 36 wk gestation – marker of term; proximal tibia at 38–40 wk)
1–13 years	**Left hand & wrist** – Greulich–Pyle atlas or Tanner–Whitehouse (TW3)
Adolescents	Elbow (Sauvegrain), iliac crest (Risser sign)

- Carpal bones: **capitate (first, 2–3 months)** and hamate (3 months) → triquetral (3 y) → lunate (4 y) → trapezium (5 y) → trapezoid (6 y) → scaphoid (7 y) → **pisiform last (10–12 y)**. Rule of thumb: number of carpal ossification centres ≈ age in years (1–7 y).
- **Bone age < chronological age**: constitutional delay, **hypothyroidism (most delayed)**, GH deficiency, malnutrition, chronic systemic disease, Cushing syndrome.
- **Bone age > chronological age**: precocious puberty, CAH, hyperthyroidism, obesity, McCune–Albright, androgen excess.
- Bone age = chronological age: **familial short stature**, primordial dwarfism (e.g. Russell–Silver), skeletal dysplasia (variable).
- **Epiphyseal dysgenesis** (stippled, fragmented epiphyses) – **congenital hypothyroidism**; stippled epiphyses also in Zellweger, warfarin embryopathy, chondrodysplasia punctata.

1.7 Short Stature

Definition: height < **3rd percentile** or < **−2 SD** for age and sex, or height velocity < 25th percentile, or height well below mid-parental target.

- **Mid-parental height (target height)** = (father's height + mother's height) / 2 **+ 6.5 cm for boys**, **− 6.5 cm for girls** (range ± 8.5 cm).



Diagnostic approach to short stature

Constitutional delay vs familial short stature

Feature	Constitutional delay of growth & puberty (CDGP)	Familial (genetic) short stature
Family history	Delayed puberty in a parent ("late bloomer")	**Short parents**
Birth length	Normal	Normal
Growth pattern	Crosses down percentiles in 1st 2–3 years, then grows parallel below 3rd centile	Parallel to and below 3rd centile
Growth velocity	Normal	Normal
Bone age	**Delayed** (BA < CA, = height age)	**= Chronological age**
Puberty	Delayed	Normal timing
Final height	**Normal** (reaches target)	**Short but within target range**

PATHOLOGICAL SHORT STATURE CLUES

Height velocity < 4 cm/year, height < −3 SD, disproportion, dysmorphism, height far below mid-parental target, weight age < height age (systemic disease / malnutrition) vs **weight age > height age (endocrine: hypothyroid, GH deficiency, Cushing – "short & chubby")**.

Causes of short stature

Proportionate	Disproportionate (skeletal dysplasias & rickets)
Prenatal: IUGR (~10% don't catch up), Russell–Silver, Turner, Down, Noonan, Prader–Willi, Seckel, fetal alcohol	**Short limbs** (↑ US:LS): achondroplasia, hypochondroplasia, rickets, thanatophoric dysplasia, Ellis–van Creveld
Postnatal: malnutrition (MCC in India), chronic diseases (celiac, CKD, IBD, cyanotic CHD, thalassemia, asthma), psychosocial deprivation	**Short trunk** (↓ US:LS): spondyloepiphyseal dysplasia, mucopolysaccharidoses, Morquio, spinal TB/scoliosis
Endocrine: hypothyroidism, GH deficiency, Cushing, pseudohypoparathyroidism, poorly controlled diabetes (Mauriac)	

- **Most common cause of short stature overall**: familial short stature and CDGP (normal variants). **Most common pathological cause in India**: undernutrition.
- **Psychosocial dwarfism**: functional GH deficiency; reverses on removal from adverse environment.
- Investigations: CBC, ESR, RFT, LFT, electrolytes, urinalysis, **TSH/T4**, **tTG-IgA (celiac)**, karyotype in all short girls (**Turner**), bone age, **IGF-1/IGFBP-3**, GH stimulation tests (clonidine, glucagon, insulin, arginine) – peak GH < 10 ng/mL suggests deficiency.

Growth hormone therapy – approved indications

GH deficiency, **Turner**, **Prader–Willi**, **CKD**, SGA without catch-up by 2–4 y, **Noonan**, SHOX deficiency, idiopathic short stature (US). Side effects: **SCFE**, pseudotumor cerebri, scoliosis progression, gynecomastia, insulin resistance.

Tall stature

Familial (MCC), obesity (exogenous – tall in childhood, normal adult height), **Marfan**, **homocystinuria**, **Klinefelter**, **Sotos** (cerebral gigantism – macrocephaly, advanced bone age), **Weaver**, **Beckwith–Wiedemann**, pituitary gigantism, precocious puberty (tall child → short adult), hyperthyroidism, CAH.

1.8 Dysmorphology – High-Yield Syndromes

Syndrome	Key features
Russell–Silver	IUGR, triangular face, **body hemihypertrophy/asymmetry**, normal HC (relative macrocephaly), clinodactyly 5th finger; hypomethylation 11p15 / maternal UPD 7
Seckel ("bird-headed dwarf")	Severe IUGR, microcephaly, beak-like nose, ID; AR
Cornelia de Lange (Amsterdam dwarfism)	**Synophrys** (eyebrows meet), long philtrum, thin lips, hirsutism, micromelia/limb defects, ID; NIPBL
Rubinstein–Taybi	**Broad thumbs & great toes**, beaked nose, ID; CREBBP
Cri-du-chat (5p–)	Cat-like cry, microcephaly, hypertelorism, ID
Williams (7q11.23 del)	**Elfin facies**, supravalvular aortic stenosis, **hypercalcemia**, "cocktail party" personality, stellate iris
Noonan (PTPN11, AD)	Male Turner phenotype, normal karyotype, **pulmonary stenosis** (dysplastic valve), HCM, webbed neck, bleeding diathesis, cryptorchidism
Prader–Willi (paternal 15q11–13 del / maternal UPD)	Neonatal hypotonia & feeding difficulty → hyperphagia, obesity, hypogonadism, small hands/feet, almond eyes
Angelman (maternal 15q11–13 del / paternal UPD; UBE3A)	"Happy puppet": paroxysmal laughter, ataxic gait, seizures, severe ID, absent speech
Beckwith–Wiedemann (11p15)	Macrosomia, **macroglossia**, omphalocele, hemihypertrophy, ear lobe creases/pits, neonatal **hypoglycemia**; risk of **Wilms**, hepatoblastoma
Sotos	Cerebral gigantism, macrocephaly, frontal bossing, pointed chin, advanced bone age; NSD1
Treacher Collins (TCOF1, AD)	Mandibulofacial dysostosis: malar & mandibular hypoplasia, **lower lid coloboma**, down-slanting palpebral fissures, conductive deafness, normal intelligence
Pierre Robin sequence	Micrognathia → glossoptosis → **U-shaped cleft palate**; airway obstruction (prone position)
Goldenhar (oculo-auriculo-vertebral)	Hemifacial microsomia, **epibulbar dermoid**, preauricular tags, vertebral anomalies
Waardenburg (PAX3)	**White forelock**, heterochromia iridis, dystopia canthorum, congenital **SNHL**
Cleidocranial dysostosis (RUNX2)	Absent/hypoplastic **clavicles** (shoulders opposable), large late-closing fontanelles, supernumerary teeth, delayed dentition
Kabuki	Long palpebral fissures with eversion of lower lid, arched eyebrows, persistent fetal finger pads
Smith–Lemli–Opitz	Cholesterol synthesis defect (7-DHC reductase); **2–3 toe syndactyly**, microcephaly, ptosis, genital anomalies
Potter sequence	Renal agenesis → **oligohydramnios** → pulmonary hypoplasia, flat (Potter) facies, limb deformities
VACTERL	Vertebral, Anal atresia, Cardiac, TE fistula, Renal, Limb (radial) defects
CHARGE (CHD7)	Coloboma, Heart defects, Atresia choanae, Retarded growth, Genital hypoplasia, Ear anomalies/deafness

TERMINOLOGY

Malformation – intrinsic abnormal development (cleft lip, CHD). **Deformation** – mechanical force on normal tissue (clubfoot from oligohydramnios). **Disruption** – breakdown of normal tissue (amniotic bands). **Sequence** – cascade from a single primary defect (Pierre Robin, Potter). **Syndrome** – multiple anomalies from a single cause (Down). **Association** – non-random co-occurrence without known single cause (VACTERL, CHARGE historically).

Digits & hands

- **Polydactyly**: post-axial (ulnar side) – most common, familial, AD in Africans; syndromic – **trisomy 13**, Ellis–van Creveld, **Laurence–Moon–Bardet–Biedl**, Meckel–Gruber, Carpenter. Pre-axial (thumb side) – Carpenter, Greig cephalopolysyndactyly, Holt–Oram (triphalangeal thumb).
- **Syndactyly**: **Apert** (mitten hands/"sock feet"), Carpenter, Poland, Smith–Lemli–Opitz (2–3 toes).
- **Arachnodactyly**: Marfan, homocystinuria, congenital contractural arachnodactyly (Beals).
- **Clinodactyly** (5th finger incurved): Down, Russell–Silver. **Camptodactyly**: flexion contracture.
- **Absent radius/thumb**: **TAR** (thrombocytopenia-absent radius – **thumbs present**), **Fanconi anemia** (thumbs absent/hypoplastic), Holt–Oram (ASD + upper limb defects), VACTERL.
- **Knuckle–dimple signs**: **Down** – short 5th middle phalanx; **Turner** – short **4th metacarpal** (knuckle-knuckle-dimple-knuckle); **pseudohypoparathyroidism (Albright)** – short **4th & 5th** metacarpals.
- **Simian crease**: Down (also 5% normal).

Practice MCQs · Normal and Abnormal Growth

81 questions · correct option highlighted in green · cover the answer strip to self-test

Anthropometry & Measurements

1. All are true about anthropometric measurements except:

A. Birth weight triples by 1 year
B. Skinfold thickness is measured in sub scapular region
C. Weight is measured to the nearest 100 gram
D. Standing height is about 1.7 cm less than recumbent length

Ans D Growth Standards, 2008
- If a child < 2 years old, measure recumbent length.
- If ≥ 2 years age & able to stand, measure standing height.
- Standing height is about 0.7 cm less than recumbent length.

2. In a healthy child, the head and chest circumference equal each other around the age of:

A. 2nd year
B. 3rd year
C. 5th year
D. 15th year

Ans C 9-12 months

3. True about head circumference measurement:

A. Measured at supraorbital ridge
B. Measures hydrocephalus/microcephaly
C. Serial measurement is useful
D. Helps in measurement of neurological development
E. Is an indicator of brain growth DENTITION

Ans A b. Measures hydrocephalus/microcephaly; c. Serial measurement is useful; d. Helps in measurement of neurological development; e. Is an indicator of brain growth DENTITION

Growth Charts & WHO Standards

4. Which of the following is not true in assessing skin fold thickness?

A. Measured at the triceps level
B. Measured to the nearest of 1 cm
C. Tanner's chart contains the normal values
D. Indirectly indicates the caloric reserve of the body

Ans B Tanner skinfold standards (1975), were considerably higher than subsequent reported measurements.

5. All are unique features of WHO growth charts 2006 except:

A. Breast-fed infant as normative growth model
B. Sample collected from six States in the US
C. Includes new indicators such as skinfold thickness
D. Reiterates children grow similarly across the World

Ans B WHO growth charts are based on multicentre growth reference study (MGRS)Q conducted in 6 countries all over the world including: Brazil, Oman, Norway, Ghana, US and India (New Delhi).

6. Mid adolescence is the age between:

A. 10–13 years
B. 14–16 years
C. 17–19 years
D. 14–18 years

Ans B

7. A child is below the third percentile for height. His growth velocity is normal, but chronologic age is more than skeletal age. The most likely diagnosis is:

A. Constitutional delay in growth
B. Genetic short stature
C. Primordial dwarfism
D. Hypopituitarism

Ans A The child in this Question has: Height below 3rd percentile, i.e., short stature. Normal growth velocity, i.e., normal variant of short stature and Skeletal age is less than chronological age. So, this child has Constitutional delay in growth.

Weight, Height & Growth Velocity

8. A newborn is double his height at birth by what age?

A. 2½ years
B. 3½ years
C. 4½ years
D. 5½ years

Ans C Weight doubles at 5 months while height at 4–4½ years.

9. Postnatally when is the growth velocity maximum?

A. In the first year of life
B. In the second year of life
C. In the seventh year of life
D. In adolescence

Ans A Postnatally there are 2 periods of accelerated growth: first year of life and growth spurt during puberty.

10. Birth weight doubles by?

A. 3 months
B. 5 months
C. 9 months
D. 12 months

Ans B

11. Weight of newborn quadruples by:

A. 9 months
B. 12 months
C. 2 years
D. 3 years

Ans C

12. The weight of the child at 3 years is usually—times the birth weight:

A. 3
B. 4
C. **5**
D. 7

Ans C

13. Birth length doubles by what age?

A. 1 year
B. 2 years
C. 3 years
D. **4 years**

Ans D

14. A normal healthy child has a height of 100 cm and weighs 16 kg. What is his most likely age?

A. 3 years
B. **4 years**
C. 5 years
D. 6 years

Ans B At 4 years, height becomes 100 cm and weight becomes 2 × 4 + 8 = 16 kg.

15. The normal gain in length in a full term baby, at first 6 months of life is:

A. 6 cm
B. 9 cm
C. **15 cm**
D. 24 cm

Ans C • Length at birth = 50 cm
• Length at 3 months = 60 cm; So, gain in length in 3 months = 60–50 = 10 cm
• Length at 9 months = 70 cm; So, gain in length in 9 months = 70–50 = 20 cm
• So, gain in length in 6 months is between 10 and 20 cm, so 15 cm is the best answer.

16. Average gain of length in first year is:

A. **1 year**
B. 2 years
C. 4 years
D. 6 years

Ans A 25 cm Gain of Length in first year is 75–50 cm = 25 cm.

17. Increase in height in first year is by:

A. 40%
B. **50%**
C. 60%
D. 75%

Ans B In 1st year, From 50 cm, the height increases by 25 cm (i.e. 50%) to 75 cm.

18. Increase in height of the child from 1–3 years:

A. 10 cm
B. 15 cm
C. **20 cm**
D. 25 cm

Ans C Height at 1 year is 75 cm and at 4 years is 100 cm; So increase in height from 1–4 years is 100 – 75 = 25 cm. Height at 1 year is 75 cm and at 2 years is 90 cm; So increase in height from 1–2 years is 90 – 75 = 15 cm. So increase in height from 1–3 years lies between 15 and 25 cm, so 20 cm is the best answer.

19. Maximum growth spurt in boys occurs at:

A. 8–10 years
B. 10–11 years
C. 12–13 years
D. **14–15 years HEAD CIRCUMFERENCE**

Ans D 14–15 years Height velocity curves for males (solid line) and females (dashed line) showing maximum growth velocity in:
• Boys at 14–15 yrs
• Girls at 11–13 yrs

20. Growth spurt occurs:

A. Just before appearance of axillary hair
B. **Just before menarche**
C. After 16 years
D. Before thelarche

Ans B

21. A 13 years old boy has bilateral gynecomastia. His height is 148 cm, weight 58 kg, the sexual maturity rating is stage 2. The gynecomastia is most likely due to:

A. Prolactinoma
B. Testicular tumor
C. **Pubertal gynecomastia**
D. Chronic liver disease SHORT STATURE

Ans C Pubertal gynecomastia:
• It is a benign subareolar glandular breast enlargement seen in up to 65% adolescent males.
• Typically appears at least 6 months after onset of secondary sex characteristics.

22. A child's growth velocity is normal but bone development is not according to the chronological age. What is diagnosis?

A. Genetic
B. Dwarfism
C. **Constitutional delay**
D. Familial short stature

Ans C

23. A newborn is noted to have a large head and short limbs. On further examination, short broad fingers, a small face, and low-normal length are noted. The trunk appears long and narrow, to confirm the diagnosis you should:

A. Order an ophthalmologic examination
B. **Obtain skeletal radiographs**
C. Order chromosome analysis
D. Examine the parents16 Section 1: General Pediatrics

Ans B The given description suggests a diagnosis of Achondroplasia; Skeletal X-rays will help in diagnosing it. What is the proubeasbtleio ange of this baby? Q 1 a. 2 weeks b. 6 weeks c. 3 months d. 4 months
• Development quotient (DQ) = Developmental age/actual age * 100 M neMonic M
• A child learns to walk in 1 year & sits in half of that time, i.e. 6 months
• 'a' comes before 'e', so crawling comes before creeping When does the muielessttioonne shown in this picture apQpear? 2 a. 6-7 months b. 10-11 months c.

Head Circumference & Skull

24. True for a 2 years old boy is:

A. **Weight four times that of birth weight**
B. Rides tricycle
C. Climbs up and down the stairs with alternating steps
D. Both upper and lower canines erupted
E. Ossification centre for radius head appears

Ans A d. Both upper and lower canines erupted a. True At 1 year weight becomes 3 times & at 2 years, 4 times of birth weight b. False A child is able to ride a tricycle usually at 3 years age c. False A child is able to climb up & down stairs with alternating steps at 4 years d. True Upper & lower canines usually erupt at around 16–20 weeks age e. False Ossification centre for radius head appears between 3 and 4 years

25. Which of the fontanelle is the last to close?

A. Posterior fontanelle
B. **Anterior fontanelle**
C. Mastoid fontanelle
D. Sphenoidal fontanelle

Ans B Time of closure of different fontanelle:
• Posterior fontanelle—2 to 3 months after birth
• Sphenoidal fontanelle—6 months after birth
• Mastoid fontanelle—6 to 18 months after birth
• Anterior fontanelle—10–24 months after birth. a. Maxillary hypoplasia; c. Hydrocephalus; e. Mandibular prognathism Crouzon syndrome:
• Autosomal dominant (FGFR2 gene)
• Craniosynostosis involving coronal suture
• Wide-set, bulging eyes & beaked nose
• Maxillary hypoplasia weiveR • Underdeveloped upper jaw & protruding lower jaw

26. Crouzon syndrome consists of:

A. **Maxillary hypoplasia**
B. Syndactyly
C. Hydrocephalus
D. Macrocephaly
E. Mandibular prognathism

Ans A Crouzon syndrome (craniosynostosis) is characterized by premature fusion of cranial sutures leading to maxillary hypoplasia with relative mandibular prognathism and shallow orbits causing proptosis. Unlike Apert syndrome, it does NOT feature syndactyly.

27. Turricephaly is:

A. **Tall head**
B. Narrow head
C. Wide head
D. Short head

Ans A Turricephaly is cone-shaped head from premature fusion of the coronal, sphenofrontal & frontoethmoidal sutures.

28. Causes of macrocephaly includes:

A. **Galactosemia**
B. Weaver syndrome
C. Rett syndrome
D. Soto's syndrome
E. Morquio syndrome

Ans A b. Weaver syndrome, d. Soto's syndrome, e. Morquio syndrome Causes of Megalencephaly:
• Lysosomal storage disorders: MPS, Gangliosidosis
• Carbohydrate disorder: Galactosemia
• Aminoacidurias: MSUD, Glutaric aciduria type I
• Neurodegenerative ds: Alexander ds, Canavan ds, Metachromatic leukodystrophy
• Neurocutaneous ds: Neurofibromatosis, Tuberous sclerosis Soto's syndrome, Achondroplasia
• Benign familial megalencephaly

29. Following feature differentiate hydranencephaly from hydrocephalus:

A. Hydranencephaly is static while hydrocephalus continue to increase in size
B. Hydranencephaly does not cause increase in head size
C. **Cerebral cortex is deficient/ hypoplastic in hydranencephaly**
D. All of the above

Ans C Cerebral cortex is deficient/hypoplastic in hydranencephaly In Hydranencephaly, cerebral hemispheres are absent or represented by membranous sacs with remnants of frontal, temporal, or occipital cortex dispersed over the membrane, while the midbrain and brainstem are relatively intact

30. Anterior fontanelle closes at:

A. 2–3 months
B. 4–7 months
C. **12–20 months**
D. 24–30 months

Ans C Anterior fontanelle is a diamond-shaped structure at the junction of the frontal and parietal bones, that is open at birth and usually measures 2 × 2 cm. The average time of closure is 18 mo, but the fontanelle can close normally as early as 9 months.

31. Anterior fontanelle is located between which two bones?

A. Frontal & occipital
B. **Frontal & parietal**
C. Parietal & occipital
D. Frontal & zygomatic

Ans B Frontal and parietal

32. Premature fusion of coronal, sphenofrontal and fronto-ethmoidal sutures is seen in:

A. Turricephaly
B. Dolichocephaly
C. Plagiocephaly
D. Trigoncephaly

Ans A Discussing about the options one by one, a. Turricephaly Cone-shaped head caused by premature fusion of coronal, sphenofrontal and frontoethmoidal sutures b. Dolichocephaly Due to premature closure of sagittal suture produces a long and narrow skull Also called scaphocephaly; It is the most common form of craniosynostosis with c. Plagiocephaly Asymmetrical distortion or flattening of one side of the skull d.

33. Microcephaly is defined as HC:

A. 2SD less than the mean
B. 3SD less than the mean
C. <33 cm at birth
D. <40 cm at 6 months

Ans B

34. The rate of increase in HC in the first 3 months:

A. 2 cm/month
B. 1 cm/month
C. 0.5 cm/month
D. 0.25 cm/month

Ans A Rate of increase in head circumference in 1st 3 months of life is 2 cm/month.

35. Sagittal sutures overlapping causes:

A. Oxycephaly
B. Dolichocephaly
C. Plagiocephaly
D. Cephalocephaly

Ans B

Dentition & Teeth Eruption

36. True about constitutional growth delay is/are:

A. Normal length and weight at birth
B. Catch up growth at 6–12 months
C. Height velocity assume normal rate at 2–3 years
D. Bone age > chronological age
E. Puberty occurs at normal age

Ans A In constitutional growth delay, weight and length (height) are normal at birth, but decrease near the end of infancy, parallel the norm through middle childhood (6–11 years age), and accelerate toward the end of adolescence. The bone age is less than chronological age, but comparable to the height age. Adult size is normal.

37. Delayed eruption is usually considered when there are no teeth by approximately by the age of:

A. 18 months of age
B. 9 months of age (mean +3 SD)
C. 16 months of age (mean +3 SD)
D. 13 months of age (mean +3 SD)

Ans D

38. Eruption of 1st molar permanent tooth occurs at the age of:

A. 4–5 years
B. 6–7 years
C. 7–8 years
D. 8–10 years

Ans B Permanent dentition begins at 6 years and is completed by 12 years (except for 3rd molar).

39. Most common location of supernumerary teeth is:

A. Between incisors
B. Between canines
C. Between molars
D. Between premolars

Ans A Supernumerary teeth most commonly involves maxillary incisor region.

40. True about dentition are all except:

A. Hypothyroidism causes delayed dentition
B. Premolar is not seen in primary dentition
C. Third molar is the last to appear in secondary dentition
D. Canine is the first in primary dentition

Ans D The first tooth to erupt in primary dentition is lower central incisor, while in secondary dentition, it is the first molar.

41. Which of the following are the first incisors to erupt in an infant?

A. Lower central
B. Lower lateral
C. Upper central
D. Upper lateral

Ans A

42. Primary dentition begins to show teeth eruption by:

A. 6 weeks
B. 12 weeks
C. 6 months
D. 12 months14 Section 1: General Pediatrics

Ans C

43. Ist permanent teeth to appear:

A. Molar
B. Premolar
C. Incisor
D. Canine

Ans A

44. Milk teeth—Total no. in human being:

A. 20
B. 28
C. 32
D. 24

Ans A 20 Primary dentition comprises of 20 teeth, which begins at 6 months of age and completes by 3 years.

45. **Delayed dentition is seen in:**
A. Down syndrome
B. Congenital hypothyroidism
C. Rickets
D. **All of the above**

Ans D

46. **By ___ years all milk teeth are erupted:**
A. 1.5
B. 2
C. 2.5
D. **3**

Ans D

47. **First teeth to erupt in primary dentition is:**
A. First molar
B. Premolar
C. **Incisor**
D. Canine

Ans C

48. **Which of the following is true about eruption of teeth?**
A. Premolar appears first in primary dentition
B. Incisors appear first in secondary dentition
C. **Third molar is last to develop**
D. Hypothyroidism delays dentition
E. Canines are last to appear in primary dentition NORMAL PUBERTY

Ans C d. Hypothyroidism delays dentition

Puberty & Sexual Maturation

49. **Early adolescent age is:**
A. 8–11 years
B. **10–13 years**
C. 14–15 years
D. 16–19 years

Ans B

50. **Earliest indication of sexual maturation in females:**
A. Menarche
B. **Thelarche**
C. Pubarche
D. Genital maturation

Ans B Earliest indication of sexual maturation in females is thelarche, while in males, it is testicular enlargement.

51. **What is the first visible sign of puberty and the hallmark of SMR2 in boys and at what age does it occur?**
A. **Testicular enlargement and 9 and ½ years**
B. Pubic hair and 11 and ½ years
C. Testicular enlargement and 11 and ½ years
D. Pubic hair and 9 and ½ years

Ans A

52. **What age range does the Phallic/Oedipal stage of Freud' theory correspond to:**
A. 0–1 year
B. **3–6 years**
C. 2–3 years
D. 6–12 years

Ans B Phallic/Oedipal stage of Freud theory correspond to the preschool age (3–6 years); At the core of Freudian theory is the idea of body- centered (or, broadly, "sexual") drives.

53. **Earliest sign of puberty in females is?**
A. Growth spurt
B. Pubarche
C. **Thelarche**
D. Menarche

Ans C (breast budding) is the earliest sign of puberty in most girls, typically preceding pubarche, the growth spurt, and menarche.

54. **In boys which is the first visible sign of puberty?**
A. **Testicular enlargement**
B. Penile growth
C. Breast hypertrophy
D. Dark scrotum

Ans A

55. **True about Tanner stage III:**
A. **Penis increases in length**
B. Penis increases in width
C. Scanty hair at base of penis
D. Darkening of scrotum
E. More growth occur in boys than girls

Ans A For Classification of Sexual Maturity Status in Boys, In Tanner's stage 3 Pubic hair is darker, starting to curl Penis lengthens and testes becomes Larger Note: Peak increase in height in boys is usually seen in Tanner's stage 4.

56. **What is thelarche?**
A. Pubertal breast enlargement in boys
B. Breast enlargement in pregnancy
C. **Hormone related breast enlargement in girls**
D. Post hormonal therapy breast enlargement in postmeno-pausal females

Ans C

57. **Adolescent age starts at the age of:**
A. 7 years
B. **10 years**
C. 14 years
D. 17 years

Ans B

58. **Which one of the following is the correct order of events at puberty in a girl?**
A. Thelarche-pubarche-menarche-growth spurt
B. Pubarche-thelarche-growth spurt-menarche
C. Menarche-growth spurt-thelarche-pubarche
D. **Thelarche-pubarche-growth spurt-menarche**

Ans D

59. WHO defines adolescent age between:

A. 10–19 years
B. 10–14 years
C. 10–25 years
D. 9–14 years

Ans A

60. Areola and papilla forming secondary mound in adolescent girls is classified under which stage of sexual maturity rating (SMR)?

A. SMR stage
B. SMR stage
C. SMR stage
D. SMR stage

Ans C

Short Stature & Growth Disorders

61. Embryonic period is up to:

A. 8 weeks
B. 10 weeks
C. 12 weeks
D. 6 weeks

Ans A Embryo refers to the product of conception from the day of fertilization (generally 14th day post ovulation) to 8 weeks.

62. Which of the following age group, falls under early adolescence age group:

A. 8 – <10 years
B. 10–13 years
C. 14–16 years
D. 17–19 years

Ans B Adolescent group Early Middle Late Age range 10–13 years 14–16 years 17–19 years c. Thelarche

63. GH deficiency is diagnosed by:

A. Bone age less than chronological age
B. Bone age more than chronological age
C. Bone age equal to chronological age
D. Ratio of upper to lower segment of body is increased

Ans A In GH deficiency bone age is less than chronological age by 2 years

64. Which of the following statement regarding 'familial short stature' is incorrect?

A. Puberty is delayed
B. Bone age is normal
C. Growth is slow from birth
D. Growth hormone levels are normal

Ans A Normal variants of short stature: Birth weight Skeletal Timing of Cause and length maturation puberty Family history Other features Familial short stature Normal Normal Normal Of short stature Normal growth velocity Constitutional growth delay Normal Delayed Delayed Of delayed puberty Normal growth velocity

65. Which of the following is true about constitutional delay in growth?

A. Neonates with constitutional delay show anomalies at birth
B. IGF-1 levels are low for chronological age
C. Bone age is normal
D. IGF-1 levels are low for bone age

Ans B In constitutional delay, bone age is less than chronological age and IGF-1 levels are also low for chronological age.

66. Short stature, secondary to growth hormone deficiency is asso ciated with:

A. Normal body proportion
B. Low birth weight
C. Normal epiphyseal development
D. Height age equal to skeletal age

Ans A Short stature due to human growth hormone deficiency is characterized by:
• Normal ratio of upper to lower segment, so normal body proportion
• Bone age or epiphyseal development is less than chronological age by about 2 years
• Normal height and weight at birth; Delay in growth is usually observed after the age of one year.

67. Short stature are found in all except:

A. Malnutrition
B. Emotional deprivation
C. Congenital heart disease
D. Klinefelter syndrome
E. Hypothyroidism

Ans D Klinefelter syndrome with

68. True about constitutional delay in growth:

A. Normal length and weight at birth
B. Catch up growth at 6–12 months
C. Height velocity assume normal rate at 2–3 years
D. Bone age < chronological age
E. Puberty occurs at normal age

Ans A d. Bone age < chronological age

69. Bone age is less than chronological age in all except:

A. Familial short stature
B. Constitutional short stature
C. Under nutrition
D. Hypopituitarism

Ans A

70. Growth hormone deficiency is associated with all except:

A. Micropenis
B. Doll like facies
C. IUGR
D. Obesity

Ans C Growth hormone deficiency is associated with Hypoglycemia, Hypothyroidism, Micropenis, Truncal adiposity.

71. Cretinism is:

A. Disproportionate dwarfism
B. Short stature with long trunk
C. Short stature with short trunk
D. Long stature with long trunk

Ans A In hypothyroidism, patient has disproportionate short stature with short limbs.

72. Most common cause of short stature:

A. Familial
B. Constitutional
C. Hypothyroidism
D. Growth hormone deficiency

Ans B

Bone Age & Skeletal Development

73. Which of the following is/are true regarding development of child during 6 to 12 years of age:

A. Weight increases by 1–2 kg/yr
B. Head grow at the rate of 2–3 cm/yr
C. Growth occur continuously
D. Growth occur in spurts
E. Height increases at rate of 6–7 cm/yr

Ans D e. Height increases at rate of 6–7 cm/year During 6–11 years (middle childhood): growth averages 3–3.5 kg/year and 6–7 cm/year. Growth occurs discontinuously, in 3–6 irregularly timed spurts/year. Head grows only 2 cm in circumference throughout the entire period, reflecting a slowing of brain growth.

74. You order a wrist X-ray for a 2 months old child. Which carpal bone are you expecting to be present?

A. Capitate
B. Scaphoid
C. Lunate
D. Trapezoid

Ans A At birth, there is no calcification in the carpal bones. Although there is great individual variability, approximate ossification times are as follows: Capitate: 1–3 months Scaphoid: 4–6 years Hamate: 2–4 months Trapezium: 4–6 years Triquetrum: 2–3 years Trapezoid: 4–6 years Lunate: 2–4 years Pisiform: 8–12 years M neMonic Remember order of ossification of carpal bones Excluding the pisiform, an easy way to remember the order of ossification is

75. Which of the following X-ray should be advised for age determination between 1 and 13 years of age?

A. Shoulder
B. Wrists
C. Elbow
D. Iliac bones

Ans B

76. At the end of 1 year of age, the number of carpal bones seen in the skiagram of the hand is:

A. Nil
B. 1
C. 2
D. 3

Ans C

77. The first epiphyseal centre appears in:

A. Femur
B. Cuboid
C. Ossicles
D. Clavicle

Ans A First epiphyseal centre to appear are: Dorsal side of right hand The calcaneal and talar ossification centers & the distal femoral & proximal tibial epiphyseal ossification centers are found at the level of the knee.

Dysmorphology & Syndromes

78. Fetal growth is maximally affected by:

A. Insulin
B. Growth hormone
C. Cortisol
D. Thyroxine

Ans A Insulin and Insulin-like growth factor stimulate fetal growth.

79. Fetal alcohol syndrome comprises of:

A. Microcephaly
B. Deafness
C. Short palpebral fissure
D. All of the above

Ans D 896 Fetal alcohol syndrome:
• Growth retardation
• Microcephaly, small palpebral fissure
• Smooth philtrum, thin upper lip
• Increased risk of hearing loss of both conductive and sensorineural origin.

80. Infantile proportion in adult is seen in:

A. Morquio's disease
B. Achondroplasia
C. Hypothyroidism
D. Malnutrition

Ans B Normal body proportion In adults In infants Height = arm span Height (length) > arm span Upper segment = Lower segment Upper segment > Lower segment Infantile proportion refers to increased upper segment: Lower segment ratio, as seen in an infant. It is seen in causes of short limb dwarfism like Achondroplasia and Hypothyroidism. Reverse infantile body proportion is seen in Eunuchoidism, Marfan syndrome, Homocystinuria and Klinefelter's syndrome.

81. Upper segment to lower segment ratio in 3 years age child is:

A. 1.2:1
B. 1.3:1
C. 1.4:1
D. 1.6:1

Ans B Normal upper segment to lower segment ratio in children: Newborn → 1:7 – 1.9:1 ⇒ 3 years → 1:3 : 1 ⇒ 7 years → 1:1Q.

02

CHAPTER 02

Development



WHAT THIS CHAPTER COVERS

- Motor Milestones
- Fine Motor & Hand Skills
- Language & Speech Development
- Social & Adaptive Behavior
- Reflexes
- Cognitive & Learning
- Sexual Development
- Sleep Patterns
- Developmental Assessment
- Developmental Disorders

108 MCQs **10** subtopics

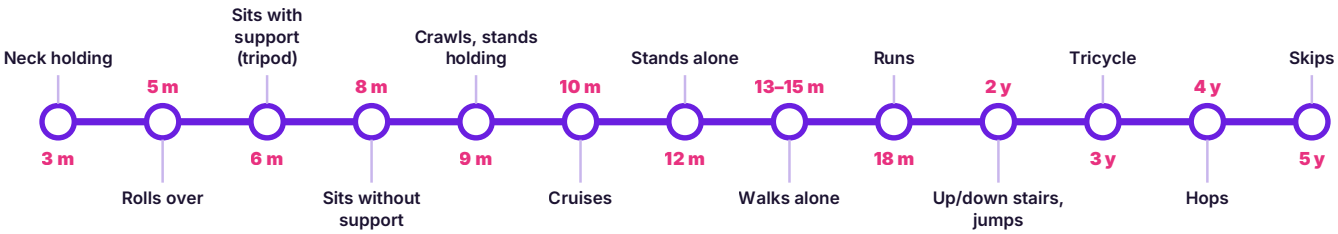
2.1 Principles of Development

- Development proceeds **cephalocaudal** (head → toe) and **proximodistal** (trunk → fingers); from **generalised mass activity to specific** responses.
- Sequence is constant; **rate varies** between children.
- Four domains: **gross motor, fine motor (vision), language (hearing), personal-social (adaptive)**.
- For preterm infants use **corrected age** (chronological age − weeks of prematurity) until **2 years**.
- **Developmental delay**: failure to attain milestone by the **upper limit** of normal age. **Global developmental delay**: significant delay in ≥ 2 domains in a child < 5 years.
- **Developmental quotient (DQ)** = developmental age / chronological age × 100. DQ < 70 = delay.

KEY MOTOR MILESTONES (MEAN AGE) — REMEMBER THE 'RUNG' AGES

Neck holding 3 m · Rolls over 5 m · Sits with support 6 m (tripod) · Sits without support 8 m · Crawls 9 m · Stands with support 9 m · Cruises 10 m · Stands alone 12 m · Walks alone 13 m · Runs 18 m · Walks upstairs (2 feet/step) 2 y · Jumps 2 y · Rides tricycle 3 y · Stairs alternate feet up 3 y, down 4 y · Hops on one foot 4 y · Skips 5 y.

2.2 Gross Motor Milestones



Gross-motor ladder — average ages

Age	Gross motor
1 month	Prone: lifts chin momentarily; head lag on pull-to-sit
3 months	**Neck holding**; prone – lifts head & chest on forearms (45–90°)
4 months	Prone – weight on hands, chest off; no head lag
5 months	**Rolls over** (front to back first); sits with back straight when supported
6 months	**Sits with support (tripod)**; bears weight on legs
8 months	**Sits without support**
9 months	**Crawls**, pulls to stand, **stands holding furniture**
10 months	Creeps; **cruises** (walks holding furniture)
11 months	Walks with one hand held
12 months	**Stands alone**, walks with one hand held / few steps
13–15 months	**Walks alone** (delay if not walking by **18 months**); creeps upstairs
18 months	**Runs** stiffly, walks upstairs with hand held, pulls toys

Age	Gross motor
2 years	Walks up & down stairs **2 feet per step**, **jumps**, kicks ball
3 years	**Rides tricycle**, alternate feet going **up** stairs, stands on one foot momentarily
4 years	**Hops on one foot**, alternate feet going **down** stairs
5 years	**Skips** on alternate feet, hops in a line

2.3 Fine Motor / Adaptive Milestones

Age	Fine motor & adaptive
1 month	Hands fisted; follows object to midline
2 months	Follows object **past midline**; hands open half the time
3 months	Hands open, **holds rattle** if placed; follows 180°
4 months	**Reaches for objects** with both hands (**bidextrous reach**); hands to midline, plays with hands
5 months	Grasps with whole hand (palmar grasp); takes objects to mouth (**mouthing** 5–6 m)
6 months	**Unidextrous reach**, **transfers objects hand to hand**, palmar grasp
7 months	Radial palmar grasp, rakes small objects
9 months	**Immature (inferior) pincer grasp**, bangs two cubes together
12 months	**Mature (neat) pincer grasp**, releases object on request, casting
15 months	**Tower of 2 cubes**, scribbles (15–18 m), feeds with spoon spilling
18 months	**Tower of 3–4 cubes**, turns **2–3 pages** at a time, scribbles spontaneously
2 years	**Tower of 6–7 cubes**, turns **single pages**, imitates **vertical line**, train of cubes
3 years	**Tower of 9–10**, copies **circle** (imitates cross), builds bridge of 3 cubes
4 years	Copies **cross (+)** and square (4–4½ y), builds gate of 5 cubes
5 years	Copies **triangle**; steps of 6 cubes
6 years	Copies **diamond**

COPYING SHAPES: 1-2-3-4-5-6

Vertical line 2 y · Circle 3 y · Cross 4 y · Square 4½ y · Triangle 5 y · Diamond 6 y. Tower of cubes ≈ age in years × 3 (2 y → 6 cubes; 3 y → 9 cubes).

2.4 Language & Hearing

Age	Language
Newborn	Startles to sound; alerts to voice
1–2 months	**Social smile** (6 weeks); coos (vowel sounds) at **2–3 months**
3–4 months	**Laughs aloud** (4 m), turns head towards sound
6 months	**Monosyllables** (ba, da, ma); babbles; turns to sound at ear level
9 months	**Bisyllables** (mama, dada – non-specific); understands "No"; waves bye-bye
12 months	**1–2 meaningful words** (mama/dada with meaning); follows 1-step command with gesture
15 months	**Jargon**; 4–6 words; points to 1 body part

Age	Language
18 months	**10–20 words**; points to 3 body parts; names one picture
2 years	**2–3 word sentences** (phrases), ~50–300 words, uses pronouns (I, me, you); 50% intelligible
3 years	**Telegraphic → 3–4 word sentences**, knows **full name, age and gender**, asks questions; 75% intelligible
4 years	Tells a story, counts 4 objects, sings song; 100% intelligible
5 years	Asks meaning of words, knows colours, counts 10 objects

RED FLAGS IN LANGUAGE

No babbling by **12 months**, no single words by **16–18 months**, no 2-word phrases by **24 months**, **any loss of language or social skills at any age** (regression → autism, Rett, Landau–Kleffner). **First step in any speech delay = formal hearing assessment (BERA/OAE).**

2.5 Social & Adaptive

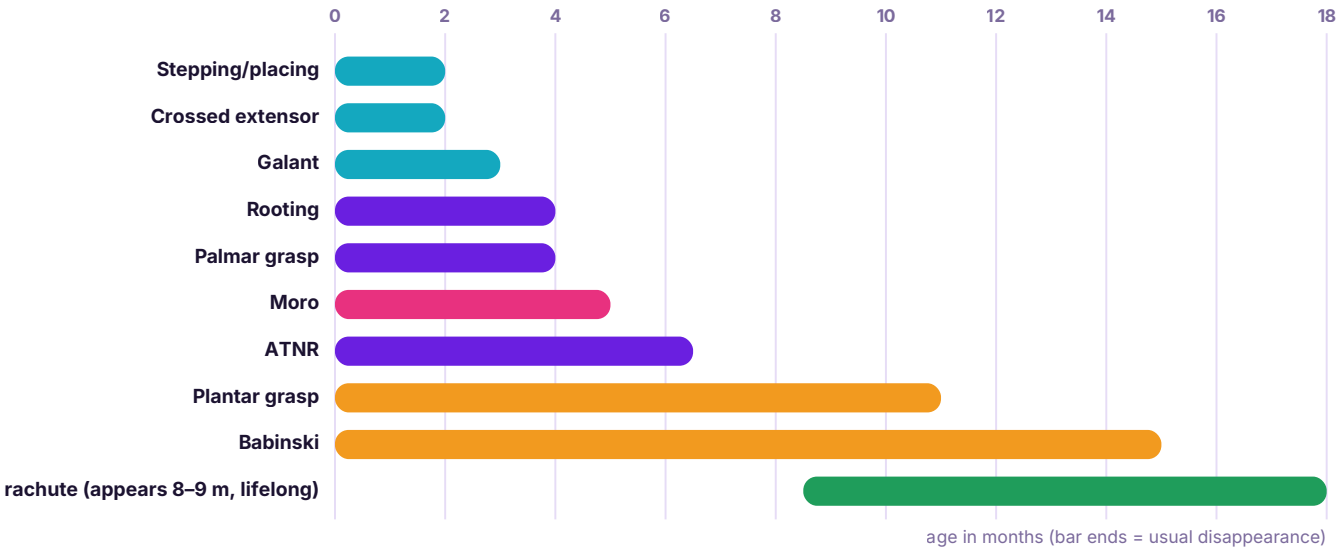
Age	Social / personal
6 weeks	**Social smile** (first social milestone)
3 months	Recognises mother, anticipates feeds
6 months	Recognises strangers – **stranger anxiety** begins (peaks 8–9 m); smiles at mirror image
9 months	Waves bye-bye, plays peek-a-boo, **object permanence**, separation anxiety
12 months	Comes when called, plays simple ball game, **drinks from cup**, cooperates in dressing
15 months	Feeds self; hugs parents; indicates wet pants
18 months	**Copies parents in household tasks** (domestic mimicry), **dry by day (partial)**, uses spoon well, takes off shoes/socks
2 years	**Parallel play**, asks for toilet, puts on simple clothes, **dry by day**
3 years	**Shares toys, knows gender**, **dry by night**, eats with spoon & fork, **associative/group play**
4 years	**Cooperative play**, goes to toilet alone, dresses & undresses, plays with imagination
5 years	Dresses & ties shoelaces, asks meaning of words, competitive games

- **Hand preference (dominance)** established at **3 years** (if earlier than 18 m → suspect contralateral hemiparesis).
- Bladder control: day by **2 y**, night by **3–5 y**; enuresis is diagnosed only after **5 years**.

2.6 Vision Milestones

Age	Vision
Birth	Fixes on face; acuity ~6/240 (20/400); prefers human face
1–2 months	Follows to midline/past midline; **binocular fixation** by 3 m
3 months	Follows 180°, convergence; **visual fixation & following fully developed**
4 months	Accommodation; looks at own hands
6 months	Adult-like colour vision, stereopsis begins; any **squint beyond 6 months is abnormal**
2–3 years	Acuity ~6/12
5–6 years	**Adult acuity 6/6**

2.7 Primitive Reflexes



Primitive reflexes – time of disappearance

Reflex	Appears	Disappears	Clinical note
Moro	28–32 wk GA (complete by 37 wk)	**3–6 months**	Asymmetric → **Erb's palsy, clavicle fracture**, hemiparesis; absent → severe CNS depression; persistence > 6 m → brain damage
Rooting	32 wk	3–4 months	
Sucking	32–34 wk (coordinated with swallow at **34 wk**)	4 months (becomes voluntary)	Basis of oral feeding readiness at 34 wk
Palmar grasp	28 wk	**3–4 months** (by 6 m)	Must disappear before voluntary grasp
Plantar grasp	28 wk	**9–12 months**	Disappears before walking
ATNR (fencing)	35 wk / 2 wk postnatal	**6–7 months**	Obligatory ATNR at any age = abnormal (CP)
Stepping / placing	35–37 wk	2 months	
Galant (trunk incurvation)	32 wk	2–4 months	
Crossed extensor	28 wk	1–2 months	
Babinski (extensor plantar)	Birth	**12–18 months** (up to 2 y)	

Postural (protective) reflexes – appear and persist for life

Reflex	Appears
Neck righting	4–6 months
Landau	3 months (fully 10 m), disappears ~2 y
Parachute (forward)	**8–9 months** – **persists for life**; asymmetry → hemiparesis
Lateral propping (side)	6–7 months
Backward propping	9–10 months

REFLEX RULES

Primitive reflexes are **brainstem/spinal**; they disappear as **cortical inhibition** matures. **Persistence beyond expected age** or **reappearance** = upper motor neuron lesion (cerebral palsy). Moro is the most commonly tested; **parachute** is the reflex that **never disappears**.

2.8 Red Flags & Developmental Assessment

By this age	Refer if NOT achieved
2 months	Social smile
4 months	Neck holding, following objects
6 months	Reaching for objects, turning to sound
9 months	**Sitting without support**
18 months	**Walking alone**, single words, pincer grasp lost or absent
2 years	Two-word phrases, running
3 years	Speech understood by strangers, copying circle
Any age	**Loss of acquired skills (regression)**, asymmetry of movement

Screening & assessment tools

Tool	Type / Age	Notes
DDST-II (Denver)	Screening, birth–6 y	125 items in 4 domains
Trivandrum Developmental Screening Chart (TDSC)	Screening, 0–6 y (0–2 y version)	Indian, simple, used by health workers
Baroda Development Screening Test	Screening	Indian adaptation of Bayley
Ages & Stages Questionnaire (ASQ)	Parent-completed screening	
M-CHAT-R/F	Autism screening at **18–24 months**	
Goodenough–Harris Draw-a-Person	Non-verbal IQ, 3–13 y	
Bayley Scales (BSID-III)	**Diagnostic**, 1–42 months	Gold standard for infants
Developmental Assessment Scale for Indian Infants (DASII)	Diagnostic, 0–30 months	Indian norms of Bayley
Stanford–Binet, **WISC** (6–16 y), WPPSI (2½–7 y)	IQ testing	
Vineland Social Maturity Scale	Adaptive behaviour / social quotient	
Gesell Developmental Schedules	Diagnostic	

2.9 Developmental Disorders – Overview

- **Intellectual disability (ID)**: IQ < 70 with deficits in adaptive functioning, onset in developmental period. Mild **50–69** (educable, ~85% of ID), Moderate **35–49** (trainable), Severe **20–34**, Profound **< 20**.
- MCC of ID (genetic): **Down syndrome**; MC **inherited** cause: **Fragile X**; MC **preventable** cause: **iodine deficiency/congenital hypothyroidism**; fetal alcohol syndrome is the MC **preventable non-genetic** cause in Western data.
- **Cerebral palsy, autism, ADHD, learning disability** – see Neurology & Behaviour chapters.

- **Hearing assessment**: OAE (screening, newborn), **BERA/ABR** (confirmatory, objective, any age); universal newborn hearing screening – **1-3-6 rule** (screen by 1 m, diagnose by 3 m, intervene by 6 m).

CLASSIC CASE STEMS

- "Child sits without support, transfers objects, says 'mama' non-specifically, has stranger anxiety" → **9 months**.
- "Walks alone, says 2 words, pincer grasp" → **12–13 months**.
- "Rides tricycle, copies circle, knows gender" → **3 years**.
- "Hops, copies cross, goes to toilet alone, tells story" → **4 years**.
- "Skips, copies triangle, ties shoelaces" → **5 years**.

2.10 Sleep in Children

Age	Total sleep / day
Newborn	16–18 h (REM ~50% of sleep; **sleep onset through REM**)
Infant (4–12 m)	12–16 h incl. naps
1–2 y	11–14 h
3–5 y	10–13 h
6–12 y	9–12 h
Adolescent	8–10 h (physiological **delayed sleep phase**)

Parasomnia	Sleep stage	Features
Night terrors (pavor nocturnus)	**NREM stage 3 (slow wave)**, first third of night	Child screams, inconsolable, autonomic arousal, **no recall** next morning
Sleepwalking (somnambulism)	NREM 3	Amnesia; safety measures
Nightmares	**REM**, last third of night	Child awakens, **vivid recall**, consolable
Bruxism	Stage N2	
Enuresis	NREM (any stage)	
Head banging / body rocking	Sleep onset	Usually benign, < 4 y

- **Obstructive sleep apnea** in children – MCC **adenotonsillar hypertrophy**; snoring, mouth breathing, behaviour problems, poor growth; investigation of choice **polysomnography**; Rx **adenotonsillectomy**.

Practice MCQs · Development

108 questions · correct option highlighted in green · cover the answer strip to self-test

Motor Milestones

1. A child of 4 years can do all of the following except:

A. Skip
B. Goes down stairs one foot per step
C. Tell a story
D. **Copies triangle**

Ans D Copying a triangle comes at 5 years.

2. When can a child kick a ball?

A. DACB
B. DCAB
C. **ACDB**
D. ADCB

Ans C 24 months A child can run and kick a ball at 2 years age and can go upstairs and downstairs 2 feet/step.

3. A baby can follow an object till 180°, can hold neck, can sit with support but cannot sit without support. The age of the baby is:

A. 1 month
B. 3 months
C. 5 months
D. **6 months**
E. 9 months

Ans D Following an object up to 180° comes at 2 months, Neck holding comes at 3 months, Sitting with support is attained at 6 months; So, the age of this baby is 6 months.

4. A child sits with support, identifies parts of body and builds tower of 4 cubes. Age is:

A. **1–2 yrs**
B. 2 yrs
C. 2–3 yrs
D. 4 yrs

Ans A Sitting with support comes at 6 months, identify 1 or more parts of body at 18 months and builds tower of 4 cubes at 18 months. So the age of the child is between 1 and 2 years.

5. An infant can sit with leaning forward on his hands. He bounces actively when made to stand. He laughs aloud and becomes concerned when the mother moves away. What is his most likely age?

A. 12 weeks
B. 16 weeks
C. 22 weeks
D. **28 weeks**

Ans D • An infant can sit with leaning forward on his hands (sitting with support) at around 5–6 months
• He bounces actively when made to stand at 7 months, laughs aloud at 4 months and becomes concerned when the mother moves away at around 7 months So, his most likely age is 7 months (28 weeks).

6. A female child has recently learned to eat with spoon without spilling, to dress and undress herself with supervision and to understand that she is a girl. These skills are first mastered between the ages of: (AIIMS Nov 2008) 30 Section 1: General Pediatrics

A. **2 and 3 years**
B. 3 and 4 years
C. 4 and 5 years
D. 5 and 6 years

Ans A

7. A child climbs with alternate steps, builds a tower of 8–9 cubes, tells 'I' but not his name and cannot say his age and sex, the probable age is:

A. 36 months
B. 24 months
C. **30 months**
D. 48 months

Ans C A child climbs with alternate steps at around 30 months, builds a tower 8–9 cubes between 2–3 years, tells 'I' but not his name at 2 years and tells his age and sex at 3 years. So, the probable age of this child is a little less than 3 years. Hence, among the given options, 30 months is the best answer.

8. The age at which the infant achieve early head control with bobbing motion when pulled to sit is

A. 2 mo
B. **3 mo**
C. 4 mo
D. 5 mo
E. 6 mo

Ans B This is a developmental skill of 3 months of age which include also; lifts head and chest with arms extended on prone; head above plane of body on ventral suspension; reaches toward and misses objects; waves at toy; listens to music and says "aah, ngah".

9. The child who's able to stands momentarily on 1 foot, makes tower of 10 cubes; imitates construction of "bridge" of 3 cubes; copies circle and imitates cross is

A. 24 mo old
B. 30 mo old
C. **36 mo old**
D. 42 mo old
E. 48 mo old

Ans C

10. A mother to a 4-year-old child who has pauses and repetitions of initial sounds visited outpatients department. Of the following, the MOST appropriate advice is

A. this is a normal phenomenon affecting about 5% of preschool children
B. there is no need for action as 80% of affected children recover by their own
C. **tries to reduce pressures associated with speaking**
D. I'll refer him to ENT specialist for further evaluation
E. I'll refer him to speech therapist

Ans C Developmental dysfluency or stuttering it includes a pauses and repetitions of initial sounds. Stress or excitement exacerbates these difficulties, which generally resolve on their own. Although 5% of preschool children will stutter, it will resolve in 80% of those children by age 8 yr. Children with stuttering should only be referred for evaluation if it is severe, persistent, or associated with anxiety, or if parental concern is elicited.

Fine Motor & Hand Skills

11. Child rolls over by:

A. 3 months
B. **6 months**
C. 7 months
D. 8 months

Ans B Once the Asymmetric Tonic Neck reflex (ATNR) disappears, baby learns to roll over from prone to supine and vice versa.

12. A child is able to copy a cross at what age?

A. **4 years**
B. 5 years
C. 2 and half years
D. 3 years

Ans A Copying a cross or rectangle comes at 4 years age.

13. Child is able to make tower of 5–6 cubes at which age?

A. 18 months
B. 24 months
C. **21 months**
D. 30 months

Ans C • Tower of 2 cubes: 15 months • Makes "bridge" of 3 cubes: 3 years
• Tower of 4 cubes: 18 months • Makes "gate" of 5 cubes: 4 years dna • Tower of 6–7 cubes: 24 months

14. Bidextrous grasp appears at:

A. 3 months
B. 4 months
C. **5 months**
D. 6 months

Ans C • 16 weeks: Tries to reach cube, but overshoots and misses.
• 20 weeks: Able to grasp voluntarily. Bidextrous approach
• 24 weeks: More mature grasp. Drops one cube when another is given
• 28 weeks: Unidextrous approach. Bangs cube on table. Transfers. Retains one when another is given.

15. Child starts making tower of 9 cubes by:

A. 18 months
B. 24 months
C. 30 months
D. **36 months**

Ans D

16. A baby can draw a triangle at the age of:

A. 3 years
B. **5 years**
C. 6 years
D. 7 years

Ans B

17. A child draws circle at:

A. 12 months
B. 24 months
C. 30 months
D. **36 months**

Ans D

18. A child can make a 3 cube tower at the age of:

A. 36 months
B. **16 months**
C. 20 months
D. 24 months

Ans B Milestones based on Tower of cubes: Age 15 months 18 months 2 years 3 years Number of cubes in tower 2 4 6–7 9–10

19. Handedness develops by:

A. 24 months
B. 36 months
C. 48 months
D. **60 months**

Ans D 36 months Handedness appears at 24 months, but is usually established by or developed by the 3rd year (36 months).

20. A child can ride a tricycle, copy a circle and knows his age and sex by the age of:

A. 30 months
B. 42 months
C. **36 months**
D. 48 months

Ans C

21. A child draws triangle at what age?

A. 3 years
B. 4 years
C. **5 years**
D. 7 years

Ans C 5 years with

22. Pincer grasp develops at what age?

A. 5–8 months
B. **9–11 months**
C. 12–15 months
D. 15–18 months

Ans B Immature pincer grasp usually develops by 9 months and mature pincer grasp by 12 months.

23. A child is able to build blocks of 4 cubes. His devel op-mental age is:

A. 12 months
B. 15 months
C. **18 months**
D. 30 months

Ans C

24. Pincer grasp is attained at ..... months:

A. 4
B. **10**
C. 12
D. 18

Ans B 10 As 9 months is not given in the options, b. 10 months is the best answer.

25. Handedness gets established at what age?

A. 12 months
B. 24 months
C. **36 months**
D. 48 months

Ans C

26. A boy can grasp a rattle and recently he is able to transfer objects from hand to hand. He should also be able to:

A. **Babble**
B. Say 'mama' or 'dada'
C. Sit with support
D. Stand with support
E. Able to walk

Ans A c. Sit with support The boy is able to transfer objects. So, his age is around 7 months. At 7 months he can also babble and sit with support.

27. A boy draws triangle but not diamond shape; what is his age?

A. 3 years
B. 4 years
C. **5 years**
D. 6 years

Ans C

28. Child changes a rattle from one hand to another at the age of:

A. 3 months
B. 5 months
C. **7 months**
D. 9 months

Ans C

29. The age at which the infant can reach an object, grasp it and bring it to mouth and seems exited when see the food is

A. **4 mo**
B. 5 mo
C. 6 mo
D. 7 mo
E. 8 mo

Ans A By this age he also able to lifts head and chest, with head in approximately vertical axis; achieve no head lag when pulled to sitting position, enjoys sitting with full truncal support when held erect, pushes with feet when sees raisin, but makes no move to reach for it, laughs out loud; and may show displeasure if social contact is broken.

30. You are specialist explaining the developmental implication of a 6-month-old baby boy who "transfers object hand to hand" to college students. Of the following, the BEST statement describing that is a/an

A. visuomotor coordination
B. voluntary release
C. **comparison of objects**
D. ability to explore small objects
E. increasing autonomy

Ans C The developmental implication reflects cognitive development.

31. Building a tower of 6 cubes by a 22-month-old child requires

A. visualmotor coordination
B. ability uses objects in combination
C. **visual, gross, and fine motor coordination**
D. able to link actions to solve problems
E. symbolic thought

Ans C

32. The age by which the child can makes tower of 9 cubes and imitates circular stroke is

A. 24 mo
B. **30 mo**
C. 36 mo
D. 42 mo
E. 48 mo

Ans B A 30 months old child also able to goes up stairs alternating feet, makes vertical and horizontal strokes, refers to self by pronoun "I"; knows full name, helps put things away and pretends in play.

33. The child who helps to undress; puts 3 words together (subject, verb, object); and handles spoon well, his/her MOST appropriate age is around

A. 15 mo
B. 18 mo
C. **24 mo**
D. 30 mo
E. 48 mo

Ans C

34. **The child who plays simple games (in "parallel" with other children); helps in dressing and washes hands, his/her MOST appropriate developmental age is around**

A. 15 mo
B. 18 mo
C. 24 mo
D. **30 mo**
E. 48 mo

Ans D

35. **The child who is able to imitates construction of "gate" of 5 cubes; draws a man with 2-4 parts besides head and identifies longer of 2 lines, his/her MOST appropriate developmental age is around**

A. 24 mo
B. 30 mo
C. **48 mo**
D. 54 mo
E. 60 mo

Ans C

Language & Speech Development

36. **Which of the following can be done by 15 months child?**

A. **Walks alone**
B. Transfers object hand-to-hand
C. Builds tower of 2 cubes
D. Speaks 10–15 words
E. Speaks two-word sentences

Ans A b.T ransfers object hand-to-hand; c. Builds tower of 2 cubes A child walks without support 13 months onwards, transfers objects at 7 months, builds a tower of 2 cubes at 15 months, speaks 10 words with meaning at 18 months, while speaks 2 word sentences at 2 years.

37. **Children can reach for objects by:**

A. **4 months**
B. 6 months
C. 8 months
D. 10 months

Ans A At 4 months a child tries to reach for an object but overshoots.

38. **Object permanence in a child is seen after:**

A. 6 months
B. **9 months**
C. 12 months
D. 15 months

Ans B Object permanence is the understanding that objects continue to exist, even when not seen and appears at 9 months.

39. **Child knows his/her sex at the age of:**

A. 1 year
B. **3 years**
C. 5 years
D. 7 years

Ans B

40. **A normal child has dysfluency of speech between:**

A. **2 and 4 years**
B. 4 and 6 years
C. 6 and 8 years
D. 8 and 10 years

Ans A Normal (physiological) dysfluency of speech is common between 2 and 4 years of age as expressive language rapidly outpaces speech-motor coordination; it usually resolves spontaneously.

41. **In development of language, a child is able to tell stories by:**

A. 1 year
B. 2 years
C. 3 years
D. **4 years**

Ans D Telling a story/poem or singing a song comes at 4 years age.

42. **Vocabulary of a 2-year-old child is:**

A. 20 words
B. 30 words
C. **100 words**
D. 200 words

Ans C Vocabulary at 18 months is 10-15 words, while at 2 years, it is 50–100 words.

43. **A child is able to say sentence of 6 words at:**

A. 2 years
B. 3 years
C. 4 years
D. **5 years**

Ans D M neMonic Between ages of 2 and 5 years, number of words in a typical sentence equals the child's age (e.g. 2 by age 2 year, 3 by age 3 year); So, a child speaks 5 word sentences at 5 years; So, out of the given options 5 years is the best answer

44. **Child starts speaking monosyllables at what age:**

A. 4 months
B. **6 months**
C. 8 months
D. 10 months

Ans B

45. **A normal child develops the ability to use 10 words with meaning at the age of:**

A. 12 months
B. 15 months
C. **18 months**
D. 24 months

Ans C

Social & Adaptive Behavior

46. A 2-month-old child is most likely to:

A. Show a positive parachute protective reflex
B. Hold head steady in a seated position
C. Lift head and chest off a flat surface with extended elbows
D. Sustain head level with the body when placed in ventral suspension

Ans D Discussing about the options one by one: Milestone Age at which attained a. Show a positive parachute protective reflex 9–10 months b. Hold head steady in a seated position 4 months c. Lift head and chest off a flat surface with extended elbows 3 months d. Sustain head level with the body when placed in ventral suspension 2 months

47. A 15-month-old child is able to do all the following except:

A. Crawl upstairs
B. Make a tower of 5 cubes
C. Name familiar objects
D. Point to familiar things

Ans B

48. At what age a child can eat food without spilling, identify her gender, say her full name, can dress self with supervision?

A. 2 yrs
B. 3 yrs
C. 4 yrs
D. 5 yrs

Ans B Eating food without spilling comes at 2 years, identifying her gender and saying her full name and dressing self with supervision comes at 3 years.

49. A child plays a simple ball game at:

A. 52 weeks
B. 36 weeks
C. 12 weeks
D. 48 weeks

Ans A Playing a simple ball game comes at 1 year or 52 weeks.

50. A 3-year-old child is expected to have all except:

A. Speak in sentences
B. Copy a circle
C. Hobble 5 steps
D. Climbs upstairs and downstairs

Ans C

51. All the following are recognizable teratogens EXCEPT

A. ethanol
B. antiepileptic medications
C. toxoplasmosis
D. hypothermia
E. mercury

Ans D Teratogens associated with gross physical and mental abnormalities include various infectious agents (toxoplasmosis, rubella, syphilis); chemical agents (mercury, thalidomide, antiepileptic medications, and ethanol), high temperature, and radiation.

52. A 16-year-old G1P1 mother complaining of reduced milk production and breasts engorgement. She stated that she feels unhappy and scared most of the times. Her husband is a soldier and she is living with his family. Of the following, the MOST appropriate action is to

A. refer her to obstetrician
B. explain the appropriate way of milk expression
C. advice milk stimulant medications
D. apply Edinburgh postnatal scale
E. stop breast feeding

Ans D Pediatricians must be alert to detect postpartum depression. Edinburgh Postnatal Depression Scale is the standard to apply; if the score achieved, the mother should be referred to psychiatrist.

53. A mother of a 2-week-fullterm baby noted doll's-eye movement of her baby eyes. Of the following, The MOST appropriate next action is to

A. reassures her by informing her that, this is a normal reaction
B. refer the baby to an ophthalmologist
C. takes a detailed history of perinatal period
D. order brain ultrasound
E. order brain MRI

Ans A This is normal finding in neonatal period.

54. A worried mother of a 4-year-old boy describing attacks of inconsolable crying episodes of her child, taking long time, she stated also that he prefers to play alone. Of the following, the MOST appropriate action is to

A. reassures her that this is a normal phenomenon of temper tantrum
B. seek more history regarding other skills and developmental domains
C. refer her to pediatric psychiatry
D. investigate social issues of the family
E. investigate for the child abuse

Ans B Classical temper tantrums of children usually result from fear, overtiredness, inconsistent expectations, or physical discomfort. Tantrums normally appear toward the end of the 1st yr of life and peak in prevalence between 2 and 4 years of age. Tantrums lasting more than 15 min or regularly occurring more than 3 times/day may reflect underlying medical, emotional, or social problems. In the scenario the episodes takes long time to resolve and the mother points to problem in social development.

55. A young couples of a 2-month-old baby girl with excessive crying seek your medical advice. Mother said that the baby is crying about 3 hr in a day, 2-3 days per week. She is intermittently spitting but she is gaining weight adequately. She is bottle fed baby. Of the following, the LEAST important advice is to

A. master the situation in relaxed manner
B. adhere to precry cues
C. change milk formula
D. avoid sensory overstimulation
E. reassure about benign nature of illness

Ans C Prolong crying episodes is present in about 20% of infants younger than 2 mo of age.Crying (longer than 3 hr/day, longer than 3 days/wk lasting longer than 1 wk) and more often crying/fussiness persisting longer than 3-5 mo might be associated with; child abuse; behavioral problems when an older child (anxiety, aggression, hyperactivity); decreased duration of breastfeeding and postnatal depression (uncertain which is the cause or effect).

56. A highly careful mother of a 10-month-old baby boy complaining that her baby has inadequate weight gain. She explained to you how she is so strict and so careful to feed her baby by spoon, but he was always refusing. Of the following, the LEAST helpful advice is to

A. respect infant independence
B. offer softer diet
C. use 2-spoons (1 for the child and 1 for the parent)
D. use finger foods
E. use high chair with tray table

Ans B Poor weight gain at this age often reflects a struggle between an infant's emerging independence and parent's control of the feeding situation.

57. A mother of 4-month-old baby boy complaining that the baby never goes to sleep unless he is rocked for quite long time. She needs to repeat the same issue whenever he is awake at night. She was surprised as no one of her previous babies had the similar behavior. She feels tired because of insufficient sleep. Of the following, the MOST common cause of this baby problem is

A. an early signs of ADHD
B. behavioral insomnia of childhood, sleep-onset association
C. primary sleep disorder
D. restless legs syndrome
E. sleep terrors

Ans B In this situation, the child learns to fall asleep only under certain conditions or associations, which typically require parental presence, such as being rocked or fed, and does not develop the ability to self-soothe. In babies, gradually weaning the child from dependence, and in older children introduction of more appropriate sleep associations that will be readily available to the child during the night (transitional objects, such as a blanket or toy), in addition to positive reinforcement (i.e., stickers for...

58. Parents of a 4-year-old boy complaining that he is refusing to go to bed, he remains active and playing. Of the following, the MOST likely cause of this child problem is

A. thyrotoxicosis
C. primary sleep disorder
D. limit setting; behavioral insomnia of childhood
E. behavioral insomnia of childhood, sleep-onset association

Ans D Is often the result of parental difficulties in setting limits and managing behaviour in general, and the inability or unwillingness to set consistent bedtime rules and enforce a regular bedtime in particular.

Reflexes

59. Parachute reflex appears at?

A. 3 months
B. 5 months
C. 7 months
D. 9 months

Ans D

60. A normally developing 10-month-old child should be able to do all of the following except:

A. Stand alone
B. Play peek a boo
C. Pick up a pellet with thumb and index finger
D. Build a tower of 3–4 cubes SOCIAL MILESTONES

Ans D Build a tower of 3–4 cubes

61. Which one of the following activities cannot be performed by a 7 month old infant?

A. Pivot
B. Cruise
C. Transfer objects
D. Enjoy mirror

Ans B Cruising or walking holding on to furniture comes at around 10–12 months.

62. A 3 months baby can do:

A. Social smile
B. Can sit without support
C. Transfer objects from right to left
D. Hold his neck
E. Can change position from prone to supine

Ans A d. Hold his neck Sitting without support appears at 8 months, transfers objects at 7 months and prone to supine at around 6–7 months. dna 55. c. 3 years

63. A 3-year-old child can do all, EXCEPT:

A. Eat with a spoon
B. Climb stairs running
C. Walk properly
D. Vocabulary of 20–100 words

Ans B

64. **Prenatal exposure to cigarette smoke is associated with**

A. lower birthweight
B. shorter length
C. changes in neonatal neurodevelopmental status
D. **neonatal diabetes**
E. learning problems

Ans D Prenatal exposure to cigarette smoke is associated with lower birthweight, shorter length, and smaller head circumference, as well as changes in neonatal neurodevelopmental assessments. Later, these children are at increased risk for learning problems, externalizing behavior disorders, and long-term health effects.

65. **A medical student asked you during the morning round about his observation of infants cry in response to the cry of another infant. Of the following, the MOST appropriate answer is that it represent**

A. **an early sign of empathy development**
B. a sign of good hearing
C. a startle reflex to a loud sound
D. an early sign of fear development
E. an early sign of autistic behavior

Ans A

Cognitive & Learning

66. **An 8-week-infant can do all the following except:**

A. **Head control (PGI Dec 11)**
B. Lift its head up to horizontal line in ventral suspension
C. Follows red object up to 180 degrees
D. Social smile
E. Turns head towards sound

Ans A Head control; e. Turns head towards sound Milestone Age at which attained a. Head control 3 months b. Lift its head up to horizontal line in ventral suspension 2 months c. Follows red object up to 180 degrees 2 months d. Social smile 2 months e. Turns head towards sound 4 months

67. **Child knows his sex by what age?**

A. 12 months
B. 24 months
C. **36 months**
D. 48 months

Ans C A child knows his sex by 36 months.

68. **Range of IQ in mild mental retardation is?**

A. <
B. **50–70**
C. 70–100
D. >100

Ans B Mild intellectual disability (ID) is IQ between 50-70, while severe ID is IQ <50

69. **Child copies a circle at the age of?**

A. 1 year
B. 2 years
C. **3 years**
D. 4 years

Ans C A child can copy a circle at 3 years, a cross at 4 years, a square at 5 years, and a triangle at 6 years — a classic developmental milestone sequence.

70. **A 6-year-old child has a DQ of 50. Which of the following can the child do?**

A. **Identify colors**
B. Read a sentence
C. Ride a bicycle
D. Copy a triangle

Ans A DQ = Developmental age x 100/ actual age. In the given question, a 6 year old child has a DQ of 50; so his developmental age is 3 years. So, he can do milestones corresponding to 3 years age only; At 3 years, a child can name 2 colors

71. **By the age of 7 months the infant is able to do all the following EXCEPT**

A. transfer object from hand to hand
B. bounces actively
C. **cruises**
D. grasp uses radial palm
E. roll over

Ans C or walks' holding to furniture is by 10 months of age.

72. **These facts are true regarding the developmental stage of preschool children**

A. handedness is achieved by 3 years of age
B. boys are usually later than girls in achieving bladder control
C. **knowing gender by 4 years**
D. egocentric thinking
E. musturbation

Ans C Bowel and bladder control emerge during this period, with "readiness" for toileting having large individual and cultural variation. Bed-wetting is normal up to age 4 yr in girls and age 5 yr in boys. Thinking in this period characterized by magical thinking (the child might believe that people cause it to rain by carrying umbrellas, that the sun goes down because it is tired), and egocentrism which refers to a child's inability to take another's point of view.

73. **Regarding the physical growth of preschool children (3-5 yr); all the following are true EXCEPT**

A. **4-5 kg weight increment/yr**
B. 7-8 cm height increment/yr
C. head will grow only an additional 5-6 cm up to18 yr
D. all 20 primary teeth have erupted by 3 yr
E. average wt 20 kg and height 40 inch by 4 yr

Ans A 2kg/yr.

Sexual Development

74. **A 4-year-child can perform which one of the following?**

A. **Can hop on single leg for 15 feet (AIIMS Dec 08)**
B. Can skip without falling to either side
C. Can stand on one feet for 20 seconds
D. Can walk down the stairs with alternating steps holding on the iron rail FINE MOTOR

Ans A Can hop on single leg for 15 feet; d. Can walk down the stairs with alternating steps holding on the iron rail Milestone Age at which attained a. Can hop on single leg for 15 feet 4 years b. Can skip without falling to either side 5 years c. Can stand one feet for 20 seconds 5 years d. Can walk down the stairs with alternating steps holding on the iron rail 4 years Note: (d) is a better answer than (c), because child hops on a single leg at 4 years, but hopping on 1 leg for a distance as long as 15

75. The developmental implication of developmental miles tone uncovers toys after seeing it hidden is:

A. Symbolic thought
B. Beginning of labelling
C. Object permanence
D. Self-discovery
E. Cause and effect

Ans C Uncovers toy (after seeing Pretend play with doll (e.g., Milestone Stares at own hand it hidden) gives doll bottle) Developmental implication Self-discovery Object permanence Symbolic thought

76. A 2-year-old child can do:

A. Can ride tricycle
B. Climb up and downstairs with one step each time
C. Knows sex and age
D. Handles spoon well
E. Can read story with picture

Ans B d. Handles spoon well Discussing about the options one by one. Milestone Age at which attained a. Can ride tricycle 3 years b. Climb up and downstairs with one step each time (means with 2 feet per step) 2 years c. Knows sex and age 3 years d. Handles spoon well 2 years e. Can read story with picture 4–5 years

77. Which of the following milestones is not developed at 1 year?

A. Stand without support
B. Plays peek a boo
C. Can speak "mama" and "papa"
D. Draws a circle

Ans D A child is able to draw a circle at around 3 years age. 57. Ans a. 2 and 4 years fo Language development occurs most rapidly between 2 and 5 years of age. The period of rapid language acquisition is also when weiveR developmental dysfluency and stuttering are most likely to emerge

78. A 10-month-old child cannot do:

A. Change cube from one hand to another
B. Can build a tower of six cubes
C. Can pull to stand
D. Can speak a sentence of 4 to 5 words
E. Pincer grasp

Ans B d. Can speak a sentence of 4 to 5 words Building a tower of 6 cubes comes around 2 years age and talking a sentence of 4–5 words the given milestones appear within or at the age of 10 months.

79. A 10-month-old child can do all the following EXCEPT

A. follows one-step command without gesture
B. says "mama" or "dada"
C. points to objects
D. speaks first real word
E. inhibition to "no"

Ans D This is a 12 months developmental skill.

80. The birthweight usually quadruples by the age of

A. 1.5 yr
B. 2 yr
C. 2.5 yr
D. 3 yr
E. 3.5 yr

Ans C

Sleep Patterns

81. Absence of which of the given milestones in a 3-year- old child should be called 'delayed' development?

A. Hopping on one leg
B. Drawing square
C. Feeding by spoon
D. Catching a ball reliably

Ans C • Hopping on one leg, drawing a square & catching a ball reliably appear at 4 years.
• Feeding by spoon comes by 18 months, so its absence in a 3 year child is considered abnormal.

82. Which of the following cannot be done by a 3 years old child?

A. Draw a triangle
B. Draw a circle
C. Can arrange 9 cubes
D. Can go up and down the stairs

Ans A

83. A 40-week-old infant can do all the following except:

A. Waves bye bye
B. Transfer object from one hand to another
C. Sits without support
D. Makes a tower of 3–4 cubes

Ans D 40 week old infant is around 10 months old; Tower of 3–4 cubes comes by around 18 months age.

84. Behavioral observation audiometry is used as a screening test for hearing in age group:

A. <6 months
B. 6–30 months
C. 30–60 months
D. >60 months

Ans A Behavioral observation Visual reinforcement Type of audiometry Play audiometry audiometry audiometry Used in age group <5 months 6–30 months 30 months to 5 years

85. An 18-month-old infant can do all except:

A. Climbing upstairs (PGI Dec 03)
B. Can follow mother's activities
C. Can turn 2–3 pages at a time
D. Can say 2–3 words
E. Can make tower of 8 cubes Chapter 2: Development 31 Answers

Ans E Can make tower of 8 cubes Discussing about the options one by one: Milestone Time of appearance a. Climbing upstairs A child can climb upstairs holding the rail at 18 months b. Can follows mother's activities This is domestic mimicry, that appears at 18 months c. Can turn 2–3 pages at a time 18 months d. Can say 2–3 words At 1 year, a child can speak 2–3 words with meaning e. Can make tower of 8 cubes Between 2 and 3 years age Answers and at 5 years, 4 colors. 20/e p 65-77 comes at 4–5 years. Rest of with M

86. You are observing a 15-month-old toddler, he was able to do all the following

A. walks alone
B. makes tower of 3 cubes
C. inserts raisin in a bottle
D. responds to his/her name
E. identifies 1 or more parts of body

Ans E This is an 18 months developmental skill.

87. Between 2-6 months of life, the infant start to achieve a regular sleep–wake cycles. All the following are true about infant sleep during this period EXCEPT

A. total sleep hours are about 14-16 hr/24 hr
B. sleeps about 9-10 hr concentrated at night
C. sleeps 2 naps/day
D. sleep electroencephalogram shows the mature pattern
E. the sleep cycle time is similar to that of adults

Ans E The sleep cycle remains shorter than in adults (50-60 min vs approximately 90 min). As a result, infants who arouses frequently during the night, considered as behavioral sleep problem.

88. The following statements about sleep are true EXCEPT

A. melatonin which is secreted in dark-light cycles is secreted from hypothalamus
B. slow-wave sleep is the first cycle of sleep
C. rapid eye movement (REM) sleep is responsible for dreams
D. both cycles are needed for sufficient sleep
E. REM sleep protect brain from injury

Ans A Melatonin is secreted from penile body. Two sleep cycles; the slow-wave sleep (SWS) (i.e., n3, delta, or deep sleep) appears to be the most restorative form of sleep; it is entered into relatively quickly after sleep onset, it is preserved in the face of reduced total sleep time, and it increases (rebounds) after a night of restricted sleep. Rapid eye movement (REM) sleep (stage r or "dream" sleep) appears to be involved in (1) completing vital cognitive functions, such as the consolidation of memory;

89. The MOST common cause of sleeping difficulty in the first 2 months of life is

A. gastro-esophageal reflux
B. formula intolerance
C. colic
D. developmentally sleeping behavior
E. recurrent self resolving intussusceptions

Ans D Most sleep issues that are perceived as problematic at this stage represent a discrepancy between parental expectations and developmentally appropriate sleep behaviors. Newborns that are noted by parents to be extremely fussy and persistently difficult to console are more likely to have one of the mentioned medical issues.

90. All the following are included in basic principles of healthy sleep EXCEPT

A. set bedtime routine
B. avoid stimulating activities as playing computer games
C. makes sure your child spends time outside every day
D. gives a heavy meal within an hour or 2 of bedtime
E. keeps the television set out of your child's bedroom

Ans D Other advices also include; don't send your child to bed hungry. A light snack (such as milk and cookies) before bed is a good idea. Heavy meals within an hour or 2 of bedtime, however, may interfere with sleep. Avoid products containing caffeine for at least several hours before bedtime. These include caffeinated sodas, coffee, tea, and chocolate. Keep your child's bedroom quiet and dark. A low-level night light is acceptable for children who find completely dark rooms frightening.

91. All the following are compatible with the definition of obstructive sleep apnea

A. repeated episodes of prolonged upper airway obstruction
B. apnea
C. ≥30% reduction in airflow
D. ≥ 30% O2 desaturation
E. disrupted sleep

Ans D ≥ 3% O2 desaturation.

92. Of the following, the MOST common cause of obstructive sleep apnea in children is

A. adenotonsillar hypertrophy
B. obesity
C. allergies
D. craniofacial abnormalities
E. pharyngeal reactive edema due to gastroesophageal reflux

Ans A

93. Persons with Down syndrome are at particularly high risk for obstructive sleep apnea with up to 70% prevalence. All the following are considered as risk factors EXCEPT

A. peculiar facial anatomy
B. hypotonia
C. developmental delay
D. central adiposity
E. hypothyroidism

Ans C Macroglosia.

94. All the following are parasomniac disorders EXCEPT

A. narcolepsy
B. sleepwalking
C. sleep terror
D. confusional arousal
E. nightmare

Ans A is a recurrent episode of excessive daytime sleepiness, while parasomnia is an episodic nocturnal behavior that often involves cognitive disorientation and autonomic and skeletal muscle disturbance.

Developmental Assessment

95. The milestones of a 3-year-old child are considered delayed if he is unable to:

A. Hop on 1 foot
B. Use spoon effectively
C. Copy a square
D. Reliably catch a ball

Ans B Hop on 1 foot, copying a square & catching a ball reliably, comes at 48 m (4 yrs) A child is able to feed self at 18 months, but handles spoon effectively at 24 months (2 yrs)

96. An 18 months baby can do?

A. Hide and seek game
B. Write alphabet
C. Say short sentence
D. Run

Ans A Hide and seek game or peek a boo comes at around 10 months age.

97. All of the following suggest a developmental delay in Questions milestones except:

A. Absence of pincer grasp at 9 months
B. Not climbing stairs up and down at 2 years
C. Not speaking 2 word phrases at 18 months
D. Not able to sit at 9 months

Ans A

98. In 'Milestones of Development' 'Listening' refers to which type of development?

A. Motor
B. Language
C. Adaptive
D. Socio-personal

Ans B

99. In embryonic period, all the following are true EXCEPT

A. formation of blastocyst by 8 days
B. formation of endoderm and ectoderm by 2 weeks
C. formation of mesoderm by 6 weeks
D. formation of human like embryo by 8 weeks
E. the crown-rump length is about 3 cm

Ans C Formation of mesoderm by 4 weeks.

100. The child who continue to search for a hidden subject, has achieved the developmental age of

A. 7 mo
B. 9 mo
C. 11 mo
D. 13 mo
E. 15 mo

Ans B A major milestone is the achievement by 9 mo of object permanence (constancy), the understanding that objects continue to exist, even when not seen. At 4-7 month of age, infants look down for ball that has been dropped but quickly give up if it is not seen. With object constancy, infants persist in searching. They will find objects hidden under a cloth or behind the examiner's back.

101. Regarding the physical growth of middle childhood (6-11 yr); all the following are true EXCEPT

A. 3-3.5 kg wt increment/yr
B. 6-7 cm height increment/yr
C. brain myelinization stops by 8 yr
D. first deciduous tooth falls by 6 yr
E. risk for future obesity

Ans C Brain myelinization continues into adolescence, with peak gray matter at 12-14 yr.

Developmental Disorders

102. Which of the following milestones is developed in child between 6 to 9 months of age?

A. Can point something with index finger
B. Swaps object from one palm to another
C. Can hold object with thumb and index finger
D. Can voluntarily drop object
E. Can extend arm

Ans B c. Can hold object with thumb and index finger Milestone Age at which attained a. Can point something with index finger 15 months b. Swaps object from one palm to another 7 months c. Can hold object with thumb and index finger 9 months d. Can voluntarily drop object 12 months e. Can extend arm (helps to undress) 2 years

103. Red flag sign in child development, if not attained?

A. Vocalization at 2 months
B. Walking at 12 months
C. Single word at 12 months
D. Social smile at 6 months

Ans D For red flag signs for some important milestones

104. Which of the following appears 1st in child?

A. Creeping
B. Crawling
C. Mirror play
D. Pincer grasp

Ans A (moving with the abdomen on the ground) appears before crawling (on hands and knees), pincer grasp, and mirror play in the normal developmental sequence.

105. **A 4-year-old child cannot do which of the following?**

A. Count 10 pennies
B. Hop on one foot
C. Copy a bridge
D. Identify the longer of two lines

Ans A A child is able to count 10 pennies at the age of 5 years. Important milestones attained at 4 year age: Gross motor Fine motor Social Language
• Hops on 1 foot • Copies bridge from a model • Plays with several children • Counts 4 pennies accurately
• Throws ball overhand • Imitates" gate" of 5 cubes • Role-playing • Tells a story or a poem
• Climbs well • Copies cross and square • Goes to toilet alone • Sings a song
• Draws man with 2-4 parts
• Identifies longer of 2 lines
• Uses scissors to cut out pictures

106. **In fetal period, all the following are true EXCEPT**

A. by 10 weeks the midgut returns to abdomen
B. by 12 weeks external genitalia becomes clearly distinguishable
C. by 24 weeks surfactant production begun
D. by 26 weeks recognizable human face formed
E. during third trimester the weight triples

Ans D Usually by 10 weeks.

107. **Of those babies who have prolonged crying episodes in the first 2 mo of life, the percentage that will remain having similar episodes is about**

A. 1%
B. 5%
C. 10%
D. 30%
E. 50%

Ans B

108. **The first permanent tooth to erupt is**

A. central incisor at 6 yr
B. molar at 6 yr
C. premolar at 6-7 yr
D. lower canine at 6-7 yr
E. upper canine at 6-7 yr

Ans B The first permanent molars usually erupt between ages 6 and 7 years. For that reason, they often are called the "six-year molars." They are among the "extra" permanent teeth in that they don't replace an existing primary tooth. These important teeth sometimes are mistaken for primary teeth. The six-year molars also help determine the shape of the lower face and affect the position and health of other permanent teeth. The first deciduous teeth to fall are central incisors.

03

CHAPTER 03

Nutrition and Malnutrition



WHAT THIS CHAPTER COVERS

- Breastfeeding & Breast Milk
- Formula Feeding & Weaning
- Nutritional Requirements & RDA
- Protein-Energy Malnutrition
- Kwashiorkor & Marasmus
- Vitamin A Deficiency
- Vitamin D & Rickets
- Vitamin K Deficiency
- Vitamin B Complex Deficiency
- Vitamin C & Scurvy
- Iron Deficiency & Anemia
- Zinc, Copper & Trace Elements
- Iodine Deficiency & Goiter
- Obesity & Overnutrition
- Nutritional Rehabilitation

205 MCQs **15** subtopics

3.1 Breastfeeding

- **Initiate within 1 hour** of birth (including after caesarean section) – "golden hour"; skin-to-skin contact.
- **Exclusive breastfeeding for 6 months** (no water, no honey, no ghutti); continue breastfeeding **till 2 years or beyond** with complementary feeds from 6 months.
- Feed on demand, **8–12 times/day**; adequacy: **≥ 6 wet nappies/day**, sleeps 2–3 h after feeds, weight gain ≥ 20 g/day (500 g/month).
- **Prelacteal feeds are not given.**

Types of breast milk

Milk	Period	Features
Colostrum	First **3–4 days**	Thick yellow, small volume (10–40 mL/day); **rich in protein, IgA, lactoferrin, vitamins A, E, K, zinc**, leucocytes; low fat & lactose; laxative (helps meconium) – "**first immunisation**"
Transitional milk	Day 4–14	↑ fat & lactose
Mature milk	After 2 weeks	65–70 kcal/dL
Foremilk	Start of feed	Watery, rich in **protein, lactose, water** – quenches thirst
Hindmilk	End of feed	**Rich in fat (energy)** – satiety & weight gain; so **empty one breast before switching**
Preterm milk	Mother of preterm	More protein, Na, Cl, IgA, lactoferrin & calories than term milk

Composition (per 100 mL)

	Human milk	Cow's milk
Energy	67 kcal	67 kcal
Protein	**1.1 g** (whey : casein **60 : 40** → 70:30 early)	**3.3 g** (whey : casein **20 : 80**)
Fat	3.5–4.5 g – PUFA, **DHA**, **lipase**	3.5–4 g – saturated
Lactose	**7 g**	4.5 g
Calcium : phosphorus	2 : 1 (better absorption)	1.2 : 1 (high phosphate → neonatal tetany)
Iron	0.05 mg (low, but **50% absorbed**)	Low, 10% absorbed
Sodium	Low (renal solute load ↓)	High
Vitamins	**Deficient in vitamin K, D**; adequate vit C if mother well-fed	Deficient in **vitamin C, iron**

WHAT BREAST MILK LACKS

Vitamin K (→ give 1 mg IM at birth), **vitamin D** (→ 400 IU/day from birth for breastfed infants), and **iron** after 6 months; also low in vitamin B12 in vegan mothers.

- **Anti-infective factors**: secretory **IgA** (most important), **lactoferrin** (binds iron, bacteriostatic to E. coli), **lysozyme**, **bifidus factor** (promotes **Lactobacillus bifidus** → acidic stool), macrophages & lymphocytes, interferon, complement, **PABA** (anti-malarial), oligosaccharides, mucins, **anti-staphylococcal factor**.
- Main whey protein of human milk: **α-lactalbumin**; of cow's milk: **β-lactoglobulin** (allergen).
- Taurine, cystine-rich; low methionine & phenylalanine (preterm-friendly).

Physiology & technique

- **Prolactin** – milk secretion; secreted **more at night** (night feeds important); **oxytocin** – milk ejection ("let-down" reflex, inhibited by anxiety/pain).
- Good **attachment**: chin touching breast, mouth wide open, lower lip everted, **more areola visible above than below**. Good **positioning**: head & body in line, facing breast, body close, whole body supported.
- Expressed breast milk storage: **6–8 h at room temperature**, **24 h in refrigerator**, **3 months in freezer** (−20 °C); never boil/microwave – warm in warm water.

Contraindications to breastfeeding

Absolute	NOT contraindications (continue BF)
Classic galactosemia (infant)	Maternal **hepatitis B** (give HBIG + vaccine), hepatitis C
Maternal **HTLV-1/2**	Maternal TB (start INH prophylaxis to baby; separate only if mother is sputum+ and untreated in some protocols)
Untreated **brucellosis**	**HIV in India** – exclusive BF with **maternal ART + infant nevirapine prophylaxis** (avoid mixed feeding)
Maternal anticancer drugs, radioactive isotopes (temporary), drugs of abuse	Mastitis (continue from affected side too), breast abscess (feed from other side)
Active HSV lesions on breast (that side)	Maternal fever, common cold, COVID-19 (with mask)
PKU – partial BF with Phe-free formula (not absolute)	Jaundice in infant (breast milk jaundice – continue)

DRUGS & LACTATION

Contraindicated: **antineoplastics (methotrexate, cyclophosphamide), lithium (relative), amiodarone, ergotamine, bromocriptine (suppresses lactation), radioactive iodine, chloramphenicol, tetracyclines (prolonged), gold salts, oestrogen-containing pills (↓ milk)**. Safe: penicillins, cephalosporins, paracetamol, ibuprofen, heparin/warfarin, insulin, most anti-epileptics (monitor phenobarbital), **progestin-only pill/LAM**. Galactagogues: **domperidone**, metoclopramide (not routinely recommended).

Breastfeeding problems

- **Lactational amenorrhoea method (LAM)**: > 98% effective if exclusive BF, amenorrhoeic, < 6 months postpartum.
- **Breast milk jaundice**: onset after 1st week, peaks 2–3 wk, due to **β-glucuronidase** / pregnanediol in milk; continue BF.
- **Breastfeeding jaundice**: 1st week, due to inadequate intake/dehydration.
- Inverted nipples – syringe method; sore/cracked nipple – correct attachment; engorgement – frequent feeding & expression.
- **Baby Friendly Hospital Initiative (BFHI)** – WHO/UNICEF **"Ten steps to successful breastfeeding"** (1991). **IMS Act** (India 1992, amended 2003) prohibits promotion of infant milk substitutes for children **< 2 years**. **World Breastfeeding Week: 1–7 August** (WABA). MAA programme (2016).

3.2 Complementary Feeding & Weaning

- Start at **6 completed months**; continue BF.
- Principles: **soft, energy-dense, home-made** (cereal-pulse mix e.g. khichdi), add **oil/ghee** (energy density ≥ 0.8–1 kcal/g), introduce one new food at a time, **responsive feeding**, hygiene, no bottles.
- **Frequency**: 6–8 m → 2–3 meals/day; 9–11 m → 3–4 meals + 1–2 snacks; 12–24 m → 3–4 meals + 1–2 snacks.
- Avoid **honey < 1 y** (infant botulism), **cow's milk as main drink < 1 y** (occult GI blood loss, iron deficiency), added salt/sugar, whole nuts (aspiration).

IYCF – COMPLEMENTARY FEEDING 'FATVAH'

Frequency, **A**mount, **T**exture (thickness), **V**ariety, **A**ctive/responsive feeding, **H**ygiene.

3.3 Nutritional Requirements

Age	Energy	Protein
0–6 months	~90–100 kcal/kg/day (ICMR 2020: ~92)	1.16–1.4 g/kg/day
6–12 months	~80 kcal/kg/day	1.7 g/kg
1–3 years	~1000–1100 kcal/day (100 kcal/kg)	1.2 g/kg
Rough rule	**100 kcal/kg for first 10 kg**, +50 kcal/kg for next 10 kg, +20 kcal/kg thereafter	10–15% of energy

- Distribution of energy: carbohydrate 50–60%, fat 25–35% (infants up to 50%), protein 10–15%.
- **Water**: Holliday–Segar (100 / 50 / 20 mL/kg) – see Fluids chapter.
- Essential fatty acids: **linoleic (ω-6)** and **α-linolenic (ω-3)**; deficiency → **scaly dermatitis**, alopecia, thrombocytopenia, poor wound healing.
- Essential amino acids: PVT TIM HALL — Phenylalanine, Valine, Threonine, Tryptophan, Isoleucine, Methionine, **Histidine (essential in infants)**, Arginine (semi-essential), Leucine, Lysine.

Infant formula

- Standard formula (cow's milk based) – modified to resemble breast milk; **1 scoop to 30 mL water**.
- **Soy formula** – galactosemia, primary lactase deficiency, vegan families; **not** for preterm or prevention of allergy.
- **Extensively hydrolysed formula** – cow's milk protein allergy (CMPA); **amino acid formula** – severe CMPA/ multiple allergies.
- **Preterm formula / human milk fortifier** – for VLBW infants (< 1.5 kg) on breast milk.

3.4 Protein-Energy Malnutrition (PEM)

Classification systems

System	Basis	Grades
IAP (1972)	Weight-for-age (% of 50th centile)	Normal > 80%; **I 71–80**; **II 61–70**; **III 51–60**; **IV ≤ 50%**; add "K" if oedema
Gomez	Weight-for-age	Normal > 90%; I 75–89; II 60–74; III < 60
Waterlow	**Wasting** (weight-for-height) & **stunting** (height-for-age)	Distinguishes acute vs chronic
Wellcome	Weight-for-age + oedema	60–80% + oedema = **kwashiorkor**; 60–80% no oedema = **undernutrition**; < 60% no oedema = **marasmus**; < 60% + oedema = **marasmic kwashiorkor**
WHO (current)	Z-scores	See below

Severe acute malnutrition (SAM) – WHO / IAP definition (6–59 months)

Any one of: 1. **Weight-for-height/length < −3 SD**, or 2. **MUAC < 11.5 cm**, or 3. **Bilateral pitting pedal oedema** (nutritional oedema).

- **Moderate acute malnutrition (MAM)**: WFH −2 to −3 SD or MUAC 11.5–12.5 cm.
- Infants < 6 months: SAM = WFL < −3 SD or oedema (MUAC not used).

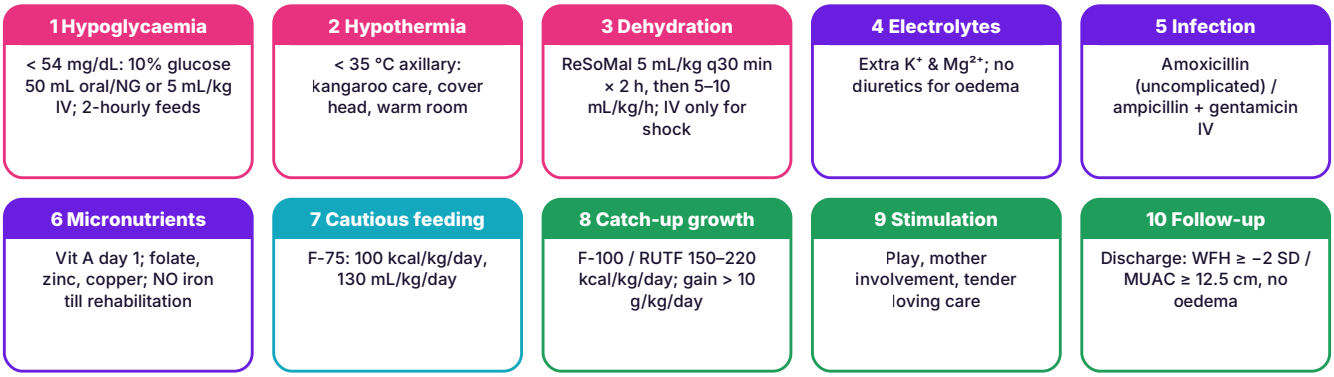
Kwashiorkor vs Marasmus

Feature	Kwashiorkor	Marasmus
Meaning	"Disease of the displaced child" (Ga language, Cicely Williams)	"Wasting"
Deficiency	Relative protein deficiency; **aflatoxin, free radicals**, infection	Total energy (calorie) deficiency
Age	1–4 y (after weaning)	< 1 y
Oedema	**Present (essential for diagnosis)**	**Absent**
Weight	60–80% expected	**< 60%**
Muscle wasting	Present, masked by oedema	**Severe, "skin & bones", old-man facies**
Subcutaneous fat	Preserved	Lost (**buccal pad of fat last to go**)
Mental change	**Apathetic, irritable, miserable**	Alert, hungry
Appetite	**Anorexia**	Good/voracious
Hair	**Flag sign** (alternate bands of hypopigmentation), sparse, easily pluckable	Normal-ish
Skin	**Flaky-paint dermatosis**, "crazy pavement"	Dry, loose folds
Fatty liver (hepatomegaly)	**Present**	Absent
Serum albumin	**Very low** (< 2.5 g/dL)	Normal/slightly low
Prognosis	Worse	Better

ORDER OF FAT LOSS IN MARASMUS

Buttocks/axilla → abdomen → chest/back → buccal pad of fat (last) ("Bichat's fat pad"). Loss of buccal fat = grade IV wasting.

WHO 10 steps of SAM management



WHO 10 steps – stabilisation (days 1–7) and rehabilitation (weeks 2–6)

Step	Key points
1. **Hypoglycaemia**	Blood glucose < **54 mg/dL (3 mmol/L)**: conscious – **50 mL 10% glucose/sucrose orally/NG**; unconscious – **10% dextrose 5 mL/kg IV** then 50 mL 10% by NG; feed **2-hourly day & night**
2. **Hypothermia**	Axillary < 35 °C (rectal < 35.5 °C): **kangaroo care**, cover with blankets, warm room, feed
3. **Dehydration**	Use **ReSoMal** (low Na 45, high K 40 mmol/L): **5 mL/kg every 30 min for 2 h**, then 5–10 mL/kg/h for 4–10 h orally/NG. **Avoid IV** except shock; if shock: **15 mL/kg over 1 h of half-strength Darrow's with 5% dextrose or Ringer lactate with 5% dextrose**

Step	Key points
4. **Electrolytes**	Extra **potassium (3–4 mmol/kg/day)** & **magnesium (0.4–0.6 mmol/kg/day)** for ≥ 2 weeks; **do NOT treat oedema with diuretics**
5. **Infection**	Assume infection in all: no complications – **oral amoxicillin**; complicated – **IV ampicillin + gentamicin** (ceftriaxone if no improvement in 48 h); measles vaccine if > 6 m and unvaccinated
6. **Micronutrients**	**Withhold iron in stabilisation phase**; start iron in rehabilitation (week 2); **vitamin A** on day 1 (< 6 m: 50,000 IU; 6–12 m: 1 lakh; > 12 m: 2 lakh; repeat day 2 & 14 if eye signs); folic acid 5 mg day 1 then 1 mg/day; zinc 2 mg/kg/day; copper
7. **Initial (cautious) feeding**	**F-75** (75 kcal & 0.9 g protein/100 mL): 100 kcal/kg/day, 1–1.5 g protein/kg/day, fluid 130 mL/kg/day (100 if severe oedema); small frequent feeds
8. **Catch-up growth**	**F-100** (100 kcal & 2.9 g protein/100 mL) or RUTF: **150–220 kcal/kg/day**, protein 4–6 g/kg/day; aim weight gain > **10 g/kg/day**
9. Sensory stimulation & emotional support	Tender loving care, play therapy, mother's involvement
10. **Prepare for follow-up**	Discharge when **WFH ≥ −2 SD / MUAC ≥ 12.5 cm and no oedema for 2 weeks**, eating well

DANGER: REFEEDING SYNDROME

Rapid refeeding → **hypophosphatemia** (hallmark), hypokalemia, hypomagnesemia, fluid overload & cardiac failure. Prevented by cautious **F-75** and electrolyte supplementation.

- **Ready-to-use therapeutic food (RUTF)**: peanut-paste based, energy ~520–550 kcal/100 g; used in **community-based management (CMAM)** of uncomplicated SAM with good appetite (appetite test passed).
- **Nutrition Rehabilitation Centres (NRC)** in India manage complicated SAM (anorexia, oedema, medical complications).
- Most common cause of death in SAM: **infection (sepsis, pneumonia, diarrhoea)**, hypoglycaemia, hypothermia, electrolyte imbalance, heart failure from overhydration.

3.5 Vitamin A

- Sources: liver, fish-liver oil, egg, milk; carotenoids (**β-carotene**) in yellow/green vegetables, papaya, mango, carrot.
- Functions: vision (**rhodopsin**), epithelial integrity, immunity, growth.

WHO classification of xerophthalmia

Code	Sign
XN	**Night blindness** – **earliest symptom**
X1A	Conjunctival xerosis
X1B	**Bitot's spots** (foamy triangular, temporal, keratinised)
X2	Corneal xerosis
X3A	Corneal ulceration/keratomalacia < 1/3 of cornea
X3B	Keratomalacia ≥ 1/3 cornea
XS	Corneal scar
XF	Xerophthalmic fundus

- **Earliest sign**: conjunctival xerosis (X1A); **earliest symptom**: night blindness. **Most specific**: Bitot's spots. **X3** = medical emergency.
- Other features: **phrynoderma** (follicular hyperkeratosis – "toad skin"), recurrent infections, growth failure. Severe vitamin A deficiency increases mortality in **measles** – give vitamin A in all measles cases.

VITAMIN A TREATMENT & PROPHYLAXIS (INDIA)

Treatment (xerophthalmia/measles/SAM): Day **1, 2 and 14** – < 6 m: **50,000 IU**; 6–12 m: **1 lakh IU**; > 1 y: **2 lakh IU** (oral). **Prophylaxis (National programme)**: **1 lakh IU at 9 months (with MR-1)**, then **2 lakh IU every 6 months from 16–18 months up to 5 years** → **9 doses total (17 lakh IU)**.

- **Hypervitaminosis A**: acute → **raised ICT (bulging fontanelle, pseudotumor cerebri)**, vomiting; chronic → bone pain, **cortical hyperostosis**, hepatomegaly, alopecia, dry skin; teratogenic (isotretinoin) – CNS, cardiac, craniofacial defects. Hypercarotenemia → yellow palms, **sclera spared**.

3.6 Vitamin D & Rickets

- Sources: **sunlight (UV-B, 290–315 nm)** – skin 7-dehydrocholesterol → cholecalciferol (D3); diet (fish oil, egg yolk; ergocalciferol D2 from plants).
- Liver **25-hydroxylation** (CYP2R1) → 25(OH)D – **best indicator of vitamin D status** (deficient < 12 ng/mL; insufficient 12–20 ng/mL per IAP/Global consensus).
- Kidney **1α-hydroxylation** (stimulated by PTH, low phosphate) → **1,25(OH)$_2$D – active form (calcitriol)**.
- Daily requirement: **400 IU/day in infants**, 600 IU/day in children > 1 y.

Rickets = failure of mineralisation of growing bone (**growth plate**) before epiphyseal fusion. **Osteomalacia** = failure of mineralisation of osteoid after fusion (also present in children with rickets).

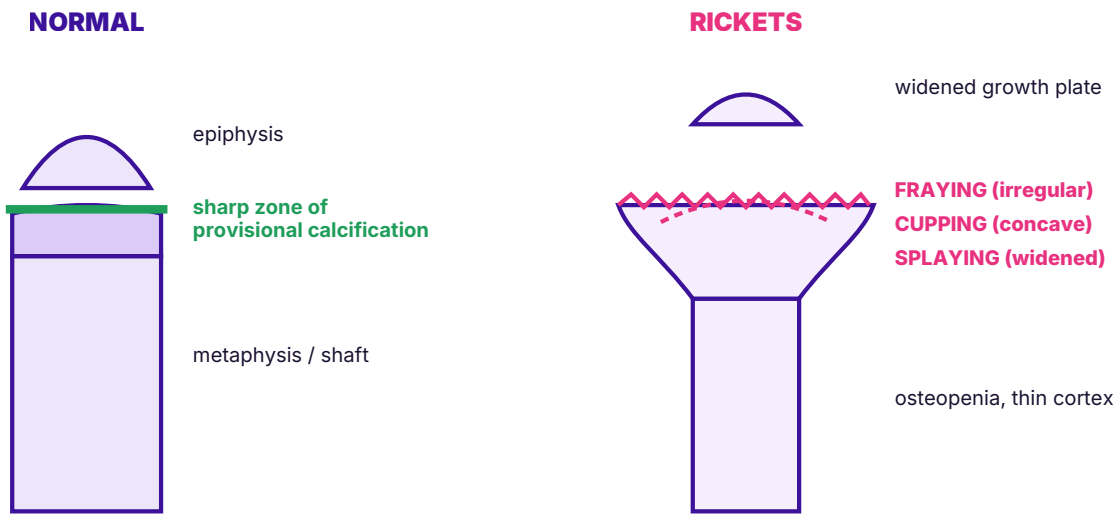
Clinical features (with earliest sites)

Region	Feature
Skull	**Craniotabes** – earliest sign (ping-pong ball feel of occipital/parietal bones; **< 6 m physiological in preterm**), frontal & parietal **bossing (hot cross bun skull)**, delayed AF closure, caput quadratum
Teeth	Delayed eruption, enamel hypoplasia, caries
Chest	**Rachitic rosary** (enlarged costochondral junctions – **painless, rounded, beaded**), **Harrison's sulcus** (groove along diaphragm insertion), pigeon chest, pneumonia
Limbs	**Widening of wrist (first evident at wrists)**, double malleoli (Marfan's sign), **genu varum (bow legs, < 2 y)** / genu valgum (knock knees, > 2–3 y), windswept deformity
Spine & pelvis	Kyphoscoliosis, triradiate pelvis (contracted)
General	Hypotonia, **pot belly**, delayed motor milestones, **hypocalcaemic tetany/seizures** (esp. < 6 m), short stature

RACHITIC VS SCORBUTIC ROSARY

Rachitic rosary (vitamin D) – **painless, rounded, nodular**, at costochondral junction. **Scorbutic rosary** (vitamin C) – **painful, sharp/angular ("step-like")** due to sternal depression with subperiosteal haemorrhage.

Radiology (X-ray wrist/knee – IOC for diagnosis & follow-up)



Rickets at the distal radius – cupping, fraying and splaying of the metaphysis

- **Cupping, fraying, splaying** of metaphysis; widened growth plate (↑ distance between metaphysis & epiphysis); osteopenia, coarse trabeculae; pseudofractures (Looser zones).
- **Earliest radiological sign**: loss of the normal **zone of provisional calcification** (indistinct metaphyseal margin).
- **First sign of healing**: **white line of provisional calcification** at metaphysis within **2–4 weeks** of treatment (Thacher scoring).

Biochemistry

Type	Ca	PO_4	ALP	PTH	25(OH)D	$1,25(OH)_2D$
Nutritional (vit D deficiency)	N/↓	↓	**↑↑**	**↑**	**↓↓**	N/↑/↓
Calcium deficiency rickets	↓	↓	↑	↑	N	↑
VDDR type 1A (1α-hydroxylase def., CYP27B1)	↓	↓	↑	↑	N	**↓↓**
VDDR type 2A (receptor defect, VDR) – **alopecia**	↓	↓	↑	↑	N	**↑↑↑**
X-linked hypophosphatemic rickets (PHEX → ↑FGF-23)	**N**	**↓↓**	↑	N/mild ↑	N	N/↓ (inappropriately)
Renal (CKD) osteodystrophy	↓	**↑**	↑	↑↑	N	↓
Hypophosphatasia	N/↑	N	**↓↓ (low ALP)**	N	N	N

- **Raised ALP** is the most sensitive marker of rickets activity (and of response to treatment); **serum 25(OH)D** is the best test of vitamin D status; **low phosphate** is consistently present (secondary hyperparathyroidism → phosphaturia).
- **Hypophosphatemic rickets** (most common heritable form): **X-linked dominant**, short stature with bowing, **no tetany/myopathy**, dental abscesses; Rx **oral phosphate + calcitriol**; **burosumab** (anti-FGF23 antibody).
- **Fanconi syndrome**, distal RTA – rickets due to phosphate wasting / acidosis.

TREATMENT OF NUTRITIONAL RICKETS (IAP)

- **Daily**: vitamin D3 **2000 IU/day (< 1 y) or 3000–6000 IU/day (> 1 y)** for 12 weeks, **or**
- **Stoss therapy** (Ghai/Indian practice): single large oral dose of **3–6 lakh IU** (commonly **6 lakh IU** in children > 1 year, e.g. 60,000 IU × 10 doses). Global Consensus 2016 prefers smaller stoss doses: **50,000 IU (3–12 m), 1.5 lakh IU (1–12 y), 3 lakh IU (> 12 y)**.
- **Plus calcium 500 mg/day (elemental)**; maintenance 400–600 IU/day thereafter.
- Hypocalcaemic seizures/tetany: **IV 10% calcium gluconate 1–2 mL/kg slowly with cardiac monitoring**.
- Review X-ray at **4 weeks** (healing line); ALP normalises by ~3 months.

- **Hypervitaminosis D** → **hypercalcaemia**: polyuria, vomiting, constipation, nephrocalcinosis, **metastatic calcification**; Rx stop vit D, hydration, steroids.
- Infants at risk of rickets: exclusively breastfed without supplementation, dark skin, purdah, preterm (**metabolic bone disease of prematurity** – mainly phosphate deficiency), anticonvulsants (phenytoin, phenobarbitone), liver/kidney disease, malabsorption.

3.7 Vitamin E & Vitamin K

- **Vitamin E** (tocopherol, antioxidant): deficiency in **preterm infants** → **haemolytic anaemia, thrombocytosis, oedema**; in fat malabsorption/cholestasis → **spinocerebellar degeneration** (ataxia, areflexia, loss of proprioception), **retinopathy**, peripheral neuropathy. Ataxia with isolated vitamin E deficiency (AVED – α-TTP gene).
- **Vitamin K** – see Hemorrhagic disease of the newborn: give **1 mg IM** (0.5 mg if < 1000 g) at birth to **all newborns**.

HDN (VKDB)	Timing	Cause/site
Early	First **24 h**	Maternal drugs: **phenytoin, phenobarbitone, warfarin, rifampicin, INH** – cephalhaematoma, ICH
Classic	**Day 2–7**	Exclusive BF without prophylaxis – **GI bleed, umbilical stump, circumcision**
Late	**2–12 weeks** (up to 6 m)	Exclusive BF + malabsorption/cholestasis (biliary atresia, CF) – **intracranial haemorrhage** (50%)

Lab: ↑ **PT (first)**, ↑ aPTT, normal platelets & fibrinogen. Rx: IV vitamin K 1–2 mg; FFP if severe.

3.8 Vitamin B-Complex

Vitamin	Deficiency	Key features
B1 – Thiamine	**Beriberi**	**Infantile beriberi (2–6 m, breastfed by deficient mother)**: acute **cardiac (wet)** – tachycardia, cardiomegaly, CCF; **aphonic (laryngeal nerve palsy – soundless cry)**; pseudomeningitic. Wet = cardiac (high-output failure); dry = peripheral neuropathy; **Wernicke** (ophthalmoplegia, ataxia, confusion). Test: **RBC transketolase**. Polished rice, thiaminase in raw fish.
B2 – Riboflavin	Ariboflavinosis	**Angular stomatitis, cheilosis, glossitis (magenta tongue)**, seborrhoeic dermatitis, corneal vascularisation; test: RBC glutathione reductase; phototherapy ↓ riboflavin
B3 – Niacin	**Pellagra**	**4 Ds – dermatitis (sun-exposed, Casal's necklace), diarrhoea, dementia, death**; maize/jowar (leucine excess) diets, **Hartnup disease**, carcinoid, INH
B5 – Pantothenic acid		Burning feet syndrome
B6 – Pyridoxine		**Pyridoxine-dependent seizures** (neonatal, refractory, ALDH7A1 – respond to IV B6), INH neuropathy, microcytic sideroblastic anaemia, glossitis

Vitamin	Deficiency	Key features
B7 – Biotin		Dermatitis, alopecia, hypotonia, organic aciduria (raw egg white – avidin; biotinidase deficiency)
B9 – Folate		**Megaloblastic anaemia**, glossitis, **neural tube defects** (periconceptional 400 µg; 4 mg if previous NTD); goat's milk (deficient in folate & B12)
B12 – Cobalamin		Megaloblastic anaemia + **neurological (subacute combined degeneration)**, **knuckle hyperpigmentation**, developmental regression & tremors in infants of vegetarian mothers;

INFANTILE TREMOR SYNDROME (ITS)

Seen in **Indian infants 5–18 months**, exclusively breastfed by vegetarian mothers: **tremors, developmental regression, hypopigmented brown hair, skin hyperpigmentation (knuckles), pallor (megaloblastic anaemia)** – attributed to **vitamin B12 deficiency**. Rx: B12, propranolol for tremors.

3.9 Vitamin C & Scurvy

- Functions: hydroxylation of proline & lysine in **collagen**, iron absorption, antioxidant.
- Age: **6–24 months** (infants on boiled/pasteurised milk, no fruits); also autism with restricted diets.
- Features: **irritability, pseudoparalysis ("frog-leg position", painful limbs – cries on handling/changing diapers)**, subperiosteal haemorrhage, **bleeding gums (only if teeth have erupted)**, perifollicular haemorrhages, **corkscrew hair**, **scorbutic rosary**, poor wound healing, anaemia.
- **X-ray signs**: **ground-glass osteopenia**, **Wimberger ring sign** (pencil-thin cortex around epiphysis), **white line of Frankel** (dense zone of provisional calcification), **Trümmerfeld zone** (zone of rarefaction below white line), **Pelkan spur** (lateral metaphyseal spur), subperiosteal haemorrhage calcifies on healing.
- Diagnosis: **leukocyte (buffy coat) ascorbic acid** – best; plasma ascorbate.
- Rx: **vitamin C 100–200 mg/day** orally – rapid response within 1 week.

WIMBERGER – TWO DIFFERENT SIGNS

Wimberger ring (epiphyseal ring) → **scurvy**. **Wimberger sign** (bilateral destruction of medial proximal tibial metaphysis) → **congenital syphilis**.

3.10 Iron, Zinc, Iodine & Trace Elements

Iron

- Term infant iron stores last **4–6 months**; preterm 2–3 months → supplement preterm from **2 weeks–6 weeks** at 2 mg/kg/day.
- **Anaemia Mukt Bharat (IFA prophylaxis)**: 6–59 months – **IFA syrup 1 mL (20 mg iron + 100 µg folic acid) biweekly**; 5–9 y – 45 mg iron + 400 µg FA weekly (pink tablet); 10–19 y – 60 mg + 500 µg weekly (blue tablet); deworming **albendazole biannually**.
- Details of IDA → Hematology chapter.

Zinc

- Deficiency: growth failure, **diarrhoea**, impaired immunity, delayed wound healing, **acrodermatitis enteropathica** (AR, SLC39A4 defect in zinc absorption): triad of **periorificial & acral dermatitis, diarrhoea, alopecia**; appears after weaning from breast milk; Rx lifelong oral zinc.
- **Zinc in diarrhoea**: **10 mg/day (< 6 months), 20 mg/day (> 6 months) × 14 days** – reduces duration, severity & recurrence (next 2–3 months).

Other trace elements

Element	Deficiency
Copper	**Menkes kinky hair disease (ATP7A, X-linked recessive)** – steely brittle hair (pili torti), neurodegeneration, hypothermia, **low serum copper & ceruloplasmin**; acquired – neutropenia, microcytic anaemia, osteoporosis
Selenium	**Keshan disease** (cardiomyopathy), Kashin–Beck (osteoarthropathy)
Iodine	Goitre, **cretinism** (neurological – deaf-mutism, spastic diplegia, squint; myxoedematous – hypothyroid), **MC preventable cause of ID**; iodized salt **30 ppm at production, 15 ppm at consumer level**
Fluoride	Excess → **dental fluorosis (> 1.5 ppm)**, **skeletal fluorosis (> 3–10 ppm)**, genu valgum; optimal 0.5–0.8 ppm; Nalgonda technique for defluoridation
Chromium	Impaired glucose tolerance
Manganese / Molybdenum	Rare; molybdenum cofactor deficiency – seizures, lens dislocation (sulfite oxidase)

3.11 Childhood Obesity

- Definition (IAP 2015, 5–18 y): **BMI > 23-adult-equivalent = overweight; > 27-adult-equivalent = obese**. WHO < 5 y: weight-for-height > +2 SD overweight, > +3 SD obese.
- **Exogenous (simple/nutritional) obesity** (> 95%): **tall for age**, normal/advanced bone age, normal intelligence, family history.
- **Endocrine/genetic obesity**: **short stature**, delayed bone age, dysmorphism, ID – **hypothyroidism, Cushing, GH deficiency, pseudohypoparathyroidism**, **Prader–Willi**, **Laurence–Moon–Bardet–Biedl** (obesity, retinitis pigmentosa, polydactyly, hypogonadism, ID, renal anomalies), Alström, Cohen, **MC4R mutation (MC monogenic obesity)**, leptin deficiency.
- Complications: **acanthosis nigricans** (marker of insulin resistance), type 2 DM, NAFLD, **SCFE**, Blount disease, OSA, pseudotumor cerebri, PCOS, hypertension, dyslipidaemia.
- Management: lifestyle – diet, **≥ 60 min/day of moderate-to-vigorous physical activity**, **screen time < 2 h/day**, sleep; metformin in insulin resistance; bariatric surgery for severe adolescent obesity.

Practice MCQs · Nutrition and Malnutrition

205 questions · correct option highlighted in green · cover the answer strip to self-test

Breastfeeding & Breast Milk

1. Which of the following is a contraindication for breast feeding?

A. Hep A
B. CMV
D. Active untreated TB

Ans D

2. Which of the following is true regarding storage of expressed breast milk?

A. EBM can be stored at room temperature for about 6 hours and 24 hours in refrigerator
B. EBM can be stored at room temperature for about 24 hours and 6 days in refrigerator
C. EBM can be stored at room temperature for about 18 hours and 1 month in refrigerator
D. EBM can be stored at room temperature for about 6 hours and 6 days in refrigerator

Ans A

3. Which one of the following is a bioactive factor of human milk when compared to formula milk?

A. Lipoprotein A
B. Transforming growth factor
C. Immunoglobulin-M
D. Polysaccharides

Ans B • TGFβ is the most abundant cytokine in human milk; It has 3 isoforms, of which TGFβ2 predominates
• Milk-borne TGF-β regulates inflammation, wound repair & helps prevent allergic diseases.

4. Breastfeeding should begin within ... hours after cesarean delivery:

A. 2
B. 4
C. 8
D. 24

Ans B 4 Breastfeeding should be initiated within 30 mins. of a normal vaginal delivery & 4 hours of cesarean section.

5. Breast milk storage in a refrigerator is up to:

A. 4 hours
B. 8 hours
C. 12 hours
D. 24 hours

Ans D Site of storage/temperature Room temperature Refrigerator Freezer at –20° C Time for which EBM can be stored 8–10 hoursQ 24 hoursQ 3 months

6. Colostrum is rich in:

A. IgA
B. IgE
C. IgG
D. IgM

Ans A Colostrum contains more antibodies (IgA), WBCs and protein. Also called 'first immunization' of the baby.

7. Which of the following are true about breast milk?

A. It is a rich source of Iron
B. It contains more carbohydrates than cow's milk
C. It contains more proteins than cow's milk
D. It contains less DHA than cow's milk

Ans B

8. Nipple confusion means?

A. Baby fed with a bottle finding it difficult and confusing to suckle at breast
B. Baby not able to suckle with bottle
C. Baby not able to feed with spoon
D. Baby not able to feed with paladin

Ans A

9. The average whey/casein ratio in breast milk is:

A. 60:40
B. 80:20
C. 20:80
D. 40:60

Ans A Human milk contains two types of proteins 60% is: whey and 40% is casein; helps in quick & easy digestion.

10. Amount of protein present in 100 mL of breast milk is:

A. 2.2 g
B. 1.1 g
C. 0.55 g
D. 3.3 g

Ans B

11. Compared to breast milk, colostrum is richer in:

A. Protein
B. Fat
C. Lactose
D. Water

Ans A

12. Breast milk at room temperature stored for:

A. 4 hours
B. 8 hours
C. 12 hours
D. 24 hours

Ans B

13. **Breastfeeding for a new born to be started:**
A. As soon as possible
B. After 6 hours
C. After 1 day
D. After meconium has passed

Ans A

14. **Breast milk is rich in ___ when compared to cow's milk:**
A. Lactose
B. Protein
C. Fat
D. Vitamin K

Ans A Lactose Answers

15. **All are true about breastfeeding except:**
A. It is available at proper temperature
B. Breast milk contains antibodies
C. Exclusive breastfeeding up to 6 months of age
D. To be started after 4 hours of normal delivery

Ans D

16. **Exclusive breastfeeding is done till:**
A. 4 months
B. 6 months
C. 8 months
D. 10 months

Ans B

17. **Fatty acid present in breast milk which is important for growth & CNS development is:**
A. Docosahexaenoic acid
B. Palmitic acid
C. Linoleic acid
D. Linolenic acid

Ans A Human milk contains 30 times more DHA (10 mg/dl) than cow's milk, which is a predominant fatty acid in brain and retina. with

18. **Breast milk is maximum at:**
A. 1 month
B. 3 months
C. 6 months
D. 12 months

Ans C 5–6 months Breast milk output is maximum at 5–6 months of lactation ~ 730 mL/day.

19. **Paraaminobenzoic acid of breast milk prevent the infection of:**
A. Plasmodium vivax
B. Klebsiella pneumoniae
C. Giardia
D. E. coli

Ans A

20. **Which of the following is not the correct sign of good attachment of a baby to the breast:**
A. Baby's mouth wide open
B. Lower areola more visible
C. Baby's lower lip everted
D. Baby's chin touching the breast

Ans B

21. **True about breastfeeding:**
A. Best for both preterm and term
B. 50% energy from protein
C. Promote lactobacillus growth in bowel
D. Predispose to necrotizing enterocolitis
E. Decrease allergic disorder

Ans A c. Promote lactobacillus growth in bowel; e. Decrease allergic disorder 20/e p 286-290, Ghai 8/e p 150-161 Discussing about the options one by one: a Composition of breast milk varies at different stages of lactation to suit the needs of the baby b Energy from different constituents of breast milk is Carbohydrate: 30–40%, from Protein 7–10%; from fat 45–60% c Bifidus factor supports the growth of lactobacillus in gut thereby preventing growth of harmful bacteria

22. **In breastfed infant less chances of enteric infection is due to:**
A. Alkaline pH of stool
B. Breast milk nutrients, beneficial effect on immune system
C. IgA in breast milk
D. Bacteroides and Clostridium in gut
E. Sterile nature of breast milk

Ans B c. IgA in breast milk; e. Sterile nature of breast milk Note: pH of stool in breastfed babies is usually acidic & not alkaline.

23. **Benefits of breast milk are:**
A. Better nutrition
B. Less infection
C. More allergy
D. Less allergy
E. High sodium contents

Ans A b. Less infection; d. Less allergy

24. **Breast milk is known to transmit:**
A. Tuberculosis
B. CMV
C. Varicella
D. Rubella

Ans B Breast milk transmits (with definitive evidence) → HIV & CMV.

25. **Breast milk is rich in _____ when compared to cow's milk:**
A. Lactose
B. Protein
C. Fat
D. Vitamin K

Ans A

26. The protective effects of breast milk are known to be Questions associated with:

A. IgM antibodies
B. Lysozyme
C. Mast cells
D. **IgA antibodies**

Ans D Although antibodies & lysozyme, both are important for protective effects of breast milk, antibodies (IgA) play a major role.

27. The current recommendation for breastfeeding is that:

A. **Exclusive breastfeeding should be continued till 6 months of age followed by supplementation with additional foods**
B. Exclusive breastfeeding should be continued till 4 months of age followed by supplementation with additional foods
C. Colostrum is the most suitable food for a newborn baby but it is best avoided in first 2 days
D. The baby should be allowed to breastfeed till one year of age MALNUTRITION

Ans A Exclusive breastfeeding should be continued till 6 months of age followed by supplementation with additional foods MALNUTRITION

28. Breast feeding has been suggested to have a possible protective effect against all the following EXCEPT

A. otitis media
B. urinary tract infections
C. septicemia
D. childhood cancer
E. **skin infections**

Ans E Diarrhea, otitis media, urinary tract infections, necrotizing enterocolitis, septicemia, infant botulism, celiac disease, childhood cancer, allergy, hospitalizations, and infant mortality all are conditions in which breastfeeding has been suggested to have a possible protective effect.

29. An absolute contraindication to breastfeeding is

A. hepatitis c infection of the mother
B. alcohol intake
C. herpes simplex lesion of the lips
D. **active pulmonary tuberculosis**
E. maternal high grade fever

Ans D is a real contraindication to breastfeeding. Herpes simplex infection of the breast is another contraindication.

30. Regarding breast engorgement; All the following are true EXCEPT

A. **usually happens in the first stage of lactogenesis**
B. poor breast feeding technique can cause engorgement
C. breastfeeding immediately at signs of infant hunger will eventually prevent this
D. to reduce engorgement, breasts should be softened prior to infant feeding with a combination of hot compresses and expression of milk
E. between feedings, cold compresses applied, and oral nonsteroidal anti- inflammatory medications administered

Ans A Breast engorgement happens in the second stage of lactogenesis.

Formula Feeding & Weaning

31. Milk is deficient in?

A. **Iron and Vit C**
B. Iron and Vit A
C. Phosphorus and Vit C
D. Phosphorus and Vit A

Ans A About the options: Iron Iron content (1 mg/L) in cow & breast milk makes it difficult to maintain body iron & meet RDA of infant (8–10 mg) Breastfed infants absorb iron 2–3 times more efficiently than infants fed cow's milk Vit C Infants consuming pasteurized or boiled animal milk are at significant risk of developing Vitamin C deficiency Phosphorus Cow's milk is rich in phosphate; So, an infant on a diet rich in cow's milk has chances of developing hypocalcemia

32. A 15monthold child feeding on cow milk with water with severe wasting and bipedal edema presents with poor appetite. Diagnosis?

A. **Kwashiorkor**
B. Marasmus
C. Both (a) and (b)
D. None of the above

Ans A

33. Best indicator of growth monitoring in children:

A. Weight
B. Mid-arm circumference
C. **Rate of increase in height and weight**
D. Head circumference

Ans C Serial measurements of Height & weight (using growth charts) are more important than one time measurement.

34. A 6yearold girl on a predominantly maize based diet presents with history of recurrent diarrhea, dermatitis with rashes involving the neck area is seen in:

A. **Pellagra**
B. Tyrosinemia
C. Phenylketonuria
D. Albinism

Ans A Maize based diet predisposes to Niacin deficiency, which gives rise to Pellagra.

35. All the following organisms can cause mastitis EXCEPT

A. Staphylococcus aureus
B. Escherichia coli
C. Haemophilus influenza
D. Klebsiella pneumoniae
E. **group B streptococcus**

Ans E Group A streptococcus, not group B.

36. The MOST common cause of using infant formula is

A. **parental preference**
B. inborn error of metabolism
C. maternal tuberculosis
D. masititis
E. inadequate weight gain

Ans A

37. The hallmark of refeeding syndrome is the development of severe

A. hypophosphatemia
B. hypokalemia
C. hypomagnesemia
D. hypernatremia
E. hyperglycemia

Ans A The hallmark of refeeding syndrome is the development of severe hypophosphatemia after the cellular uptake of phosphate duringthe 1st wk of starting to refeed. Serum phosphate levels of ≤0.5 mmol/L can produce weakness, rhabdomyolysis, neutrophil dysfunction, cardio-respiratory failure, arrhythmias, seizures, altered level of consciousness, or sudden death.

38. The following conditions are risk factors for carotenemia EXCEPT

A. liver disease
B. food faddist
C. diabetes mellitus
D. hypothyroidism
E. malabsorption

Ans E is not associated with any risk of carotenemia.

Nutritional Requirements & RDA

39. Compared with cow's milk, mother's milk has more?

A. Lactose
B. Vitamin D
C. Proteins
D. Fat

Ans A

40. Calorie requirement per day of a child weighing 15 kg would be:

A. 1150 kcal
B. 1250 kcal
C. 1450 kcal
D. 1550 kcal

Ans B For children with normal body weights, energy requirement can be calculated as: For 1st 10 kg: 100 kcal/kg ⇒ For next 10 kg: 50 kcal/kg ⇒ Beyond 20 kg: 20 kcal/kg. So, for 15 kg, caloric requirement is 10 × 100 + 5 × 50 = 1000 + 250 = 1250 kcal/day.

41. With reference to a malnourished child, the following statements are correct except:

A. Skin and mucosa are not effective barriers to infection
B. There is impairment of chemotaxis and defective candidacidal, bactericidal capacities of polymorphs
C. There is impairment of cell mediated immunity and delayed hypersensitivity
D. There is impairment of humoral response to immunizing agents and reduced number of B-cells

Ans D There is impairment of humoral response to immunizing agents and reduced number of Bcells In PEM, Humoral immunity is relatively spared; Number & proportion of B lymphocytes are not altered.
• Levels of all Immunoglobulins are normal or increased except IgAQ.

42. One of the following is false regarding complications of proteinenergy malnutrition:

A. Hyperglycemia
B. Hypothermia
C. Septic shock
D. Electrolyte imbalance

Ans A

43. Initial protein requirement of a Kwashiorkor child (g/kg/day):

A. 0.5–1.0
B. 1.0–1.5
C. 1.5–2.0
D. 2–3

Ans B Recommended daily energy and protein intake from initial feeds in a child with SAM is 100 kcal/kg & 11.5 g/kg.

44. Daily protein requirement in a 2 years old child is?

A. 13 gm
B. 25 gm
C. 40 gm
D. 60 gm

Ans A The recommended daily protein intake for a 2-year-old child is approximately 13 g/day, per standard pediatric nutrition guidelines (WHO/ICMR safe levels of intake for toddlers).

45. For which vitamin, RDA is determined according to protein intake:

A. Thiamine
B. Niacin
C. Pyridoxine
D. Vitamin B12

Ans C Because of the importance of Vit B6 (Pyridoxine) in amino acid metabolism high protein intake can increase the requirement for pyridoxine.

46. Regarding nutritional requirement during childhood, all the following are true

A. nutrition and growth during 3-6 years of life predict adult stature and some health outcomes
B. major risk period for growth stunting is between 4 and 24 months of age
C. estimated average requirement (EAR) is the average daily nutrient intake level estimated to meet the requirements for 50% of the population
D. recommended dietary allowance (RDA) is an estimate of the daily average nutrient intake to meet the nutritional needs of >97% of the individuals in a population
E. the 3 components of energy expenditure are the basal metabolic rate, thermal effect of food, and energy for physical activity

Ans A The first 3 years.

47. All the following statements concerning iron requirement in children are true EXCEPT

A. breast milk provides optimal intake of iron for the first 4 months
B. iron present in animal protein is more bioavailable than that found in vegetables
C. iron deficiency is the most common micronutrient deficiency
D. cow's milk is a good source of bioavailable iron
E. iron supplements can interfere with zinc absorption

Ans D Cow's milk is a poor source of bioavailable iron.

Protein-Energy Malnutrition

48. The following component of human milk is protective against Giardia lamblia infection?

A. Lysozyme
B. Lactoferrin
C. Bifidus factor
D. Glycoconjugate

Ans B Discussing about the options one by one: Component of breast milk Protects against a. Lysozyme E. histolytica b. Lactoferrin G. lamblia, Plasmodium falciparum, T. gondii, E. histolytica & Pneumocystis carinii c. Bifidus factor Shigella, Salmonella, E. coli d. Glycoconjugate V. cholerae, E. coli

49. Definition of severe acute malnutrition includes which of the following?

A. Wt for Ht < – 2 SD
B. Wt for Ht < – 3 SD
C. Wt for age < – 2 SD
D. Wt for age < – 3 SD

Ans B Wt for Ht < 3 SD Wt for Ht < – 3 SD on WHO growth standard is one of the diagnostic criteria for severe acute malnutrition.

50. Proximal muscle weakness & atrophy in a child with PEM is due to which electrolyte disturbance?

A. Hypokalemia
B. Hypomagnesemia
C. Hypophosphatemia
D. Hypocalcemia

Ans C • Chronic hypophosphatemia causes proximal muscle weakness & atrophy
• Hypokalemia in skeletal muscle may cause muscle weakness & cramps
• Paralysis is a possible complication, generally only at K < 2.5 mEq/L. d. Phosphate Chronic hypophosphatemia causes proximal muscle weakness and atrophy. However, hypokalemia in skeletal muscle may cause muscle weakness and cramps. Paralysis is a possible complication, generally only at potassium levels <2.5 mEq/L.

51. Proximal muscle weakness atrophy in a child with PEM due to which electrolyte disturbance:

A. Magnesium
B. Calcium
C. Zinc
D. Phosphate

Ans D Hypophosphatemia is a well-recognized cause of proximal muscle weakness and atrophy in malnourished children, particularly during refeeding (refeeding syndrome).

52. While assessing a child for severe acute malnutrition, all are criteria except:

A. Mid arm circumference
B. Edema
C. Weight for height
D. Height for age

Ans D In children 6–59 months of age, severe acute malnutrition (SAM) is defined as: Weight/height or Weight/length < – 3 Z score, using WHO Growth Charts or presence of bipedal edema of nutritional origin or Mid-upper arm circumference (MUAC) < 115 mm.

53. Best marker for chronic PEM is:

A. Weight for age
B. Height for age
C. Weight for height
D. Head circumference

Ans B

54. According to WHO, severe acute malnutrition in children is defined as:

A. Weight for height Z score < –3
B. Weight for height Z score < –2
C. Weight for height Z score < –1
D. Weight for height Z score between –2 and –3

Ans A Weight for height Z score < –

55. In a child with severe PEM which of the following should not be included in the initial treatment:

A. Vitamin A
B. Iron
C. Zinc
D. Folic acid

Ans B Iron should be started in rehabilitation phase Answers

56. In PEM hypoglycemia is corrected by:

A. 5% dextrose
B. 10% dextrose
C. 25% dextrose
D. 50% dextrose

Ans B Hypoglycemia is defined as blood glucose < 54 mg/dL in a severely malnourished child. It is treated by immediately giving a 50 mL bolus of 10% glucose orally or through NG tube.

57. Mostly death in PEM is due to all except:

A. Hypothermia
B. CCF
C. Worm infestation
D. Electrolyte imbalance

Ans C M neMonic Mnemonic for indicators of poor prognosis in malnutrition "SHIELDED SCALD" S: Sugar deficiency or Hypoglycemia (blood glucose < 54 mg/dL) S Severe anemia H: Hypothermia (Rectal temperature < 35.5°C) CA: Cardiac failure I: Infections (Systemic infections) L: Liver failure EL: Electrolyte imbalance D: Diarrhea DE: Dehydration or Overhydration D: Deficiency of micronutrients like potassium, Iron

58. **Acute complication of PEM:**
A. Hypothermia
B. Hypoglycemia
C. Hypokalemia
D. Hypermagnesemia
E. Eosinophilia

Ans A b. Hypoglycemia; c. Hypokalemia

59. **The following hereditary forms of rickets may help to diagnose an apparently healthy mother with the same disease**
A. X- linked hypophosphatemic rickets
B. autosomal dominant hypophosphatemic rickets
C. autosomal recessive hypophosphatemic rickets
D. Fanconi syndrome
E. hereditary hypophosphatemic rickets with hypercalciuria

Ans A Undiagnosed disease in the mother is not unusual in X-linked hypophosphatemia.

Kwashiorkor & Marasmus

60. **Which of the following is not a component of Kangaroo mother care (KMC)?**
A. Skin to skin contact
B. Supplementary nutrition
C. Exclusive breastfeeding
D. Early discharge and follow-up

Ans B Components of Kangaroo Mother Care (KMC) are: a. Kangaroo position: skintoskin + b. Exclusive breastfeeding ± forti- + c. Early discharge and follow-up contact fication

61. **Specific sign of Kwashiorkor is:**
A. Pitting edema
B. Weight loss
C. Flag sign
D. Muscle wasting

Ans A Pitting edema is the main sign of Kwashiorkor and it usually starts from feet.

62. **Which is the most specific clinical feature for diagnosis for Kwashiorkor?**
A. Fatty liver
B. Easy pluckable hair
C. Low serum albumin
D. Edema

Ans D Kwashiorkor is primarily a clinical diagnosis. There is no specific diagnostic test for the condition. Edema is the defining characteristic.

63. **Which of the following is false about Kwashiorkor?**
A. Flaky paint dermatosis
B. Atrophic changes in liver on biopsy
C. Can pull loose hair easily
D. Serum albumin < 3 gm per 100 mL

Ans B In kwashiorkor, hepatomegaly with fatty liver is usually seen.

64. **Kwashiorkor is diagnosed in growth retarded children along with:**
A. Edema and mental changes
B. Hypopigmentation and anemia
C. Edema and hypopigmentation
D. Hepatomegaly and anemia

Ans A

65. **Select the false statement regarding marasmus:**
A. There is muscle atrophy and resultant hypotonia
B. Loss of fat from sucking pads of cheeks is earliest sign
C. Besan panjiri is an energy dense food used in treatment
D. This is non-edematous severe childhood undernutrition

Ans B Loss of fat from sucking pads of the cheeks is the earliest sign Buccal pad of fat is usually the last to go in severe malnutrition.

66. **All are seen in marasmus except:**
A. Hepatomegaly
B. Muscle wasting
C. Voracious appetite
D. Weight loss

Ans A

67. **Fatty liver in Kwashiorkor is due to:**
A. Decrease in carbohydrates
B. Decrease in protein substrates
C. Abnormal protein synthesis
D. Reduced synthesis of lipoprotein carrier

Ans D In kwashiorkor there is protein & calorie deficiency, hence the liver is incapable of making apolipoproteins which are needed for transportation of the lipids; So lipids accumulate in liver causing fatty liver.

68. **Not seen in Kwashiorkor:**
A. Apathy
B. Flaky paint dermatosis
C. Baggy pant appearance
D. Increased transaminase

Ans C d. Increased transaminase In Kwashiorkor, there is generalized edema, so loose folds of skin (baggy pants appearance) are usually not seen; Though fatty liver & hepatomegaly are seen in Kwashiorkor, Liver function tests are usually normal.

69. **Which of the following is a feature of marasmus and not Kwarshiorkar?**
A. Voracious appetite
B. Fatty change in liver
C. Hypoalbuminemia
D. Edema

Ans A

70. **Kwashiorkor, not an essential criteria:**
A. Weight for height
B. Skin changes
C. Edema
D. Muscle wasting

Ans C Hepatomegaly Hepatomegaly may be seen in Kwashiorkor, but it is not an essential criteria.

71. **False about marasmus are all except:**
A. Hepatomegaly
B. Edema
C. **Good appetite**
D. Flaky paint appearance

Ans C

72. **All of the following conditions are observed in marasmus, except:**
A. **Hepatomegaly**
B. Muscle wasting
C. Low insulin levels
D. Extreme weakness

Ans A

73. **All of the following are characteristic features of Kwashiorkor except:**
A. **High blood osmolarity**
B. Hypoalbuminemia
C. Edema
D. Fatty liver

Ans A

74. **Kwashiorkor in Ghanaian language means:**
A. Condition seen in 2nd child
B. **Condition seen in the displaced child**
C. Condition seen in cousin
D. Condition seen in the stepchild
E. Condition seen in the 3rd child MICRONUTRIENTS & THEIR DEFICIENCY

Ans B The name Kwashiorkor, comes from the Ghanian language, meaning disease of the displaced baby when the next one is born. MICRONUTRIENTS &

Vitamin A Deficiency

75. **After premature delivery, mother's milk is low in:**
A. **Lactose**
B. Fat
C. Protein
D. Sodium

Ans A Lactose For Mnemonic,

76. **Dose of treatment of vitamin A deficiency in 10 kg child who is 2 years old is:**
A. 5,000 IU
B. 100,000 IU
C. 150,000 IU
D. **200,000 IU**

Ans D In children > 1 yr age with weight > 8 kg, therapeutic dose of vit A is 200,000 IU

77. **When can a severely malnourished child be safely discharged from the hospital?**
A. The child attains his height for age
B. The child reaches his ideal weight for height
C. **The child loses edema and starts gaining weight**
D. The child attains weight for his age

Ans C

78. **Dose of vitamin A for a 18monthold baby, with keratomalacia, weighing 10 kg is:**
A. 50,000 IU
B. 1,00,000 IU
C. **2,00,000 IU**
D. 5,00,000 IU

Ans C

79. **First clinical sign of vitamin A deficiency:**
A. Poor growth
B. **Conjunctival xerosis**
C. Hydrocephalus
D. Phrynoderma

Ans B First clinical sign of vitamin A deficiency is conjunctival xerosis, while 1st symptom is night blindness

80. **Stage X3B refers to the following in the classification of vitamin A deficiency:**
A. Night blindness
B. Corneal xerosis
C. Conjunctival xerosis
D. **Keratomalacia**

Ans D

81. **The recommended oral dose of vitamin A to be given in 124. a 10 months child with deficiency on each of day 1, 2 and 28 is:**
A. 50,000 IU
B. **1,00,000 IU**
C. 2,00,000 IU
D. 6,00,000 IU

Ans B Dose of oral vitamin A for the treatment of Vitamin A deficiency consists of: Age < 6 months 6–12 months > 1 year Dose of vitamin A 50,000 IU 1,00,000 IU 2,00,000 IU

82. **Encephalopathy in refeeding syndrome is mainly a result of**
A. hypophosphatemia
B. hypokalemia
C. hypomagnesemia
D. **thiamine deficiency**
E. hyperglycemia

Ans D causes encephalopathy, lactic acidosis, and death.

83. **The MOST characteristic lesion of vitamin A deficiency is**
A. **xerophthalmia**
B. corneal ulcers
C. Bitot spots
D. keratomalacia
E. keratoconus

Ans A In early vitamin A deficiency, the cornea keratinizes, becomes opaque, is susceptible to infection, and forms dry, scaly layers of cells (xerophthalmia).

84. **All the following are helpful for diagnosis of marginal vitamin A deficiency EXCEPT**

A. dark adaptation test
B. conjunctival impression cytology
C. relative dose response
D. **plasma retinol level**
E. diet history

Ans D The plasma retinol level is not an accurate indicator of vitamin A status unless the deficiency is severe and liver stores are depleted, in which case low plasma retinol is likely to be evident.

Vitamin D & Rickets

85. **True about cow's milk are all except:**

A. **Cow's milk contains 80% whey protein not casein**
B. Cow milk has less carbohydrate than mother's milk
C. Has more K+ and Na+ than infant formula feeds
D. Has more protein than breast milk

Ans A

86. **Monkey facies and Baggy pants refers to:**

A. Peculiar facies of monkeypox infection
B. Peculiar dress of African tribe
C. **Look in marasmus because of loss of fat**
D. Look in monkey bite

Ans C

87. **The following statement about Gomez classification is false:**

A. **Based on height retardation**
B. Based on 50th centile Boston standards
C. Between 75–89% implies mild malnutrition
D. This classification has prognostic value for hospitalization of children

Ans A

88. **True abut vitamin D deficiency in children is/are:**

A. RDA for vitamin D is 400 IU
B. **PTH & 1,25-(OH) vitamin D play key roles in the 2 regulation of calcium and phosphate balance**
C. Serum calcium is always low
D. Serum phosphorus is low
E. PTH is normal

Ans B PTH & 1,25(OH) vitamin D play key roles in the regulation of calcium & phosphate balance; d. Serum phosphorus is low RDA of vitamin D for infants who are breastfed is 400 IU, while for older children it is 600 IU/day. In vitamin D deficiency
• PTH level gets elevated
• Hypocalcemia is a variable finding as a result of actions of elevated PTH to increase serum calcium
• Hypophosphatemia is caused by PTH-induced renal losses of phosphate & a decrease in intestinal absorption.

89. **The American academy of pediatrics change the vitamin D intake recommendation in 2010 to**

A. 200 IU/day
B. 400 IU/day
C. **600 IU/day**
D. 800 IU/day
E. 1000 IU/day

Ans C

90. **The outward sign of cell damage in malnutrition is**

A. repeated infections
B. wasting
C. **edema**
D. hepatomegaly
E. facial appearance

Ans C and hair/skin changes are outward signs of cell damage.

91. **Thrombocytopenia in refeeding syndrome is mainly a result of**

A. **hypophosphatemia**
B. hypokalemia
C. hypomagnesemia
D. thiamine deficiency
E. hyperglycemia

Ans A causes leucocyte dysfunction, hemolysis, and thrombocytopenia.

92. **Brown-Vialetto-Van Laere syndrome (BVVLS), a neurologic disorder characterized by progressive neurologic deterioration, sensorineural hearing loss, and pontobulbar palsy usually responds to treatment with high doses of**

A. niacin
B. biotin
C. **riboflavin**
D. pyridoxine
E. thiamin

Ans C Mutations in genes coding for riboflavin transporter proteins have been identified in children with BVVLS.

93. **All the following are recognized causes of rickets EXCEPT**

A. Mccune-Albright syndrome
B. epidermal nevus syndrome
C. **tuberus sclerosis**
D. neurofibromatosis
E. aluminum-containing antacids

Ans C Aluminum-containing antacids cause phosphorus deficiency. McCune-Albright syndrome, epidermal nevus syndrome, and Neurofibromatosis enhance renal losses.

94. **Dominant features in X-linked hypophasphatemic rickets are**

A. head; craniotabes, frontal bossing, and caries
B. back; scoliosis, lordosis, and kyphosis
C. chest; rachitic rosary and Harrison groove
D. **extremities; coxa vara and windswept deformity**
E. general; listlessness and failure to thrive

Ans D

95. Inorganic phosphorus in rickets is raised in

A. vitamin D deficiency
B. dietary calcium deficiency
C. Fanconi syndrome
D. **chronic renal failure**
E. tumor-induced rickets

Ans D All other distracters are associated with decreased level of inorganic phosphorus.

96. 25-hydroxy vitamin D level is reduced in

A. **vitamin D deficiency**
B. autosomal recessive hypophosphatemic rickets
C. autosomal dominant hypophosphatemic rickets
D. Fanconi syndrome
E. tumor-induced rickets

Ans A

97. The standard method for determining a patient's vitamin D status is

A. 25-hydroxylase level
B. **25-hydroxy vitamin D level**
C. 1, 25-dihydroxy vitamin D level
D. D-binding protein
E. PTH level

Ans B The level of 1, 25-D is only low when there is severe vitamin D deficiency.

Vitamin K Deficiency

98. Compared with cow's milk, mother's milk has more:

A. **Lactose**
B. Vit D
C. Proteins
D. Fats

Ans A

99. Which of the following is the best indicator of long term nutritional status?

A. Mid-arm circumference
B. **Height for age**
C. Weight for age
D. Weight for height

Ans B

100. Common to both acute and chronic malnutrition is:

A. **Weight for age**
B. Weight for height
C. Height for age
D. BMI

Ans A

101. In children having mild vitamin K deficiency, prothrombin time may be normal and estimation of protein Induced in vitamin K absence (PIVKA) is useful to detect early vitamin K deficiency. What is the exact form of PIVKA?

A. Undercarboxylated factor VII
B. **Undercarboxylated factor II**
C. Carboxylated factor VII
D. Carboxylated factor II

Ans B • In mild vitamin K deficiency, PT is normal, but there are elevated levels of undercarboxylated forms of proteins induced by vitamin K absence (PIVKA) that are normally carboxylated in the presence of vitamin K.
• Measurement of undercarboxylated factor II (PIVKA-II) can be used to detect mild vitamin K deficiency.
• Determination of blood vitamin K level is less useful because of significant variation based on recent dietary intake & levels do not always reflect tissue stores.

102. Baby fed exclusively on goat's milk develop:

A. **Craniofacial malformations**
B. Renal agenesis
C. Long bone defects
D. Chondrodysplasia punctata 50 Section 1: General Pediatrics
E. Acrodermatitis enteropathica Which of the following congenital malformation is seen in a child of a mother who is on treatment with oral anticoagulants?

Ans A Megaloblastic anemia Goat's milk is a poor source of folate; Hence, babies fed exclusively on Goat's milk may develop Megaloblastic Anemia.

103. Concerns to be considered among vegetarians are all the following EXCEPT

A. **higher bioavailability of iron**
B. lower B12 levels
C. risk of having lower level of fatty acids
D. lower levels of calcium and vitamin D3
E. lower bioavailability of zinc

Ans A The level is similar in both vegetarian and non-vegetarian diets but the bioavailability is lower in vegetarians.

104. The recommended transition time from stabilization to rehabilitation phase in the treatment of malnutrition is

A. 1 day
B. **3 days**
C. 5 days
D. 7 days
E. 10 days

Ans B A controlled transition over 3 days is recommended to prevent the "refeeding syndrome."

105. Adeposity rebound, the period when the body fat is typically at the lowest level, is best represented at the age of

A. 1 year
B. 3 years
C. **6 years**
D. 9 years
E. 12 years

Ans C Body fat levels decrease for approximately 5.5 yr until the period called adiposity rebound, when body fat is typically at the lowest level. Adiposity then increases until early adulthood.

106. **The following are recognized causes of craniotabes EXCEPT**

A. osteogenesis imperfect
B. rickets
C. syphilis
D. **Sotos syndrome**
E. normal newborn

Ans D Soto's syndrome causes large head.

107. **The following conditions/agents are incorporated in a similar presentation to vitamin K deficiency bleeding EXCEPT**

A. liver disease
B. hereditary factor II deficiency
C. high dose salicylate
D. **heparin**
E. rodent poison

Ans D Anticoagulant warfarin and rodent poisons contain Coumarin derivatives that inhibit the action of vitamin K by preventing its recycling to an active form.

108. **The first factor to be affected by deficiency of vitamin K is**

A. Factor I
B. Factor II
C. **Factor VII**
D. Factor IX
E. Factor X

Ans C has the shortest half life of the coagulation factors.

109. **The baby that is least vulnerable to develop vitamin K bleeding tendency is**

A. **a three-day old neonate of a healthy mother that startedimmediate breastfeeding after birth**
B. a three-year old child spent 21 days in intensive care unit receiving total parenteral nutrition
C. a nine-month old infant with prolonged history of diarrhea and multiple antibiotics therapy
D. a six-month old infant with cystic fibrosis missed to be diagnosed during early neonatal period
E. a six-hour neonate of an epileptic mother that received phenobarbital during pregnancy

Ans A Delayed breast feeding is a risk factor to vitamin K bleeding tendency.

Vitamin B Complex Deficiency

110. **Hind milk is richer in:**

A. Carbohydrate
B. Protein
C. **Fat**
D. Minerals

Ans C

111. **Admission to nutrition rehabilitation center for severe acute malnourished child between 6month and 5year is all except:**

A. Wt/ht < −3 SD
B. MUAC < 11.5 cm
C. **Wt < 2 SD for age**
D. Bilateral pedal edema

Ans C

112. **Acute malnutrition is assessed by:**

A. Weight for age
B. **Weight for height**
C. Height for age
D. Broca's index
E. Ponderal index

Ans B

113. **All of the following are vitamin B6 dependent syn dromes, except:**

A. Pyridoxine dependent seizures
B. Xanthurenic aciduria
C. Cystanthioninuria
D. **Darier disease**

Ans D • Vitamin B6 dependence syndromes include: pyridoxine-dependent epilepsy, a vitamin B6–responsive anemia, xanthurenic aciduria, cystathioninuria, and homocystinuria.
• Pyridoxine-dependent epilepsy involves mutations in ALDH7A1 gene causing deficiency of antiquitin, an enzyme involved in dehydrogenation of l-alpha-aminoadipic semialdehyde.

114. **Thiamine deficiencycauses:**

A. Pellagra
B. **Beri-beri**
C. Keshan's disease
D. Rickets

Ans B Beriberi Clinical features of Beri-beri (caused by thiamine deficiency):
• Peripheral neuritis (tingling, burning, paresthesias of toes and feet), decreased deep tendon reflexes, loss of vibration sense, tenderness and cramping of leg muscles, heart failure and psychological disturbances.
• Hoarseness or aphonia caused by paralysis of the laryngeal nerve is a characteristic sign.
• Muscle atrophy and tenderness of nerve trunks, ataxia, loss of coordination and loss of deep sensation.

115. **Vitamin B12 gets absorbed from:**

A. Stomach
B. Duodenum
C. **Ileum**
D. Colon

Ans C

116. **Recommended dose of folic acid during pregnancy is:**

A. 200 mcg/day
B. 300 mcg/day
C. **400 mcg/day**
D. 500 mcg/day

Ans C All females of childbearing age should take 400 mcg of folic acid/day.

117. **The symptoms of dietary deficiency of niacin (which results in pellagra) will be less severe if the diet has a high content of:**

A. **Tryptophan**
B. Tyrosine
C. Thymine
D. Thiamine

Ans A Tryptophan can be converted to nicotinamide adenine dinucleotide (NAD) 60 mg tryptophan gives rise to 1 mg niacin.

118. Consumption of which of the following cereal as staple diet is associated with Pellagra:

A. Rice
B. Wheat
C. **Maize**
D. Bajara

Ans C Maize Nelson 20/e 324-326

119. The classic clinical triad of Wernicke encephalopathy of thiamine deficiency is

A. **mental status changes, ocular signs, and ataxia**
B. cardiac involvement, peripheral neuritis, and aphonia
C. depression, drowsiness, and poor mental concentration
D. increased intracranial pressure, meningismus, and coma
E. deterioration of school performance, seizures, and headache

Ans A This triad is rarely reported in infants and young children with severe deficiency secondary to malignancies or feeding of defective formula.

120. Death from thiamine deficiency is usually due to

A. renal involvement
B. repeated infections
C. lactic acidosis
D. **cardiac involvement**
E. increased intracranial pressure

Ans D Death from thiamine deficiency usually is secondary to cardiac involvement. The initial signs are cyanosis and dyspnea, but tachycardia, enlargement of the liver, loss of consciousness, and convulsions can develop rapidly. The heart, especially the right side is enlarged.

121. Regarding thiamine deficiency

A. features develop within 2-3 weeks of deficient intake
B. poor mental concentration and depression are late features of the disease
C. **hoarseness or aphonia is a characteristic sign**
D. the term (dry) beriberi reflects the features of raised intracranial pressure
E. the term (wet) beriberi reflects the features of renal dysfunction

Ans C It can develop within 2-3 months of deficient intake. Poor mental status is an early feature. Hoarseness or aphonia caused by paralysis of the laryngeal nerve is a characteristic sign. Dry beriberi reflects neuronal involvement while wet beriberi reflects cardiac involvement.

122. Thiamine-responsive megaloblastic anemia syndrome is a rare disorder, it is characterized by the following EXCEPT

A. diabetes mellitus
B. **peripheral neuritis**
C. megaloblastic anemia
D. sensorineural hearing loss
E. autosomal recessive inheritance

Ans B This is an autosomal recessive disorder, characterized by Megaloblastic anemia, diabetes mellitus, and sensorineural hearing loss.

123. The MOST convenient way to confirm a diagnosis of pellagra in children is

A. skin biopsy
B. urinary 2-pyridone
C. **response to niacin treatment**
D. urinary n1-methyl-nicotinamide
E. physical signs of glossitis and dermatitis

Ans C Rapid clinical response to niacin treatment is an important confirmatory test.

124. The MOST characteristic manifestation of pellagra is

A. **dermatitis**
B. dementia
C. diarrhea
D. anorexia
E. lassitude

Ans A the most characteristic manifestation of pellagra, can develop suddenly or insidiously and may be initiated by irritants, including intense sunlight.

125. The dietary reference intake of pyridoxine for a healthy eight-month-old infant is

A. 0.1 mg/day
B. **0.3 mg/day**
C. 0.5 mg/day
D. 1.0 mg/day
E. 3 mg/day

Ans B 0-6 months: 0.1 mg/ day; 7-12 months: 0.3 mg/ day; 1-3 yr: 0.5 mg/day; 4-8 yr: 0.6 mg/day; 9-13 yr: 1.0 mg/day 14-18 yr: 1.2 - 1.3 mg/day.

126. All the following manifestations of pyridoxine deficiency are seen in children

A. seizures
B. cheilosis
C. listlessness
D. **peripheral neuritis**
E. seborrheic dermatitis

Ans D is a feature of deficiency in adults but is not usually seen in children.

127. Pyridoxine is not affected in patients receiving

A. isoniazid
B. **valproate**
C. phenytoin
D. penicillamine
E. corticosteroids

Ans B Other distracters cause pyridoxine deficiency.

128. Folate is important during embryogenesis for the development of

A. lungs
B. genital organs
C. gastrointestinal system
D. **central nervous system**
E. cardiovascular system

Ans D

129. **All the following can cause folate deficiency EXCEPT**

A. celiac disease
B. sickle cell anemia
C. **6-mercaptopurine therapy**
D. inflammatory bowel disease
E. methylene tetrahydrofolate reductase deficiency

Ans C Methotrexate, not 6-mercaptopurine.

130. **Clinical features of cerebral folate deficiency include the following EXCEPT**

A. **lethargy**
B. microcephaly
C. atatxia
D. ballismus
E. blindness

Ans A It manifests usually at 4-6 months of age. Other features are irritability (not lethargy), developmental delay, pyramidal tract signs, choreoathetosis and seizures.

131. **One of the recognized non-hematological manifestations of folic acid deficiency in children is**

A. repeated respiratory abscesses
B. recurrent abdominal pain
C. recurrent seizures
D. **growth retardation**
E. teeth decay

Ans D Non hematologic manifestations include glossitis, listlessness, and growth retardation not related to anemia.

132. **All the following are features of hereditary folate malabsorption EXCEPT**

A. **anal ulcers**
B. chronic diarrhea
C. failure to thrive
D. megaloblastic anemia
E. opportunistic infections

Ans A Hereditary folate malabsorption manifests at 1-3 months of age with recurrent or chronic diarrhea, failure to thrive, oral ulcerations, neurologic deterioration, megaloblastic anemia, and opportunistic infections.

133. **The best indicator of chronic folate deficiency is**

A. serum folate level
B. **RBC folate level**
C. urinary folate level
D. bone marrow aspiration
E. CSF 5- methyltetrahydrofolate level

Ans B The normal RBC folate level is 150-600 ng/ mL of packed cells.

134. **Effective supplementation of folate for prevention of neural tube defect is**

A. **started at least 1 mo before conception, and continued through the first 2-3 mo of pregnancy**
B. started at least 3 mo before conception, to be stopped after positive pregnancy test.
C. started after positive pregnancy test and continued through the first 2-3 mo of pregnancy.
D. started at least 1 mo before conception, to be stopped after positive pregnancy test.
E. started at least 3 mo before conception, and continued through the first 2-3 mo of pregnancy

Ans A All women desirous of becoming pregnant should consume 400-800 µg folic acid daily; the dose is 4 mg/ day in those having delivered a child with neural tube defect.

135. **The dietary reference intake (DRI) of folate for a healthy eight-month-old infant is**

A. **80 microgm/day**
B. 150 microgm/day
C. 200 microgm/day
D. 300 microgm/day
E. 400 microgm/day

Ans A The DRIs for folate are 65 µg for infants 0-6 months of age and 80µg between 6-12 months of age. For older children, the DRIs are 150 µg for ages 1-3 years; 200 µg for ages 4-8 years; 300 µg for ages 9-13 years; and 400 µg for ages 14-18 years.

136. **Massive doses of folate given by injection have the potential to cause**

A. ototoxicity
B. **neurotoxicity**
C. nephrotoxicity
D. severe abdominal pain
E. respiratory difficulty

Ans B

137. **Important mechanism for maintaining vitamin B12 nutriture include the following**

A. direct absorption
B. **acidic pH in the ileum**
C. enterohepatic circulation
D. binding with intrinsic factor
E. intestinal bacterial synthesis

Ans B Alkaline pH not acidic.

138. **Vitamin B12 is important for all of the following EXCEPT**

A. hematopoiesis
B. mental development
C. **skeletal development**
D. psychomotor development
E. central nervous system myelination

Ans C

139. **All the following are real risk factors for vitamin B12 deficiency EXCEPT**

A. celiac disease
B. ileal resection
C. Imerslund-gräsbeck disease
D. helicobacter pylori infection
E. **use of proton pump inhibitors**

Ans E and/or histamine 2 receptor antagonists may increase the risk of deficiency but is not a real cause of deficiency.

140. **Common observations with vitamin B12 deficiency in children include all the following EXCEPT**

A. hypotonia
B. **peripheral neuritis**
C. developmental regression
D. involuntary movements
E. hyperpigmentation of the knuckles

Ans B Irritability, developmental delay, developmental regression, and involuntary movements are the common neurologic symptoms in infants and children, whereas sensory deficits, paresthesias, and peripheral neuritis are seen in adults. Hyperpigmentation of the knuckles and palms is another common observation with B12 deficiency in children.

141. **The dietary reference intake of cobalamin for a healthy eight-month-old infant is**

A. **0.5 microgm/day**
B. 1.0 microgm/day
C. 1.5 microgm/day
D. 2.0 microgm/day
E. 2.5 microgm/day

Ans A 0-6 months: 0.4 µg/day; 7-12 months: 0.5 µg/day; 1-3 yr: 0.9 µg/day 4-8 years: 1.2 µg/day 9-13 years: 1.8 µg/day 14-18 years: 2.4 µg/day.

Vitamin C & Scurvy

142. **Adaptive starvation with increased metabolism:**

A. Bulimia
B. Kwashiorkor
C. **Marasmus**
D. Cachexia

Ans C Marasmus is a chronic, fairly well-adapted form of starvation rather than an acute illness.

143. **Waterlow classification of malnutrition in children takes into account:**

A. Weight for height (wasting)
B. Height for age (stunting)
C. **Weight for height (wasting) and height for age (stunting)**
D. Percent of reference weight for age

Ans C Gomez Criteria Waterlow Criteria Grade of malnutrition Weight for age (%) Weight for height (%) Height for age (%) Normal 90–110 90–110 >95 Mild 75–89 80–89 90–94 Moderate 60–74 70–79 85–89 Severe <60 <70 <85

144. **Deficit in weight for height in a 3yearold child indicates:**

A. **Acute malnutrition**
B. Chronic malnutrition
C. Concomitant acute and chronic
D. Underweight

Ans A

145. **In which stage corneal xerosis is seen?**

A. Stage X1
B. **Stage X2**
C. Stage X3A
D. Stage X3B

Ans B Stage X2 with

146. **The adequate intake (AI) for the total digestible carbohydrates in a nine-month-old boy is**

A. 60 gm/day
B. **95 gm/day**
C. 120 gm/day
D. 145 gm/day
E. 170 gm/day

Ans B The AI is 60 gm/day for 0-6 months and 95 gm/day for 7-12 months.

147. **All the following are dimensions of food security EXCEPT**

A. access
B. utilization
C. **preparation**
D. stability
E. availability

Ans C

148. **The WHO recommendation for antibiotic cover in the stabilization phase of treating malnourished boy free of initial complications is to use**

A. **oral amoxicillin**
B. parenteral penicillin
C. oral second generation cephalosporin
D. parenteral gentamicin
E. parenteral third generation cephalosporin

Ans A If no complications; oral Amoxicillin 25 mg/kg twice daily for 5 days. If there are any complications (shock, hypoglycemia, hypothermia, skin lesions, urinary tract or respiratory tract infections, or lethargy); Gentamicin 7.5 mg/kg IV or IM once daily for 7 days and Ampicillin 50 mg/kg IV or IM every 6 hours for 2 days then oral Amoxicillin 25- 40 mg/kg every 8 hours for 5 days.

149. **The gastrointestinal hormone/response that stimulates appetite in children is**

A. cholecystokinin
B. glucagon-like peptide-1
C. peptide yy
D. vagal neuronal feedback
E. **ghrelin**

Ans E Gastrointestinal hormones, including cholecystokinin, glucagon-like peptide-1, peptide YY, and vagal neuronal feedback promote satiety. Ghrelin stimulates appetite.

150. **Radiographic features of scurvy are similar to that of**

A. **copper deficiency**
B. magnesium deficiency
C. vitamin A excess
D. vitamin D excess
E. mercury exposure

Ans A

151. **The best indicator of the body stores of vitamin C is**

A. plasma ascorbate concentration
B. **leukocyte ascorbate concentration**
C. urinary ascorbate level
D. urinary amino acid level
E. plasma amino acid level

Ans B

152. **The MOST specific but late radiographic feature of scurvy is**

A. ground-glass appearance of the shafts of the long bones
B. pencil outlining of the epiphysis and diaphysis
C. the white line of fränkelat the metaphysis
D. **Trümmerfeld zone at the metaphysis**
E. Pelkan spur at cortical ends

Ans D The more specific but late radiologic feature of scurvy is a zone of rarefaction under the white line at the metaphysis . is zone of rarefaction (Trümmerfeld zone), a linear break in the bone that is proximal and parallel to the white line, represents area of debris of broken-down bone trabeculae and connective tissue.

153. **Ingestion of large doses of vitamin C can cause**

A. musculoskeletal pain
B. **osmotic diarrhea**
C. severe headache
D. respiratory symptoms
E. hematemesis

Ans B Larger doses of vitamin C can cause gastrointestinal problems, such as abdominal pain and osmotic diarrhea.

Iron Deficiency & Anemia

154. **Which of the following would be affected if malnutrition is longstanding?**

A. Weight for age
B. **Height for age**
C. Weight for height
D. All of the above46 Section 1: General Pediatrics

Ans B

155. **In protein deficiency, all are seen except:**

A. Flaky paint like skin
B. **Glossitis**
C. Nail change
D. Cherry like skin

Ans B Glossitis is a feature of vitamin B complex deficiency.

156. **Casal necklace is seen in?**

A. Scurvy
B. Kwashiorkar
C. **Pellagra**
D. Indicanuria

Ans C a. 13 gm Daily protein requirement is: 1.5 g/kg/day for infants (~ 10 g/day), 1.1 g/kg/day for 1–3 years (~ 13 g/day), 0.95 g/kg/day for 4–13 years, (4–8 years → 19 g/day, 9–13 years → 34 g/day) & 0.85 g/kg/day for 14–18 years, (~52 g/day for males & 46 g/day for females)

157. **A rare autosomal recessive disorder caused by mutation in SLC19A2 gene and characterized by megaloblastic anemia, diabetes mellitus and sensorineural hearing loss responds to which of the following nutrient?**

A. Vitamin C
B. Vitamin B12
C. **Vitamin B1**
D. Folic acid

Ans C Thiamineresponsive megaloblastic anemia (TRMA) syndrome:
• It is a rare autosomal recessive disorder characterized by megaloblastic anemia, diabetes mellitus, and sensorineural hearing loss, responding in varying degrees to thiamine treatment.
• It occurs because of mutations in the SLC19A2 gene, encoding a thiamine transporter protein, leading to abnormal thiamine transportation and cellular vitamin deficiency. Note: Thiamine may improve the outcome in children with Leigh's disease and type 1 diabetes mellitus.

158. **The adequate intake (AI) for the total fat in a nine-month-old boy is**

A. 10 gm/day
B. 20 gm/day
C. **30 gm/day**
D. 40 gm/day
E. 50 gm/day

Ans C The AI is 31 gm/day for 0-6 months and 30 gm/day for 7-12 months.

159. **The TRUE hemoglobin cutoff to define anemia is**

A. 120 g/L for children 6-59 mo
B. **115 g/L for children 5-11 yr**
C. 110 g/L for children 12-14 yr
D. 110 g/L for non-pregnant women
E. 120 g/L for men

Ans B Hemoglobin cutoffs to define anemia are 110 g/L for children 6-59 mo, 115 g/L for children 5-11 yr, and 120 g/L for children 12-14 yr. Cutoffs to define anemia for non- pregnant women are 120 g/L, 110 g/L for pregnant women, and 130 g/L for men.

160. **During treatment of malnutrition, the signal of entry to the rehabilitation phase is**

A. **reduced edema**
B. resolution of infection
C. disappearance of signs of micronutrient deficiency
D. constant blood sugar level
E. constant body temperature

Ans A The signals for entry to this phase are reduced or minimal edema and return of appetite.

161. **The following physical features might be discovered during physical examination of an obese 10-year-old boy, and each reflect the associated morbidity EXCEPT**

A. developmental delay suggests genetic disorder
B. day time sleepiness suggests sleep apnea
C. **polyuria and nocturia suggest type 1 diabetes**
D. hip pain suggests blount disease
E. abdominal pain suggests non alcoholic fatty liver disease

Ans C Type 2 diabetes mellitus is influenced in obesity, not type 1.

162. **Parathyroid hormone level is reduced in**

A. vitamin D dependent rickets
B. X- linked hypophosphatemic rickets
C. **hypophosphatasia**
D. dietary calcium deficiency
E. vitamin D deficiency

Ans C

Zinc, Copper & Trace Elements

163. **A 4yearold child with severe wasting and voracious appet ite without pedal edema:**

A. Kwashiorkor
B. **Marasmus**
C. Both
D. None

Ans B

164. **In order to discharge from hospital, malnourished child should have minimum weight gain of:**

A. **5 gm/kg/day**
B. 10 gm/kg/day
C. 15 gm/kg/day
D. 20 gm/kg/day

Ans A

165. **Which of the following are the qualities of a green light food as per traffic light diet plan for children?**

A. Nutrition dense, but higher in calories and fat
B. **Low calories, high fiber, low fat, Nutrition-dense**
C. High in calories, sugar and fat
D. None of the above

Ans B Low calories, high fiber, low fat, Nutritiondense The "traffic light" diet groups foods into the following categories: Category Characteristics Recommended intake Examples dna Red High in calories, sugar, and fat Reserved for infrequent treats Fatty meats, sugar, sweet beverages, fried foods Yellow Nutrient-dense, but higher in calories & fat In moderation Lean meats, dairy, starches, grains Green Low-calorie, high-fiber, low-fat, nutrient-dense Without any limitations Fruits, vegetables

166. **Cardiac edema and neuropathy is seen in which micronutrient deficiency?**

A. Biotin
B. Pyridoxine
C. **Thiamine**
D. Riboflavin

Ans C Thiamine deficiency gives rise to cardiomyopathy and peripheral neuropathy.

167. **Zinc deficiency leads to all except?**

A. Delayed sexual maturation
B. Impaired immune function
C. Skeletal abnormalities
D. **Excessive appetite**

Ans D

168. **Abnormalities of copper metabolism are implicated in the pathogenesis of all the following except:**

A. Wilson's disease
B. Menke's Kinky-hair syndrome
C. Indian childhood cirrhosis
D. **Keshan disease**

Ans D Keshan disease is a disorder of selenium metabolism. Diseases of copper metabolism are Wilson's disease, Menkes kinky hair syndrome, Indian childhood cirrhosis.

169. **Selenium deficiency is seen in:**

A. **1 mg/day for 10–14 days**
B. 10 mg/ day for 10–14 days
C. 15 mg/ day for 10–14 days
D. 20 mg/ day for 10–14 days

Ans A Keshan disease

170. **Zinc deficiency is associated with increased risk for all the following EXCEPT**

A. stunting
B. impaired immune function
C. increased risk for respiratory diseases
D. increased risk for diarrheal diseases
E. **increased risk for skin infections**

Ans E Zinc is Essential for proper growth and development, and an important catalyst for about 100 specific enzymes.

171. **For children <5 yr, the highest global prevalence of micronutrient and trace elements deficiencies is that of**

A. **vitamin A deficiency**
B. zinc deficiency
C. iron deficiency
D. iodine deficiency
E. copper deficiency

Ans A For children <5 yr, the global prevalence is estimated to be 33% for vitamin A deficiency, 29% for iodine deficiency, 17% for zinc deficiency, and 18% for iron- deficiency anemia. Prevalence of micronutrient deficiencies tends to be highest in Africa.

172. **The following vitamins and trace elements are recommended to be given during the stabilization phase of malnutrition EXCEPT**

A. **iron**
B. vitamin A
C. folic acid
D. zinc
E. multivitamins

Ans A should be avoided in this phase.

173. **One of the following matching is TRUE regarding the trace elements deficiencies and their consequences**

A. zinc and hypogonadism
B. selenium and dental caries
C. iron and hypothyroidism
D. fluoride and cardiomyopathy
E. manganese and decreased alertness

Ans A Selenium deficiency causes cardiomyopathy and myopathy; Iron deficiency causes anemia and learning impairment; Manganese deficiency causes hypercholesterolemia and weight loss; Fluoride deficiency causes dental caries.

174. **One of the following matching is TRUE regarding the trace element excess and its consequence**

A. copper and congenital hypothyroidism
B. manganese and hyperuricemia
C. iron and abdominal pain
D. zinc and cholestatic jaundice
E. fluoride and nail changes

Ans C Copper excess causes acute (Nausea, vomiting and hepatic coma) and chronic toxicity (liver and brain injury); Manganese excess causes neurological manifestations and cholestatic jaundice; Zinc excess causes abdominal pain, diarrhea and vomiting; Fluoride excess causes dental fluorosis.

Iodine Deficiency & Goiter

175. **Decrease in weight for height indicates?**

A. Acute malnutrition
B. Chronic malnutrition
C. Both acute and chronic malnutrition
D. Short stature

Ans A

176. **Flaky paint appearance of skin is seen in:**

A. Dermatitis
B. Pellagra
C. Marasmus
D. Kwashiorkor

Ans D

177. **Recommended daily dose of Iodine in a child is:**

A. 120–200 mg
B. 90–120 mg
C. 30–60 mg
D. 500 mg

Ans B The recommended dietary allowance of iodine is 30 mg/kg/24 hr for infants, 90–120 mg/24 hr for children, and 150 mg/24 hr for adolescents and adults.

178. **A child presents with malnutrition, persistent diarrhea. Apart from antibiotics what would you like to add in your prescription?**

A. Zinc, Vit A, Iron
B. Zinc, Iron
C. Zinc, Vit A
D. B complex, Zinc

Ans C An episode of diarrhea, of presumed infectious etiology, which starts acutely but lasts for > 14 days Principles of management are: (i) correction of dehydration, electrolytes & hypoglycemia, (ii) evaluation for infections & their management, and (iii) nutritional therapy with supplemental multivitamins & minerals, twice the RDA Vitamin A (1 lac–2 lac IU) & zinc (10–20 mg/day) are supplemented as both of them enhance the recovery from persistent diarrhea.

179. **The adequate intake (AI) for the total protein in a nine-month-old boy is**

A. 9 gm/day
B. 11 gm/day
C. 13 gm/day
D. 15 gm/day
E. 17 gm/day

Ans B The AI is 9 gm/day for 0-6 months and 11 gm/day for 7-12 months.

180. **The MOST commonly used index for nutritional status is**

A. height-for-age
B. weight-for-height
C. body mass index
D. mid-upper arm circumference
E. weight-for-age

Ans E is the most commonly used index of nutritional status, although a low value has limited clinical significance as it does not differentiate between wasting and stunting. Weight-for-age has the advantage of being somewhat easier to measure than indices that require height measurements.

181. **In consideration of preventing infections in stabilization phase of malnutrition in unimmunized 10-month-old boy. Of the following, the vaccine that is recommended to be given is**

A. oral polio vaccine
B. acellular pertussis vaccine
C. measles vaccine
D. pneumococcal vaccine
E. H. influenza vaccine

Ans C is given to unimmunized children age >6 mo.

182. **Chronic hypervitaminosis A is associated with all the following features EXCEPT**

A. seborrhea
B. alopecia
C. hepatosplenomegaly
D. bulging fontanel
E. echymosis

Ans E Manifestations of vitamin C deficiency.

183. **Urinary phosphorus is reduced in**

A. **chronic renal failure**
B. autosomal dominant hypophosphatemic rickets
C. autosomal recessive hypophosphatemic rickets
D. dietary calcium deficiency
E. Fanconi syndrome

Ans A Reduced in chronic kidney disease and dietary phosphorus deficiency only while elevated in all other forms of rickets.

Obesity & Overnutrition

184. **In severe malnutrition feeding is started with diet which has all except:**

A. Osmolarity less than 350 mOsmol/L
B. Lactose 2–3 g/kg/day
C. **Calories from protein > 15%**
D. Renal solute load urinary < 600 mOsmol/L

Ans C For a child with severe malnutrition, start feeding as soon as possible with a diet, which has:
• Osmolarity <350 mOsm/L; Lactose < 2–3 g/kg/day • Adequate bioavailability of micronutrients
• Appropriate solute load (urinary osmolarity <600 mOsm/L) • Low viscosity, easy to prepare and socially acceptable
• Initial percentage of calories from protein of 5% • Adequate storage, cooking and refrigeration

185. **Flag sign is seen in:**

A. **Kwashiorkor**
B. Beriberi
C. Pellagra
D. Marasmus

Ans A

186. **Acrodermatitis enteropathica is seen with deficiency of:**

A. **Zinc**
B. Iron
C. Copper
D. Vitamin A

Ans A Deficiency of zinc leads to acrodermatitis enteropathica, in which rashes shown on skin, especially around the orifices.

187. **All of the following are features of vitamin E deficiency except:**

A. Areflexia
B. **Seizure**
C. Myopathy
D. Ataxia

Ans B Clinical Features of Vitamin E Deficiency: Loss of deep tendon reflexes (areflexia) is usually the initial finding Progressive cerebellar disease & posterior column dysfunction Limb ataxia and truncal ataxia, dysarthria, nystagmus, positive Romberg test, decreased vibratory sensation seen Some patients have pigmentary retinopathy & visual field constriction that can progress to blindness Cognition & behavior can also be affected; Myopathy and cardiac arrhythmias are less-common findings, but may be seen

188. **All the following amino acids are indispensable in human diet EXCEPT**

A. leucine
B. methionine
C. threonine
D. valine
E. **alanine**

Ans E aspartic acid, asparagine, glutamic acid, and serine are dispensable amino acids.

189. **The MOST profound consequence of undernutrition is**

A. **premature death**
B. repeated infections
C. stunting
D. cell damage
E. developmental delay

Ans A

190. **Emergency treatment in severe malnutrition includes all the following EXCEPT**

A. shock
B. hypoglycemia
C. dehydration
D. severe anemia
E. **infections**

Ans E Treatment of infections is one of the therapeutic directives for stabilization phase.

191. **Across all racial groups, the factor that confers protection against childhood obesity is**

A. gestational weight gain
B. high birth weight
C. maternal smoking
D. breast feeding
E. **maternal education**

Ans E Prenatal factors including high preconceptual weight, gestational weight gain, high birth weight, and maternal smoking are associated with increased risk for later obesity. Paradoxically, intrauterine growth restriction with early infant catch-up growth is associated with the development of central adiposity and adult-onset cardiovascular risk. Breastfeeding is only modestly protective for obesity.

192. **All the following are childhood obesity-associated comorbidities later in life EXCEPT**

A. hypertension
B. gallbladder disease
C. tibia vara
D. asthma
E. **type 1 diabetes mellitus**

Ans E Type 2 diabetes mellitus is a childhood obesity-associated comorbidity.

193. **Genetic disorders associated with obesity can have all the following features**

A. dysmorphic features
B. cognitive impairment
C. vision abnormalities
D. hearing abnormalities
E. **tall stature**

Ans E Short stature is a common finding in children with obesity due to genetic disorders.

194. **The following are well recognized causes of secondary obesity EXCEPT**

A. myelodyplasia
B. muscular dystrophy
C. anti-psychotic medications
D. achondroplasia
E. **anti-migraine medications**

Ans E Antimigraine medications do not influence obesity in children. All other distracters are true causes of secondary obesity.

195. **The investigations to be done as part of initial evaluation of a newly identified obesity in a 12-year-old girl are the following EXCEPT**

A. fasting plasma glucose
B. triglyceride level
C. lipoprotein level
D. liver function tests
E. **renal function tests**

Ans E Glucose, Insulin, Triglyceride level, LDL and HDL, total cholesterol, and liver function tests are the initial markers to be done during evaluation of an obese 12-year girl.

196. **The only Food and Drug Administration (FDA)-approved medication for obesity in children <16 yr old is**

A. **orlistat**
B. phentermine
C. topiramate
D. amylin
E. lorcaserin

Ans A decreases absorption of fat, resulting in modest weight loss. Complications include flatulence, oily stools, and spotting.

Nutritional Rehabilitation

197. **In a child with malnutrition, the first line of management would be:**

A. Prevention of hypothermia
B. **Prevention of hypoglycemia**
C. Treatment of infection
D. Treatment of dehydration

Ans B Management of hypoglycemia, hypothermia & dehydration are the first 3 steps in a child with severe acute malnutrition

198. **Poor indicator to response during treatment of malnu trition includes:**

A. Failure to gain appetite by day
B. **Presence of edema on day**
C. Failure to gain at least 5 g/kg/day by day
D. Failure to gain at least 5 g/kg/day by day
E. Failure to gain at least 5 g/kg/day by day for 3 consecutive days during the rehabilitation phase

Ans B 10; d. Failure to gain at least 5 g/kg/day by day 10; e. Failure to gain at least 5 g/kg/day by with day for 3 consecutive days during the rehabilitation phase;

199. **The disease which is characterized by three D's—diarrhea, dermatitis and dementia is due to deficiency of?**

A. Vitamin A
B. **Niacin**
C. Folate
D. Vitamin C

Ans B Diarrhea, Dermatitis & Dementia are seen in Pellagra, which is due to Niacin deficiency.

200. **Administration of which of the following can prevent 125. neural defects in newborn?**

A. **Folic acid**
B. Vit B12
C. Pyridoxine
D. Vitamin C

Ans A

201. **Essential fatty acids deficiency is associated with all the following features EXCEPT**

A. desquamating skin rashes
B. alopecia
C. **thrombocytosis**
D. impaired immunity
E. growth deficits

Ans C Thrombocytopenia.

202. **"Reductive adaptation" process of evolution to a state of full malnutrition, all the following events will be ensued EXCEPT**

A. the liver makes glucose less readily
B. less heat production
C. gut produces less gastric acid and gut enzymes
D. **evident responses to infection**
E. reduced red cell mass

Ans D Immune function is impaired, especially cell-mediated immunity. The usual responses to infection may be absent, even in severe illness, increasing the risk of undiagnosed infection.

203. **The aim of the rehabilitation phase in the treatment of malnutrition is to**

A. repair cellular function
B. correct fluid and electrolyte imbalance
C. restore homeostasis
D. **catch-up growth**
E. prevent death from infection

Ans D The aim of the rehabilitation phase is to restore wasted tissues (i.e., catch-up growth).

204. **The following are characterstic features of acute hypervitaminosis A EXCEPT**

A. nausea and vomiting
B. drowsiness
C. diplopia
D. cranial nerve palsies
E. **seizures**

Ans E Nausea, vomiting, and drowsiness; less-common symptoms include diplopia, papilledema, cranial nerve palsies, and other symptoms suggesting pseudotumor cerebri are the hall mark of acute Hypervitaminosis A. Seizures are unlikely.

205. **Regarding vitamin E deficiency in premature infants, one of the following is TRUE**

A. it does not cross the placenta
B. hemolysis develops in the first week of life
C. premature formula that contains poly unsaturated fatty acids may carry a protective effect against hemolysis
D. **excessive iron supplementation for premature infants may augment the . oxidative stress .**
E. causes thrombocytopenia

Ans D It crosses the placenta significantly during the third trimester, which brings premature infants at risk. Hemolysis in premature infants typically started in the second month of life. Formulas containing poly unsaturated fatty acids may lead to exacerbation of deficiency as they make the red blood cells susceptible to oxidative stresses. In premature infants, deficiency causes hemolytic anemia and thrombocytosis, not thrombocytopenia.

04

CHAPTER 04

Fluid and Electrolyte Disturbances



WHAT THIS CHAPTER COVERS

- Body Water & Fluid Compartments
- Dehydration & Rehydration
- Hyponatremia & Hypernatremia
- Hypokalemia & Hyperkalemia
- Hypocalcemia & Hypercalcemia
- Acid-Base Disorders
- Metabolic Acidosis
- Metabolic Alkalosis
- Respiratory Acidosis & Alkalosis
- Oral Rehydration Therapy
- IV Fluids & Maintenance
- Diabetic Ketoacidosis

104 MCQs **12** subtopics

4.1 Body Water & Compartments

Age	Total body water (% body weight)	ECF	ICF
Fetus (early)	~90%		
Preterm	80–85%	ECF > ICF	
Term newborn	**75%**	**45%**	33%
1 year	**60%** (adult level)	20–25%	35–40%
Adult male / female	60% / 50%	20%	40%

- **ECF : ICF reverses** (ICF becomes larger) by **1 year**. Neonates have larger ECF → greater susceptibility to dehydration.
- Intravascular volume (blood volume): **neonate 85–90 mL/kg**, infant 80 mL/kg, adult 70 mL/kg.
- Plasma osmolality = **2 × Na + glucose/18 + BUN/2.8** (normal 285–295 mOsm/kg).

Maintenance fluids – Holliday–Segar

Weight	Per day	Per hour ("4-2-1 rule")
First 10 kg	**100 mL/kg**	4 mL/kg/h
Next 10 kg (11–20)	**+50 mL/kg**	2 mL/kg/h
Each kg > 20	**+20 mL/kg**	1 mL/kg/h

- Example: 25 kg child → 1000 + 500 + 100 = **1600 mL/day** (~65 mL/h).
- Maintenance electrolytes: **Na 2–3 mEq/kg/day, K 1–2 mEq/kg/day**.
- Current practice (AAP 2018): **isotonic fluids (0.9% NaCl/Ringer lactate/Plasma-Lyte with 5% dextrose)** for maintenance in children 28 days–18 y to **prevent hospital-acquired hyponatraemia**; avoid hypotonic fluids (0.18% NaCl).
- **Neonatal fluids**: day 1 – **60–80 mL/kg/day** (10% dextrose, no Na/K first 48 h), increase by **10–20 mL/kg/day** to **150 mL/kg/day** by day 7; preterms need more (insensible losses ↑ under radiant warmer, phototherapy +10–20%).
- **Insensible water loss**: **~300–400 mL/m²/day** (≈ 1/3 of maintenance); ↑ by **12% for every 1 °C rise of temperature**.

Common IV fluids (mEq/L)

Fluid	Na	K	Cl	Other	Use
0.9% NS	154	–	154	Osmolarity 308	Bolus, shock, resuscitation
Ringer lactate	130	4	109	Lactate 28, Ca 3	Bolus, severe dehydration (WHO Plan C)
N/2 (0.45%) saline	77	–	77		
N/5 (0.18%) + 5% D	31	–	31		(Old neonatal maintenance – avoid in children)
Isolyte-P	25	20	22	Acetate 23, Mg 3, PO_4 3	Paediatric maintenance (hypotonic)
3% saline	513	–	513		Symptomatic hyponatraemia, raised ICP
10% dextrose	–	–	–	100 g/L, 340 kcal/L	Neonatal fluid, hypoglycaemia

4.2 Dehydration & Diarrhoea Management (WHO / IMNCI)

Assessment of dehydration

Sign	No dehydration	Some dehydration (≥ 2 signs)	Severe dehydration (≥ 2 signs)
General condition	Well, alert	**Restless, irritable**	**Lethargic or unconscious**
Eyes	Normal	**Sunken**	Sunken
Thirst	Drinks normally	**Drinks eagerly, thirsty**	**Drinks poorly / not able to drink**
Skin pinch	Goes back quickly	**Goes back slowly (< 2 s)**	**Goes back very slowly (≥ 2 s)**
% loss (approx.)	< 5%	5–10%	> 10%
Plan	**Plan A** (home)	**Plan B** (ORS in health facility)	**Plan C** (IV fluids)

Plan A – no dehydration

- ORS after each loose stool: **< 2 y: 50–100 mL**; **2–10 y: 100–200 mL**; > 10 y: as much as wanted.
- Continue feeding; **zinc**; return if danger signs.

Plan B – some dehydration

- **ORS 75 mL/kg over 4 hours** (by weight); reassess after 4 h.

Plan C – severe dehydration (IV Ringer lactate 100 mL/kg)

Age	First 30 mL/kg in	Then 70 mL/kg in
Infant < 12 months	**1 hour**	**5 hours**
≥ 12 months	**30 minutes**	**2½ hours**

- Repeat 30 mL/kg once if radial pulse still weak. Reassess every 1–2 h; give ORS (5 mL/kg/h) as soon as child can drink (usually after 3–4 h in infants, 1–2 h in older children).
- If IV not possible → **NG ORS 20 mL/kg/h for 6 h (120 mL/kg)**.
- **SAM**: avoid Plan C; use ReSoMal (see Nutrition).

ZINC & OTHER RULES IN DIARRHOEA

Zinc: 10 mg/day < 6 m, 20 mg/day ≥ 6 m for 14 days. Antibiotics only for **dysentery (ciprofloxacin/ceftriaxone/azithromycin; IAP prefers ceftriaxone/azithromycin), suspected cholera with severe dehydration (doxycycline/azithromycin)**, and giardiasis/amoebiasis (metronidazole). **No antimotility drugs, no antiemetics routinely** (single-dose ondansetron allowed in older children with vomiting).

Oral rehydration solution

Component (mmol/L)	Low-osmolarity WHO-ORS (2002)	Old WHO-ORS (1975)	ReSoMal
Sodium	**75**	90	**45**
Potassium	**20**	20	**40**
Chloride	**65**	80	70
Citrate	**10**	10	7
Glucose	**75**	111	125
Total osmolarity	**245 mOsm/L**	311	300

- Composition in grams/L: **NaCl 2.6, glucose anhydrous 13.5, KCl 1.5, trisodium citrate 2.9**.

- **Mechanism**: **sodium–glucose co-transport (SGLT-1)** in small-intestinal villi is **intact in secretory diarrhoea**; glucose : Na ratio ~1 : 1.
- Low-osmolarity ORS → **↓ stool output by 20–25%, ↓ vomiting by 30%, ↓ need for IV fluids by 33%**.
- Prepared ORS is used within **24 hours**.
- Home-available fluids: salted rice water, lassi, soup, coconut water (not soft drinks/sweet juices – high osmolarity).

4.3 Sodium Disorders

Hyponatraemia (Na < 135 mEq/L)

Volume status	Causes
Hypovolaemic	GI losses (diarrhoea treated with hypotonic fluids – MCC), **CAH (salt-wasting 21-OH def.)**, diuretics, cerebral salt wasting, pseudohypoaldosteronism, CF (sweat)
Euvolaemic	**SIADH** (meningitis, pneumonia, CNS injury, drugs), hypothyroidism, glucocorticoid deficiency, water intoxication (diluted formula)
Hypervolaemic	Nephrotic syndrome, CCF, cirrhosis, AKI/CKD
Pseudohyponatraemia	Hyperlipidaemia, hyperproteinaemia (isotonic)
Translocational	**Hyperglycaemia** (Na falls 1.6 mEq/L per 100 mg/dL glucose rise), mannitol

- Symptoms (esp. Na < 125 or acute fall): nausea, headache, lethargy, **seizures**, coma (cerebral oedema).
- **Symptomatic hyponatraemia**: **3% NaCl 2–5 mL/kg over 10–20 min** (raises Na by ~2–4 mEq/L); repeat till symptoms resolve.
- Correction rate for chronic: **≤ 8–10 mEq/L in 24 h** – faster → **osmotic demyelination (central pontine myelinolysis)**.
- Sodium deficit (mEq) = (desired Na – actual Na) × 0.6 × weight (kg).

	SIADH	Cerebral salt wasting
Volume	Euvolaemic / mild ↑	**Hypovolaemic**
Urine Na	> 20–40	**Very high**
Urine output	Low	**High**
Uric acid	Low	Low
Treatment	**Fluid restriction** (vaptans)	**Saline + Na replacement** (fludrocortisone)

Hypernatraemia (Na > 145–150 mEq/L)

- Causes: **inadequate breastfeeding in neonates** (hypernatraemic dehydration), diarrhoea with hypertonic fluids (improperly made ORS/formula), **diabetes insipidus**, salt poisoning, osmotic diuresis.
- Features: **doughy skin**, irritability, high-pitched cry, hypertonia, seizures; **skin pinch less marked** (ECF preserved) → dehydration underestimated; **intracranial haemorrhage** (shrunken brain tears bridging veins), thrombosis.
- Correction slowly over **48–72 h**, fall **≤ 10–12 mEq/L per 24 h** (≤ 0.5 mEq/L/h) – rapid fall → **cerebral oedema & seizures** (idiogenic osmoles).

4.4 Potassium Disorders

	Hypokalaemia (< 3.5)	Hyperkalaemia (> 5.5)
Causes	Diarrhoea, vomiting/**pyloric stenosis**, diuretics, **DKA treatment (insulin)**, β-agonists (salbutamol), alkalosis, **Bartter/Gitelman**, RTA (type 1, 2), hyperaldosteronism, Liddle, SAM	**Haemolysed sample (pseudo)**, AKI/CKD, **CAH**, Addison, tumour lysis, rhabdomyolysis, acidosis, K-sparing diuretics, ACEi, succinylcholine, massive transfusion
Clinical	Muscle weakness, **ileus**, paralysis, polyuria, arrhythmia	Weakness, paraesthesia, **arrhythmias (VF, asystole)**
ECG	**Flat/inverted T, U waves**, ST depression, prolonged QU	**Tall peaked T (earliest)** → prolonged PR → flat P → **wide QRS** → **sine wave** → VF
Treatment	Oral KCl; IV KCl **max 0.5–1 mEq/kg/h, concentration ≤ 40 mEq/L peripheral (≤ 80 central)**; never IV push	See below

TREATMENT OF HYPERKALAEMIA – 'C BIG K DROP'

1. **Stabilise myocardium – 10% calcium gluconate 0.5–1 mL/kg IV over 5–10 min** (first step if ECG changes; acts in minutes, doesn't lower K).
2. **Shift K into cells** – **insulin + glucose** (0.1 U/kg insulin with 0.5 g/kg glucose), **nebulised salbutamol**, sodium bicarbonate (if acidotic).
3. **Remove K** – **Kayexalate (sodium polystyrene sulfonate)** 1 g/kg, loop diuretics, **dialysis** (definitive).

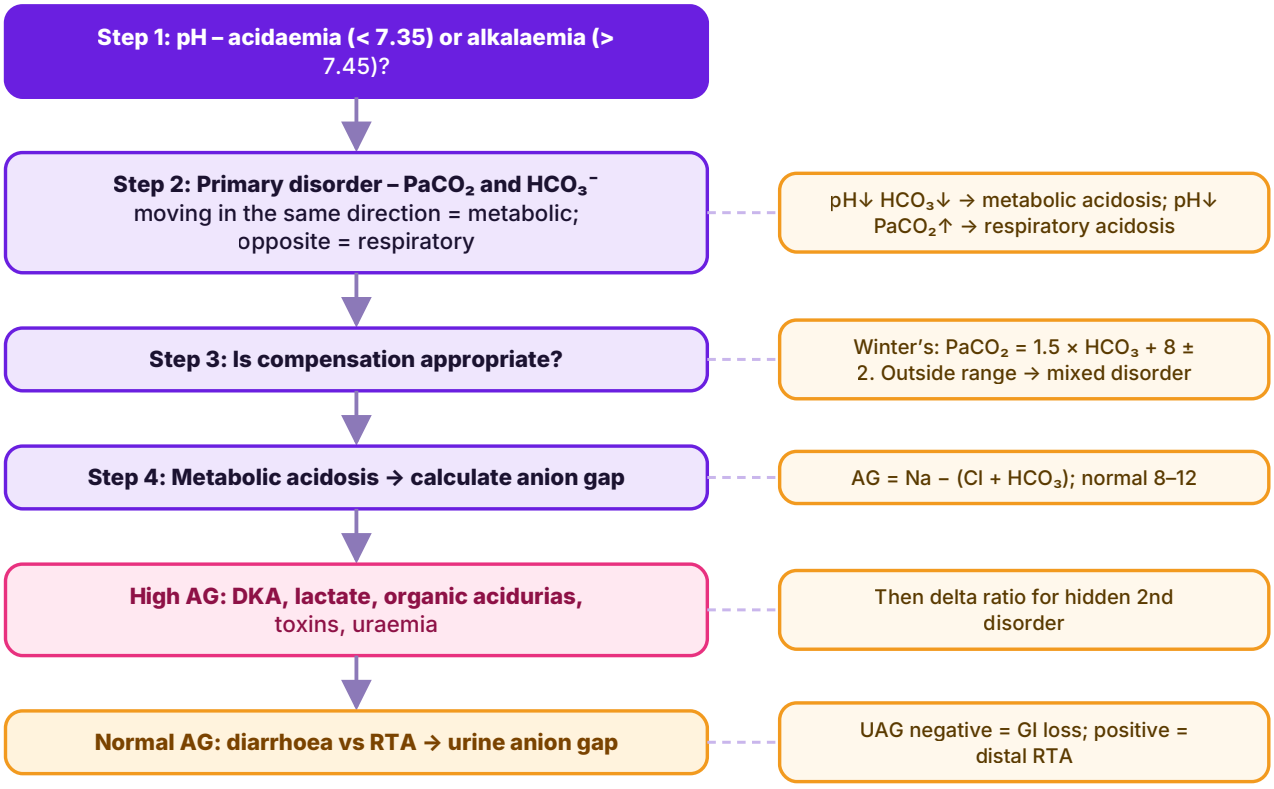
4.5 Calcium & Magnesium

- Normal serum Ca 8.8–10.8 mg/dL (neonate 7–11); **ionised Ca 4.4–5.4 mg/dL** (1.1–1.35 mmol/L). Correct total Ca for albumin: +0.8 mg/dL per 1 g/dL fall in albumin below 4.

Hypocalcaemia (total < 8 / neonate < 7, ionised < 4)	Hypercalcaemia (> 11)
Early neonatal (< 72 h): prematurity, **infant of diabetic mother**, birth asphyxia, IUGR, maternal hyperparathyroidism	**Williams syndrome**, idiopathic infantile hypercalcaemia (CYP24A1)
Late neonatal (5–10 days): **high-phosphate cow's milk feeding**, hypomagnesaemia, **hypoparathyroidism (DiGeorge)**, maternal vitamin D deficiency	Hypervitaminosis D/A, immobilisation, malignancy, **subcutaneous fat necrosis** (newborn), hyperparathyroidism, familial hypocalciuric hypercalcaemia
Older child: vitamin D deficiency (rickets), hypoparathyroidism, **pseudohypoparathyroidism**, CKD, pancreatitis, **tumour lysis**	
Signs: jitteriness, seizures, **Chvostek & Trousseau**, **laryngospasm, carpopedal spasm**, **prolonged QTc**	Polyuria, vomiting, constipation, nephrocalcinosis, **short QT**
Rx: **10% Ca gluconate 1–2 mL/kg IV slowly** (dilute, monitor HR, avoid extravasation)	Rx: hydration + furosemide, steroids, bisphosphonates, calcitonin

- **Hypomagnesaemia** (< 1.5 mg/dL) → **refractory hypocalcaemia** (↓ PTH secretion & action); Rx **50% $MgSO_4$ 0.1–0.2 mL/kg IM/IV**. Causes: IDM, Gitelman, malabsorption, aminoglycosides, amphotericin, cisplatin, PPI.

4.6 Acid–Base Disorders



Stepwise ABG interpretation

Disorder	Primary change	Expected compensation
Metabolic acidosis	↓ HCO_3^-	**Winter's formula: expected $PaCO_2 = 1.5 \times HCO_3^- + 8$ (± 2)**
Metabolic alkalosis	↑ HCO_3^-	$PaCO_2$ ↑ 0.7 mmHg per 1 mEq/L ↑ HCO_3^-
Acute respiratory acidosis	↑ $PaCO_2$	HCO_3^- ↑ 1 per 10 mmHg ↑ $PaCO_2$
Chronic respiratory acidosis	↑ $PaCO_2$	HCO_3^- ↑ 3.5 per 10 mmHg
Acute respiratory alkalosis	↓ $PaCO_2$	HCO_3^- ↓ 2 per 10 mmHg
Chronic respiratory alkalosis	↓ $PaCO_2$	HCO_3^- ↓ 4–5 per 10 mmHg

- Normal: **pH 7.35–7.45, $PaCO_2$ 35–45 mmHg, HCO_3^- 22–26 mEq/L**; anion gap = Na – (Cl + HCO_3) = **8–12 (± 4)**; correct AG for albumin (+2.5 per 1 g/dL fall).
- **Delta ratio** ($\Delta AG/\Delta HCO_3$): < 1 → coexisting normal-AG acidosis; > 2 → coexisting metabolic alkalosis.

Metabolic acidosis

High anion gap – "MUDPILES/ GOLDMARK"	Normal AG (hyperchloraemic)
Methanol, Uraemia, **DKA**, Propylene glycol/paraldehyde, Iron/INH, **Lactic acidosis** (shock, sepsis, asphyxia, mitochondrial), Ethylene glycol, **Salicylates**; **inborn errors – organic acidurias** (MMA, propionic, isovaleric, MSUD)	**Diarrhoea (MCC in children)**, **renal tubular acidosis**, ureterosigmoidostomy, carbonic anhydrase inhibitors (acetazolamide), excess NS infusion, early CKD

- **Urinary anion gap** (UNa + UK − UCl): **negative** in diarrhoea (↑ NH_4^+ excretion), **positive** in distal RTA.
- Bicarbonate therapy: only for **severe acidosis (pH < 7.1–7.15)** with normal-AG losses; generally **not** in DKA or lactic acidosis (treat the cause). HCO_3 deficit = 0.3 × weight × base deficit.

Metabolic alkalosis

Chloride-responsive (urine Cl < 10–20)	Chloride-resistant (urine Cl > 20)
Vomiting – hypertrophic pyloric stenosis (hypochloraemic, hypokalaemic metabolic alkalosis with paradoxical aciduria), NG suction, **cystic fibrosis** (sweat Cl loss), congenital chloride diarrhoea, post-hypercapnia, diuretics (remote)	**Bartter**, **Gitelman**, Liddle, primary hyperaldosteronism, Cushing, 11β-hydroxylase/ 17α-hydroxylase CAH, licorice, current diuretic use
Rx: **0.9% NaCl + KCl**	Rx: treat cause, K-sparing diuretics

PYLORIC STENOSIS BIOCHEMISTRY

Loss of HCl → **hypochloraemic metabolic alkalosis**; volume depletion → aldosterone ↑ → K^+ loss (**hypokalaemia**) and, when K^+ is depleted, H^+ secreted in exchange for Na^+ → **paradoxical aciduria**. Correct fluids/electrolytes (**NS + KCl**) **before** Ramstedt pyloromyotomy (surgery is never an emergency).

Respiratory disorders

- **Respiratory acidosis**: hypoventilation – airway obstruction, severe asthma (late, ominous normal/high $PaCO_2$), pneumonia, neuromuscular disease (GBS, SMA, DMD), CNS depression (opioids, benzodiazepines), BPD.
- **Respiratory alkalosis**: hyperventilation – anxiety, fever, early salicylate poisoning (**mixed: respiratory alkalosis + high-AG metabolic acidosis**), hypoxaemia, hepatic encephalopathy, sepsis, mechanical over-ventilation.

4.7 Bartter vs Gitelman

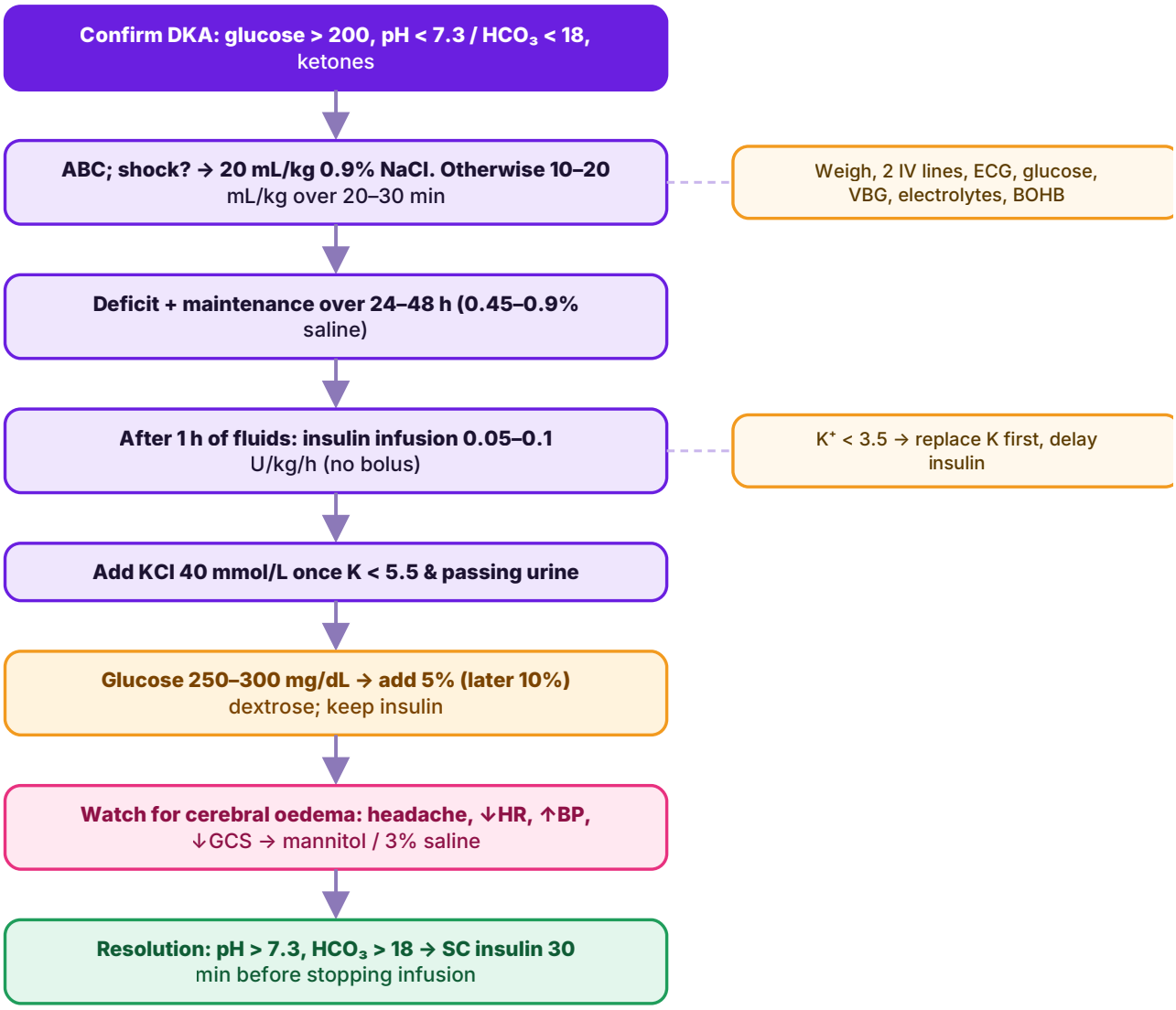
	Bartter	Gitelman
Defect	Thick ascending limb (NKCC2, ROMK, ClC-Kb, barttin) – **like loop diuretic**	Distal convoluted tubule (**NCC thiazide-sensitive** – SLC12A3) – **like thiazide**
Age	Neonatal/infancy (antenatal **polyhydramnios**, prematurity)	Older child/adolescent
Features	Polyuria, failure to thrive, dehydration	Tetany, weakness, cramps; milder
Blood	**Hypokalaemic metabolic alkalosis**, normal BP, high renin & aldosterone	Same + **hypomagnesaemia**
Urine Ca	**High (hypercalciuria, nephrocalcinosis)**	**Low (hypocalciuria)**
Rx	Indomethacin, K^+, spironolactone	Mg & K supplements

- **Liddle syndrome**: ENaC gain-of-function → **hypertension**, hypokalaemic alkalosis, **low renin & low aldosterone**; Rx **amiloride/triamterene**.
- **Pseudohypoaldosteronism type 1**: aldosterone resistance → salt wasting, **hyperkalaemia, metabolic acidosis**, high renin & aldosterone.

4.8 Diabetic Ketoacidosis (ISPAD 2022)

Diagnosis: blood glucose **> 200 mg/dL (11 mmol/L)** + **pH < 7.3 or HCO_3^- < 18 mmol/L** + **ketonaemia (β-hydroxybutyrate ≥ 3 mmol/L)** or moderate/large ketonuria.

Severity	pH	HCO_3^-
Mild	< 7.3	< 18
Moderate	< 7.2	< 10
Severe	**< 7.1**	**< 5**



DKA management in children (ISPAD 2022 principles)

KEY STEPS

1. **Fluids first**: shock → 20 mL/kg 0.9% NaCl bolus; otherwise **10–20 mL/kg NS over 20–30 min**, then deficit (assume 5–10% dehydration) + maintenance **over 24–48 h** with 0.45–0.9% saline.
2. **Insulin**: regular insulin infusion **0.05–0.1 U/kg/h**, started **1–2 h after fluids**; **no insulin bolus**.
3. **Potassium**: add **40 mmol/L KCl** once K < 5.5 and urine output confirmed (start immediately if hypokalaemic; delay insulin if K < 3.5).
4. **Glucose**: add 5% dextrose when blood glucose falls to **250–300 mg/dL**; target fall 50–100 mg/dL/h.
5. **Bicarbonate – not recommended** (↑ risk of cerebral oedema) except life-threatening hyperkalaemia/ cardiac compromise.
6. Monitor: hourly glucose & neuro obs; electrolytes, VBG every 2–4 h.

CEREBRAL OEDEMA – MOST COMMON CAUSE OF DEATH IN PAEDIATRIC DKA

Occurs **4–12 h after starting treatment**. Risk: **young age, new-onset diabetes, severe acidosis, low $PaCO_2$, high BUN, bicarbonate use, failure of Na to rise as glucose falls**, rapid fluids. Signs: headache, **bradycardia + hypertension**, falling GCS, irritability, recurrence of vomiting. Rx: **mannitol 0.5–1 g/kg IV over 10–15 min or 3% saline 2.5–5 mL/kg**, head end up 30°, reduce fluids.

- **Corrected sodium** = measured Na + 1.6 × (glucose − 100)/100; should **rise** as glucose falls.
- Pseudo-normokalaemia: total body K is always depleted despite normal/high serum K.

Practice MCQs · Fluid and Electrolyte Disturbances

104 questions · correct option highlighted in green · cover the answer strip to self-test

Body Water & Fluid Compartments

1. At what age extracellular fluid is equal to the inter cellular fluid?

A. 14 days
B. **4 weeks**
C. 2 months
D. 3 months

Ans B • In fetus & newborn, ECF volume is larger than ICF volume.
• Normal postnatal diuresis causes an immediate decrease in the ECF volume.
• This is followed by continued expansion of ICF volume, which results from cellular growth.
• By 1 year of age, the ratio of the ICF to ECF volume approaches adult levels.

2. Total body water at birth is what proportion of body weight?

A. 60%
B. 65%
C. 70%
D. **75%**

Ans D • Total body water (TBW) constitutes 75% of the body weight at birth
• During the 1st year of life, TBW decreases to 60% of body weight & remains at this level until puberty
• By the end of puberty TBW in males remains at 60%, but TBW in females decreases to 50% of body weight

3. Parklands formula used for Burn's resuscitation is:

A. **4 mL/kg/% TBSA**
B. 5 mL/kg/%TBSA
C. 6 mL/kg/%TBSA
D. 8 mL/kg/% TBSA

Ans A • For most children with burns, the Parkland formula is an appropriate starting guideline for fluid resuscitation.
• According to it, 4 mL lactated Ringer solution/kg/% BSA burned is used
• Half of the fluid is given over 1st 8 hr, calculated from the time of onset of injury; remaining over next 16 hours.

4. The ratio of the intracellular fluid volume to the extracellular fluid volume approaches adult levels at the age of

A. **1 yr**
B. 2 yr
C. 3 yr
D. 4 yr
E. 5 yr

Ans A

5. The MOST common cause of hyperphosphatemia is

A. acute hemolysis
B. vitamin D intoxication
C. **renal insufficiency**
D. cow's milk intake
E. hypoparathyroidism

Ans C

6. For each 1°C increase in temperature above 38°C leads to increase in maintenance water needs by

A. 5-10%
B. **10-15%**
C. 15-20%
D. 20-25%
E. 25-30%

Ans B

Dehydration & Rehydration

7. Which one of these is a 3rd space fluid loss?

A. Fluid loss through a stoma
B. **Tissue edema**
C. Urinary losses
D. Ryle's tube aspirate ACID BASE BALANCE

Ans B "Third spacing" Refers to fluid loss into interstitial spaces, caused by increases in vascular permeability and/or a reduction in oncotic pressure (hypoalbuminemia) "
• This occurs in sepsis syndrome, burns, pancreatitis, nutritional hypoalbuminemia, and peritonitis
• Third-space losses are isotonic & can cause significant volume depletion, leading to ↑ADH production and water retention, which can cause hyponatremia if the patient receives hypotonic fluid.

8. Hypernatremic dehydration in children leads to irrita bility, restlessness, weakness, lethargy and fever. What is the most devastating consequence of hypern atremia in children?

A. Seizures
B. Hyperglycemia
C. **Brain hemorrhage**
D. Hypocalcemia

Ans C • Brain hemorrhage is the most devastating consequence of untreated hypernatremia. dna • Seizures & coma are possible sequelae of hemorrhage, although seizures are more common during correction of hypernatremia. Mechanism of Intracranial hemorrhage in hypernatremia: Due to hypernatremia, extracellular osmolality increases ↓ Water moves out of brain cells, leading to a decrease in brain volume ↓ Tearing of intracerebral veins and bridging blood vessels as the brain moves away from skull and meninges fo ↓

9. Hypernatremic dehydration is characterized by:

A. S. Sodium > 150 mmol/L
B. Signs of dehydration are minimal
C. ECF volume increased
D. Rapid correction is required
E. Shift of water from ECF to ICF

Ans A and b. Signs of dehydration are minimal Discussing about the options one by one Answers a. True It is the definition of hypernatremia b. True In hypernatremic dehydration, shift of water from intracellular space to extracellular space → blood pressure and urine output maintained → minimal signs of dehydration c. False ECF volume usually remains normal d. False Rapid correction of hypernatremia is harmful; The goal is to decrease the serum sodium by < 12 mEq/L every 24 with hours, a rate of 0.5 mEq/L/hour e.

10. Concentration of K+ (mEq/L) in most commonly used ORS is:

A. 10
B. 20
C. 65
D. 75

Ans B

11. The total osmolality of lowosmolality WHO ORS is?

A. 200 mOsm/L
B. 245 mOsm/L
C. 305 mOsm/L
D. 311 mOsm/L

Ans B

12. Dosage of intravenous fluid for 2 months old child in dia rrhea with severe dehydration:

A. 100 mL/kg in 6 hours
B. 50 mL/kg in 6 hours
C. 75 mL/kg in 6 hours
D. 80 mL/kg in 6 hours

Ans A Treatment of severe dehydration in children: Age First give Then give Total treatment < 12 months 30 mL/kg in 1 hour 70 mL/kg in 5 hours 100 mL/kg in 6 hours 1–5 years 30 mL/kg in 30 minutes 70 mL/kg in 2½ hours 100 mL/kg in 3 hours

13. Low osmolar ORS composition:

A. Na 90 + 311 mOsm/L
B. Na 75 + 245 mOsm/L
C. Na 60 + 245 mOsm/L
D. Na 60 + 240 mOsm/L

Ans B Na 75 + 245 mOsm/L see ans 26 below

14. A 5yearchild comes with no dehydration. Plan A was proposed for treatment of dehydration. How much ORS should be given per loose stool?

A. 0–50 mL
B. 50–100 mL
C. 100–200 mL
D. 200–300 mL

Ans C Plan A: For Patients without physical signs of dehydration, amount of ORS to be given after each loose stool: Age < 24 months 2–10 years > 10 years Amount of ORS 50–100 mL 100–200 mL As much as wants

15. Molar ratio of Sodium and Glucose in WHO ORS is:

A. 1 :
B. 1 :
C. 1 :
D. 2 :

Ans A 1 Molarity of sodium and glucose in ORS should be 1:1, which helps in the efficient transport of sodium and water.

16. Sodium content in mmol/L in WHO oral rehydration solution is:

A. 20
B. 80
C. 90
D. 111

Ans B 80 WHO ORS contains 75 mmol/L sodium; 80 is the closest option to it; So it is the best answer amongst the given options.

17. Amount of ORS given in a 4 kg child with diarrhea with some dehydration:

A. 200 mL
B. 300 mL
C. 400 mL
D. 800 mL

Ans A 300 mL 75 mL/kg ORS is to be given in some dehydration, i.e. 75 × 4 = 300 mL in this case.

18. What is the amount of trisodium citrate in WHO ORS?

A. 1.5 gm
B. 2.5 gm (MAHA PG 11)
C. 2.9 gm
D. 3.9 gm

Ans C 2.9 g

19. What is the correct composition (per liter) of WHO reduced osmolarity ORS?

A. NaCl 2.29 g; KCl 1.1 g; Glucose 13.1 g; Citrate 2.5 g
B. NaCl 2.49 g; KCl 1.3 g; Glucose 13.3 g; Citrate 2.7 g
C. NaCl 2.69 g; KCl 1.5 g; Glucose 13.5 g; Citrate 2.9 g
D. NaCl 2.89 g; KCl 1.7 g; Glucose 13.7 g; Citrate 3.1 g

Ans C

20. Concentration of K+ in ORS is _____ mEq/L

A. 20
B. 40
C. 90
D. 10 INTRAVENOUS FLUID ADMINISTRATION

Ans A

21. What percentage of weight loss in children constitutes severe dehydration?

A. >4% loss of body weight
B. >5% loss of body weight
C. 5% loss of body weight
D. 7% loss of body weight

Ans D Severe dehydration is defined as weight loss of >10% of body in an infant; >6% in an older child.

22. A child with acute diarrhea with signs of dehydration, Na+ 125, K+3, HCO– 16, pH 7.23; IV Fluid to be given is: 3

A. Normal saline
B. 3% saline
C. N/3 saline + 10% dextrose
D. N/3 saline + 5% dextrose

Ans A In the given scenario, the child has dehydration with low sodium, along with that metabolic acidosis, which is most probably secondary to hypovolemia; So, the fluid of choice in this case is an isotonic fluid to correct dehydration metabolic acidosis; So normal saline is the best answer.

23. A 2yearold child presents with loose stools. On examination she has signs of severe dehydration. If her weight is 10 kg, what would be the volume of fluid to be given in the first 4 hours?

A. 1000 mL
B. 100 mL
C. 200 mL
D. 500 mL

Ans A In a 2yearold child with severe dehydration, fluid given over 1st 30 minutes is 30 mL/kg over next 2 and 1/2 hour is 70 mL/kg; So with fluid given over 3 hours is 100 mL/kg. In this case, as weight of child is 10 kg, amount of fluid to be given is 10 x 100 = 1000 mL.

24. Rehydration therapy in a 2yearold child with diarrhea with some dehydration:

A. 30 mL/kg in 1 hour – 70 mL in 5 hours
B. 30 mL/kg in 30 mm – 70 mL/kg in 2½ hours
C. 20 mL/kg in 30 mm – 50 mL/kg in 2½ hours
D. 75 mL/kg in 4 hours

Ans D

25. When severe dehydration in a neonate occurs, amount of fluid replacement in 1st hour:

A. 20–40 mL/kg
B. 5–10 mL/kg
C. 10–15 mL/kg
D. 15–20 mL/kg

Ans A In a child < 1 year old, with severe dehydration, Fluid given over 1st hour is 30 mL/kg.

26. In malnourished children the following parameters are reliable indicators of dehydration except:

A. Skin turgor
B. Dry buccal mucosa
C. Oliguria
D. Thirst

Ans A Skin turgor is not a reliable parameter for assessing dehydration in a child with malnutrition, as it may appear to be decreased due to decreased subcutaneous fat.

27. Following pathogenetic mechanisms operate in septic shock except?

A. Increased peripheral vascular resistance
B. Venoconstriction
C. Direct toxic endothelial injury
D. Activation of complement

Ans A In Septic shock in children, there is decreased systemic vascular resistance, due to peripheral vasodilatation;

28. Septic Shock is due to:

A. Vasodilatation
B. Decreased cardiac output
C. Endothelial damage
D. All of the above

Ans D

29. Rehydration therapy in a 2yearold severely dehydrated child is:

A. 30 mL/kg in 1 hour, 70 mL in 5 hours
B. 30 mL/kg in 30 minutes, 70 mL/kg in 2½ hours
C. 20 mL/kg in 30 minutes, 80 mL/kg in 2½ hours
D. 75 mL/kg in 4 hours

Ans B • Intraosseous (IO) cannulation is recommended for patients for whom IV access proves difficult or unattainable
• If venous access is not available within 1 minute in a child with cardiopulmonary arrest, an IO needle should be placed in anterior proximal tibia

30. A child presents with hypovolemic shock, anuria and respiratory acidosis, treatment is:

A. RL
B. Dopamine
C. Dobutamine
D. Inj. Sodium Bicarbonate

Ans A RL Initial resuscitation of any child with shock consists of Pushing IV fluid boluses (RL or NS) of 20 mL/kg up to 60 mL/kg until perfusion improves or unless rales or hepatomegaly develop; Next step is use of Inotropes like Dopamine.

31. In a child with hypernatremic dehydration, the first priority is restoration of intravascular volume by

A. 3% saline
B. normal saline
C. lactated Ringer solution
D. 1/3 glucose saline
E. 1/2 glucose saline

Ans B Repeated boluses of normal saline (10-20 mL/kg) may be required to treat hypotension, tachycardia, and signs of poor perfusion (poor peripheral pulses and capillary refill time).

32. The best fluid bolus giving to a child with isolated vomiting and severe dehydration is

A. normal saline
B. ringer lactate
C. half-normal saline
D. hypertonic (3%) saline
E. 5% dextrose + half-normal saline

Ans A Ringer lactate should not be used because the lactate would worsen the alkalosis.

33. The MOST common manifestation of cerebral edema from an overly rapid decrease of serum sodium concentration during correction of hypernatremic dehydration is

A. irritability
B. hyperreflexia
C. spastisity
D. seizure
E. coma

Ans D

Hyponatremia & Hypernatremia

34. A child with diarrhea has deep & rapid respiration. Most likely diagnosis is:

A. Respiratory acidosis
B. Respiratory alkalosis
C. Metabolic acidosis
D. Metabolic alkalosis

Ans C

35. A breastfed baby presents with hypernatremia (S. Sodium >170 mEq/L). His urine sodium is 70 mEq/L. Which of the following is the most likely cause?

A. Diabetes insipidus
B. Acute tubular necrosis
C. Severe dehydration
D. Excessive intake of sodium

Ans D This is a controversial question. Discussing about the options one by one, a. Diabetes insipidus → serum sodium is usually high and urinary sodium low b. Acute tubular necrosis → It is a cause of intrinsic ARF in which though urinary sodium is high with a low urine osmolality but Serum Sodium is usually low c. Severe Dehydration → It represents a prerenal state with a low urine sodium d. Excessive intake of sodium can lead to high serum sodium in blood and urine

36. Sodium content of ReSoMal is:

A. 90 mmoL/L
B. 60 mmoL/L
C. 45 mmoL/L
D. 30 mmoL/L

Ans C iter ReSoMAL (Rehydration solution for severely Malnourished child): dna As severely malnourished children are deficient in potassium and have relatively high levels of sodium, ReSoMal has a lower sodium concentration and higher potassium concentration to meet the electrolyte needs of these children. ReSoMal Low Osmolarity New Composition of WHO ORS (Low osmolarity) Component (mEq/L) WHO ORS (mEq/L) Name of the constituent Amount in gm Glucose 125 75 Sodium 45 75 Sodium chloride 2.6 Potassium 40 20

37. Na+ content of RL in mmol/L:

A. 120
B. 125
C. 130
D. 135

Ans C

38. Which of the following would be plasma osmolality of child with plasma Na+ 125 mEq/L, glucose of 108 mg/dL, and blood urea Nitrogen (BUN) of 140 mg/dL?

A. 360 mOsm/kg
B. 306 mOsm/kg
C. 312 mOsm/kg
D. 318 mOsm/kg

Ans B Formula for calculating Plasma Osmolality = 2 x (Na+) + glucose/ 18 + BUN/2.8. So, in this child, plasma osmolality = 2 x 125 + 108/18 + 140/2.8 = 306 mOsm/L.

39. The critical site for the renal regulation of sodium balance is the

A. nephron
B. proximal tubule
C. loop of Henle
D. distal tubule
E. collecting duct

Ans E Even though the amount of sodium resorbed in this segment is less than in any other segment.

40. More than 40% of total body sodium is in the

A. intravascular spaces
B. interstitial spaces
C. intracellular fluid
D. bone
E. gastrointestinal fluid

Ans D

41. The MOST devastating consequence of untreated hypernatremia is

A. brain hemorrhage
B. seizures
C. central pontinemyelinolysis
D. extra pontinemyelinolysis
E. brain edema

Ans A As the extracellular osmolality increases, water moves out of brain cells, leading to a decrease in brain volume, this decrease can result in tearing of intra cerebral veins and bridging blood vessels as the brain moves away from the skull and the meninges, patients may have subarachnoid, subdural, and parenchymal hemorrhages. Seizures and brain edema are more common during correction of hypernatremia.

42. Pseudohyponatremia is present in all the following EXCEPT

A. multiple myeloma
B. immunoglobulin infusion
C. protein losing enteropathy
D. hypertriglyceridemia
E. hypercholesterolemia

Ans C Hypoalbuminemia caused by gastrointestinal disease (protein losing enteropathy) cause hypervolemichyponatremia, while pseudohyponatremia is a laboratory artifact that is present when the plasma contains very high concentrations of protein (multiple myeloma, intravenous immunoglobulin infusion) or lipid.

43. Hypervolemic hyponatremia is caused by

A. cirrhosis
B. cerebral salt wasting
C. pseudohypoaldosteronism type I
D. obstructive uropathy
E. bowel obstruction

Ans A Other distracters are causes of hypovolemic hyponatremia.

44. **Asymptomatic hyponatremia is seen in**

A. cirrhosis
B. tap water enema
C. child abuse
D. **hyperglycemia**
E. tubule -interstitial nephritis

Ans D Because the manifestations of hyponatremia are a result of the low plasma osmolality, patients with hyponatremia resulting from hyperosmolality do not have symptoms of hyponatremia.

Hypokalemia & Hyperkalemia

45. **All of the following are used in the treatment of hyperkalemia except:**

A. Calcium gluconate
B. **Hypertonic saline**
C. Sodium bicarbonate
D. Insulin

Ans B Drugs used in the treatment of acute hyperkalemia include: Calcium gluconate, Sodium bicarbonate, Insulin infusions & salbutamol nebulisation

46. **The clinical consequences of hypokalemia in skeletal muscle include muscle weakness and cramps. What is the level of Serum Potassium at which Paralysis Is a possible complication of hypokalemia?**

A. **Serum Potassium at 2.0 mEq/L**
B. Serum Potassium at 2.5 mEq/L
C. Serum Potassium at 3.0 mEq/L
D. Serum Potassium at 3.5 mEq/L

Ans A Paralysis is a possible complication, generally only at potassium levels <2.5 mEq/L. It usually starts in legs and moves to arms;

47. **Immediate treatment for hyperkalemia + bradycardia is?**

A. Sodium bicarbonate
B. **Ca gluconate**
C. Salbutamol
D. Kayexalate

Ans B IV calcium gluconate (10%) is used in treatment of Severe hyperkalemia (K+ > 8 mEq/L), to reverse the cardiac effects of hyperkalemia immediately.

48. **Which of the following inherited disorders is a Potassium Channelopathy?**

A. Hyperkalemic periodic paralysis
B. Hypokalemic periodic paralysis
C. Beckers's disease
D. **Anderson Tawil syndrome**

Ans D Channelopathies are progressive syndromic cluster of symptoms not localizing to single lesion; Important Channelopathies: (All of these are autosomal dominant & show episodic weakness) Calcium Channel Sodium Channel Potassium Channel Feature Hypokalemic PP Hypokalemic PP Paramyotonia Congenita Andersen-Tawil syndrome Age of onset Adolescence Early childhood Early childhood Early childhood Myotonia No Yes Yes No Frequency of Daily to yearly May be 2-3/day With cold, usually rare Daily to yearly attacks of weakness

49. **Requirement of potassium in child is:**

A. **Blood glucose**
B. Serum protein
C. Serum bilirubin
D. Oxygen saturation

Ans A 1–2 mEq/kg • Daily requirement of potassium in a child is 1–2 mEq/kg/day and sodium is 3 mEq/kg/day.

50. **Spurious hyperkalemia or pseudohyperkalemia in children is usually exaggerated by**

A. blood transfusions
B. digitalis intoxication
C. **thrombocytosis**
D. renal failure
E. malignant hyperthermia

Ans C This phenomenon is exaggerated with thrombocytosis because of potassium release from platelets, for every 100,000/m3 increase in the platelet count; the serum potassium level rises by approximately 0.15 mEq/L.

51. **The MOST common cause of hypokalemia in children is**

A. alkalemia
B. distal rental tubular acidosis
C. **gastroenteritis**
D. diabetic ketoacidosis
E. loop diuretic

Ans C

52. **Spurious hypokalemia occurs in patients with**

A. laxative abuse
B. **high white blood cell count**
C. hypomagnesaemia
D. posthypercapnia
E. Cushing syndrome

Ans B Spurious hypokalemia occurs in patients with leukemia and very elevated white blood cell counts if sample for analysis is left at room temperature, permitting the white blood cells to take up potassium from the plasma.

53. **The mechanism of hypokalemia in emesis is mainly due to**

A. gastric loss of potassium
B. **gastric loss of hydrochloric acid**
C. low aldosterone level
D. dehydration
E. hypomagnesaemia

Ans B The gastric loss of hydrochloric acid (HCl), leading to a metabolic alkalosis and a state of volume depletion. The kidney compensates for the metabolic alkalosis by excreting bicarbonate in the urine, but there is obligate loss of potassium and sodium with the bicarbonate. The volume depletion raises aldosterone levels, further increasing urinary potassium losses and preventing correction of the metabolic alkalosis and hypokalemia until the volume depletion is corrected.

54. Renal manifestations of hypokalemia include all the following EXCEPT

A. polyuria
B. urinary retention
C. decrease ammonia production
D. interstitial nephritis
E. renal cysts

Ans C Hypokalemia stimulates renal ammonia production, an effect that is clinically significant if hepatic failure is present, because the liver cannot metabolize the ammonia. Hypokalemia impairs bladder function, potentially leading to urinary retention.

Hypocalcemia & Hypercalcemia

55. Diagnostic criteria for Syndrome of Inappropriate Antidiuretic hormone secretion include all except:

A. Urine Osmolality >100 mOsm/kg (Usually >Plasma)
B. Serum Osmolality <280 mOsm/kg and serum sodium <135 mEq/L
C. Urine Sodium <30 mEq/L
D. Reversal of "Sodium Wasting" and correction of hypona tremia with water restriction

Ans C In SIADH, Urine sodium should be more than 30 mEq/L

56. Green IV cannula is:

A. 16G
B. 18G
C. 20G
D. 22 G

Ans B

57. Daily water requirement in child weighing 30 kg, height 123 cm and BSA of 1 m2 is:

A. 1300 mL
B. 1700 mL
C. 2000 mL
D. 2500 mL

Ans B Fluid requirement in a child in this child weighing 30 kg is: For first 10 kg (1–10 kg) 100 × 10 = 1000 mL For next 10 kg (11–20 kg) 50 × 10 = 500 mL; For next 10 kg (21–30 kg) 20 × 10 = 200 mL; So, total requirement = 1700 mL.

58. Expansion of the intravascular volume and increased intravascular pressure are the main causes of edema in

A. lymphatic obstruction
B. heart failure
C. protein-losing enteropathy
D. nephrotic syndrome
E. sepsis

Ans B There is an increase in venous hydrostatic pressure from expansion of the intravascular volume, which is caused by impaired pumping by the heart, and the increase in venous pressure causes fluid to move from the intravascular space to the interstitial space.

59. Systemic calcification occurs when plasma calcium × plasma phosphorus, both measured in mg/dL is

A. >90
B. >80
C. >70
D. >60
E. >50

Ans C

Acid-Base Disorders

60. All of the following are causes of normal anion gap metabolic acidosis, except:

A. Renal Tubular acidosis
B. Diarrhea
C. Acidosis due to Intestinal bacterial overgrowth
D. Urinary tract diversions

Ans C In patients with intestinal bacterial overgrowth, excessive bacterial metabolism of glucose into dlactic acid can cause a lactic acidosis; so it is a high anion gap metabolic acidosis;

61. High osmolality with high anion gap is seen in:

A. Diarrhea
B. Distal renal tubular acidosis
C. Ammonium chloride intake
D. Urinary tract diversion
E. Methanol poisoning

Ans E In Methanol poisoning, there is high osmolality with high anion gap; In all others given in the options, anion gap is usually normal;

62. High anion gap acidosis is seen in all except:

A. Diarrhea
B. Salicylate poisoning
C. Acute renal failure
D. Lactic acidosis

Ans A

63. An 11yearchild with failure to thrive, metabolic acidosis with increased anion gap. Not true regarding the differential diagnosis:

A. Insulin dependent diabetes mellitus
B. Chronic renal failure
C. Renal tubular acidosis
D. Inborn errors of metabolism

Ans C

64. Widened anion gap is not seen in:

A. Acute renal failure
B. Diarrhea
C. Lactic acidosis
D. Diabetic ketoacidosis DISORDERS OF SODIUM & POTASSIUM

Ans B DISORDERS OF SODIUM & POTASSIUM

65. The predominant buffering system in the blood plasma is:

A. Hemoglobin
B. Plasma proteins
C. K HPO & KH PO
D. **NaHCO & H CO 2 4 2 4 3 2**

Ans D NaHCO & H CO 3 2 3 The bicarbonate buffer system is very effective as a result of the high concentration of bicarbonate in the body (24 mEq/L) and the fact that it is an open system, so lungs increase CO2 excretion when required

66. Normal anion gap metabolic acidosis can occur in

A. **renal failure**
B. liver failure
C. severe anemia
D. malignancy
E. starvation ketoacidosis

Ans A In renal failure, there is retention of unmeasured anions, including phosphate, urate, and sulfate. The increase in unmeasured anions in renal failure is usually less than the decrease in the bicarbonate concentration. Renal failure is thus a mix of an increased gap and a normal gap metabolic acidosis.

67. Metabolic acidosis with increased anion gap occurs in

A. ammonium chloride intake
B. posthypocapnia
C. urinary tract diversions
D. hyporeninemic hypoaldosteronism
E. **severe anemia**

Ans E Because of tissue hypoxia.

Metabolic Acidosis

68. A Normalaniongap metabolic acidosis occurs in patients with?

A. Diabetic ketoacidosis
B. Methyl alcohol poisoning
C. **Diarrhea**
D. Acute kidney injury

Ans C Diarrhea and Renal tubular acidosis (RTA) are two important causes of normal anion gap metabolic acidosis.

69. In case of massive bleeding during surgery which sized cannula is used?

A. **16G**
B. 20G
C. 22G
D. 24G

Ans A During massive bleeding wide bore cannula is to be used. Among the given options, 16G is the widest and so the best answer. Size of IV cannula 14G 16G 18G 20G 22G 24G 26G Color coding Orange Gray Green Pink Blue Yellow Violet

70. Parkland formula is:

A. **Percentage of burn area × body wt ×**
B. Percentage of burn area × body wt/4
C. Percentage of burn area × body surface area ×
D. Percentage of burn area × body surface area/4

Ans A Percentage of burn area × body wt × 4 /e p

71. Syndrome of inappropriate antidiuretic hormone secretion (SIADH) is characterized by

A. extravascular volume expansion
B. high serum uric acid
C. high blood urea nitrogen
D. **euvolemic hyponatremia**
E. urine sodium <30 mEq/L

Ans D Because SIADH is a state of intravascular volume expansion, low serum uric acid and BUN levels are supportive of the diagnosis, the kidney increases sodium excretion in an effort to decrease intravascular volume to normal; thus, the patient has a mild decrease in body sodium(urine sodium >30 mEq/L).

72. All the following phosphorus binders are used in chronic renal insufficiency EXCEPT

A. calcium carbonate
B. calcium acetate
C. sevelamer
D. lanthanum
E. **aluminum**

Ans E Because of the risk of toxicity.

73. The MOST important indication of mechanical ventilation in respiratory acidosis is

A. **Pco2>75 mm Hg**
B. concomitant metabolic acidosis
C. slowly responsive underlying disease
D. hypoxia that responds poorly to oxygen
E. tiring patient

Ans A

Metabolic Alkalosis

74. Tall T wave is seen in?

A. **Hyperkalemia**
B. Hypokalemia
C. Hypercalcemia
D. Hypocalcemia

Ans A

75. In a child who has diarrhea and vomiting with inadequate water intake, which of the following is seen:

A. **Intracellular dehydration with hypernatremia**
B. Intracellular dehydration with hyponatremia
C. Extracellular dehydration with hyponatremia
D. Extracellular dehydration with hypernatremia ORAL REHYDRATION (ORS)

Ans A • Diarrhea results in depletion of both sodium and water; Because diarrhea is hypotonic, with typical sodium concentration of 35 65 meq/L, water losses exceeds sodium losses, potentially leading to hypernatremia
• Most children with gastroenteritis do not have hypernatremia because they drink enough hypotonic fluid to compensate for stool water losses. But in this case, as the baby has inadequate water and fluid intake, he is at risk of developing hypernatremic dehydration. ORAL REHYDRATION (ORS)

76. **A 6yearold drowsy child came in emergency with history of vomiting, loose motion for 3 days. On exami nation he had very sunken eyes, hypothermia, skin on pinching takes time to revert; the diagnosis is: (Recent Question 2013, AIIMS June 2002) 66 Section 1: General Pediatrics**

A. No dehydration
B. Mild dehydration
C. Some dehydration
D. **Severe dehydration**

Ans D Parameters No dehydration Some dehydration Severe dehydration Look at Sensorium Well alert "Restless, irritable" "lethargic", floppy Eyes Normal Sunken Very sunken and dry Tears Present Absent Absent Mouth & Tongue Moist Dry Very dry Thirst Drinks normally, not thirsty "Thirsty, drinks eagerly" "Drinks poorly/ not able to drink" Feel Skin pinch Goes back quickly "Goes back slowly" "Goes back very slowly" Decide No signs of dehydration If the patient has 2 or more signs, including at least 1 "sign"

77. **In a dehydrated child with hypertrophic pyloric stenosis the fluid of choice is:**

A. **Normal saline**
B. 5% dextrose
C. Ringers lactate
D. Darrows solution

Ans A • Surgical patients with pyloric stenosis can develop hypochloremic, hypokalemic, or metabolic alkalosis
• Hydrogen ion reabsorption occurs with an accompanied potassium ion excretion.
• The resulting hypokalemia leads to the excretion of hydrogen ions in the face of alkalosis, a paradoxic aciduria
• Treatment includes replacement of volume deficit with isotonic saline & potassium once adequate urine output is ensured.

78. **Kallu, a 2yearold child weighing 6.7 kg presents in the casualty with h/o vomiting and diarrhea for last 2 days. On examination, skin pinch over the anterior abdominal wall goes quickly to its original position. Interpretation of skin pinch test in this child will be:**

A. No dehydration
B. Some dehydration
C. Severe dehydration
D. **Skin pinch cannot be evaluated in this child SHOCK**

Ans D Skin pinch cannot be evaluated in this child The given child in question has a weight of 6.7 kg at 2 years, against expected of 12 kg (2 × 2 + 8 =12). So this child is malnourished; For assessment of dehydration in a malnourished child, skin pinch is less useful. SHOCK

79. **An alert 6monthold child is brought with vomiting and diarrhea. RR45/min, HR130/min, SBP85 mm Hg. Capillary refilling time is 4 seconds, Diagnosis is**

A. **Early compensated hypovolemic shock**
B. Early decompensated hypovolemic shock
C. Late compensated hypovolemic shock
D. Late decompensated shock due to SVT

Ans A This is a case of hypovolemia as h/o vomiting and diarrhea is present. In this case the infant has poor peripheral perfusion (shock) as evident from a prolonged capillary refill time, but BP is normal, so it is a compensated shock. Hypotension is defined as a systemic blood pressure less than the 5th percentile of normal for age, as given below:
• < 60 mm Hg in term neonates (0–28 days) • < 70 mm Hg + (2 x age in years) in children 1–10 years
• < 70 mm Hg in infants (1–12 months) • < 90 mm Hg for >10 years of age.

80. **Metabolic alkalosis with urinary chloride >20 meq/l occurs in**

A. loop diuretics
B. cystic fibrosis
C. **Cushing syndrome**
D. emesis
E. post-hypercapnia

Ans C Other distracters are causes of metabolic alkalosis and urinary chloride less than 15 meq/L.

81. **In metabolic alkalosis secondary to gastric loss, all the following mechanisms prevent renal bicarbonate loss EXCEPT**

A. **mineralocorticoid escape phenomenon**
B. reduce GFR
C. sodium and bicarbonate resorption
D. increase aldosterone level
E. hypokalemia

Ans A

82. **Measuring serum concentrations of renin and aldosterone differentiates children with metabolic alkalosis; both renin and aldosterone are elevated in**

A. **renovascular disease**
B. glucocorticoid-remediable aldosteronism
C. Liddle syndrome
D. Cushing syndrome
E. 11β-hydroxylase deficiency

Ans A Aldosterone is high and renin is low in patients with adrenal adenomas or hyperplasia and glucocorticoid-remediable aldosteronism. Renin and aldosterone are low in children with Cushing syndrome, Liddle syndrome, licorice ingestion, 17α-hydroxylase deficiency, 11β-hydroxylase deficiency, and 11β-hydroxysteroid dehydrogenase deficiency.

Respiratory Acidosis & Alkalosis

83. **Metabolic compensation in a child with respiratory acidosis takes how much time?**

A. < 1 day
B. 1–2 days
C. **3–4 days**
D. More than 7 days

Ans C 34 days Unlike respiratory compensation, which occurs rapidly, it takes 3–4 days for the kidneys to complete appropriate metabolic compensation.

84. **Respiratory acidosis is characterized by primary:**

A. Deficit of carbonic acid
B. **Excess of carbonic acid**
C. Deficit of bicarbonate
D. Excess of bicarbonate

Ans B Respiratory acidosis is caused by CO retention → excess carbonic acid.

85. **A 5yearold boy passed 18 loose stools in last 24 hours and vomited twice in last 4 hours he is irritable but drinking fluids. The optimal therapy for this child is:**

A. Intravenous fluids
B. **Oral rehydration therapy**
C. Intravenous fluid initially for 4 hours followed by oral fluids
D. Plain water

Ans B The child is irritable but drinking fluids. He thus falls in category of some dehydration and needs to be managed with Plan B. Treatment plan B is oral Rehydration therapy and hence the answer here.

86. **Medications that cause renal losses of magnesium include all the following EXCEPT**

A. amphotericin
B. cisplatin
C. **lithium**
D. proton pump inhibitors
E. mannitol

Ans C Mild hypermagnesemia may occur in lithium ingestion.

87. **The time for the kidneys to complete appropriate metabolic compensation for a primary respiratory process is**

A. 12-24 hr
B. 24-36 hr
C. 36-48 hr
D. 48-72 hr
E. **72-96 hr**

Ans E While the respiratory compensation for a metabolic process happens quickly and is complete within 12-24 hr.

88. **CNS manifestations of respiratory alkalosis include**

A. psychosis
B. anxiety
C. asterixis
D. **paresthesia**
E. hallucinations

Ans D The paresthesia, tetany, and seizures may be partially related to the reduction in ionized calcium that occurs because alkalemia causes more calcium to bind to albumin.

89. **Respiratory acidosis occurs in**

A. severe anemia
B. carbon monoxide poisoning
C. **hypophosphatemia**
D. hypotension
E. Liddle syndrome

Ans C cause respiratory muscle weakness.

Oral Rehydration Therapy

90. **Sine wave in ECG is seen in?**

A. **Hyperkalemia**
B. Hypokalemia
C. Hypercalcemia
D. Hypocalcemia

Ans A Hyperkalemia produces a sequence of changes in ECG:
• 1st ECG finding: Narrowing and peaking (tenting) of the T wavesQ
• Further elevation of K+ leads to AV conduction disturbances, ↓ Pwave amplitude & widening of QRS interval
• Severe hyperkalemia eventually causes with a slow sinusoidal "sinewave" pattern followed by asystole. Peaked T waves Sine wave

91. **In children with burns maintenance intravenous fluid normally given is:**

A. **Ringer's lactate**
B. 5% Dextrose
C. Normal saline
D. Dextrose saline

Ans A Ringer's lactate Ringer lactate is usually used for fluid resuscitation in a child with burns.

92. **A 5yearold child presented with continuous fever and features of sepsis with a BP of 90/60 mm Hg, Pulse rate 144/min and Respiratory rate of 30/min. What is the initial fluid of choice for management?**

A. 10 mL/kg of hydroxyethyl starch
B. 10 mL/kg of 10% dextrose
C. **20 mL/kg of 0.9% normal saline**
D. 20 mL/kg of 0.45% normal saline

Ans C 20 mL/kg of 0.9% Normal Saline Ghai 8/e p718;

93. **ECG changes in hypomagnesaemia is characterized by**

A. peaking of the T waves and ST-segment depression
B. flattened T wave and depressed ST segment
C. **flattening of the T wave and lengthening of the ST segment**
D. prolonged PR, QRS, and QT intervals
E. appearance of a U wave

Ans C ECG changes in hyperkalemia include peaking of the T waves, this is followed, as the potassium level increases, by ST-segment depression, an increased PR interval, flattening of the P wave, and widening of the QRS complex. In hypokalemia a flattened T wave, a depressed ST segment, and the appearance of a U wave. In hypermagnesemia ECG changes include prolonged PR, QRS, and QT intervals.

94. **Contraction alkalosis occurs in**

A. metformin
B. **diuretic use**
C. propofol
D. methanol
E. toluene

Ans B causes fluid loss without bicarbonate; thus, the remaining body bicarbonate is contained in a smaller total body fluid compartment.

IV Fluids & Maintenance

95. Initial Fluid of choice for diarrhea in an infant?

A. ORS
B. Salt water
C. Sugar water
D. Dextrose

Ans A • Oral rehydration solution (ORS) containing appropriate quantities of sodium and glucose, promote optimum absorption of fluid from the intestine
• Other clear liquids, such as rice water, fruit juice, and sports drinks, are inappropriate for rehydration of young children with significant stool loss.

96. Na content (mEq/L) in Normal saline is:

A. 111
B. 130
C. 135
D. 154

Ans D

97. Daily maintenance fluid for a child weighing 10 kg is:

A. 1000 mL/day
B. 800 mL/day
C. 500 mL/day
D. 1200 mL/day

Ans A

98. Clinical manifestations of hypermagnesemia include all the following EXCEPT

A. hypotonia
B. hyporeflexia
C. sleepiness
D. hypertension
E. vomiting

Ans D Elevated magnesium values are associated with hypotension because of vascular dilatation, which also causes flushing.Hypotension can be profound at higher concentrations from a direct effect on cardiac function.

99. Increased production of CO2 occurs in all the following EXCEPT

A. fever
B. emesis
C. excess caloric intake
D. high levels of physical activity
E. increased respiratory muscle work

Ans B

Diabetic Ketoacidosis

100. Through a peripheral line, max % of dextrose that can be given is:

A. 10%
B. 12.5%
C. 15%
D. 25%

Ans B 12.5% , AIIMS NICU Protocols 2014

101. Maximum concentration of dextrose that can be given through peripheral vascular line in neonate:

A. 5%
B. 10%
C. 12.5%
D. 25%

Ans C AIIMS NICU Protocols 2014\ It is recommended to avoid using > 12.5% dextrose infusion through a peripheral vein due to risk of thrombophlebitis.

102. The most sensitive indicator of depletion of intravascular volume in infant is:

A. Cardiac output
B. Stroke volume
C. Heart rate
D. Blood pressure Chapter 4: Fluid and Electrolyte Disturbances 67 Answers

Ans C In infants with shock, compensatory increases in cardiac output are achieved primarily by "tachycardia" rather than by increase in stroke volume. Heart rates of 190–210/min are common in infants with compensated shock. What does theu egsivteino nKaryot ype • show? Q 1
• • Classification a. Normal male b. Normal female Important c. Down syndrome
• d. Turner syndrome
• •
• •
• •
• Overall, missense or nonsense TYPES muta tions are the most common
• Gain-of-function mutations typi cally

103. Treatment of hypermagnesmia include all the following EXCEPT

A. intravenous hydration
B. loop diuretics
C. Kayexalate
D. exchange transfusion
E. intravenous calcium gluconate

Ans C used for treatment of hypokalemia.

104. Very-low birth weight infants can have insensible losses of

A. 10-20 mL/kg/24 hr.
B. 30-40 mL/kg/24 hr.
C. 50-60 mL/kg/24 hr.
D. 70-80 mL/kg/24 hr.
E. 100-200 mL/kg/24 hr.

Ans E

05

CHAPTER 05

Genetics and Genetic Disorders

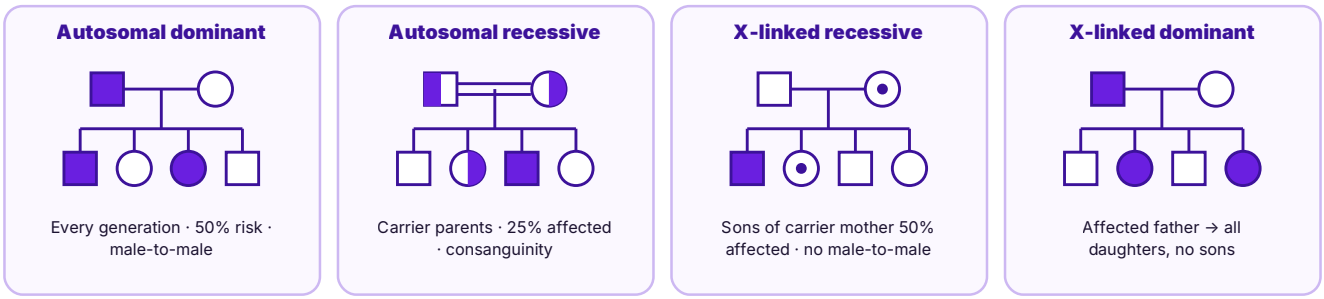


WHAT THIS CHAPTER COVERS

- Basic Genetics & Inheritance Patterns
- Chromosomal Disorders
- Down Syndrome
- Turner & Klinefelter Syndromes
- Muscular Dystrophies
- Hemoglobinopathies & Thalassemias
- Cystic Fibrosis
- Inborn Errors & Metabolic Disorders
- Neurocutaneous Syndromes
- Genetic Counseling & Screening
- Molecular Genetics & Mutations

218 MCQs **11** subtopics

5.1 Patterns of Inheritance



Four Mendelian patterns at a glance

Pattern	Key features	Examples
Autosomal dominant (AD)	Vertical transmission, every generation, M = F, **50% risk** to offspring of affected parent, male-to-male transmission possible; **variable expressivity, incomplete penetrance, new mutations (paternal age)**; usually **structural proteins**	Achondroplasia, **Marfan**, NF-1, NF-2, tuberous sclerosis, **Huntington**, myotonic dystrophy, **hereditary spherocytosis**, vWD (type 1), osteogenesis imperfecta (I–IV), familial hypercholesterolaemia, ADPKD, polyposis coli, Treacher Collins, Noonan, Waardenburg, retinoblastoma (hereditary), Ehlers–Danlos (most), Gilbert, Alport (rare AD)
Autosomal recessive (AR)	Horizontal (sibs affected), parents carriers, **consanguinity**, **25% risk** for each sib, 2/3 of unaffected sibs are carriers; usually **enzyme defects**	**Inborn errors of metabolism** (PKU, galactosemia, MSUD, GSD, most lysosomal storage – **except Hunter & Fabry**), **cystic fibrosis**, **sickle cell, thalassaemia**, **SMA**, Wilson, haemochromatosis, CAH, Friedreich ataxia, ataxia telangiectasia, albinism, Crigler–Najjar, Dubin–Johnson, Rotor
X-linked recessive (XLR)	**Males affected**, females carriers; **no male-to-male transmission**; affected male → all daughters carriers; carrier mother → 50% sons affected	**Haemophilia A & B**, **DMD/Becker**, **G6PD deficiency**, **Hunter (MPS II)**, **Fabry**, **Lesch–Nyhan**, **Bruton XLA**, **Wiskott–Aldrich**, CGD (X-linked form), colour blindness, ocular albinism, **Menkes**, nephrogenic DI, SCID (common γ chain), Alport (85%), adrenoleukodystrophy, OTC deficiency, Duncan (XLP)
X-linked dominant (XLD)	Affected father → **all daughters affected, no sons**; females more often affected, milder	**Hypophosphataemic (vitamin-D-resistant) rickets**, **Rett syndrome** (lethal in males), **incontinentia pigmenti**, Alport (some), **fragile X** (X-linked, behaves like dominant with reduced penetrance), Aicardi, oral-facial-digital type 1
Mitochondrial	**Maternal inheritance** only; all children of affected mother at risk; **heteroplasmy** → variable severity	**MELAS, MERRF, Leber hereditary optic neuropathy (LHON)**, Kearns–Sayre, Pearson, NARP, Leigh (some)
Multifactorial	Polygenic + environment; risk ↑ with number of affected relatives & severity; sex predilection (Carter effect)	**NTD, cleft lip/palate, CHD, pyloric stenosis, DDH, clubfoot, Hirschsprung**, diabetes, hypertension

X-LINKED RECESSIVE – 'BE WISE, FOOL'S GOLD HEEDS SILLY HOPE'

Bruton, **W**iskott–Aldrich, **F**abry, **G**6PD, **O**cular albinism, **L**esch–Nyhan, **D**uchenne, **H**unter, **S**CID (IL2RG), **H**aemophilia A/B, **O**TC deficiency.

Non-Mendelian mechanisms

Concept	Meaning / example
Genomic imprinting	Expression depends on parent of origin: **Prader–Willi** (loss of **paternal** 15q11–13), **Angelman** (loss of **maternal** UBE3A), **Beckwith–Wiedemann**, Russell–Silver

Concept	Meaning / example
Uniparental disomy (UPD)	Both homologues from one parent – **maternal UPD 15 → Prader–Willi**; **paternal UPD 15 → Angelman**
Trinucleotide repeat expansion – anticipation	Severity ↑/onset earlier in successive generations: **Fragile X (CGG, FMR1, maternal expansion)**, **myotonic dystrophy (CTG; congenital form from mother)**, **Huntington (CAG; paternal anticipation)**, Friedreich ataxia (GAA – AR), SCA
Mosaicism	Two cell lines from post-zygotic event – mosaic Down/Turner (milder); **gonadal (germline) mosaicism** – recurrence in sibs of "new" AD mutation (OI, achondroplasia, DMD)
Pleiotropy	One gene → many unrelated effects (Marfan, PKU)
Locus heterogeneity	Same phenotype from different genes (retinitis pigmentosa, OI)
Allelic heterogeneity	Different mutations in same gene (CF – ΔF508 and others; β-thalassaemia)
Penetrance vs expressivity	Penetrance – all-or-none (retinoblastoma ~90%); expressivity – degree (NF-1)
Lyonisation	Random X-inactivation in females → **manifesting carriers** (DMD, haemophilia)

5.2 Chromosomal Disorders – Overview

- Most common chromosomal abnormality in **liveborns**: **trisomy 21**; in **first-trimester abortions**: **trisomy 16** (most common trisomy) and overall **45,X (Turner)** is the most common single karyotype in abortuses.
- Most common sex-chromosome abnormality: **Klinefelter (47,XXY)** in males; **47,XXX** in females.
- Most common cause of trisomy: **maternal meiosis I non-disjunction** (risk ↑ with maternal age).
- Karyotyping is done on **metaphase** (colchicine arrest), **G-banding (Giemsa)**; **FISH** for rapid aneuploidy/microdeletions (DiGeorge 22q11, Williams 7q11, Prader–Willi); **chromosomal microarray (CMA)** – first-tier test for ID/autism/multiple anomalies; **whole exome sequencing** for monogenic disorders.

Trisomy	Syndrome	Key features	Survival
21	**Down**	Hypotonia, flat face, upslanting eyes, epicanthus, Brushfield spots, single palmar crease, sandal gap, AVSD	Into adulthood
18	**Edwards**	**Clenched fist with overlapping fingers (2nd over 3rd, 5th over 4th)**, **rocker-bottom feet**, prominent occiput, micrognathia, low-set ears, **short sternum**, VSD, horseshoe kidney, polyhydramnios, IUGR	90% die in 1st year; **F > M**
13	**Patau**	**Holoprosencephaly**, **cleft lip/palate**, **microphthalmia/cyclopia**, **post-axial polydactyly**, **scalp defects (cutis aplasia)**, omphalocele, **polycystic kidneys**, CHD	Most die in 1st month

EDWARDS = ELECTION AGE 18; PATAU = PUBERTY AT 13; DOWN = DRINKING AGE 21

Edwards (18): Rocker-bottom feet, Overlapping fingers, Clenched hand, Kidney (horseshoe), Ears low-set, Retardation, small jaw (**"ROCKER"**). **Patau (13)**: Polydactyly, Midline defects (holoprosencephaly, cleft), Microphthalmia, Cutis aplasia.

5.3 Down Syndrome (Trisomy 21)

Mechanism	%	Recurrence risk
Meiotic non-disjunction (maternal, meiosis I)	**~95%**	~1% (or age-related, whichever higher)
Robertsonian translocation (14;21 most common, 21;21)	3–4%	**Mother carrier 10–15%**, father carrier 2–3%; **21;21 carrier → 100%**

Mechanism	%	Recurrence risk
Mosaicism	1–2%	Low; milder phenotype

- Maternal-age risk: 1/1500 at 20 y, **1/350 at 35 y**, 1/100 at 40 y, 1/30 at 45 y. But most Down babies are born to **younger mothers** (more births).
- Translocation Down: **karyotype parents** (always karyotype the baby first).

Clinical features

- **Hypotonia (most common neonatal feature)**, flat facial profile, **upslanting palpebral fissures**, **epicanthic folds**, **Brushfield spots**, small low-set ears, protruding tongue, short neck with excess nuchal skin, **single palmar (simian) crease**, **clinodactyly of 5th finger**, **sandal gap**, brachycephaly, **third fontanelle**, joint hyperflexibility.
- **Intellectual disability** – MC genetic cause of ID (IQ 25–70).
- **Cardiac (40–50%)**: **AVSD (endocardial cushion defect) – most characteristic**, VSD (MC in some series), ASD (ostium primum), PDA, TOF.
- **GI (10–12%)**: **duodenal atresia ("double bubble")**, **Hirschsprung disease**, TEF, imperforate anus, **annular pancreas**, celiac disease.
- **Haematological**: **transient myeloproliferative disorder (TMD)** in newborn (GATA1), **AMKL (M7) – most common leukaemia < 4 y in DS**, **ALL (more common overall)** – 10–20 × risk; polycythaemia, macrocytosis.
- **Endocrine**: **congenital & acquired hypothyroidism** (screen at birth, 6 m, 12 m then yearly), diabetes, obesity.
- **Others**: **atlantoaxial instability** (screen before sports), **early-onset Alzheimer's** (APP gene on chr 21), OSA, hearing loss, refractive errors & cataract, **infertile males**, keratoconus.
- Prenatal screening: **1st trimester – NT ↑, PAPP-A ↓, free β-hCG ↑**; **2nd trimester quadruple – AFP ↓, uE3 ↓, hCG ↑, inhibin-A ↑**; **cell-free fetal DNA (NIPT)** – best screening; confirm by **CVS (10–13 wk) / amniocentesis (15–18 wk)** karyotype/QF-PCR.

SCREENING MARKERS – MEMORISE THE ARROWS

Down: 'HI' is high (**h**CG & **i**nhibin ↑), rest low. **Edwards: all low** (including hCG). **NTD: AFP ↑ only.** Ultrasound soft markers of Down: **↑ nuchal translucency (most sensitive single US marker in 1st trimester)**, absent/hypoplastic nasal bone, echogenic bowel, short femur, echogenic intracardiac focus, duodenal atresia.

5.4 Turner & Klinefelter

Feature	Turner (45,X)	Klinefelter (47,XXY)
Incidence	1 : 2500 live **female** births (99% of 45,X conceptions abort)	1 : 500–1000 males (MC cause of male hypogonadism/infertility)
Origin	Loss of **paternal X** in ~70–80%; mosaics 45,X/46,XX; isochromosome Xq	Maternal or paternal non-disjunction
Newborn	**Lymphoedema of hands & feet**, webbed neck (**cystic hygroma** in utero), **low posterior hairline**	Usually normal
Stature	**Short stature (MC feature)** – SHOX haploinsufficiency	**Tall, eunuchoid** (long legs)
Gonads	**Streak ovaries** – primary amenorrhoea, ↑ FSH (hypergonadotropic hypogonadism)	**Small firm testes**, azoospermia, ↑ FSH & LH, ↓ testosterone; gynaecomastia
Cardiac	**Bicuspid aortic valve (MC)**, **coarctation of aorta**, aortic dissection	MVP
Renal	**Horseshoe kidney**, duplicated collecting system	—
Others	**Shield chest with widely spaced nipples**, **cubitus valgus**, short 4th metacarpal, nail dysplasia, multiple naevi, hypothyroidism (Hashimoto), celiac, hearing loss; **normal intelligence** (visuospatial deficit)	Learning/language difficulty, ↑ breast cancer, SLE, germ cell tumours (mediastinal)

Feature	Turner (45,X)	Klinefelter (47,XXY)
Treatment	**GH** for height; **oestrogen** from ~11–12 y; Y-material mosaic → **gonadectomy** (gonadoblastoma risk)	Testosterone from puberty

- **Noonan syndrome** (PTPN11, AD, both sexes, **normal karyotype**): Turner-like phenotype + **pulmonary valve stenosis (dysplastic)**, **HCM**, ptosis, pectus, cryptorchidism, **bleeding diathesis (factor XI deficiency)**, ID mild, JMML risk.
- **47,XYY**: tall, normal appearance, behavioural problems, normal fertility. **48,XXXY / 49,XXXXY**: more severe ID.

5.5 Microdeletion & Other Important Syndromes

Syndrome	Defect	Features
DiGeorge / velocardiofacial	**22q11.2 deletion (TBX1)**, 3rd & 4th pharyngeal pouch	**CATCH-22**: Cardiac (**interrupted aortic arch type B, truncus arteriosus**, TOF), Abnormal facies, **Thymic hypoplasia (T-cell deficiency)**, Cleft palate, **Hypocalcaemia (hypoparathyroidism)**; schizophrenia later
Williams	7q11.23 (**elastin**)	Elfin face, **supravalvular AS**, peripheral PS, **hypercalcaemia**, friendly "cocktail party" personality, hyperacusis
Prader–Willi	Paternal 15q11–13 deletion (70%) / maternal UPD (25%)	Hypotonia → hyperphagia & obesity after 2 y, hypogonadism, short stature, small hands, ID; **methylation test first**
Angelman	Maternal 15q (UBE3A)	Happy puppet, seizures, ataxia, absent speech, microcephaly
Cri-du-chat	5p deletion	Cat cry, microcephaly, hypertelorism, ID
Wolf–Hirschhorn	4p deletion	"**Greek warrior helmet**" facies, seizures
Smith–Magenis	17p11.2	Self-injury, sleep disturbance, hugs self
WAGR	11p13 (WT1 + PAX6)	**Wilms, Aniridia, GU anomalies, Retardation**
Miller–Dieker	17p13 (LIS1)	**Lissencephaly**
Alagille	JAG1 (AD)	Paucity of bile ducts, **peripheral pulmonary stenosis**, **butterfly vertebrae**, posterior embryotoxon, triangular face
Fragile X	**FMR1 CGG repeat > 200** (Xq27.3); premutation 55–200	**MC inherited cause of ID & autism**; long face, **large ears**, prominent jaw, **macro-orchidism (post-pubertal)**, hyperextensible joints, MVP; premutation carriers – **premature ovarian failure**, FXTAS; Dx **PCR/Southern blot** (not karyotype)
Marfan	**FBN1 (fibrillin-1), chr 15q21, AD**	Tall, **arachnodactyly**, arm span > height, **ectopia lentis (upward & outward/superotemporal)**, **aortic root dilatation/dissection (MCC death)**, **MVP**, pectus, high-arched palate, scoliosis, pneumothorax; **thumb (Steinberg) & wrist (Walker–Murdoch) signs**; Rx β-blocker/losartan
Homocystinuria	**Cystathionine β-synthase (AR)**	Marfanoid + **ID**, **downward & inward (inferonasal) lens dislocation**, **thrombosis**, osteoporosis; **cyanide-nitroprusside test +**; Rx **pyridoxine (B6)**, methionine restriction, betaine
Ehlers–Danlos	Collagen (COL5 classic; COL3 vascular)	Skin hyperextensibility, joint hypermobility, **"cigarette-paper" scars**, vascular type → arterial rupture

5.6 Muscular Dystrophies & SMA (genetic view)

	Duchenne (DMD)	Becker (BMD)
Gene	**Dystrophin, Xp21** – largest human gene; **frameshift (out-of-frame) deletion** → absent dystrophin	**In-frame deletion** → reduced/abnormal dystrophin

	Duchenne (DMD)	Becker (BMD)
Onset	**3–5 years** (delayed walking, frequent falls)	5–15 years, milder
Signs	**Gowers' sign**, waddling gait, toe-walking, **calf pseudohypertrophy** (fat/fibrous), lordosis, proximal > distal weakness; **mild ID** in 1/3	Same, slower
Course	**Wheelchair by ~12 y**, scoliosis, dilated cardiomyopathy; death in 20s from **respiratory failure** (± cardiomyopathy)	Ambulant beyond 16 y; cardiomyopathy prominent
Investigations	**CPK markedly ↑ (10–100× – earliest, even at birth)**; **MLPA/genetic testing for deletion = first test of choice**; muscle biopsy – absent dystrophin on immunostaining; EMG myopathic	
Rx	**Corticosteroids (prednisolone/deflazacort) from ~4–5 y** – only drugs proven to prolong ambulation; ACEi/β-blocker for heart; exon-skipping (eteplirsen), gene therapy	

- **Myotonic dystrophy type 1** (DMPK, CTG repeat, AD): distal weakness, **myotonia** (can't release grip), **hatchet face**, frontal balding, cataract, cardiac conduction defects; **congenital form** (mother affected) – neonatal hypotonia, polyhydramnios, respiratory failure, **tented upper lip**.
- **Spinal muscular atrophy** (SMN1 deletion, chr 5q, AR): anterior horn cell loss; **type 1 (Werdnig–Hoffmann)** – onset < 6 m, never sits, **tongue fasciculations**, frog-leg posture, **areflexia**, bell-shaped chest, alert face, death < 2 y; type 2 – sits, never walks; type 3 (Kugelberg–Welander) – walks. Copy number of **SMN2** modifies severity. Rx **nusinersen** (antisense), **onasemnogene abeparvovec** (gene therapy), risdiplam.

5.7 Hemoglobinopathies (genetic aspects)

- **β-thalassaemia**: AR, chr 11; **most common mutation in India – IVS 1-5 (G→C) splice mutation**; > 200 mutations. **Point mutations** (not deletions) in β-thal; **deletions** in α-thalassaemia (chr 16).
- **Sickle cell**: point mutation **GAG → GTG at codon 6 (glutamic acid → valine)** of β-globin – classic **missense** mutation.
- Details → Haematology chapter.

5.8 Cystic Fibrosis

- **AR, CFTR gene on chr 7q31**; most common mutation **ΔF508 (class II – protein misfolding/processing defect)**; most common lethal AR disease in Caucasians (rare in India).
- CFTR = cAMP-regulated **chloride channel** → thick secretions.
- Features: **meconium ileus (earliest manifestation, 10–20%)**, recurrent pneumonia (**S. aureus** early, **Pseudomonas aeruginosa** (mucoid) later, **Burkholderia cepacia** – poor prognosis), bronchiectasis (upper lobes), nasal polyps, pancreatic insufficiency (steatorrhoea, FTT, fat-soluble vitamin deficiency), **rectal prolapse**, **DIOS**, CF-related diabetes, **male infertility (congenital bilateral absence of vas deferens – 98%)**, salty-tasting skin, **hypochloraemic metabolic alkalosis/pseudo-Bartter** in hot climates, focal biliary cirrhosis, **digital clubbing**.
- Diagnosis: **sweat chloride (pilocarpine iontophoresis) ≥ 60 mmol/L** – gold standard (30–59 intermediate); newborn screening **immunoreactive trypsinogen (IRT)**; nasal potential difference; CFTR genotyping; **faecal elastase** for pancreatic function.
- Rx: airway clearance (chest physio), **dornase alfa (rhDNase)**, **hypertonic (7%) saline**, inhaled tobramycin/aztreonam, **azithromycin**, pancreatic enzymes + ADEK, high-calorie diet; **CFTR modulators – ivacaftor (gating, G551D), elexacaftor-tezacaftor-ivacaftor (ΔF508)**; lung transplant.

5.9 Neurocutaneous Syndromes

Feature	NF-1 (von Recklinghausen)	Tuberous sclerosis (TSC)	Sturge–Weber
Genetics	**AD, chr 17q11 (neurofibromin)**; 50% new mutations	**AD, TSC1 (hamartin, 9q34) / TSC2 (tuberin, 16p13)** – mTOR pathway	**Sporadic – somatic GNAQ mutation**
Skin	**≥ 6 café-au-lait macules** (> 5 mm prepubertal, > 15 mm post-pubertal), **axillary/inguinal freckling (Crowe sign)**, neurofibromas (cutaneous, plexiform)	**Ash-leaf macules (hypomelanotic – earliest; Wood's lamp)**, **facial angiofibromas (adenoma sebaceum)**, **shagreen patch** (lumbosacral), **periungual fibromas (Koenen tumours)**, confetti lesions	**Port-wine stain (naevus flammeus)** in **ophthalmic (V1) trigeminal** distribution
Eye	**Lisch nodules** (iris hamartomas, ≥ 2), **optic glioma**	Retinal hamartomas (phakomas)	**Glaucoma (buphthalmos)**, choroidal haemangioma
CNS	Learning disability, macrocephaly, optic pathway glioma, **UBOs (T2 bright spots)**	**Cortical tubers, subependymal nodules ("candle-dripping"), SEGA (giant cell astrocytoma)**; **infantile spasms (West) – vigabatrin is DOC in TSC**, ID, autism	**Leptomeningeal angiomatosis**, **tram-track (gyriform) calcification** on X-ray/CT, seizures, hemiparesis, ID
Others	Scoliosis, **pseudarthrosis of tibia**, **sphenoid wing dysplasia**, pheochromocytoma, hypertension (renal artery stenosis), JMML, MPNST	**Cardiac rhabdomyoma (MC fetal cardiac tumour; regress)**, **renal angiomyolipoma**, **lymphangioleiomyomatosis (LAM)**, dental pits	—
Rx	Surveillance; **selumetinib** for plexiform neurofibromas	**mTOR inhibitors – everolimus/sirolimus**	Anticonvulsants, laser (pulsed-dye) for stain, hemispherectomy

- **NF-2**: **chr 22 (merlin)**, **bilateral vestibular schwannomas (acoustic neuromas)**, meningiomas, juvenile posterior subcapsular cataract; fewer café-au-lait spots.
- **Von Hippel–Lindau**: chr 3p (VHL, AD) – cerebellar & retinal **haemangioblastomas**, renal cell carcinoma, pheochromocytoma, pancreatic cysts.
- **Ataxia telangiectasia**: ATM, chr 11q22, AR – see Immunology.
- **Incontinentia pigmenti**: XLD (NEMO) – lethal in males; skin stages: vesicular → verrucous → **hyperpigmented whorls (Blaschko lines)** → atrophic; dental & eye anomalies.
- **Hypomelanosis of Ito**: hypopigmented whorls + CNS anomalies.

5.10 Genetic Counselling & Prenatal Diagnosis

Test	Timing	Comment
Chorionic villus sampling	**10–13 weeks**	Earliest invasive test; limb reduction defects if < 10 wk; confined placental mosaicism
Amniocentesis	**15–20 weeks** (≥ 15 wk)	Karyotype, AFP, acetylcholinesterase (NTD), DNA
Cordocentesis (PUBS)	> 18 weeks	Rapid karyotype, fetal anaemia, transfusion
NIPT (cell-free fetal DNA)	**From 10 weeks**	Best screening test for trisomy 21; positive result needs invasive confirmation
Pre-implantation genetic testing (PGT)	IVF embryo (blastomere/ trophectoderm)	Avoids termination

- **PCPNDT Act (1994, amended 2003)** – prohibits sex selection/disclosure of fetal sex in India.
- **MTP (Amendment) Act 2021** – termination allowed **up to 24 weeks** for special categories; **beyond 24 weeks for substantial fetal abnormalities** on Medical Board approval.

- **Hardy–Weinberg**: $p^2 + 2pq + q^2 = 1$; carrier frequency ≈ 2√(disease incidence). E.g. incidence 1/2500 → q = 1/50 → carriers ≈ 1/25.
- Risk communication: AR – both carriers → 25% affected each pregnancy; AD → 50%; X-linked carrier mother → 50% of sons affected, 50% daughters carriers.

5.11 Molecular Genetics – Quick Concepts

Mutation	Example
Missense (amino acid change)	**Sickle cell (Glu6Val)**
Nonsense (premature stop)	β^0-thalassaemia (codon 39 – Mediterranean)
Frameshift	DMD (out-of-frame deletion), Tay–Sachs (4-bp insertion)
Splice-site	**β-thalassaemia IVS1-5 (India)**
Trinucleotide repeat	Fragile X, Huntington, myotonic dystrophy, Friedreich
Deletion	α-thalassaemia, DMD (60–65%), SMA (SMN1 exon 7)
Gain-of-function (dominant)	**Achondroplasia (FGFR3 G380R – nearly all cases)**, Huntington, Liddle
Dominant negative	Osteogenesis imperfecta, Marfan

- Techniques: **PCR** (amplification), **Southern blot (DNA)**, Northern (RNA), Western (protein) – "SNoW DRoP"; **MLPA** (exon deletions/duplications – DMD, SMA); **Sanger** (single gene); **NGS/WES** (many genes); **FISH** (microdeletions, rapid aneuploidy); **CMA** (CNVs; cannot detect balanced translocations).
- **Lyon hypothesis**, **Barr body** = inactivated X (number = X − 1): Turner 0, Klinefelter 1, XXX 2.

Practice MCQs · Genetics and Genetic Disorders

218 questions · correct option highlighted in green · cover the answer strip to self-test

Basic Genetics & Inheritance Patterns

1. What is proband in pedigree?

A. Male child of diseased
B. Female child of diseased
C. **Diseased individual**
D. Pregnant lady

Ans C

2. Inheritance of Beckers muscular dystrophy is:

A. **X-linked recessive**
B. X-linked dominant
C. Autosomal recessive
D. Autosomal dominant

Ans A Both Duchenne's & Becker's muscular dystrophy have x linked recessive inheritance.

3. Which of the following is an autosomal dominant metabolic disorder?

A. **Hereditary hypercholesterolemia**
B. Tay sachs disease
C. Gaucher's disease
D. Tyrosinemia

Ans A

4. In a family, mother is normal while father has a genetic disease. All their daughters are carriers and sons are normal, what is the pattern of inheritance of disease?

A. **X-linked recessive**
B. X-linked dominant
C. AD
D. AR

Ans A • In a X-linked recessive disease; affected male transmits the disorder to all his daughters (carriers)
• Sons get their X chromosome from their mother & not from their father
• Hence sons of a diseased father are unaffected in X-linked diseases In an X-linked dominant disorder all the daughters of an affected father would be diseased and not carriers.

5. Which of the following is inherited as autosomal recessive?

A. Achondroplasia
B. Tuberous sclerosis
C. **Hemochromatosis**
D. Osteogenesis imperfecta

Ans C

6. Which of the following is autosomal recessive inherited cancer syndrome?

A. **Ataxia telangiectasia**
B. Cowden syndrome
C. Retinoblastoma
D. HNPCC

Ans A Cowden syndrome, Retinoblastoma and HNPCC have autosomal dominant inheritance

7. Inheritance of familial hypercholesterolemia:

A. **Autosomal dominant**
B. Autosomal recessive
C. X-linked dominant
D. X-linked recessive

Ans A

8. Inheritance of congenital adrenal hyperplasia:

A. Autosomal dominant
B. **Autosomal recessive**
C. X-linked dominant
D. X-linked recessive

Ans B Congenital adrenal hyperplasia (CAH) are autosomal recessive diseases resulting from mutations of genes for enzymes mediating the biochemical steps of production of cortisol from cholesterol by the adrenal glands. d. Hypertension
Polygenic disorders:
• Disorders caused by interactions between variant forms of genes and environmental factors.
• Occur when many polymorphisms, each with a modest effect and low penetrance, are co-inherited
• E.g.

9. Inheritance of Alport syndrome:

A. Autosomal recessive
B. **X-linked dominant**
C. X-linked recessive
D. Mitochondrial inheritance

Ans B

10. Pedigree Chart is:

A. Telecanthus
B. Epicanthus
C. **Epiphora**
D. Strabismus

Ans C Used to see genetic transmission

11. Find Correct match about inheritance:

A. Hypophosphatemic rickets- X linked recessive
B. **Duchenne muscular dystrophy- X linked recessive**
C. Sickle cell: AR
D. Osteogenesis imperfecta-1 :AR
E. Achondroplasia : AR

Ans B Duchenne muscular dystrophy-X linked recessive & (c) Sickle cell: AR a. False; In X linked Hypophosphatemic rickets, female carriers are affected, so it is an X-linked dominant disorder. b. True; Duchenne muscular dystrophy has X linked recessive c. True; Sickle cell has Autosomal recessive inheritance d. False; All osteogenesis imperfecta are AD except type II, III & VII e. False; Achondroplasia has autosomal dominant inheritance

12. Study the following carefully: Read the pedigree. Inheritance pattern of the disease in the family is:

A. Autosomal recessive
B. Autosomal dominant
C. X-linked dominant
D. **X-Linked recessive**

Ans D Only males are manifesting disease and females are acting as carrier; so it is X-linked recessive inheritance.

13. Which of the following is the most likely inheritance pattern in the pedigree given below?

A. Autosomal dominant
B. Mitochondrial
C. Autosomal recessive
D. **X-linked dominant**

Ans D It is an X-linked dominant pattern of inheritance, all the female offsprings of a diseased male (XY) receive abnormal X chromosome & express the disease, whereas none of the male offsprings of diseased father have the disease.

14. Which of the following is X-linked recessive?

A. **Duchenne muscular dystrophy**
B. Hypophosphatemic rickets
C. Marfan's syndrome
D. Downs syndrome

Ans A

15. Which of the following diseases/disorders is NOT transmitted as a recessive sex linked trait?

A. Hemophilia
B. **Cystic fibrosis**
C. G6PD deficiency
D. Retinitis pigmentosa

Ans B Cystic fibrosis with

16. Which of the following has autosomal dominant inheritance?

A. **Hereditary spherocytosis**
B. Thalassemia
C. Sickle cell disease
D. G6PD deficiency

Ans A

17. Which one of the following conditions is not inherited in autosomal dominant pattern?

A. Neurofibromatosis
B. **Friedrich's ataxia**
C. Marfan's syndrome
D. Hereditary spherocytosis

Ans B

18. All of the following conditions have autosomal dominant inheritance except:

A. **Fabry disease**
B. Marfan's syndrome
C. Osteogenesis imperfecta
D. Ehlers-Danlos syndrome

Ans A

19. True about autosomal dominant type of inheritance:

A. 25% affected and 50% carrier if one parent affected
B. 50% affected and 75% carrier if both parent affected
C. **75% affected if both parent affected**
D. 50% affected if one parent affected
E. All carrier irrespective of either one parent affected or both parent affected

Ans C and d. 50% affected if one parent affected In an autosomal dominant disorder, If one parent is affected: If both parents are affected: So, 50% children are affected, if only one parent is affected 75% affected if both parents affected

20. All the following are autosomal dominant except:

A. **Cronkhite Canada syndrome**
B. Bannayan Ruvalcaba Riley syndrome
C. Peutz Jeghers syndrome
D. Gardner's syndrome86 Section 1: General Pediatrics

Ans A Discussing about options one by one, a. Cronkhite Canada syndrome is a non-inherited condition occurring in old age with features of loss of taste, intestinal polyps, hair loss and nail growth problem b. Bannayan-Riley-Ruvalcaba syndrome is an autosomal dominant condition characterized by a large head size (macrocephaly), multiple noncancerous tumors and hamartomas, and dark freckles on penis in males c.

21. The population frequency for phenylketonuria is 1 in 10,000. What is the carrier frequency for the disease?

A. **1/50**
B. 1/100
C. 1/500
D. 1/1000

Ans A It is based on Hardy Weinberg principle: Simplifying it, Carrier frequency for an autosomal recessive disorder = twice the square root of disease incidence So 2 X square root of 1/10,000 gives 1/50 as the answer.

22. What is the risk of an affected individual transmitting an autosomal dominant disease to his/her offspring?

A. **50%**
B. 100%
C. 25%
D. 60%

Ans A 50% a. Autosomal dominant

23. What is the mode of inheritance of Hereditary spherocytosis?

A. Autosomal dominant
B. Autosomal recessive
C. X-linked recessive
D. X-linked dominant

Ans A Hereditary spherocytosis is inherited in an autosomal dominant pattern in about 75% of cases; a minority are autosomal recessive or arise from new mutations.

24. Which is X-linked dominant condition?

A. Hypophosphatemic Rickets
B. Hemophilia
C. Gaucher disease
D. Cystic fibrosis

Ans A

25. The inheritance of hemophilia is:

A. Autosomal dominant
B. Autosomal recessive
C. X-linked dominant
D. X-linked recessive

Ans D

26. X-linked dominant disease is:

A. Fragile X syndrome
B. Vit D resistant rickets
C. Lesch-Nyhan syndrome
D. Adrenoleukodystrophy

Ans B

27. True statement about inheritance of an X-linked recessive trait is:

A. Single nucleotide mutation
B. Trinucleotide repeat mutation
C. Base excision repair
D. DNA breakage

Ans A 50% of boys of carrier mother are affected For a X-linked recessive disease: If mother is a carrier: If father is diseased: So, 50% of boys of carrier mothers are affected All daughters are carriers Mothers transmit defective gene to 50% of the sons (diseased) as well Fathers transmit defective gene to daughters as daughters (carriers)

28. Which of the following is autosomal dominant type of genetic disorder?

A. Colour blindness
B. Hemophilia
C. Phenylketonuria
D. Tuberous sclerosis

Ans D

29. A particular genetic disorder appears in three consecutive generations of a family without any sex predilection. It was also noticed that phenotypically normal family members were having healthy offspring, What is the pattern of inheritance of this disorder?

A. Autosomal recessive
B. Autosomal dominant
C. Mitochondrial inheritance
D. Uniparental disomy

Ans B Features of Autosomal Dominant Inheritance:
• Affected individuals present in every generation and Males and females are equally affected
• Any child of an affected parent has a 50% risk of inheriting the disorder
• Phenotypically normal family members do not transmit the condition to the offspring

30. The following are autosomal dominant disorders except:

A. Myotonic dystrophy
B. Von Willebrand disease
C. Hemochromatosis
D. Marfan's syndrome

Ans C Hemochromatosis is autosomal recessive.

31. Pedigree analysis—Analyze the following pedigree and give the mode of inheritance:

A. Autosomal recessive
B. Autosomal dominant
C. Mitochondrial inheritance
D. X-linked dominant

Ans C This disease is manifesting in both males and females and all children of a diseased female have disease; Both these are characteristics of mitochondrial diseases.

32. In an autosomal Recessive (AR) disorder, one parent is normal and the other is carrier and the child is also affected. What is the reason?

A. Germline mosaicism
B. Genomic imprinting
C. Penetration
D. Uniparental disomy

Ans D • If one parent is normal for an autosomal recessive disorder and the other is carrier. No child should be affected.
• But there is exception to this rule, i.e. uniparental disomy, when a person receives 2 copies of a chromosome, from one parent and no copies from the other parent.

33. Which of the following diseases is an autosomal dominant disorder:

A. Wilson's disease
B. Tuberous sclerosis
C. Huntington's disease
D. Schizophrenia

Ans C Maturity onset diabetes of the young Maturity-onset diabetes of the young (MODY):
• Now referred to as monogenic diabetes; it is autosomal dominant and has strong family history
• MODY results from mutations in genes for pancreatic β-cell or liver glucokinase or hepatocyte nuclear factor (1 , 4 , or 1β); there is no autoimmune destruction of β cells and no HLA association
• It may require insulin treatment or can be treated with sulfonylureas with varying degrees of success.

34. A is hesitant about having children because her two sisters had sons who had died form kinky hair disease. Her mother's brother also died of the same condition. Which of the following is the possible mode of inheritance in her family?

A. X-linked recessive
B. X-linked dominant
C. Autosomal recessive
D. Autosomal dominant

Ans A The clues in this Question are: Only males are manifesting disease and Females are acting as carriers (sister's sons had suffered from the disease); So, this disease has X-linked recessive inheritance

35. An albino girl gets married to a normal boy. What are the chances of their having an affected child and what are the chances of their children being carriers?

A. None affected, all carriers
B. All normal
C. 50% carriers
D. 50% affected, 50% carriers

Ans A • Albinism is autosomal recessive; Female here is affected (2 defective genes), while male is normal (both genes normal).
• So each offspring (son/daughter) will inherit one defective gene only, from the mother.
• So all children will be carriers of albinism and none will be affected.

36. A parent is homozygous and a parent heterozygous for an autosomal recessive gene. What will be the outcome:

A. 75% children affected
B. No child affected, but all are carriers
C. 50% children affected, rest are carriers
D. 25% children affected, rest are carriers DOWN SYNDROME

Ans C As it is autosomal recessive, 50% affected and 50% carrier

37. Type of inheritance in MELAS:

A. AD
B. AR
C. Mitochondrial
D. X-linked

Ans C

38. The following are indications for genetic counseling EXCEPT

A. paternal age > 50 years
B. maternal age > 45 years
C. family history of intellectual disability
D. consanguinity
E. infertility

Ans B Maternal age > or equal to 35 is an indication for genetic counseling. Genetic counseling is a communication process in which the genetic contribution to health is explained, along with specific risks of transmission of a trait and options to manage the condition and its inheritance. The counselor is expected to present information in a neutral, nondirective manner and to provide support to the individual and family to cope with decisions that are made.

39. The following statements are true about genetic counseling EXCEPT

A. constructing family pedigree up to the third degree
B. collecting health files about affected family members
C. performing appropriate genetic testing
D. refer to support groups
E. talking about rewarding gene therapy

Ans E Gene therapy still experimental and rewarding therapy is still a hope.

40. One of the following is explained by digenetic inheritance

A. congenital lactic acidosis
B. glycogen storage disease type VI
C. biotinidase deficiency

Ans B Digenic inheritance explains the occurrence of retinitis pigmentosa (RP) in children of parents who each carry a mutation in a different RP-associated gene. Both parents have normal vision, as would be expected, but their offspring who are double heterozygotes—having inherited both mutations—develop RP. Digenic pedigrees can exhibit characteristics of both autosomal dominant (vertical transmission) and autosomal recessive inheritance (1 in 4 recurrence risk).

41. Parent-to-child transmission (vertical transmission) is a character of autosomal dominant inheritance, for many patients with an autosomal dominant disorder there is no history of an affected family member. All the following are true explanations EXCEPT

A. variable expressiveness
B. new mutation
C. somatic mutations (mosaicism)
D. digenetic inheritance
E. incomplete penetrance

Ans D Double heterozygotes

42. The following are characteristic feature of autosomal dominant inheritance EXCEPT

A. vertical transmission
B. male to male transmission
C. males and females are equally affected
D. skipped generation
E. both parents carried the affected gene

Ans E Autosomal dominant inheritance is determined by the presence of 1 abnormal gene on 1 of the autosomes (chromosomes 1-22). Autosomal genes exist in pairs, with each parent contributing 1 copy. In an autosomal dominant trait, a change in 1 of the paired genes has an effect on the phenotype.

43. All the following are characteristic features of autosomal recessive inheritance

A. vertical transmission
B. no affected family members in other generations
C. males and females are equally affected
D. consanguinity plays important role
E. 25% recurrence risk

Ans A Horizontal transmission is the characteristic feature of autosomal recessive inheritance, the observation of multiple affected members of kindred in the same generation, but no affected family members in other generations.

44. **Pseudodominant inheritance happens when**

A. an x-linked disorder appears as autosomal dominant
B. an autosomal dominant appears as recessive trait
C. **an autosomal recessive disorder appears as autosomal dominant**
D. skipped generations appears
E. an autosomal dominant appears as x-linked disorder

Ans C Pseudodominant inheritance refers to the observation of apparent dominant (parent to child) transmission of a known autosomal recessive disorder. This occurs when a homozygous affected individual has a partner who is a heterozygous carrier, and it is most likely to occur for relatively common traits, such as sickle cell anemia.

45. **In genetic disorders, male to male transmission occurs in Y- linked inheritance and in**

A. X- linked inheritance
B. pseudodominant inheritance
C. digenetic inheritance
D. **autosomal dominant disorder**
E. pseudogenetic inheritance

Ans D

46. **Females sometimes manifest signs of X- linked recessive disorders. All the following are possible explanations EXCEPT**

A. homozygosity for an X-linked trait
B. 45,XO female
C. 46,XY female
D. **uniparental disomy**
E. nonrandom X-inactivation

Ans D

47. **All the following are features of mitochondrial inheritance EXCEPT**

A. non-traditional inheritance
B. maternal inheritance
C. **male to offspring transmission**
D. both sexes are affected
E. mitochondrial DNA point mutations

Ans C Mitochondrial inheritance represents a non-traditional inheritance (not follow the usual Mendelian inheritance), both sexes can be affected from the affected mitochondrial DNA genome in the ovum (sperm contain few mitochondria which finishes during fertilization), so no male to offspring transmission.

48. **The following represent a non-traditional inheritance EXCEPT**

A. mitochondrial disorders
B. triplet repeat expansion diseases
C. imprinting defects
D. **familial clustering**
E. uniparental disomy

Ans D The first three represent the non-traditional inheritance; familial clustering represents a form of pseudogenetic transmission, while uniparental disomy is a type of imprinting defects.

49. **The recurrence risk of multifactorial inheritance are characterized by all the following EXCEPT**

A. related to the incidence of the disease in community
B. similar among all 1st-degree relatives
C. **no sex predilection**
D. increased when multiple family members are affected
E. greater when the disorder is more severe

Ans C Some multifactorial inherited disorders have a sex predilection, as indicated by an unequal male: female incidence. Pyloric stenosis, for example, is more common in males, whereas congenital dislocation of the hips is more common in females. Where there is an altered sex ratio, the risk is higher for the relatives of an index case whose gender is less commonly affected than relatives of an index case of the more commonly affected gender.

50. **A chromosomal study of 22-year-old mother of a baby with Down syndrome t (14; 21) shows that; she is the carrier for the translocation. You explained that the recurrence rate will be approximately**

A. 2-4%
B. **5-7%**
C. 8-10%
D. 11-13%
E. 33%

Ans B Translocation (21; 21) carriers have a 100% recurrence risk for a chromosomally abnormal child, and other Robertsonian translocations, such as t(14;21), have a 5-7% recurrence risk when transmitted by females.

Chromosomal Disorders

51. **A form of fetal programming where two genes are inherited but one is turned off through environmentally induced modification is called as:**

A. **Epigenetic imprinting**
B. Polymorphism
C. Gene modification
D. Gene heterogeneity

Ans A

52. **The structural abnormality of chromosome in which one arm is lost and remaining arm is duplicated is called:**

A. Ring chromosome
B. **Isochromosome**
C. Translocation
D. Mutation

Ans B Iso-chromosome results when 1 arm of a chromosome is lost and the remaining arm is duplicated or when the axis of division occurs perpendicular to the normal axis of division.

53. **RET proto oncogene is located on which chromosome:**

A. 9
B. **10**
C. 11
D. 12

Ans B 10 Important genes and their chromosomal location: Gene (Associated diseases) Chromosomal location Gene (Associated diseases) Chromosomal location Rb (Retinoblastoma) 13q14.3 NF2 (Neurofibromatosis 2) 22p12 p53 (Li-Fraumeni syndrome, Sarcomas) 17q13.1 WT1 (Wilm's tumor) 11p13 APC (Carcinoma Colon) 5q21 RET (MEN2 syndrome) 10q NF1 (Neurofibromatosis 1) 17q11

54. **Trisomy 13 is:**
A. Patau syndrome
B. Edward syndrome
C. Down syndrome
D. Di George syndrome

Ans A

55. **Abnormality involving which chromosome is seen in Hunter disease?**
A. X chromosome
B. Y chromosome
C. Chromosome
D. Chromosome

Ans A 9/e p 640 All types of mucopolysaccharidoses have autosomal recessive inheritance except Hunter disease, which is X linked recessive.

56. **Alpha1 antitrypsin gene is on which chromosome?**
A. Chr
B. Chr
C. Chr
D. Chr

Ans D 14 α1-Antitrypsin deficiency
• It is caused by mutation in SERPINA1 gene (chr 14q), which has autosomal recessive inheritance.
• α1-Antitrypsin, a protease inhibitor synthesized by liver, protects lung tissue from destruction by neutrophil elastase;
• Most common allele of protease inhibitor (Pi) system is M & the normal phenotype is PiMM.

57. **Trisomy seen in Down syndrome is:**
A. 21
B. 22
C. 18
D. 13

Ans A 21 In Down syndrome, Trisomy 21 is usually due to meiotic non-disjunction.

58. **Among the following, the most sensitive USG finding of Trisomy 21 is:**
A. Absent nasal bone
B. Thickened nuchal fold
C. Short femur
D. Echogenic bowel

Ans B Prenatal USG markers of Down syndrome include thickness of nuchal fold (75% sensitive), absence of nasal bone (58% sensitive), cardiac abnormalities, duodenal atresia, shortened femur, shortened humerus, renal pyelectasis, a hyperechogenic bowel, and a choroid plexus cyst.

59. **Identify the syndrome in a male child with the following karyotype:**
A. Turner syndrome
B. Edward syndrome
C. Down syndrome
D. Patau syndrome

Ans C The given karyotype shows trisomy 21 (3 chromosomes on chr no 21) and so is suggestive of Down syndrome.

60. **One of the parents who have a child with Down syndrome, has a balanced translocation between chromosome 15 and 21. What advice will you provide to the couple to prevent a child being born with Down syndrome?**
A. Prenatal diagnosis and advice abortion
B. Artificial insemination with donor's sperm
C. Adoption
D. Does not matter

Ans A For details about Recurrence risk of Down syndrome, Taking into view the chances of baby affected with Downs syndrome, prenatal diagnosis is advocated and abortion advised, if fetus is affected.

61. **A 35-years-old lady has chromosomal translocation 21/21. The risk of Down syndrome in the child is:**
A. 100%
B. 0%
C. 10%
D. 50%

Ans A A parent carrying a 21;21 Robertsonian translocation can only produce gametes with either two copies or zero copies of chromosome 21 — every liveborn child will have trisomy 21, giving a 100% recurrence risk.

62. **Identify the syndrome in following given karyotype?**
A. 47, XX +
B. 46, XXXY
C. 47, XXY
D. 45, XO 90 Section 1: General Pediatrics 134. True about Turner syndrome:
E. Gonadal dysgenesis The following statements regarding Turner syndrome are true except:

Ans A Turner syndrome The given karyotype shows single X chromosome and no Y chromosome and a total of 45 chromosomes which suggests 45, XO i.e Turner syndrome.

63. **Identify the disease shown in the karyotype:**
A. Cri-du-chat syndrome
B. Bloom syndrome
C. Angelman syndrome
D. Fragile X syndrome

Ans A Longman 11/e p 21; In the karyotype, there is deletion of chromosome 5, which leads to Cri-du-chat syndrome.

64. **Chromosome 22 deletion syndrome is:**
A. Down syndrome
B. Di George syndrome
C. Turner syndrome
D. Klinefelter syndrome

Ans B

65. **The karyotype in testicular feminising syndrome is?**
A. XX
B. XY
C. XXY
D. XXXY

Ans B

66. **Trisomy 13 is seen in:**
A. Edward syndrome
B. **Patau syndrome**
C. Down syndrome
D. Turner syndrome

Ans B

67. **About Trisomy 13, Which the following are true statements?**
A. **Bilateral microphthalmia**
B. Neurofibroma
C. Rocker bottom feet
D. Dermoid cyst

Ans A

68. **Which of the following is an example of disorders of sex chromosomes?**
A. Marfan's syndrome
B. Testicular feminization syndrome
C. **Klinefelter syndrome**
D. Down syndrome

Ans C Klinefelter syndrome is 47, XXY; In none of the other 3 disorders, abnormality of X or Y chromosomes is seen

69. **Males who are sexually underdeveloped with rudimentary testes and prostate glands, sparse pubic and facial hairs, long arms and legs and large hands and feet are likely to have the chromosome complement:**
A. 45XXY
B. 46XY
C. **47XXY**
D. 46X

Ans C 47 XXY This is clearly a case of Klinefelter syndrome:
• The male has a normal external genitalia but there is microorchidism and decreased testosterone level.
• The male in Kleinfelter syndrome is tall with long arms and legs.

70. **Common ocular manifestation in Trisomy 13 is:**
A. Capillary hemangioma
B. **Bilateral microphthalmos**
C. Neurofibroma
D. Dermoid cyst

Ans B Bilateral microphthalmos 140 above

71. **The maternal uniparental disomy for chromosome 15 results in:**
A. Angelman Syndrome
B. **Prader – Willi Syndrome**
C. Pallister – Killian Syndrome
D. Hypomelanosis of ITO

Ans B Maternal uniparental disomy of chromosome 15 (both copies inherited from the mother, none from the father) silences paternally-expressed genes and causes Prader-Willi syndrome. Paternal UPD15 causes Angelman syndrome.

72. **Maternal disomy of chromosome 15 is seen in:**
A. **Prader- Willi syndrome**
B. Klinefelter's syndrome
C. Angelman syndrome
D. Turner's syndrome DIAGNOSTIC TOOLS

Ans A Prader-Willi syndrome dna

73. **Submicroscopic deletions of any size can be detected by:**
A. **Multiplex ligation-Dependent probe amplification (MLPA)**
B. Southern blotting
C. Cytogenomic array technology
D. Chromosome painting

Ans A Multiplex ligation-Dependent probe amplification Multiplex Ligation-Dependent Probe Amplification (MLPA): It is a technique which mixes DNA hybridization, ligation, & PCR amplification to detect deletions & duplications of any size, including anomalies that are too large to be detected by PCR and too small to be identified by FISH.

74. **A 17-year-old adolescent female with trisomy 21 became pregnant, the chance of affection of her offspring with the same problem is**
A. Not possible
B. 25%
C. **50%**
D. 75%
E. 100%

Ans C Most males with Down syndrome are sterile, but some females have been able to reproduce, with a 50% chance of having trisomy 21 pregnancies.

75. **All the following are features of Edwards syndrome (Trisomy 18) EXCEPT**
A. low birth weight
B. closed fists
C. cardiac malformations
D. short sternum
E. **slopping forehead**

Ans E is a feature of Trisomy 13.

76. **All the following are features of Patau syndrome (Trisomy 13) EXCEPT**
A. scalp defects
B. corneal abnormalities
C. holoprosencephaly
D. capillary hemangiomas
E. **prominent occiput**

Ans E is a feature of Edwards's syndrome.

77. **Robertsonian translocations involve 2 acrocentric chromosomes that fuse near the centromeric region with a subsequent loss of the short arms. It can occur in the following chromosomes EXCEPT**
A. 13
B. 15
C. **17**
D. 21
E. 22

Ans C It includes also chromosome number 14.

78. **One of the common chromosomal deletions which can lead to retinoblastoma is**

A. 4p-
B. 5p-
C. 9p-
D. **13q-**
E. 18p-

Ans D

79. **All the following are examples of microdeletion syndromes EXCEPT**

A. Williams
B. Prader-Willi
C. Angelman
D. DiGeorge
E. **Wolf-Hirschhorn**

Ans E Microdeletions involve loss of small chromosome regions so that the affected individuals can have a distinctive phenotype depending on the number of genes involved. All the mentioned syndromes are common microdeletions and the board candidate should be familial with their phenotypic descriptions and clinical manifestations. Wolf-Hirschhorn syndrome is chromosomal deletion syndrome.

80. **An 18-month-old female child had been diagnosed as Turner syndrome (45, XO), FISH analysis shows Y chromosome mosaicism. Of the following, the MOST appropriate next step is to**

A. refer to endocrinologist
B. reassure as the course will be milder
C. advise for feminizing hormones
D. **arrange for laparoscopic removal of gonads**
E. rear the child as male gender

Ans D Phenotypic females with 45, X/46, XY mosaicism have a 15-30% risk of developing gonadoblastoma. The American Academy of Pediatrics has recommended the use of FISH analysis to look for Y-chromosome mosaicism in all 45, X patients. If Y chromosome material is identified, laparoscopic gonadectomy is recommended.

81. **All the following are characteristic features of fragile X chromosome of a 3-year-old male child EXCEPT**

A. intellectual disability
B. autistic behavior
C. **macroorchidism**
D. hyperextensible finger joints
E. characteristic facial features

Ans C The macroorchidism may not be evident until puberty. The facial features, which include a long face, large ears, and a prominent square jaw, become more obvious with age.

Down Syndrome

82. **Regarding 'Davidson body', all are correct except:**

A. **Present in males**
B. Present in neutrophils
C. Present in 4 – 6 % of cells
D. Drumstick appearance

Ans A Davidson bodies are small nuclear buds of chromatin shaped like a drumstick found in neutrophils of females, but absent in males.

83. **Jeune syndrome is transmitted as:**

A. **Autosomal recessive**
B. Autosomal dominant
C. X-linked Dominant
D. X-linked Recessive

Ans A Jeune syndrome
• It is a rare autosomal recessive genetic disorder that affects the cartilages and bones
• It mainly affects the rib cage (bell-shaped thorax), pelvis, arms and legs
• Thoracic insufficiency is the most important feature of this syndrome.

84. **Most common cause of Down syndrome is:**

A. **Meiotic non-disjunction**
B. Robertsonian translocation
C. Mosaicism
D. Partial trisomy

Ans A

85. **In the first trimester screening for Down syndrome which of the following test is to be done?**

A. **Beta HCG, PAPPA**
B. MSAFP, PAPPA
C. Inhibin, PAPPA
D. Unconjugated Estriol, PAPPAs

Ans A

86. **In Down syndrome, which of the following is decreased?**

A. Nuchal thickness
B. Beta HCG
C. Inhibin
D. **Alfa fetoprotein**

Ans D

87. **Definitive diagnosis of Down syndrome is by:**

A. **Karyotyping**
B. Triple test
C. Fetal ultrasonography
D. All of above

Ans A

88. **Risk of having a child with Down syndrome:**

A. Is unaffected by maternal age
B. Decreases with increase in maternal age
C. Increases with increase in paternal age
D. **Increases with increase in maternal age**

Ans D

89. **Most common heart lesion in down syndrome:**

A. **Endocardial cushion defect**
B. ASD with ostium secundum
C. VSD
D. Coarctation of aorta

Ans A

90. All the following are features of Down syndrome except:
A. Brushfield's spots in iris
B. Simian crease
C. Mental retardation
D. **Hypertonia**

Ans D Hypotonia is a feature of Down syndrome and not hypertonia

91. In Down syndrome the shape of the head is:
A. Oxycephalic
B. Scaphocephalic
C. **Brachicephalic**
D. Plagiocephalic

Ans C

92. In Down syndrome, all are seen except:
A. t (14;21)
B. Trisomy
C. **t (11;14)**
D. t (15;21)

Ans C t (11 ; 14) 3 copies of chr 21 should be present to cause Down syndrome

93. Prenatal diagnosis of Down syndrome is by: (Recent Question 2014) 88 Section 1: General Pediatrics
A. Karyotyping
B. Triple test
C. Fetal ultrasonography
D. **All of the above**

Ans D All of above

94. Most common CVS lesion in Down syndrome is:
A. VSD
B. **Endocardial cushion defect**
C. TOF
D. COA

Ans B Most common cardiac defect in Down syndrome is Endocardial cushion defect or Atrioventricular septal defect

95. Down syndrome in advanced maternal age is most commonly due to:
A. **Non disjunction**
B. Translocation
C. Mosaicism
D. Deletion

Ans A

96. Down syndrome most commonly occurs due to:
A. Reciprocal translocation
B. **Non-disjunction in maternal meiosis**
C. Translocation defect
D. Non-disjunction in paternal meiosis

Ans B

97. A Down syndrome child is mentally retarded. All cytogenetic abnormalities may occur except?
A. **Deleted chromosome**
B. Trisomy
C. Robertsonian translocation
D. Mosaic

Ans A 21 An extra copy of chr 21 is required to cause trisomy 21 (Down syndrome), so deletion of chr 21 will not cause it.

98. A Down syndrome patient is posted for surgery, the necessary preoperative investigation to be done is:
A. **Echocardiography**
B. CT brain
C. X-ray cervical spine
D. USG abdomen

Ans A > c. X-ray cervical spine Congenital heart diseases are more common in Down syndrome (in 50%), than atlantoaxial instability (20%); so a preoperative echocardiogram is warranted, so that management of child can be planned accordingly.

99. All of the following are features of Down syndrome except:
A. **Increased PAPPA**
B. Increased free beta HCG levels
C. Absent nasal bone
D. Abnormal ductus venosus flow velocity

Ans A Decreased PAPPA (pregnancy associated plasma protein A) level is used in 1st trimester screening for Down syndrome. M neMonic
• Levels of all biochemical markers decrease in Down syndrome except 'HI' that increases:
• So level of HCG and Inhibin increase, while, Level of α-fetoprotein, unconjugated estriol & PAPPA decrease

100. True about Down syndrome are all except:
A. Mental retardation
B. Hypotonia
C. **Coarctation of aorta is the most common cardiac lesion**
D. Epicanthal folds

Ans C

101. In Down syndrome, which of the following are seen?
A. **Sandal gap**
B. Antimongoloid slant
C. Clinodactyly
D. Hypotonia
E. Hypertonia

Ans A Sandle gap, c. Clinodactyly, d. Hypotonia

102. Down syndrome predisposes to which cancer:
A. **AML**
B. CML
C. ALL
D. CLL

Ans A & c. ALL • Acute leukemia occurs 15-20 times more frequently in children with Down syndrome than in general population.
• The ratio of ALL to AML in patients with Down syndrome is the same as that in the general population.
• The exception is during the 1st 3 yr of life, when AML is more common.

103. **Triple test for diagnosis of Down syndrome includes all of the following except:**

A. HCG
B. Alfa Fetoprotein
C. **Serum Prolactin level**
D. Serum estriol level

Ans C

104. **Infants with Down syndrome have all except:**

A. VSD
B. Duodenal atresia
C. Leukemia
D. **Normal intelligence**
E. Delayed skeletal maturation

Ans D

105. **Down syndrome is associated with:**

A. **Congenital heart disease**
B. ALL
C. Early onset Alzheimer's disease
D. CNS tumors
E. Infection

Ans A b. ALL, c. Early onset Alzheimer's disease, e. Infection Frequent infections (sinusitis, nasopharyngitis, pneumonia) are seen in Down Syndrome

106. **Commonest cause of intestinal obstruction in Down syndrome is:**

A. Colonic atresia
B. Intestinal atresia
C. **Duodenal atresia**
D. Esophageal atresia

Ans C • Most common cause of Intestinal obstruction in Down's syndrome is duodenal atresia.

107. **All are seen in Down syndrome except:**

A. Hypertonia
B. Simian crease
C. Lymphedema
D. **Mongoloid slant**
E. Anti-mongoloid slant

Ans D Respiratory tract infection uncommon

108. **All the following are recognized gastrointestinal features of Down syndrome in neonatal period EXCEPT**

A. annular pancreas
B. tracheoesophageal fistula
C. **diaphragmatic hernia**
D. imperforate anus
E. neonatal cholestasis

Ans C It also includes duodenal atresia and Hirschsprung disease.

109. **The MOST important reason for chromosomal analysis in persons suspected of having Down syndrome is to**

A. confirm clinical diagnosis
B. detect mosaicism
C. reassure the family
D. **detect translocations**
E. detect rare cases of ring chromosome

Ans D Chromosome analysis is indicated in every person suspected of having Down syndrome. If a translocation is identified, parental chromosome studies must be performed to determine whether one of the parents is a translocation carrier, which carries a high recurrence risk for having another affected child.

110. **A highly educated young couple has their first baby with Down syndrome; they are asking you about his development. Of the following, the MOST appropriate answer is**

A. programs of stimulation will achieve eventual normal development
B. motor development is the mostly affected domain with fair cognitive delay
C. developmental delay will be in the initial first few years, but it will normalize later on
D. developmental delay is universal, all domains of development almost equally affected
E. **developmental delay is universal, social development is relatively spared but he will have problems with expressive language**

Ans E Developmental delay is universal. Cognitive impairment does not uniformly affect all areas of development. Social development is relatively spared, but children with Down syndrome have considerable difficulty using expressive language. Understanding these individual developmental strengths will maximize the educational process for children with Down syndrome. Persons with Down syndrome often benefit from programs aimed at stimulation, development, and education.

111. **You are explaining the motor development to a mother with a baby with Down syndrome, you told her that there is a wide range of time to achieve his milestone and as example the baby can walk between**

A. 10-18 mo
B. 12-24 mo
C. **12-45 mo**
D. 15-50 mo
E. 18-60 mo

Ans C

112. **A 5-year-old child with Down syndrome complaining of intermittent symptoms of torticollis, weakness of the lower limbs and gait disturbances. Of the following, the NEXT important step is to**

A. reassure the family
B. **send for radiological assessment of the neck**
C. send for thyroid antibodies
D. send for radiological assessment of the airway
E. send for thyroid function test

Ans B The incidence of atlantoaxial sublaxation or instability in Down syndrome is about 10-30%, affected patient should not allowed to participate in high risk sports as include diving starts in swimming, butterfly stroke, diving, pentathlon, high jump, equestrian sports, gymnastics, football, soccer, alpine skinning and warm up exercises placing stress on the head and neck

113. Quad screen is the four maternal serum tests that should be done for all pregnant women in second trimester as a screening for Down syndrome, which includes the following tests EXCEPT

A. free β-human chorionic gonadotropin
B. unconjugated estriol
C. **androsterone**
D. inhibin
E. α-fetoprotein

Ans C is generally considered to be an inactive metabolite of testosterone, and it is not related to quad test.

Turner & Klinefelter Syndromes

114. Atavism means child resembles with his:

A. Father
B. Siblings
C. **Grand parents**
D. Neighbour MENDELIAN DISORDERS

Ans C Atavism means the appearance of a characteristic is presumed to have been present in some remote ancestor due to chance recombination of genes or environmental conditions favourable to their expression in the embryo.

115. The following is not a feature of Pierre-Robin syndrome:

A. Hearing defect
B. **Coloboma Iris**
C. Respiratory distress
D. Mandibular hypoplasia

Ans B J Laryngol Otol 1996 Sep;110(9):830-5; Pierre Robin syndrome Components • It consists of micrognathia usually accompanied by a high arched or cleft palate
• Tongue is usually of normal size, but the floor of the mouth is foreshortened (mandibular hypoplasia)
• The air passages can become obstructed, particularly on inspiration (respiratory distress) Associations • 30-50% of these patients have Stickler syndrome, an autosomal dominant condition with prominent joints,

116. Nuchal thickness more than _____ mm at 11-14 weeks of pregnancy is suggestive of Down's syndrome:

A. 2
B. **3**
C. 4
D. 5

Ans B 3 Nuchal pad thickening is > 3 mm at 11-14 wk or > 6 mm at 15-20 wk Important causes: Cystic hygroma, Trisomy 21, Trisomy 18, Turner syndrome (45, XO), Normal (~25%).

117. Transient Myeloproliferative disorders occur with increased incidence in children with:

A. **Down syndrome**
B. Turner's syndrome
C. Ataxia telangiectasia
D. Neurofibromatosis

Ans A Transient Myeloproliferative disorders in Down syndrome
• 10% of children with Down syndrome may develop a transient leukemia or myeloproliferative syndrome with high leukocyte counts, blast cells in peripheral blood, anemia, thrombocytopenia and hepatosplenomegaly
• 20–30% of these will develop typical leukemia (often acute megakaryocytic leukemia) by 3 yr of life
• GATA1 mutations (a transcription factor that controls megakaryopoiesis) are present in blasts from patients with Down syndrome who

118. Which of the following is true about 45,XO?

A. Webbed neck has 10 times more risk of congenital heart disease
B. If webbed neck is present, coarctation of aorta is absent
C. **Male Turner (Noonan) has higher risk of cardiac diseases**

Ans C pathogenic association in 45 X-O Turner syndrome? Teratology 29(3):355-61 dna Discussing about options one by one, a. False; Incidence of congenital heart disease has been found to be 30% in those with webbed neck and 9% of those with normal eck (p value < 0.0005). b. False c. True; Cardiac disease is seen in almost 80% of individuals with Noonan syndrome; Noonan syndrome is one of the most common genetic diseases associated with congenital heart defect, being second for frequency only to Down syndrome. d. False;

119. Turner syndrome is associated with which of the following cardiac abnormality?

A. Atrial septal defect
B. Ventricular septal defect 130.
C. Patent ductus arteriosus
D. **Coarctation of aorta**

Ans D

120. 136. In Turner syndrome which of the following is not seen:

A. **Short stature**
B. Widely spaced nipple
C. Webbed neck
D. Mental retardation

Ans A 5p Cri-du-chat syndrome occurs due to deletion on chr 5p15.2 "Cat-like" cry, microcephaly, growth deficiency, intellectual disability and congenital heart disease are seen.

121. Klinefelter's syndrome is associated with:

A. 47, XYY
B. 45, XO
C. 46, XY
D. **47, XXY**

Ans D

122.Not a feature of Edward syndrome:

A. **Long neck**
B. Low set ears
C. Rocker bottom foot
D. Renal malformations

Ans A

123.NARP is a:

A. Lipid storage disorder
B. Glycogen storage disorder
C. **Mitochondrial disorder**
D. Lysosomal storage disorder GENOMIC IMPRINTING & UNIPARENTAL DISOMY

Ans C GENOMIC

124.Karyotyping is done in which phase of cell cycle:

A. Anaphase
B. **Metaphase**
C. Telophase
D. S phase

Ans B

125.Predispostional genetic testing is more useful in

A. numerical chromosomal disorders
B. **multifactorial disorders**
C. single gene disorders
D. structural chromosomal disorders
E. pre arrangement for gene therapy

Ans B In a disease cause by many genes and affected by environmental factors, controlling of environmental factors may reduce disease progression as in cigarette smoking and α1-antitrypsin deficiency, familial breast cancer and mammography.

126.The following are a triplet repeat expansion disorders EXCEPT

A. fragile X syndrome
B. myotonic dystrophy
C. Huntington disease
D. spinocerebellar ataxias
E. **Kearns-Sayre syndrome**

Ans E Represent mitochondrial disorder.

127. All the following are recognized features of Turner syndrome EXCEPT

A. female phenotype
B. male phenotype
C. hypothyroidism
D. **type I diabetes mellitus**
E. gonadal dysgenesis

Ans D Male phenotype can occur with mosaic type of Turner syndrome, and type II insulin resistance is the true answer.

Muscular Dystrophies

128.Discrete nodule in the iris called as lisch nodule representing melanocytic hamartomas of the iris seen most commonly in:

A. Tuberous sclerosis
B. **Neurofibromatosis**
C. Incontinentia Pigmenti
D. I cell disease

Ans B

129.An affected male infant born to normal parents could be an example of all of the following except:

A. **An autosomal dominant disorder**
B. An autosomal recessive disorder
C. A polygenic disorder
D. A vertically transmitted disorder

Ans A with In autosomal dominant disorder, parents of an affected child cannot be normal, as, even if a parent has 1 copy of defective gene, parent will be diseased.

130.Which of the following syndromes is due to non-disjunction?

A. Edward
B. **Down**
C. Patau
D. Klinefelter

Ans B 95% cases of Down Syndrome are due to Non-disjunction

131. All of the following may occur in Down's syndrome except:

A. Hypothyroidism
B. **Undescended testis**
C. Ventricular septal defect
D. Brushfield's spots

Ans B

132.Cystic hygroma is found in:

A. Klinefelter's syndrome
B. Down's syndrome
C. Edward's syndrome
D. **Turner's syndrome**

Ans D

133.A child presents with congenital cyanotic heart disease and polydactyly. Likely diagnosis is:

A. **Ellis-van Creveld syndrome**
B. Rubinstein- Taybi syndrome
C. McKusick – Kaufman syndrome
D. Edward syndrome 150.

Ans A Discussing about the options, one by one, a. Ellis-van Creveld syndrome Also called 'chondroectodermal dysplasia' Chondrodystrophy, polydactyly, ectodermal dysplasia, CHD (Single atrium, VSD) seen b. Rubinstein- Taybi syndrome Microcephaly, ptosis, beaked nose with low-lying philtrum, broad thumbs and large toes, intellectual disability , VSD c. McKusick – Kaufman also called Cartilage-hair hypoplasia; Skeletal defects involving legs & rib cage; Immunodeficiency, celiac

134. **Not seen in William syndrome?**
A. Elfin facies
B. **Subvalvular aortic stenosis**
C. Hypercalcemia
D. Hypertension

Ans B Williams syndrome classically causes SUPRAvalvular (not subvalvular) aortic stenosis, along with elfin facies and hypercalcemia; mild hypertension is also a recognized feature.

135. **Patau syndrome feature include all except:**
A. Cleft lip
B. Hypotelorism
C. Holoprosencephaly
D. **Rocker bottom foot**

Ans D Rocker bottom foot is a feature of Trisomy 18, not trisomy13

136. **A child with a small head, minor anomalies of the face including a thin upper lip, growth delay, and develop- mental disability can have all of the following, except:**
A. A chromosomal syndrome
B. A teratogenic syndrome
C. A Mendelian syndrome
D. **A polygenic syndrome**

Ans D The given child has microcephaly, thin upper lip & growth & developmental delay. These features can be seen in: a. A chromosomal syndrome like Tetrasomy 12p (Pallister-Killian syndrome) b. A teratogenic syndrome like fetal alcohol syndrome c. A mendelian syndrome like Rubinstein-Taybi syndrome Hence, a polygenic syndrome is least likely in the given scenario In a polygenic disease, multiple genes contribute to the disease like hypertension & diabetes

137. **Which of the following hormones are raised in Prader-Willi syndrome?**
A. Growth Hormone (GH)
B. Leuteinizing Hormone (LH)
C. Follicle Stimulating Hormone
D. **Ghrelin**

Ans D Prader-Willi syndrome:
• It is caused by lack of expression of genes on the paternally inherited chromosome 15q11.2-q13
• In infancy it is characterized by hypotonia with poor suck resulting in failure to thrive
• As the child ages, developmental delay, cognitive disability, behavior problems and obesity become evident
• Hypothalamic dysfunction may lead to hyperphagia, temperature instability, high pain threshold, sleep disorders

138. **Microarray is:**
A. **Study of multiple genes**
B. Study of diseases
C. Study of organisms
D. Study of blood group

Ans A

139. **A 4-year-old female child, a known case of ALL, she is developing an increasing toxicity to methotrexate therapy. Of the following, the MOST useful test is**
A. homocysteine level
B. prothrombin test
C. factor V leiden
D. **MTHFR DNA testing**
E. folate level

Ans D Knowledge of individual genotypes (pharmacogentic genetic testing) will guide pharmacologic therapy, allowing customization of choice of drug and dosage to avoid toxicity and provide a therapeutic response. An example of this is testing for polymorphisms within the methylenetetrahydrofolate reductase (MTHFR) gene for susceptibility of potentially increased toxicity to methotrexate antimetabolite therapy for treatment of acute lymphoblastic leukemia.

140. **The observation of increasing severity of the disease and early age of onset in subsequent generation is called**
A. genetic imprinting
B. **genetic anticipation**
C. uniparental diasomy
D. non expressiveness
E. non penetrance

Ans B

Hemoglobinopathies & Thalassemias

141. **Lisch Nodule is seen in:**
A. **Neurofibromatosis type**
B. Neurofibromatosis type
C. Tuberous sclerosis
D. MEN

Ans A

142. **The chances of having an unaffected baby, when both parents have achondroplasia are:**
A. 0%
B. **25%**
C. 50%
D. 100%

Ans B • Achondroplasia is an autosomal dominant disorder:
• If both parents have Achondroplasia, chances of a child being affected with Achondroplasia are 75%

143. **Mutation leading to sickle cell anemia:**
A. Crossover mutation
B. Frameshift
C. Deletion
D. Non-disjunction
E. **Point mutation**

Ans E

144. **Transient myeloproliferative disorder of newborn is seen in association with:**
A. Turner syndrome
B. **Down syndrome**
C. Neurofibromatosis
D. Ataxia telangiectasia

Ans B

145. **Streak ovaries are seen in which one of the following syndrome?**
A. Triple X
B. Klinefelter's
C. **Turner**
D. Swyer's

Ans C

146. **Which of the following is true about Treacher Collins syndrome:**
A. Autosomal recessive
B. **First and second arch affected**
C. Dental structure preserved
D. Hyperplastic mandible

Ans B Treacher Collins syndrome is a congenital disorder involving structures developing from the first and second branchial arches.

147. **The disorder in Patau syndrome involves which of the following?**
A. Chromosome
B. Chromosome
C. **Chromosome**
D. Chromosome

Ans C

148. **Edward syndrome is:**
A. Trisomy
B. **Trisomy**
C. Trisomy
D. Trisomy

Ans B

149. **A baby presenting with multiple deformities, cleft lip, cleft palate, microcephaly, small eyes, scalp defect and polydactyly, seen in which syndrome:**
A. **Trisomy**
B. Trisomy
C. Trisomy
D. Monosomy

Ans A Trisomy 13 140 above

150. **Angelman syndrome is due to:**
A. Digenic inheritance
B. Inversion
C. **Uniparental disomy**
D. Mitochondrial disorder

Ans C

151. **What is the diagnostic test you would do for a P2L2 pregnant female at 18 weeks of gestation having a history of 1 baby with thalassemia?**
A. CVS
B. NIPT
C. Amniocentesis
D. **Cordocentesis**

Ans D • Thalassemia major is caused by numerous mutations, prenatal diagnosis is difficult
• Prior identification of the familial mutation is required to offer prenatal diagnosis In the given scenario, the lady has already 1 child with thalassemia, so both she & her husband are carriers; As she has presented late in pregnancy (18 weeks), CVS or Amniocentesis cannot be done. Hence, Cordocentesis is the investigation of choice.

152. **Enzyme replacement therapy are available for the following disorders EXCEPT**
A. **Wolman disease**
B. Gaucher disease
C. Fabry disease
D. Mucopolysaccharidosis type VI
E. Pompe disease

Ans A

153. **The following genetic scenarios are possible to explain Angelman syndrome EXCEPT**
A. inheriting both chromosomes 15 from the father
B. deletion of the maternal 15q12
C. mutation in an imprinted gene
D. mutation in the imprinting center
E. **genetic anticipation**

Ans E The first statement is called uniparental disomy when both copies of a chromosome is inherited from one parent, the second call genetic imprinting when one gene copy is silenced in one side; either in mother or father resulting in each situation in different syndromes, third and fourth distracters is possible but are uncommon causes.

Cystic Fibrosis

154. **Hypophosphaemic Vit D Resistant Rickets is?**
A. AD
B. AR
C. **XD**
D. XR

Ans C

155. **For a normal husband and wife the first child was diagnosed to have cystic fibrosis. What is the percentage of chances for the second child be affected?**
A. **25**
B. 50 (PGI June 06)
C. 0
D. 75
E. 100

Ans A 25 • Cystic fibrosis is an autosomal recessive disorder
• If husband and wife are clinically normal and the child has developed the disease that means both husband and wife are carriers of cystic fibrosis
• If both father & mother are carriers of an autosomal recessive disorder, each child has 25% chances (1 in 4) of being affected by the disease

156. **In Downs syndrome there is:**

A. Translocation
B. Mutations
C. Paternal nondisjunction
D. **Maternal nondisjunction**

Ans D • In trisomy 21, the additional number 21 chromosome is derived from mother in 95% of cases
• It is due to non-dysjunction of 21st chromosome at the time of meiosis
• In non-dysjunction, chromosomal pair fails to separate in 1st meiotic division during gametogenesis.

157. **Increased nuchal fold thickness is a feature of:**

A. Paul-Bunnell syndrome
B. DiGeorge syndrome
C. **Down syndrome**
D. Cri-du chat syndrome

Ans C

158. **Edema of hands and feet in infants is characteristic of:**

A. Klinefelter's syndrome
B. Noonan syndrome
C. **Turner syndrome**
D. Fragile-X syndrome

Ans C

159. **Which of the following about Pierre Robins syndrome is 151. false?**

A. **Abdominal muscles are absent**
B. Floor of mouth is shortened 152.
C. Prone position is relieving
D. Achieves normal growth in 5 years of life

Ans A Pierre Robin syndrome
• It consists of micrognathia usually accompanied by a high arched or cleft palate.
• The tongue is usually of normal size, but the floor of the mouth is foreshortened.
• The infant should be maintained in a prone position to relieve respiratory obstruction.
• In some children, the jaw may achieve normal growth by 4-6 years of age

160. **F body is?**

A. **Y chromatin**
B. X chromatin
C. Chromosome
D. Chromosome

Ans A Quinacrine, a fluorescence dye, binds strongly to Y chromosome forming a bright fluorescent spot (F body). This is clearly visible in stained interphase cells from various tissues from human male and in mature spermatozoa

161. **Which one of the following is a distinguishing feature of Edward's syndrome?**

A. Hypotonia
B. Hypotelorism
C. Holoprosencephaly
D. **Rocker bottom feet**

Ans D

162. **Most common chromosomal syndrome is:**

A. Fragile X-syndrome
B. Trisomy
C. **Trisomy**
D. Trisomy 13 92 Section 1: General Pediatrics
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Ans C 21 • Down syndrome (Trisomy 21) is the most common of the chromosomal disorder.
• Trisomy 16 is the most common trisomy overall, however most do not survive and die in utero Hence, among the live births, or among children, most common chromosomal abnormality is Trisomy 21

163. **All of the following are associated with uniparental disomy except:**

A. Prader willi syndrome
B. **Bloom syndrome**
C. Angelman syndrome
D. Russell silver syndrome

Ans B

164. **Which of the following procedures is routine technique for karyotyping using light microscopy:**

A. C-banding
B. **G-banding**
C. Q-banding
D. Brd V-staining GENE THERAPY

Ans B G-banding which includes staining with Giemsa is the most common technique used for karyotyping.

165. **DiGeorge syndrome is an example of**

A. **contiguous gene disorders**
B. single gene disorders
C. mitochondrial inheritance disorders
D. numeric chromosomal disorders
E. imprinting disorders

Ans A

166. **The MOST common genetic cause of Prader-Willi syndrome is**

A. inheriting both chromosomes 15 from the mother
B. deletion of the maternal 15q12
C. **deletion of the paternal 15q12**
D. mutation in an imprinted gene
E. mutation in the imprinting center

Ans C In about 70% of cases missing paternal 15q12 lead to what is called genomic imprinting. The same clinical effect if the child inherted the same chromosome (15) from the mother (25-29%). In Angelman syndrome, paternal UPD of chromosome 15 is rarer and is observed in approximately 5% of the cases (missing the maternal chromosome 15).

Inborn Errors & Metabolic Disorders

167. Abnormal color vision is 20 times more common in men than women because most cases are caused by an abnormal:

A. Dominant gene on Y chromosome
B. Recessive gene on Y chromosome
C. Dominant gene on X chromosome
D. **Recessive gene on X chromosome**

Ans D /e p 643
• Red and green cone pigments are encoded on the X chromosome, and the blue cone pigment on chromosome 7.
• Mutations of the red and green pigments cause congenital X-linked color blindness in 8% of males
• Because color blindness is almost exclusively X-linked, it is worth screening only male children.

168. All of the following are chromosomal breakage syndromes except:

A. Fanconi's anemia
B. **Ehler-Danlos syndrome**
C. Bloom syndrome
D. Ataxia telangiectasia

Ans B Chromosome instability syndromes (previously called chromosome breakage syndromes)
• Disorders resulting from specific defects in DNA repair, cell cycle control and apoptosis with autosomal recessive inheritance → chromosomal instability → increased risk of developing neoplasms
• E.g. Fanconi anemia, ataxia telangiectasia, Nijmegen syndrome, ICF (immunodeficiency, centromere instability, and facial anomalies) syndrome, Roberts syndrome, Werner syndrome and Bloom syndrome.

169. All the following are biochemical serum markers in second trimester screening of Down's syndrome except:

A. **PAPPA**
B. Beta HCG
C. Inhibin
D. Unconjugated oestriol

Ans A PAPPA is used for screening for Down syndrome in 1st trimester

170. stenosis, short stature and undescended testis. The likely diagnosis is:

A. Hypoparathyroidism
B. **Noonan syndrome**
C. Klinefelter syndrome
D. XYY sex chromosomes

Ans B Prader Willi syndrome

171. Which of the following feature is not seen in Prader Willi syndrome?

A. Obesity
B. Mental retardation
C. **Ataxia**
D. Hypotonia

Ans C

172. Patau's syndrome is due to:

A. Trisomy
B. Trisomy
C. Trisomy
D. **Trisomy I3**

Ans D Trisomy

173. Common features of Marfan syndrome & Homocystinuria are:

A. **Arm span > Height**
B. Ectopia lentis
C. Hypermobility of joint
D. Mental retardation
E. Arachnodactyly

Ans A Arm spam > Height, b. Ectopia lentis, c. Hypermobility of joint and e. Arachnodactyly For details about Homocystinuria, Lens dislocation in Marfan syndrome is Supero-lateral (Remember: "M-S-L"), while in Homocystinuria it is Infero-nasal. In homocystinuria, skeletal abnormalities resembling those of Marfan syndrome seen like tall stature, arachnodactyly, scoliosis, pectus excavatum or carinatum, genu valgum, pes cavus, high-arched palate, & crowding of the teeth are seen and progressive

174. Which of the following is a trinucleotide repeat disorder?

A. Duchenne muscular dystrophy
B. **Becker muscular dystrophy**
C. Limb girdle dystrophy
D. Myotonic dystrophy

Ans B Myotonia dystrophica

175. Cordocentesis is done in which time of pregnancy

A. **18-20 weeks**
B. 14-16 weeks
C. 12-14 weeks
D. 8-10 weeks

Ans A

176. When an individual is at-risk genotype and not clinically express the condition; this phenomena is called

A. genetic imprinting
B. genetic anticipation
C. uniparental diasomy
D. non expressiveness
E. **non penetrance**

Ans E

177. The MOST important screening tool for genetic disorders is

A. genetic study
B. **family history**
C. karyotyping
D. prenatal diagnosis
E. preimplantation genetic screen

Ans B The family history remains the most important screening tool for pediatricians in identifying a patient's risk for developing a wide range of diseases, from multifactorial conditions, such as diabetes and attention-deficit disorder, to single-gene disorders such as sickle cell anemia and cystic fibrosis.

178. **Chromosomal analysis is indicated in all the following EXCEPT**

A. **recurrent premature deliveries**
B. postnatal problems in growth and development
C. unexplained intellectual disability
D. primary amenorrhea
E. recurrent miscarriages (≥3)

Ans A The specific indications for studies include advanced maternal age (>35 yr) or multiple abnormalities on fetal ultrasound (prenatal testing), multiple congenital anomalies, unexplained growth restriction in the fetus or postnatal problems in growth and development, ambiguous genitalia, unexplained intellectual disability with or without associated anatomic abnormalities, primary amenorrhea or infertility, recurrent miscarriages (≥3) or prior history of stillbirths and neonatal deaths, a 1st- degree relative with a...

Neurocutaneous Syndromes

179. **Testicular feminization syndrome is:**

A. XX
B. **XY**
C. XYY
D. XXY

Ans B XY Testicular feminization syndrome
• It is also called the complete androgen insensitivity syndrome (AIS);
• XY fetuses with this disorder are insensitive (unresponsive) to androgens; So they are born looking externally like normal girls; Internally, there is a short blind-pouch vagina & no uterus, fallopian tubes or ovaries
• There are testes in abdomen or inguinal canal; AIS is the most common cause of male DSD.

180. **MC congenital heart defect in Down's syndrome:**

A. Patent ductus arteriosus
B. Atrial septal defect
C. **Atrioventricular septal defect**
D. Ventricular septal defect

Ans C Atrioventricular septal defect or Endocardial cushion defect is the most common congenital heart disease in Down syndrome.

181. **Simian crease is not seen in:**

A. Cognitive impairment
B. Delayed dentition
C. Recurrent chest infection
D. **Constipation**

Ans D Noonan synd rome Important information about Simian crease:
• The term "simian crease" is not used much anymore, since it tends to have a negative meaning (the word "simian" refers to a monkey or ape); The crease is usually just referred to as a single transverse palmar crease.
• A simian crease is a single line that runs across the palm of the hand. Conditions in which similar crease is seen:
• Down syndrome • Fetal alcohol syndrome • Rubella syndrome
• Pseudohypoparathyroidism

182. **micrognathia, Tripod skull and fifth finger clinodactyly:**

A. **Russell Silver syndrome**
B. Beckwith–Wiedemann syndrome
C. Angelman syndrome
D. Prader–Willi syndrome

Ans A Gynecomastia with long thin limbs Given Karyotype is 47 XXY → Kilnefelters syndrome with Gynecomastia with long thin limbs as expected findings.

183. **Which of the following is seen in Noonan syndrome with pulmonic stenosis?**

A. Chromosomal aberration
B. Coarctation of aorta
C. Pulmonary lymphangiectasia
D. **Abnormal intelligence**

Ans D

184. **Absence of clavicle is a feature of:**

A. **Cleidocranial dysplasia**
B. Multiple epiphyseal dysplasia
C. Fibrous dysplasia
D. Cranio-Metaphyseal dysplasia

Ans A Cleido cranial dysplasia Cleidocranial dysostosis, also called cleidocranial dysplasia or mutational dysostosis, is a hereditary congenital disorder, where there is delayed ossification of midline structures; Absent clavicles and delayed dentition.

185. **An infant with cleft lip, cleft palate, polydactyly, micro-cephaly with holoprosencephaly, ectodermal scalp defect is suffering from:**

A. Trisomy
B. Trisomy
C. **Trisomy**
D. Turner syndrome

Ans C

186. **Which is the most common genetic cause of mental retardation:**

A. Tuberous sclerosis
B. **Fragile-X-syndrome**
C. Cri-du-chat syndrome
D. Angelman's syndrome

Ans B

187. **For karyotyping, the dividing cells are arrested by the addition of colchicines in the following mitotic phase**

A. Prophase
B. **Metaphase**
C. Anaphase
D. Telophase

Ans B Chromosomes are examined after arresting dividing cells in metaphase with mitotic spindle inhibitors (e.g., N-diacetyl- N-methylcolchicine), followed by staining.

188. **The genetic testing in an asymptomatic child with positive family history of Huntington chorea is called**

A. predisposional testing
B. **predictive testing**
C. diagnostic testing
D. pharmacogentic testing
E. linkage testing

Ans B

189. **The risk of a genetic disorder for the offspring of a first-cousin marriage (consanguinity marriage) is**

A. 3-5%
B. **6-8%**
C. 9-11%
D. 12-14%
E. 15-17%

Ans B The risk of a genetic disorder for the offspring of a first-cousin marriage (6-8%) is about double the risk in the general population (3-4%).

190. **Chromosomal abnormalities in live births constitute around**

A. **1-2%**
B. 3-4%
C. 5-6%
D. 7-8%
E. 9-10%

Ans A Clinical cytogenetics is the study of chromosomes: their structure, function, inheritance, and abnormalities. Chromosome abnormalities are very common and occur in approximately 1-2% of live births, 5% of stillbirths, and 50% of early fetal losses in the 1st trimester of pregnancy.

Genetic Counseling & Screening

191. **A 4-year-baby is having large face, large jaw, large ear and macroorchidism. Diagnosis?**

A. Mc Cune Albright syndrome
B. Down's syndrome
C. Cri-du chat syndrome
D. **Fragile X syndrome**

Ans D

192. **Triple test includes all the following except:**

A. AFP
B. HCG
C. **Inhibin**
D. Estradiol

Ans C

193. **Thirteen pair of Ribs can be seen in all of the following except:**

A. **Down syndrome**
B. Holt-Oram syndrome
C. Turner syndrome
D. Incontinentia pigmenti

Ans A Thirteen pairs of ribs may be seen in:
• Aarskog syndrome • Turner syndrome • Holt-Oram syndrome
• Alagille syndrome • Incontinentia pigmenti • Fetal akinesia deformation sequence
• Idiopathic

194. **Hypoglycemia, jaundice, microphallus with absent septum pellucidum is characteristic of which syndrome?**

A. **De Morsier syndrome**
B. Silver–Russell syndrome
C. Kallmann syndrome
D. Kearns–Sayre syndrome

Ans A • Also called septo-optic dysplasia
• Underdevelopment of the optic nerve, pituitary gland dysfunction, and absence of the septum pellucidum
• Two of these features need to be present for a clinical diagnosis—only 30% of patients have all three.
• De Mosier first recognized the relation of a rudimentary or absent septum pellucidum with hypoplasia of the optic nerves and chiasm in 1956. with
• Intracranial malformations associated with septo-optic dysplasia include agenesis of septum pellucidum, schizencephaly, and

195. **Triangular facies is indicative of?**

A. **Double stranded break repair**
B. Base excision repair
C. Mismatch repair
D. Nucleotide excision repair

Ans A Russel Silver syndrome In Russell silver syndrome, short stature, triangular face (small jaw & pointed chin), & limb anomalies are seen; It is an example of genomic imprinting disorder involving chromosome 11; Triangular facies is also seen in Alagille syndrome, 145. Ans. a. Trisomy 18 Single umbilical artery
• It is seen in 5-10/1,000 births & 35-70/1,000 in twin births (more common in twins).
• 30% of these infants have congenital abnormalities; Trisomy 18 is one of the more frequent abnormalities.

196. **In Treacher Collins all are seen except:**

A. Coloboma of inf. eyelid
B. Mandibular angle obtuse
C. **Low intelligence**
D. Cleft palate

Ans C Treacher Collins syndrome Also called Mandibulofacial dysostosis Inheritance Autosomal dominant syndrome that primarily affects the face Identifying • Down-slanting eyes (anti-mongoloid slant) features • Drooping part of the lateral lower eyelids & coloboma of lower eyelids
• Micrognathia (small jaw) with underdeveloped & sunken cheek bones
• Malformed or absent ears &/or deafness
• Cleft palate; Teeth may be hypoplastic, or displaced
• Intelligence is usually normal Associated with Breathing and feeding problems

197. **Cat eye syndrome is:**

A. Partial trisomy
B. Partial trisomy
C. Partial trisomy
D. **Partial trisomy**

Ans D 22 Harrison's 19/e p 83e6 Cat eye syndrome: It is due to trisomy or tetrasomy of short arm & small part of long arm of chromosome 22. Features seen in cat Answers eye syndrome include:
• Ocular coloboma of iris, choroid or optic nerve • Renal malformation—U/L renal agenesis
• Preauricular skin tags or pits • Antimongoloid slant of eye
• Congenital heart disease • Microphthalmia with

198.Fragile X syndrome is associated with all except:

A. Large nose
B. Large face
C. Large ears
D. Large testis MITOCHONDRIAL INHERITANCE

Ans A

199.Karyotyping is useful in diagnosis of:

A. Autosomal recessive disorders
B. X-linked recessive disorders
C. Chromosomal abnormalities
D. Biochemical abnormalities

Ans C

200.A 5-year-old asymptomatic male child with strong positive family history of Huntington chorea, mother is highly concern about her child possible genetic affection, she asked for predictive genetic testing. Of the following, the MOST appropriate explanation is that

A. whatever the result it will not affect the disease process
B. this is an age dependent type of genetic disorder and may not manifest ever
C. leave the decision to the child when he is grown up
D. it may lead to discrimination
E. we should weigh the benefits VS risks of testing

Ans C In fact, all the mentioned explanations about the disease are true but it is generally agreed that predictive genetic tests should be performed for children if the results of the test will benefit the medical management of the child. Otherwise, the test should be deferred until the child has an understanding of the risks and benefits of testing and can provide informed consent.

201.The organ that can be spared in mitochondrial disorders is

A. brain
B. eye
C. small intestine
D. liver
E. muscle

Ans C The mitochondria are the cell's suppliers of energy, so the organs that are most affected by the presence ofabnormal mitochondria are those that have the greatest energyrequirements, such as the brain, muscle, heart, and liver. Common manifestations include developmentaldelay, seizures, cardiac dysfunction, decreased muscle strengthand tone, and hearing and vision problems.

Molecular Genetics & Mutations

202.Children heterozygous for having _____ {this is a genetic phenomenon) for KCNE2 gene (Potassium Channel voltage gated) have higher chance of having life threatening arrhythmias after administration of Erythromycin?

A. Gene Polymorphism
B. Anticipation
C. Ontogeny
D. Phylogeny

Ans A

203.Different mutations in the same genetic locus causing similar or identical phenotype is:

A. Phenotypic heterogeneity
B. Allelic heterogeneity
C. Locus heterogeneity
D. Non-allelic heterogeneity

Ans B • "Allelic Heterogeneity" refers to the fact that different mutations in the same genetic locus can cause an identical or similar phenotype. For example, many different mutations of the β-globin locus can cause β-thalassemia
• "Phenotypic Heterogeneity" occurs when more than one phenotype is caused by alletic mutations, e.g. Identical mutations in the FGFR2 gene can result in very distinct phenotypes: Crouzon syndrome, or Pfeiffer syndrome

204.Frameshift mutation occurs due to:

A. Penetrance
B. Inheritance
C. Co-dominance
D. Pleiotropism

Ans C Insertion A frameshift mutation is caused by a deletion or insertion in a DNA sequence that shifts the way the sequence is read.

205.Name the proposed mechanism where one gene (among two inherited genes) is turned off through environmentally induced modifications leading to fetal programming?

A. The one-two selection mechanism
B. Fetal programming service
C. Cytogenetic manipulation
D. Epigenetic imprinting

Ans D

206.Differential expression of same gene depending on parent of origin is referred to as:

A. Genomic imprinting
B. Mosaicism
C. Anticipation
D. Non-penetrance

Ans A

207.Single gene defect causing multiple unrelated problems:

A. Pleiotropism
B. Pseudodominance
C. Penetrance
D. Anticipation

Ans A Pleiotropism refers to single gene defect causing multiple traits.

208.Waardenburg syndrome is due to mutation of:

A. PAX2 gene
B. PAX3 gene
C. PAX6 gene
D. PAX9 gene

Ans B Waardenburg syndrome (WS):
• Autosomal dominant inherited condition due to mutations in EDN3, EDNRB, MITF, PAX3, SNAI2 or SOX10 genes

209. **The law "Relative frequencies of each gene allele tends to remain constant from generation to generation", was given by:**

A. Co-dominance
B. **AD**
C. AR
D. Mitochondrial inheritance

Ans B Hardy Weinberg Hardy-weinberg's law
• It describes a theoretical situation in which a population is undergoing no evolutionary change
• It explains that if the evolutionary forces are absent, the population is large and its individuals have random mating
• Thus each parent produces equal number of gametes
• Such gametes combine at random and the gene frequency remains constant. Finally the genetic equilibrium of the genes is maintained and the variability present in the population is preserved.

210. **All are mitochondrial disorders except:**

A. Pearson Syndrome
B. MERRF (Myoclonic Epilepsy Ragged Red Fibre)
C. MELAS (Mitochondrial Encephalopathy Lactic Acidosis and Stroke-like episode)
D. **Fragile X Syndrome**

Ans D Mitochondrial Disorders, include Leber Hereditary Optic Neuropathy, MELAS (Mitochondrial encephalomyopathy, lactic acidosis, and stroke like episodes), MERRF (Myoclonic epilepsy, ragged red fibers) in muscle, & Pearson syndrome

211. **Genomic imprinting is seen in:**

A. **Prader Willi syndrome**
B. Marfan syndrome
C. Down syndrome
D. Osteogenesis imperfect

Ans A Prader-Willi syndrome • Genomic imprinting occurs when the phenotypic expression of a gene depends on the parent of origin
• Classic examples: Prader-Willi syndromeQ and Angelman syndromeQ, Beckwith-Wiedemann syndromeQ, Russell- Silver syndromeQ, pseudohypoparathyroidism and neonatal diabetes.

212. **Known gene loci can be diagnosed by:**

A. **FISH**
B. Comparative gene hybridization
C. PCR
D. Chromosomal painting

Ans A FISH involves DNA probes that recognize sequences specific to particular chromosomal regions (diagnosis of known loci)

213. **Gene therapy is most commonly targeted against:**

A. **SCID**
B. cancer
C. cystic fibrosis
D. Leukemia

Ans A FDA-approved gene therapy is used for ADA-SCID, Leber's congenital amaurosis, multiple myeloma & Adrenoleukodystrophy;

214. **Which of the following disease can NOT be treated by Gene Therapy:**

A. Adenosine deaminase deficiency
B. **Leukemia**
C. Cystic Fibrosis
D. Thalassemia

Ans B Gene therapy has been found to be potentially useful in disorders like SCID (due to Adenosine deaminase deficiency), Cystic fibrosis, Thalassemia, not in leukemia (till now), where chemotherapy is required.

215. **Following are potential complications of gene therapy except:**

A. Genotoxicity
B. **Genome integration**
C. Gene silencing
D. Immunotoxicity

Ans B Problems with gene therapy especially with viral vectors include toxicity, immune and inflammatory responses.

216. **Which of the following techniques is used in gene therapy?**

A. **Electroporation (PGI 01)**
B. Electrofocusing
C. Selectively targeted recombination
D. Intranuclear injection Chapter 5: Genetics and Genetic Disorders 93 Answers

Ans A Electroporation, d. Intranuclear injection Gene therapy: An experimental technique that uses genes to treat or prevent disease. Delivery of DNA into cells, can be done using Recombinant viruses or 'viral vectors' or naked DNA or DNA complexes Methods used include: Injection, Electroporation, 'gene-gun', sonoporation, magnetofection, & use of oligonucleotides, lipoplexes or nanoparticles. 6 Chapter Inborn Metabolism When to Suspect a Metabolic Disorder in Neonates Infants?

217. **The phenomena of changing phenotypes (clinical presentations) associated with the same single-gene disorder is called**

A. **variable expressiveness**
B. variable penetrance
C. false positive
D. false negative
E. validity

Ans A

218. **The observation of significantly increasing number of imprinting disorders as Beckwith-Wiedemann and Angelman syndrome are MOST likely due to**

A. new methods of prenatal diagnosis (amniocentesis)
B. new technology of gene therapy
C. **assisted reproductive technology**
D. increase radiation exposure
E. high pressure electricity chains in large cities

Ans C Several studies suggest that there may be a small but significantly increased incidence of imprinting disorders, specifically Beckwith-Wiedemann and Angelman syndrome, associated with assisted reproductive technologies such as in vitro fertilization and intracytoplasmic sperm injection. However, the overall incidence of these disorders in children conceived using assisted reproductive technologies is likely to be <1%.

06

CHAPTER 06

Inborn Errors of Metabolism



WHAT THIS CHAPTER COVERS

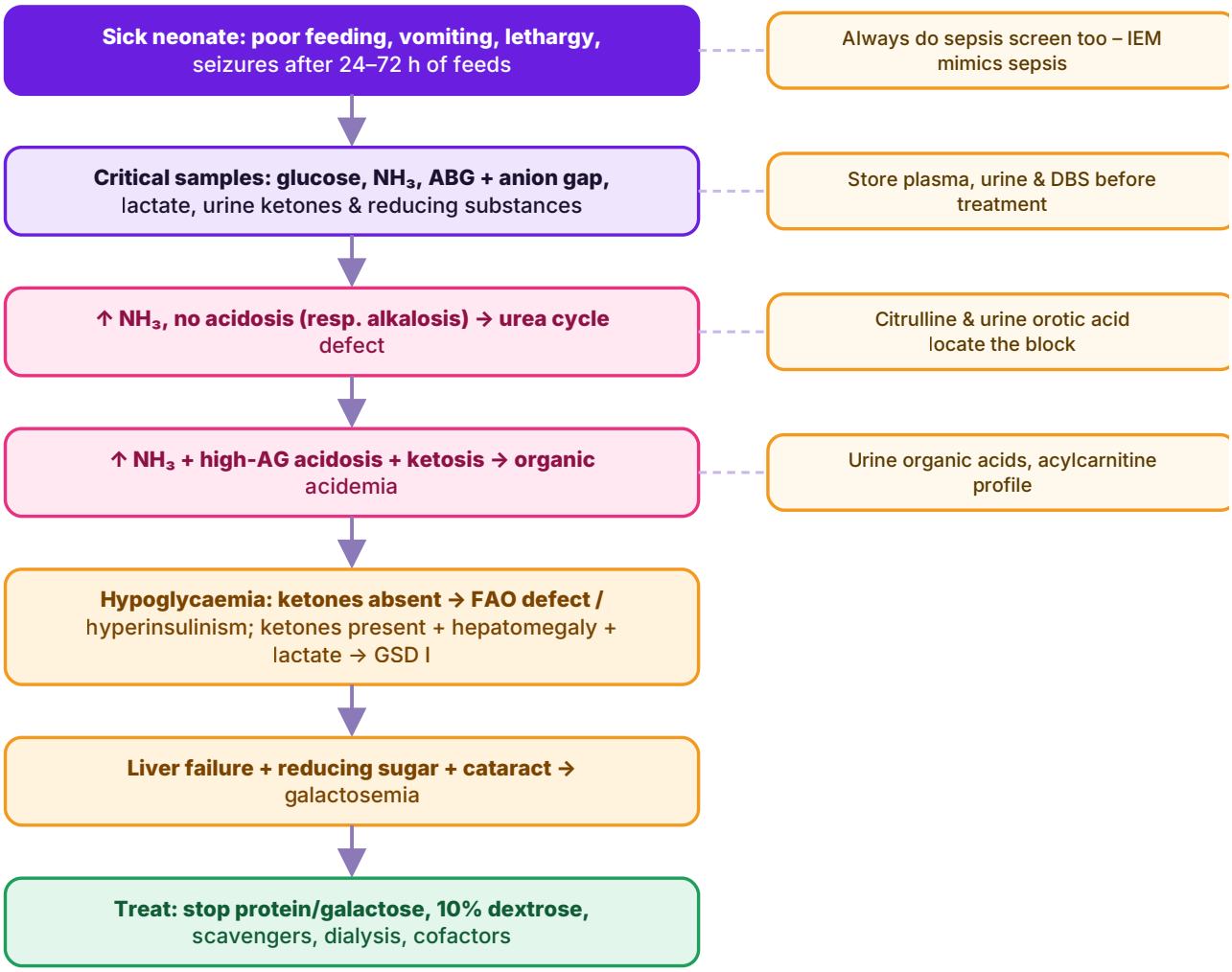
- Phenylketonuria (PKU)
- Galactosemia
- Maple Syrup Urine Disease
- Glycogen Storage Diseases
- Mucopolysaccharidoses
- Lipid Storage Disorders
- Amino Acid Disorders
- Organic Acidurias
- Urea Cycle Disorders
- Mitochondrial Disorders
- Fatty Acid Oxidation Disorders
- Newborn Screening

204 MCQs **12** subtopics

6.1 Approach to Inborn Errors of Metabolism (IEM)

- Mostly **autosomal recessive** (exceptions: **OTC deficiency – X-linked**; Hunter, Fabry – XLR; some porphyrias – AD); consanguinity, unexplained sib deaths.
- **Clue**: a well baby at birth who deteriorates after **24–72 h of feeding** (protein/galactose exposure) – poor feeding, vomiting, lethargy, seizures, coma – **mimics sepsis**.

Presentation group	Examples
Intoxication type (acute, after feeds)	**Urea cycle defects, organic acidurias (MMA, PA, IVA), MSUD**, galactosemia, hereditary fructose intolerance
Energy deficiency	Fatty-acid oxidation defects, mitochondrial disorders, GSD, gluconeogenesis defects
Complex molecule (storage) – slowly progressive	Lysosomal storage (MPS, sphingolipidoses), peroxisomal (Zellweger, X-ALD), CDG



First-line metabolic work-up in a sick neonate

Finding	Think of
Hyperammonaemia + respiratory alkalosis, no acidosis, no ketosis	**Urea cycle defect**
Hyperammonaemia + **high-AG metabolic acidosis + ketosis**	**Organic aciduria** (propionic, methylmalonic, isovaleric)

Finding	Think of
Hypoketotic hypoglycaemia	**Fatty-acid oxidation defect (MCAD)**, hyperinsulinism
Hypoglycaemia + lactic acidosis + **hepatomegaly**	**GSD type I (von Gierke)**, fructose-1,6-bisphosphatase deficiency
Liver failure + **reducing substances in urine** + E. coli sepsis + cataract	**Galactosemia**
Lactic acidosis + normal glucose, ↑ L:P ratio	Mitochondrial disorder, PDH deficiency
Intractable neonatal seizures	**Pyridoxine dependency**, NKH (↑ CSF:plasma glycine), molybdenum cofactor/ sulfite oxidase deficiency, GLUT1 deficiency (low CSF glucose), folinic-acid responsive

Abnormal urine/body odour

Odour	Disorder
Mousy / musty	**Phenylketonuria** (phenylacetate)
Maple syrup / burnt sugar	**MSUD** (sotolone)
Sweaty feet	**Isovaleric acidemia**, glutaric aciduria type II
Cat's urine	Multiple carboxylase deficiency (3-MCC)
Boiled cabbage / rancid butter	**Tyrosinaemia type I**, hypermethioninaemia
Rotting fish	**Trimethylaminuria**
Swimming pool (chlorine)	Hawkinsinuria

EMERGENCY MANAGEMENT OF SUSPECTED IEM

Stop protein & galactose feeds, give **10% dextrose at 1.5× maintenance** (glucose infusion rate 8–10 mg/kg/min) ± insulin to stop catabolism, correct acidosis, **ammonia scavengers – sodium benzoate, sodium phenylbutyrate/phenylacetate, L-arginine** (for urea cycle defects; arginine contraindicated in arginase deficiency), **carnitine** (organic acidurias), **haemodialysis** if NH_3 > 400–500 µmol/L or rising; cofactors (biotin, B12, B6, thiamine) empirically. Collect **critical samples before treatment**: blood glucose, ammonia, lactate, ABG, plasma amino acids, acylcarnitine profile (dried blood spot), urine organic acids, urine ketones & reducing substances.

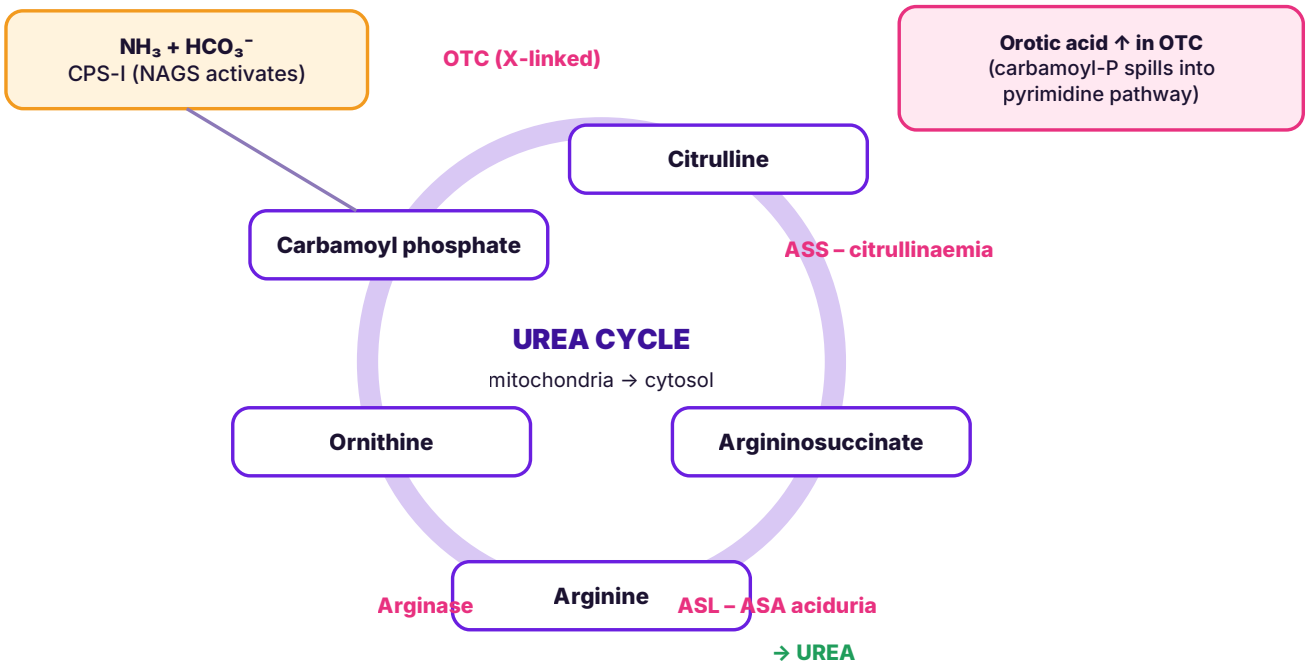
6.2 Phenylketonuria (PKU)

- **Phenylalanine hydroxylase (PAH) deficiency** (chr 12, AR) → Phe can't convert to **tyrosine**. Cofactor **tetrahydrobiopterin (BH4)** deficiency (1–3%) – **malignant PKU** (also ↓ dopamine & serotonin; needs L-dopa, 5-HTP, BH4).
- Normal at birth (maternal clearance) → **ID (most important), seizures, eczema, fair skin/hair & blue eyes (↓ melanin), mousy odour**, hyperactivity, microcephaly.
- Screening: **newborn screen after 48–72 h of protein feeding** – **Guthrie bacterial inhibition test** (historic), now **tandem mass spectrometry (Phe:Tyr ratio)**; **ferric chloride test** – green colour.
- Rx: **Phe-restricted diet for life** (target blood Phe **2–6 mg/dL / 120–360 µmol/L**); tyrosine becomes essential; **sapropterin (BH4)** in responsive cases; **pegvaliase** (adults). Avoid **aspartame**.
- **Maternal PKU**: uncontrolled Phe in pregnancy → **microcephaly, ID, CHD (TOF, CoA), IUGR** in a **non-PKU fetus** – strict control before conception.

6.3 Other Amino Acid Disorders

Disorder	Defect	Key features	Rx
Tyrosinaemia type I (hepatorenal)	**Fumarylacetoacetate hydrolase**	Acute liver failure, cirrhosis, **renal Fanconi (rickets)**, **succinylacetone in urine (diagnostic)**, porphyria-like crises, **↑↑ AFP**, high risk of **hepatocellular carcinoma**; cabbage odour	**Nitisinone (NTBC)** + low Tyr/Phe diet; liver transplant
Tyrosinaemia type II (Richner–Hanhart)	Tyrosine aminotransferase	**Painful palmoplantar keratosis, corneal dendritic ulcers**, ID	Diet
Alkaptonuria	**Homogentisic acid oxidase**	Urine **turns black on standing**, ochronosis (ear cartilage, sclera), arthritis in adults; benign in children	Nitisinone, vitamin C
Albinism	Tyrosinase (OCA1)	Photophobia, nystagmus, ↓ visual acuity, skin cancers	Sun protection
MSUD	**Branched-chain α-ketoacid dehydrogenase (BCKAD)** – leucine, isoleucine, valine	Day 4–7: poor feeding, **opisthotonus, fencing/bicycling movements, alternating hypertonia–hypotonia**, cerebral oedema, coma; **maple-syrup odour**; **alloisoleucine (diagnostic)**; DNPH test +	Dialysis, BCAA-free diet, **thiamine** trial
Homocystinuria	**Cystathionine β-synthase**	Marfanoid, **downward lens dislocation**, ID, thrombosis, osteoporosis; **cyanide-nitroprusside +**	**Pyridoxine**, betaine, ↓ methionine, B12/folate
Non-ketotic hyperglycinaemia	Glycine cleavage system	Neonatal intractable seizures, apnoea, hiccups, **burst-suppression EEG**; **↑ CSF/plasma glycine > 0.08**	Sodium benzoate, dextromethorphan
Hartnup disease	Neutral amino-acid transporter (SLC6A19)	**Pellagra-like rash**, cerebellar ataxia; neutral aminoaciduria	**Nicotinamide**, high-protein diet
Cystinuria	Dibasic AA transporter (**COLA**: cystine, ornithine, lysine, arginine)	**Hexagonal crystals**, radiolucent-ish stones; cyanide-nitroprusside +	Hydration, alkalinise urine, penicillamine/ tiopronin
Cystinosis	Lysosomal cystine transporter (cystinosin, CTNS)	**MC cause of inherited Fanconi syndrome**, rickets, FTT, **corneal crystals (photophobia)**, ESRD by 10 y	**Cysteamine**

6.4 Organic Acidurias & Urea Cycle Disorders



Urea cycle – enzyme defects and their key markers

Urea cycle defect	Inheritance	Key lab
CPS-I deficiency	AR	↑ NH_3, **low citrulline, normal/low urinary orotic acid**
OTC deficiency (MC urea cycle defect)	**X-linked**	↑ NH_3, low citrulline, **↑↑ urinary orotic acid**
Citrullinaemia (ASS)	AR	**↑↑↑ citrulline**
Argininosuccinic aciduria (ASL)	AR	↑ citrulline, ↑ **argininosuccinate**, **trichorrhexis nodosa (brittle hair)**
Arginase deficiency	AR	**↑ arginine**, **progressive spastic diplegia** (rarely hyperammonaemic crises)
NAGS deficiency	AR	Like CPS-I; **carglumic acid** responsive

OROTIC ACIDURIA: UREA CYCLE VS PYRIMIDINE DEFECT

OTC deficiency: orotic aciduria **+ hyperammonaemia, no megaloblastic anaemia**. **Hereditary orotic aciduria (UMP synthase)**: orotic aciduria **+ megaloblastic anaemia unresponsive to B12/folate, no hyperammonaemia** – Rx **uridine**.

Organic acidemia	Defect	Features	Rx
Methylmalonic acidemia	Methylmalonyl-CoA mutase / **B12 (cobalamin) metabolism**	Neonatal ketoacidosis, hyperammonaemia, **neutropenia/ pancytopenia**, metabolic stroke of **basal ganglia (globus pallidus)**, renal failure	Protein restriction, **B12 (hydroxocobalamin)**, carnitine, metronidazole (↓ gut propionate)
Propionic acidemia	Propionyl-CoA carboxylase (**biotin**-dependent)	As above, cardiomyopathy	Biotin, carnitine, diet
Isovaleric acidemia	Isovaleryl-CoA DH	**Sweaty-feet odour**, acidosis, pancytopenia	**Glycine**, carnitine, leucine restriction

Organic acidemia	Defect	Features	Rx
Glutaric aciduria type I	Glutaryl-CoA DH	**Macrocephaly**, acute encephalopathic crisis with **striatal injury → dystonia**, **subdural haematomas (mimic NAI)**, frontotemporal atrophy	Lysine restriction, carnitine
Multiple carboxylase / **biotinidase deficiency**	Biotin recycling	Seizures, **alopecia, dermatitis**, hypotonia, hearing loss, optic atrophy	**Biotin** (dramatic response)

6.5 Disorders of Carbohydrate Metabolism

Galactosemia (classic)

- **Galactose-1-phosphate uridyl transferase (GALT)** deficiency, AR (chr 9).
- After milk feeds: vomiting, **jaundice (conjugated + unconjugated), hepatomegaly → liver failure/cirrhosis**, **E. coli sepsis (MCC death)**, **oil-drop cataract**, renal Fanconi, ID, **ovarian failure (late)**, bleeding.
- Dx: **urine reducing substances positive (Benedict) but glucose oxidase (dipstick) negative**; RBC GALT assay (falsely normal after transfusion – use DNA); newborn screening.
- Rx: **lifelong lactose/galactose-free diet (soy formula)**; **breastfeeding absolutely contraindicated**.
- **Galactokinase deficiency** – only **cataract** (galactitol); no liver/brain disease.
- **Hereditary fructose intolerance** (**aldolase B**, AR): symptoms on **weaning** to fruit/sucrose (sucrose, honey, sorbitol) – vomiting, **hypoglycaemia**, liver failure, Fanconi; **aversion to sweets, no dental caries**. Rx avoid fructose/sucrose/sorbitol.
- **Essential fructosuria** (fructokinase) – benign.

Glycogen storage diseases (GSD)

Type	Enzyme	Organ	Key features
I – von Gierke (MC hepatic GSD)	**Glucose-6-phosphatase** (Ia) / G6P translocase (Ib)	Liver, kidney	**Severe fasting hypoglycaemia (seizures in early morning), lactic acidosis, hyperuricaemia (gout), hyperlipidaemia (xanthomas)**, massive **hepatomegaly + nephromegaly**, **doll-like (cherubic) face**, short stature, bleeding (platelet dysfunction); **no rise in glucose with glucagon**; Ib – **neutropenia, IBD-like**; Rx **frequent feeds, uncooked cornstarch**, allopurinol; hepatic adenomas
II – Pompe	**Acid maltase (lysosomal α-1,4-glucosidase)** – the only **lysosomal** GSD	Heart, muscle (generalised)	Infantile: **massive cardiomegaly (hypertrophic CMP), short PR + giant QRS**, hypotonia ("floppy"), **macroglossia**, hepatomegaly; **normal glucose**; death < 1–2 y; Rx **enzyme replacement – alglucosidase alfa**
III – Cori / Forbes	**Debranching enzyme (amylo-1,6-glucosidase)**	Liver, muscle	Like type I but **milder, normal lactate & uric acid**, **↑ transaminases & CK**, myopathy, limit dextrin
IV – Andersen	Branching enzyme	Liver	**Progressive cirrhosis** in infancy, amylopectin-like glycogen
V – McArdle	**Muscle glycogen phosphorylase (myophosphorylase)**	Muscle	Adolescents: **exercise intolerance, cramps, myoglobinuria**, "**second-wind**" phenomenon; **no rise in lactate on ischaemic forearm exercise test**, ↑ ammonia
VI – Hers	Liver phosphorylase	Liver	Mild hepatomegaly, mild hypoglycaemia
VII – Tarui	Muscle phosphofructokinase	Muscle, RBC	Like McArdle + **haemolysis**; no second-wind

'VERY POOR CARBOHYDRATE METABOLISM' = TYPES I, II, III, V

Von Gierke (I), **P**ompe (II), **C**ori (III), **M**cArdle (V). Pompe "**Pompe trashes the Pump**" (heart, liver, muscle).

6.6 Lysosomal Storage Disorders

Mucopolysaccharidoses (MPS)

Type	Enzyme	Inheritance	Distinguishing features
I – Hurler (severe)	**α-L-iduronidase**	AR	**Corneal clouding**, coarse (gargoyle) facies, ID, **dysostosis multiplex**, hepatosplenomegaly, umbilical hernia, **gibbus (lumbar kyphosis)**, cardiac (valvular), death by 10 y
I – Scheie (mild)	α-L-iduronidase	AR	Corneal clouding, stiff joints, **normal intelligence**, aortic regurgitation
II – Hunter	**Iduronate-2-sulfatase**	**X-linked recessive**	**No corneal clouding**, aggressive behaviour, ID, **pebbly ivory skin lesions over scapulae**, retinitis pigmentosa; males only
III – Sanfilippo (A–D)	Heparan-N-sulfatase etc.	AR	**Severe ID & behavioural problems** with **mild somatic features**; heparan sulfate only
IV – Morquio	Galactose-6-sulfatase (A) / β-galactosidase (B)	AR	**Normal intelligence**, **short-trunk dwarfism**, severe skeletal dysplasia (**odontoid hypoplasia → atlanto-axial instability**), **corneal clouding**, joint laxity; keratan sulfate
VI – Maroteaux–Lamy	Arylsulfatase B	AR	Hurler-like with **normal intelligence**
VII – Sly	β-glucuronidase	AR	Hydrops fetalis

- **Dysostosis multiplex** on X-ray: J-shaped sella, **oar/paddle-shaped ribs**, **beaked (hook-shaped) vertebrae** (L1–L2), bullet-shaped phalanges, flared iliac wings.
- Dx: **urinary glycosaminoglycans (GAG)** screen → enzyme assay; Rx **haematopoietic stem cell transplant (Hurler < 2 y)**, enzyme replacement (laronidase MPS I, idursulfase MPS II, elosulfase MPS IVA, galsulfase MPS VI).

CORNEAL CLOUDING IN MPS

Present in Hurler, Scheie, Morquio, Maroteaux–Lamy; **ABSENT in Hunter and Sanfilippo**. "**Hunters need clear eyes** to aim."

Sphingolipidoses

Disease	Enzyme	Inheritance	Features
Tay–Sachs	**Hexosaminidase A** (GM2)	AR (Ashkenazi)	**Cherry-red spot**, **exaggerated startle (hyperacusis)**, **macrocephaly**, neuroregression from 3–6 m, blindness, seizures; **no hepatosplenomegaly**; "**onion-skin**" lysosomes
Sandhoff	Hexosaminidase A + B	AR	Like Tay–Sachs **+ hepatosplenomegaly**
Niemann–Pick type A/B	**Sphingomyelinase**	AR	**Hepatosplenomegaly**, cherry-red spot (50%), neurodegeneration (A), **foamy (zebra-body) cells**; type C – NPC1 cholesterol trafficking, **vertical supranuclear gaze palsy**, cataplexy
Gaucher (MC lysosomal storage disease)	**β-glucocerebrosidase**	AR	Type 1 (non-neuronopathic, MC): **massive splenomegaly**, bone pain, **Erlenmeyer-flask deformity of femur**, avascular necrosis, pancytopenia; **Gaucher cells – "crumpled tissue paper / wrinkled silk" cytoplasm**; ↑ chitotriosidase, ACE, acid phosphatase; Rx **ERT – imiglucerase**, substrate reduction (eliglustat)
Krabbe	**Galactocerebrosidase**	AR	Irritability, hypertonia, **optic atrophy**, **globoid cells**, peripheral neuropathy, ↑ CSF protein

Disease	Enzyme	Inheritance	Features
Metachromatic leukodystrophy	**Arylsulfatase A**	AR	Late-infantile gait disturbance, regression, **peripheral neuropathy**, metachromatic granules (urine sulfatides)
Fabry	**α-galactosidase A**	**XLR**	**Angiokeratomas** (bathing-trunk area), **painful acroparaesthesia**, hypohidrosis, **corneal verticillata**, renal failure, stroke, cardiomyopathy; Rx ERT (agalsidase), migalastat
Farber	Ceramidase	AR	Hoarse cry, painful swollen joints, subcutaneous nodules
Wolman	Lysosomal acid lipase	AR	**Adrenal calcification**, hepatosplenomegaly, steatorrhoea

CHERRY-RED SPOT AT MACULA

Tay–Sachs, Sandhoff, Niemann–Pick (A), GM1 gangliosidosis, sialidosis (cherry-red spot myoclonus), Farber, metachromatic leukodystrophy (some), and **central retinal artery occlusion**. **Not** in Gaucher type 1.

- **I-cell disease (mucolipidosis II)**: defective **N-acetylglucosamine-1-phosphotransferase** → enzymes can't be tagged with **mannose-6-phosphate** → secreted outside; **↑ plasma lysosomal enzymes**; Hurler-like features, gingival hyperplasia, death by 5–8 y.

6.7 Peroxisomal & Mitochondrial Disorders

Disorder	Features
Zellweger (cerebro-hepato-renal) – PEX genes	**Severe neonatal hypotonia**, **large fontanelle, high forehead**, seizures, hepatomegaly, **renal cortical cysts**, **stippled calcification of patella**, ↑ **VLCFA**; death < 1 y
X-linked adrenoleukodystrophy (ABCD1)	Boys 4–10 y: **ADHD-like behaviour → regression, vision/hearing loss, Addison disease (bronze skin)**; **↑ VLCFA**; MRI – **posterior (parieto-occipital) white-matter** demyelination; Rx **HSCT** early, **Lorenzo's oil** (limited), gene therapy (elivaldogene)
Refsum disease	Phytanic acid ↑ – retinitis pigmentosa, neuropathy, ichthyosis, ataxia
Leigh syndrome (subacute necrotising encephalomyelopathy)	Regression, brainstem signs (abnormal breathing, eye movements), lactic acidosis; **bilateral symmetric basal ganglia & brainstem lesions**
MELAS	Mitochondrial Encephalopathy, Lactic Acidosis, **Stroke-like episodes** (posterior, not in vascular territory) – tRNA-Leu (A3243G)
MERRF	**Myoclonic epilepsy with ragged red fibres** (Gomori trichrome) – tRNA-Lys
Kearns–Sayre	Onset < 20 y: **chronic progressive external ophthalmoplegia, pigmentary retinopathy, heart block**, ↑ CSF protein; mtDNA deletion (sporadic)
Pearson syndrome	**Sideroblastic anaemia + exocrine pancreatic insufficiency**; survivors evolve into Kearns–Sayre
LHON	Young males – acute painless bilateral sequential **optic neuropathy**

6.8 Fatty Acid Oxidation Defects

- **MCAD deficiency** – **most common FAO disorder**; previously well infant 3–24 m presents after **fasting/illness** with **hypoketotic hypoglycaemia**, vomiting, lethargy, hepatomegaly, **Reye-like** picture, **sudden infant death**; Dx **acylcarnitine profile (↑ C8 – octanoylcarnitine)**; Rx **avoid fasting**, frequent carbohydrate feeds, IV glucose in illness, **avoid** long-chain fats only in LCHAD.
- **LCHAD** – cardiomyopathy, rhabdomyolysis, **retinopathy**, maternal **AFLP/HELLP** when fetus affected; Rx MCT oil.
- **Carnitine deficiency**: primary (OCTN2 transporter) – cardiomyopathy, hypoketotic hypoglycaemia; Rx **L-carnitine**.

6.9 Other Metabolic Disorders (selected)

Disorder	Key points
Wilson disease	ATP7B (chr 13, AR) – see Hepatology
Menkes	ATP7A (X-linked) – kinky hair, ↓ Cu
Lesch–Nyhan	**HGPRT deficiency (XLR)** – **self-mutilation (lip & finger biting)**, dystonia, choreoathetosis, ID, **hyperuricaemia (orange "sand" in diapers), gout**, uric acid stones; Rx allopurinol (doesn't help neuro)
Smith–Lemli–Opitz	7-dehydrocholesterol reductase – low cholesterol; 2–3 toe syndactyly, ptosis, ambiguous genitalia
Congenital disorders of glycosylation (PMM2-CDG)	**Inverted nipples, abnormal fat pads (buttocks)**, cerebellar hypoplasia, strabismus; **transferrin isoelectric focusing**
Acute intermittent porphyria	PBG deaminase (AD) – abdominal pain, neuropsychiatric, **port-wine urine on standing**; precipitated by barbiturates, sulfonamides
Crigler–Najjar / Gilbert	UGT1A1 – see Neonatal jaundice

6.10 Newborn Screening

- **Sample**: heel-prick **dried blood spot on Guthrie (filter) card**, ideally at **48–72 h of age** (after protein feeds); preterm/sick – repeat at 2 weeks.
- **Tandem mass spectrometry (MS/MS)** – screens amino-acid disorders, organic acidemias & FAO defects simultaneously.
- **Indian (RBSK / state programmes) core panel**: **congenital hypothyroidism (TSH) – most common**, **CAH (17-OHP)**, **G6PD deficiency**, **galactosemia**, **PKU**, **biotinidase deficiency**, **cystic fibrosis (IRT)**, haemoglobinopathies (sickle cell) in endemic areas; **hearing** (OAE/BERA) and **CCHD (pulse oximetry)** screening.
- **Wilson–Jungner criteria** for screening: important health problem, treatable, recognisable latent stage, suitable acceptable test, cost-effective.

FREQUENTLY ASKED ONE-LINERS

- IEM with **cataract**: galactosemia, galactokinase def. · with **lens dislocation**: homocystinuria (down), sulfite oxidase def. · **cherry-red spot**: Tay–Sachs · **macrocephaly**: Tay–Sachs, Canavan (aspartoacylase, ↑ NAA), Alexander (GFAP), glutaric aciduria I.
- Hypoglycaemia with **no ketones**: MCAD/FAO, hyperinsulinism. Hypoglycaemia with **lactic acidosis + hepatomegaly**: GSD I.
- **Self-mutilation**: Lesch–Nyhan. **Black urine**: alkaptonuria. **Red/port-wine urine**: porphyria. **Blue diaper**: tryptophan malabsorption (Drummond). **Orange sand**: Lesch–Nyhan.

Practice MCQs · Inborn Errors of Metabolism

204 questions · correct option highlighted in green · cover the answer strip to self-test

Phenylketonuria (PKU)

1. Metabolic abnormalities in which jaundice is seen are all of the following except?

A. Von Gierke disease
B. Galactosemia
C. Tyrosinemia
D. Hereditary fructose intolerance

Ans A In Von Gierke disease, hepatic carbohydrate metabolism is affected, which is responsible for plasma glucose homeostasis. So, fasting ypoglycemia and hepatomegaly is seen. Despite, marked hepatomegaly, the liver transaminase levels are usually normal or only slightly elevated. Jaundice is usually not seen.

2. Hyperphenylalaninemia is due to defect in the enzyme:

A. Phenylalanine hydroxylase:
B. Tyrosinase
C. Homogentisic acid oxidase
D. Ornithine transcarbamylase
E. Phenylalanine oxidase

Ans A Phenylalanine hydroxylase Deficiency of the enzyme phenylalanine hydroxylase or of its cofactor tetrahydrobiopterin (BH4) causes accumulation of phenylalanine in body fluids and in the brain.

3. Tom cat urine odor is seen in?

A. Multiple carboxylase deficiency
B. Hawkinsinuria
C. Tyrosinemia
D. Phenylketonuria

Ans A

4. Enzyme deficient in phenylketonuria:

A. Tyrosinase
B. Phenylalanine hydroxylase
C. Tyrosine transaminase
D. Homogentisic oxidase

Ans B

5. Phenylketonuria–true statement:

A. Urine phenylalanine metabolites are increased
B. Deficiency of tyrosine hydroxylase
C. Serum tyrosine levels are increased
D. Mental retardation does not occur if patient is advised to have phenyl alanine free diet

Ans A Discussing about the options, one by one, Option a. True Phenylalanine metabolites like phenylacetate, phenylpyruvate increase in blood and urine Option b. False Phenylalanine hydroxylase is the enzyme deficient in this condition Option c. False Serum tyrosine levels are decreased, as tyrosine is not getting synthesized Option d. False If PKU is detected by neonatal screening and low phenylalanine diet is started immediately and continued life long

6. The enzyme deficient in phenylketonuria:

A. Phenylalanine hydroxylase
B. Tyrosinase
C. Aminotransferase
D. Homogentisic acid oxidase

Ans A Phenylalamine hydroxylase

7. Which one of the following is not a feature of Phenylketonuria?

A. Severe mental retardation
B. Reduced tendon reflexes
C. Enamel hypoplasia
D. Vomiting in early morning

Ans B Clinical Manifestations of Phenylketonuria include:
• Blonde hair, blue iris, fair skin, musty or mousy body odor, Vomiting, enamel hypoplasia, growth retardation. Mental retardation — Not present at birth, develops gradually; > 90% of untreated patients have low IQ
• Neurological signs — Irritability, tremors, convulsions, hypertonia, exaggerated reflexes and microcephaly

8. In phenylketonuria, the treatment of choice is:

A. Limit intake of substrate for the enzyme
B. Provide the deficient amino acid
C. Correct the enzyme defect
D. Symptomatic management (AIIMS Nov 10)

Ans A Low phenylalanine diet is the treatment of choice for phenylketonuria.

9. PKU is a congenital amino acid metabolic disorder. In one of the following rare variants of PKU, dihydrobiopterin synthesis is affected. The enzyme deficient is:

A. Histidine decarboxylase (PGI June 08)
B. Phenylalanine hydroxylase
C. Dihydropteridine reductase
D. Tyrosine deficiency

Ans C • In 1-3% of infants with hyperphenylalaninemia, the defect resides in 1 of the enzymes necessary for production or recycling of the cofactor BH4;
• If these infants are misdiagnosed as having PKU, they may deteriorate neurologically despite adequate control of plasma phenylalanine
• BH4 is synthesized from guanosine triphosphate (GTP) by following enzymes: pterincarbinolamine dehydratase, dihydrobiopterin reductase, GTP cyclohydrolase, 6pyruvoyl tetrahydropterin synthase.

10. True statement regarding a 3weekold child with phenylketonuria are all, except:

A. Provocative protein meal tests help in the diagnosis
B. Tyrosine becomes an essential amino acid in diet
C. Serum phenylalanine is increased and urinary phenyl pyruvate level is elevated
D. **Phenylalanine should be completely stopped in diet**

Ans D In Phenylketonuria, low phenylalanine diet is used; small amount of phenylalanine should be added to diet.
• Serum and urine phenylalanine and its metabolites are increased; Tyrosine becomes an essential amino acid
• Blood for screening should be obtained after 24–48 hrs (Protein feeding) to prevent false –ve result.

11. All the following are widely used modalities for treatment of inborn errors of metabolism (IEM) EXCEPT

A. special diets
B. **gene therapy**
C. peritoneal dialysis
D. liver transplantation
E. administration of the deficient metabolite

Ans B Replacement of the mutant gene with a normal one (gene therapy) is still in the experimental phase.

12. The mainstay of treatment of phenylketonurea (PKU) is especial diet. All the following statements are true EXCEPT

A. phenylalanine not totally restricted
B. all patients kept on a phenylalanine-restricted diet lifelong
C. especial diet should be used before and during pregnancy in mother with PKU
D. especial diet should be started when blood phenylalanine levels above 10 mg/dL
E. **especial diet is not advocated in mild hyperphenylalaninemia whose levels are ≥ 6 mg/Dl**

Ans E The mainstay of treatment of PKU is a low–phenylalanine diet. Most physicians advocate phenylalanine-restricted diet in patients with mild hyperphenylalaninemia whose levels are persistently above 6 mg/ dL (360 µmole/L). It is generally accepted that infants with persistent (more than a few days) plasma levels of phenylalanine ≥6 mg/dL (360 µmole/L) should be treated with a phenylalanine-restricted diet similar to that for classic PKU.

13. All the following lipidoses are inherited as an autosomal recessive traits EXCEPT

A. **Fabry disease**
B. Krabbe disease
C. Gaucher disease
D. Tay-sachs disease
E. Niemann-pick disease

Ans A inherited as X-linked recessive.

14. All the following should be restricted in hereditary fructose intolerance EXCEPT

A. **milk**
B. sorbitol
C. fruit juice
D. table sugar
E. sweetened cereal

Ans A In hereditary fructose intolerance, treatment consists of the complete elimination of all sources of sucrose, fructose, and sorbitol from the diet. It may be difficult because these sugars are widely used additives, found even in most medicinal preparations.

15. All the following drugs are unsafe in acute intermittent porphyria EXCEPT

A. nifedipine
B. diclofenac
C. **cimetidine**
D. valproic acid
E. carbamazepine

Ans C

16. A 3-year-old boy presented to emergency unit at 9:00 AM with history of difficult arousal from sleep, flue like illness, and low grade fever; the mother stated that he had slept without having his supper. Fasting blood sugar was 42 mg/dL. The following metabolic changes are seen in this condition EXCEPT

A. ketonuria
B. low insulin level
C. **high alanine level**
D. high cortisol level
E. high growth hormone level

Ans C Children with ketotic hypoglycemia have plasma alanine concentrations that are markedly reduced in the basal state after an overnight fast and decline even further with prolonged fasting. Alanine, produced in muscle, is a major gluconeogenic precursor.

17. All the following are features of neonatal adrenoleukodystrophy (ALD) EXCEPT

A. hepatomegaly
B. impaired liver function
C. severely impaired hearing
D. **chondrodysplasia punctata**
E. pigmentary degeneration of the retina

Ans D and renal cysts are absent.

Galactosemia

18. A child who was normal at birth, develops chronic liver failure and muscle weakness at 3 months of age. On investigation, Serum Glucose is low, along with ketoacidosis and decreased pH. ALT and AST are raised. Blood lactate and uric acid levels are normal. Intravenous glucagon given after meals raises the blood glucose levels but does not raise glucose when given after an overnight fast. Liver biopsy shows increased glycogen in liver. Which is the enzyme likely to be defective in this child?

A. Glucose-6-phosphatase
B. Muscle Phosphorylase
C. Branching enzyme
D. **Debranching enzyme**

Ans D The given clinical scenario points towards an underlying glycogen storage disease, with both liver and muscle involvement; weiveR During childhood, type III GSD (Debrancher deficiency) may be indistinguishable from type I GSD (Glucose6-Phosphatase deficiency), because hepatomegaly, hypoglycemia, hyperlipidemia, & growth retardation are seen in both. Difference between Type I and Type III GSD: Type I GSD Type III GSD

19. In children with classical galactosemia all are true except:

A. E. coli neonatal sepsis is common
B. **Elimination of galactose in diet will not reverse cataract**
C. Galactose converts to galactitol which is toxic to brain and liver
D. Duarte variant of galactosemia were asymptomatic

Ans B E. coli sepsis is common & the onset of sepsis often precedes the diagnosis of galactosemia Elimination of galactose reverses growth failure, cataract, renal & hepatic dysfunction GALT deficiency → Galactose-1-P accumulates → Injury to kidney, liver & brain Galactokinase deficiency → accumulation of galactose &galactitol The Duarte variant, a single amino acid substitution, has 50% of normal RBC enzyme activity, but usually no clinical significance.

20. True regarding galactosemia:

A. **Mental retardation occurs**
B. Absent disaccharidase in intestine
C. Defect in epimerase
D. Defect in galactose 1 PO uridyl transferase
E. Reducing substance positive in urine

Ans A c. Defect in epimerase, d. Defect in galactose 1 PO uridyl transferase, e. Reducing substance positive in urine Galactosemia is caused by deficiency of: GALT (Galactose 1 PO uridyl transferase) [Most common], Galactokinase or Epimerase. Mental retardation can occur if the intake of milk or milk products is continued, in a child with Galactosemia.

21. Oochronosis is found in:

A. **Alkaptonuria**
B. Tyrosinemia
C. Phenylketonuria
D. Homocystinuria

Ans A Ochronosis (dark spot on sclera/ear cartilage) is seen in Alkaptonuria which is an autosomal recessive disorder due to deficiency of enzyme homogentisic acid oxidase.

22. If the tyrosinase gene is defective, it results in:

A. **Lack of pigmentation**
B. Maple syrup urine disease
C. Phenylketonuria
D. Kidney stones

Ans A Defect in Tyrosinase leads to Albinism, which presents with hypopigmentation.

23. Deficiency of homogentisate oxidase leads to:

A. Homocystinuria
B. **Alkaptonuria**
C. Maple syrup urine disease
D. Phenylketonuria

Ans B

24. Crumpled tissue paper appearance of cytoplasm in bone marrow examination is seen due to accumulation of which of the following inside the cell?

A. Cerebrosides
B. Ganglioside accumulation
C. Sphingomyelin
D. **Glucocerebroside accumulation**

Ans D

25. Lorenzo oil used in treatment of:

A. Gaucher disease
B. **Adrenoleukodystrophy**
C. Fabry's disease
D. Hurles disease

Ans B Therapeutic approaches to ALD include administration of glycerol trioleate and glycerol trierucate (Lorenzo's oil), bone marrow transplantation, and lovastatin.

26. Deep white matter lesion with bilateral deep bright thalamic appearance on CT head is suggestive of:

A. Alexander disease
B. Canavan's disease
C. **Krabbe's disease**
D. Metachromatic leukodystrophy

Ans C 1 05. d. Exaggerated tendon reflexes In metachromatic leukodystrophy (MLD), a disorder of myelin metabolism, there is hypotonia and absent reflexes.

27. Menke's disease is due to defect in the metabolism of:

A. Zinc
B. **Copper**
C. Selenium
D. Iron

Ans B In Menke's disease, the defective protein is a copper-binding ATPase (ATP7A), responsible for distribution & metabolism of copper in tissues. Chapter 6: Inborn

28. **A 28-day-old girl presented with lethargy, poor feeding, and repeated vomiting for last 5 days; IV fluid and empirical antibiotics were started; later she develops repeated seizures not responding to IV calcium, glucose, B6, and anticonvulsant drugs. Septic screen is negative; serum ammonia is elevated with normal anion gap and normal pH. Family history reveals 2 siblings died with same scenario. Of the following, the MOST likely diagnosis is**

A. galactosemia
B. hyperglycinemia
C. organic acidemias
D. phenylketonurea
E. **urea cycle defects**

Ans E Elevation of blood ammonia is usually caused by defects of urea cycle enzymes. Infants with elevated blood ammonia levels from urea cycle defects commonly have normal serum pH and bicarbonate values; without measurement of blood ammonia, they may remain undiagnosed and succumb to their disease.

29. **A 27-day-old girl presented with abnormal bicycling movement, lethargy, poor feeding, and repeated vomiting for the last 7 days, and then she developed rapid breathing, opisthotonos, and hypertonicity with bad odor urine. Of the following, the MOST effective mode of therapy is**

A. good hydration
B. **peritoneal dialysis**
C. liver transplantation
D. sufficient calories and nutrients
E. diet low in branched-chain amino acids

Ans B Treatment of the acute state of maple syrup urine disease is aimed at hydration and rapid removal of the branched-chain amino acids and their metabolites from the tissues and body fluids. Because renal clearance of these compounds is poor, hydration alone may not produce a rapid improvement. Peritoneal dialysis or, preferably, hemodialysis is the most effective mode of therapy in critically ill infants.

30. **Niemann-pick disease type A is characterized by all the following EXCEPT**

A. failure to thrive
B. hepatosplenomegaly
C. **progressive lung disease**
D. psychomotor retardation
E. rapidly progressive neurodegenerative

Ans C The progressive deposition of sphingomyelin in the central nervous system results in the neurodegenerative course seen in type A, and in nonneural tissue in the systemic disease manifestations of type B, including progressive lung disease in some patients.

31. **Irreversible complication of untreated galactosemia is**

A. sepsis
B. cataract
C. renal injury
D. myocardial damage
E. **intellectual disability**

Ans E In galactosemia when the diagnosis is not made at birth, damage to the liver (cirrhosis) and brain (intellectual disability) becomes increasingly severe and irreversible.

32. **All the following are common features between hereditary fructose intolerance and galactosemia EXCEPT**

A. failure to thrive
B. impaired renal function
C. impaired hepatic function
D. reducing substance in urine
E. **impaired intellectual development**

Ans E In hereditary fructose intolerance, intellectual development is usually unimpaired.

33. **At 10:00 AM Lylaa`s mother brought her 3-year-old daughter to emergency department with difficult arousal from sleep, low grade fever, and cough for the last 2 days. Fasting blood sugar was 36 mg/dL with ketonuria. Of the following, the MOST likely diagnosis is**

A. galactosemia
B. propionic acidemia
C. **ketotic hypoglycemia**
D. hereditary fructose intolerance
E. medium-chain acyl-CoA dehydrogenase (MCAD) deficiency

Ans C During acute episodes of medium-chain acyl-CoA dehydrogenase (MCAD) deficiency, hypoglycemia is usually present; plasma and urinary ketone concentrations are inappropriately low (hypoketotic hypoglycemia).

34. **All the following are characteristic features of infantile Refsum disease EXCEPT**

A. ataxic gait
B. **hypertonia**
C. hearing loss
D. impaired cognition
E. retinal degeneration

Ans B Early hypotonia and hepatomegaly with impaired function are common.

Maple Syrup Urine Disease

35. **Which disorder of carbohydrate metabolism typically has cardiac involvement?**

A. Glycogen Storage Disease Type I (Von Gierke disease)
B. **Glycogen Storage Disease Type II (Pompe disease)**
C. Hereditary fructose intolerance
D. Galactosemia

Ans B

36. **Lactose intolerance in a suspected patient is diagnosed by giving an oral load of lactose and measuring concentration of:**

A. CO
B. **H 2**
C. One/two carbon compounds
D. N

Ans B Breath hydrogen tests can be used to determine a specific carbohydrate malabsorption. It can be used to diagnose bacterial overgrowth or lactose/fructose malabsorption.

37. **Dried blood spot test in neonates is used to check for:**

A. Inborn error of metabolism
B. Blood group
C. Total cell count
D. Creatinine and bilirubin

Ans A Tandem mass spectrometry (TMS) on dried blood spot can be done for screening for metabolic disorders

38. **Branched chain keto acids are excreted in urine in:**

A. Maple syrup urine disease
B. Hartnup disease
C. Albinism
D. Alkaptonuria122 Section 1: General Pediatrics

Ans A Maple syrup urine disease is an autosomal recessive disorder of branched chain amino acid, i.e. valine, leucine and Isoleucine

39. **Which of the following is a branched chain keto acid disorder?**

A. Propionic acidemia
B. Methylmalonic acidemia
C. Maple syrup urine disease
D. Homocystinuria

Ans C

40. **Maple syrup urine disease is due to defect in the metabolism of:**

A. Branched chain amino acids
B. Essential fatty acids
C. Essential amino acids
D. Glucogenic amino acids

Ans A

41. **Maple syrup urine disease is due to defective decarboxylation of:**

A. Branched chain amino acids
B. Unbranched chain amino acids
C. Sulfur containing amino acids
D. Hydroxyl amino acids

Ans A

42. **Globoid cells is a diagnostic feature of:**

A. Krabbe's disease
B. Progressive multifocal leukoencephalopathy
C. Tay Sach's disease
D. Metachromatic leukodystrophy

Ans A Krabbe disease (KD) is also called Globoid Cell Leukodystrophy;

43. **All are seen in metachromatic leukodystrophy except:**

A. Mental retardation
B. Optic atrophy
C. Decerebrate posture
D. Exaggerated tendon reflexes

Ans D MLD classically shows DIMINISHED or absent deep tendon reflexes (from an associated peripheral demyelinating neuropathy) despite central white-matter disease — exaggerated reflexes are not a feature.

44. **Metabolic disease in which intestinal obstruction is seen?**

A. Hereditary fructose intolerance
B. Phenylketonuria
C. Hurler disease
D. Cystic fibrosis

Ans D In cystic fibrosis, neonates may present with meconium ileus, meconium plug & meconium peritonitis, with distal intestinal obstruction syndrome.

45. **You suspect a metabolic problem in a 30-day-old girl presented with poor feeding, vomiting, lethargy, and convulsion. Previous sibling died with the same condition. Serum ammonia, pH, HCO3, and anion gap are normal. Of the following, the MOST likely diagnosis is**

A. porphyria
B. galactosemia
C. organic acidemia
D. urea cycle defect
E. aminoacidopathy

Ans E

46. **The following are ocular manifestations of albinism EXCEPT**

A. red reflex
B. strabismus
C. refractive errors
D. foveal hyperplasia
E. lack of binocular vision

Ans D Ocular manifestations of albinism include hypopigmentation of iris and retina, foveal hypoplasia, reduced visual acuity, refractive errors, nystagmus, alternating strabismus, and a red reflex.

47. **A 7-month-old boy presented with recurrent attacks of hypoglycemic seizures especially during an acute illness. On examination, there are thin extremities, protuberant abdomen, and hepatomegaly. All the following are biochemical hallmarks of this disease EXCEPT**

A. lactic acidosis
B. hypoglycemia
C. hyperuricemia
D. hyperlipidemia
E. markedly elevated transaminase levels

Ans E In type I GSD (von Gierke disease), despite marked hepatomegaly, the liver transaminase levels are usually normal or only slightly elevated.

48. **A 9-year-old boy presented with tiredness on exertion; cardiac auscultation reveals a murmur in aortic area; echo study showed aortic valve disease. On examination he had mild coarse facial features, corneal clouding, joint stiffness, and mild dysostosis multiplex but normal intelligence and stature. Of the following, the MOST likely diagnosis is**

A. Hurler disease
B. **Scheie disease**
C. Hunter disease
D. Morquio disease
E. Sanfilippo disease

Ans B Scheie is a comparatively mild disorder characterized by joint stiffness, aortic valve disease, corneal clouding, and mild dysostosis multiplex. Patients with Scheie disease have normal intelligence and stature but have significant joint and ocular involvement.

49. **All the following are clinical features of acute intermittent porphyria EXCEPT**

A. neuropathy
B. tachycardia
C. abdominal pain
D. bladder dysfunction
E. **cutaneous photosensitivity**

Ans E is a feature of Porphyria cutanea tarda.

50. **A 5-year-old child with recurrent attacks of ketotic hypoglycemia. Of the following, the BEST advice to the parents during any intercurrent illness is to**

A. administrate a high-protein diet
B. **test the child's urine for ketones**
C. administrate a high-carbohydrate diet
D. do frequent monitoring of blood sugar
E. administrate intravenous glucose administration

Ans B During intercurrent illnesses, parents should be taught to test the child's urine for the presence of ketones, the appearance of which precedes hypoglycemia by several hours. In the presence of ketonuria, liquids of high carbohydrate content should be offered to the child.

51. **The diagnostic assay of Zellweger syndrome is by elevated level of**

A. bile acid
B. plasmalogen
C. phytanic acid
D. pristanic acid
E. **very long chain fatty acid**

Ans E 🆘 Neonatal adrenoleukodystrophy ---- Phytanic acid (increased). 🆘 Infantile Refsum disease ---- Pristanic acid (increased), Bile acid (Increased), and Plasmalogen level (decreased).

Glycogen Storage Diseases

52. **All of the following diseases/syndromes are Glycogen storage disorders except?**

A. Andersen's disease
B. Her's disease
C. **Scheie's disease**
D. Tarui's disease

Ans C Hurler/Scheie is a Mucopolysaccharidosis and not a Glycogen storage disease.

53. **Hypoglycemia, hepatomegaly, growth retardation, muscle weakness and accumulation of limit dextrins is caused by which glycogen storage disease?**

A. **Cori's disease**
B. Von Gierke's disease
C. Anderson disease
D. Pompe's disease

Ans A Important Glycogen Storage Diseases: Type Name Enzyme deficient Characteristics Type I Von Gierke's disease Glucose-6-phosphatase Liver and renal tubule cells loaded with glycogen. Hypo- glycemia, lactic acidemia, ketosis, hyperlipidemia. Type II Pompe's disease Lysosomal α-1 → 4 and 1 → Fatal, accumulation of glycogen in lysosomes, heart failure; muscle 6-glucosidase (acid maltase) involvement seen Type III Limit dextrinosis, or Debranching enzyme Accumulation of a characteristic branched polysaccharide

54. **Coarse facies, hepatosplenomegaly and tall QRS on ECG are characteristic feature of: (AIIMS May 2015, AIIMS Nov 2001) a.Glycogen storage disease type II**

A. **Pompe disease (GSD type II)**
B. Hurler's disease
C. Hunter's disease
D. Hemochromatosis

Ans A Glycogen storage disease type II The given features along with ECG changes is suggestive of Pompe disease (Type II GSD).

55. **Which glycogen storage disease does not affect muscles?**

A. **Type**
B. Type 2 (APPG 08)
C. Type
D. Type

Ans A 1 Type 2 Pompe disease predominantly involves muscles, while type I GSD involves liver. Although type 2 and type 3 GSD are predominantly liver glycogenoses, skeletal muscle involvement may also be seen.

56. **Glycogen storage diseases includes:**

A. **Von Gierke's disease**
B. Fabry's disease
C. McArdle's disease
D. Fragile X syndrome
E. Krabbe disease OTHER DISORDERS OF CARBOHYDRATE METABOLISM

Ans A c. McArdle's disease

57. **A 5dayold child presents with intractable seizures. He Questions had rashes all over the body. Blood examination showed hyperammonemia and lactic acidosis. The probable diagnosis is:**

A. **Organic acidemia**
B. Mitochondrial encephalopathy with lactic aciduria
C. Phenylketonuria
D. Urea cycle enzyme deficiency

Ans A Presence of hyperammonemia along with lactic acidosis in a child with neurological and cutaneous manifestations suggests a diagnosis of Organic acidemia.

58. **A oneyearold boy presented with hepatosplenomegaly and delayed milestones. The liver biopsy and bone mar row biopsy revealed presence of histiocytes with PAS positive Diastase resistant material in the cytoplasm. Electron microscopic examination of these histiocytes is most likely to reveal the presence of:**

A. Birbeck granules in the cytoplasm
B. Myelin figures in the cytoplasm
C. **Parallel rays of tubular structures in lysosomes**
D. Electron dense deposit in the mitochondria

Ans C Hepatosplenomegaly with delayed development and given biopsy findings are suggestive of Gaucher disease. In Gaucher disease electron microscopy reveals lipid material stored as bilayer in lysosomes.

59. **Male child with hyper intensities on T2 weighted MRI in bilateral frontal lobe. Most probable diagnosis is:**

A. Canavan disease
B. Krabbe disease
C. X-linked adrenoleukodystrophy
D. **Alexander disease**

Ans D Radiopaedia.org; Caffey's 12e/ p337 Name of disease MRI Brain finding Alexander disease T2 hyperintensities in frontal white matter; shows contrast enhancement; Canavan disease Macrocephaly; bilateral symmetric T2 white matter hyperintensity diffusely throughout cerebral white matter; does not enhance at CT & MRI; Large N acetyl aspartate (NAA) peak on MRS Adrenoleukodystrophy Symmetrical cerebral white matter signal change involving posterior (occipitoparietal) periven tricular white matter

60. **KF ring is seen in:**

A. **Low ceruloplasmin, high urine copper**
B. Low ceruloplasmin, low urine copper
C. High ceruloplasmin, high urine copper
D. High ceruloplasmin, low urine copper

Ans A Wilson disease Wilson disease (hepatolenticular degeneration) is an autosomal recessive disorder characterized by degenerative changes in the brain, liver disease, and Kayser-Fleischer rings in the cornea.

61. **A child presented with anemia, kinky hair, intellectual disability and seizures. Which of the following is the most likely diagnosis?**

A. **Menkes disease**
B. Down syndrome
C. Iron deficiency anemia
D. Lesch Nyhan syndrome

Ans A Menke disease is a X-linked recessive multisystem disorder that mainly involves CNS & usually presents become hypopigmented, sparse, short, and brittle. Hair microscopy reveals pili torti (twisted hair) & trichorrhexis of the hair shaft at regular intervals).

62. **A 3-year-old boy presented with history of hyperactivity and mild mental retardation. Past history revealed repeated projectile vomiting with normal abdominal sonography. On examination, there are eczematoid rash with lighter skin, microcephaly, and mild spasticity with exaggerated tendon reflexes. Of the following, the MOST likely urine odor of this baby is**

A. **musty**
B. rotting fish
C. sweaty feet
D. boiled cabbage
E. swimming pool

Ans A Phenylketonurea is the most likely diagnosis; these children have an unpleasant odor of phenylacetic acid, which has been described as musty or mousey. Hawkinsinuria ----------Swimming pool Isovaleric acidemias ---------- Sweaty feet Trimethylaminuria ---------- Rotting fish Tyrosinemia --- Boiled cabbage, rancid butte

63. **A 6-year-old tall and thin boy who had failed preparatory exam for school entry found to have myopia and subluxation of the ocular lens. Of the following, the MOST important challenge is to know whether he is responsive to**

A. folic acid
B. vitamin C
C. vitamin E
D. **vitamin B6**
E. vitamin B12

Ans D Homocystinuria; this is the most common inborn error of methionine metabolism. Approximately 40% of affected patients respond to high doses of vitamin B6 and usually have milder clinical manifestations than those who are unresponsive to vitamin B6 therapy. These patients possess some residual enzyme activity.

64. **A 2-year-old girl presented with rapid deep breathing, hypoglycemia, recurrent epistaxis, easy bruising, failure to thrive, fat cheek, and hepatomegaly. Of the following, the MOST likely cause of bleeding tendency is**

A. vascular disorders
B. clotting factors deficiency
C. Impaired hepatic function
D. depressed platelets count
E. **impaired platelet aggregation and adhesion**

Ans E In type I GSD (von Gierke disease), easy bruising and epistaxis are common and are associated with a prolonged bleeding time as a result of impaired platelet aggregation and adhesion.

65. **The definitive diagnosis of type I glycogen storage disease GSD is by**

B. **liver biopsy**
C. muscle biopsy
D. analysis of gene-based mutation
E. enzyme assay of cultured skin fibroblasts

Ans B The diagnosis of type I GSD is suspected on the basis of clinical presentation and the laboratory findings of hypoglycemia, lactic acidosis, hyperuricemia, and hyperlipidemia. Neutropenia is noted in GSD Ib patients, typically before 1 yr of age. Administration of glucagon or epinephrine results in little or no rise in blood glucose level, but the lactate level rises significantly.

66. **In von Gierke disease; treatment is designed to maintain normal blood glucose levels which is achieved by all the following EXCEPT**

A. restriction of fructose
B. restriction of galactose
C. **administration of table sugar**
D. continuous nasogastric infusion of glucose
E. oral administration of uncooked corn starch

Ans C Sucrose (table sugar, cane sugar, other ingredients), fructose (fruit, juice, high fructose corn syrup), lactose (dairy foods), and sorbitol should be avoided or limited.

67. **The MOST specific and important laboratory finding in X-linked adrenoleukodystrophy (ALD) is elevated level of**

A. cortisol
B. aldosterone
C. epinephrine
D. **very long chain fatty acid**
E. adrenocorticotrophic hormone

Ans D The most specific and important laboratory finding is the demonstration of abnormally high levels of VLCFA in plasma, red blood cells, or cultured skin fibroblasts. The test should be performed in a laboratory that has experience with this specialized procedure. Positive results are obtained in all male patients with ALD and in approximately 85% of female carriers of ALD.

68. **The normally functioning organ in Hutchinson-Gilford progeria syndrome (progeria) is**

A. eye
B. ear
C. skin
D. **liver**
E. heart

Ans D In progeria; normally functioning systems are; Liver, kidney, thyroid, immune, gastrointestinal, and neurologic systems (other than stroke-related) remain intact. Intellect is normal for age.

Mucopolysaccharidoses

69. **An Infant presents with hepatomegaly, hypoglycemia, hyperlipidemia and acidosis. Most probable underlying diagnosis is:**

A. **Von Gierke's**
B. Con's disease
C. Pompe's disease
D. All of the above

Ans A Presence of hepatomegaly, hypoglycemia, hyperlipidemia & acidosis, point towards Von Gierke disease.

70. **A 6monthchild presents with episodes of vomiting after ingesting fruit juice. Which of following enzyme deficiency is likely?**

A. **Aldolase B**
B. Fructokinase
C. Glucose 6-phosphatase
D. Hexokinase

Ans A In the given scenario, the child becomes symptomatic after ingesting fruit juice, which is often sweet, containing sucrose and hence, glucose and fructose. So, he is suffering from Hereditary Fructose intolerance.

71. **A 5yearold male child with developmental delay was brought with squint & bilateral lens subluxation. After 4 years, he dies of massive stroke. Autopsy reveals middle cerebral artery thrombosis and old renal infracts. Disease course could have been modified by supplement of:**

A. Thiamine
B. **Pyridoxine**
C. Tyrosine
D. Methionine

Ans B The given history of developmental delay, lens subluxation & stroke (thrombosis) points to a diagnosis of homocystinuria. Pyridoxine (Vit B6) is used in the treatment of homocystinuria, cystathioninuria, pyridoxine-dependent epilepsy, a vitamin B6–responsive anemia & xanthurenic aciduria.

72. **Mousy odor of urine is seen in:**

A. Isovaleric acidemia
B. **Phenylketonuria**
C. MSUD
D. Tyrosinemia

Ans B

73. **Mental retardation is not a feature in this aminoaciduria:**

A. Phenylketonuria
B. Homocystinuria
C. **Albinism**
D. Maple syrup urine disease

Ans C Intellectual disability is common in Phenylketonuria, Homocystinuria and MSUD.

74. **Mousy odor urine is seen in:**

A. **Maple syrup urine ds**
B. Phenylketonuria
C. Isovaleric acidemia
D. Cystinuria
E. Homocystinuria LYSOSOMAL STORAGE DISEASES

Ans A LYSOSOMAL

75. **Hunter's syndrome is due to deficiency of:**

A. **Iduronate sulfatase**
B. Hexosaminidase
C. Glucocerebrosidase
D. L-Iduronidase

Ans A

76. **Enzyme deficiency in Hunter disease is:**

A. **L-iduronidase**
B. Iduronate sulfatase
C. Heparan sulfamidase
D. Hyaluronidase

Ans A

77. **Wilson's disease is caused by defect in:**

A. ATP 7A mutation
B. **ATP 7B mutation**
C. Ceruloplasmin
D. ATP 7C mutation

Ans B The abnormal gene for Wilson disease is ATP7B on long arm of chromosome 13 (13q14.3).

78. **All are metabolic causes of liver disease except:**

A. **Histiocytosis**
B. Hemochromatosis
C. Gaucher disease
D. Wilson disease
E. Galactosemia

Ans A Metabolic causes of Liver disease:
• Carbohydrate disorders: Galactosemia, HFI, GSD
• Amino acid disorders: Tyrosinemia, Urea cycle defects, MSUD
• Lipid disorders: Gaucher disease, Niemann-Pick disease, Wolman disease
• Others: Zellweger syndrome, Wilson disease, alpha1 AT deficiency, CF, Hemochromatosis Langerhans cell histiocytosis refers to reactive proliferation of dendritic cells

79. **A 6-year-old boy with eczematoid rash, abnormal face, and prominent maxilla complains from deterioration in school performance, hyperactivity, and seizures for the last 2 years. Of the following, the MOST likely enzymatic deficiency is**

A. acid β-glucosidase
B. β-hexosaminidases
C. acid sphingomyelinase
D. **phenylalanine hydroxylase**
E. fumarylacetoacetate hydrolase

Ans D Phenylketonurea is the most likely diagnosis; deficiency of the enzyme phenylalanine hydroxylase (PAH) or of its cofactor tetrahydrobiopterin (BH4) causes accumulation of phenylalanine in body fluids and in the brain.

80. **A 6-year-old girl that looks tall and thin with light skin. On examination she has a peculiar malar flush, subluxation of the ocular lens, developmental delay, and severe hypertension. Of the following,the MOST likely cause of hypertension is**

A. atherosclerosis
B. hyperthyroidism
C. **thromboembolism**
D. coarctation of aorta
E. renal artery stenosis

Ans C In Homocystinuria, thromboembolic episodes involving both large and small vessels, especially those of the brain, are common and may occur at any age. Optic atrophy, paralysis, cor pulmonale, and severe hypertension (from renal infarcts) are among the serious consequences of thromboembolism, which is caused by changes in the vascular walls and increased platelet adhesiveness secondary to elevated homocystine levels.

81. **A 2-year-old boy presented with growth retardation, hepatomegaly, and recurrent attacks of hypoglycemia; biochemical study reveals elevated blood lactate, triglyceride, and uric acid levels. All the following are complications of this disease EXCEPT**

A. pancreatitis
B. renal failure
C. **cardiomyopathy**
D. hepatic adenoma
E. pulmonary hypertension

Ans C Type I GSD (von Gierke disease) complicated by pancreatitis, renal failure, hepatic adenoma, and pulmonary hypertension, while cardiomyopathy occure in type II GSD (pompe disease).

82. **The earliest radiographic sign in Hurler syndrome is**

A. **thick ribs**
B. thickened calvarium
C. enlarged J-shaped sella
D. abnormal spacing of teeth
E. premature closure of lambdoid suture

Ans A Hurler syndrome; Radiographs show a characteristic skeletal dysplasia known as dysostosis multiplex. The earliest radiographic signs are thick ribs and ovoid vertebral bodies. Skeletal abnormalities include enlarged, coarsely trabeculated diaphyses of the long bones with irregular metaphyses and epiphyses.

83. **Enzyme replacement is the main therapy of Hurler disease, it improve all the following EXCEPT**

A. growth rate
B. joint mobility
C. organomegaly
D. **cognition functions**
E. episodes of sleep apnea

Ans D Enzyme replacement using recombinant α-L-iduronidase has been approved for patients with MPS-I. It reduces organomegaly and ameliorates rate of growth, joint mobility, reduces the number of episodes of sleep apnea and urinary GAG excretion. The enzyme does not cross the blood–brain barrier and does not prevent deterioration of cognition and other neurologic functions.

84. **Hunter disease is characterized by all the following features EXCEPT**

A. short stature
B. **corneal clouding**
C. dysostosis multiplex
D. intellectual disability
E. coarse facial features

Ans B Patients with severe MPS-II have features similar to those of Hurler disease except for the lack of corneal clouding and the somewhat slower progression of somatic and central nervous system (CNS) deterioration.

85. **In mucopolysaccharidosis, disproportionate severe CNS involvement with mild physical features is unique to**

A. Scheie disease
B. Hurler disease
C. Hunter disease
D. Morquio disease
E. **Sanfilippo disease**

Ans E Patients with Sanfilippo disease are characterized by slowly progressive, severe CNS involvement with mild somatic disease. Such disproportionate involvement of the CNS is unique to MPS-III.

86. **All the following are skeletal manifestations of Morquio disease EXCEPT**

A. kyphosis
B. genua valga
C. waddling gait
D. short trunk and neck
E. **bullet-shaped phalanges**

Ans E in addition to metacarpals and phalanges are abnormally short, wide and deformed with proximal pointing of the metacarpals are feature of Hurler disease.

87. **The characteristic radiographic finding of Hurler disease is**

A. kyphosis
B. genua valgus
C. V-shaped configuration of the digits
D. **hook-shaped appearance of L1 vertebra**
E. ovoid configuration of the vertebral bodies

Ans D Hurler disease, anterosuperior hypoplasia of L-1 resulting in hook-shaped appearance.Sanfilippo disease, immature, ovoid configuration of the vertebral bodies.Morquio disease, genua valgus and kyphosis. Scheie disease, the carpal bones are small leading to V-shaped configuration of the digits.

Lipid Storage Disorders

88. **Hers disease is due to deficiency of:**

A. **Liver phosphorylase**
B. Muscle phosphorylase
C. Branching enzyme
D. RBC phosphofructokinase

Ans A Liver phosphorylase with

89. **All are seen in lactose intolerance except: a.Benedict test positive in urine**

B. **Alkaline urine**
C. Acidic stool
D. Lactase enzyme deficiency

Ans B Acidic stool (pH < 5.5), Benedict's test +ve and Lactase enzyme deficiency are all features of Lactose intolerance.

90. **A 7dayold infant born with insignificant antenatal and neonatal history presents with poor feeding, vomiting, lethargy, rapidly progressive coma, metabolic acidosis, a peculiar odour of his urine, hypertonicity with severe opisthotonus which alternate with bouts of flaccidity. He was initially empirically diagnosed as having CNS infection and sepsis but was found to have hypoglycemia and the correction of hypoglycemia did not improve clinical condition. What is the most likely differential for this clinical presentation?**

A. Phenylketonuria
B. Tyrosinemia
C. **Maple syrup urine disease**
D. Isovaleric acidemia

Ans C Important clinical features of MSUD:
• Infants are normal at birth; develop poor feeding and vomiting in 1st wk of life followed by lethargy and coma.
• Physical examination reveals hypertonicity and muscular rigidity with severe opisthotonos.
• Periods of hypertonicity may alternate with flaccidity, manifested as repetitive movements of extremities
• Seizures occur in most infants, & hypoglycemia is common (correction of blood glucose does not improve clinical condition).

91. **A 7monthold infant presents with history of vomiting & failure to thrive. Patient improved with IV glucose. After one month, returns with same complaints. On evaluation, found to have high glutamine and uracil. Which is the likely enzyme defect?**

A. CPS1
B. **Ornithine transcarbamylase**
C. Arginase
D. Arginosuccinase lyase

Ans B Nelson's 20/e p 669-674 with Discussing about the findings of each of the above enzyme deficiencies, one by one. a. CPS1 • Hyperammonemia with markedly increased glutamine and alanine • Urinary orotic acid is usually low or may be absent b. Ornithine • Hyperammonemia with elevated plasma glutamine and alanine transcar- bamylase • Marked increase in urinary excretion of orotic acid* (diff. from CPS deficiency) c. Arginase • Marked elevations of arginine in plasma and CSF

92. **Ferric chloride test is used to diagnose:**

A. **Phenylketonuria**
B. Alkaptonuria
C. Hartnup disease
D. Cystinuria

Ans A Pheylketonuria Ferric chloride test is used to determine the presence or absence of phenols in a given sample. FeCl3 test with urine gives green colour in Phenylketonuria (detects phenylalanine in urine).

93. **A 5yearold child, with developmental delay, pre sented with nasal obstruction and a large head with hearing defect. There was also an associated history of sleep apnea. On examination hepatosplenomegaly was present. ECHO was suggestive of cardiac valve fibrosis. What is the most likely diagnosis?**

A. Hunter syndrome
B. **Hurler syndrome**
C. Tay-Sachs disease
D. Fragile X syndrome

Ans B Hurler disease (MPS-IH)
• Severe, progressive disorder with multiple organ & tissue involvement that results in premature death, usually by 10 yr of age.
• An infant with Hurler syndrome appears normal at birth, but inguinal hernia may be present.
• Diagnosis is usually made between 6 and 24 months with evidence of hepatosplenomegaly, coarse facial features, corneal Answers clouding, large tongue, prominent forehead, joint sti ness, short stature, and skeletal dysplasia.

94. **Gaucher disease is produced due to defect in the enzyme:**

A. **β-glucosidase**
B. Glucokinase
C. Sphingomyelinase
D. Glucose-6-phosphate dehydrogenase

Ans A βglucosidase Gaucher disease is the most common lysosomal storage disorder with a defect of lysosomal enzyme acid β glucosidase.

95. **True about Gaucher's disease:**

A. **PAS positive cells**
B. Oil Red O positive cells
C. Deficiency of acid sphingomyelinase
D. Deficiency of glucocerebrosidase
E. Gaucher cells are present

Ans A d. Deficiency of glucocerebrosidase, e. Gaucher cells are present

96. **About Wilson disease, all are true except:**

A. Autosomal recessive
B. KF ring
C. Raised copper level
D. **Raised ceruloplasmin level**

Ans D

97. **Dried blood spot test in neonates is used in testing for:**

A. **Inborn error of metabolism**
B. Blood group
C. Total cell count
D. Creatinine and bilirubin

Ans A Tandem Mass spectrometry (TMS) done on dried blood spots is one of the screening tests for inborn errors

98. **A 5-year-old mentally retarded girl presented with repeated convulsions and abnormal EEG finding. On examination, she had a light skin, small head, and exaggerated reflexes. Of the following, the MOST appropriate method to confirm the diagnosis is**

A. Guthrie test
B. flow cytometry
C. tandem mass spectrometry
D. measurement of phenylketones in the urine
E. **measurement of plasma phenylalanine concentration**

Ans E In infants with positive screening results, diagnosis should be confirmed by quantitative measurement of plasma phenylalanine concentration.

99. **An 11-month-old boy presented with repeated convulsions, poor eye contact, exaggerated startle response to noise, and large head. Ophthalmic exam revealed cherry-red spot. Of the following, the MOST likely diagnosis is**

A. Tyrosinemia
B. Fabry disease
C. Gaucher disease
D. **Tay-Sachs disease**
E. Niemann-pick disease

Ans D results from the deficiency of β-hexosaminidase activity. Clinical manifestations in infancy including loss of motor skills, increased startle reaction, and macular pallor and retinal cherry-red spots.

100. **A 3-year-old girl presented with growth retardation, recurrent attacks of otitis media, diarrhea, and hypoglycemic seizures. On examination, there is hepatomegaly and her absolute neutrophil count (ANC) is 600/µL. Of the following, the MOST likely diagnosis is**

A. Kostmann disease
B. cyclic neutropenia
C. drug-induced neutropenia
D. **glycogen storage disease Ib**
E. Shwachman-Diamond syndrome

Ans D In patients with GSD Ib, the loss of mucosal barrier function as a result of inflammation, which is likely related to the disturbed neutrophil function, seems to be the main cause of diarrhea.

101. **All the following mucopolysaccharidoses are inherited as autosomal recessive**

A. Hurler disease
B. Scheie disease
C. **Hunter disease**
D. Morquio disease
E. Sanfilippo disease

Ans C inherited as X- linked recessive.

102. **The least common presentation of acute intermittent porphyria is**

A. sensory loss
B. mental symptoms
C. muscle weakness
D. **abdominal tenderness**
E. pain in the extremities

Ans D The common manifestations include mental symptoms; pain in the extremities, head, neck, or chest; muscle weakness; and sensory loss. Because all these manifestations are neurologic rather than inflammatory, there is little or no abdominal tenderness, fever, or leukocytosis.

103. **In X-linked adrenoleukodystrophy (ALD),MRI shows characteristic cerebral white matter lesions mainly in**

A. frontal lobes
B. temporal lobes
C. frontoparietal lobes
D. **parietoccipital lobes**
E. temporoparietal lobes

Ans D In 80% of patients, the lesions are symmetric and involve the periventricular white matter in the posterior parietal and occipital lobes. Approximately 50% show a garland of accumulated contrast material adjacent and anterior to the posterior hypodense lesions. In 12% of patients, the initial lesions are frontal.

104. **The MOST common initial manifestation of X-linked adrenoleukodystrophy (ALD) is**

A. seizure
B. **hyperactivity**
C. visual disturbance
D. increased intracranial pressure
E. impaired auditory discrimination

Ans B In the childhood cerebral form of ALD, symptoms are first noted most commonly between the ages of 4 and 8 yr. The most common initial manifestations are hyperactivity, which is often mistaken for an attention deficit disorder, and worsening school performance in a child who had previously been a good student.

Amino Acid Disorders

105. **A child presents with hepatomegaly and hypoglycemia. There is no improvement in blood sugar even after administration of epinephrine. What is the likely diagnosis?**

A. **Von Gierke's disease**
B. Anderson's disease
C. Pompe's disease
D. McArdle's disease

Ans A • Epinephrine increases blood glucose level by stimulating glycogenolysis and gluconeogenesis, mediated by the enzyme glucose 6 phosphatase, that converts Glucose-6-phosphate to Glucose
• Von Gierke disease is due to deficiency of Glucose 6 phosphatase enzyme; Hence, epinephrine is not able to increase blood glucose in a child with Von Gierke disease, but lactate levels increase.

106. **Second degree consanguineous marriage, baby with diarrhea, perianal diaper area redness suggests ?**

A. **Lactose intolerance**
B. Shigella diarrhea
C. Salmonella
D. Fungal infection

Ans A Ref: Lactose intolerance by Shinjini Bhatnagar. BMJ. 2007 Jun 30; 334(7608): 1331–1332. Lactose intolerance
• It is due to lactase deficiencyQ, which can be primary, secondary, congenital, or developmental
• It should be suspected in people with abdominal symptoms after ingestion of milkQ and milk products
• It may lead to diarrhea, dehydration and growth failure; Perianal excoriationsQ due to acidic stools are common.

107. **Mutation seen in Hartnup's disease:**

A. SLC6A
B. **SLC6A**
C. SLC6A
D. SLC6A

Ans B 19 Hartnup's disease is due to mutation of SLC6A19 gene encoding the neutral amino acid transporter, resulting in deficiency of nicotinamide synthesis

108. **In Hartnup disorder, metabolism of which of the follo wing amino acids is affected?**

A. Tyrosine
B. **Tryptophan**
C. Phenylalanine
D. Homocysteine

Ans B In Hartnup disorder, there is a defect in transport of monoamino-monocarboxylic amino acids (neutral amino acids), including tryptophan, by intestinal mucosa & renal tubules due to mutation of SLC6A19 gene located on chr 5p15

109. **In Hartnup disorder what is seen?**

A. **Pellagra like rash**
B. Burn feet rash
C. Alopecia
D. Vitiligo

Ans A

110. **Treatment of Alkaptonuria:**

A. Vitamin A
B. **Vitamin C**
C. Vitamin D
D. Vitamin K

Ans B Some patients with Alkaptonuria benefit from highdose vitamin C; In Alkaptonuria, Vitamin C has been shown to decrease the buildup of brown pigment in cartilage & may slow the development of arthritis.

111. **Which of the following is false about alkaptonuria?**

A. **Genitourinary system not involved**
B. Homogentisic acid oxidase deficiency
C. Black urine
D. Calcification in vertebral bodies

Ans A Genitourinary system is also involved in Alkaptonuria, as this enzyme is expressed only in liver and kidneys; darkening of urine on standing is an important feature of Alkaptonuria. c. Mental retardation Clinical features of Hartnup disease:
• Most children with Hartnup defect remain asymptomatic
• Major clinical manifestation in symptomatic patient is cutaneous photosensitivity: pellagralike rash on sun exposure

112. **Not a feature of Hartnup's disease:**

A. Pellagroid skin lesion
B. Cerebellar ataxia
C. **Mental retardation**
D. Psychological disturbances

Ans C Hartnup disease features pellagra-like photosensitive skin lesions, cerebellar ataxia, and psychiatric disturbance from tryptophan malabsorption, but intellectual disability is not a consistent feature — many patients are asymptomatic or have normal cognition.

113. **Massive aminoaciduria without a corresponding increase in plasma amino acid level is characteristic of which one of the following diseases?**

A. Homocystinuria
B. **Hartnup disease**
C. Tyrosinemia
D. Maple syrup urine disease

Ans B Laboratory findings in Hartnup disorder: fo Aminoaciduria, involving neutral amino acids (alanine, serine, threonine, valine, leucine, isoleucine, phenylalanine, tyrosine, weiveR tryptophan, histidine); Plasma concentrations of neutral amino acids are usually normal. This seemingly unexpected finding occurs because these amino acids are absorbed as dipeptides and the transport system for small peptides is intact in Hartnup disorder.

114. **False about hartnup's disease:**

A. Defect in neutral amino acid transport
B. **Mental retardation is the common presentation**
C. Most children are asymptomatic
D. Photosensitivity

Ans B

115. **Deficiency of enzyme alphaketo acid decarboxylase leading to a block in the metabolism of branch chain amino acids is observed in:**

A. Maple syrup urine disease
B. Hartnup's disease
C. Alkaptonuria
D. Phenylketonuria

Ans A

116. **The MOST appropriate method to confirm the diagnosis of tyrosinemia type 1 is by elevated level of**

A. α-fetoprotein
B. plasma tyrosine
C. serum methionine
D. serum succinylacetone
E. urinary 5-aminolevulinic acid

Ans D The presence of elevated levels of succinylacetone in serum and urine is diagnostic for tyrosinemia type I. Increased levels of α-fetoprotein are present in the cord blood of affected infants, indicating intrauterine liver damage. Plasma tyrosine levels are usually elevated at diagnosis but this is a nonspecific finding and is dependent on dietary intake. Plasma levels of other amino acids, particularly methionine, may also be elevated in patients with liver damage.

117. **The MOST appropriate treatment of homocystinuria is**

A. betaine
B. folic acid
C. vitamin C
D. vitamin B6
E. methionine restriction

Ans D Treatment with high doses of vitamin B6 (200-1,000 mg/24 hr) causes dramatic improvement in most patients who are responsive to this therapy.

118. **The diagnosis of homocystinuria is usually made after 3 yr of age when the ophthalmologist found**

A. cataracts
B. glaucoma
C. astigmatism
D. ectopia lentis
E. retinal detachment

Ans D Infants with homocystinuria are normal at birth. Clinical manifestations during infancy are nonspecific and may include failure to thrive and developmental delay. The diagnosis is usually made after 3 yr of age, when subluxation of the ocular lens (ectopia lentis) occurs.

119. **The MOST common electrolyte abnormality in acute intermittent porphyria is**

A. hyperkalemia
B. hyponatremia
C. hypercalcemia
D. hypomagnesemia
E. hypophosphatemia

Ans B is common during acute attacks. Inappropriate antidiuretic hormone secretion is often the most likely mechanism, but salt depletion from excess renal sodium loss, gastrointestinal loss, and poor intake have been suggested as causes of hyponatremia in some patients. Other electrolyte abnormalities may include hypomagnesemia and hypercalcemia.

120. **All the following are secondary causes of hypercholesterolemia EXCEPT**

A. obesity
B. cholestasis
C. hypothyroidism
D. anorexia nervosa
E. nephrotic syndrome

Ans A is secondary cause of hypertriglyceridemia.

121. **Xanthomas are a feature of all the following EXCEPT**

A. familial hypercholesterolemia
B. familial dysbetalipoproteinemia
C. familial combined hyperlipidemia
D. familial chylomicronemia (Frederickson type I)
E. familial hypertriglyceridemia(Frederickson type V)

Ans C an autosomal dominant condition characterized by moderate elevation in plasma LDL cholesterol and triglycerides, and reduced plasma HDL cholesterol.

Organic Acidurias

122. **A child presents with massive hepatomegaly and hypoglycemia. There is no improvement in blood glucose on administration of glucagon. The probable diagnosis is:**

A. Von Gierke disease
B. McArdle disease
C. Coris disease
D. Forbe's disease

Ans A

123. **A child with low blood glucose is not able to do glyco geno lysis or gluconeogenesis. Which of the following enzyme is missing in the child?**

A. Fructokinase
B. Glucokinase
C. Glucose 6-phosphatase
D. Transketolase

Ans C Glucose 6 phosphatase Glycogenolysis or gluconeogenesis is mediated by glucose 6 phosphatase which is absent in Von Gierke disease. c. Galactose The given clinical features of vomiting, abdominal distension, diarrhea and presence of reducing substance in urine, in a 3-day-old neonate, who is probably on exclusive breast feeding, suggest an underlying diagnosis of Galactosemia.

124. **Nitisinone is used in the treatment of:**

A. Phenylketonuria
B. Alkaptonuria
C. Homocystinuria
D. Tyrosinemia

Ans D Nitisinone is the treatment of choice is for Tyrosinemia type I;

125.An infant presents with history of seizures and skin rashes. Investigations show metabolic acidosis increased blood ketone levels and normal NH . This child is likely 3 to be suffering from:

A. Propionic aciduria
B. Urea cyclic disorder
C. Phenylketonuria
D. **Multiple carboxylase deficiency**

Ans D Discussing each option one by one: Option a. Skin rashes are not seen in Propionic aciduria Option c. Normal NH level excludes urea cycle defect Option b. Absence of hypopigmentation, presence of metabolic acidosis & increased blood ketones excludes PKU Option d. Skin rashes with presence of ketones and metabolic acidosis are all features of Multiple carboxylase deficiency

126.The enzyme that is deficient in homocysteinuria:

A. Tyrosinase
B. Sphingomyelinase
C. **Cystathionine synthase**
D. Acid lipase

Ans C

127. A 4monthboy presents with failure to thrive. He had several upper respiratory tract infections during the last two months. On examination, hepatosplenomegaly and mild hypotonia are noted. Blood tests reveal pancytopenia & transaminitis. Chest Xray shows reticulonodular pattern and calcified nodules. Biopsy of liver shows foamy histiocytes. What is the expected ophthalmological finding in this patient?

A. **Cherry red macula**
B. Cataracts
C. Kayser-Fleisher ring
D. Foveal hypoplasia

Ans A The given history of failure to thrive, recurrent respiratory infections with the given X-ray findings and foamy histiocytes on liver biopsy, point to a diagnosis of NiemannPick disease, in which cherry red spot is seen.

128.Enzyme replacement therapy is not available for which of the following?

A. Gaucher's disease
B. Pompe disease
C. **Sanfilippo disease**
D. Fabry's disease

Ans C

129.Metachromatic leukodystrophy is due to deficiency of:

A. β galactosidase
B. **Arylsulfatase A**
C. Glycogen phosphorylase
D. Medium chain acyl-coA synthetase

Ans B Metachromatic leukodystrophy results from deficiency of arylsulfatase A, causing accumulation of sulfatides and progressive demyelination.

130.Which of the following is true for Wilson disease?

A. High ceruloplasmin
B. **Low serum ceruloplasmin and high urinary copper**
C. Low serum copper
D. Low urinary copper

Ans B

131. Patient with growth retardation, delayed milestones with kinky hair. What is the diagnosis?

A. **Menke's disease**
B. Trisomy
C. Lesch Nyhan syndrome
D. Wilson disease

Ans A Menke's disease
• It is caused by mutations in the gene encoding Cu2+ transporting ATPase (ATP7A)
• Copper level in liver & brain are low in contrast to an increase in enterocytes & fibroblasts.
• Progressive cerebral degeneration (seizures), feeding difficulties, failure to thrive, hypothermia, apnea, hair), hypopigmentation, bone changes & cutis laxa seen;
• Patients with the classic form of Menke's disease usually die before 3 yr age.

132.In the classic phenylketonurea, the affected infant is normal at birth, intellectual disability develops gradually if the infant remains untreated, and cognitive delay may not be evident for the first few months. All the following are true about neonatal screening program of this condition EXCEPT

A. Guthrie test was the 1st method used
B. **optimal time for Guthrie test at 7-14 of life**
C. blood phenylalanine may rise to a diagnostic level as early as 4 hr after birth
D. fluorometric and tandem mass spectrometry give a low false-positive rate
E. diagnosis must be confirmed by measurement of plasma phenylalanine concentration

Ans B It is recommended that the blood for screening be obtained in the first 24-48 hr of life after feeding protein to reduce the possibility of false-negative results, especially in the milder forms of the condition.

133.One of the following is a characteristic feature for both Infantile Sandhoff disease and Tay-Sachs disease

A. splenomegaly
B. hepatomegaly
C. cardiac involvement
D. **retinal cherry-red spots**
E. mild bony abnormalities

Ans D Both of Sandhoff disease and Tay-Sachs disease in infant characterize by a neurologic features and retinal cherry-red spots. Infants with Sandhoff disease have hepatosplenomegaly, cardiac involvement, and mild bony abnormalities.

134.A 9-month-old boy presented with persistent head lag, rapid breathing, difficult feeding, and hepatomegaly. Chest X-ray showed cardiomegaly. Of the following, the MOST appropriate confirmatory test is

A. liver biopsy
B. muscle biopsy
C. electrocardiography
D. serum creatine phosphokinase
E. enzyme assay in dried blood spots

Ans E The confirmatory step for a diagnosis of Pompe disease is enzyme assay demonstrating deficient acid α-glucosidase. The enzyme assay is usually done in dried blood spots, leukocytes, blood mononuclear cells, muscle, and cultured skin fibroblasts.

135.Which of the following mucopolysaccharidoses have normal intelligence?

A. Sly disease
B. Hurler disease
C. Hunter disease
D. Morquio disease
E. Sanfilippo disease

Ans D is characterized by short-trunk dwarfism, fine corneal deposits, a skeletal dysplasia that is distinct from other mucopolysaccharidoses, and preservation of intelligence.

136.All the following are features of congenital erythropoietic porphyria (CEP) EXCEPT

A. neuropathy
B. reddish urine
C. erythrodontia
D. photosensitivity
E. neonatal hyperbilirubinemia

Ans A Neuropathic symptoms are absent, and there is no sensitivity to drugs, hormones, and carbohydrate restriction.

137. All the following drugs are secondary causes of hypertriglyceridemia EXCEPT

A. tegretol
B. estrogen
C. thiazides
D. β-blockers
E. anabolic steroids

Ans A is secondary causes of hypercholesterolemia.

Urea Cycle Disorders

138.All are liver glycogenosis except:

A. Von Gierke disease
B. Her's disease
C. Type III glycogenosis
D. Pompe's disease

Ans D

139.A 3daychild vomits everything he feeds, has a distended abdomen and diarrhea. The urine is positive for Benedict's test for reducing substance. The substance in urine is:

A. Sucrose
B. Glucose
C. Galactose
D. Fructose

Ans C A neonate with vomiting, diarrhea, abdominal distension after milk feeds and a reducing substance in the urine (Benedict's test) that is not glucose points to galactosemia — the reducing sugar is galactose, derived from lactose in milk.

140.A child with subluxation of lens and mental retardation died. Which of the following would have helped in his treatment?

A. Biotin
B. Pyridoxine
C. Zinc
D. Riboflavin

Ans B Subluxation of lens and mental retardation in a child who died suggests underlying homocystinuria; Pyridoxine (vitamin B6) is useful in its treatment;

141. HHH syndrome is caused by defect in:

A. Ornithine permease
B. Ornithine transcarbamyolase
C. Arginase
D. Argininosuccinase lyase

Ans A Hyperammonemia-Hyperornithinemia-Homocitrullinemia (HHH) Syndrome: In this rare autosomal recessive disorder, defect is in transport system of ornithine from cytosol into mitochondria, which causes an accumulation of ornithine in the cytosol and a deficiency of ornithine inside mitochondria.

142.Not a correct match:

A. Alkaptonuria-homogentisate oxidase
B. Albinism-tyrosinase
C. Tyrosinemia-fumaryl acetoacetate hydrolase
D. Phenylketonuria-aryl sulphatase A

Ans D

143.NiemannPick disease is due to deficiency of one of the following enzymes–which one?

A. Arylsulfatase
B. Acid Sphingomyelinase
C. Hexosaminidase A
D. Ceramidase

Ans B

144.TaySachs disease is due to deficiency of:

A. Beta-galactosidase
B. Hexosaminidase
C. Sphingomyelinase
D. Glucocerebrosidase

Ans B Tay-Sachs disease is due to Hexosaminidase A deficiency.

145. **White matter is involved in which of the following diseases?**

A. Canavan disease
B. Gaucher disease
C. GM1 Gangliosidosis
D. Mucopolysaccharidosis

Ans A

146. **The gene for Wilson's disease is on:**

A. Long arm of Chromosome
B. Long arm of Chromosome
C. Short arm of Chromosome
D. Short arm of Chromosome

Ans A

147. **Hyperuricemia is caused in newborn due to defect in:**

A. Glucose 6 phosphate dehydrogenase
B. Glucose 6 phosphatase
C. Xanthine oxidase
D. Hypoxanthine guanine ribosyltransferase
E. Ornithine transcarbamylase

Ans B d. Hypoxanthine guanine ribosyl transferase Plasma uric acid is elevated in Von Gierke disease & Lesch Nyhan-disease while it is very low in xanthine

148. **A 4-month-old boy presented with failure to thrive, rapid breathing, and repeated vomiting. On examination, there is hepatomegaly and abdominal sonography revealed renal calcification. Of the following, the MOST likely urine odor of this infant is**

A. mousey
B. rotting fish
C. sweaty feet
D. boiled cabbage
E. swimming pool

Ans D Tyrosinemia is the most likely diagnosis; these children have an unpleasant odor which has been described as boiled cabbage, rancid butter.

149. **A 12-year-old boy presented with epistaxis, mild bruising, intermittent bone pain, and massive splenomegaly. Complete blood count shows thrombocytopenia and mild anemia. Radiological study revealed lytic lesions and Erlenmeyer flask deformity of the distal femur. Of the following, the MOST likely diagnosis is**

A. Gaucher disease
B. Sandhoff disease
C. Tay-Sachs disease
D. von Gierke disease
E. Niemann-pick disease

Ans A is a multisystemic lipidosis characterized by hematologic abnormalities, organomegaly, and skeletal involvement, the latter usually manifesting as bone pain and pathologic fractures. It is one of the most common lysosomal storage diseases, results from the deficient activity of the lysosomal hydrolase, acid β-glucosidase, which is encoded by a gene located on chromosome 1q21-q31. The pathologic hallmark of Gaucher disease is the Gaucher cell in the reticuloendothelial system, particularly in the bone marrow.

150. **A 7-month-old girl presented with recurrent chest infection, chocking during feeding, large protruded tongue, persistent head lag, and hepatomegaly. Chest X-ray showed cardiomegaly. All the following are beneficial in the treatment of this patient EXCEPT**

A. exercise therapy
B. high-protein diet
C. cardiac transplantation
D. nocturnal ventilatory support
E. specific enzyme replacement therapy

Ans C Specific enzyme replacement therapy (ERT) with recombinant human acid α- glucosidase is available for treatment of Pompe disease and is capable of preventing deterioration or reversing abnormal cardiac and skeletal muscle functions. A high- protein diet and exercise therapy may also be beneficial. Nocturnal ventilatory support, when indicated, should be used. It has been shown to improve the quality of life and is particularly beneficial during a period of respiratory decompensation.

151. **A 5-year-old boy presented with chronic diarrhea. On examination he had hepatosplenomegaly, coarse facial features, joint stiffness, clear cornea, and short stature. Of the following, the MOST likely diagnosis is**

A. Hurler disease
B. Hunter disease
C. Scheie disease
D. Morquio disease
E. Sanfilippo disease

Ans B

152. **The MOST important screening test of acute intermittent porphyria is**

A. plasma porphyrins
B. fibroblast porphyrins
C. fecal porphobilinogen
D. erythrocyte porphyrins
E. urinary porphobilinogen

Ans E 🅿🅾 Porphyria cutanea tarda ---------- Plasma (or urine) porphyrins. 🅿🅾 Erythropoietic protoporphyria ---- Erythrocyte (or plasma) porphyrins.

153. **Smith Lemli-Opitz Syndrome (SLOS) is characterized by all the following EXCEPT**

A. epispadias
B. cleft palate
C. microcephaly
D. retromicrognathia
E. mental retardation

Ans A Genital anomalies; hypospadias, cryptorchidism, and sexual ambiguity (<50%).

Mitochondrial Disorders

154. **An infant has hepatosplenomegaly, hypoglycemia, hyper lipidemia, acidosis and normal structured glycogen depo sition in liver. What is the diagnosis?**

A. Her's disease
B. **Von Gierke's disease**
C. Cori's disease
D. Anderson's disease
E. Pompe's disease

Ans B Hepatorenomegaly, hypoglycemia, hyperlipidemia, acidosis and normal structured glycogen deposition in liver is suggestive of Von Gierke's disease; Normal structured glycogen excludes options c. Cori's and d. Anderson's disease.

155. **Child of Vasanthi was weaned from breast milk on the 5th day and was given sugarcane juice. The child developed hypoglycemia and hepatomegaly. Biochemical examination showed hypophosphatemia and reducing substances in urine. The child is probably suffering from which of the following enzyme deficiencies?**

A. Fructokinase
B. **Aldolase B**
C. Glucose 6-phosphatase
D. Beta-galactosidase DISORDERS OF AMINO ACID METABOLISM Questions

Ans B In this question as the symptoms have developed following administration of sugarcane juice, so this suggests hereditary fructose ntolerance in which there is deficiency of the enzyme aldolase B.

156. **Boiled cabbage urine odor is seen in:**

A. **Tyrosinemia**
B. Isovaleric aciduria
C. Maple syrup urine disease
D. Phenylketonuria

Ans A

157. **Intervertebral disc calcification is seen in which of the following?**

A. Rickets
B. **Alkaptonuria**
C. Scurvy
D. Gaucher disease

Ans B

158. **Treatment of multiple carboxylase deficiency:**

A. **Biotin**
B. Pyridoxine
C. Thiamine
D. Folic acid (AIPGMEE 07)

Ans A

159. **Wrinkled paper appearance of macrophage in bone marrow aspirate is seen in:**

A. Niemann Pick disease
B. **Gaucher disease**
C. Hemophagocytic lymphohistiocytosis
D. Langerhan cell histiocytosis

Ans B • Gaucher cells are Lipid Laden Macrophages have a characteristic histologic appearance of "wrinkled tissue paper" • Membranebound inclusions filled with glucocerebroside are seen with electron microscopy.

160. **The following is an Xlinked mucopolysaccharidoses:**

A. Hurler's syndrome
B. **Hunter's syndrome**
C. Scheie's syndrome
D. Sanfilippo's syndrome

Ans B All mucopolysaccharidoses are autosomal recessive except Hunter's disease which is inherited as X-linked recessive.

161. **An infant presents with irritability, increased tone of extremities and recurrent seizures. Tissue examination reveals globoid cells in parenchyma around blood vessels. What is the most probable diagnosis?**

A. Tay Sach's disease
B. **Krabbe disease**
C. Adrenoleukodystrophy
D. Canavan's disease

Ans B

162. **Congenital Wilson's disease is characterized by:**

A. KF ring is present at birth
B. **May present as acute hepatitis**
C. Decreased urinary copper excretion
D. Decreased hepatic copper concentration

Ans B KayserFleischer rings are absent in young patients with hepatic Wilson disease up to 50% of the time but are present in 95% of patients with neurologic symptoms.

163. **A child who had derangement of milestones and was hyperactive later on developed self mutilating behavior and hair pulling behavior. The child died a few years later. Which of the following enzyme is most likely deficient in the child?**

A. **HGPRT**
B. Phenylalanine hydroxylase
C. Adenosine deaminase
D. Hexosaminidase A 126 Section 1: General Pediatrics

Ans A Delayed development, hyperactivity and self-mutilating behavior in a child who died later is suggestive due to HGPRT deficiency. Errors of Metabolism 133 Answers while infants may present in early infancy. Scalp hair with nodosa (fractures of metabolism. hair abnormalities (kinky oxidase deficiency. of Lesch- Nyhan disease A 1 month old buaebsyt piorensents with Q the following picture. He h 1ad a his- tory of neonatal sepsis requiring hospitalization. His Hb was 15 g/dl, Total leucocyte count was 18,000

164. **A 10-month-old boy presented with failure to thrive, fever, jaundice, hepatomegaly, and severe rickets. Investigations revealed hypoglycemia and normal anion gap metabolic acidosis. Of the following, the MOST likely enzyme deficiency is**

A. acid β-glucosidase
B. β-hexosaminidases
C. acid sphingomyelinase
D. phenylalanine hydroxylase
E. **fumarylacetoacetate hydrolase**

Ans E Tyrocinemia type 1 is a severe disease of the liver, kidney, and peripheral nerves are caused by a deficiency of the enzyme fumarylacetoacetate hydrolase. 🆘 acid β-glucosidase ---------- Gaucher disease 🆘 phenylalanine hydroxylase ------ Phenylketonurea 🆘 acid sphingomyelinase ---- Niemann-pick disease 🆘 fumarylacetoacetate hydrolase ------- Tyrosinemia 🆘 β-hexosaminidases ----------=-- Tay-Sachs disease

165. **A 9-year-old boy admitted to the hospital because of fracture of left femur due to a minor trauma. Past history revealed recurrent attacks of epistaxis and intermittent generalized bone pain. On examination, there is massive splenomegaly. Of the following, the MOST appropriate practical treatment of this disease is**

A. gene therapy
B. liver transplantation
C. **enzyme replacement**
D. substrate reduction agents
E. bone marrow transplantation

Ans C Treatment of patients with Gaucher disease type 1 includes enzyme replacement therapy. The efficacy of enzyme replacement therapy with mannose- terminated recombinant human acid β-glucosidase has definitively been demonstrated. Most symptoms (organomegaly, hematologic indices, bone pain) are reversed by enzyme replacement therapy (60 IU/kg) administered by intravenous infusion every other week and the bone involvement can be stabilized or improved.

166. **A 28-day-old boy presented with prolonged neonatal jaundice, lethargy, poor feeding, and failure to regain birth weight. On examination there are hepatomegaly and cataract. Of the following, the MOST likely injured organs in this disease are**

A. eyes, liver, and brain
B. kidney, liver, and eyes
C. **kidney, liver, and brain**
D. kidney, heart , and brain
E. pancreas, liver, and brain

Ans C In galactosemia, without the transferase enzyme, the infant is unable to metabolize galactose-1-phosphate, the accumulation of which results in injury to kidney, liver, and brain.

167. **Which of the following mucopolysaccharidoses is susceptible to life-threatening atlantoaxial instability and dislocation?**

A. Hurler disease
B. Scheie disease
C. Hunter disease
D. **Morquio disease**
E. Sanfilippo disease

Ans D In Morquio disease, instability of the odontoid process and ligamentous laxity is regularly present and can result in life-threatening atlantoaxial instability and dislocation.

168. **In older infants and children, hypoglycemia is defined as whole blood glucose concentration less than**

A. 45 mg/dL
B. 50 mg/dL
C. **55 mg/dL**
D. 60 mg/dL
E. 65 mg/dL

Ans C In older infants and children, a whole blood glucose concentration of <55 mg/dL (10-15% higher for serum or plasma) represents hypoglycemia, because counter regulatory mechanisms are activated at these glucose concentrations.

169. **A 3-year-old boy presented to emergency unit at 10:00 AM with history of difficult arousal from sleep, lethargy, vomiting, and seizures; the mother stated that he had slept without having his supper. Fasting blood sugar was 38mg/dL and urinary ketone was low. Of the following, the MOST likely diagnosis is**

A. galactosemia
B. propionic acidemia
C. ketotic hypoglycemia
D. hereditary fructose intolerance
E. **medium-chain acyl-CoA dehydrogenase (MCAD) deficiency**

Ans E MCAD ; Previously undiagnosed affected patients usually present in the first 3 mo-5 yr of life with episodes of acute illness triggered by prolonged fasting (longer than 12-16 hr). During acute episodes of MCAD deficiency, hypoglycemia is usually present. Plasma and urinary ketone concentrations are inappropriately low (hypoketotic hypoglycemia).

170. **The MOST common presentation of mitochondrial β-oxidation of fatty acids disorders is**

A. cardiomyopathy
B. muscle weakness
C. renal tubulopathy
D. **hypoglycemic coma**
E. acute rhabdomyolysis

Ans D The most common presentation is an acute episode of life-threatening coma and hypoglycemia induced by a period of fasting because of defective hepatic ketogenesis.

Fatty Acid Oxidation Disorders

171. **An 8 days old neonate presents with hypoglycemia (Glucose–17 mg%) and jaundice is diagnosed to have metabolic disorder. Most likely diagnosis:**

A. Hypothyroidism
B. **Galactosemia**
C. Glycogen storage disease
D. Fatty acid oxidation defect

Ans B Hypoglycemia and jaundice in a neonate, after initiation of breastfeeding, suggests a diagnosis of Galactosemia.

172. **Mental retardation is seen in:**

A. **Phenylketonuria**
B. Alkaptonuria
C. Gaucher disease
D. Von Gierke disease

Ans A

173. **Most sensitive test to identify metabolic disease in a newborn:**

A. Tandem mass spectrometry
B. Arterial blood gas
C. Complete hemogram
D. Liver function test

Ans A Tandem mass spectrometry (TMS) is the latest technical advance which requires a few drops of blood to be placed on a filter paper and mailed to a central laboratory for assay to diagnose a large number of genetic conditions. As it is a very sensitive test, it is used as one of the screening tests for suspected metabolic diseases.

174. **A normal born child presents with mental retardation, blonde hair and convulsions at 1 year of age. Most probable diagnosis is:**

A. Albuminuria
B. Phenylketonuria
C. Gaucher's disease
D. Tyrosinemia

Ans B Hypopigmentation and CNS involvement with seizures and mental retardation suggest Phenylketonuria.

175. **Darkening of urine on standing is associated with:**

A. Alkaptonuria
B. Cystinuria
C. Fabry's disease
D. Tyrosinemia

Ans A Darkening of urine on standing is associated with Alkaptonuria, due to Homogentisic acid oxidase deficiency.

176. **Deposition of cells with crumpled tissue paper appearance of cytoplasm is seen in:**

A. Gaucher disease
B. Niemann Pick disease
C. Tay Sach disease
D. Mucopolysaccharidosis124 Section 1: General Pediatrics

Ans A

177. **Only males are affected in:**

A. Scheie's syndrome
B. Hunter's syndrome
C. Hurler's syndrome
D. Gaucher's disease

Ans B As Hunter's disease is a X linked recessive, males are mostly affected by it.

178. **Hyperdense basal ganglia and hypodensity of the white matter on CT scan is diagnostic of:**

A. Alexander's disease
B. Krabbe's disease
C. Canavan's disease
D. Metachromatic leukodystrophy

Ans B

179. **True about Wilson's disease:**

A. Increase in urinary copper and increased serum ceruloplasmin and copper
B. Increased serum ceruloplasmin levels with increased urinary copper
C. Elevated hepatic copper level and increased serum ceruloplasmin levels
D. Increased in urinary copper and decreased serum ceruloplasmin

Ans D

180. **Inborn errors of metabolism (IEM) are hereditary biochemical disorders caused by single-gene mutations that result in alteration of**

A. chromosomal number or structure
B. mitochondrial synthesis or function
C. primary fat structure or the amount of fat synthesized
D. primary protein structure or the amount of protein synthesized
E. primary carbohydrate structure or the amount of carbohydrate synthesized

Ans D Many childhood conditions are caused by single-gene mutations that encode specific proteins. These mutations can result in the alteration of primary protein structure or the amount of protein synthesized. The function of a protein, whether it is an enzyme, receptor, transport vehicle, membrane component, or structural element, may be compromised or abolished.

181. **A 15-month-old boy had recurrent hospital admissions because of fever, jaundice, subcutaneous bleeding, and hypoglycemic fits; on examination, there is hepatomegaly. In the last attack, the child developed severe legs pain associated with retraction of the neck and trunk. Of the following, the MOST likely diagnosis is**

A. galactosemia
B. organic acidemia
C. urea cycle defect
D. tyrosinemia type
E. maple syrup urine disease

Ans D Tyrocinemia type 1 is a severe disease of the liver, kidney, and peripheral nerves are caused by a deficiency of the enzyme fumarylacetoacetate hydrolase. An acute hepatic crisis commonly heralds the onset of the disease and is usually precipitated by an intercurrent illness that produces a catabolic state. Episodes of acute peripheral neuropathy occur in approximately 40% of affected children. Renal nvolvement is manifested as a Fanconi-like syndrome.

182. **A 2-year-old boy presented with failure to thrive, regression of developmental milestone, and difficult to contact with surroundings. On examination, there are hepatosplenomegaly and moderate lymphadenopathy. Of the following, the MOST likely diagnosis is**

A. Gaucher disease
B. Sandhoff disease
C. Tay-Sachs disease
D. Von Gierke disease
E. **Niemann-pick disease**

Ans E (NPD), inherited as autosomal recessive traits, the clinical manifestations and course of type A NPD is uniform and is characterized by a normal appearance at birth. Hepatosplenomegaly, moderate lymphadenopathy, and psychomotor retardation are evident by 6 mo of age, followed by neurodevelopmental regression and death by 3 yr. With advancing age, the loss of motor function and the deterioration of intellectual capabilities are progressively debilitating; and in later stages, spasticity and rigidity are evident.

183. **A 20-day-old girl presented with prolonged neonatal jaundice, lethargy, and poor feeding treated as sepsis by empirical antibiotics and nothing by mouth. The patient got partial response, when oral feeding resumed, the condition deteriorate again. In this condition, the blood culture MOST likely reveals growth of**

A. **Escherichia coli**
B. Staphylococcal aureus
C. group B streptococcus
D. Listeria monocytogens
E. Pseudomonas aeruginosa

Ans A Patients with galactosemia are at increased risk for Escherichia coli neonatal sepsis; the onset of sepsis often precedes the diagnosis of galactosemia.

184. **A definitive diagnosis of mucopolysaccharidoses disorder is established by**

A. liver biopsy
B. **enzyme assay**
C. radiographic features
D. bone marrow examination
E. urinary glycosaminoglycan (GAG) test

Ans B Any individual who is suspected of a mucopolysaccharidoses disorder based on clinical features, radiographic results, or urinary GAG screening tests should have a definitive diagnosis established by enzyme assay. Serum, leukocytes, or cultured fibroblasts are used as the tissue source for measuring lysosomal enzymes.

185. **The least well-defined long-term sequelae of severe prolonged hypoglycemia is**

A. cerebral palsy
B. **personality affection**
C. cognitive impairment
D. recurrent seizure activity
E. autonomic dysregulation

Ans B Subtle effects on personality are also possible but have not been clearly defined.

186. **All the following are recognized laboratory findings of medium-chain acyl-CoA dehydrogenase (MCAD) deficiency EXCEPT**

A. hypoglycemia
B. hypoketonuria
C. hypoketonemia
D. **metabolic acidosis**
E. elevated liver enzyme

Ans D During acute episodes of MCAD deficiency, hypoglycemia is usually present. Plasma and urinary ketone concentrations are inappropriately low (hypoketotic hypoglycemia). Because of the hypoketonemia, there is little or no metabolic acidosis, which is expected to be present in many children with hypoglycemia.

187. **The MOST common presentation of primary carnitine deficiency is**

A. liver disease
B. **cardiomyopathy**
C. skeletal myopathy
D. endocardial fibroelastosis
E. hypoketotic hypoglycemia

Ans B The most common presentation of primary carnitine deficiency is progressive cardiomyopathy with or without skeletal muscle weakness beginning at 1-4 yr of age.

188. **The specific clue to the diagnosis of fatty acid oxidation disorders may be the finding of hypoglycemia and**

A. **hypoketonuria**
B. metabolic acidosis
C. elevated liver enzyme
D. elevated blood ammonia
E. prolonged prothrombin time

Ans A Fatty acid oxidation disorders are easily overlooked because the only specific clue to the diagnosis may be the finding of inappropriately low concentrations of urinary ketones in an infant who has hypoglycemia.

Newborn Screening

189. **A 5month old baby presents to emergency with history of vomiting, irritability & jaundice. The baby was previously healthy. History of introducing fruit juices into the diet one week back. USG revealed hepatomegaly and liver function test were abnormal. The infants most likely has deficiency of which of the following enzymes?**

A. Primary biliary cirrhosis
B. **Galactosemia**
C. Paralysis agitans
D. Hepatolenticular degeneration

Ans B Aldolase B The given history of symptoms, on introduction of fruit juices, including hepatic dysfunction, points to a diagnosis of Hereditary fructose intolerance, which is due to deficiency of Aldolase B.

190.Ectopia lentis in a child is seen in which of the following diseases?

A. Sarcoidosis
B. **Homocystinuria**
C. Alkaptonuria
D. Urea cycle defects

Ans B Ectopia lentis is one of the features of Homocystinuria, where the direction of lens subluxation is infero-medial, unlike supero-lateral weiveR subluxation in Marfan syndrome.

191. A child has microcephaly, blue eyes, fair skin, and mental retardation; ferric chloride test is positive. What is the likely diagnosis?

A. **Phenylketonuria (PKU)**
B. Homocystinuria
C. Tyrosinosis
D. Alkaptonuria

Ans A Microcephaly, blue eyes, fair skin and mental retardation along with positive ferric chloride test (which detects phenylalanine in urine) is suggestive of Phenylketonuria caused by deficiency of phenylalanine hydroxylase.

192.A child presents with pellagra like dermatitis and aminoaciduria. Two siblings have the condition and it is absent in two siblings while it is absent in both parents. Which of the following condition is most likely?

A. **Homocystinuria**
B. Isovaleric acidemia
C. Phenylketonuria
D. Alkaptonuria

Ans A Hartnup's disease

193.Guthrie test can be used for the diagnosis of:

A. Tyrosinemia
B. Galactosemia
C. Alkaptonuria
D. **Phenylketonuria**

Ans D The bacterial inhibition assay of Guthrie is used for Phenylketonuria.

194.A male child with coarse facies, macroglossia, thick lips with hepatosplenomegaly presents with copious mucus discharge from nose. Probable underlying diagnosis is:

A. **Hurler disease**
B. Beckwith-Wiedemann syndrome
C. Proteus syndrome
D. Hypothyroidism

Ans A Hurler disease (deficiency of L-iduronidase)
• Diagnosis is usually made between 6–24 months of age with hepatosplenomegaly, coarse facial features, corneal clouding, large tongue, prominent forehead, joint stiffness, short stature and skeletal dysplasia
• Most patients have recurrent upper respiratory tract infections, noisy breathing and copious nasal discharge
• Valvular heart disease, obstructive airway disease, respiratory infection, and cardiac complications are common.

195.For which of the following diseases enzyme replacement therapy is available?

A. Albinism
B. Niemann-Pick disease
C. Metachromatic leukodystrophy
D. **Gaucher's disease LEUKODYSTROPHY**

Ans D

196.Macrocephaly is seen in which of the following syndromes?

A. Metachromatic leukodystrophy
B. Adrenoleukodystrophy
C. **Canavan's disease**
D. Krabbe's disease

Ans C In Canavan disease, there is Hypotonia, head lag & macrocephaly.

197. A 12yearold girl with the mood and emotional liability has a golden brown discoloration in descemet membrane. Most likely diagnosis is:

A. Fabry's disease
B. **Wilson's disease**
C. Glycogen storage disease
D. Acute rheumatic fever OTHER METABOLIC DISORDERS Questions

Ans B KF ring in Wilson disease is due to deposition of Copper in the Descemet membrane layer of cornea.

198.Inborn errors of metabolism (IEM) are hereditary biochemical disorders caused by single-gene mutations that encode specific proteins. The following are the common characteristics of IEM EXCEPT

A. the majority of conditions are inherited as autosomal recessive
B. **the later the appearance of clinical symptoms, the more severe is the disease**
C. early diagnosis is of paramount importance before irreversible organ damage
D. the affected infant is normal at birth and becomes symptomatic later on in life
E. there is a variation in the severity of the phenotype based on the gene mutation

Ans B The earlier the appearance of clinical symptoms, the more severe is the disease.

199. **A 10-day-old boy presented with lethargy, poor feeding, repeated vomiting, and recurrent hypoglycemic fits not responding to IV glucose. On examination, he was hypertonic with severe opisthotonos alternating with flaccidity. Of the following, the MOST likely diagnosis is**

A. tyrocinemia
B. galactosemia
C. phenyletonuria
D. homocystinurea
E. **maple syrup urine disease**

Ans E In maple syrup urine disease, affected infants who are normal at birth develop poor feeding and vomiting in the 1st wk of life; lethargy and coma may ensue within a few days. Physical examination reveals hypertonicity and muscular rigidity with severe opisthotonos. Periods of hypertonicity may alternate with bouts of flaccidity manifested as repetitive movements of the extremities (boxing and bicycling). Cerebral edema may be present; convulsions occur in most infants, and hypoglycemia is common.

200. **All the following are recognized feature of Niemann-pick disease type B EXCEPT**

A. normal IQ
B. progressive lung disease
C. **prolonged neonatal jaundice**
D. subtle or no neurologic involvement
E. cherry-red maculae in some patients

Ans C Niemann-pick disease type B patients do not have neurologic involvement and have a normal IQ. Some patients with type B disease have cherry-red maculae or haloes and subtle neurologic symptoms (peripheral neuropathy). In some type B patients, decreased pulmonary diffusion caused by alveolar infiltration becomes evident in late childhood or early adulthood and progresses with age. Prolonged neonatal jaundice occurs in type C.

201. **A 9-month-old boy with exclusive breast feeding presented with jaundice, vomiting, lethargy, hepatomegaly, and hypoglycemic fits when sweetened cereal added. Laboratory findings showed prolonged clotting time, hypoalbuminemia, elevation of bilirubin and transaminase levels, and proximal tubular dysfunction. Of the following, the MOST likely diagnosis is**

A. tyrosinemia
B. galactosemia
C. organic acidemias
D. maple syrup urine disease
E. **hereditary fructose intolerance**

Ans E Patients with hereditary fructose intolerance HFI are asymptomatic until fructose or sucrose (table sugar) is ingested (usually from fruit, fruit juice, or sweetened cereal). Symptoms may occur early in life, soon after birth if foods or formulas containing these sugars are introduced into the diet.

202. **Acute intermittent porphyria (AIP) could be exacerbated by all the following**

A. surgery
B. puberty
C. **pregnancy**
D. premenstrual period
E. decreased carbohydrate intake

Ans C Surprisingly, pregnancy is usually well tolerated, suggesting that beneficial metabolic changes may ameliorate the effects of high levels of progesterone.

203. **The following are counter regulatory hormones which are increased as blood glucose falls EXCEPT**

A. cortisol
B. glucagon
C. epinephrine
D. **thyroid hormone**
E. growth hormone

Ans D is not a counter regulatory hormone in response to hypoglycemia.

204. **All the following are clinical abnormalities of Zellweger syndrome EXCEPT**

A. cataract
B. micrognathia
C. high forehead
D. **small fontanells**
E. redundant skin fold of neck

Ans D Craniofacial dysmorphisms are; high forehead, flat occiput, large fontanelle(s), wide sutures, shallow orbital ridges, low/broad nasal bridge, epicanthus, high arched palate, external ear deformity, and micrognathia.

07

CHAPTER 07

Diseases of Immune System



WHAT THIS CHAPTER COVERS

- Immunoglobulins & Complement
- Primary Immunodeficiencies
- SCID & Combined Immunodeficiency
- Agammaglobulinemia & CVID
- Phagocyte Disorders
- Complement Deficiencies
- Wiskott-Aldrich & Ataxia Telangiectasia
- DiGeorge Syndrome
- Autoimmune Disorders
- Vaccines & Immune Response
- Allergies & Anaphylaxis

215 MCQs **11** subtopics

7.1 Immunoglobulins & Complement – Developmental Facts

Ig	Key facts in children
IgG	**Only Ig that crosses placenta** (active transport via FcRn, mostly in 3rd trimester) → maternal IgG protects for 3–6 m; **physiological nadir at 3–6 months** (transient hypogammaglobulinaemia of infancy); adult levels by 5–7 y; **IgG2 subclass matures last** (poor response to polysaccharide antigens < 2 y)
IgM	**First Ig produced by fetus (by 20 wk)**; **cord IgM > 20 mg/dL → intrauterine infection**; largest (pentamer), best complement activator; adult levels by 1 y
IgA	Secretory IgA in colostrum; serum IgA adult levels by **adolescence** (last); **selective IgA deficiency = MC primary immunodeficiency**
IgE	Allergy, parasites; ↑ in hyper-IgE, Wiskott–Aldrich
IgD	B-cell surface receptor

- **Complement**: classical pathway (C1q–C4–C2, antibody-triggered), alternative (C3, factor B, D, properdin), lectin (MBL). **C3 is central**; MAC = C5b–C9.
- **CH50** – screens classical pathway; **AH50** – alternative pathway.
- **Neonatal immune deficits**: low complement (esp. alternative), poor neutrophil chemotaxis & storage pool, poor opsonisation, Th2 bias → susceptible to **GBS, E. coli, Listeria, HSV**.

7.2 Primary Immunodeficiency (PID) – Approach

10 WARNING SIGNS OF PID (JEFFREY MODELL)

≥ 4 ear infections/year · ≥ 2 serious sinus infections/year · ≥ 2 months of antibiotics with little effect · ≥ 2 pneumonias/year · **failure to thrive** · recurrent deep skin/organ abscesses · persistent oral thrush/fungal infection > 1 y · need for IV antibiotics · ≥ 2 deep-seated infections (sepsis, meningitis) · **family history of PID**.

Defect	Age of onset	Typical organisms	Screening test
B-cell / antibody	**After 6 months** (maternal IgG wanes)	**Encapsulated bacteria** (pneumococcus, Hib), **enteroviruses**, **Giardia**	Serum IgG, IgA, IgM; vaccine antibody titres; CD19/CD20 B-cell count
T-cell / combined	**Early infancy (< 3–6 m)**	**Viruses, fungi (Candida, PCP)**, mycobacteria (BCG-osis), opportunists; **FTT, chronic diarrhoea**	**Absolute lymphocyte count** (low), CD3/CD4/CD8 flow cytometry, **TREC** (newborn screen), mitogen proliferation; **delayed hypersensitivity skin test (anergy)**
Phagocyte	Early	**Catalase-positive bacteria** (S. aureus, Serratia, Burkholderia, Nocardia), **Aspergillus**; skin & deep abscesses, poor wound healing, **delayed cord separation**	Neutrophil count & morphology, **DHR flow cytometry / NBT test**, CD18
Complement	Any	**Neisseria** (terminal C5–C9), pyogenic (C3), **SLE-like (C1q, C2, C4 – early components)**	**CH50, AH50**



Where the defect lies – classic primary immunodeficiencies

7.3 Antibody (B-cell) Deficiencies

Disorder	Defect	Features
X-linked (Bruton) agammaglobulinaemia	**BTK (Bruton tyrosine kinase)** – pre-B cells fail to mature	**Boys > 6 months**, recurrent sinopulmonary infections (pneumococcus, Hib), **enteroviral meningoencephalitis**, **vaccine-associated polio (OPV)**, arthritis (Mycoplasma); **absent tonsils & lymph nodes**; **all Ig classes very low; B cells (CD19) < 2%**; T cells normal. Rx **IVIG lifelong (400–600 mg/kg every 3–4 wk)**; **no live vaccines**
Common variable immunodeficiency (CVID)	Heterogeneous (TACI, ICOS…)	Onset **> 2 y – adolescence/adult**; low IgG + IgA ± IgM, **normal B-cell numbers**, poor vaccine responses; **autoimmunity (ITP, AIHA), granulomas, lymphoma, gastric cancer**, nodular lymphoid hyperplasia; Rx IVIG
Selective IgA deficiency		**MC PID** (1 : 500); mostly asymptomatic; sinopulmonary infections, **giardiasis, celiac disease (false-negative IgA-tTG → test IgG-based)**, autoimmunity; **anaphylaxis to blood products containing IgA (anti-IgA antibodies)** – use washed RBCs/IgA-deficient products
Transient hypogammaglobulinaemia of infancy	Delayed maturation	Low IgG after 6 m, **normal vaccine responses, normal B cells**, recovers by 2–4 y
Hyper-IgM syndrome (X-linked – CD40 ligand)	Defective class switching (CD40L on T cells)	**↑/normal IgM, ↓ IgG, IgA, IgE**; **Pneumocystis pneumonia**, Cryptosporidium (sclerosing cholangitis), neutropenia; it is a **combined** immunodeficiency; Rx IVIG, HSCT
IgG subclass deficiency	IgG2 most significant	Polysaccharide-antigen infections

7.4 Combined Immunodeficiencies – SCID

- **Most severe PID**; presents **in first months** with **FTT, chronic diarrhoea, persistent oral thrush, PCP pneumonia, severe viral infections (CMV, RSV), disseminated BCG**, absent thymic shadow, **lymphopenia (ALC < 2500/µL in infants – key clue)**, GvHD from maternal T cells or **non-irradiated blood transfusions**.
- **Medical emergency – fatal in first 1–2 years without HSCT**.

SCID type	Defect	Phenotype
X-linked SCID (MC, ~45%)	**IL2RG – common γ chain** (IL-2, 4, 7, 9, 15, 21 receptors)	**T^- B^+ NK^-**
ADA deficiency (MC AR form)	Adenosine deaminase → toxic dATP	**T^- B^- NK^-**; **costochondral cupping/flaring**, neurological & hepatic features; Rx **PEG-ADA, gene therapy**, HSCT
JAK3 deficiency	AR	T^- B^+ NK^-
RAG1/RAG2	V(D)J recombination	**T^- B^- NK^+**; hypomorphic → **Omenn syndrome** (erythroderma, eosinophilia, ↑ IgE, lymphadenopathy, hepatosplenomegaly)
IL-7Rα	AR	T^- B^+ NK^+
Artemis	DNA repair (radiosensitive)	T^- B^- NK^+
Reticular dysgenesis	AK2	Absent lymphocytes & neutrophils, deafness
Bare lymphocyte syndrome	**MHC class II** (CIITA, RFX)	**Low CD4**, hypogammaglobulinaemia
ZAP-70 deficiency	TCR signalling	**Absent CD8**, normal CD4 (non-functional)

MANAGEMENT OF SCID

HSCT (ideally < 3.5 months, before infections) – curative; IVIG; PCP prophylaxis (co-trimoxazole); **irradiated, leucoreduced, CMV-negative blood products only**; **no live vaccines (BCG, OPV, rotavirus, MMR, varicella)**; gene therapy (ADA, X-SCID). Newborn screening by **TREC assay** (T-cell receptor excision circles).

7.5 Syndromic Immunodeficiencies

Disorder	Genetics	Classic triad / features
Wiskott–Aldrich syndrome	**XLR, WAS gene (WASP)**	**Eczema + thrombocytopenia with SMALL platelets + recurrent infections** (encapsulated organisms); bloody diarrhoea/bleeding in infancy; **↓ IgM, ↑ IgA & IgE, normal IgG**; ↑ autoimmunity & **lymphoma (EBV)**; poor polysaccharide response. Rx **HSCT** (curative), gene therapy; splenectomy ↑ sepsis risk
Ataxia telangiectasia	**AR, ATM (chr 11q22) – DNA repair**	**Progressive cerebellar ataxia (from walking age, ~1–2 y; first)**, **oculocutaneous telangiectasia (3–6 y, bulbar conjunctiva)**, sinopulmonary infections, **↑ AFP (diagnostic marker)**, **↓ IgA, IgG2, IgE**, **radiosensitivity**, ↑ **lymphoma/leukaemia**, oculomotor apraxia, insulin-resistant DM; avoid X-rays
DiGeorge (22q11.2 del)	TBX1; 3rd & 4th pharyngeal pouches	**Hypocalcaemic tetany (neonatal), conotruncal heart defects (interrupted aortic arch type B, truncus arteriosus, TOF)**, thymic hypoplasia → ↓ T cells (variable; complete DiGeorge = SCID-like), **cleft palate, characteristic facies** (hypertelorism, low-set ears, short philtrum), learning disability, psychiatric illness; absent thymic shadow; Dx **FISH/CMA**; Rx Ca + vitamin D, **thymus transplant** (complete)
Hyper-IgE (Job) syndrome – AD (STAT3)	STAT3 → ↓ Th17	**FATED**: coarse **F**acies, **A**bscesses ("**cold**" staphylococcal abscesses, no inflammation), retained primary **T**eeth, ↑↑ **IgE (> 2000 IU/mL)**, **D**ermatologic (eczema); **pneumatoceles**, eosinophilia, fractures with minor trauma, scoliosis; **AR form (DOCK8)** – severe viral skin infections, allergy
Chronic mucocutaneous candidiasis	AIRE (APECED), STAT1 GOF, IL-17 defects	Candida of skin/nails/mucosa; **APECED: hypoparathyroidism, Addison's, candidiasis**
X-linked lymphoproliferative disease (Duncan)	SH2D1A (SAP)	**Fulminant/fatal EBV infection (infectious mononucleosis)**, HLH, lymphoma
Chediak–Higashi	AR, LYST (lysosomal trafficking)	**Partial oculocutaneous albinism, silvery hair**, recurrent pyogenic infections, **giant cytoplasmic granules in neutrophils**, peripheral neuropathy, bleeding; **accelerated phase = HLH**; Rx HSCT
IPEX	FOXP3 (XLR) – Treg defect	**I**mmune dysregulation, **P**olyendocrinopathy (**neonatal type 1 DM**), **E**nteropathy, **X**-linked; eczema
Griscelli	RAB27A	Partial albinism, HLH, no giant granules

7.6 Phagocyte Disorders

Disorder	Defect	Features & diagnosis
Chronic granulomatous disease (CGD)	**NADPH oxidase** (gp91phox **CYBB – X-linked, 65–70%**; others AR) → no respiratory burst	Recurrent infections with **catalase-positive organisms – S. aureus, Serratia, Burkholderia cepacia, Nocardia, Aspergillus (MCC death)**; **liver abscess**, suppurative lymphadenitis, osteomyelitis, perianal abscess, **granulomas (GI/GU obstruction)**, colitis. **Dx: DHR (dihydrorhodamine) flow cytometry – test of choice**; **NBT test – no blue colour (negative)**. Rx **co-trimoxazole + itraconazole prophylaxis, interferon-γ**, HSCT

Disorder	Defect	Features & diagnosis
Leucocyte adhesion deficiency type 1 (LAD-1)	**CD18 (β2 integrin)** – AR	**Delayed umbilical cord separation (> 3–4 weeks), omphalitis**, recurrent **necrotic skin infections without pus**, severe gingivitis/periodontitis, poor wound healing; **marked neutrophilia (even without infection)**; Dx **flow cytometry for CD18/CD11**; Rx HSCT
LAD-2	Fucose transporter (sialyl-Lewis X)	Bombay blood group, ID, short stature
Myeloperoxidase deficiency	MPO	**MC phagocyte defect**, mostly asymptomatic; Candida infections in diabetics; **NBT normal**
Severe congenital neutropenia (Kostmann)	ELANE (AD), HAX1 (AR)	ANC < 200, **maturation arrest at promyelocyte stage**, omphalitis, abscesses; risk of MDS/AML; Rx **G-CSF**
Cyclic neutropenia	ELANE	Neutropenia every **21 days** lasting 3–6 days – fever, **oral ulcers**, gingivitis
Shwachman–Diamond syndrome	SBDS	**Neutropenia + exocrine pancreatic insufficiency + metaphyseal dysostosis**, short stature, MDS/AML risk

NBT TEST INTERPRETATION

Normal neutrophils **reduce yellow NBT to blue-black formazan**. In **CGD → no colour change** (stays yellow = abnormal). DHR test: **no fluorescence shift in CGD** (X-linked carriers show **two populations – mosaic pattern**).

7.7 Complement Deficiencies

Deficiency	Clinical
C1q, C1r, C1s, C2, C4 (early classical)	**SLE-like disease**, glomerulonephritis; **C2 deficiency = MC complement deficiency**
C3	Severe recurrent **pyogenic** infections (pneumococcus), MPGN
C5–C9 (terminal, MAC)	**Recurrent Neisseria (meningococcal, gonococcal) infections** – vaccinate
Properdin (X-linked)	**Fulminant meningococcal disease** (alternative pathway); normal CH50, low AH50
Factor H / I	**Atypical HUS**, MPGN (C3 glomerulopathy)
C1-esterase inhibitor (AD)	**Hereditary angioedema** – recurrent non-pruritic, non-urticarial swelling of face, limbs, **larynx (fatal)**, abdominal pain; triggered by trauma/dental work; **low C4 (best screening), low C1-INH level/function, normal C3**; bradykinin-mediated → **no response to antihistamines/steroids/adrenaline**; Rx **C1-INH concentrate, icatibant (bradykinin B2 antagonist), ecallantide, FFP**; prophylaxis – danazol, lanadelumab; **avoid ACE inhibitors & oestrogens**
MBL deficiency	Common; mild infections in infancy

7.8 Autoimmune & Vasculitic Disorders (Overview)

Kawasaki disease (mucocutaneous lymph node syndrome)

- **Most common cause of acquired heart disease in children in developed countries** (RHD in India); age **< 5 y** (peak 1–2 y), boys, Asians.
- **Diagnosis**: **fever ≥ 5 days + ≥ 4 of 5** principal features:
  1. **Bilateral non-exudative bulbar conjunctival injection (limbal sparing)**
  2. **Oral changes** – red cracked lips, **strawberry tongue**, diffuse erythema

3. **Polymorphous rash** (no vesicles)
4. **Extremity changes** – erythema/oedema of palms & soles (acute), **periungual desquamation (2–3 wk)**, **Beau's lines** later
5. **Cervical lymphadenopathy ≥ 1.5 cm, usually unilateral** (least common)

- **Incomplete Kawasaki**: fever ≥ 5 d + 2–3 criteria + supportive labs (**CRP ≥ 3 mg/dL, ESR ≥ 40**, albumin ≤ 3, anaemia, ↑ ALT, **platelets ≥ 4.5 lakh after day 7**, WBC ≥ 15,000, sterile pyuria) or echo changes. Infants < 6 m often incomplete – high coronary risk.
- Other: **BCG scar reactivation (erythema/induration) – specific clue in BCG-vaccinated children**, irritability (aseptic meningitis), arthritis, gallbladder hydrops, uveitis, perineal desquamation.
- Labs: **thrombocytosis in 2nd–3rd week (subacute phase)**, ↑ ESR/CRP, anaemia, hypoalbuminaemia, ↑ NT-proBNP.
- **Coronary artery aneurysms** in **~25% untreated → < 5% with IVIG**; giant aneurysm (Z ≥ 10 or ≥ 8 mm) → thrombosis, MI (MCC death). **2D echo** at diagnosis, 1–2 wk and 4–6 wk.

TREATMENT OF KAWASAKI DISEASE (AHA 2017)

IVIG 2 g/kg single infusion over 10–12 h, ideally within 10 days of fever onset (give even after 10 days if fever/inflammation persists) **+ aspirin** – moderate (30–50 mg/kg/day) or high dose (80–100) until afebrile 48–72 h, then **low-dose 3–5 mg/kg/day for 6–8 weeks** (until echo normal; long-term if aneurysm). **IVIG-resistant** (fever ≥ 36 h after IVIG, ~10–20%): **2nd IVIG dose**, **methylprednisolone/prednisolone**, **infliximab**, ciclosporin. **Live vaccines (MMR, varicella) deferred 11 months after IVIG**. Aspirin + influenza/varicella → **Reye syndrome** risk.

MIS-C (Multisystem inflammatory syndrome in children – post-COVID)

- 2–6 weeks after SARS-CoV-2 infection; **older (6–12 y)**, **prominent GI symptoms**, **shock & myocardial dysfunction (LV dysfunction)**, ↑ troponin/BNP, **lymphopenia, thrombocytopenia** (vs thrombocytosis in KD); Rx IVIG + steroids, aspirin, anticoagulation.

IgA vasculitis (Henoch–Schönlein purpura) – MC vasculitis of childhood

- Age 3–10 y, often after **URTI (group A strep)**; **IgA-immune complex** small-vessel vasculitis.
- **Tetrad**: **palpable purpura** (lower limbs & buttocks – dependent areas, **normal platelet count**), **arthritis/arthralgia** (knees/ankles, non-deforming), **abdominal pain** (colicky; **intussusception – ileo-ileal**, GI bleed), **renal involvement** (haematuria ± proteinuria, 20–55%; **long-term morbidity**).
- Labs: **normal platelets & coagulation**, ↑ serum IgA (50%), skin biopsy – **leucocytoclastic vasculitis with IgA deposits**; renal – mesangial IgA deposition.
- Rx: supportive (NSAIDs for arthritis if renal function normal); **steroids (prednisolone 1–2 mg/kg)** for severe abdominal pain/GI haemorrhage, orchitis; nephritis/nephrotic-range proteinuria → steroids ± immunosuppression; **urinalysis & BP monitoring for 6–12 months**.

Feature	Kawasaki	HSP
Vessel	**Medium-sized** (coronaries)	**Small** vessels
Age	< 5 y	3–10 y
Rash	Polymorphous, non-purpuric	**Palpable purpura**
Major complication	**Coronary aneurysm**	**Nephritis**, intussusception
Rx	**IVIG + aspirin**	Supportive / steroids

- **Takayasu arteritis** (large vessel): young females, **pulseless disease**, absent/unequal pulses, BP difference, renovascular hypertension, claudication; India – **MC cause of renovascular hypertension in children** (with fibromuscular dysplasia).
- **Polyarteritis nodosa** (medium): **Hep B association**, **microaneurysms**, renal artery involvement, **spares lungs**, cutaneous PAN after strep.
- **SLE in children** (cSLE): more severe renal & haematological disease; **ANA (sensitive), anti-dsDNA & anti-Smith (specific)**; neonatal lupus – **congenital complete heart block (anti-Ro/SSA)**, annular rash.

- **Juvenile dermatomyositis**: **heliotrope rash, Gottron papules**, proximal weakness, **calcinosis cutis** (more in children), vasculopathy, ↑ CK, MRI muscle oedema; Rx steroids + methotrexate.

7.9 Vaccines in Immunodeficiency

Condition	Live vaccines
SCID, complete DiGeorge, combined defects	**All live vaccines contraindicated** (BCG, OPV, MMR, varicella, rotavirus, yellow fever, LAIV)
XLA, CVID	**OPV contraindicated** (VAPP), avoid other live vaccines (ineffective on IVIG); give IPV
CGD	**No live bacterial vaccines (BCG, Ty21a)**; live viral vaccines OK
Complement deficiency	All vaccines safe; **pneumococcal, meningococcal, Hib** essential
Selective IgA deficiency	All except OPV (relative)
Asymptomatic HIV (CD4 ≥ 15%)	**MMR & varicella allowed**; **BCG contraindicated in symptomatic HIV** (in India give BCG at birth to all HIV-exposed)
Steroids ≥ **2 mg/kg/day or ≥ 20 mg/day for ≥ 14 days**	**Defer live vaccines for 1 month** after stopping
After IVIG / blood products	Defer measles/varicella vaccines 3–11 months (dose dependent)

7.10 Allergy & Anaphylaxis

- **Atopic march**: atopic dermatitis (infancy) → food allergy → allergic rhinitis → **asthma**.
- **Cow's milk protein allergy (CMPA)**: MC food allergy in infants; IgE-mediated (urticaria, vomiting, anaphylaxis) or non-IgE (proctocolitis – **blood-streaked stools in breastfed infant**, FPIES – repetitive vomiting & lethargy 1–4 h after ingestion, enteropathy); Rx **extensively hydrolysed formula** (amino-acid formula if severe), mother's dairy elimination if breastfed; most outgrow by 3–5 y.
- Common food allergens: milk, egg, **peanut**, tree nuts, soy, wheat, fish, shellfish; peanut/tree-nut/fish/shellfish allergies usually **persist**.
- **Early introduction of peanut (4–6 m) prevents peanut allergy** (LEAP trial) in high-risk infants.
- Tests: **skin prick test**, specific IgE (ImmunoCAP), **oral food challenge (gold standard)**.

ANAPHYLAXIS

IM adrenaline 1:1000 (1 mg/mL) 0.01 mg/kg (0.01 mL/kg), max 0.5 mg (0.3 mg in prepubertal), into anterolateral thigh; repeat every **5–15 min**. Supine with legs raised, oxygen, **IV fluid bolus 20 mL/kg** NS for hypotension; adjuncts – **H1 antihistamine, nebulised salbutamol** (bronchospasm), **steroids (prevent biphasic reaction)**. Observe ≥ 4–6 h (biphasic in up to 20%). Auto-injector: 0.15 mg (15–30 kg), 0.3 mg (> 30 kg). Tryptase ↑ within 1–3 h confirms mast-cell activation.

- **Urticaria**: acute (< 6 weeks) – viral infections (MCC in children), foods, drugs; chronic (> 6 wk) – idiopathic/autoimmune; Rx **2nd-generation non-sedating antihistamines** (cetirizine, levocetirizine, fexofenadine, loratadine – up to 4× dose).
- **Serum sickness** (type III) – 7–14 days after drug (cefaclor, penicillin) or antiserum: fever, urticaria, arthralgia, lymphadenopathy.

Practice MCQs · Diseases of Immune System

215 questions · correct option highlighted in green · cover the answer strip to self-test

Immunoglobulins & Complement

1. What does 'A' depict in the given image of serum immunoglobulin levels in a pediatric patient?

A. IgM
B. IgG
C. Maternal and Fetal IgM
D. **Maternal and Fetal IgG**

Ans D • A small amount of IgM is present from 20th week onwards
• Rapid synthesis of IgG starts within 1st few weeks of postnatal life
• Final adult values of all subclasses of IgG is usually reached between 8-10 years age
• Last class of Ig to reach adult serum levels is IgA

2. Children with Chediak Higashi syndrome have light skin, silvery hair, solar sensitivity, photophobia, frequent infections, neuropathy, prolonged bleeding times with normal platelet count. What is the treatment which improves the clinical status of some children in the stable phase?

A. Vitamin B12
B. Vitamin B1
C. **Vitamin C**
D. Vitamin E

Ans C High-dose ascorbic acid or Vitamin C (200 mg/day for infants, 2,000 mg/day for adults) may improve clinical status of some children in stable phase of Chediak Higashi disease

3. Chediak Higashi syndrome is characterized by the following except:

A. **Neutrophilia**
B. Defective degranulation
C. Delayed microbial killing
D. Giant granules

Ans A Main leukocyte abnormalities in Chediak-Higashi syndrome are:
• Neutropenia (most common); Leukocytes contain giant granules,Q (characteristic) due to aberrant phagolysosome fusionQ
• Defective degranulation, Impaired chemotaxis, Delayed microbial killing; NK cell function is also impaired

4. Which is found in DiGeorge's syndrome:

A. **Tetany**
B. Eczema
C. Mucocutaneous candidiasis
D. Absent B and T cells
E. Total absence of T cells HENOCH-SCHÖNLEIN PURPURA

Ans A c. Mucocutaneous candidiasis, e. Total absence of T cells. Disease Primary defect Secondary defect DiGeorge Syndrome Familial cases: • T-cell defect (Thymic Hypoplasia)Q failure of development of 3rd & 4th pharyn- • TetanyQ geal pouchesQ • Congenital defects of heart and great vessels Non-familial cases: 22q11 deletion • Mucocutaneous Candidiasis with HENOCH-SCHÖNLEIN PURPURA

5. Henoch-Schönlein purpura is characterized by the deposition of the following immunoglobulin around the vessels:

A. IgM
B. IgG (AIIMS Nov 05)
C. **IgA**
D. IgE KAWASAKI DISEASE

Ans C Biopsy finding in HSP is deposition of IgA, specifically IgA1 in the affected tissue, by immunofluorescence, accompanied to a lesser extent by deposition of C3, fibrin & IgM.

6. All are features of DiGerorge syndrome except:

A. Recurrent infection
B. **Thymic hyperplasia**
C. Impaired T cell function
D. Hypocalcemic seizures

Ans B DiGeorge syndrome results from thymic HYPOPLASIA/ aplasia (not hyperplasia), leading to impaired T-cell function, recurrent infections, and hypocalcemic seizures from associated parathyroid hypoplasia.

7. A very high serum concentrations of one or more immunoglobulin classes, suggest all the following EXCEPT

A. HIV infection
B. chronic inflammation
C. **intestinal lymphangiectasia**
D. chronic granulomatous disease
E. autoimmune lymphoproliferative syndrome

Ans C There is a possible loss of immunoglobulins through the urinary or gastrointestinal tracts (nephrotic syndrome, protein-losing enteropathies, intestinal lymphangiectasia).

8. A 3-year-old girl presented with recurrent attacks of chest infection and frequent episodes of chronic diarrhea, stool examination revealed Giardia lamblia and serum IgA level was very low. Regarding this condition, all the following are true EXCEPT

A. autosomal dominant inheritance
B. phenotypically normal blood B cells
C. administration of IVIG is not indicated
D. environmental factors may trigger the disease
E. **it is associated with a celiac-like syndrome with dramatic response to gluten- free diet**

Ans E An isolated absence or near absence (<10 mg/dL) of serum and secretory IgA is the most common well-defined immunodeficiency disorder. IgA deficiency is associated with a celiac-like syndrome, which may or may not respond to a gluten-free diet.

9. Of the following, the findings that are MOST suggestive of DiGeorge syndrome are

A. cataract and a congenital heart defect
B. hypotonia and a congenital heart defect
C. cutis aplasia and a congenital heart defect
D. **hypocalcemia and a congenital heart defect**
E. mongoloid slant to the eyes and a congenital heart defect

Ans D Digeroge syndrome; associated with congenital heart disease (conotruncal, atrial, and ventricular septal defects). The diagnosis is often first suggested by hypocalcemic seizures during the neonatal period.

10. The MOST appropriate method to correct the immune deficiency in complete DiGeorge syndrome is

A. vaccination
B. administration of IVIG
C. judicious use of antibiotics
D. **transplantation of thymic tissue**
E. transplantation of hematopoietic stem cells

Ans D

11. In patient with Chédiak-Higashi syndrome, the only curative therapy is hematopoietic stem cell transplantation which correct all the following EXCEPT

A. **neuropathy**
B. immunologic function
C. hematopoietic function
D. natural killer cell deficiency
E. conversion to the accelerated phase

Ans A

12. All the following matching about genetic deficiencies of plasma complement components and associated clinical findings are true EXCEPT

A. **C4 ---------- discoid lupus erythematosus**
B. C1q---------- systemic lupus erythematosus
C. C1 INH ---------- hereditary angioedema
D. factor H---- atypical hemolytic-uremic syndrome
E. properdin ---------- N. meningitidis meningitis

Ans A Systemic lupus erythematosus (SLE) is very common in C4 deficiency.

13. The complement component of both the classical and alternative pathways is

A. C2
B. **C3**
C. C4
D. C5
E. C6

Ans B

14. The best method for detection of allergen-specific IgE is

A. **skin tests**
B. serum immune assay for specific IgE
C. nasal smear study
D. eosinophilia in blood film
E. total serum IgE concentration

Ans A Epicutaneous skin tests provide the best method for detection of allergen- specific IgE (positive predictive value of 48.7% for the epidemiologic diagnosis of AR). They are inexpensive and sensitive, and the risks and discomfort are minimal.

15. All the following are triggers for pruritus in atopic dermatitis EXCEPT

A. grass
B. tree nuts
C. **high humidity**
D. herpes simplex
E. excessive sweating

Ans C Foods (cow milk, egg, peanut, tree nuts, soy, wheat, and shellfish), aeroallergens (pollen, grass, animal dander, and dust mites), infection (staphylococcus, herpes simplex, and molluscum), reduced humidity, excessive sweating, and irritants (wool, acrylic, soaps, toiletries, fragrances, and detergents) can exacerbate (trigger) pruritus and scratching.

16. IgE anti-bodies against Malassezia furfur have been found in patients with atopic dermatitis of

A. **head and neck**
B. hands and arms
C. legs and feet
D. upper chest
E. lower abdomen

Ans A There has been particular interest in the role of Malassezia furfur in atopic dermatitis because it is lipophilic yeast commonly present in the seborrheic areas of the skin.

17. Systemic allergic responses to insects are attributed to IgE antibody response caused primarily by

A. ticks
B. spiders
C. **hymenoptera**
D. scorpions
E. triatoma (kissing bug)

Ans C

18. The MOST reliable diagnostic modality for detection of venom-specific IgE is

A. in vitro serum assay for venom-specific IgE
B. **skin test**
C. serum tryptase level
D. plasma histamine
E. radioallergosorbent assay

Ans B

19. Non IgE mediated urticaria can be caused by

A. milk
B. hymenoptera
C. **Epstein-barr virus**
D. latex
E. blood

Ans C Acute urticaria can also result from non–IgE-mediated stimulation of mast cells, caused by radiocontrast agents, viral agents (including hepatitis B and Epstein-Barr virus), opiates, and nonsteroidal anti-inflammatory agents.

20. In cases of food-associated exercise-induced anaphylaxis, the parents are advised that children must not take exercise after ingestion of the triggering food for

A. **2-3 hours**
B. 4-6 hours
C. 8-12 hours
D. 13-18 hours
E. 19-24 hours

Ans A

21. Which of the following results of investigations is unlikely in serum sickness?

A. elevated erythrocyte sedimentation rate
B. **thrombocytosis**
C. reduced C3 activity
D. negative microbial cultures
E. microscopic hematuria

Ans B Thrombocytopenia is often present.

22. All the following are symptoms of acute IgE mediated food allergy EXCEPT

A. urticaria
B. **pruritis**
C. abdominal pain
D. diarrhea
E. wheezing

Ans B

23. One of the following presentations is a non-IgE mediated food allergy

A. **Heiner syndrome**
B. gastrointestinal anaphylaxis
C. rhino conjunctivitis
D. oral allergy syndrome
E. angioedema

Ans A (food induced pulmonary hemosiderosis) is a non-IgE mediated food allergy.

24. Wheezing occurs in approximately which percent of IgE-mediated food allergic reactions?

A. 15%
B. **25%**
C. 35%
D. 45%
E. 55%

Ans B

Primary Immunodeficiencies

25. Combined T & B cell immunodeficiency includes:

A. DiGeorge syndrome
B. Chronic granulomatous disease
C. **Adenosine deaminase deficiency**
D. Wiskott-Aldrich syndrome
E. Ataxia telangiectasia

Ans C d. Wiskott-Aldrich syndrome & e. Ataxia telangiectasia

26. The commonest primary immunodeficiency is:

A. Common variable immunodeficiency
B. Isolated IgA immunodeficiency
C. Wiskott-Aldrich syndrome
D. **AIDS**

Ans D Nitroblue tetrazolium test is useful for screening

27. All the following are indicated in the long-term therapy of children with coronary abnormalities in Kawasaki disease except:

A. **Intravenous immunoglobulins**
B. Aspirin
C. Clopidogrel
D. Warfarin

Ans A Treatment of Kawasaki Disease Acute stage • Intravenous immunoglobulin 2 g/kg over 10-12 hr and
• Aspirin 80-100 mg/kg/day divided every 6 hr orally until patient is afebrile for at least 48 hr Convalescent stage • Aspirin 3-5 mg/kg once daily until 6-8 wk after illness onset if normal coronary findings throughout Long-term therapy for • Aspirin 3-5 mg/kg once daily orally or Clopidogrel 1 mg/kg/day (maximum: 75 mg/day)

28. Silk Road disease is:

A. **Behcet's syndrome**
B. Giant cell arteritis
C. Henoch-Schonlein purpura
D. Wegener's granulomatosis

Ans A • Behçet disease is characterized by recurrent oral ulcerations, uveitis & skin abnormalities
• It has a high prevalence in countries along the Silk Road, extending from Japan to the eastern Mediterranean.

29. Lung is characteristically involved in:

A. **Churg-Strauss syndrome**
B. HSP
C. PAN
D. ITP

Ans A Churg Strauss syndrome: It is a small vessel necrotizing granulomatous vasculitis associated with a history peripheral eosinophilia.

30. A 9-month-old boy presented with recurrent attacks of diarrhea, thrush, and failure to thrive shortly after birth. You suspect immune deficiency. Of the following, the MOST common cause of this condition is

A. hyper-IgE syndrome
B. defect in phagocytic cells
C. **deficiencies in T-cell function**
D. defect in antibody production
E. defect in complement proteins

Ans C Children with defects in antibody production, phagocytic cells, or complement proteins have recurrent infections with encapsulated bacteria and may grow and develop normally despite their recurring infections, unless they develop bronchiectasis from repeated lower respiratory tract bacterial infections or persistent enteroviral infections of the central nervous system.

31. Leukemoid reactions (WBC count > 50,000/µL) are characterized by abundant

A. blasts
B. **band form**
C. myelocytes
D. promyelocytes
E. metamyelocytes

Ans B Leukemoid reactions are usually neutrophilic, and unlike true leukemia, show only small proportions of immature myeloid cells, consisting primarily of band forms, occasional metamyelocytes, and progressively rarer myelocytes, promyelocytes, and blasts.

32. The diagnosis of allergic rhinitis is established by the time the child reaches age of

A. 2 yr
B. 4 yr
C. **6 yr**
D. 8 yr
E. 10 yr

Ans C The symptoms may appear in infancy; with the diagnosis generally established by the time the child reaches age 6 yr. The prevalence peaks late in childhood.

33. Allergic rhinitis may be complicated by secondary bacterial infection as sinusitis. Of the following, the MOST likely clue to infection is

A. hyperemia
B. swollen turbinates
C. **thick nasal secretions**
D. bluish mucus membranes
E. edematous mucus membranes

Ans C Thick, purulent nasal secretions indicate the presence of infection.

34. Approximately 80% of all asthmatic patients report disease onset prior to the age of

A. 2 yr
B. 4 yr
C. **6 yr**
D. 8 yr
E. 10 yr

Ans C

35. A patient with exacerbation of asthma being handled and managed in the ER may be discharged home if all the following parameters are met EXCEPT

A. normal physical activity
B. PEF >70% of predicted or personal best
C. **exercise tolerance**
D. oxygen saturation >92% while the patient is breathing room air for 4 hr
E. no more use of accessory muscles

Ans C

36. All the following are a recognized features of a ''well-controlled'' asthma in a 7- year-old boy EXCEPT

A. FEV1:FVC ratio >80%
B. FEV1of >80% of predicted
C. daytime symptoms ≤2 days/wk
D. **≥4 exacerbations in the past year**
E. need a rescue bronchodilator ≤2 days/wk

Ans D Children with well-controlled asthma have daytime symptoms ≤2 days/wk and need a rescue bronchodilator ≤2 days/wk; an FEV1of >80% of predicted (and an FEV1: FVC ratio >80% for children 5-11 yr of age); no interference with normal activity; and <2 exacerbations in the past year.

37. In infancy, atopic dermatitis is usually acute and spares the

A. extensor surfaces of the extremities
B. forehead
C. scalp
D. **diaper area**
E. cheeks

Ans D

38. The best choice of antibiotic in treating localized impetiginous lesions in patients with atopic dermatitis is

A. oral erythromycin
B. oral cephalexin
C. **topical mupirocin**
D. topical ampicillin
E. topical gentamycin

Ans C Erythromycin and azithromycin are usually beneficial for patients who are not colonized with a resistant S. aureus strain; a first-generation cephalosporin (cephalexin) is recommended for macrolide-resistant S. aureus. Topical mupirocin is useful in the treatment of localized impetiginous lesions, with systemic antibiotics for widespread infections.

39. Reactions to stinging and biting insects may cause

A. **a limited lesion confined to the primary site**
B. a pronounced localized reaction
C. a pronounced systemic reaction due to immediate hypersensitivity
D. a pronounced systemic reaction due to delayed hypersensitivity
E. no reaction

Ans A

40. **The eye is a common target of allergic disorders because of its marked vascularity and direct contact with allergens in the environment. Of the following, the MOST immunologically active tissue of the external eye is**

A. iris
B. sclera
C. eyelids
D. eyelashes
E. **conjunctiva**

Ans E

41. **Acute urticaria is a self-limited illness requiring little treatment. All the following can be used EXCEPT**

A. loratadine
B. hydroxyzine
C. epinephrine IM
D. **H2 antihistamines**
E. oral corticosteroid

Ans D used in chronic urticaria.

42. **Food protein–induced enterocolitis syndrome is characterized by all the following**

A. manifests in the first months of life
B. vomiting occurs 1-3 hours after feeding
C. hypotension occurs in approximately 15% of cases
D. **commonly improved with ingestion of soy protein based formula**
E. usually resolve by the age of three years of life

Ans D Commonly provoked by ingestion of milk and soy based protein formula.

43. **One of the following drug-induced allergic reactions is not included as an indictment of allopurinol**

A. exanthem
B. Stevens-Johnson syndrome
C. toxic epidermal necrolysis
D. **pulmonary fibrosis**
E. interstitial nephritis

Ans D Nitrofurantoin, bleomycin and methotrexate are implicated in causing pulmonary fibrosis.

SCID & Combined Immunodeficiency

44. **True about SCID in children is:**

A. **It is due to adenosine deaminase deficiency**
B. Humoral immunity is predominantly affected
C. Child should receive double dose of all vaccines
D. Patients with SCID have very small thymuses
E. Tonsils & adenoids are absent or underdeveloped

Ans A d. Patients with SCID have very small thymuses; e. Tonsils & adenoids are absent or underdeveloped SCID is a characteristic example of combined antibody and cellular defects; Live vaccines are contraindicated in SCID

45. **All of the following are not seen in SCID except?**

A. C1 inhibitor
B. S protein
C. **C3b inactivator**
D. C2 kinins

Ans C GVHD SCID (Severe Combined Immunodeficiency)
• It is caused by diverse genetic mutations that lead to absence of all adaptive immune function
• Combined deficiency of B & T cells seen; so Autoimmune ds, Granuloma formation & Graft rejection are absent
• While, GVHD which is due to immune-competent cells attacking a immunosuppressed host can still be seen

46. **All are true regarding SCID except:**

A. Due to deficiency of adenosine deaminase
B. **Toxicity of leucocytes due to accumulation of cAMP**
C. Both cellular and humoral immunity affected
D. Bone marrow transplantation is the treatment

Ans B In SCID, deficiency of ADA leads to accumulation of deoxyadenosine and its derivatives (e.g., deoxy-ATP) (not AMP), which are toxic to rapidly dividing immature lymphocytes, especially those of the T-cell lineage.

47. **Which is not a characteristic of Wegener's granulomatosis:**

A. Polyarteritis nodosa
B. Wegener's granulomatosis
C. Microscopic polyangiitis
D. **Churg-Strauss syndrome**

Ans D Involves large vessels Wegener's granulomatosis involves small vessels Chapter 7: Diseases

48. **A 17-month-old girl presented with history of recurrent attacks of pneumonia since the age of 8 months. You suspect a predominant B-cell defect. Of the following, the BEST simple initial screening test is**

A. **IgA measurement**
B. IgG measurement
C. IgM measurement
D. IgE measurement
E. isohemagglutinins titer

Ans A If the IgA level is normal, selective IgA deficiency, which is the most common B- cell defect, is excluded, as are most of the permanent types of hypogammaglobulinemia, as IgA is usually very low or absent in those conditions. If IgA is low, IgG and IgM should also be measured.

49. **Which of the following statements is TRUE regarding diurnal variation of the absolute eosinophil count (AEC)?**

A. higher in the evening
B. lower in the afternoon
C. higher in the afternoon
D. lower in the early morning
E. **higher in the early morning**

Ans E The absolute eosinophil count (AEC) is calculated as the white blood cell count/μL × percent of eosinophils, it is usually <450 cells/μL and varies diurnally, with eosinophil numbers higher in the early morning and diminishing as endogenous glucocorticoid levels rise.

50. **Leukemoid reactions (WBC count >50,000/µL) are caused by all the following**

A. shigellosis
B. septicemia
C. salmonellosis
D. **chronic granulomatous disease**
E. leukocyte adhesion defect with infection

Ans D

51. **A significant risk factor in the development of allergic rhinitis in children is**

A. **positive family history**
B. serum IgA higher than 100 iu/ml
C. alcoholic mother
D. diabetic mother
E. father with renal disease

Ans A is the major risk factor. Other risk factors are raised serum IgE more than 100 IU/ml, smoking mother, and those with heavy exposure to indoor allergens.

52. **Causes of hormonal rhinitis include all the following EXCEPT**

A. hypothyroidism
B. exercise
C. cancer
D. vaculitides
E. **systemic lupus erythematosis**

Ans E

53. **Of the following, the major risk factor for persistent asthma is**

A. **eczema**
B. allergic rhinitis
C. wheezing apart from colds
D. food allergen sensitization
E. ≥4% peripheral blood eosinophils

Ans A Early childhood risk factors for persistent asthma have been identified and have been described as major (parent asthma, eczema, inhalant allergen sensitization) and minor (allergic rhinitis, wheezing apart from colds, ≥4% peripheral blood eosinophils, food allergen sensitization) risk factors.

54. **All the following are considered as risk factors for asthma morbidity and mortality**

A. poverty
B. air pollution exposure
C. **female gender**
D. poor response to systemic steroid therapy
E. sudden asphyxia episodes

Ans C Male gender.

55. **Use of a stoplight zone system (green, yellow, red) tailored to each child's "personal best" PEF values can optimize effectiveness and interest. Of the following, the PEF value in yellow zone is**

A. <10%
B. 10-30%
C. 30-50%
D. **50-80%**
E. 80-100%

Ans D The green zone (80-100% of personal best) indicates good control; the yellow zone (50-80%) indicates less-than-optimal control and necessitates increased awareness and treatment; the red zone (<50%) indicates poor control and greater likelihood of an exacerbation, requiring immediate intervention.

56. **Older children with chronic atopic dermatitis have lichenification and a tendency to be localized to**

A. **flexural folds of the extremities**
B. forehead
C. scalp
D. diaper area
E. cheeks

Ans A

57. **One of the following statements is TRUE regarding skin tests in identifying food allergies in patients with atopic dermatitis**

A. negative skin and blood test results for allergen-specific ige have a low predictive value for excluding suspected allergens
B. positive results of skin or blood tests using foods often correlate with clinical symptoms and no need to be confirmed with controlled food challenges
C. extensive elimination diets are commonly required
D. even with multiple majority of patients react to more than 3 foods
E. **potential allergens can be identified by a careful history and performing selective skin prick tests or in vitro blood testing for allergen- specific IgE**

Ans E For the other stems; A- high predictive value, B- Positive skin test often do not correlate with clinical symptoms, C- Rarely required, and D- The majority react to less than 3 foods.

58. **The MOST notorious stinging insect is**

A. honeybee
B. harvester ants
C. hornet
D. bumblebee
E. **yellow jackets**

Ans E are aggressive and ground dwelling, and they linger near activities involving food.

59. **The MOST common presenting complain of allergic conjunctivitis is**

A. pain
B. **itching**
C. redness
D. blurred vision
E. purulent discharge

Ans B Patients with allergic conjunctivitis complain of variable ocular itching, rather than pain, with increased tearing.

60. **The differential diagnosis of chronic urticaria includes the following EXCEPT**

A. cutaneous mastocytosis
B. systemic mastocytosis
C. complement- mediated mast cell degranulation in malignancies
D. cutaneous blistering disorders
E. **dermatographism**

Ans E

61. **The MOST common cause of food protein induced enteropathy is**

A. peanuts
B. fish
C. **cow's milk**
D. eggs
E. tree nuts

Ans C

62. **Drug Rash with Eosinophilia and Systemic Symptoms (DRESS) can be caused by all the following EXCEPT**

A. anticonvulsants
B. sulfonamides
C. **B-lactam antibiotics**
D. minocycline
E. allopurinol

Ans C They can cause anaphylaxis.

Agammaglobulinemia & CVID

63. **A 3-year-old child presents with recurrent pneumonia, eczema & thrombocytopenia. Which protein synthesis is abnormal in this child?**

A. **Wasp**
B. Hamartin
C. Adenosine deaminase
D. HLA 1a

Ans A Recurrent infections, eczema & thrombocytopenia in a child suggest a diagnosis of Wiskott Aldrich syndrome; In this X-linked recessive syndrome, WASP gene & its product are defective;

64. **All are seen in Henoch Schönlein purpura except:**

A. **Thrombocytopenia**
B. Glomerulonephritis
C. Arthralgia
D. Abdominal pain

Ans A Palpable purpura without thrombocytopenia is seen in Henoch Schönlein purpura (HSP).

65. **Kawasaki disease is associated with all except:**

A. Erythema
B. Posterior cervical lymphadenopathy
C. **Thrombocytopenia**
D. Conjunctivitis

Ans C M neMonic Diagnostic Criteria for Kawasaki Disease Fever persisting at least 5 days with presence of at least 4 features out of 5 (CREAM): Conjunctivitis: Rash, Edema & erythema of palms & soles, Adenopathy and Mucosal involvement: Strawberry tongue

66. **In Wegener's granulomatosis cytoplasmic anti neutrophilic antibodies are directed against:**

A. Proteinase
B. Proteinase
C. **Proteinase**
D. Proteinase

Ans C

67. **A 3-year-old boy presented with recurrent attacks of pneumonia and otitis media since the 1st birthday. Your diagnosis is X-linked agammaglobulinemia. Of the following, the MOST likely offending organism is**

B. Mycoplasma
C. Pneumocystis jiroveci
D. Staphylococcus aureus
E. **Streptococcus pneumonia**

Ans E They acquire infections with extracellular pyogenic organisms, such as Streptococcus pneumoniae and Haemophilus influenzae, unless they are given prophylactic antibiotics or immunoglobulin therapy.

68. **Eosinophilia can be seen in all the following EXCEPT**

A. scabies
B. urticaria
C. **corticosteroid therapy**
D. Hodgkin disease
E. hypersensitivity drug reactions

Ans C Exogenous and endogenous glucocorticoid suppresses absolute eosinophil count.

69. **Shifting to the left (proportion of immature neutrophil cells >5%) may occasionally be caused by all the following EXCEPT**

A. burns
B. trauma
C. hemorrhage
D. acute hemolysis
E. **megaloblastic anemia**

Ans E Higher degrees of left shift with more immature neutrophil precursors are indicative of serious bacterial infections and may be a direct sign of depletion of the bone marrow reserve pool of neutrophils. Marked left shift may occasionally be encountered with trauma, burns, surgery, acute hemolysis, or hemorrhage.

70. **The critical period for the development of allergic rhinitis exists when the genetically susceptible individual at greatest risk of sensitization in**

A. early adolescence
B. late adolescence
C. school age
D. toddlerhood
E. **early infancy**

Ans E

71. **The reported rate of remission of allergic rhinitis among children is about**

A. 1-10%
B. **11-20%**
C. 21-30%
D. 31-40%
E. 41-50%

Ans B

72. **Of the following, the strongest identifiable factor for the persistence of childhood asthma is**

A. **allergy**
B. male gender
C. low birth weight
D. parental asthma
E. lower respiratory tract infection

Ans A

73. **The best ''rescue'' medication in the treatment of acute asthma symptoms is**

A. oral SABA
B. **inhaled SABA**
C. oral corticosteroid
D. inhaled ipratropium
E. inhaled corticosteroid

Ans B up to 3 treatments in 1 hr. If the child has an incomplete response to initial treatment with rescue medication (persistent symptoms and/or a PEF value <80% of personal best), a short course of oral corticosteroid therapy (prednisone 1-2 mg/kg/day [not to exceed 60 mg/day] for 4 days), in addition to inhaled β-agonist therapy, should be instituted.

74. **All levels of persistent asthma should be treated with daily medications to improve long-term control. All the following are long-term controller medications EXCEPT**

A. **albuterol**
B. omalizumab
C. montaleukast
D. inhaled corticosteroid
E. sustained-release theophylline

Ans A Long-term controller medications are inhaled corticosteroid, LABAs, leukotriene modifiers, sustained-release theophylline, and nonsteroidal antiinflammatory agents. An anti-IgE preparation, omalizumab (Xolair), is approved by the FDA for use as an add- on therapy in children ≥12 yr that have moderate to severe allergic asthma that is difficult to control.

75. **The following are the major clinical features of atopic dermatitis (AD) EXCEPT**

A. pruritus
B. family history
C. relapsing dermatitis
D. facial eczema in infants
E. **extensor eczema in adolescents**

Ans E The major clinical features in atopic dermatitis are (pruritus, facial and extensor eczema in infants and children, flexural eczema in adolescents, relapsing dermatitis and personal or family history).

76. **One of the following medications used in treatment of atopic dermatitis should be discontinued after failure to achieve good results within 4-6 weeks**

A. methotrexate
B. azathioprine
C. cyclosporine
D. **mycophenolate mofetil**
E. omalizumab

Ans D

77. **Delayed/Late reaction to stinging venom may precipitate all the following EXCEPT**

A. vasculitis
B. serum sickness
C. encephalopathy
D. **hemolytic anemia**
E. nephrotic syndrome

Ans D

78. **Topical ophthalmic medications usually cause**

A. **contact allergy**
B. allergic conjunctivitis
C. vernal keratoconjunctivitis
D. atopic keratoconjunctivitis
E. giant papillary conjunctivitis

Ans A typically involves the eyelids but can also involve the conjunctivae. It is being recognized more frequently in association with increased exposure to topical medications, contact lens solutions, and preservatives.

79. **Sudden collapse in the presence of cutaneous symptoms in a previously healthy child should raise suspicion of**

A. vasovagal collapse
B. **anaphylaxis**
C. aspiration
D. pulmonary embolism
E. seizure disorder

Ans B Other choices are to be considered in cases where there are no cutaneous manifestations.

80. **The MOST severe form of food protein-induced enteropathy is**

A. **celiac disease**
B. eosinophilic esophagitis
C. oral allergy syndrome
D. acute gastrointestinal allergy
E. food protein proctocolitis

Ans A

81. **All the following are risk factors for adverse drug reactions EXCEPT**

A. prior exposure
B. route of administration
C. dosing schedule
D. genetic predisposition
E. **atopy**

Ans E does not appear to predispose patients to allergic reactions to low- molecular-weight compounds, but atopic patients in whom an allergic reaction develops have a significantly increased risk of serious reaction.

Phagocyte Disorders

82. A child having delayed separation of umbilical cord with leukocytosis with Down syndrome and recurrent infections. Diagnosis:

A. L eucocyte adhesion deficiency
B. Neonatal sepsis
C. Histiocytosis-X
D. All of above

Ans A Leucocyte adhesion deficiency Characteristic Clinical Patterns in important Primary Immunodeficiencies: Diagnosis Features In newborns and young infants (0 to 6 months) DiGeorge anomaly HypocalcemiaQ, heart disease, unusual facies Leukocyte adhesion defect Delayed umbilical cord detachment, leukocytosis, recurrent infections Severe combined immunodeficiency Diarrhea, pneumonia, thrush, failure to thrive Wiskott-Aldrich syndrome Bloody stools, draining ears, eczema

83. Which one of the following statements is correct regarding chronic granulomatous disease:

A. It is an autosomal dominant disease
B. It is characterized by abnormal bacterial phagocytosis
C. Recurrent streptococcal infections are usual in this disease
D. Nitroblue tetrazolium test is useful for screening

Ans D The nitroblue tetrazolium (NBT) test — and now more commonly the DHR flow cytometry assay — is used to screen for chronic granulomatous disease, which impairs the oxidative (respiratory burst) killing of catalase-positive organisms, not phagocytosis itself.

84. Kawasaki disease is associated with all except:

A. Thrombocytopenia
B. Cervical lymphadenopathy
C. Red eye
D. Rash OTHER CONNECTIVE TISSUE DISEASES

Ans A OTHER

85. Characteristic feature of Goodpasture's syndrome?

A. Lumpy-bumpy deposits on immunofluorescence
B. Serum antibodies against alpha 3 NC 1 domain of collagen–IV
C. Serum antibodies against alpha 1 NC 1 domain of collagen III
D. Anti DNAse antibodies positive

Ans B Serum antibodies against alpha 3 NC1 domain of collagen – IV • In Goodpasture syndrome, type 1 RPGN & type II hypersensitivity reaction are seen.
• It is characterized by serum antibodies against alpha 3 NC1 domain of collagen – IV.

86. A 4.5-year-old boy presented with history of recurrent attack of secretary otitis media and purulent nasal discharge, Haemophilus influenza revealed by culture of ear discharge, on physical examination there is no tonsillar tissue and no palpable lymph nodes. Of the following, the MOST appropriate test to confirm the diagnosis is measurement of

A. flow cytometry
B. IgA concenteration
C. isohemagglutinins titer
D. IgG and IgM concenteration
E. antibodies to antigens of routine immunizations

Ans A The diagnosis of X-linked agammaglobulinemia (XLA) should be suspected if lymphoid hypoplasia is found on physical examination (minimal or no tonsillar tissue and no palpable lymph nodes). Flow cytometry is an important test to demonstrate the absence of circulating B cells, which will distinguish this disorder from common variable immunodeficiency, the hyper-IgM syndrome, and transient hypogammaglobulinemia of infancy.

87. The following conditions can cause eosinophilia EXCEPT

A. malaria
B. filariasis
C. giardiasis
D. ascariasis
E. amebiasis

Ans C Eosinophilia are caused by tissue-invasive as helminth infections; ascariasis, filariasis, amebiasis, malaria, and scabies whereas entrobius vermicularis and Giardia lamblia are solely intraluminal in gastrointestinal tract.

88. Severe neutropenia is defined as absolute neutrophil count (ANC) less than

A. 500/µL
B. 750/µL
C. 1000/µL
D. 1250/µL
E. 1500/µL

Ans A Mild neutropenia, is an absolute neutrophil count (ANC) of 1,000-1,500/µL. Moderate neutropenia, is an absolute neutrophil count (ANC) of 500- 1,000/µL. Severe neutropenia, is an absolute neutrophil count (ANC) <500/µL.

89. Chronic neutropenia by definition lasts longer than

A. 1 mo
B. 3 mo
C. 6 mo
D. 9 mo
E. 12 mo

Ans B Chronic neutropenia by definition lasts longer than 3 months and arises from reduced production, increased destruction or excessive splenic sequestration of neutrophils.

90. Chronic neutropenia may be induced by

A. adenovirus
B. enteroviruses
C. cytomegalovirus
D. influenzas A and B
E. respiratory syncytial virus

Ans C Chronic neutropenia often accompanies infection with Epstein-Barr virus, cytomegalovirus, or HIV while the other distracters cause acute neutropenia.

91. **Recurrent infections with neutropenia are a distinctive feature of**

A. Pompe disease
B. Hurler disease
C. Gaucher disease
D. Glycogen storage disease Ia (GSDIa)
E. **Glycogen storage disease Ib (GSDIb)**

Ans E Recurrent infections with neutropenia are a distinctive feature of glycogen storage disease (GSD) type Ib. As in classic von Gierke disease (GSDIa), glycogen storage in GSDIb causes massive hepatomegaly and severe growth retardation.

92. **All the following drugs can induce neutropenia EXCEPT**

A. penicillin
B. **tetracycline**
C. phenobarbital
D. phenothiazine
E. chloramphenicol

Ans B The process of neutropenia likely arises from effects of drugs, such as propylthiouracil or penicillin, that act as haptens to stimulate antibody formation, or drugs, such as quinine, that induce immune complex formation. Late-onset neutropenia can occur after rituximab therapy. Idiosyncratic reactions, for example to chloramphenicol, are unpredictable with regard to dose or duration of use.

93. **A 15-year-old boy with rheumatoid arthritis, on quinine for 6 months, presented with fever, aphthous stomatitis, and fatigue for the last 5 days. His absolute neutrophil count (ANC) was 700/µL. The MOST next appropriate therapeutic measure to be done when quinine withdrawal fails to improve neutropenia is**

A. administration of IVIG
B. aggressive use of antibiotics
C. transplantation of hematopoietic stem cells
D. administration of pulse methylprednisolone
E. **administration of recombinant human granulocyte colony-stimulating factor**

Ans E

94. **The MOST common pathogen in chronic granulomatous disease (CGD) is**

A. **S. aureus**
B. Aspergillus
C. Salmonella
D. Mycobacterium
E. Candida albicans

Ans A The most common pathogen is S. aureus, although any catalase-positive microorganism may be involved.

95. **The diagnosis of chronic granulomatous disease CGD is MOST often made by**

A. DNA analysis
B. **flow cytometry**
C. neutrophil G6PD assay
D. erythrocyte G6PD assay
E. nitroblue tetrazolium dye test

Ans B The diagnosis is most often made by performing flow cytometry using dihydrorhodamine 123 (DHR) to measure oxidant production through its increased fluorescence when oxidized by H2O2.

96. **To reduce the number of bacterial infection in patients with chronic granulomatous disease (CGD), they should be given daily**

A. penicillin
B. cloxacillin
C. interferon-γ
D. itraconazole
E. **trimethoprim-sulfamethoxazole**

Ans E A placebo-controlled study found that interferon-γ significantly reduces the number of hospitalizations and serious infections. Itraconazole administered prophylactically reduces the frequency of fungal infections.

97. **In chronic granulomatous disease (CGD), the best method to detect and follow up deep-seated infection is**

A. **erythrocyte sedimentation rate (ESR)**
B. blood culture
C. WBC count
D. C-reactive protein
E. immature to mature neutrophil ratio

Ans A The erythrocyte sedimentation rate (ESR) can be quite helpful. If the child does not have a deep-seated infection, the ESR will be normal or will normalize within several days with standard management. However, if it does not, a search for deep tissues is warranted, as is consideration of empiric antibiotics.

98. **All the following medications are encountered to cause serum sickness EXCEPT**

A. **ciprofloxacin**
B. meropenem
C. rituximab
D. trimethoprim sulfate
E. carbamazepine

Ans A

99. **The only way to establish the diagnosis of cell-mediated food reactions is**

A. breath hydrogen test
B. endoscopy
C. **elimination and challenge test**
D. IgE testing
E. skin test

Ans C There are no laboratory studies to help identify foods responsible for cell- mediated reactions. Consequently, elimination diets followed by food challenges are the only way to establish the diagnosis.

100. **Of the following, the agent that does not share an identical side chain with penicillin is**

A. cephalexin
B. cefotaxime
C. **cefixime**
D. ceftazidime
E. ceftriaxone

Ans C

Complement Deficiencies

101. **Both B and T cell defect is present in:**

A. **SCID**
B. Common Variable immunodeficiency
C. Wiskott-Aldrich syndrome
D. X-linked Agammaglobulinemia
E. Chronic mucocutaneous candidiasis

Ans A b. Common Variable immunodeficiency Common variable Immunodeficiency is predominantly a B cell defect, but it also shows defect in T cell activation.

102. **Henoch-Schönlein purpura is characterized by the following except?**

A. Glomerulonephritis
B. Hematochezia
C. **Thrombocytopenia**
D. Palpable purpura

Ans C Kidney involvement (glomerulonephritis) & bowel involvement (blood in stools, abdominal pain) can be seen in HSP.

103. **Joint erosions are typically absent in:**

A. Rheumatoid arthritis
B. **Systemic lupus erythematosus**
C. Psoriatic arthritis
D. Osteoarthritis

Ans B Arthritis seen in SLE is Non-erosive arthritis (without deformity).

104. **ANCA positive entities are?**

A. Polyarteritis nodosa
B. **Wegener's granulomatosis**
C. Microscopic polyangitis
D. Giant cell arteritis
E. Churg-Strauss syndrome

Ans B c. Microscopic polyangiitis, e. Churg-Strauss syndrome

105. **A 2-year-old boy presented with failure to thrive, neonatal hypocalcemic seizure, and three attacks of sepsis. Examination reveals cleft palate and holo-systolic murmur. Of the following, the genetic predisposition of this condition is**

A. **microdeletions of chromosome 22q11.2**
B. interstitial deletion of the long arm of chromosome 15 (15q11-13)
C. presence of two copies of paternally derived chromosome 11p15.5
D. presence of mutated gene on long arm of chromosome 11 (11q22-23)
E. presence of abnormal gene, on the proximal arm of the X chromosome at Xp11.22-11.23

Ans A A. DiGeorge syndrome, B. Prader-Willi syndrome, C. Beckwith-Wiedemann syndrome, D. Wiskott-Aldrich syndrome, and E. ataxia-telangiectasia.

106. **The highest white blood cells count is seen at**

A. **birth**
B. 12 hour of life
C. 1 week of life
D. 1 year of life
E. adolescence

Ans A For newborns, the mean WBC count at birth is high, followed by a rapid fall beginning at 12 hr through the 1st wk of life. Thereafter, values are stable until 1 yr of age, after which a slow steady decline in the WBC count continues throughout childhood until adult values are reached during adolescence.

107. **A 15-year-old boy presented with high fever, lymphadenopathy, hepatosplenomegaly, recurrent skin infections, silvery hair, and photophobia. Last year, he developed ataxia. The MOST characteristic finding in this condition is**

A. neutropenia
B. thrombocytopenia
C. **prolonged bleeding time**
D. prolonged prothrombine time
E. prolonged partial prothrombine time

Ans C Patients with Chédiak-Higashi syndrome (CHS) have prolonged bleeding times with normal platelet counts, resulting from impaired platelet aggregation associated with a deficiency of the dense granules containing adenosine diphosphate and serotonin.

108. **The occurrence of 3 or more episodes of rhinorrhea in the first year of life is associated with allergic rhinitis (AR) at age of**

A. 11 yr
B. 9 yr
C. **7 yr**
D. 5 yr
E. 3 yr

Ans C

109. **What percentage of allergic rhinitis patients has allergic conjunctivitis?**

A. >30%
B. >40%
C. >50%
D. **>60%**
E. >70%

Ans D Allergic conjunctivitis, characterized by itching, redness and swelling of the conjunctivae, has been reported in at least 20% of the population and in more the 70% of patients with allergic rhinitis, most frequently in older children and young adults.

110. **The following actions may help reduce the likelihood of asthma development**

A. healthy diet
B. **immunization**
C. active lifestyle
D. avoidance of tobacco smoking
E. prolonged breastfeeding > 4 months

Ans B

111. The best treatment option for step -6- severe persistent asthma in a 6-year-old boy is

A. medium dose inhaled corticosteroids with long acting B-agonists
B. high dose inhaled corticosteroids with leukotriene receptor antagonist
C. low dose inhaled corticosteroids with leukotriene receptor antagonist
D. high dose inhaled corticosteroids with long acting B-agonist and oral corticosteroids
E. high dose inhaled corticosteroids with long acting B-agonist and oral corticosteroidsalong with omalizumab therapy

Ans D

112. Adverse effects of frequently administered β-agonist therapy in asthma include all the following EXCEPT

A. tremor
B. irritability
C. tachycardia
D. hypokalemia
E. mouth dryness

Ans E

113. A high index of suspicion of which of the following conditions is to be undertaken in a patient with atopic dermatitis and failure to thrive

A. Wiscott-Aldrich syndrome
B. severe combined immune deficiency
C. Histiocytosis
D. hyper IgE syndrome
E. chronic granulomatous disease

Ans C other mentioned disorders are associated with infections.

114. Systemic corticosteroids are rarely indicated in the treatment of atopic dermatitis because

A. toxic side effects after long term use
B. rebound flare after therapy discontinuation
C. cannot do more than what topical can
D. tapering is required even after short term use
E. antimetabolites can do better in modifying disease course

Ans B The dramatic clinical improvement that may occur with systemic corticosteroids is frequently associated with a severe rebound flare of atopic dermatitis after therapy discontinuation.

115. In the presence of convincing history of a severe systemic reaction, the next diagnostic step for those with initially negative skin test is

A. in vitro serum assay for venom-specific IgE
B. repeat skin test after 4-6 weeks
C. serum tryptase level
D. plasma histamine
E. basophil histamine release test

Ans A is recommended if skin test results are negative in the presence of a convincing history of a severe systemic reaction.

116. Contact lenses are associated with

A. contact allergy
B. allergic conjunctivitis
C. vernal keratoconjunctivitis
D. atopic keratoconjunctivitis
E. giant papillary conjunctivitis

Ans E has been linked to chronic exposure to foreign bodies, such as contact lenses, both hard and soft, ocular prostheses, and sutures.

117. The MOST common childhood symptoms of hereditary angioedema is

A. cutaneous non-pitting but pruritic edema associated with urticaria
B. cutaneous pitting but non-pruritic edema not associated with urticaria
C. cutaneous pitting and pruritic edema associated with urticaria
D. cutaneous non-pitting and non-pruritic edema associated with urticaria
E. cutaneous non-pitting and non-pruritic edema not associated with urticaria

Ans E

118. Unpredictable drug reactions include

A. dose dependent
B. drug toxicity
C. drug interactions
D. adverse effects
E. allergic reaction

Ans E

Wiskott-Aldrich & Ataxia Telangiectasia

119. All of the following are true except:

A. NADPH oxidase acts via superoxide ions
B. Chediak-Higashi syndrome is due to defective phago-lysosome formation
C. In Brutons agammaglobulinemia opsonisation is not affected
D. Myeloperoxidase action is mainly due to OCl-

Ans C In Brutons agammaglobulinemia opsonization is not affected IgM is important for opsonization, so in Brutons agammaglobulinemia opsonisation is affected.

120. About Wiskott Aldrich not true is?

A. Common variable immunodeficiency
B. Severe combined immunodeficiency
C. Chronic granulomatous disease
D. Nezelof syndrome

Ans C Reduced IgA Remember IgE & IgA are ElevAted in Wiskott Aldrich syndrome.

121. **In which of the following c-ANCA is seen?**

A. **Wegener's granulomatosis**
B. Polyarteritis nodosa
C. Churg-Strauss syndrome
D. Bechet's syndrome

Ans A C-ANCA P-ANCA (against Myeloperoxidase) Wegener's GranulomatosisQ • Microscopic PAN (microscopic polyangitis) • Crescentic GlomerulonephritisQ (90-95%) • Churg-Strauss syndromeQ • GoodpasteurQ's syndromeQ
• Drugs (hydralazine, propythiouracil) • Ulcerative colitis
• Primary sclerosing cholangitisQ

122. **In Wegener's glomerulonephritis characteristic feature seen is:**

A. Granuloma in the vessel wall
B. **Focal necrotizing glomerulonephritis**
C. Nodular glomerulosclerosis
D. Interstitial granuloma

Ans B Note: Granuloma in vessel wall is seen in Wegener's Granulomatosis, but presence of granuloma alone Wegener's; It should be granulomatous necrotizing vasculitis.

123. **In addition to infection and bleeding, the MOST common cause of death in Wiskott- Aldrich syndrome is**

A. renal failure
B. graft versus host disease
C. protracted bloody diarrhea
D. **EBV-associated malignancies**
E. development of autoimmune disease

Ans D Wiskott-Aldrich syndrome, an X-linked recessive syndrome, is characterized by atopic dermatitis, thrombocytopenic purpura with normal-appearing megakaryocytes but small defective platelets, and undue susceptibility to infection. Survival beyond the teens is rare; infections, bleeding, and EBV-associated malignancies are major causes of death.

124. **The treatment of choice for a 3-year-old boy with Wiskott-Aldrich syndrome is**

A. splenectomy
B. use of killed vaccines
C. aggressive antibiotics
D. administration of IVIG
E. **bone marrow or cord blood transplantation**

Ans E Treatment; Good supportive care includes appropriate nutrition, routine IVIG, use of killed vaccines, aggressive management of eczema and associated cutaneous infections, platelet transfusion for serious bleeding episodes, splenectomy if a transplant is not going to be done, and high-dose IVIG with systemic steroids for autoimmune complications. Bone marrow or cord blood transplantation is the treatment of choice and is usually curative.

125. **A 3-year-old boy presented with recurrent subcutaneous abscesses, lymphadenitis, osteomyelitis, and family history of recurrent infections. The hallmark of this condition is**

A. hypocalcemia
B. atopic dermatitis
C. thrombocytopenia
D. **granuloma formation**
E. prolonged bleeding time

Ans D and inflammatory processes are a hallmark of chronic granulomatous disease CGD and may be the presenting symptoms that prompt testing for CGD if they cause pyloric outlet obstruction, bladder outlet or ureter obstruction, or rectal fistulas and granulomatous colitis simulating Crohn disease.

126. **Early introduction of all the following decrease the risk of asthma and allergic rhinitis**

A. rye
B. fish
C. egg
D. **meat**
E. wheat

Ans D There is a decreased risk of asthma, allergic rhinitis (AR), and atopic sensitization with early introduction to wheat, rye, oats, barley, fish and eggs.

127. **The percentage of allergic rhinitis in asthmatic patients is 78%, while the percentage of asthma in patients with allergic rhinitis is**

A. 18%
B. 28%
C. **38%**
D. 48%
E. 58%

Ans C

128. **Recurrent coughing and wheezing occurs in 35% of preschool-age children. Those who continue to have persistent asthma into later childhood are approximately**

A. one-fifth
B. one-fourth
C. **one-third**
D. one-half
E. two-thirds

Ans C Of these, approximately one-third who continue to have persistent asthma into later childhood, and approximately two-thirds improve on their own through their teen years.

129. **A common finding of chest radiograph in a child with asthma is**

A. **peribronchial thickening**
B. atelectasis
C. pneumothorax
D. bronchiectasis
E. pneumomediastinum

Ans A and flattening of diaphragm.

130. **Atopic dermatitis affects many children worldwide, the exact percentage approximating**

A. 5-10%
B. **10-30%**
C. 30-50%
D. 50-70%
E. 70-90%

Ans B

131. **One of the following metabolic conditions is not deemed in the differential diagnosis of atopic dermatitis**

A. zinc deficiency
B. pyridoxine deficiency
C. **cobalamin deficiency**
D. niacin deficiency
E. phenylketonuria

Ans C

132. **The least potent topical steroid used in treatment of atopic dermatitis is**

A. fluticasone
B. desonide
C. betamethasone dipropionate
D. **hydrocortisone**
E. clobetasol propionate

Ans D is labeled as group 7 topical steroids which is the least potent one.

133. **Sting sites rarely become infected possibly owing to**

A. cleansing the area immediately after insect
B. the over use of antibacterial creams for the area
C. **venom constituents have antibacterial action**
D. vasospasm after stinging impedes bacterial invasion
E. venom immunotherapy has antibacterial properties

Ans C

134. **Conjunctivitis medicamentosa is a consequence of chronic use of**

A. antihistamines
B. steroids
C. **decongestants**
D. anti-inflammatory
E. mast cell stabilizers

Ans C Topical decongestants act as vasoconstrictors; adverse effects of topical include burning and rebound hyperemia or conjunctivitis medicamentosa with chronic use.

135. **Once the offending agent causing serum sickness is discontinued, the symptoms resolve spontaneously within**

A. 1-4 days
B. **1-4 weeks**
C. 5-8 weeks
D. 9-12 weeks
E. 13-18 weeks

Ans B Symptoms lasting longer suggest another diagnosis.

136. **Which of the following drugs can cause Stevens - Johnson syndrome?**

A. dapsone
B. hydralazine
C. **sulfonamide**
D. procainamide
E. bleomycin

Ans C Antibacterial sulfonamides, anticonvulsants, oxicam NSAIDs, and allopurinol have been implicated in causing Stevens - Johnson syndrome and toxic epidermal necrolysis.

DiGeorge Syndrome

137. **In genetic deficiency of MPO, the increased susceptibility to infection is due to:**

A. Defective production of Prostaglandins
B. Defective rolling of' neutrophils
C. **Inability to produce hydroxyl halide radicals**
D. Inability to produce hydrogen peroxide

Ans C In MPO deficiency there is decreased microbial killing because of defective MPO—H O system, so there is an inability to 2 2 produce hydroxyl halide radicals.

138. **Frequency of renal involvement in Henoch Schönlein purpura is?**

A. 1-5%
B. 5-10%
C. **10-50%**
D. 50-70%

Ans C Renal involvement in HSP:
• It occurs in up to 50% of children with HSP
• Manifests as microscopic hematuria, proteinuria, hypertension, frank nephritis, nephrotic syndrome, and acute or chronic renal failure; Progression to end-stage renal disease is uncommon in children (1-2%)

139. **A 9-year-girl presents with difficulty in combing, climbing stairs and erythematous lesion at MCP joint. Gower sign is positive. What is the next step for diagnosis? (AIIMS Nov 2015) 144 Section 1: General Pediatrics**

A. CK leve
B. Rheumatoid factor
C. **Electromyography**
D. Nerve conduction studies

Ans C This girl is probably suffering from Juvenile Dermatomyositis, as she has proximal muscle weakness & skin lesions suggestive of Gottron's papules. Discussing about the options one by one, a. CK level Creatine kinase level is usually elevated, but may be normal on initial presentation b. Rheumatoid factor It is negative in JDM c. Electromyography Shows signs of myopathy as well as muscle fiber necrosis d. Nerve conduction studies Typically normal unless severe muscle necrosis and atrophy are present

140. **ANCA positive is:**

A. Goodpasture syndrome
B. **Wegener's granulomatosis**
C. Sjogren syndrome
D. Relapsing polychondritis

Ans B

141. **All the following are features of ataxia-telangiectasia EXCEPT**

A. hepatic manifestation
B. high incidence of malignancy
C. chronic sinopulmonary disease
D. oculocutaneous telangiectasias
E. variable humoral and cellular immunodeficiency

Ans A The most prominent clinical features are progressive cerebellar ataxia, oculocutaneous telangiectasias, chronic sinopulmonary disease, a high incidence of malignancy, and variable humoral and cellular immunodeficiency.

142. **A 4-year-old boy presented with high fever, decreased appetite, aphthous stomatitis, and cellulitis of the right calf muscle. His absolute neutrophil count (ANC) was 600/μL. The MOST likely organisms causing this condition are**

A. Staphylococcus aureus and Histoplsmosis
B. Staphylococcus aureus and rickettsial pox
C. Staphylococcus aureus and Cytomegalovirus
D. Staphylococcus aureus and Pneumocystis carinii
E. Staphylococcus aureus and gram-negative bacteria

Ans E Infections commonly associated with neutropenia include cellulitis, furunculosis, perirectal inflammation, colitis, sinusitis, and otitis media, as well as more serious infections such as pneumonia, deep tissue abscess, and sepsis.

143. **All the following are characteristic in properdin deficiency EXCEPT**

A. dermal vasculitis
B. male predominance
C. systemic lupus erythematosus
D. susceptibility to N. meningitidis meningitis
E. normal serum hemolytic complement activity

Ans C Typical discoid lupus erythematosus is common.

144. **The main differentiating item between intermittent versus persistent allergic rhinitis is**

A. school performance
B. troublesome symptoms
C. daily activities
D. number of attacks per week
E. sleep pattern

Ans D The symptoms of intermittent allergic rhinitis occur on <4 days per week or for <4 consecutive weeks. In persistent AR symptoms occur on >4 days per week and/or for >4 consecutive weeks.

145. **To avoid false-negative skin test results in atopic host, most sedating antihistamine should be withheld for 3-4 days, and nonsedating antihistamines for 5-7 days, while montelukast should be withheld for**

A. 1 day
B. 3 day
C. 5 day
D. 7 day
E. 9 day

Ans A

146. **Indications of severe exacerbation of asthma include the following EXCEPT**

A. breathlessness
B. accessory muscles use
C. labored breath
D. PEF or FEV1 value <70% of personal best values
E. mental status changes

Ans D Severe airflow limitation (PEF or FEV1 value <50% of personal best or predicted values).

147. **Bronchodilator response to an inhaled β-agonist (e.g., albuterol) is greater in asthmatic patients than nonasthmatic persons, the rate of improvement in FEV1 consistent with asthma is**

A. ≥10%
B. ≥12%
C. ≥14%
D. ≥16%
E. ≥18%

Ans B In asthmatic patients, an improvement in FEV1≥12% or >200 mL in response to bronchodilator is consistent with asthma, while FEV1 typically decreases during or after exercise by >15%.

148. **The hallmark of atopic dermatitis is**

A. intense pruritus
B. cutaneous reactivity
C. lichenification
D. fibrotic papules
E. dry skin

Ans E The hallmark of atopic dermatitis is dry skin, is results from compromise of the epidermal barrier, which leads to excess transepidermal water loss, allergen penetration, and microbial colonization.

149. **The first line therapy of atopic dermatitis (AD) is**

A. moisturizers
B. cyclosporine
C. antihistamine
D. tar preparations
E. topical corticosteroids

Ans A Because patients with AD have impaired skin barrier function from reduced lipid levels, they present with diffuse, abnormally dry skin, or xerosis. Moisturizers are first- line therapy.

150. **Which topical steroid is preferable to use for treatment of atopic dermatitis in the face?**

A. fluticasone 0.005%
B. desonide 05%
C. betamethasone dipropionate 0.05%
D. hydrocortisone 1%
E. clobetasol propionate 0.05%

Ans D Use the least potent steroid in the face and intertriginous areas.

151. **A TRUE indication of venom immunotherapy in a six-year-old boy is**

A. large local reaction with positive skin test and positive in vitro test
B. generalized cutaneous reaction with positive skin test and negative in vitro test
C. generalized cutaneous reaction with positive in vitro test and negative skin test
D. **systemic reaction with positive skin test and negative in vitro test**
E. systemic reaction with negative skin test and negative in vitro test

Ans D Systemic reaction at any age with positive skin test / in vitro test is an indication for venom immunotherapy. Large local reaction and generalized cutaneous reactions in children are not indications for both diagnostic tests (skin and in vitro tests) and venom immunotherapy.

152. **Chronic urticaria may be caused by**

A. latex
B. peanut
C. IV immunoglobulin
D. streptococcal pharyngitis
E. **systemic lupus erythematosus**

Ans E Autoimmune disease, like SLE, Juvenile rheumatoid arthritis, thyroid disease, inflammatory bowel diseases and celiac disease, are well established causes of chronic urticaria.

153. **Skin biopsy is not usually necessary for confirming the diagnosis of serum sickness because**

A. the complement C3 and C4 yield better results
B. the clinical examination is enough for the diagnosis
C. skin test is superior to biopsy
D. **the histopathological findings are not specific**
E. it may be complicated by local infection

Ans D Not specific and not diagnostic.

154. **Fixed drug eruption can be caused by**

A. allopurinol
B. cephalosporin
C. **nonsteroidal anti-inflammatory drug**
D. nitrofurantoin
E. infliximab

Ans C Tetracycline, NSAIDs, and carbamazepine are implicated in causing fixed drug eruption.

Autoimmune Disorders

155. **Nitroblue tetrazolium test is used for?**

A. **Phagocytes**
B. Complement
C. T cell
D. B cell

Ans A Nitroblue-tetrazolium (NBT) test tells whether or not NADPH oxidase enzymes are present in phagocytes.

156. **About Henoch-Schönlein purpura, all true except:**

A. **Thrombocytopenia**
B. Hematuria
C. Palpable purpura
D. Intususception

Ans A

157. **Most common vasculitis in children:**

A. Kawasaki disease
B. Takayasu disease
C. **HSP**
D. Microscopic PAN

Ans C Henoch-Schönlein purpura (HSP) is the most common vasculitis of childhood.

158. **Small vessel vasculitis includes:**

A. **Anti GBM antibody disease**
B. Takayasu disease
C. Kawasaki disease
D. Systemic Lupus erythematosus
E. IgA vasculitis

Ans A d. Systemic lupus erythematosus & e. IgA vasculitis Note: Anti GBM antibody disease (Goodpasture disease) & IgA vasculitis (Henoch-Schonlein purpura) are examples of small vessel vasculitis b. Thymic hyperplasia Thymic hypoplasia is seen in DiGeorge syndrome.

159. **Most sensitive antibody for SLE:**

A. **Anti-nuclear**
B. Anti-ds DNA
C. Anti-Smith
D. Anti-histone

Ans A Autoantibodies in Systemic Lupus Erythematosus (SLE)Q Antibody Antigen Recognized Clinical Utility Antinuclear antibodies Multiple nuclear Best screening test/Most sensitiveQ Anti-dsDNA DNA (double-stranded) SpecificQ, correlate with disease activity, nephritis, vasculitis Anti-Sm U1 RNA SLEQ Most Specific for Anti-RNP U1 RNA Not specific for SLE Anti-Ro/La (SS-A) hY RNA Sicca syndrome, neonatal lupus with congenital heart blockQ Antihistone Histones Drug-induced lupusQ

160. **Most specific autoantibodies found in Systemic Lupus Erythematosus are?**

A. Anti-RO
B. **Anti-Sm**
C. Anti-RNP
D. Anti dsDNA

Ans B

161. **Class IV lupus nephritis is:**

A. Mesangial lupus nephritis
B. Proliferative lupus nephritis
C. Membranous lupus nephritis
D. **Diffuse lupus nephritis**

Ans D Classification of Lupus Nephritis (International Society of Nephrology) Class I: Minimal Mesangial Lupus Nephritis (least common) Class IV: Diffuse Lupus Nephritis (most common) Class II: Mesangial Proliferative Lupus Nephritis Class V: Membranous Lupus NephritisQ Class III: Focal Lupus NephritisQ Class VI: Advanced Sclerotic Lupus Nephritis

162. **Commonest renal lesion in SLE is:**

A. Focal proliferative
B. **Diffuse proliferative**
C. Membranous
D. Minimal change disease

Ans B Class I is the Least common & class IV is the most common pattern of lupus nephritis.

163. **Antibody seen in neonatal lupus with congenital heart block:**

A. Anti-ribosomal
B. **Anti-Ro (SS-A)**
C. Anti-neuronal
D. Anti-histone

Ans B Anti-Ro/La (SS-A against hY RNA is seen in Sicca syndrome, neonatal lupus with congenital heart block.

164. **False statement regarding musculoskeletal manifestations of SLE:**

A. Intermittent polyarthritis
B. Commonly involves hands, wrist, knees
C. **Erosions seen on X-rays**
D. Ischemic necrosis of bone can occur, when the patient is on glucocorticoids

Ans C

165. **Acute febrile illness with vasculitis having predilection for the coronary arteries is seen in:**

A. **Kawasaki disease**
B. Adenovirus infection
C. Diphtheria
D. Measles

Ans A Coronary artery aneurysm is an important complication of Kawasaki disease

166. **ANCA positive vasculitis:**

A. Henoch-Schönlein purpura
B. Behcet's syndrome
C. **Wegener's granulomatosis**
D. None

Ans C

167. **Which is associated with vasculitis of medium size vessels:**

A. Temporal arteritis
B. Wegener's granulomatosis
C. **Classic PAN**
D. Tuberous sclerosis

Ans C Mnemonic: 'Milk is Poor in vit K': Medium size vessel vasculitis are Polyarteritis nodosa & Kawasaki disease

168. **ANCA positive vasculitis are:**

A. **Churg-Strauss syndrome**
B. Polyarteritis nodosa
C. Wegener granulomatosis
D. Takayasu arteritis
E. Henoch-Schönlein purpura

Ans A c. Wegener granulomatosis , Ghai 8/e

169. **Large vessel vasculitis is/are:**

A. **Takayasu disease**
B. Temporal arteritis
C. PAN
D. Giant cell arteritis
E. Wegener granulomatosis

Ans A b. Temporal arteritis, d. Giant cell arteritis

170. **Small vessel vasculitis is/are:**

A. Classical PAN
B. **Wegener's granulomatosis**
C. Giant cell arteritis
D. HSP
E. Churg-Strauss syndrome Chapter 7: Diseases of Immune System 145 Answers

Ans B d. HSP, e. Churg-Strauss syndrome , of Immune System 149 Answers with 20/e p 1221-1223 is not characteristic of p 631-635 of refractory asthma & Ghai 8/e p 631-635 M neMonic M Days on which rash appear in differ- ent diseases: "Very Sick People Must Take Daily Exercise" Day 1: V—Varicella Day 2: S—Scarlet fever Day 3: P—Pox (Smallpox) Day 4: M—Measles Day 5: T—Typhus Day 6: D—Dengue Day 7: E—Enteric fever
• Fetuses infected with Rubella after 16 weeks suffer no major abnormality

171. **Skin biopsy for diagnosis of possible urticarial vasculitis is recommended for**

A. urticarial lesions that persist at different locations for >24 hr
B. those with non- pigmented or non-purpuric components
C. those that burn more than itch
D. **those with associated collagen vascular diseases**
E. those appeared after peanuts ingestion

Ans D Skin biopsy is recommended for urticarial lesions that persist at the same location for >24 hr, those with pigmented or purpuric components, and those that burn more than itch. Collagen vascular diseases such as systemic lupus may manifest urticarial vasculitis as a presenting feature.

172. **Which one of the following agents is implicated in causing cutaneous lupus?**

A. corticosteroids
B. **calcium-channel blockers**
C. allopurinol
D. B-lactam antibiotics
E. sulfonamide

Ans B Hydrochlorothiazide and calcium channel blockers can cause cutaneous lupus.

173. **Systemic lupus erythematosus can be caused by**

A. **isoniazid**
B. sulfonamide
C. hydrochlorothiazide
D. allopurinol
E. nonsteroidal anti-inflammatory drug

Ans A Along with Hydralazine and procainamide.

174. **Vasculitis can be cause by**

A. isoniazid
B. sulfonamide
C. **penicillamine**
D. calcium channel blockers
E. tetracycline

Ans C Along with Hydralazine and propylthiouracil.

Vaccines & Immune Response

175. **Lack of leucocyte adhesion molecules (LAM) is associated with:**

A. **Delayed fall of umbilical cord**
B. Normal chemotaxis
C. Complement opsonization
D. Neutropenia

Ans A Leukocyte adhesion deficiency (LAD) Salient features LAD type 1 LAD type 2 Inheritance Autosomal recessive Autosomal recessive Basic defect Defect in CD 18 moleculeQ required for biosynthesis of Absence of Sialyl-Lewis X, the fucose-containing B2 chain shared by LFA-1 & Mac-1 integrins ligand for P & E-selection, due to defect in fucosyl- transferase responsible for binding fucose moieties to protein backbones

176. **True about Henoch-Schönlein pupura:**

A. **Palpable purpura**
B. Thrombocytopenia
C. C-ANCA positive
D. Kidney commonly affected
E. Thrombocytosis

Ans A d. Kidney commonly affected, e. Thrombocytosis

177. **Gottron's papules are found on?**

A. Face
B. Trunk
C. Spine
D. **Knee**

Ans D /e p Classic Gottron papules are bright pink or pale, shiny, thickened or atrophic plaques over the proximal interphalangeal joints & distal interphalangeal joints and occasionally on the knees, elbows, small joints of the toes, and ankle malleoli.

178. **All of the following condition(s) are associated with raised ANCA except:**

A. Wegener's granulomatosis
B. **Polyarteritis nodosa**
C. Microscopic polyangiitis
D. Churg-Strauss syndrome

Ans B

179. **The earliest clinical feature of ataxia-telangiectasia is**

A. **ataxia**
B. jaundice
C. inability to walk
D. ocular telangiectasias
E. cutaneous telangiectasias

Ans A typically becomes evident soon after these children begin to walk and progresses until they are confined to a wheelchair, usually by the age of 10-12 yr. The telangiectasias begin to develop at 3-6 yr of age.

180. **A 9-year-old boy with acute lymphoblastic leukemia on chemotherapy, presented with fever, aphthous stomatitis, and fatigue for the last 5 days. His absolute neutrophil count (ANC) was 450/µL and hemoglobin level was 10gm/dl. The NEXT appropriate therapeutic measure is**

A. administration of IVIG
B. transplantation of hematopoietic stem cells
C. administration of pulse methylprednisolone
D. **aggressive use of broad-spectrum antibiotics**
E. administration of recombinant human granulocyte colony-stimulating factor

Ans D A decline in the WBC count typically occurs 7-10 days after administration of the anticancer drug and may persist for 1-2 wk. Patients with chemotherapy/ radiation- related neutropenia and fever must be treated aggressively with broad-spectrum antibiotics.

181. **All the following conditions can induce lymphopenia EXCEPT**

A. sepsis
B. typhoid
C. **brucellosis**
D. corticosteroid use
E. cartilage-hair hypoplasia

Ans C Chronic bacterial infections such as tuberculosis and brucellosis may lead to a sustained lymphocytosis.

182. **Allergic shiners represent**

A. an upward rubbing of the nose with an open palm
B. continuous open- mouth breathing
C. **dark circles under the eyes**
D. transverse crease of the nose
E. conjunctival edema and itching

Ans C (A) represent allergic salute, while (B) represent allergic gape.

183. **Second generation antihistamines are preferable to first generation antihistamines because they have**

A. longer action
B. **less sedation**
C. more potency
D. more palatable effect
E. more shelf life

Ans B

184. **Spirometry is a helpful objective measure of airflow limitation; it depends on patient's ability to properly perform a full, forceful, and prolonged expiratory maneuver. Spirometry usually feasible in children**

A. ≥4 yr of age
B. **≥6 yr of age**
C. ≥8 yr of age
D. ≥10 yr of age
E. ≥12 yr of age

Ans B

185. **The diurnal variation in Peak Expiratory Flow (PEF) that is consistent with asthma is more than**

A. 10%
B. **20%**
C. 30%
D. 40%
E. 50%

Ans B

186. **Atopic dermatitis typically begins in**

A. **infancy**
B. toddlerhood
C. preschooler age
D. school age
E. adolescence

Ans A Approximately 50% of patients experience symptoms in the 1st yr of life, and an additional 30% are diagnosed between 1 and 5 yr of age.

187. **Prevention of atopic dermatitis in infancy includes the following measures EXCEPT**

A. breast feeding
B. feeding with a hypoallergenic hydrolyzed formula
C. use of probiotics
D. elimination of implicated food allergen from the mother diet
E. **use of special type napkins**

Ans E

188. **In atopic dermatitis, the presence of punched out erosions, vesicles, and infected skin lesions that fail to respond to oral antibiotics suggests infection with**

A. Herpes zoster
B. **Herpes simplex**
C. cutaneous warts
D. Trichophyton rubrum
E. Molluscum contagiosum

Ans B virus (HSV) can provoke recurrent dermatitis and may be misdiagnosed as S. aureusinfection, which can be diagnosed by a Giemsa-stained Tzanck smear of cells scraped from the vesicle base or by viral polymerase chain reaction or culture.

189. **Skeeter syndrome is a large local reaction to stinging and biting insects. Of the following, the TRUE statement is**

A. Ig E mediated response
B. usually occurs in older children
C. usually followed by anaphylaxis
D. **mosquito is the responsible agent**
E. misdiagnosed as erythema nodosum

Ans D Skeeter syndrome is a large local reaction to mosquito bite, it happens usually in young children and misdiagnosed as cellulitis. This is not an Ig E mediated reaction but because of the vasoactive and irritant material from the mosquito, it does not cause anaphylaxis.

190. **The drug of choice for cold-induced urticaria is**

A. loratadine
B. epinephrine IM
C. **cyproheptadine**
D. diphenhydramine
E. oral corticosteroid

Ans C

191. **All the following are complications of serum sickness EXCEPT**

A. carditis
B. glomerulonephritis
C. Guillain-barre syndrome
D. peripheral neuritis
E. **colitis**

Ans E

192. **Administration of the following vaccine is contraindicated in children with egg allergy**

B. Influenza
D. **yellow fever**
E. rabies

Ans D

Allergies & Anaphylaxis

193. **A 6-year-old child presented with abdominal pain and red, elevated skin rashes, mainly involving the lower limbs, as shown in the given image: What is the probable diagnosis?**

A. Kawasaki disease
B. **Henoch Schonlein purpura**
C. Varicella infection
D. Meningococcemia

Ans B Abdominal pain and palpable purpura, mainly involving the lower limbs, suggests a diagnosis of Henoch Schonlein purpura.

194. **Which of the following is not seen in adults?**

A. **Kawasaki disease**
B. HSP
C. Susac syndrome
D. Takayasu arteritis

Ans A

195. **Anti-histone antibodies are specific for:**

A. CNS lupus
B. **Drug induced lupus**
C. Neonatal lupus
D. Cutaneous lupus

Ans B Anti histone antibodies are directed against Histones & are seen in Drug-induced lupus.

196. **Wegener's granulomatosis involve:**

A. **Lung**
B. Liver
C. Kidney
D. Upper respiratory tract
E. Heart

Ans A c. Kidney, d. Upper respiratory tract

197. **Severe eosinophilia, defined as an absolute eosinophil count (AEC) more than**

A. 750 cells/μL
B. 1,000 cells/μL
C. 1,500 cells/μL
D. **5,000 cells/μL**
E. 7,500 cells/μL

Ans D Moderate (AEC 1,500-5,000 cells/μL) and severe (AEC >5,000 cells/μL).

198. **A 13-month-old girl presented with failure to thrive, repeated chest infection, and fatty offensive diarrhea. Hemoglobin level 8.8gm/dl, platelate count 150/mm3, white blood cell count 3.500/mm3, with absolute neutrophil count (ANC) 500/μL. Of the following, the MOST likely diagnosis is**

A. Kostmann disease
B. cyclic neutropenia
C. drug-induced neutropenia
D. acute lymphoblastic leukemia
E. **Shwachman-Diamond syndrome**

Ans E (SDS) is an autosomal recessive disorder classically characterized by neutropenia, pancreatic insufficiency, and short stature with skeletal abnormalities. The initial symptoms are usually steatorrhea and failure to thrive because of malabsorption, which usually develops by 4 mo of age, although the gastrointestinal symptoms may be subtle in some patients and go unrecognized. Virtually all patients with SDS have neutropenia, with the ANC periodically <1000/μL.

199. **The recognition of allergic rhinitis as a major chronic respiratory disease of children rests largely on all the following features EXCEPT**

A. high prevalence
B. effect on quality of life
C. school performance
D. other comorbidities
E. **presentation in infancy**

Ans E Recognition of allergic rhinitis as a major chronic respiratory disease of children rests largely on its high prevalence, detrimental effects on quality of life and school performance, and comorbidities. The prevalence peaks late in childhood.

200. **The exposure to which of the following early in childhood protects against the development of atopy**

A. smoking
B. alcohol
C. rodents
D. **dogs**
E. indoor allergen

Ans D The exposure to dogs, cats and endotoxin early in childhood protects against the development of atopy.

201. **The MOST effective therapy for persistent allergic rhinitis is**

A. oral desloratadine
B. oral montelukast
C. subcutaneous omalizumab
D. **nasal budesonide spray**
E. nasal ipratropium bromide spray

Ans D Patients with more persistent, severe symptoms require intranasal corticosteroids, the most effective therapy for allergic rhinitis, a treatment that may be beneficial also for concomitant allergic conjunctivitis.

202. **The MOST vital initial treatment in the management of severe asthma exacerbations is**

A. **supplemental oxygen**
B. inhaled β -agonist
C. intramuscular injection of epinephrine
D. inhaled ipratropium
E. intramuscular injection of β-agonist

Ans A is the best to start with.

203. **In emergency department, the patient may be discharged to home if there is symptomatic improvement, normal physical findings, PEF >70% of predicted or personal best, and oxygen saturation >92% in room air for 4hr. Of the following, the MOST likely discharge medication used is**

A. inhaled β-agonist only
B. oral corticosteroid only
C. inhaled corticosteroid only
D. **inhaled β-agonist plus oral corticosteroid**
E. oral β-agonist plus inhaled corticosteroid

Ans D Discharge medications include administration of an inhaled β-agonist up to every 3-4 hr plus a 3-7 day course of an oral corticosteroid.

204. **The cardinal feature of atopic dermatitis is**

A. skin rash
B. lichenification
C. fibrotic papules
D. **intense pruritus**
E. dry skin

Ans D especially at night, and cutaneous reactivity are the cardinal features of atopic dermatitis.

205. **Predictive factors of a poor prognosis for atopic dermatitis includes all the following**

A. widespread atopic dermatitis in childhood
B. filaggrin gene null mutations
C. concomitant allergic rhinitis and asthma
D. family history of atopic dermatitis in parents or siblings
E. **late age at onset of atopic dermatitis**

Ans E

206. **Exfoliative dermatitis may develop in patients with atopic dermatitis and extensive skin involvement, usually caused by inappropriate therapy or superinfection with**

A.
B. Herpes zoster
C. **Herpes simplex**
D. Streptococcal pyogen
E. Trichophyton rubrum

Ans C Exfoliative dermatitis associated with generalized redness, scaling, weeping, crusting, systemic toxicity, lymphadenopathy, and fever, and is usually caused by superinfection (e.g., with toxin-producing S. aureus or HSV infection) or inappropriate therapy. In some cases, the withdrawal of systemic glucocorticoids used to control severe AD precipitates exfoliative erythroderma.

207. **Anaphylactic reactions after a Hymenoptera sting are treated exactly like anaphylaxis from any cause. Of the following, the drug of choice is**

A. oxygen
B. **epinephrine**
C. antihistamines
D. corticosteroids
E. intravenous fluids

Ans B Adjunctive treatment includes antihistamines, corticosteroids, intravenous fluids, oxygen, and transport to the emergency room.

208. **The MOST common hypersensitivity response of the eye is**

A. **allergic conjunctivitis**
B. vernal keratoconjunctivitis
C. atopic keratoconjunctivitis
D. giant papillary conjunctivitis
E. contact allergy

Ans A is the most common hypersensitivity response of the eye, affecting approximately 25% of the general population and 30% of children with atopy.

209. **The MOST sight-threatening type of ocular allergy is**

A. contact allergy
B. **vernal keratoconjunctivitis**
C. giant papillary conjunctivitis
D. seasonal allergic conjunctivitis
E. perennial allergic conjunctivitis

Ans B and atopic keratoconjunctivitis are sight-threatening entities and causes visual morbidity and suspected cases need to be referred to an ophthalmologist.

210. **All the following factors can amplify anaphylaxis EXCEPT**

A. exercise
B. **vaccination**
C. upper respiratory tract infection
D. fever
E. emotional stress

Ans B is a trigger, not a cofactor in anaphylaxis, others are cofactors.

211. **One of the principal pathologic features in fatal anaphylaxis is**

A. **acute bronchial obstruction**
B. hypotension
C. behavioral change
D. abdominal pain
E. vomiting

Ans A Principal pathologic features in fatal anaphylaxis include acute bronchial obstruction with pulmonary hyperinflation, pulmonary edema, intraalveolar hemorrhaging, visceral congestion, laryngeal edema, and urticaria and angioedema. Acute hypotension is attributed to vasomotor dilation and/or cardiac dysrhythmias.

212. **Cutaneous symptoms may be absent in anaphylaxis in**

A. 5%
B. 10%
C. **20%**
D. 30%
E. 40%

Ans C

213. **The best diagnostic test for anaphylaxis in a child exposed to an allergen 3 hours before arrival to hospital is**

A. plasma histamine
B. skin test
C. radioallergosorbent assay
D. **plasma tryptase**
E. immunoCAP IgE test

Ans D Plasma histamine is elevated for a brief period but is unstable and difficult to measure in a clinical setting. Plasma tryptase is more stable and remains elevated for several hours but often is not elevated, especially in food-induced anaphylactic reactions.

214. **After institution of treatment of anaphylaxis, the patient should be monitored in the emergency room for at least**

A. 1 hour
B. **4 hours**
C. 6 hours
D. 12 hours
E. 24 hours

Ans B Patients may experience biphasic anaphylaxis, which occurs when anaphylactic symptoms recur after apparent resolution. More than 90% of biphasic responses occur within 4 hours, so patients should be observed for at least 4 hr before being discharged from the emergency department.

215. **The onset of presentation of food allergy is late in**

A. hen's egg white
B. peanuts
C. **shellfish**
D. soybean
E. cow's milk

Ans C The onset is usually during adulthood. Others are earlier in presentation.

08

CHAPTER 08

Infectious Diseases



WHAT THIS CHAPTER COVERS

- Viral Exanthems (Measles, Mumps, Rubella)
- Chickenpox & Herpes Zoster
- Dengue & Arboviral Infections
- Tuberculosis
- Typhoid & Enteric Fever
- Malaria
- Leishmaniasis & Kala Azar
- Meningitis
- Pneumonia
- UTI & Pyelonephritis
- Gastroenteritis & Diarrhea
- Hepatitis
- HIV & AIDS
- Polio & Enteroviruses
- Diphtheria, Pertussis & Tetanus
- Streptococcal & Staphylococcal Infections
- Sepsis & Septicemia
- Congenital & Neonatal Infections
- Fungal Infections
- Parasitic & Helminthic Infections
- Antibiotics & Antimicrobial Therapy
- Fever & Fever Management

292 MCQs **24** subtopics

8.1 Fever with Rash – Viral Exanthems

Disease	Agent	Incubation	Rash – onset & features	Hallmarks / complications
Measles (rubeola) – "1st disease"	Paramyxovirus (Morbillivirus)	**10–14 days**	Day **4** of fever; **maculopapular, starts behind ears/hairline → face → trunk → limbs (centrifugal, downward)**; becomes confluent; fades with **brownish staining & branny desquamation** (sparing palms/soles)	Prodrome **3 Cs – cough, coryza, conjunctivitis** + high fever; **Koplik spots** (buccal mucosa opposite lower molars, pathognomonic, appear 1–2 days before rash); **pneumonia = MCC death**; diarrhoea, otitis media (MC complication), **giant-cell (Hecht) pneumonia**, encephalitis, **SSPE (7–10 y later)**, **TB flare (tuberculin anergy)**, malnutrition, **vitamin A deficiency (xerophthalmia)**
Rubella (German measles) – "3rd disease"	Togavirus	**14–21 days**	Day 1–2; pink maculopapular, face → body in 24 h, **gone by day 3 ("3-day measles")**, no staining	**Postauricular, suboccipital & posterior cervical lymphadenopathy**; **Forchheimer spots** (soft-palate petechiae); arthralgia in adolescent girls; **congenital rubella syndrome**
Erythema infectiosum – "5th disease"	**Parvovirus B19**	4–14 days	**"Slapped-cheek"** facial erythema → **lacy reticular rash** on extremities, recurs with heat/sun	**Aplastic crisis in sickle cell/ spherocytosis** (P antigen on RBC precursors), **hydrops fetalis** in pregnancy, arthropathy, papular-purpuric gloves-and-socks
Roseola infantum (exanthem subitum) – "6th disease"	**HHV-6 (HHV-7)**	9–10 days	**High fever 3–5 days in infant (6 m–2 y); rash appears as fever DISAPPEARS** – rose-pink macules on trunk	**Febrile seizures** (MC cause in < 2 y viral), bulging fontanelle; **Nagayama spots** (uvulopalatoglossal ulcers)
Scarlet fever – "2nd disease"	**Group A Streptococcus (erythrogenic/ pyrogenic toxin)**	2–4 days	Day 1–2; **sandpaper (goose-skin) rash**, starts on neck/axilla → trunk, **Pastia lines** (in skin folds), **circumoral pallor**, flushed cheeks; **desquamation of palms & soles** in 2nd week	**White → red strawberry tongue**, exudative pharyngitis; rheumatic fever, AGN; Rx **penicillin**
Varicella (chickenpox)	VZV (HHV-3)	**14–16 (10–21) days**	Day 1; **centripetal (trunk > limbs)**, **pleomorphic – macule → papule → vesicle ("dewdrop on rose petal") → crust in all stages at the same time**; itchy; involves scalp & mucosa	Secondary **bacterial skin infection (MC complication – S. aureus/GAS)**, pneumonia (adults), **cerebellar ataxia (MC neuro complication, benign)**, encephalitis, **Reye syndrome (aspirin)**, purpura fulminans, congenital/neonatal varicella
Hand-foot-mouth disease	**Coxsackie A16, Enterovirus 71**	3–6 days	Oval vesicles on **palms, soles, buttocks** + **oral ulcers**	EV-71: **brainstem encephalitis**, pulmonary oedema; **onychomadesis** later
Herpangina	Coxsackie A		**Vesicles/ulcers on posterior pharynx, soft palate, tonsillar pillars**	Fever, sore throat
Dengue	Flavivirus	4–7 days	Flushing early; **convalescent "islands of white in a sea of red"** rash	See below

Disease	Agent	Incubation	Rash – onset & features	Hallmarks / complications
Infectious mononucleosis	EBV	30–50 days	**Maculopapular rash after ampicillin/amoxicillin (~90%)**	Fever, exudative pharyngitis, **posterior cervical LN**, splenomegaly (avoid contact sports – rupture), atypical lymphocytes (Downey cells), **heterophile (Paul–Bunnell/Monospot) antibodies** (often negative < 4 y – do VCA-IgM)

NUMBERED EXANTHEMS – 'NEVER SHALL RUBY DUST FIND ROSY'

1 **M**easles · 2 **S**carlet fever · 3 **R**ubella · 4 **D**ukes' (filatov–Dukes, staph scalded skin – obsolete) · 5 **F**ifth (parvovirus B19) · 6 **R**oseola (HHV-6).

RASH APPEARS WITH DEFERVESCENCE

Roseola (rash as fever falls) vs **measles** (rash at peak fever, day 4) vs **dengue** (rash in convalescence) vs **Kawasaki** (fever ≥ 5 d persists with rash).

Measles – key management & prevention

- Isolation **4 days before to 4 days after rash onset**; infectivity highest in **prodromal (catarrhal) stage**.
- **Vitamin A** to all cases: < 6 m 50,000 IU, 6–11 m 1 lakh, ≥ 12 m 2 lakh IU – day 1 & 2 (third dose after 2–4 weeks if eye signs).
- **Post-exposure**: **measles vaccine within 72 h**; **immunoglobulin within 6 days** for infants < 6 m, pregnant & immunocompromised.
- **SSPE**: persistent defective measles virus; years after measles (often < 2 y at infection); **behaviour change → myoclonic jerks → dementia → death**; EEG **periodic complexes (Radermecker)**, **↑ CSF anti-measles antibody (diagnostic)**; Rx isoprinosine (inosiplex) – limited.

Varicella – management

- **Acyclovir** 20 mg/kg (max 800 mg) **4–5 times/day for 5 days** for children > 12 y, chronic skin/lung disease, on steroids/salicylates, secondary household cases; **IV acyclovir 10 mg/kg 8-hourly** in immunocompromised/ neonates/complicated disease.
- Infectious from **1–2 days before rash until all lesions crusted** (~5 days).
- **VZIG** (within **10 days**, ideally 96 h) for: immunocompromised, pregnant women, **newborn whose mother develops varicella 5 days before to 2 days after delivery** (neonatal varicella – severe, 30% mortality untreated), premature infants.
- **Congenital varicella syndrome** (maternal infection **8–20 wk**): **cicatricial skin scars (dermatomal)**, **limb hypoplasia**, eye defects (chorioretinitis, cataract), microcephaly, cortical atrophy.
- **Herpes zoster** in children: after intrauterine/early infancy varicella; milder.

8.2 Congenital (TORCH) Infections

Infection	Classic findings
Toxoplasmosis	**Classic triad – chorioretinitis, hydrocephalus, diffuse (scattered) intracranial calcifications**; seizures; risk of transmission ↑ with gestation, severity ↓; Rx **pyrimethamine + sulfadiazine + folinic acid** (spiramycin in pregnancy)
Rubella (CRS)	**Gregg's triad – cataract (pearly nuclear), sensorineural deafness (MC single defect), PDA / peripheral pulmonary artery stenosis**; "blueberry muffin" rash (extramedullary haematopoiesis), microcephaly, IUGR, **salt-and-pepper retinopathy** (MC eye finding), hepatosplenomegaly, thrombocytopenia, **celery-stalk metaphyses**, late – diabetes, thyroid disease, PRP. Risk **highest in 1st trimester (up to 90% < 11 wk)**; infant sheds virus for up to 1 year

Infection	Classic findings
CMV (MC congenital infection)	**MC cause of non-genetic SNHL**; **periventricular calcification**, microcephaly, chorioretinitis, hepatosplenomegaly, petechiae (thrombocytopenia), IUGR, **blueberry muffin**; 90% asymptomatic at birth (10–15% develop SNHL later); Dx **urine/saliva CMV PCR within 3 weeks**; Rx **valganciclovir 6 months** (symptomatic)
HSV (neonatal)	Mostly **perinatal (85%)**, HSV-2; 3 forms – **SEM (skin-eye-mouth)**, **CNS (temporal lobe; day 14–21)**, **disseminated (liver failure, DIC; day 7–14)**; Rx **IV acyclovir 20 mg/kg 8-hourly – 14 days (SEM) / 21 days (CNS, disseminated)**
Congenital syphilis	**Early (< 2 y)**: **snuffles** (rhinitis – highly infectious), maculopapular/bullous rash on **palms & soles (desquamation)**, hepatosplenomegaly, **osteochondritis & periostitis → Parrot's pseudoparalysis**, **Wimberger sign**, condylomata lata. **Late (> 2 y)**: **Hutchinson's triad – Hutchinson teeth, interstitial keratitis (5–20 y), 8th nerve deafness**; **saddle nose**, **saber shins**, **Clutton's joints (painless knee effusion)**, **frontal bossing (Olympian brow)**, **rhagades**, mulberry molars, Higoumenakis sign. Rx **aqueous crystalline penicillin G 50,000 U/kg IV 12-hourly × 7 d then 8-hourly to complete 10 days**
Zika	**Severe microcephaly** with collapsed skull, subcortical calcifications, arthrogryposis
Varicella	See above
Parvovirus B19	**Non-immune hydrops fetalis** (anaemia), myocarditis
HIV / Hepatitis B	Perinatal; see below

INTRACRANIAL CALCIFICATION PATTERN

CMV → Periventricular ("**C**MV = **C**ircles the ventricles"); **Toxoplasma → diffuse/scattered (cortical & basal ganglia)**; **Sturge–Weber → tram-track gyral**; **TSC → subependymal nodules**; Zika – subcortical.

8.3 Dengue (WHO 2009 / NVBDCP)

- **Aedes aegypti** (day-biting, breeds in clean stored water); 4 serotypes (DENV 1–4); **secondary infection with a different serotype → severe dengue (antibody-dependent enhancement)**.
- **Phases**: febrile (2–7 d) → **critical phase (around defervescence, day 3–7) – plasma leakage** → recovery.

Category	Features
Dengue without warning signs	Fever + 2 of: nausea/vomiting, rash, aches & pains, **positive tourniquet test**, leucopenia
Warning signs	**Abdominal pain/tenderness**, **persistent vomiting**, **clinical fluid accumulation (ascites, pleural effusion)**, **mucosal bleed**, **lethargy/restlessness**, **liver enlargement > 2 cm**, **rising haematocrit with rapid fall in platelets**
Severe dengue	**Severe plasma leakage → shock (DSS)/fluid accumulation with respiratory distress**; **severe bleeding**; **severe organ involvement (AST/ALT ≥ 1000, impaired consciousness, myocarditis)**

- **Tourniquet test**: BP cuff at midpoint of SBP & DBP for **5 min**; **≥ 10 petechiae/sq inch** = positive (≥ 20 in older definitions).
- Diagnosis: **NS1 antigen** (day 1–5, best early test), RT-PCR (day 1–5); **IgM ELISA from day 5** (MAC-ELISA); IgG ↑↑ in secondary.
- **Haematocrit rise ≥ 20%** = plasma leakage; thrombocytopenia ≤ 1 lakh.
- **Shock**: narrow pulse pressure (**≤ 20 mmHg**) is early sign; hypotension late.

DENGUE FLUID MANAGEMENT

- Without warning signs: oral fluids, **paracetamol only** (no NSAIDs/aspirin, no IM injections).
- Warning signs: **crystalloid (NS/RL) 5–7 mL/kg/h for 1–2 h**, then 3–5 then 2–3 mL/kg/h guided by haematocrit.
- **Compensated shock**: **10–20 mL/kg crystalloid over 1 h**; if improves taper (10 → 7 → 5 → 3 mL/kg/h) over 24–48 h; if not – repeat bolus/colloid; **rising Hct → colloid; falling Hct with instability → suspect bleeding → blood transfusion (10 mL/kg PRBC / 20 mL/kg whole blood)**.
- **Hypotensive shock**: 20 mL/kg bolus over 15–30 min.
- **Platelet transfusion** only if **< 10,000/µL** with bleeding or significant bleeding (not prophylactically).
- Watch for **fluid overload in recovery** (reabsorption).

- **Chikungunya** (alphavirus, Aedes): **severe polyarthralgia (stooped posture)**, rash, **nose hyperpigmentation (chik sign)**; neonatal encephalopathy; arthritis may persist months.
- **Japanese encephalitis** (Culex, pigs = amplifier host, ardeid birds = reservoir): children 1–15 y, **thalamic & basal ganglia lesions**, Parkinsonian sequelae; vaccine SA-14-14-2 / JENVAC in UIP endemic districts.
- **Zika**: Aedes; mild febrile illness with conjunctivitis; fetal microcephaly; **Guillain–Barré**.

8.4 Malaria in Children

- **P. vivax** most common overall in India; **P. falciparum** causes severe malaria (cerebral malaria, severe anaemia – **MC severe manifestation in young children**).
- **Severe falciparum malaria (WHO)**: impaired consciousness (cerebral malaria – Blantyre coma score ≤ 2), **prostration**, multiple convulsions (> 2 in 24 h), **acidosis**, **hypoglycaemia (< 40 mg/dL)**, **severe anaemia (Hb < 5 g/dL)**, AKI, jaundice, pulmonary oedema/ARDS, bleeding, shock, **hyperparasitaemia (> 10%)**.
- Dx: **thick smear (detection – most sensitive)**, thin smear (species, parasite density); **RDT (HRP-2 for Pf; pLDH for pan)**.

NATIONAL (NVBDCP 2013+) TREATMENT

- **P. vivax**: **chloroquine 25 mg/kg over 3 days (10, 10, 5)** + **primaquine 0.25 mg/kg/day × 14 days** (radical cure; **check G6PD; contraindicated < 1 year & pregnancy**).
- **Uncomplicated P. falciparum**: **ACT – artesunate + sulfadoxine-pyrimethamine (AS+SP) × 3 days** (**artemether–lumefantrine in north-eastern states**) + **single-dose primaquine 0.75 mg/kg on day 2** (gametocidal; not < 1 y/pregnancy).
- **Severe malaria**: **IV/IM artesunate 2.4 mg/kg (3 mg/kg if < 20 kg) at 0, 12, 24 h then daily** until oral possible, then full ACT course; alternative – **quinine** (watch hypoglycaemia); artemether IM, arteether.
- Pregnancy 1st trimester Pf: quinine (ACT now acceptable per WHO 2022).

- **Kala-azar (visceral leishmaniasis)** – **L. donovani**, **sandfly (Phlebotomus argentipes)**; Bihar, Jharkhand, WB, UP; **fever with massive splenomegaly**, hepatomegaly, **darkening of skin**, pancytopenia, **hypergammaglobulinaemia** (aldehyde test historic); Dx **rK39 RDT**, splenic aspirate (most sensitive, LD bodies); Rx **single-dose liposomal amphotericin B 10 mg/kg (DOC, India)**, miltefosine + paromomycin; **PKDL** after treatment.

8.5 Typhoid (Enteric) Fever

- **Salmonella** Typhi / Paratyphi A, B; faeco-oral; incubation **7–14 days**.
- **Week 1**: step-ladder fever, headache, abdominal pain, constipation/diarrhoea, **relative bradycardia** (less common in children), coated tongue; **rose spots** (trunk, 2nd week); hepatosplenomegaly.
- **Complications (week 2–3)**: **intestinal perforation (terminal ileum – Peyer's patches)** & haemorrhage, encephalopathy, myocarditis, hepatitis, cholecystitis, **chronic carriage (gallbladder)**.

- Investigations by week – "**BASU**": **B**lood culture (week 1 – **IOC**; 40–80% positive), **A**ntibodies (Widal – week 2; **≥ 1:160 O & H** single titre in endemic areas, 4-fold rise), **S**tool culture (week 3), **U**rine culture (week 4). **Bone marrow culture = most sensitive (unaffected by prior antibiotics)**. Typhidot (IgM).
- Leucopenia/relative lymphocytosis, eosinopenia.

TYPHOID TREATMENT (IAP 2022 / ICMR)

- **Uncomplicated (OPD)**: **oral cefixime 20 mg/kg/day × 14 days** or **azithromycin 10–20 mg/kg/day × 7 days**.
- **Complicated / unable to take orally**: **IV ceftriaxone 75–100 mg/kg/day × 14 days**; add **azithromycin** if slow response; **meropenem** for suspected XDR (Pakistan strains).
- **Fluoroquinolones not first-line** (widespread resistance in India).
- **Dexamethasone** (3 mg/kg then 1 mg/kg 6-hourly × 48 h) in severe typhoid with shock/encephalopathy.
- Defervescence takes **4–5 days** with effective therapy.

8.6 Tuberculosis in Children (NTEP 2022)

- Most children have **primary (pulmonary) TB** with paucibacillary disease; **contact history** crucial; household contacts of sputum+ adult.
- **Primary complex (Ghon complex)**: Ghon focus (subpleural, mid-zone) + lymphangitis + **hilar lymphadenopathy** (Ranke complex when calcified).
- **Extrapulmonary**: **lymph node TB (MC EPTB – cervical)**, **TB meningitis** (most serious, < 5 y), **miliary TB**, spine (Pott's – thoracolumbar), bone & joint, abdominal, pericardial.

Diagnosis

- Specimens: **gastric aspirate (early morning, 2 consecutive days)**, induced sputum, nasopharyngeal aspirate, BAL, stool (Ultra).
- **CBNAAT/Xpert MTB/RIF or Truenat – first test of choice (NTEP)** – detects MTB & rifampicin resistance in 2 h.
- **Mantoux (TST)**: **5 TU PPD (or 2 TU RT-23)** intradermal, read at **48–72 h**; **≥ 10 mm induration = positive** (≥ 5 mm in HIV, SAM, immunosuppressed). **False negative**: malnutrition, **measles**, HIV, steroids, miliary TB, TBM, recent live vaccine. IGRA – alternative (> 5 y).
- CXR: hilar/paratracheal LN (most common finding), consolidation, miliary mottling, effusion.
- TB meningitis: CSF – lymphocytic pleocytosis, **↑ protein, ↓ glucose**, cobweb coagulum; MRI – **basal exudates, hydrocephalus, infarcts, tuberculomas**.

PAEDIATRIC ANTI-TB TREATMENT (NTEP – WEIGHT BANDS, DAILY FDC)

- **Drug-sensitive TB**: **2 HRZE + 4 HRE** (6 months) for pulmonary & most EPTB.
- **TB meningitis / osteoarticular / CNS TB**: **2 HRZE + 10 HRE (12 months total)** + **steroids** (dexamethasone/ prednisolone **4–8 weeks** in TBM, also pericardial).
- Doses: **H 10 (7–15) mg/kg, R 15 (10–20) mg/kg, Z 35 (30–40) mg/kg, E 20 (15–25) mg/kg** daily.
- **Pyridoxine** with INH in malnourished/HIV/adolescents.
- **TB preventive therapy (TPT)** for household contacts **< 5 y (after ruling out active TB)**, HIV+: **6 months INH (6H)** or **3HP (rifapentine + INH weekly × 12, ≥ 2 y)** / 4R.
- **Neonate of mother with TB**: if mother sputum+ → rule out congenital TB, give **INH prophylaxis 6 months**, then BCG (if not given); **continue breastfeeding**.
- DR-TB: shorter all-oral bedaquiline-containing regimens; **BPaLM** for ≥ 14 y.

- **Congenital TB** (Cantwell criteria): primary hepatic complex/caseating hepatic granulomas, lesions in first week, maternal genital/placental TB.
- **BCG lymphadenitis** – non-suppurative: observe; suppurative: needle aspiration.

8.7 Bacterial Meningitis (also see Neurology)

Age	Common organisms (India)
Neonate (< 1 m)	**E. coli, Klebsiella**, GBS (worldwide MC), Listeria
1–3 months	Neonatal organisms + pneumococcus, Hib
> 3 months	**S. pneumoniae (MC)**, **N. meningitidis**, Hib (↓ after vaccine)

- Features: fever, headache, vomiting, neck rigidity (unreliable < 18 m), **Kernig & Brudzinski**, bulging fontanelle, seizures, altered sensorium; **meningococcaemia – petechial/purpuric rash, Waterhouse–Friderichsen (adrenal haemorrhage)**.
- CSF: **↑ polymorphs, ↑ protein, ↓ glucose (CSF/blood < 0.4)**; Gram stain, culture, latex agglutination, PCR.
- Empirical Rx (> 1 m): **ceftriaxone 100 mg/kg/day** (or cefotaxime) **± vancomycin** (resistant pneumococcus); duration – **meningococcus 7 d, Hib 7–10 d, pneumococcus 10–14 d, GBS 14–21 d, gram-negative 21 d**.
- **Dexamethasone 0.15 mg/kg 6-hourly × 2–4 days, given before/with first antibiotic dose** – ↓ **hearing loss in Hib** meningitis.
- **Most common sequela: sensorineural hearing loss** (do BERA before discharge). Subdural effusion (MC in Hib – benign, usually resolves).
- **Chemoprophylaxis** of contacts: meningococcus – **rifampicin 10 mg/kg 12-hourly × 2 days** (or single-dose ciprofloxacin/ceftriaxone); Hib – rifampicin 20 mg/kg/day × 4 days.

8.8 Diphtheria, Pertussis & Tetanus

	Diphtheria	Pertussis (whooping cough)	Tetanus (neonatal)
Agent	**Corynebacterium diphtheriae** (exotoxin – **phage-encoded**, inhibits EF-2)	**Bordetella pertussis**	**Clostridium tetani** (tetanospasmin – blocks GABA/glycine release)
Features	**Faucial – greyish-white adherent pseudomembrane, bleeds on removal**, **bull neck** (cervical LN + oedema), low-grade fever; **laryngeal → stridor**; nasal – serosanguinous discharge	**Catarrhal (1–2 wk, most infectious) → paroxysmal (2–6 wk): inspiratory whoop, post-tussive vomiting, cyanosis; apnoea in young infants (no whoop) → convalescent ("100-day cough")**	**Day 3–14**: baby stops sucking (**trismus**, earliest), **risus sardonicus**, generalised spasms, **opisthotonus**, **conscious** child; unclean cord cutting
Complications	**Myocarditis (2nd week – MCC death)**, **neuritis (palatal palsy – 3rd week, nasal regurgitation)**, airway obstruction	**Pneumonia (MCC death)**, apnoea, seizures, **subconjunctival haemorrhage**, rectal prolapse, hernia, **bronchiectasis**	Aspiration, laryngospasm, autonomic instability
Lab	**Albert stain (metachromatic granules), Loeffler serum slope, tellurite agar**; Elek's gel precipitation (toxigenicity)	**Lymphocytosis (absolute, > 15,000) – marked**; PCR of NP swab; **Bordet–Gengou medium**	Clinical diagnosis
Rx	**Antidiphtheritic serum (ADS) – immediately on suspicion** (after test dose; 20,000–1,00,000 U by severity) **+ penicillin or erythromycin × 14 days**	**Azithromycin (DOC) 10 mg/kg × 5 days** (erythromycin – **infantile hypertrophic pyloric stenosis** in < 1 m); contacts – same	**TIG (human 500 IU IM)**, metronidazole (DOC antibiotic), **diazepam/midazolam** for spasms, $MgSO_4$, quiet dark room, NG feeds; active immunisation after recovery (disease doesn't confer immunity)

	Diphtheria	Pertussis (whooping cough)	Tetanus (neonatal)
Prevention	DPT; carriers – erythromycin	DTaP/DTwP; **Tdap in pregnancy (27–36 wk)**	**Td to pregnant women (2 doses / booster)**, clean delivery practices ("6 cleans") – **India eliminated MNT (2015)**

8.9 HIV in Children

- **MC mode: vertical (mother-to-child) transmission** – in utero, **intrapartum (MC)**, breastfeeding. Risk without intervention 25–45%; with ART **< 2%**.
- **Diagnosis in infants < 18 months: HIV DNA PCR (NAT)** (maternal antibodies persist up to 18 m) – at **6 weeks**, repeat at 6 months, 12 months and 6 weeks after stopping breastfeeding; antibody tests at 18 months.
- **PPTCT (NACO)**: **lifelong ART (TLD) to all pregnant/breastfeeding women** regardless of CD4; infant **nevirapine prophylaxis for 6 weeks** (high-risk infant – mother on ART < 8–12 weeks or high viral load – **dual AZT + NVP, extended up to 12 weeks**); **exclusive breastfeeding for 6 months**; **co-trimoxazole prophylaxis from 6 weeks** until HIV ruled out.
- **Treat all HIV-infected children** irrespective of CD4/clinical stage. First-line (NACO 2021): **< 20 kg/< 10 y – ABC + 3TC + DTG (or LPV/r)**; ≥ 20 kg – **TLD (TDF + 3TC + DTG)**.
- Clinical clues: **FTT, recurrent pneumonia, persistent oral candidiasis, parotid enlargement, generalised lymphadenopathy, hepatosplenomegaly, chronic diarrhoea, LIP (lymphoid interstitial pneumonitis – clubbing, reticulonodular shadows, good prognosis)**, **PCP pneumonia (3–6 m, MC AIDS-defining illness in infants – Rx high-dose co-trimoxazole + steroids)**, encephalopathy (microcephaly, developmental regression, basal ganglia calcification).

8.10 Hepatitis & Other Notable Infections

- **Hepatitis A** – MC cause of acute viral hepatitis in children (faeco-oral); mostly anicteric < 6 y; **acute liver failure** (MCC in Indian children); IgM anti-HAV; vaccine (live attenuated – single dose).
- **Hepatitis E** – faeco-oral, epidemics, fulminant in pregnancy.
- **Hepatitis B** – vertical (HBeAg+ mother → **90% chronicity**); **birth-dose vaccine within 24 h + HBIG (0.5 mL within 12 h) for HBsAg+ mother's baby**; tenofovir to HBeAg+/high viral load mothers in 3rd trimester.
- **Infectious mononucleosis** – see above; **X-linked lymphoproliferative** – fatal EBV.
- **Mumps** – paramyxovirus; parotitis (bilateral 70%), **aseptic meningitis (MC extrasalivary, often without parotitis)**, **orchitis (post-pubertal; sterility rare)**, pancreatitis, **SNHL (unilateral – MCC acquired unilateral deafness in children)**, oophoritis; ↑ serum amylase.
- **Scrub typhus** (**Orientia tsutsugamushi**, trombiculid mite larvae): fever, **eschar** (painless, black, groin/axilla), lymphadenopathy, hepatosplenomegaly, pneumonitis, myocarditis, meningoencephalitis, AKI, shock; Dx IgM ELISA/Weil-Felix (OX-K); Rx **doxycycline (all ages, 5–7 d)** or azithromycin.
- **Leptospirosis** – rat urine, flooding; conjunctival suffusion, calf tenderness, jaundice + AKI (Weil's); Rx penicillin/ doxycycline.
- **Brucellosis** – raw milk; undulant fever, arthritis (sacroiliitis); Rx doxycycline + rifampicin (TMP-SMX + rifampicin < 8 y).
- **Rabies** – see Immunization chapter.
- **COVID-19 in children** – mostly mild; MIS-C (see Immunology).

8.11 Parasitic & Fungal

Infection	Key points
Ascariasis	MC helminth; Loeffler syndrome (pulmonary eosinophilia), intestinal obstruction (bolus), biliary ascariasis; Rx **albendazole 400 mg single dose (200 mg for 1–2 y)**; obstruction – conservative + piperazine (paralyses worm)
Hookworm	IDA, hypoproteinaemia; ground itch
Enterobius (pinworm)	**Perianal itch at night**, vulvovaginitis; **cellophane (scotch) tape test** in morning; Rx albendazole, repeat after 2 weeks, treat family
Giardia	Steatorrhoea, malabsorption, FTT; IgA deficiency; Rx metronidazole/tinidazole/nitazoxanide
Amoebiasis	Dysentery, **liver abscess (right lobe, anchovy-sauce pus)**; Rx metronidazole + luminal agent (diloxanide/paromomycin)
Neurocysticercosis	**MC cause of focal seizures in Indian children**; **single small enhancing CT lesion (ring/disc, < 2 cm)**; Rx **albendazole 15 mg/kg/day × 7–28 days + steroids** (not for calcified lesions; check eyes first) + anticonvulsants
Hydatid cyst	Liver > lung; Rx albendazole + PAIR/surgery
Toxocara (visceral larva migrans)	Pica, dog contact – eosinophilia, hepatomegaly; ocular – leukocoria mimicking retinoblastoma
Candida	Oral thrush (nystatin), diaper candidiasis (satellite lesions – clotrimazole); neonatal invasive candidiasis (VLBW) – amphotericin/fluconazole

Practice MCQs · Infectious Diseases

292 questions · correct option highlighted in green · cover the answer strip to self-test

Viral Exanthems (Measles, Mumps, Rubella)

1. The following age group is most severely affected by rubella infection:

A. Females aged 25–35 year
B. Young girls
C. Adolescent girls
D. Unborn child

Ans D • The pathologic findings for Congenital Rubella syndrome are often severe and may involve nearly every organ system
• Postnatal infection with rubella is a mild disease not easily discernible from other viral infections, especially in children
• Complications following postnatal infection with rubella are infrequent and generally not lifethreatening.

2. Which of the following is not a common manifestation of congenital rubella?

A. Deafness
B. PDA
C. Aortic stenosis
D. Mental retardation VARICELLA

Ans C Aortic stenosis VARICELLA

3. Which statement regarding measles is false?

A. Koplik's spots cannot be seen in conjunctival or vaginal mucosa
B. Vitamin A administration is part of management of clinical measles
C. CD 150 and PVRL4 are receptors for measles virus
D. Pneumonia is the common cause of death in children with measles

Ans A Measles
• Prodromal phase:fever, conjunctivitis, coryza
• Koplik spots on inner cheeks at level of 1st& 2nd molars
• Rash begins behind the ears & spreads downwards
• Rash fades in 7 days in the same progression as it evolved
Koplik spots
• Are the pathognomonic sign of measles
• Appear 1-4 days prior to the onset of the rash
• First appears as discrete red lesions with bluish white spots in the center on the inner aspects of the cheeks at the level of premolars.

4. Most common complication of measles is:

A. Orchitis
B. Pneumonia
C. Otitis media
D. Seizures

Ans C Acute otitis media is the most common complication of measles.

5. Incubation period of measles is:

A. 1-4 days
B. 5-7 days
C. 10-14 days
D. 15-20 days

Ans C 1014 days Incubation period of Measles is 8-12 days.

6. Which is not a complication of measles?

A. Cataract
B. Encephalitis
C. Pancreatitis
D. Meningitis

Ans A Acute otitis media, Croup, tracheitis, bronchiolitis, sinusitis, mastoiditis, encephalitis & meningitis are common complications in infants & toddlers with measles; Pneumonia is the most common cause of death in measles.

7. True about measles:

A. Rash appear first on leg
B. Koplik spots are seen in retina
C. Long term complications follow in form of SSPE
D. Incubation period is 2–3 days
E. Caused by RNA virus

Ans C and e. Caused by RNA virus Measles virus is a single-stranded, lipid-enveloped RNA virus in the family Paramyxoviridae and genus Morbillivirus.

8. True about measles:

A. Koplik spots appear in prodromal stage
B. Fever stops after the onset of rash
C. Vaccine given at 9 months
D. It is not diagnosed when coryza and rhinitis is absent.
E. It is not infective before the rash appears

Ans A b. Fever stops after the onset of rash & c. Vaccine given at 9 months Measles consists of 4 phases: incubation period, prodromal illness, exanthematous phase, and recovery.
• Incubation period usually lasts for 8-12 days.
• Prodromal phase: mild fever followed by conjunctivitis, coryza, fever & Koplik spots (in 50- 70%).
• Exanthematous phase: Rash begins on forehead, behind the ears, as a red maculopapular eruption.
• Recovery: With onset of rash, symptoms begin to subside;

9. Which of the following is true of mumps?

A. Salivary gland involvement is limited to the parotids
B. The patient is not infectious prior to clinical parotid enlargement.
C. Meningoencephalitis can precede parotitis
D. Mumps orchitis frequently leads to infertility

Ans C a. False; Mumps virus may involve salivary glands (parotid ± submandibular), CNS, pancreas, testes b. False; Virus appears in saliva from up to 7 days before to 7 days after onset of parotid swelling c. True; Meningoencephalitis may occur before, along with, or following the parotitis d. False; Mumps orchitis can lead to atrophy of testes, but Sterility is rare even with bilateral involvement

10. Incubation period of mumps is:

A. 5–7 days
B. 8–15 days
C. 14–24 days
D. 24–30 days

Ans C The incubation period for mumps ranges from 12-25 days but is usually 16-18 days.

11. Most common complication of mumps in children:

A. Orchitis
B. Aseptic meningitis
C. Myocarditis
D. Pancreatitis

Ans B Common complications of Mumps:
• Most common complications of mumps are meningitis, with or without encephalitis, and gonadal involvement
• In adolescent and adult males, orchitis is second only to parotitis as a common finding in mumps.
• Orchitis is extremely rare in prepubescent boys, but occurs in 30-40% of males after puberty.

12. All of the following are features of mumps, except:

A. Caused by paramyxovirus
B. Aseptic meningitis is a complication in children
C. Orchitis is a complication in adults
D. Incubation period is less than 2 weeks

Ans D Mumps virus is a single stranded RNA virus in the family Paramyxoviridae and the genus Rubulavirus.

13. Commonest complication of mumps is:

A. Orchitis and oophritis
B. Encephalitis
C. Pneumonia
D. Myocarditis HIV

Ans A Orchitis and oophritis HIV

14. Risk of congenital defects in a baby is maximum, if the gestational age at the time of maternal rubella infection is:

A. < 11 weeks
B. 11-12 weeks
C. 13-14 weeks
D. 15-16 weeks

Ans A Risk of congenital defects in a baby is maximum, if mother acquires Rubella in the first trimester (before 11 weeks).

15. The minimum interval between the 2 doses of MMR is

A. 2 wk
B. 4 wk
C. 2 mo
D. 6 mo
E. 1 yr

Ans B

16. The portal of entry of measles virus is through the respiratory tract or conjunctivae following contact with large droplets or small-droplet aerosols in which the virus is suspended. Patients with measles are infectious

A. 1 day before to up to 4-6 days after the onset of rash
B. 2 days before to up to 4-6 days after the onset of rash
C. 3 days before to up to 4-6 days after the onset of rash
D. 4 days before to up to 4-6 days after the onset of rash
E. 5 days before to up to 4-6 days after the onset of rash

Ans C

17. Measles is a serious infection characterized by high fever, an enanthem, cough, coryza, conjunctivitis, and a prominent exanthem. All the following are true EXCEPT

A. incubation period is 8-12 days
B. Koplik spots have been reported in 50-70% of measles cases
C. the rash begins on the forehead, behind the ears, and on the upper neck
D. the rash fades over about 3 days in the same progression as it evolved
E. in more severe cases, generalized lymphadenopathy may be present

Ans D The rash fades over about 7 days in the same progression as it evolved, often leaving a fine desquamation of skin in its wake.

18. The MOST common complication of measles is

A. aseptic meningitis
B. pharyngitis
C. otitis media
D. pneumonia
E. cystitis

Ans C Acute otitis media is the most common complication of measles and pneumonia is the most common cause of death in measles.

19. Subacute sclerosing panencephalitis (SSPE) is a chronic complication of measles with a delayed onset and an outcome that is nearly always fatal. SSPE is characterized by

A. females are affected twice as often as males
B. clinical manifestations begin insidiously 3-6 yr after primary measles infection
C. the hallmark of the 1st stage is massive myoclonus
D. the 3rd stage is characterized by loss of critical centers
E. clinical trials using isoprinosine suggest significant benefit

Ans E Males are affected twice as often as females, and there appear to be more cases reported from rural than urban populations. Clinical manifestations of SSPE begin insidiously 7-13 yr after primary measles infection. The hallmark of the 2nd stage is massive myoclonus, which coincides with extension of the inflammatory process site to deeper structures in the brain, including the basal ganglia. The 4th stage is characterized by loss of critical centers that support breathing, heart rate, and blood pressure.

20. The MOST common finding among infants with congenital rubella syndrome is

A. psychomotor retardation
B. cataracts
C. deafness
D. patent ductus arteriosus
E. neonatal purpura

Ans C Nerve deafness is the single most common finding among infants with congenital rubella syndrome.

21. Mumps virus is in the family Paramyxoviridae and the genus Rubulavirus. Mumps is characterized by

A. incubation period ranges from 7-11 days
B. unilateral parotitis rarely becomes bilateral
C. pale opening of the Stensen duct
D. parotid swelling peaks in approximately 7 days
E. a morbilliform rash is rarely seen

Ans E The incubation period for mumps ranges from 12-25 days but is usually 16-18 days. Parotitis may be unilateral initially but becomes bilateral in approximately 70% of cases. The opening of the Stensen duct may be red and edematous. The parotid swelling peaks in approximately 3 days and then gradually subsides over 7 days.

22. The MOST common complication of mumps is

A. meningitis
B. conjunctivitis
C. optic neuritis
D. pneumonia
E. thrombocytopenia

Ans A The most common complications of mumps are meningitis, with or without encephalitis, and gonadal involvement. Uncommon complications include conjunctivitis, optic neuritis, pneumonia, nephritis, pancreatitis, and thrombocytopenia.

23. Intrauterine mumps infection have been associated with

A. glaucoma
B. limb anomalies
C. pulmonary stenosis
D. renal agenesis
E. no fetal malformations

Ans E have been associated with intrauterine mumps infection.

24. Mumps virus is neurotropic and is thought to enter the CNS via the choroid plexus and infect the choroidal epithelium and ependymal cells. CNS involvement is characterized by

A. symptomatic CNS involvement occurs in 40-60% of infected individuals
B. encephalitis most commonly manifests 10 days after the parotitis
C. CNS symptoms usually resolve in 3 days
D. CSF protein content is usually elevated
E. facial palsy is a less-common CNS complication

Ans E Symptomatic CNS involvement occurs in 10-30% of infected individuals, but CSF pleocytosis has been found in 40-60% of patients with mumps parotitis. The meningoencephalitis may occur before, along with, or following the parotitis. It most commonly manifests 5 days after the parotitis. In typical cases, symptoms resolve in 7- 10 days. The CSF protein content is normal or mildly elevated. Less-common CNS complications of mumps include transverse myelitis, aqueductal stenosis, and facial palsy.

Chickenpox & Herpes Zoster

25. All of the following are true about Chickenpox except:

A. Incubation period can range from 10-21 days.
B. Subclinical varicella is rare
C. Simultaneous presence of lesions in various stages of evolution is characteristic
D. Umbilicated vesicles are the earliest lesions to appear

Ans D • Initial exanthem in Chicken pox consists of pruritic erythematous macules that evolve through papular stage to form clear, fluid- filled vesicles. Clouding & umbilication of the lesions begin in 24-48 hr.
• Simultaneous presence of lesions in various stages of evolution is characteristic of varicella.
• Distribution of rash is predominantly centripetal with greatest concentration on trunk.

26. Following are the complications of chickenpox except:

A. Meningitis
B. Pneumonia
C. Enteritis
D. Reye's syndrome

Ans C Recognized complications of varicella include meningoencephalitis, secondary bacterial pneumonia, and Reye syndrome (linked to aspirin use); enteritis is not a typical complication.

27. A girl child is suffering from varicella. And child's aunt is pregnant. When is it earliest that the child can meet her aunt?

A. Incubation period 2–3 days
B. Scabs are infective
C. Centrifugal rash
D. Rash appears on first day
E. Rash can occur in axilla

Ans A When the lesions have crusted , CDC website
• The period of communicability of varicella begins 1–2 days before the onset of rash and ends when all lesions are crusted, typically 4–7 days after onset of rash in immunocompetent people, longer in immunocompromised people
• People with varicella should be isolated until the lesions crust over.

28. Most common complication of chickenpox in children:

A. Encephalitis
B. Secondary bacterial infection
C. Pneumonia
D. Otitis media
E. Orchitis MEASLES

Ans B Secondary bacterial infections of skin, by group A Streptococcus & S. aureus, may occur in children with varicella. MEASLES

29. Sabin Feldman dye test is used for:

A. Toxoplasma
B. Plasmodium
C. Syphilis
D. Herpes

Ans A The SabinFeldman dye test is sensitive & specific for Toxoplasma; It measures primarily IgG antibodies;

30. Which of the following vaccines is contraindicated for a patient with X-linked agammaglobulinemia?

A. oral polio vaccine
B. hepatitis B
E. varicella

Ans A Oral polio, smallpox, live attenuated influenza, and BCG all these vaccines are contraindicated for this patient.

31. Leukocyte adhesion defects are caused by defects in the β chain of integrin (CD18), which is required for the normal process of neutrophil aggregation and attachment to endothelial surfaces. It is characterized by all the following EXCEPT

A. delayed cord separation
B. recurrent infections
C. ecthyma gangrenosum
D. neutropenia
E. survival is usually <10 yr

Ans D Because the defect involves leukocyte migration and adherence, the neutrophil count in the peripheral blood is usually extremely elevated but pus is not found at the site of infection.

32. Which of the following is a gram negative bacterium?

A. Staphylococcus aureus
B. Streptococcus pneumoniae
C. Actinomyces
D. Haemophilus influenza
E. Nocardia

Ans D Actinomyces, staphylococcus, streptococcus, diphtheria, enterococcus, listeria and nocardia are gram positive.

33. Human brucellosis is caused by organisms of the genus Brucella and continues to be a major public health problem worldwide. All the following are true EXCEPT

A. symptoms can be acute or insidious
B. most patients present with fever, arthralgia/arthritis, and hepatosplenomegaly
C. some present as a fever of unknown origin
D. variable fever pattern
E. invasion of the nervous system occurs in approximately 20% of cases

Ans E Headache, mental inattention, and depression may be demonstrated in patients with brucellosis, invasion of the nervous system occurs in only approximately 1% of cases.

34. Varicella-zoster virus (VZV) causes primary, latent, and recurrent infections. All the following statements are true EXCEPT

A. varicella is a serious disease in young infants
B. within households, transmission of VZV occurs at a rate of 65-86%
C. Herpes zoster is more common in winter
D. the lifetime risk for herpes zoster for individuals with a history of varicella is 20- 30%
E. Herpes zoster is very rare in healthy children younger than 10 yr of age

Ans C Herpes zoster is caused by the reactivation of latent VZV. It is not common in childhood and shows no seasonal variation in incidence.

35. Varicella is an acute febrile rash illness that is common in Iraq. Of the following, the TRUE statement is

A. it usually begins 4-6 days after exposure
B. subclinical varicella is common
C. mild abdominal pain may occur 24-48 hr before the rash appears
D. temperature elevation usually as high as 41.1°c
E. lesions often appear first on the extremities

Ans C The illness usually begins 14-16 days after exposure, although the incubation period can range from 10-21 days. Subclinical varicella is rare. Temperature elevation is usually 37.8-38.9°C but may be as high as 41.1°C. Varicella lesions often appear first on the scalp, face, or trunk.

36. All the following are true about varicella in unvaccinated individuals EXCEPT

A. simultaneous presence of lesions in the same stages of evolution
B. distribution of the rash is predominantly central
C. many children have vesicular lesions on the eyelids
D. exanthem may be much more extensive in children with skin disorders
E. hypopigmentation or hyperpigmentation of lesion sites persists for days to weeks in some children

Ans A The simultaneous presence of lesions in various stages of evolution is characteristic of varicella.

37. Encephalitis and acute cerebellar ataxia are well-described neurologic complications of varicella. All the following are true EXCEPT

A. morbidity from central nervous system complications is highest among patients younger than 5 yr and older than 20 yr
B. nuchal rigidity, altered consciousness, and seizures characterize meningoencephalitis
C. patients with cerebellar ataxia have a gradual onset of gait disturbance, nystagmus, and slurred speech
D. neurologic symptoms usually begin 2-6 days after the onset of the rash but may occur during the incubation period or after resolution of the rash
E. clinical recovery is typically occurring after 96 hr, and is usually gradual

Ans E Clinical recovery is typically rapid, occurring within 24-72 hr, and is usually complete.

38. Oral therapy with acyclovir (20 mg/kg/dose; maximum: 800 mg/dose) given as 4 doses/day for 5 days can be used to treat uncomplicated varicella in individuals at increased risk for moderate to severe varicella. It includes all the following individuals EXCEPT

A. nonpregnant individuals older than 12 yr of age
B. individuals older than 12 mo of age with chronic cutaneous disorders
C. individuals receiving short-term corticosteroid therapy
D. individuals receiving long-term salicylate therapy
E. individuals with chronic hematological disorders

Ans E

Dengue & Arboviral Infections

39. A neonate presented with cicatrising skin lesions all over the body with hypoplasia of all limbs. A MRI of the brain revealed diffuse cerebral atrophy. An ophthalmologic evaluation reveals chorioretinitis. Which of these tests is most likely to show a positive result in this patient?

A. Anti HCMV antibody
B. Anti toxoplasma antibody
C. Anti VZV antibody
D. Anti rubella antibody

Ans C Cicatrising skin lesions, hypoplasia of all limbs and chorioretinitis are all features of Congenital Varicella syndrome. So, Anti VZV antibody would be positive in this baby c. Enteritis Complications of Varicella infection:
• Hematologic: Mild thrombocytopenia (Petechiae. Purpura) • CVS: myocarditis, pericarditis
• Neurologic: Acute cerebellar ataxia, Meningoencephalitis • GI: pancreatitis, gastrointestinal bleeding
• Respiratory: Pneumonia, • Eye: Acute retinal necrosis

40. Papularpurpuric 'Gloves and socks' syndrome is caused by

A. EB virus
B. West Nile virus
C. Parvo virus B19
D. KFD virus

Ans C Papular purpuric "gloves-and-socks" syndrome (PPGSS) is a selflimited condition associated with Parvovirus B19 infection & is characterized by fever, pruritus, painful edema & erythema localized to distal extremities in a distinct "glovesandsocks" distribution, followed by acral petechiae & oral lesions.

41. Dengue shock syndrome is characterized by the following except:

A. Hepatomegaly
B. Pleural effusion
C. Thrombocytopenia
D. Decreased hemoglobin

Ans D In Dengue hemorrhagic fever, there is hemoconcentration, due to increased vascular permeability, so the hematocrit & hemoglobin level increases & not decreases;

42. Rose spot is associated with

A. Enteric fever
B. Malarial
C. Hodgkins disease
D. Meningitis PERTUSSIS

Ans A Typhoid fever Skin rash in Enteric fever:
• In 25% of cases, a macular or maculopapular rash (rose spots) may be visible around the 7th10th day of the illness
• Lesions may appear in crops of 10-15 on the lower chest and abdomen and last 2-3 days
• These lesions may be difficult to see in dark-skinned children.

43. Which microorganism is responsible for classical pres entation of hydrocephalus, chorioretinitis, intracerebral calcification?

A. Toxoplasmosis
B. Rubella
C. Measles
D. CMV FUNGAL INFECTIONS

Ans A • Intracerebral calcification can be seen in both congenital Toxoplasmosis & CMV infection
• Hydrocephalus is seen in Toxoplasmosis while microcephaly in CMV infection.

44. Which of the following vaccines is contraindicated for a patient with chronic renal disease?

A. pneumococcal
B. hepatitis B
C. live attenuated influenza
D. varicella
E. hepatitis A

Ans C

45. Penicillins are the drugs of choice for pediatric infections caused by the following

A. group A Streptococcus
B. H .influenzae
C. Treponema pallidum
D. L. monocytogenes
E. N. meningitides

Ans B Although there has been ever-increasing emergence of resistance to penicillins but they remain the drugs of choice for pediatric infections caused by group A and group B Streptococcus, Treponema pallidum (syphilis), L. monocytogenes, and N. meningitidis.

46. Regarding epidemiology of Neisseria meningitides, all the following are true EXCEPT

A. meningococci are transmitted during close contact via aerosol droplets or exposure to respiratory secretions
B. **meningococci survive for long periods in the environment**
C. smoking and respiratory viral infection are associated with increased rates of carriage and disease
D. the highest rate of meningococcal disease occurs in infancy
E. most cases of meningococcal disease are sporadic

Ans B

47. Worsening of acne in adolescent female on anti-tuberculous treatment is caused by

A. **isoniazid**
B. rifampin
C. pyrazinamide
D. ethambutol
E. streptomycin

Ans A Major adverse events include hepatotoxicity in 1% of children and approximately 3% of adults (increasing with age) and dose-related peripheral neuropathy. Minor adverse events include rash, worsening of acne, epigastric pain with occasional nausea and vomiting, decreased vitamin D levels, and dizziness.

48. Epstein-Barr virus (EBV) is shed in oral secretions after acute infection for

A. 2 wk
B. 4 wk
C. 2 mo
D. 4 mo
E. **6 mo**

Ans E EBV is shed in oral secretions consistently for more than 6 mo after acute infection and then intermittently for life.

Tuberculosis

49. Which of the following intrauterine infections is associated with limb reduction defects and scarring of skin?

A. **Varicella virus**
B. Herpes virus
C. Rubella
D. Parvo virus

Ans A

50. Palivizumab is a humanized monoclonal antibody. For which of the following conditions has it been approved for?

A. Coxsackie virus
B. Avian influenzae
C. Parainfluenzae
D. **Respiratory syncytial virus**

Ans D Drug Use in RSV infection Palivi- It is a monoclonal antibody licensed for prophylaxis in high-risk infants during the RSV season, and does prevent dna zumab about half of the expected hospitalizations in that population. Ribavirin It is an antiviral agent delivered as an aerosol that showed a modest beneficial effect on the course of RSV pneumonia, ith some reduction in the duration of both mechanical ventilation and hospitalization. However, latest studies have failed

51. Most common route of TB infection in a neonate born by caesarean section:

A. Hematogenously spread
B. Through skin contact
C. **Inhalational of aerosol**
D. Transplacental
E. By aspiration of infected amniotic fluid

Ans C Congenital TB
• Congenital transmission usually occurs from a lesion in the placenta through the umbilical vein, when tubercle bacilli infect the fetal liver, where a primary focus with periportal lymph node involvement can occur.
• Congenital tuberculosis can also be caused by aspiration or ingestion of infected amniotic fluid. However, the most common route of infection for the neonate is postnatal airborne transmission from an adult with infectious pulmonary tuberculosis.

52. A 3 month old, asymptomatic infant with, H/o TB exposure, has taken 3 months of chemoprophylaxis, what is to be done next?

A. Test sputum and then decide
B. **Continue for 3 more months**
C. Tuberculin test then decide
D. Immunise with BCG & stop prophylaxis

Ans B A child born to mother who was diagnosed to have TB in pregnancy should receive prophylaxis for 6 months, provided congenital TB has been ruled out; BCG vaccination can be given at birth even with INH chemoprophylaxis.

53. An asymptomatic newborn whose mother had smear positive TB received INH for 3 months. What should be done next?

A. **Continue INH for further 3 months**
B. Discontinue as the child is asymptomatic
C. Do a chest X Ray
D. BCG vaccination should not be given

Ans A Indian Pediatr 2013;50:301-306

54. A child presents with tuberculosis. Steroids are not indicated in:

A. Tuberculoma
B. Endobronchial tuberculosis
C. Massive pleural effusion
D. **Progressive primary pulmonary disease**

Ans D Indications of Corticosteroids in TB:
• Tuberculous meningitis: reduces vasculitis, inflammation & ICP
• Tuberculous massive pleural effusion
• Endobronchial tuberculosis • Miliary tuberculosis
• Tuberculous pericardial effusion

55. True about childhood tuberculosis is:

A. 5% prevalence
B. **Common in 5–8 years age**
C. Diagnosis by sputum is easily done
D. Highly contagious

Ans B

56. Most common age for pediatric TB is:

A. <1 year
B. **1–4 years**
C. 5–8 years
D. 8–14 years

Ans B 1–4 years Answers

57. Most common organ involved by congenital TB is:

A. **Liver**
B. Pancreas
C. Kidney
D. Lung

Ans A

58. TB in children, most common is:

A. Abscess
B. Consolidation
C. **Hilar adenopathy**
D. CNS tuberculosis

Ans C

59. Management of a newborn when mother has active Tuberculosis and is not taking ATT:

A. BCG + Rifampicin + INH + Breastfeeding
B. BCG + Isolation of baby
C. BCG + INH for 6 week + Breastfeeding
D. **BCG + INH + withhold Breastfeeding DIPHTHERIA**

Ans D BCG + INH + withhold Breastfeeding Breastfeeding is withheld only till the mother is infective (usually for around 3 weeks after starting ATT). DIPHTHERIA

Typhoid & Enteric Fever

60. Subacute sclerosing panencephalitis is a late neuro logical complication associated with infection due to:

A. **Measles virus**
B. Mumps virus
C. Rubella virus
D. Parainfluenza virus

Ans A Subacute sclerosing panencephalitis (SSPE) is a chronic complication of measles with a delayed onset & fatal outcome.

61. True regarding JE:

A. F irst case was reported in 2005
B. **Transmitted by Culex tritaeniorhynchus**
C. Permanent neurologic or psychiatric sequelae
D. Major outbreaks of JE occur every 20 years
E. Vaccine available in India

Ans B c. Permanent neurologic or psychiatric sequelae & e. Vaccine available in India; a. False Epidemics of Japanese encephalitis were reported in Japan from the late 1800s b. True Culex tritaeniorhynchus, a night-biting mosquito transmits JE in Asia c. True Sequelae are seen in 5-70% of JE cases & include mental deterioration, severe emotional instability, personality changes, motor abnormalities, and speech disturbances. d.

62. Which of the following is incorrect regarding miliary TB?

A. Usually associated with meningeal TB
B. **In children it is most commonly due to reactivation**
C. Mantoux test may yield a false negative result
D. There is hematogenous dissemination 174 Section 1: General Pediatrics

Ans B Harrison's 19/e p 1103-1113 a. True In >50% cases of Tubercular meningitis, evidence of old pulmonary lesions or a miliary pattern is found on chest X ray b. False Miliary tuberculosis usually complicates primary infection (within 2-6 mo of initial infection) in infants & children; while, in adolescents & older adults, it usually results from the breakdown of a previously healed primary pulmonary lesion. c. True In severe cases of TB like military TB, Mantoux test may be falsely negative d.

63. A child presents with complaints of cough. Characteristic inspiratory whoop present. He is not immunised. Sample for investigation is:

A. **Nasopharyngeal swab**
B. Tracheal aspiration
C. Cough plate culture
D. Sputum culture

Ans A For diagnosis of Whooping cough isolation of B. pertussis in culture remains the gold standard
• The specimen is obtained with deep nasopharyngeal aspiration or a swab
• Cough plates and postnasal swabs are unsatisfactory because of overgrowth by commensal bacteria
• DNA detection by PCR is replacing culture because of increased sensitivity & quicker results. d. Pertussis

64. Approximately 80% of blood cultures that will be positive are identified at

A. **1st 24 hr from incubation**
B. 25 – 48 hr from incubation
C. after 3 day from incubation
D. after 5 day from incubation
E. after 7 day from incubation

Ans A

65. Waterless hand hygiene products are effective in killing most microbes but do not remove dirt or debris and are ineffective against

A. Pseudomonas
B. hepatitis A
C. difficile
D.
E. S. aureus

Ans D Waterless hand hygiene products increase hand hygiene compliance and save time; these agents are the preferred agents for routine hand hygiene when hands are not visibly soiled. These products are effective in killing most microbes but do not remove dirt or debris. However, they are ineffective against C. difficile spores, requiring the use of other cleansing products during hospital C. difficile outbreaks.

66. Cefixime is a third-generation cephalosporin active against all the following EXCEPT

A. Streptococci
B. **Staphylococci**
C. H. influenza
D. Neisseria gonorrhoeae
E. Proteus vulgaris

Ans B Active against streptococci, H. influenzae, M. catarrhalis, Neisseria gonorrhoeae, Serratia marcescens, and Proteus vulgaris. No antistaphylococcal or antipseudomonal activity. Dose: 8 mg/kg/24 hr divided q 12-24 hr PO.

67. Gonorrhea is manifested by a spectrum of clinical presentations from asymptomatic carriage, to the characteristic localized urogenital infections, to disseminated systemic infection. Regarding disseminated gonococcal infection, the following statement is TRUE

A. hematogenous dissemination occurs in 10-30% of all gonococcal infections
B. men account for the majority of cases
C. meningitis and osteomyelitis are the most common manifestations
D. skin lesions found in75% of patients
E. **acute endocarditis is an uncommon but often fatal manifestation**

Ans E Hematogenous dissemination occurs in 1-3% of all gonococcal infections, more frequently after asymptomatic primary infections than symptomatic infections. Women account for the majority of cases, with symptoms beginning 7-30 days after infection and within 7 days of menstruation. The most common manifestations are asymmetric arthralgia, petechial or pustular acral skin lesions, tenosynovitis, suppurative arthritis, and, rarely, carditis, meningitis, and osteomyelitis. Only 25% of patients complain of skin lesions.

68. The least common clinical feature of typhoid fever in children is

A. diarrhea
B. abdominal pain
C. pallor
D. splenomegaly
E. **headache**

Ans E Diarrhea 36%, abdominal pain 21%, pallor 20%, splenomegaly 17% and headache 4%.

69. All the following are clinical features of typhoid fever EXCEPT

A. **incubation period is usually 30-45 days**
B. macular or maculopapular rash (rose spots) may be visible around the 7th-10th day of the illness
C. if complications not occur, the symptoms and physical findings gradually resolve within 2-4 wk
D. typhoid fever usually manifests as high-grade fever with a wide variety of associated features
E. relative bradycardia, neurologic manifestations, and gastrointestinal bleeding, are rare in children

Ans A The incubation period of typhoid fever is usually 7-14 days but depends on the infecting dose and ranges between 3 and 30 days.

70. Regarding diagnosis of typhoid fever, one of the following is TRUE

A. blood cultures are positive in 65-80% of the patients
B. urine culture results become positive within the 1st wk
C. thrombocytosis may be a marker of severe illness
D. **diagnosis by Widal test alone is prone to error**
E. leukocytosis is rare in young children

Ans D Results of blood cultures are positive in 40-60% of the patients seen early in the course of the disease, and stool and urine culture results become positive after the 1st wk. Thrombocytopenia may be a marker of severe illness and may accompany disseminated intravascular coagulopathy. Blood leukocyte counts are frequently low in relation to the fever and toxicity, there is a wide range in counts; in younger children leukocytosis is common and may reach 20,000-25,000 cells/µL.

Malaria

71. Warthin Finkeldey giant cells are seen in:

A. Mumps
B. Rubella
C. **Measles**
D. Polio

Ans C Fusion of infected cells results in multinucleated giant cells, the WarthinFinkeldey giant cells that are pathognomonic for measles, with up to 100 nuclei and intracytoplasmic and intranuclear inclusions.

72. Amplifying host of Japanese encephalitis is?

A. Human
B. **Pig**
C. Cat
D. Dog

Ans B Pigs serve as an amplifying host in Japanese encephalitis.

73. Which of the following statements is true about childhood TB?

A. 5% prevalence
B. Common in 5-8 years age
C. **Diagnosis by sputum is difficult**
D. Highly contagious

Ans C > b. Common in 58 years age Option a. False Prevalence of childhood TB is 10-20% of all TB cases Option b. True Most common in children less than 5 years age; but it is also common in 5-14 years age group Option c. True Children with TB are usually sputum negative; Moreover, collecting sputum sample is difficult in children Option d. False Childhood TB is usually a paucibacillary disease, so it is rarely contagious

74. Leucocytosis is seen in which of the following childhood infections?

A. Tuberculosis
B. Typhoid
C. Diphtheria
D. **Pertussis**

Ans D characteristically causes a marked leukocytosis with absolute lymphocytosis, unlike typhoid (leukopenia) or tuberculosis (usually normal counts).

75. What is the most common complication of malaria in children?

A. Cerebral malaria
B. Respiratory distress
C. **Severe malarial anemia**
D. Hypogylcemia

Ans C Most common serious complication of malaria is severe anemiaQ

76. The following is/are helpful in a child with severe falciparum malaria with high parasitemia:

A. IV Dexamethasone
B. **Exchange transfusion**
C. Hyperbaric oxygen
D. All the above

Ans B Supportive treatment for severe Malaria includes exchange transfusion in P. falciparum malaria with parasitemia greater than 10% with evidence of severe complications (e.g., severe malarial anemia, cerebral malaria). Nelson also says: "Exchange transfusion is thought to be useful in severe malaria with high-level parasitemia, but no randomized clinical trial has ever been conducted to assess its utility, & some groups, including CDC no longer advocate its use for severe malaria. Corticosteroids are

77. Resistant plasmodium falciparum malaria in the pediatric age group should be treated with:

A. Chloroquine
B. Tetracycline
C. **Clindamycin**
D. Doxycycline

Ans C

78. All patients who are presumed or proven to have gonorrhea should be evaluated for concurrent presence of all the following infections EXCEPT

A. syphilis
B. hepatitis B
C. trachomatis
D.
E. **HSV2**

Ans E All patients who are presumed or proven to have gonorrhea should be evaluated for concurrent syphilis, hepatitis B, HIV, and C. trachomatis infection.

79. Isoniazid is accompanied by significant drug–drug interactions, which of the following is TRUE?

A. aluminum salts increase absorption of isoniazid
B. **isoniazid increase toxicity of carbamazepine**
C. rifampin decreased hepatotoxicity of isoniazid
D. isoniazid decrease level of warfarin
E. prednisolone decreased isoniazid metabolism

Ans B Aluminum salts decrease absorption of isoniazid, rifampin increase hepatotoxicity of isoniazid, isoniazid increase level of warfarin and prednisolone increased isoniazid metabolism.

80. What percent of the world's population infected by Epstein-Barr virus (EBV)?

A. 5%
B. 25%
C. 50%
D. 75%
E. **95%**

Ans E EBV infects more than 95% of the world's population.

Leishmaniasis & Kala Azar

81. Chronic carrier state is seen in all, except:

A. **Measles**
B. Diphtheria
C. Typhoid
D. Gonorrhoea

Ans A 90% of exposed susceptible individuals experience measles

82. Roseola infantum is also known as?

A. ALL
B. **CML**
C. Pure red cell aplasia
D. Hairy cell leukemia

Ans B Exanthema subitum

83. Breastfeeding should be avoided by a mother on ATT for how many weeks?

A. 1
B. 2
C. **3**
D. 4

Ans C 3 • Once the mother and child are taking adequate therapy, it is usually safe for the mother to breastfeed, as the medications, although found in milk, are present in low concentrations
• AAP recommends continued feeding with expressed milk in mothers with pulmonary TB who are contagious, untreated or treated (< 3 wk) with isolation.
• WHO recommends breastfeeding under all circumstances, however, close contact with the baby should be reduced

84. Criteria for diagnosing SIRS all seen except:

A. Temp > 38.5 degree C or < 36 degree C
B. RR > 2 SD above normal for age
C. Children < 1 year old with persistent bradycardia over 0.5 hour
D. **> 50% immature neutrophils**

Ans D Pediatric SIRS criteria require ≥10% immature neutrophils (bands), not >50%, alongside abnormal temperature, tachypnea, and abnormal white cell count or bradycardia in infants.

85. The optimal amount of blood to collect from a pediatric patient for blood culture depend on

A. age
B. **weight**
C. length
E. laboratory standards

Ans B

86. **All the following are recognized skin infections or infestations in children in childcare EXCEPT**

A. impetigo
B. pediculosis
C. scabies
D. **erythrasma**
E. tinea corporis

Ans D

87. **Metronidazole is highly effective in the treatment of infections caused by anaerobes. It can increase the level of which of the following drugs if given at the same time**

A. carbamazepine
B. rifampin
C. phenobarbital
D. **phenytoin**
E. vaiproic acid

Ans D It may increase levels of warfarin, phenytoin, and lithium.

88. **Children who have bacteremia or arthritis caused by gonococcal infections should be treated with ceftriaxone (50 mg/kg/day; maximum: 1 g/day if weighs <45 kg) for a minimum of**

A. 3 days
B. 5 days
C. **7 days**
D. 14 days
E. 21 days

Ans C Children who have bacteremia or arthritis should be treated with ceftriaxone (50 mg/kg/day; maximum: 1 g/day if weighs <45 kg) for a minimum of 7 days. Meningitis should be treated for 10-14 days, and endocarditis for a minimum of 28 days, with ceftriaxone (50 mg/kg/ dose q12h with maximum of 1-2 g IV q12h). Neonatal gonococcal ophthalmia is treated effectively with a single dose of ceftriaxone (50 mg/kg IM, not to exceed 125 mg); a single dose of cefotaxime (100 mg/kg IM) is an acceptable alternative.

89. **NICUs with a high incidence of invasive candidiasis should consider prophylaxis with fluconazole in infants with a birthweight of**

A. <750 g
B. **<1,000 g**
C. <1,500 g
D. <2,000 g
E. <2,500 g

Ans B Twice weekly fluconazole at 3 and 6 mg/kg/dose decreases rates of both colonization with Candidaspecies and invasive fungal infections.

90. **Infectious mononucleosis is the best-known clinical syndrome caused by Epstein- Barr virus (EBV). It is characterized by**

A. symptomatic hepatitis
B. **elevated liver enzymes**
C. massive splenic enlargement
D. huge hepatomegaly
E. occasional palatal petechiae

Ans B The classic physical examination findings are generalized lymphadenopathy (90% of cases), splenomegaly (50% of cases), and hepatomegaly (10% of cases). Epitrochlear lymphadenopathy is particularly suggestive of infectious mononucleosis. Symptomatic hepatitis or jaundice is uncommon, but elevated liver enzymes are very common. Splenomegaly to 2-3 cm below the costal margin is typical (15-65% of cases) and is seen in most cases by ultrasonography; massive enlargement is uncommon.

91. **The MOST common cause of death in kala azar is**

A. severe anemia
B. bleeding
C. **secondary bacterial infection**
D. hepatic failure
E. renal failure

Ans C The late stage of the illness is often complicated by secondary bacterial infections, which frequently are a cause of death.

Meningitis

92. **Subacute sclerosing panencephalitis is a complication of:**

A. Mumps
B. Rubella
C. Cytomegalovirus
D. **Measles**

Ans D

93. **Most common cause of infectious croup is:**

A. Adenovirus
B. **Parainfluenza type I**
C. RSV
D. Influenza

Ans B

94. **In which of the following tubercular infection, steroid is used?**

A. TB Lymphadenitis
B. TB Osteomyelitis
C. **Endobronchial TB**
D. Neurotuberculosis

Ans C

95. The most common pathogenic organism causing pyog enic liver abscess In children is:

A. Staphylococcus aureus
B. H Influenzae
C. Entamoeba HIstolytic
D. Klebsiella

Ans A In children with pyogenic liver abscesses, the most common pathogenic organisms include Staphylococcus aureus, Streptococcus spp. Escherichia coli, Klebsiella pneumoniae, Salmonella & anaerobic organisms Entamoeba histolytica or Toxocara canis–associated liver abscesses have also been reported in developing countries.

96. Most common cause of neonatal meningitis is:

A. E. coli
B. Listeria
C. Staphylococcus aureus
D. Pseudomonas

Ans A CDC website Most common cause of acute Bacterial Meningitis in: INDIA WORLD Age group Organism Age group Organism Neonates E. coli Neonates Gr B Streptococcus, E. coli, Listeria 1 month -2 year H. influenzae Infants & children S. pneumoniae, N. meningitidis, H. influenzae Older children S. pneumoniae, N. meningitidis Adolescents S. pneumoniae, N. meningitidis

97. Neonatal meningitis is caused by all except:

A. N. meningitis
B. E. coli
C. Listeria
D. Group B streptococci

Ans A N.meningitidis CDC website

98. Cephalosporins are widely used in pediatric practice, both in oral and parenteral formulations. Which of the following is a 2nd generation cephalosporins?

A. cefazolin
B. cephalexin
C. cefuroxime
D. ceftazidime
E. ceftaroline

Ans C The first-generation cephalosporins (e.g., cefazolin, a parenteral formulation, and cephalexin, an oral equivalent) are commonly used for management of skin and soft-tissue infections caused by susceptible strains of S. aureus and group A Streptococcus. The second-generation cephalosporins (e.g., cefuroxime, cefoxitin) have better activity against Gram-negative bacterial infections than do first-generation cephalosporins and are used to treat respiratory tract infections, urinary tract infections, and skin and...

99. S. pneumoniae is the most frequent cause of bacteremia, bacterial pneumonia, otitis media, and bacterial meningitis in children. Children at increased risk of pneumococcal infections include those with the following conditions EXCEPT

A. megaloplastic anemia
B. deficiencies in humoral immunity
C. HIV infection
D. cerebrospinal fluid leak
E. cochlear implants

Ans A Children at increased risk of pneumococcal infections include those with sickle cell disease, asplenia, deficiencies in humoral (B cell) and complement-mediated immunity, HIV infection, certain malignancies (e.g., leukemia, lymphoma), chronic heart, lung, or renal disease (particularly nephrotic syndrome), cerebrospinal fluid leak, and cochlear implants.

100. The MOST common clinical manifestation of meningococcal infection is

A. asymptomatic carriage
B. meningococcal meningitis
C. bacteremia without sepsis
D. meningococcal septicemia
E. pneumonia

Ans A The most common clinical manifestation of meningococcal infection is asymptomatic carriage of the organism in the nasopharynx.

101. In meningococcal purpura, necrotic skin lesions are less common among children treated with

A. penicillin G
B. ampicillin
C. cefotaxime
D. ceftriaxone
E. meropenem

Ans D

102. The MOST common complication of acute severe meningococcal septicemia is

A. arthritis
B. focal skin infarction
C. endocarditis
D. pneumonia
E. peritonitis

Ans B The most common complication of acute severe meningococcal septicemia is focal skin infarction, which most commonly affects the lower limbs and can lead to substantial scarring and require skin grafting.

103. A poor prognostic factor for invasive meningococcal disease on presentation is

A. hypertension
B. leukocytosis
C. alkalosis
D. meningitis
E. normal erythrocyte sedimentation rate

Ans E Poor prognostic factors on presentation include hypothermia or extreme hyperpyrexia, hypotension or shock, purpura fulminans, seizures, leukopenia, thrombocytopenia (including disseminated intravascular coagulation), acidosis, and high circulating levels of endotoxin and tumor necrosis factor-α. The presence of petechiae for <12 hr before admission, absence of meningitis, and low or normal erythrocyte sedimentation rate indicate rapid, fulminant progression and poorer prognosis.

104. **Of the following, the MOST effective agent for prophylaxis of meningococcal disease is**

A. ceftriaxone
B. rifampin
C. ampicillin
D. penicillin
E. amoxicillin

Ans A and ciprofloxacin are the most effective agents for prophylaxis, the latter being the drug of choice in some countries. Rifampin is most widely used but fails to eradicate colonization in 15% of cases. Neither penicillin nor ampicillin treatment eradicates nasopharyngeal carriage and should not be routinely used for prophylaxis.

105. **The MOST common cause of recurrent aseptic meningitis is**

A. mumps
C. adenoviruses
D. herpes simplex virus
E. influenza virus

Ans D HSV is the most common cause of recurrent aseptic meningitis (Mollaret meningitis).

Pneumonia

106. **SSPE (Subacute Sclerosing Panencephalitis) is associated with:**

A. Mumps
B. Chickenpox
C. Herpes
D. Measles
E. Rubella MUMPS

Ans D Measles with MUMPS

107. **A newborn presents with petechiae skin lesions, hema turia, & platelet count is 22,000/L. Most likely caused by:**

A. Congenital CMV infection
B. Congenital rubella infection
C. Premature infants
D. Both CMV or rubella infection

Ans D Both congenital CMV & rubella infections can cause thrombocytopenia & Purpura due to thrombocytopenia.

108. **First line ATT in children:**

A. Streptomycin
B. Pyrazinamide
C. Ethionamide
D. Ethambutol
E. Ofloxacin

Ans A b. Pyrazinamide and d. Ethambutol with

109. **Treatment of choice for pneumonia due to Mycoplasma is?**

A. Azithromycin
B. Doxycycline
C. Amoxicillin
D. Ciprofloxacin

Ans A M. pneumoniae is sensitive to macrolides (erythromycin, clarithromycin, azithromycin), tetracyclines, and quinolones Recommended treatment is clarithromycin (15 mg/kg/day divided into 2 doses PO for 10 days) or azithromycin (10 mg/kg once PO on day 1 and 5 mg/kg once daily PO on days 2-5).

110. **The IgM response occurs earlier in the illness, generally peaking at 7-10 days after infection, and usually disappears within a few weeks, but for some infections it can persist for months such as**

A. measles
B. mumps
C. rubella
D. hepatitis A
E. varicella

Ans D Hepatitis A and West Nile Virus.

111. **A 3-year-old child diagnosed with shigella infection, he can return to his daycare**

A. 2 days after initiation of treatment
B. after diarrhea resolves
C. after diarrhea resolves and results of a single stool culture is negative for these organisms
D. after diarrhea resolves and results of 2 stool cultures are negative for these organisms
E. after diarrhea resolves and results of 3 stool cultures are negative for these organisms

Ans D

112. **Toxic shock syndrome is an acute and potentially severe illness characterized by all the following EXCEPT**

A. desquamation on the hands and feet
B. myalgias
C. focal neurologic abnormalities
D. conjunctival hyperemia
E. strawberry tongue

Ans C Toxic shock syndrome (TSS) is an acute and potentially severe illness characterized by fever, hypotension, erythematous rash with subsequent desquamation on the hands and feet, and multisystem involvement, including vomiting, diarrhea, myalgias, nonfocal neurologic abnormalities, conjunctival hyperemia, and strawberry tongue.

113. **The MOST common etiology of joint and bone infections in young children is**

A. Staphylococcus aureus
B. Streptococcus pneumoniae
C. Kingellakingae
D. Haemophilus influenza
E. Neisseria meningitidis

Ans C is being increasingly recognized as the most common etiology of joint and bone infections in young children.

114. Antiviral chemotherapy typically requires a delicate balance between targeting critical steps in viral replication without interfering with host cellular function. Which of the following antiviral blocks M2 protein ion channel?

A. acyclovir
B. **amantadine**
C. ganciclovir
D. foscarnet
E. vidarabine

Ans B While the others inhibits viral DNA polymerase.

115. Human herpesvirus 6 (HHV-6A and HHV-6B) and human herpesvirus 7 (HHV-7) cause infection in infancy and early childhood. HHV-6B is responsible for the majority of cases of roseola infantum (exanthema subitum or sixth disease). All the following are true EXCEPT

A. 95% of children being infected with HHV-6 by 2 yr of age
B. peak age of primary HHV-6B infection is 6-9 mo of life
C. congenital infection with HHV-6 occurs in 1% of newborns
D. **congenital infection with HHV-7 is well recognized**
E. breast milk does not play a role in transmission of either HHV-6 or HHV-7

Ans D Congenital infection with HHV-7 has not been demonstrated.

UTI & Pyelonephritis

116. Parotitis and orchitis are common manifestations of:

A. Diphtheria
B. Measles
C. Rubella
D. **Mumps**

Ans D Mumps is characterized by fever, bilateral or unilateral parotid swelling and tenderness, and the frequent occurrence of meningoencephalitis and orchitis.

117. A neonate, born to a mother infected with CMV, on routine examination at birth was found to have hepatomegaly. Rest of the examination was essentially unremarkable. On investigations, Anti HCMV antibodies were found to be positive in the baby. What sequelae in later life is the child at risk of?

A. Renal failure
B. **Sensorineural hearing loss**
C. Hepatic fibrosis
D. Mental retardation

Ans B Hearing loss is the most common longterm sequela of congenital CMV infection.

118. Bullneck is seen in severe cases of which of the following?

A. **Diphtheria**
B. Tubercular lymphadenitis
C. Mumps
D. Goitre

Ans A

119. Most common organism responsible for infective endo carditis in children:

A. β-hemolytic streptococci
B. a-hemolytic streptococci
C. **Staphylococcus aureus**
D. Group D. streptococci

Ans C Viridans-type streptococci (a-hemolytic streptococci) and Staphylococcus aureus remain the leading causative agents for endocarditis in pediatric patients; Staphylococcal endocarditis is more common in patients with no underlying heart disease, Strepto viridans is more common after dental procedures & Enterococcus is more common after bowel or genitourinary manipulation.

120. Vaccines are defined as whole or parts of microorganisms administered to prevent an infectious disease. Which of the following is a live attenuated vaccine?

A. hepatitis A
B. hepatitis B
C. pneumococcal
D. **varicella**
E. diphtheria

Ans D Vaccines can consist of whole inactivated microorganisms (e.g., polio and hepatitis A), parts of the organism (e.g., acellular pertussis, HPV, and hepatitis B), polysaccharide capsules (e.g., pneumococcal and meningococcal polysaccharide vaccines), polysaccharide capsules conjugated to protein carriers (e.g., Hib, pneumococcal, and meningococcal conjugate vaccines), live attenuated microorganisms (measles, mumps, rubella, varicella, rotavirus, and live-attenuated influenza vaccines), and toxoids (tetanus and...

121. Infants, children, and adolescents in Iraq are routinely immunized against

A. 10 diseases
B. **11 diseases**
C. 12 diseases
D. 13 diseases
E. 14 diseases

Ans B Infants, children, and adolescents in the Iraq routinely are immunized against 11 diseases: BCG, poliomyelitis, hepatitis B, diphtheria, tetanus, pertussis, H. influenzae type b (Hib) disease, rotavirus, measles, mumps and rubella.

122. Which cephalosporin should not be mixed or reconstituted with a calcium- containing product, such as Ringer solution or parenteral nutrition containing calcium?

A. cefazolin
B. cefotaxime
C. **ceftriaxone**
D. ceftazidime
E. cefepime

Ans C should not be mixed or reconstituted with a calcium-containing product, because particulate formation can result. Cases of fatal reactions with ceftriaxone–calcium precipitates in lungs and kidneys in neonates have been reported.

123. **Diagnosis of chancroid in infants and children is a strong evidence of sexual abuse. Chancroid is caused by**

A. **Haemophilus ducreyi**
B. Syphilis
C. trachomatis
D. Kingellakingae
E. Neisseria gonorrhea

Ans A Chancroid is caused by Haemophilus ducreyi, a fastidious Gram negative bacillus.

124. **The ideal agent for treating fungal urinary tract infections is**

A. amphotericin B
B. **fluconazole**
C. voriconazole
D. micafungin
E. caspofungin

Ans B Concentrations of fluconazole are 10-20– fold higher in the urine than blood, making it an ideal agent for treating fungal urinary tract infections.

125. **Amantadine and rimantadine are effective only against influenza A viruses and are not approved for use in children younger than**

A. 6 mo
B. 18 mo
C. 3 yr
D. **5 yr**
E. 10 yr

Ans D

Gastroenteritis & Diarrhea

126. **Which of the following is the most common cause of meningoencephalitis in children?**

A. Mumps
B. Arbovirus
C. HSV
D. **Enterovirus**

Ans D Enterovirus is the most common cause of meningoencephalitis in children.

127. **Most common cause of neonatal diarrhea worldwide is:**

A. **Rotavirus**
B. Adenovirus
C. Norwalk virus
D. Cholera

Ans A In early childhood, the single most important cause of severe dehydrating diarrhea is rotavirus infection.

128. **All of the following are manifestations of congenital syphilis except?**

A. **Gumma**
B. Hutchinson's teeth
C. Olympian brow
D. Interstitial keratitis

Ans A Hutchinson's teeth, Olympian brow & Interstitial keratitis are features of congenital syphilis.

129. **In a 3yearold child, most common cause of pyomenin gitis:**

A. H. influenza
B. Staph aureus
C. **Strepto pneumoniae**
D. E. coli

Ans C

130. **Which of the following is a T-lymphocyte independent vaccine?**

A. hepatitis A
B. hepatitis B
C. **pneumococcal**
D. varicella
E. diphtheria

Ans C T-lymphocyte independent vaccines are associated with poor immune responses in children <2 yr of age, short-term immunity, and absence of an enhanced or booster response on repeat exposure to the antigen. With some polysaccharide vaccines, repeat doses actually are associated with reduced responses, as measured by antibody concentrations, compared to 1st doses (i.e., hyporesponsive).

131. **Rotavirus vaccine should not be initiated for infants older than**

A. 11 wk
B. 13 wk
C. **15 wk**
D. 17 wk
E. 19 wk

Ans C

132. **The final dose of rotavirus vaccine must be administered no later than**

A. 6 mo of age
B. **8 mo of age**
C. 10 mo of age
D. 12 mo of age
E. 18 mo of age

Ans B Two rotavirus vaccines are available, RotaTeq (RV5) and Rotarix (RV1). With both vaccines, the 1st dose can be administered as early as 6 wk of age and must be administered by 14 wk 6 days. The final dose in the series must be administered no later than 8 mo of age. The RV5 vaccine is administered in 3 doses at least 4 wk apart. The RV1 vaccine is administered in 2 doses at least 4 wk apart. Immunization should not be initiated for infants 15 wk of age and older as stated in the immunization schedule.

133.Oral rehydration is the mainstay of treatment for pediatric traveler's diarrhea and the drug of choice is

A. metronidazol
B. **azithromycin**
C. amoxicillin
D. trimethoprim-sulfamethoxazol
E. erythromycin

Ans B The drug of choice is azithromycin (10 mg/kg once daily for up to 3 days, with maximum daily dose of 500 mg). Ciprofloxacin (10 mg/kg per dose twice a day for up to 3 days, maximum dose of 500 mg twice a day) is an alternative for children >1 yr of age. Amoxicillin, trimethoprim-sulfamethoxazole (cotrimoxazole), and erythromycin should not be prescribed for self-treatment of traveler's diarrhea, because of widespread resistance among diarrheal pathogens.

134.Cholera is a dehydrating diarrheal disease caused by Vibrio cholerae, of more than 200 serogroups; the serogroups that have been associated with epidemics are

A. O131
B. **O139**
C. O151
D. O159
E. O165

Ans B Of the more than 200 serogroups, only serogroups O1 and O139 have been associated with epidemics, although some non-O1, non-O139 V.choleraestrains (e.g., O75 and O141) are pathogenic and can cause small outbreaks.

135.Seizure in cholera is commonly due to

A. hypocalcemia
B. hyponatremia
C. hypernatremia
D. **hypoglycemia**
E. fever

Ans D

136.Rotaviruses are in the Reoviridae family and cause disease in virtually all mammals and birds. All the following statements are true EXCEPT

A. infection typically begins after an incubation period of <48 hr (range: 1-7 days)
B. fever, vomiting and frequent watery stools are present in about 50-60% of cases
C. **vomiting and fever typically abate after the 4th day of illness**
D. dehydration may develop and progress rapidly particularly in infants
E. most severe disease typically occurs among children 4-36 mo of age

Ans C Vomiting and fever typically abate during the 2nd day of illness, but diarrhea often continues for 5-7 days.

137. The leading protozoal cause of diarrhea in children worldwide is

A. Entamoeba histolytica
B. Giardia lamblia
C. Balantidium coli
D. Isospora belli
E. **Cryptosporidium**

Ans E is recognized as a leading protozoal cause of diarrhea in children worldwide and is a common cause of outbreaks in childcare centers; it is also a significant pathogen in immunocompromised patients.

Hepatitis

138.Most common viral cause of acquired aqueductal stenosis is:

A. Rubella
B. **Mumps**
C. Toxoplasma
D. Enterovirus

Ans B CNS complications of Mumps include Meningoencephalitis, transverse myelitis, aqueductal stenosis & facial palsy.

139.Symptomatic neonatal CNS involvement is most commonly seen in which group of congenital intrauterine infection?

A. CMV and toxoplasmosis
B. Rubella and toxoplasmosis
C. **Rubella and HSV**
D. CMV and syphilis

Ans C Symptomatic CNS infections in neonatal period are most commonly seen in HSV (75–100%) f/b Rubella (30–60%); CMV, toxoplasma, and syphillis become symptomatic in less than 10%–15% of cases. Characteristic clinical features of Intrauterine Infections with Infection IUGR HSM Bleeding Eye signs CNS features Others Conge- 60–70% 2/3rd of "Blue berry "Cottage cheese with Microcephaly, periven- Anemia, sensorineural

140.Hutchinson's triad in congenital syphilis include:

A. **Interstitial keratitis, eight nerve involvement, Hutchinson's teeth**
B. Interstitial keratitis, mulberry teeth, periostitis
C. Periostitis, mulberry teeth, eight nerve involvement
D. Hutchinson's teeth, eighth nerve involvement, periostitis

Ans A

141. Most common organism in sickle cell anemia causing osteomyelitis is:

A. **Salmonella**
B. Streptococci
C. E. coli
D. Staphylococci

Ans A Harrison's latest 19th ed pg 634; CDC website Nelson 20th ed says: "Salmonella & Staph aureus are the 2 most common causes of osteomyelitis in children with sickle cell anemia"; So it does not help us with the answer to this question; Harrison Latest 19th ed says: "Unusual susceptibility to osteomyelitis, which may be caused by organisms, such as Salmonella, is seen in sickle cell disease". Wintrobes Hematology also says: "Although >80% of hematogenous osteomyelitis in the general population is caused by

142. **Immunization is one of the most beneficial and cost-effective disease prevention measures. As a result of effective and safe vaccines, which of the following diseases has been eradicated?**

A. **smallpox**
B. polio
C. measles
D. rubella
E. pertussis

Ans A As a result of effective and safe vaccines, smallpox has been eradicated; polio is close to worldwide eradication.

143. **Hepatitis A vaccine, licensed for administration to children 12 mo of age and older. The 2 doses in the series should be separated by at least**

A. 2 mo
B. 4 mo
C. **6 mo**
D. 1 yr
E. 2 yr

Ans C

144. **The minimum age for the last dose of hepatitis B vaccine is**

A. 16 weeks
B. 20 weeks
C. **24 weeks**
D. 28 weeks
E. 32 weeks

Ans C The minimum interval between 2nd and 3rd dose is 8 weeks andat least 16 weeks after 1st dose. Minimum age for the last dose is 24 weeks.

145. **The MOST important clinical manifestation of M.catarrhalis infection in children is**

A. pneumonia
B. bronchitis
C. **otitis media**
D. pharyngitis
E. cystitis

Ans C The most important clinical manifestation of M.catarrhalis infection in children is otitis media. Colonization and infection with M. catarrhalis are increasing in countries in which pneumococcal conjugate vaccines are used widely.

146. **In neonates with HSV infection including CNS involvement, to improve neurodevelopmental outcome, suppressive therapy with oral acyclovir should be used for**

A. 3 mo
B. **6 mo**
C. 9 mo
D. 12 mo
E. 18 mo

Ans B The use of suppressive therapy with oral acyclovir for 6 mo has been demonstrated to improve neurodevelopmental outcome.

147. **Typically, the first sign of infection in infants with respiratory syncytial virus (RSV) is**

A. **rhinorrhea**
B. cough
C. low-grade fever
D. increased respiratory rate
E. subcostal retractions

Ans A

HIV & AIDS

148. **Preferred diagnostic test used for HIV detection in neonate born by caesarean section whose mother is HIV positive:**

A. **HIV DNA PCR**
B. ELISA
C. HIV culture
D. HIV P24 antigen assay
E. HIV RNA PCR

Ans A c. HIV culture, e. HIV RNA PCR Diagnosis of HIV in infants & children
• Presence of IgA or IgM antiHIV in the infant's circulation can indicate HIV infection
• In any child > 18 months age, demonstration of IgG antibody to HIV by ELISA & Western blot test establishes HIV infection
• Breastfed infants should have antibody testing performed 12 wk following cessation of breastfeeding
• HIV DNA or RNA PCR or HIV culture, are useful in young infants, to reach to a definitive diagnosis

149. **Best method to diagnose HIV in an infant?**

A. Western blot
B. ELISA
C. **PCR**
D. All

Ans C Ref: Update on antiretroviral regimens for treating and preventing HIV infection and update on early infant diagnosis of HIV July 2018 policy brief HIV treatment–interim guidance
• WHO recommends that HIV virological testing (HIV DNA or RNA PCR or HIV culture) be used to diagnose HIV infection among infants and children younger than 18 months
• In any child > 18 mo of age, demonstration of IgG antibody to HIV by ELISA & Western blot test establishes HIV infection

150. **Which of the following is FALSE regarding antiretroviral regime for treating infants born to HIV Infected women?**

A. Daily Nevirapine prophylaxis at birth and continued for 6 weeks if mother received ART regularly in antenatal period
B. Daily Nevirapine prophylaxis at birth and continued for 12 weeks if mother presented directly in labor ,without evidence of adequate treatment of ART (alteast 24 weeks) or did not receive any ART earlier
C. **Daily Nevirapine prophylaxis at birth and continued for 12 weeks if mother received ART regularly in antenatal period**
D. Dose of Nevirapine for treating infants born to HIV infected women is 2 mg/kg once daily in babies with birth weight <2000 g

Ans C Daily Nevirapine prophylaxis at birth and continued for 12 weeks if mother received ART regularly in antenatal period Nevirapine prophylaxis is to be given at birth & continued for 6 weeks (not 12 weeks) if mother received ART regularly in antenatal period

151. As per WHO classification of immunosuppression, how much is the CD4 level in HIV/AIDS children with severe stage in age group of 36 – 59 months:

A. < 35%
B. < 25%
C. < 20%
D. **< 15%**

Ans D CDC guidelines 2014; Severity of immune suppression based on CD4 levels children HIV-associated Age-related CD4 + cell values immunodeficiency ≤ 11 month 12–35 month 36–59 month ≥ 5 year Mild 30–35% 25–30% 20–25% 350–499 cells/ mm3 Advanced 25–30% 20–25% 15–20% 200–349 cells/mm3 cells/mm3 dna Severe < 25% or < 1500 cell/ < 20% of < 750 cells/ < 15% or < 350 cells/ < 200 or mm3 mm3 mm3 < 15% Note: The answer given in the Maharashtra PG CET 2016 key was <15%, as per the above grading, which was previously used;

152. Which of following is NOT an AIDS defining illness?

A. **Anal cancer**
B. Hodgkins lymphoma
C. BurKitt's lymphoma
D. Kaposi sarcoma
E. Toxoplasmosis of brain

Ans A b. Hodgkins lymphoma Important AIDS defining conditions according to CDC are:
• Candidiasis of bronchi, trachea, lungs or esophagus • Histoplasmosis, disseminated or extrapulmonary fo • Coccidioidomycosis, Cryptococcosis, Cryptosporidiosis or • Kaposi sarcoma/Burkitt lymphoma/Primary CNS Lymphoma Isosporiasis • Lymphoid interstitial pneumonia or Pneumocystis jirovecii pneu monia
• CMV disease (other than liver, spleen, nodes), CMV retinitis • Mycobacterium avium complex or Mycobacterium kansasii (extra

153. If HIV infected mother has received only AZT during antenatal period, the antiretroviral drug indicated for breastfeeding infant is?

A. **Nevirapine**
B. AZT
C. 3TC
D. d4T

Ans A

154. True about HIV infection in infants:

A. Mainly occur through horizontal transmission
B. **HIV DNA PCR positive at 3 months confirms diagnosis**
C. Positive antibody test for IgG antibody at 3 month confirms infected infant
D. Passive transfer of maternal antibody generally persists for 12 months

Ans B HIV DNA PCR positive at 3 month confirms diagnosis and d. Passive transfer of maternal body generally persists for 12 months

155. Parotid enlargement in a HIV infected child is char Questions acterized in which stage of AIDS, according to WHO:

A. Stage
B. **Stage**
C. Stage
D. Stage

Ans B 2 WHO 2016 ART guidelines; In WHO Clinical Stage 1, the patient is asymptomatic or has persistent generalised lymphadenopathy Unexplained persistent parotid enlargement is included in WHO Clinical Stage 2;

156. Most common symptom of AIDS in infant is:

A. GI infection
B. Persistent cough
C. Rashes
D. **Lymphadenopathy**

Ans D • In most infants with HIV infection, physical examination at birth is normal.
• Initial symptoms may be subtle, such as lymphadenopathy and hepatosplenomegaly, or nonspecific, such as failure to thrive, chronic or recurrent diarrhea, respiratory symptoms, or oral thrush, distinguishable only by their persistence.

157. According to WHO the major sign of AIDS in children in stage I is the following:

A. **Generalised lymphadenopathy**
B. Not gaining weight
C. Oral candidiasis
D. All of above

Ans A 54. None NACO 2013 recommendations for ART in infants & children: Age Clinical stage Immunological < 24 months Treat all > 24 months WHO stage 3 & 4 Treat all WHO stage 1 & 2 Treat when CD4 count at 2-5 years is < 750/mm3 & at > 5 years: < 350/mm3 As per Latest WHO recommendations (2016), ART should be initiated in all children living with HIV, regardless of WHO clinical stage or at any CD4 cell count; 55. None;

158. True statement(s) regarding feeding of HIVinfected child is/are:

A. Breastfeeding for 4–6 months then start weaning
B. Breastfeeding for 1 year then start weaning
C. Exclusively top feeding
D. **Breastfeeding for 6 months and then rapid weaning**
E. Mixed feeding should be done

Ans D Older national guidance for HIV-exposed infants recommended exclusive breastfeeding for around 6 months followed by relatively rapid weaning, to balance the nutritional/ immune benefits of breast milk against the ongoing risk of transmission (current WHO guidance instead favors continued breastfeeding with ART coverage).

159. **Drugs used to prevent HIV from mother to child:**

A. Zidovudine to mother
B. Nevirapine to mother
C. **Nevirapine to baby up to 6 weeks**
D. Zidovudine to baby (PGI Dec 08)

Ans C d. Zidovudine to baby ART in antenatal period:
• Start ART as soon as possible & continue throughout pregnancy, delivery, breastfeeding period and thereafter lifelong
• Even if the pregnant women presents very late in pregnancy, ART should be initiated promptly
• Recommended 1stline regimen in pregnancy is Tenofovir (300 mg) + Lamuvidine (300 mg) + Efavirenz (600 mg)

160. **Vaccines contraindicated in HIV positive child:**

A. **OPV**
B. MMR (PGI Dec 08)
C. Rabies
D. Influenza
E. Hepatitis

Ans A IAP, WHO, AAP, ACIP, and CDC recommend: • All the live vaccines in asymptomatic HIV1 infected children except BCG and OPV
• However, in a symptomatic child all live vaccines are forbidden
• Yellow fever vaccine is contraindicated in both symptomatic and asymptomatic.

161. **For the prevention of parent to child transmission of HIV, the NACO's recommendation is to give:**

A. Nevirapine 200 mg in active labour to mother
B. Nevirapine 200 mg four hours after rupture to membranes, to mother172 Section 1: General Pediatrics
C. Nevirapine 200 mg in active about to mother and syrup nevirapine 2 mg/kg body weight to newborn with 72 hours of delivery
D. **Syrup Nevirapine 2 mg/kg body weight to newborn within 72 hours of birth**

Ans D Syrup Nevirapine 2 mg/kg body weight to newborn within 72 hours of birth WHO guidelines 2014;

162. **Which drug is given to the baby to prevent HIV transmission from mother to child?**

A. **ELISA for HIV IgG antibody**
B. p24 antigen
C. Virus culture
D. ELISA for HIV IgA antibody

Ans A Nevirapine

163. **All of the following methods are used for the diagnosis of HIV infection in a 2 months old child, except:**

A. DNA-PCR
B. Viral culture
C. **HIV ELISA**
D. p24 antigen assay OTHER VIRAL INFECTIONS

Ans C HIV

Polio & Enteroviruses

164. **Appropriate therapy with antiretroviral agents results in a syndrome characterized by an increased inflammatory response from the recovered immune system to subclinical opportunistic infection. What is this syndrome called as?**

A. HIV related hyper- reactivity syndrome
B. AIDS related inflammatory syndrome
C. **Immune reconstitution inflammatory syndrome**
D. CD4 responsive inflammatory syndrome

Ans C Immune reconstitution inflammatory syndrome (IRIS)
• Characterized by increased inflammatory response subclinical opportunistic infections
• It is more common in patients with progressive disease & severe CD4 + T-lymphocyte depletion.
• There is fever & worsening of clinical manifestations of opportunistic infection or new manifestations (e.g., enlargement of lymph nodes, pulmonary infiltrates), typically within the 1st few weeks after initiation of antiretroviral therapy.

165. **Hand foot mouth disease is usually caused by:**

A. **Coxsackie A**
B. Adenovirus
C. Rotavirus
D. Hepatitis B virus

Ans A

166. **True for polio virus is:**

A. VDPV is usually due to type
B. **Wild polio virus mostly is type**
C. VAPP is usually due to type
D. Bivalent Vaccine does not include type
E. Wild polio type 2 is eradicated since 1999

Ans B 1, d. Bivalent does not include type 2, e. Wild polio type 2 is eradicated since 1999 a. False Since 2006, majority of cVDPV cases are due to type 2. b. True Type 2 poliovirus was declared eradicated in September 2015, with the last virus detected in India in 1999. Type 3 wild poliovirus has not been detected anywhere in the world since November 2012. c. False Type 3 has been more often associated with recipient VAPP, type 2 has been more often observed in immunodeficient

167. **True about polio:**

A. Paralytic polio is most common
B. Spastic paralysis
C. **IM injections and increased muscular activity lead to increased paralysis**
D. Polio drop given only in < 3 year

Ans C a. False 90-95% of polio infections are asymptomatic b. False Acute flaccid paralysis is seen in Polio & not spastic paralysis c. True Risk of paralytic polio is increased by Tonsillectomy, Strenous physical exercise, Tooth extraction, Cortisone administration, Adenoidectomy, Injection (intramuscular) d. False OPV drops are given to all children less than 5 yr age in pulse polio immunisation

168. **The MOST common adverse reaction to intramuscular immunoglobulin is**

A. **pain at the injection site**
B. flushing
C. headache
D. chills
E. nausea

Ans A The most common adverse reaction to immunoglobulin is pain and discomfort at the injection site and, less commonly, flushing, headache, chills, and nausea.

169. **Ingestion of dirt is an important clue to infection with**

A. amebiasis
B. giardiasis
C. malaria
D. trichinosis
E. **toxoplasmosis**

Ans E Ingestion of dirt is a particularly important clue to infection with Toxocara canis (visceral larva migrans) or Toxoplasma gondii (toxoplasmosis).

170. **Kawasaki disease closely resembles toxic shock syndrome clinically. However, many of the clinical features of toxic shock syndrome are usually absent or rare in Kawasaki disease like**

A. fever unresponsive to antibiotics
B. hyperemia of mucous membranes
C. erythematous rash
D. desquamation
E. **diffuse myalgia**

Ans E Many of the clinical features of TSS are usually absent or rare in Kawasaki disease, including diffuse myalgia, vomiting, abdominal pain, diarrhea, azotemia, hypotension, acute respiratory distress syndrome, and shock.

171. **All the following are clinical features of shigellosis EXCEPT**

A. an incubation period of 12 hr to several days
B. most children never progress to the stage of bloody diarrhea
C. **untreated diarrhea can last more than 4 weeks**
D. neurologic findings are the most common extraintestinal manifestations
E. neonatal shigellosis is rare

Ans C Untreated diarrhea can last 7-10 days; only approximately 10% of patients have diarrhea persisting for longer than 10 days. Persistent diarrhea occurs in malnourished infants, children with AIDS, and occasionally previously normal children.

172. **Acyclovir is a safe and effective therapy for herpes simplex virus (HSV) infections. All the following are true EXCEPT**

A. activity against CMV is less pronounced
B. activity against Epstein-barr virus is modest, both in vitro and clinically
C. acyclovir therapy in a nursing mother is not a contraindication to breastfeeding
D. **main route of elimination is hepatic**
E. high doses of acyclovir are associated with neurotoxicity

Ans D The main route of elimination is renal, and dosage adjustments are necessary for renal insufficiency. Toxicity is observed typically only in exceptional circumstances: for example, if administered by rapid infusion to a dehydrated patient or a patient with underlying renal insufficiency, acyclovir can crystallize in renal tubules and produce a reversible obstructive uropathy.

173. **For military services, vaccines are available for human adenoviruses types**

A. 1 and
B. 2 and
C. 3 and
D. **4 and**
E. 5 and

Ans D Vaccines are available for HAdV types 4 and 7, but are used only for military populations.

Diphtheria, Pertussis & Tetanus

174. **Indications to start ART in children under NACO all except:**

A. **<11 months infant if CD4 Count <1500 cells/mm3**
B. 12–35 months if CD4 Count <500 cells/mm3
C. 36–59 months if CD4 Count <350 cells/mm3
D. >5 years if CD4 Count <200 cells/mm3

Ans A Under NACO/WHO pediatric ART guidelines, infants under 12 months are started on ART regardless of CD4 count or clinical stage — treatment is not conditional on a CD4 threshold at this age.

175. **True regarding infectious mononucleosis are all except:**

A. Drug induced rash
B. **Agranulocytosis**
C. Pharyngeal oedema
D. Fatigue

Ans B

176. **Late complication of diphtheria is:**

A. Airway obstruction
B. Encephalitis
C. Pneumonia
D. **Neuropathy**

Ans D Cranial neuropathies Characteristically occur in the 5th wk, leading to oculomotor and ciliary paralysis, which can cause strabismus, blurred vision, or difficulty with accommodation.

177. **The special stain used to identify C. diphtheriae in a throat swab is?**

A. Albert's stain
B. Giemsa stain
C. Gram's stain
D. India ink

Ans A

178. **All of the following statements are true about diphtheria except:**

A. Dose of antitoxin depends on virulence of organism
B. Myocarditis is a dreaded complication
C. Death is due to circulatory failure
D. Pseudomembranes may be found on the vulva/perianal membrane CONGENITAL SYPHILIS

Ans A Dose of antitoxin depends upon the virulence of the organism. Diphtheria Antitoxin is administered as a single empirical dose of 20,000- 100,000 units based on the degree of toxicity, site and size of the membrane, and duration of illness.

179. **Drug of choice for pertussis is?**

A. Penicillin
B. Ceftriaxone
C. Erythromycin
D. Azithromycin

Ans D Macrolides are preferred agents; Due to increased risk of infantile hypertrophic pyloric stenosis in neonates treated with oral erythromycin, Azithromycin is the preferred agent in all age groups.

180. **Incubation period of pertussis is:**

A. One week
B. Two weeks
C. Three weeks
D. Four weeks OTHER BACTERIAL INFECTIONS

Ans A Incubation period for Pertussis ranges from 3-12 days. OTHER BACTERIAL INFECTIONS 135. > 50% immature neutrophils dna It is an inflammatory cascade that is initiated by the host response to an infectious or noninfectious trigger. 2 of 4 criteria, 1 of which must be abnormal temp. or abnormal WBC count: Core temperature >38.5°C (101.3°F) or <36°C (96.8°F) (rectal, bladder, oral, or central catheter) 2. Tachycardia:

181. **True about tetanus:**

A. Tetanus bacilli can spread through blood
B. Spread along the nerves
C. Requires oxygen
D. Incubation period is variable
E. Causes deep wound invasion CONGENITAL TOXOPLASMOSIS

Ans B Spreads along the nerves, d. Incubation period is variable. Option a False Tetanus bacillus is not an invasive organism Option b True Tetanus toxin binds at neuromuscular junction & enters motor nerve by endocytosis, after which it under- goes retrograde axonal transport to cytoplasm of neuron Option c False Clostridium tetani, is a motile, Gram-positive, spore-forming obligate anaerobe, so does not require O

182. **Diphtheria is an acute toxic infection; toxic cardiomyopathy occurs in 10-25% of patients with respiratory diphtheria and is responsible for 50-60% of deaths. Of the following, the mainstay of therapy is**

A. antitoxin
B. penicillins
C. erythromycin
D. clindamycin
E. rifampin

Ans A Specific antitoxin is the mainstay of therapy and should be administered on the basis of clinical diagnosis. The role of antimicrobial therapy is to halt toxin production, treat localized infection, and prevent transmission of the organism to contacts.

183. **Infants younger than 3 mo of age with suspected pertussis usually are admitted to hospital, as are many between 3 and 6 mo of age unless witnessed paroxysms are not severe, as well as are patients of any age if significant complications occur. Typical paroxysms that are not life threatening have the following feature**

A. duration >45 sec
B. blue color change
C. bradycardia <60 beats/min in infants
D. oxygen desaturation that spontaneously resolves at the end of the paroxysm
E. post-tussive unresponsiveness

Ans D Typical paroxysms that are not life threatening have the following features: duration <45 sec; red but not blue color change; tachycardia, bradycardia (not <60 beats/min in infants), or oxygen desaturation that spontaneously resolves at the end of the paroxysm; whooping or strength for brisk self-rescue at the end of the paroxysm; self-expectorated mucus plug; and post-tussive exhaustion but not unresponsiveness.

184. **All the following regarding assessment and care of infants with pertussis are true**

A. infants with potentially fatal pertussis may appear well between episodes
B. a paroxysm must be witnessed before a decision is made between hospital and home care
C. suctioning of nose, oropharynx, or trachea should be performed on a preventive schedule
D. feeding in the period following a paroxysm may be more successful than after napping
E. family education, recruitment as part of the team, and continued support after discharge are essential

Ans C Suctioning of nose, oropharynx, or trachea should not be performed on a “preventive” schedule.

185. **Hospital discharge of infants with pertussis is appropriate in all the following circumstances EXCEPT**

A. over a 24-hr period disease severity is unchanged or diminished
B. intervention is not required during paroxysms
C. nutrition is adequate
D. no complication has occurred
E. parents are adequately prepared for care at home

Ans A Over a 48-hr period disease severity is unchanged or diminished.

186. **The preferable antibiotic in neonate with pertussis is**

A. azithromycin
B. erythromycin
C. clarithromycin
D. trimethoprim-sulfamethoxazole
E. amoxycilline

Ans A o Azithromycin: Recommended agent 10 mg/kg/day in a single dose for 5 days o Erythromycin: Not preferred o Clarithromycin: Not recommended o trimethoprim-sulfamethoxazole: Contraindicated for infants <2 mo of age

187. **Echocardiography should be performed in critically ill infants with pertussis to detect presence of**

A. myocarditis
B. heart failure
C. congenital heart defect
D. pulmonary hypertension
E. right atrial dilatation

Ans D Progressive pulmonary hypertension in very young infants and secondary bacterial pneumonia are severe complications of pertussis and are the usual causes of death. Echocardiography should be performed in critically ill infants with pertussis to detect presence of pulmonary hypertension and to intervene expeditiously.

188. **Tetanus is an acute spastic paralytic illness historically called lock-jaw that is caused by the neurotoxin produced by Clostridium tetani. Of the following, the TRUE statement is**

A. tetanus is most often localized
B. incubation period typically is 2-6 weeks
C. patient remains conscious and there is no pain
D. tetanic paralysis becomes more severe in the 4th wk after onset
E. cephalic tetanus occurs in association with chronic otitis media

Ans E Tetanus is most often generalized but may also be localized. The incubation period typically is 2-14 days but may be as long as months after the injury. Because tetanus toxin does not affect sensory nerves or cortical function, the patient unfortunately remains conscious, in extreme pain, and in fearful anticipation of the next tetanic seizure. The tetanic paralysis usually becomes more severe in the 1st wk after onset, stabilizes in the 2nd wk, and ameliorates gradually over the ensuing 1-4 wk.

Streptococcal & Staphylococcal Infections

189. **Prophylaxis with Cotrimoxazole is recommended in the following situation except:**

A. All symptomatic HIV infected children > 5 years of age irrespective of CD4
B. All HIV exposed infants till HIV infection can be ruled out
C. All HIV infected infants less than 1 year age irrespective of symptoms or CD4 counts
D. As secondary prophylaxis after initial treatment for pneumocystis carini pneumonia

Ans A Cotrimoxazole prophylaxis in children over 5 years is guided by CD4 count/clinical staging rather than given irrespective of CD4; it is otherwise indicated for HIV-exposed infants until infection is excluded, all infected infants under 1 year, and as secondary PCP prophylaxis.

190. **Which of the following is true about roseola infantum?**

A. Defervescence following the rash
B. Caused by HHV 6 and
C. Slapped cheek appearance is seen
D. Otitis media is common complication
E. Rash appear first on face and neck

Ans B 7 & d. Otitis media is common complication

191. **All of the following are true regarding congenital syphilis except:**

A. Rhinitis is an early sign
B. Notched incisors
C. Conductive hearing loss
D. Linear scars at the angles of mouth

Ans C Sensorineural hearing loss (due to 8th CN involvement) is seen in Syphilis;

192. **Symptoms suggestive of Scarlet fever are all except:**

A. Gooseflesh
B. Nagayama spots
C. Desquamation of palms
D. Strawberry tongue

Ans B Scarlet fever
- Caused by Group A Streptococcus
- Pharyngitis & Strawberry tongue is seen
- Erythematous rash appears 24-48 hr after onset
- Skin has a goose-pimple appearance and feels rough
- Desquamation of palms & soles seen
- Identification of GAS in the pharynx confirms the diagnosis

Roseola infantum
- Also known as exanthemsubitum and sixth disease
- Caused by human herpesvirus 6 or 7 (MC in 6 months to 4 years age)
- Begins with a sudden high fever (101-105°F) that lasts for 2-5 days

193. **All of the following are major criteria for staphylococcal toxic shock syndrome, except:**

A. Acute fever (Temperature >38.8 deg C (101.8 deg F)
B. Central nervous system abnormalities
C. Hypotension (Orthostatic. shock, blood pressure below age-appropriate norms)
D. Rash

Ans B Mnemonic for Major Diagnostic Criteria of Staphylococcal Toxic Shock Syndrome: "H-A-R jagah Staphylococcus hai" → Hypotension, acute fever and rash

194. **Bulbar conjunctivitis in a child with FUO suggests**

A. leptospirosis
B. coxsackievirus infection
C. tuberculosis
D. infectious mononucleosis
E. lymphogranuloma venereum

Ans A Palpebral conjunctivitis in a febrile patient may be a clue to measles, coxsackievirus infection, tuberculosis, infectious mononucleosis, lymphogranulomavenereum, or cat-scratch disease. In contrast, bulbar conjunctivitis in a child with FUO suggests Kawasaki disease or leptospirosis.

195. **Cephalexin is a cephalosporin active against S. aureus, Streptococcus,**

A. **1st**
B. 2nd
C. 3rd
D. 4th
E. 5th

Ans A Dose: 25-100 mg/kg/24 hr divided q 6-8 hr PO.

196. **Which of the following represent a major criterion for the diagnosis of staphylococcal toxic shock syndrome?**

A. **rash**
B. conjunctival hyperemia
C. myalgia
D. thrombocytopenia
E. vomiting

Ans A MAJOR CRITERIA (ALL REQUIRED) 1. Acute fever; temperature >38.8°C (101.8°F) 2. Hypotension 3. Rash (erythroderma with convalescent desquamation) MINOR CRITERIA (ANY 3 OR MORE) 1. Mucous membrane inflammation (vaginal, oropharyngeal or conjunctival hyperemia, strawberry tongue) 2. Vomiting, diarrhea 3. Liver abnormalities (bilirubin or transaminase greater than twice upper limit of normal) 4.

197. **Group A streptococcus can be subdivided into more than 220 serotypes on the basis of the M protein antigen, M serotyping is valuable for epidemiologic studies; specific group A streptococcus diseases tend to be associated with certain M types. Of the following, the M type associated with glomerulonephritis is**

A. 1
B. 6
C. **12**
D. 18
E. 29

Ans C A pharyngeal strains (e.g., M type 12) are associated with glomerulonephritis; skin strains (e.g., M types 49, 55, 57, and 60) are considered nephritogenic. Several pharyngeal serotypes (e.g., M types 1, 3, 5, 6, 18, 29), but no skin strains, are associated with acute rheumatic fever.

198. **Scarlet fever is an upper respiratory tract infection associated with a characteristic rash. Of the following, which statement is TRUE?**

A. it is caused by an infection with pyrogenic endotoxin producing group A streptococcus
B. the rash appears 96 hr after onset of symptoms
C. the rash begins to fade after 1-2 weeks
D. before desquamation, the reddened papillae are prominent, giving the tongue a strawberry appearance
E. **the milder form can be confused with Kawasaki disease**

Ans E It is caused by an infection with pyrogenic exotoxin (erythrogenic toxin)– producing GAS. The rash appears within 24-48 hr after onset of symptoms, although it may appear with the first signs of illness. After 3-4 days, the rash begins to fade and is followed by desquamation. After desquamation, the reddened papillae are prominent, giving the tongue a strawberry appearance. The milder form can be confused with viral exanthems, Kawasaki disease, and drug eruptions.

199. **Impetigo (or pyoderma) has traditionally been classified into 2 clinical forms: bullous and nonbullous. Of the following, which statement is TRUE?**

A. bullous impetigo is more common
B. nonbullous lesions are most common on the trunk and perineum
C. **regional lymphadenitis is commonly associated with nonbullous lesions**
D. nonbullous impetigo is generally accompanied by fever
E. bullous impetigo usually involve the face and extremeties

Ans C Nonbullous impetigo is the more common form. The lesions may occur anywhere but are most common on the face and extremities. Regional lymphadenitis is common. Nonbullous impetigo is generally not accompanied by fever or other systemic signs or symptoms. Bullous impetigo is less common and occurs most often in neonates and young infants. The usual distribution involves the face, buttocks, trunk, and perineum.

200. **Because no clinical or laboratory finding is pathognomonic for acute rheumatic fever, 5 major and 4 minor criteria with evidence of recent group A streptococcus infection are required for the diagnosis. Migratory polyarthritis represent one of the major criteria, which of the following statements is TRUE regarding this major criterion?**

A. occurs in approximately 50% of patients
B. typically involves small joints
C. **the pain can precede and can appear to be disproportionate to the objective findings**
D. rheumatic arthritis is almost deforming
E. there is often a proportional relationship between the severity of arthritis and the severity of cardiac involvement

Ans C Arthritis occurs in approximately 75% of patients with acute rheumatic fever and typically involves larger joints, particularly the knees, ankles, wrists, and elbows. Involvement of the spine, small joints of the hands and feet, or hips is uncommon. Rheumatic arthritis is almost never deforming. There is often an inverse relationship between the severity of arthritis and the severity of cardiac involvement.

201. **To prevent first attack of acute rheumatic fever after acute group A streptococcus pharyngitis, appropriate antibiotic therapy should be instituted before**

A. 3rd day of illness
B. 5th day of illness
C. 7th day of illness
D. **9th day of illness**
E. 11th day of illness

Ans D

202.Group B streptococcus (GBS), or Streptococcus agalactiae, is a major cause of neonatal bacterial sepsis. Of the following, the MOST common syndrome associated with childhood GBS disease beyond early infancy is

A. bacteremia without a focus
B. meningitis
C. ventriculitis
D. septic arthritis
E. pneumonia

Ans A Early-onset neonatal GBS disease presents as sepsis; pneumonia and meningitis. Late-onset neonatal GBS disease most commonly manifests as bacteremia (45-65%) and meningitis (25-35%). Invasive GBS disease in children beyond early infancy is uncommon. Bacteremia without a focus is the most common syndrome associated with childhood GBS disease beyond early infancy.

203.Recommended duration of therapy for manifestations of group B streptococcus are as follow

A. bacteremia without a focus: 7 days
B. meningitis: 3-6 weeks
C. ventriculitis: at least 8 weeks
D. septic arthritis: 3-4 weeks
E. osteomyelitis: 6-8 weeks

Ans D o bacteremia without a focus 10 days o meningitis 2-3 wk o ventriculitis at least 4 wk o septic arthritis 3-4 wkase o osteomyelitis 3-4 wkas

Sepsis & Septicemia

204.Perinatal prevention of mother to child, which of the following steps are useful?

A. Cleaning mother's vagina with antiseptic lotion
B. Elective Caesarian section
C. Avoid breast feeding
D. ART prophylaxis

Ans A d. ART prophylaxis 237; Latest Updated guidelines for PPTCT by NACO, December 2013. This has become a controversial question now. As per previous PPTCT guidelines, Elective caesarean section helped in preventing perinatal transmission of HIV. But as per the latest recommendations, Caesarean sections in HIV positive pregnant women should be performed for obstetric indications only;

205.Which of the following is true about erythema infec tiosum?

A. Slapped cheek appearance seen
B. Caused by parvovirus
C. Defervescence before rash
D. Rash initially appears on trunk
E. Known as 'fifth disease'

Ans A b. Caused by parvovirus and e. Known as 'fifth disease' 1571, Ghai 8/e p 216;

206.Distinctive triad of interstitial keratitis, Hutchinson teeth and eighth nerve deafness is manifestation of

A. Primary syphilis
B. Secondary syphilis
C. Tertiary syphilis
D. Congenital syphilis

Ans D

207.A 6monthsold baby was brought with complaints of difficulty in feeding. The child was found to be hypotonic with a weak gag. The child is on breast milk and mother also gives honey to the child during periods of excessive crying. The causative agent is:

A. Gram positive aerobic coccus
B. Gram positive anaerobic spore bearing bacillus
C. Toxin produced by gram positive anaerobic bacillus
D. Echovirus

Ans C • Food poisoning due to ingestion of honey is caused by Clostridium botulinum
• Clostridium botulinum is a gram positive spore forming anaerobic bacilli, which acts through botulinum toxin.

208.All the following are major recommended indications for IVIG EXCEPT

A. replacement therapy for primary immunodeficiency disorders
B. Kawasaki disease
C. hepatitis A prophylaxis
D. immune-mediated thrombocytopenia
E. prophylaxis of infection following bone marrow transplantation

Ans C In hepatitis A prophylaxis, intramuscular IG is recommended.

209.Multiple blood cultures may be required to detect bacteremia associated with

A. malaria
B. pyelonephritis
C. brucellosis
D. osteomyelitis
E. pneumonia

Ans D Multiple or repeated blood cultures may be required to detect bacteremia associated with infective endocarditis, osteomyelitis, or deep-seated abscesses.

210.Acute rheumatic carditis usually presents as cardiac murmurs and

A. tachycardia
B. chest pain
C. dyspnea
D. syncopal attack
E. tachypnea

Ans A Acute rheumatic carditis usually presents as tachycardia and cardiac murmurs, with or without evidence of myocardial or pericardial involvement.

211. Complications of campylobacter jejuni can include acute and late onset complications that may present after the acute infection has resolved. Of the following, the MOST common late-onset complication is

A. reactive arthritis
B. immunoglobulin A nephropathy
C. immune complex glomerulonephritis
D. hemolytic anemia
E. carditis

Ans A The most common late-onset complications include reactive arthritis and Guillain-Barré syndrome.

212.Ribavirin is a guanosine analog that has broad-spectrum activity against a variety of viruses, particularly RNA viruses. All the following are true EXCEPT

A. its precise mechanism of action is incompletely understood
B. aerosolized ribavirin effective for parainfluenza, influenza, and measles infections
C. ribavirin is generally nontoxic, particularly when administered by aerosol
D. **ribavirin and its metabolites concentrate in hepatocytes**
E. conjunctivitis and bronchospasm have been reported following exposure to aerosolized drug

Ans D Ribavirin and its metabolites concentrate in red blood cells and can persist for several weeks and, in rare instances, may be associated with anemia.

213.Respiratory tract infections are common manifestations of human adenoviruses (HAdV) infections in children and adults. All the following are true EXCEPT

A. HAdVs cause 5-10% of all childhood respiratory disease
B. primary infections in infants may manifest as bronchiolitis or pneumonia
C. HAdV pneumonia may manifest as features more typical of bacterial disease
D. pharyngitis typically includes symptoms of coryza, sore throat, and fever
E. **HAdV can be identified in <5% of children with isolated pharyngitis**

Ans E The virus can be identified in 15-20% of children with isolated pharyngitis, mostly in preschool children and infants.

Congenital & Neonatal Infections

214.Unexplained persistent parotid enlargement in a malnourished child is due to

A. **HIV**
B. Leukemia
C. Infectious mononucleosis
D. Mumps

Ans A HIV with

215.A child presented with Microcephaly, Hepatomegaly and periventricular calcification. Best site from where specimen should be obtained for diagnosis of CMV by PCR?

A. **Urine**
B. Blood
C. Liver biopsy
D. CSF

Ans A Oski's Pediatrics 4/e p 514 The diagnosis of congenital CMV infection is best confirmed by isolation of the virus from urine PCR assays of dried blood spots from infants have not been shown to have sufficient sensitivity for the identification of most infants with congenital CMV infection Saliva PCR assays are currently being assessed as a useful screening method for congenital CMV infection.

216.Which of the following is true regarding congenital CMV infection?

A. Diagnosed only by persistent presence of IgM antibody after 6 months
B. **It is the most common cause of non-syndromic sensori-neural hearing loss**
C. All babies born are symptomatic
D. Mothers of developing countries who transmit the virus are usually symptomatic

Ans B It is the most common cause of nonsyndromic sensorineural hearing loss • Diagnosis of congenital CMV infections requires the recovery of replicating virus and/or viral nucleic acids within 1st 3 wk of life; IgM assays can also be used, but they have limited sensitivity.
• Congenital infection with CMV presents with symptoms in only 10% of infected newborns;
• Hearing loss is the most common long-term sequela associated with congenital CMV infection;
• Most common infectious cause of congenital SNHL is CMV;

217. Which of the following does not establish a diagnosis of congenital CMV infection in a neonate?

A. Urine culture of CMV
B. **IgG CMV antibodies in blood**
C. Intra-nuclear inclusion bodies in hepatocytes
D. CMV viral DNA in blood by polymerase chain reaction

Ans B Diagnosis of Congenital CMV infection Virus isolation Serology PCR for viral genome (DNA) Definitive diagnostic method Not very useful Urine or Blood may be used; Specimens that can be used are urine, IgM test can be done but lacks sensitivity Quantitative estimation possible saliva, blood, Bronchoalveolar washing & and specificity Amniotic fluid IgG tests are not diagnostic About Option C: CMV infected cells contain large intranuclear and smaller intracytoplasmic inclusions which are pathognomic.

218.In neonatal herpes, true is:

A. **Caused by HSV-II**
B. Hepatosplenomegaly is diagnostic
C. Spontaneous recovery occurs
D. If not treated, disseminates TUBERCULOSIS

Ans A Caused by HSVII and d. If not treated, disseminates Neonatal Herpes
• It is commonly caused by HSV-2 (70% cases are caused by HSV2 and 30% caused by HSV1)
• Neonatal infections usually present on 6th day postpartum can present as: Localized skin, eye and mouth infection, CNS infection, or disseminated infection; vesicular ulcerative skin lesions occur in only 30–43% children; weiveR • Pathogenesis of HSV infection in newborns is complicated by their relative immunologic immaturity.

219.Diagnosis of toxoplasmosis in newborn is done by:

A. IgG antibody against toxoplasma
B. **IgM antibodies to toxoplasma**
C. IgA antibodies to toxoplasma
D. IgE antibodies to toxoplasma

Ans B do not cross the placenta, so their presence in a newborn indicates true congenital infection, unlike IgG which is passively transferred from the mother.

220.Congenital toxoplasmosis–False is:

A. Diagnosed by detection of IgM in cord blood
B. IgA is more sensitive than IgM for detection
C. Dye test is gold standard for IgG
D. **Avidity testing must be done to differentiate IgA & IgM**

OTHER PARASITIC INFECTIONS

Ans D Avidity testing must be done to differentiate between IgA and IgM Serological tests in Congenital Toxoplasmosis: Assay Characteristics IgG Appears in 1–2 weeks, peaks at 1–2 months & persists throughout life; Transplacental IgG disappears by 6–12 m age; But presence of IgG in the serum does not confirm the diagnosis because IgG can easily cross the placenta IgA It rises rapidly & usually disappears by 7 months; IgA has greater sensitivity for neonates compared to IgM assays; So IgA ELISA is better than IgM

221.MOST Campylobacter isolates are susceptible to

A. **aminoglycosides**
B. cephalosporins
C. rifampin
D. penicillins
E. trimethoprim

Ans A Most Campylobacter isolates are susceptible to macrolides, fluoroquinolones, aminoglycosides, chloramphenicol, tetracyclines, and clindamycin, and are resistant to cephalosporins, rifampin, penicillins, trimethoprim, and vancomycin.

222.Hematogenous dissemination of HSV to the central nervous system occur in

A. **neonates**
B. individuals with eczema
C. severely malnourished children
D. infants on steroids
E. all the above

Ans A Hematogenous dissemination of virus to the central nervous system appears to only occur in neonates.

223.The MOST common long-term sequela associated with congenital CMV infection is

A. renal failure
B. **hearing loss**
C. heart failure
D. vision loss
E. hepatic failure

Ans B is the most common long-term sequela associated with congenital CMV infection, the failure of an infant to pass a newborn hearing screening exam should raise the possibility of congenital CMV infection. Hearing loss in the older infant and young child should also alert the clinician to the possibility of congenital CMV infection, as approximately 50% of infants with hearing loss associated with congenital CMV infection will pass an initial hearing screening exam but develop hearing loss in later infancy and early...

Fungal Infections

224.Prophylactic dose of Oseltamivir for infant is:

A. 1 mg/kg/day
B. **3 mg/kg/day**
C. 5 mg/kg/day
D. 7 mg/kg/day

Ans B If a child is younger than 3 months old, use of oseltamivir for chemoprophylaxis is not recommended unless situation is judged critical due to limited data in this age group. If child is 3 months or older and younder than 1 year old1 Chemoprophylaxis
• 3 mg/kg/dose once daily2 for H1N1 infection If 1 year or older, dose varies by child's weight:
• 15 kg or less, the dose is 30 mg once a day
• >15 to 23 kg, the dose is 45 mg once a day
• > 23 to 40 kg, the dose is 60 mg once a day

225.Kenny packs were used in the treatment of:

A. **Poliomyelitis**
B. Muscular dystrophy
C. Polyneuropathies
D. Nerve injury

Ans A There is no specific treatment for Poliomyelitis; Management is primary symptomatic and supportive & includes:
• Bed rest; as physical activity & trauma increase risk of paralysis & optimum position of limbs—to prevent deformity
• Relief of pain & muscle spasm: Hot moist packs applied to muscles (sister Kenny's treatment); Physiotherapy;
• Rehabilitation: Physical, emotional and psychological.

226.In Hutchnson triad, all of the following are seen except:

A. Interstitial keratitis
B. **Cataract**
C. Deafness
D. Notched incisors

Ans B

227.Most common cause of acute otitis media in children is:

A. **Strep pneumoniae**
B. Strep pyogenes
C. Staph aureus
D. Pseudomonas

Ans A • 3 pathogens predominate in Acute Otitis Media: Strepto pneumoniae, non-typeable H. influenzae, & Moraxella catarrhalis
• With the use of conjugate pneumococcal vaccine, nontypeable H. influenzae initially overtook S. pneumoniae as the most common pathogen, being found in 40-50% of cases;
• However, over time, S. pneumoniae serotypes not covered in the conjugate vaccine have emerged, with S. pneumoniae again overtaking non-typeable H. influenzae as the most common pathogen in many studies.

228.The most common fungal infection in the neonates transmitted by caregiver's hands is:

A. Candida albicans
B. Candida glabrata
C. Candida tropicalis
D. **Candida parapsilosis** Chapter 8: Infectious Diseases 177 Answers

Ans D Candida parapsilosis Candida parapsilosis
• It is ubiquitous in nature and is found as a commensal on the human skin
• It is most frequently isolated from hands (subungual space) and the gastrointestinal tract
• It is the most common fungal infection in the neonates transmitted by the hands of caregivers
• Neonatal risk factors for invasive C. parapsilosis infections are: Birth weight <1500 g, Prematurity, Prior colonization, Parenteral nutrition, Intravascular catheters Chapter TYPES OF VACCINES M

229.Primary immunodeficiencies are compromised states that result from genetic defects affecting 1 or more arms of the immune system while secondary immunodeficiencies result from infection, malignancy, or as an adverse effect of immunomodulating or immunosuppressing medications. Which of the following represent secondary immune deficiency?

A. Shwachman-Diamond syndrome
B. **cystic fibrosis**
C. Chédiak-Higashi syndrome
D. Omenn syndrome
E. ataxia-telangiectasia

Ans B All other distractors represent primary immune deficiency.

230.Patients with acute rheumatic carditis and more than minimal cardiomegaly should receive prednisone 2 mg/kg/day in 4 divided doses for

A. 7-10 days
B. **2-3 weeks**
C. 6-8 weeks
D. 2-3 months
E. 4-6 months

Ans B Patients with carditis and more than minimal cardiomegaly and/or congestive heart failure should receive corticosteroids. The usual dose of prednisone is 2 mg/kg/day in 4 divided doses for 2-3 wk followed by half the dose for 2-3 wk and then tapering of the dose by 5 mg/24 hr every 2-3 days. When prednisone is being tapered, aspirin should be started at 50 mg/kg/day in 4 divided doses for 6 wk to prevent rebound of inflammation.

231.The genus Yersinia is a member of the family Enterobacteriaceae and comprises more than 14 named species, 3 of which are established as human pathogens. Which of the following is MOST often associated with mesenteric lymphadenitis?

A. Yersinia enterocolitica
B. **Yersinia pseudotuberculosis**
C. Yersinia pestis
D. Yersinia mollaretii
E. Yersinia rohdei

Ans B Yersinia enterocolitica is by far the most common Yersinia species causing human disease and it produces fever, abdominal pain that can mimic appendicitis, and diarrhea. Yersinia pseudotuberculosis is most often associated with mesenteric lymphadenitis. Yersinia pestis is the agent of plague and most commonly causes an acute febrile lymphadenitis (bubonic plague) and less commonly occurs as septicemic, pneumonic, pharyngeal, or meningeal plague. Other Yersinia organisms are uncommon causes of infections of humans.

232.Candida is a common cause of oral mucous membrane infections (thrush) and perineal skin infections (Candida diaper dermatitis) in young infants. All the following are true EXCEPT

A. candida species are the third most common cause of bloodstream infection in premature infants
B. up to 10% of full-term infants are colonized as the result of vertical transmission from the mother at birth
C. histamine-2 blockers facilitate Candida colonization and overgrowth
D. significant risk factors for neonatal invasive candidiasis include the presence of a central venous catheter
E. **the cumulative incidence is <0.3% among infants <750 g birthweight admitted to the NICU**

Ans E The cumulative incidence is <0.3% among infants >2,500 g birthweight admitted to the NICU. The cumulative incidence increases to 8% for infants <750 g birthweight.

233.Common adverse effects of nitazoxinide include

A. **diarrhea**
B. flatulence
C. increased appetite
D. fever
E. pruritus

Ans A Common adverse effects include abdominal pain, diarrhea, and nausea. Rare side effects include anorexia, flatulence, increased appetite, fever, pruritus, and dizziness.

Parasitic & Helminthic Infections

234.Nagayama spot are seen in:

A. Measles
B. **Roseola infantum**
C. Mumps
D. Rubella

Ans B Ulcers at uvulopalatoglossal junction (Nagayama spots) are commonly reported in infants with roseola infantum. High fever is the most consistent finding associated with primary HHV-6B infection.

235.Acute flaccid paralysis is reported in a child aged:

A. 0–3 years
B. 0-5 years
C. **0–15 years**
D. 0–5 years

Ans C Acute flaccid paralysis surveillance under polio eradication programs covers all children under 15 years of age.

236.Thrombocytopenia, macerated skin lesions, rash and periostitis in a newborn are seen in:

A. Erythroblastosis fetalis
B. Cytomegalovirus infection
C. **Syphilis**
D. HIV infection

Ans C All the given features are early manifestations of Congenital Syphilis;

237.A 3monthsold male infant developed otitis media for which he was given a course of Cotrimoxazole. A few days later, he developed extensive peeling of the skin; there were no mucosal lesions and the baby was not toxic. The most likely diagnosis is:

A. Toxic epidermal necrolysis
B. **Staphylococcal scalded skin syndrome**
C. Steven Johnson syndrome
D. Infantile pemphigus

Ans B Rashes are seen in all these diseases; However, in Toxic epidermal necrolysis, Steven Johnson syndrome & Infantile pemphigus, intraoral mucosal involvement is usually seen, but in Staphylococcal scalded skin syndrome, intraoral mucosal surfaces are spared (as in this case).

238.In a rural clinic, a 3yearold girl child is brought by her mother and is emaciated. Her hemoglobin was 5 g/dL. The girl also has edema over her knees and ankles with discrete rash on her knees, ankles and elbows. The most likely worm infestation causing these manifestations is:

A. **Hookworm**
B. Roundworm
C. Whipworm
D. Pinworm

Ans A Hookworm infestation:
• Leads to intestinal blood loss → iron deficiency anemia & malnutrition → growth retardation
• Dermatitis referred to as ground itch when they penetrate human skin, manifests as vesiculation & edema
• Infection with a zoonotic hookworm, especially A. braziliense → cutaneous tracts of cutaneous larva migrans
• Cough occurs when larvae migrate through lungs ~ 1 week after exposure; Eosinophilia is often seen

239.Treatment for the child with frequent tapeworm infestation:

A. Albendazole
B. **Niclosamide**
C. Praziquantel
D. Mebendazole
E. Ivermectin

Ans B c. Praziquantel and d. Mebendazole with Infecting organism Drug of choice Alternative Taenia saginata (Beef tapeworm) Praziquantel or Niclosamide Mebendazole Taenia solium (Pork tapeworm) Praziquantel or Niclosamide Cysticercosis (Pork tapeworm larval stage) Albendazole Praziquantel

240.The drug of choice for Sydenham chorea is

A. aspirin
B. prednisone
C. **phenobarbital**
D. haloperidol
E. chlorpromazine

Ans C (16-32 mg every 6-8 hr PO) is the drug of choice. If phenobarbital is ineffective, then haloperidol (0.01-0.03 mg/kg/24 hr divided bid PO) or chlorpromazine (0.5 mg/kg every 4-6 hr PO) should be initiated. Some patients may benefit from a few week courses of corticosteroids.

241.Patients with conditions leading to iron overload are at higher risk of developing infections with

A. Aeromonas
B. Pseudomonas aeruginosa
C. **Yersinia**
D. Francisella tularensis
E. Campylobacter

Ans C Patients with conditions leading to iron overload are at higher risk of developing Yersinia infections.

242.Herpes whitlow is HSV infection of the

A. eye
B. mouth
C. **fingers**
D. ear
E. lip

Ans C Herpes whitlow is a term generally applied to HSV infection of fingers or toes, although strictly speaking it refers to HSV infection of the paronychia.

243.One of the following statements regarding tinidazole is TRUE

A. **it is FDA approved for treatment of trichomoniasis**
B. it can be in infancy
C. it should be given in 2 divided doses
D. it is excreted via urine only
E. it carries a pregnancy category A

Ans A and for giardiasis and amebiasis in children 3 yr of age and older. In the treatment of giardiasis, it has the advantages of very few side effects and only requiring a single dose. It is excreted via urine and feces. Tinidazole carries a pregnancy category C classification and can be detected in breast milk. Breastfeeding should be interrupted during treatment and for 3 days after treatment.

244.One of the following statements regarding Giardia lamblia is TRUE

A. life cycle is composed of 3 stages
B. each ingested cyst produces 2 trophozoites in the caecum
C. trophozoites contain 4 oval nuclei anteriorly
D. **cyst viability is not affected by the usual concentrations of chlorine used to purify water for drinking**
E. Giardia genotypes difference lead to wide spectrum of clinical manifestations

Ans D The life cycle of G. lamblia (also known as Giardia intestinalis or Giardia duodenalis) is composed of 2 stages: trophozoites and cysts. Each ingested cyst produces 2 trophozoites in the duodenum. Giardia trophozoites contain 2 oval nuclei anteriorly, a large ventral disk, a curved median body posteriorly, and 4 pairs of flagella. Studies suggest that different Giardia genotypes may cause unique clinical manifestations, but these findings appear to vary according to the geographic region tested.

245.All the following statements regarding giardiasis are true EXCEPT

A. incubation period of Giardia infection usually is 1-2 wk
B. most infections are asymptomatic
C. **symptomatic infections occur more frequently in adults than in children**
D. stools do not contain blood, mucus, or fecal leukocytes
E. malabsorption of sugars, fats, and fat-soluble vitamins is well documented

Ans C

246.Giardiasis should be considered in children who have the following presentations

A. **acute dysenteric diarrhea**
B. persistent diarrhea
C. failure to thrive
D. malabsorption
E. chronic crampy abdominal pain

Ans A Giardiasis should be considered in children who have acute nondysenteric diarrhea, persistent diarrhea, intermittent diarrhea and constipation, malabsorption, chronic crampy abdominal pain and bloating, failure to thrive, or weight loss.

247.Pregnant mother seek your advice about the stool examination of her asymptomatic 15 -month-old child which contain giardia cyst, your advice should be

A. reassurance, no treatment
B. inidazole 50 mg\kg once
C. nitazoxinide 200 mg bid for 3 days
D. **metronidazole15 mg/kg/day in 3 divided doses for 5-7 days**
E. albendazole 400 mg once a day for 5 days

Ans D Asymptomatic excreters generally are not treated except in specific instances such as outbreak control, prevention of household transmission by toddlers to pregnant women and patients with hypogammaglobulinemia or cystic fibrosis, and situations requiring oral antibiotic treatment where Giardia may produce malabsorption of the antibiotic.Tinidazole not recommended below 3 years old as albendazole not recommended below age of 6 years. Nitazoxanide1-3 yr: 100 mg bid for 3 days

Antibiotics & Antimicrobial Therapy

248.All are caused by Parvo virus infection except:

A. Erythema infectiosum
B. **Gianotti crosti syndrome**
C. Aplastic crisis
D. Papular- purpuric gloves and socks syndrome (PPGSS)

Ans B Diseases caused by Parvo virus B19 infection include: Erythema infectiosum (most common), Transient arrest of erythropoiesis & papular purpuric "gloves-and-socks" syndrome (PPGSS) GianottiCrosti syndrome, a symmetric, atopic dermatitis like rash on cheeks with multiple erythematous papules is associated with EBV infection.

249.An 8 years old female child following URTI developed maculopapular rash on the jaw spread onto the trunk which cleared on the 3rd day without desquamation and tender post auricular and suboccipital lymphadenopathy. The diagnosis is:

A. Kawasaki disease
B. Erythema infectiosum
C. **Rubella**
D. Measles

Ans C

250.Premature baby of 34 weeks was delivered. Baby developed bullous lesion on the skin and Xray shows periostitis. What should be the next investigation?

A. **VDRL for mother & baby (AIIMS Nov 11, AI 07)**
B. ELISA for HIV
C. PCR for TB
D. Hepatitis surface antigen for mother

Ans A VDRL for mother and baby Bullous lesion and periostitis suggest the diagnosis of congenital syphilis. So, VDRL is done for mother & baby;

251.Risk of transplacental transmission of toxoplasma gondii is maximum when mother is infected:

A. Hydrocephalus
B. Chorioretinitis
C. Hepatosplenomegaly
D. **Thrombocytopenia**

Ans D During third trimester Risk of fetal infection in acute maternal toxoplasmosis increases with each trimester of pregnancy being 15%, 25% and 60% in 1st, 2nd and 3rd trimesters respectively; But severity of fetal infection is much greater in early pregnancy.

252.Serious reactions to IVIG include all the following EXCEPT

A. anaphylactoid events
B. thromboembolic disorders
C. aseptic meningitis
D. **carditis**
E. renal insufficiency

Ans D

253.All the following are causes of very high temperatures (>41°C) EXCEPT

A. malignant hyperthermia
B. malignant neuroleptic syndrome
C. **infection**
D. drug fever
E. heat stroke

Ans C Temperatures in excess of 41°C (105.8°F) are most often associated with a noninfectious cause.

254.Meropenem is a carbapenem antibiotic with broad-spectrum activity but has no activity against

A. P. aeruginosa
B. **S. maltophilia**
C. L. monocytogenes
D. S. aureus
E. N. meningitides

Ans B Carbapenem antibiotic with broad-spectrum activity against Gram-positive cocci and Gram-negative bacilli, including P. aeruginosa and anaerobes. No activity against S. maltophilia.

255.The MOST common reason for which children receive antibiotics is

A. pneumonia
B. bronchitis
C. **otitis media**
D. pharyngitis
E. cystitis

Ans C is the most common reason for which children receive antibiotics. On the basis of culture of middle ear fluid obtained by tympanocentesis, the predominant causes of acute otitis media are Streptococcus pneumoniae, H. influenzae, and M. catarrhalis.

256.Which antibiotics should be avoided in patients with botulism?

A. penicillin
B. cephalosporin
C. macrolide
D. **aminoglycoside**
E. trimethoprim-sulfamethoxazole

Ans D antibiotics should be avoided because they may potentiate the blocking action of botulinum toxin at the neuromuscular junction.

257.All the following can cause human amoebic meningoencephalitis EXCEPT

A. Naegleria
B. Acanthamoeba
C. Balamuthia
D. **Entamoeba histolytica**
E. Sappinia

Ans D Naegleria, Acanthamoeba, Balamuthia, and Sappiniaare small, free living amebas that cause human amebic meningoencephalitis.

Fever & Fever Management

258.A 5yearold child develops fever with rash on the first day of fever. Rash disappears after few days & the child develops ataxia. Most probable diagnosis is:

A. Measles
B. Fifth disease
C. **Chicken pox**
D. Rocky mountain spotted fever

Ans C Chickenpox Rash appearing on 1st day of fever suggests Chickenpox; Ataxia can occur as a complication of varicella infection.

259.A child presents with history of high fever for 4 days along with seizures. A rash develops on trunk on the day after the fever subsides. What is the most probable diagnosis?

A. **Roseola infantum**
B. Measles
C. Rubella
D. Varicella

Ans A History of high fever & rash appearing as the fever subsides is suggestive of Roseola infantum;

260.An 8yearsold boy presented with fever and bilateral Questions cervical lymphadenopathy with prior history of sore throat. There was no hepatomegaly. The peripheral blood smear shows > 20% lymphoplasmacytoid cells. The most likely diagnosis is:

A. Influenza
B. Tuberculosis
C. **Infectious mononucleosis**
D. Acute lymphoblastic leukemia

Ans C c. 0–15 years For AFP Surveillance, every case of acute flaccid paralysis in any child under 15 years is to be reported promptly.

261.A patient who had fever and coryza for last 3 days, developed maculopapular erythematous rash, which lasted for 48 hours and disappeared without leaving behind pigmentation. This is most commonly due to:

A. Measles
B. Typhoid
C. **Roseola infantum**
D. Fifth disease

Ans C

262.A child with fever and sore throat developed acute cervical lymphadenopathy. Most likely investigation to be done is:

A. Open biopsy of node
B. Radical neck dissection
C. Neck X-ray
D. **Complete hemogram**

Ans D • Cervical lymphadenopathy with signs of infection such as fever and sore throat points towards any infectious etiology
• In this case the most likely investigation to be done should be complete hemogram. dna 99. b. Multiple papillomatosis Juvenile laryngeal papillomatosis (also referred to as respiratory papillomatosis):
• Due to HPV infection acquired by infants during passage through an infected birth canal
• Median age at diagnosis of recurrent laryngeal papillomatosis is 3 yr;

263.A 6yearold girl presents with fever for the past 5 days, Questions generalized erythematous rash, strawberry tongue and cervical lymphadenopathy. The most likely diagnosis is:

A. Kimura disease
B. Kawasaki disease
C. **Scarlet fever**
D. Rosie-Dorfman syndrome

Ans C Fever for at least 5 days, generalized erythematous rash, strawberry tongue and cervical lymphadenopathy are compatible with a diagnosis of Scarlet fever; Kawasaki disease is a close differential diagnosis (Fever + at least 4 out of 5 criteria required for its diagnosis, here aonly 3 criteria present apart from fever)

264.The diagnosis of a child with small abrasion over face developing fever with chills, induration and fiery red swelling over the face and extending to the ear:

A. Erysipelas
B. Carbuncle
C. **Cellulitis**
D. Anthrax

Ans C Discussing about the options one by one, Answers a. Erysipelas: It involves deeper layers of skin & connective tissue with swelling, redness & tenderness, sharply defined border; abrupt onset with high fever seen b. Carbuncles refer to multiple coalesced boils, usually caused by Staphylococcal infection c. Cellulitis: Refers to a non-necrotizing inflammation of skin & subcutaneous tissues, that usually follows a breach in the skin;

265.A child from West Bengal presents with fever & unconsciousness for 1 day and pallor with no focal neurodeficit. What is the most probable diagnosis?

A. **Cerebral malaria**
B. Viral encephalitis
C. Pyomeningitis
D. Tubercular meningitis

Ans A Malaria due to Plasmodium falciparum:
• Acute onset of fever, anemia with altered sensorium in an endemic area suggests cerebral malaria
• It is the most severe form of malaria and is associated with higher density parasitemia and a number of complications
• Serious complications that appear unique to P. falciparum include cerebral malaria, acute renal failure, respiratory distress from metabolic acidosis, algid malaria and bleeding diatheses. Cerebral malaria:
• Defined as presence of coma in a child with P.

266.Fever is defined as a rectal temperature of

A. ≥37.5°C
B. ≥37.6°C
C. **≥38°C**
D. ≥38.1°C
E. ≥38.5°C

Ans C Fever is defined as a rectal temperature ≥38 ° C (100.4°F) and a value >40°C (104°F) is called hyperpyrexia.

267.Three mechanisms can produce fever: pyrogens, heat production exceeding loss, and defective heat loss. Endogenous pyrogens include

A. antigen–antibody complexes
B. complement components
C. lymphocyte products
D. **interferons β and γ**
E. androgenic steroid metabolites

Ans D Endogenous pyrogens include the cytokines interleukins 1 and 6, tumor necrosis factor α, and interferons β and γ. Some substances produced within the body are not pyrogens but are capable of stimulating endogenous pyrogens. Such substances include antigen–antibody complexes in the presence of complement, complement components, lymphocyte products, bile acids, and androgenic steroid metabolites.

268.Drugs that are known to cause fever include

A. chlorpheniramine
B. **allopurinol**
C. diphenhydramine
D. acetazolamide
E. adenosine

Ans B Drugs that are known to cause fever include vancomycin, amphotericin B, and allopurinol.

269.Which type of fever is persistent and varies by more than 0.5°c (0.9°f)/day?

A. intermittent
B. hectic
C. sustained
D. **remittent**
E. relapsing

Ans D Intermittent fever is an exaggerated circadian rhythm that includes a period of normal temperatures on most days; extremely wide fluctuations may be termed septic or hectic fever. Sustained fever is persistent and does not vary by more than 0.5°C (0.9°F)/day. Remittent fever is persistent and varies by more than 0.5°C (0.9°F)/day. Relapsing fever is characterized by febrile periods that are separated by intervals of normal temperature.

270.Relative bradycardia (when the pulse rate remains low in the presence of fever) can accompany all the following conditions EXCEPT

A. typhoid fever
B. brucellosis
C. leptospirosis
D. **visceral leishmaniasis**
E. drug fever

Ans D

271.In which of the following circumstances the diagnosis of acute rheumatic fever can be made without strict adherence to Jones criteria?

A. when chorea occurs as the only major manifestation of acute rheumatic fever
B. when indolent carditis is the only manifestation months after the apparent onset of acute rheumatic fever
C. in a limited number of patients with recurrences of acute rheumatic fever in particularly high-risk populations
D. **all of the above**
E. none of the above

Ans D

272.Prophylaxis for people who have had acute rheumatic fever with carditis but without residual heart disease persist for

A. 5 yr or until 21 yr of age, whichever is longer
B. **10 yr or until 21 yr of age, whichever is longer**
C. 5 yr or until 40 yr of age, whichever is longer
D. 10 yr or until 40 yr of age, whichever is longer
E. Lifelong

Ans B o Rheumatic fever without carditis : 5 yr or until 21 yr of age, whichever is longer o Rheumatic fever with carditis but without residual heart disease : 10 yr or until 21 yr of age,whichever is longer o Rheumatic fever with carditis and residual heart disease: 10 yr or until 40 yr of age, whichever is longer; sometimes lifelong prophylaxis

Rickettsial & Leptospirosis

273.Most common vector of Japanese encephalitis is:

A. Anopheles
B. **Culex**
C. Ixodes
D. Aedes

Ans B Culex tritaeniorhynchus summarosus, a nightbiting mosquito that feeds preferentially on large domestic animals & birds but only infrequently on humans, is the principal vector of JE in northern Asia.

274.Most common manifestation of HPV infection in children:

A. Single papilloma
B. **Multiple papillomatosis**
C. Osteoma
D. Sarcoma
E. Genital wart

Ans B In children, HPV most commonly manifests as recurrent respiratory (laryngeal) papillomatosis, which is typically multiple and recurrent rather than a single lesion.

275.Hutchnson's triad is seen in:

A. Primary syphilis
B. **Congenital syphilis**
C. Secondary syphilis
D. Tertiary syphilis

Ans B Hutchinson's triad seen in Congenital syphilis consists of: Interstitial keratitis, Hutchinson teeth & Eight nerve deafness.

276.A pregnant lady had no complaints but mild cervical lymphadenopathy in first trimester. She was prescribed Spiramycin but she was noncompliant. Baby was born with hydrocephalus and intracerebral calcification. Which of these is likely cause?

A. Rubella
B. **Toxoplasmosis**
C. CM
D. Herpes

Ans B Presence of cervical Lymphadenopathy, treatment (which was not done properly) with Spiramycin and neonatal manifestation in the form of hydrocephalus and intracerebral calcification, all point to diagnosis of congenital toxoplasmosis.

277.A toxoid is a modified bacterial toxin that is made non-toxic but still able to induce an active immune response against the toxin. Which of the following vaccine is a toxoid?

A. hepatitis A
B. hepatitis B
C. pneumococcal
D. varicella
E. **diphtheria**

Ans E Toxoids are (tetanus and diphtheria).

278.The MOST common serious bacterial infection in infant aged 1-3 mo is

A. **pyelonephritis**
B. meningitis
C. pneumonia
D. septic arthritis
E. osteomylitis

Ans A is the most common serious bacterial infection in this age group and is also more common in uncircumcised infant boys and infants with urinary tract anomalies. E. coliis the most common pathogen identified in bacteremic infants, the majority having pyelonephritis.

279.A painless, slow-growing, hard mass producing cutaneous fistulas, a condition commonly known as lumpy jaw is usually caused by

A. Staphylococcus
B. **Actinomyces**
C. Nocardia
D. Yersinia
E. Leptospira

Ans B Three major forms of actinomycosis—cervicofacial, abdominal and pelvic, and pulmonary. In the patient with cervicofacial actinomycosis, there is often a history of oral trauma, oral surgery, dental procedures, or caries, facilitating entry of organisms into cervicofacial tissues. Cervicofacial actinomycosis usually manifests as a painless, slow-growing, hard mass and can produce cutaneous fistulas, a condition commonly known as lumpy jaw.

280.The MOST common complication of Y. enterocolitica infection in younger children is

A. reactive arthritis
B. erythema multiforme
C. hemolytic anemia
D. thrombocytopenia
E. **septicemia**

Ans E is more common in younger children, and reactive arthritis is more common in older patients.

281.In neonatal HSV encephalitis, skin vesicles occur in approximately

A. 20%
B. 40%
C. **60%**
D. 80%
E. 100%

Ans C Infants with encephalitis typically present at 8-17 days of life with clinical findings suggestive of bacterial meningitis, including irritability, lethargy, poor feeding, poor tone, and seizures. Fever is relatively uncommon, and skin vesicles occur in only approximately 60% of cases.

282.The largest protozoan that parasitizes human is

A. Entamoeba histolytica
B. Giardia lamblia
C. **Balantidium coli**
D. Isospora belli
E. Cryptosporidium

Ans C

COVID-19 & Emerging Infections

283.The common name for roseola infantum caused by human herpes virus 6A and 6B is:

A. **Exanthema subitum**
B. Fifth disease
C. Herpes fever
D. None of the above

Ans A

284.Which of the following statement is true for Ebola virus?

A. **It is member of filoviridae**
B. Incubation period is 2 –12 weeks
C. Transmitted by direct contact
D. Human to human transmission is by air and food
E. In severe cases lead to renal and liver impairment

Ans A c. Transmitted by direct contact & e. In severe cases lead to renal and liver impairment Harrison's 19/e p 1323 a. True Family Filoviridae includes 3 genera: Cuevavirus, Ebolavirus & Marburg virus b. False Incubation period of Ebola virus disease is 2-21 days c. True Natural human-to-human transmission takes place through direct person-to-person (usually skin-to-skin) contact or exposure to infected bodily fluids & tissues; there is no evidence of such transmission by aerosol or respiratory droplets. d. False e.

285.Congenital syphilis can be best diagnosed by:

A. **IgM FTA–ABS**
B. IgG FTA–ABS
C. VDRL
D. TPI ENTERIC FEVER

Ans A As IgM does not cross placenta, its presence in neonatal serum confirms congenital syphilis & helps differentiating it from seropositivity due to passively transferred maternal antibodies; Tests to detect IgM are FTA-ABS & Syphilis Capita M test.

286.Which of the following causes mesenteric lymphadenitis?

A. Adenovirus
B. Campylobacter
C. Yersinia pseudotuberculosis
D. **All of the above**

Ans D • Acute appendicitis, mesenteric lymphadenitis & ileocolitis have been reported in patients who have had appendectomies during C. jejuni infection.
• Yersinia pseudotuberculosis is most often associated with mesenteric lymphadenitis.
• Human Adenovirus may also cause mesenteric adenitis.

287.Preterm infant, weight 1600 gm, should not receive the following vaccine at birth

B. **hepatitis B, if born to a HBs Ag negative mother**
C. polio

Ans B Preterm infants generally can be vaccinated at the same chronologic age as full- term infants according to the recommended childhood immunization schedule. An exception is the birth dose of hepatitis B vaccine. Infants weighing ≥2 kg and who are stable may receive a birth dose. However, hepatitis B vaccination should be deferred in infants weighing <2 kg at birth until 30 days of age, if born to a HBs Ag negative mother.

288.Neutropenia is defined as an absolute neutrophil count of less than

A. **1,000 cells/mm3**
B. 1,500 cells/mm3
C. 2,000 cells/mm3
D. 2,500 cells/mm3
E. 3,000 cells/mm3

Ans A Neutropenia is defined as an absolute neutrophil count of <1,000 cells/mm3 and can be associated with significant risk for developing severe bacterial and fungal disease.

289.The MOST common secondary site involved in cases of pulmonary nocardiosis is

A. **brain**
B. skin
C. kidney
D. liver
E. bone

Ans A The brain is the most common secondary site and is involved in 15-40% of cases of pulmonary nocardiosis. Brain abscess is the most common presentation, and meningitis is the second most common presentation.

290. **Tularemia is a zoonotic infection caused by the gram-negative bacterium Francisella tularensis. Of the following, the MOST common forms of tularemia diagnosed in children is**

A. **ulceroglandular**
B. pneumonia
C. oropharyngeal
D. oculoglandular
E. typhoidal

Ans A and glandular disease are the 2 most common forms of tularemia diagnosed in children. The most common glands involved are the cervical or posterior auricular nodes owing to a tick bite on the head or neck. If an ulcer is present, it is erythematous and painful and may last from 1-3 wk. The ulcer is located at the portal of entry. After the ulcer develops, regional lymphadenopathy ensues.

291. **In patients with neonatal HSV infection who receive IV treatment for 2-3 weeks then suppressive treatment for 6 months, they should be monitored by**

A. **absolute neutrophil count ANC**
B. liver function tests
C. renal function tests
D. platelet count
E. Hb%

Ans A The absolute neutrophil count should be measured at weeks 2 and 4 after initiation treatment and then monthly.

292. **Hand-foot-mouth disease is one of the more distinctive rash syndromes; it is MOST frequently caused by**

A. coxsackievirus A6
B. **coxsackievirus A16**
C. enterovirus
D. coxsackievirus B2
E. coxsackievirus B12

Ans B Hand-foot-and-mouth disease, one of the more distinctive rash syndromes, is most frequently caused by coxsackievirus A16, sometimes in large outbreaks, and can also be caused by enterovirus 71; coxsackie A viruses 5, 6, 7, 9, and 10; coxsackie B viruses 2 and 5; and some echoviruses.

09

CHAPTER 09

Immunization

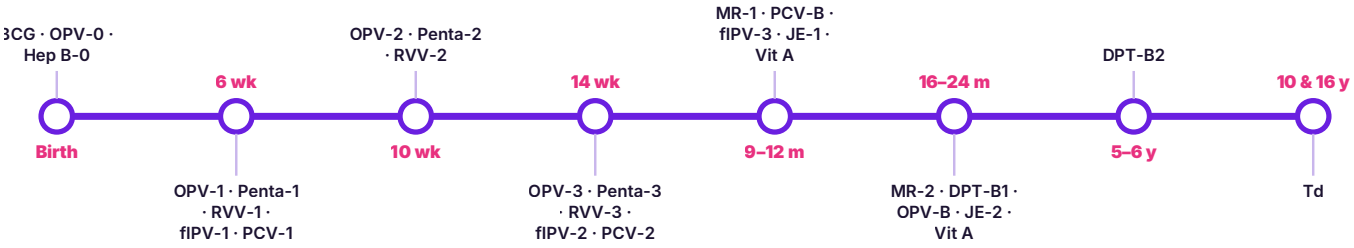


WHAT THIS CHAPTER COVERS

- National Immunization Schedule
- BCG & Tuberculosis Vaccines
- Polio Vaccines (OPV/IPV)
- DTP & Combination Vaccines
- Measles & MMR Vaccines
- Hepatitis B Vaccine
- Hib & Pneumococcal Vaccines
- Rotavirus Vaccine
- HPV Vaccine
- Typhoid & Cholera Vaccines
- Rabies & JE Vaccines
- Influenza Vaccine
- COVID-19 Vaccines
- Vaccine Contraindications & Adverse Events
- Passive Immunization
- Vaccine Storage & Cold Chain

77 MCQs **16** subtopics

9.1 National Immunization Schedule (UIP, India)



Universal Immunization Programme – infant & child schedule

Age	Vaccines (UIP)
Birth	**BCG**, **OPV-0**, **Hepatitis B birth dose (within 24 h)**
6 weeks	**OPV-1, Pentavalent-1 (DPT + Hep B + Hib), Rotavirus-1, fIPV-1, PCV-1**
10 weeks	OPV-2, Pentavalent-2, Rotavirus-2
14 weeks	OPV-3, Pentavalent-3, Rotavirus-3, **fIPV-2, PCV-2**
9–12 months	**MR-1**, **PCV booster**, **fIPV-3** (added 2023), JE-1, **Vitamin A (1st dose, 1 lakh IU)***
16–24 months	**MR-2, DPT booster-1, OPV booster**, JE-2*, Vitamin A (2nd dose, 2 lakh IU; then every 6 months to 5 y)
5–6 years	**DPT booster-2**
10 years & 16 years	**Td**
Pregnant women	**Td-1 (early), Td-2 (4 weeks later)**; **Td booster** if vaccinated within last 3 years

* JE in endemic districts. BCG can be given up to **1 year**; Hep B birth dose up to **24 h** (not after); OPV-0 within **15 days**; pentavalent up to 1 year; MR up to 5 years; DPT booster-1 up to 7 years.

UIP NUMBERS WORTH REMEMBERING

UIP launched **1985** (EPI 1978); **Mission Indradhanush 2014** (full immunisation coverage > 90%); **12 vaccine-preventable diseases** nationally (diphtheria, pertussis, tetanus, polio, measles, rubella, childhood TB, hepatitis B, Hib meningitis/pneumonia, **rotavirus diarrhoea, pneumococcal pneumonia**, JE in endemic areas). **India polio-free since 13 January 2011** (last case, Howrah); WHO certified SEAR polio-free **27 March 2014**. **tOPV → bOPV switch April 2016.** Maternal & neonatal tetanus eliminated **2015**.

9.2 IAP (ACVIP) 2023 Schedule – Key Differences from UIP

Age	IAP recommendation
Birth	BCG, OPV-0, Hep B-1
6 weeks	DTwP/DTaP-1, **IPV-1 (full dose IM)**, Hib-1, Hep B-2, Rota-1, PCV-1
10 weeks	DTP-2, IPV-2, Hib-2, Hep B-3, Rota-2, PCV-2
14 weeks	DTP-3, IPV-3, Hib-3, Hep B-4, Rota-3*, PCV-3

Age	IAP recommendation
6 months	**Influenza (IIV) – 2 doses 4 weeks apart**, then annually till 5 y; **Typhoid conjugate vaccine (TCV) from 6–9 months**
9 months	**MMR-1**
12 months	**Hepatitis A-1**, JE-1 (endemic)
15 months	**MMR-2, Varicella-1, PCV booster**
16–18 months	**DTP booster-1, IPV booster, Hib booster**
18–19 months	Hep A-2 (inactivated), **Varicella-2** (≥ 3 months after dose 1)
4–6 years	**DTP booster-2, OPV, MMR-3**
9–14 years	**HPV – 2 doses (0, 6–12 months)**; ≥ 15 y – 3 doses (0, 1–2, 6)
10–12 years	**Tdap**, then Td every 10 years

* 3rd dose for RV5 (RotaTeq), Rotavac and Rotasiil; **2 doses for RV1 (Rotarix)**.

9.3 General Principles

Principle	Rule
Live vaccines	**BCG, OPV, measles/MR/MMR, varicella, rotavirus, LAIV, yellow fever, live Hep A (H2), JE SA-14-14-2, Ty21a, cholera (oral live)**
Two **live parenteral** vaccines	**Same day or ≥ 4 weeks apart**
Minimum interval between doses of same vaccine	**4 weeks**
Interrupted schedule	**Never restart** – continue from where stopped
Preterm infants	Vaccinate as per **chronological age**, full dose; **Hep B: if < 2 kg, birth dose doesn't count – give 3 more doses** (start at 1 month)
Route & site	**Infants: anterolateral thigh (vastus lateralis)** for IM; deltoid after 1–2 y; **never gluteal** (sciatic nerve, poor immunogenicity – Hep B, rabies)
After immunoglobulin/blood	Defer measles/varicella 3–11 months
Steroids (≥ 2 mg/kg/d or ≥ 20 mg/d ≥ 14 days)	Defer live vaccines **1 month** after stopping

True contraindications vs false contraindications

True contraindications / precautions	NOT contraindications (vaccinate)
Anaphylaxis to a previous dose or component (all vaccines)	Mild illness ± low-grade fever, cold, diarrhoea
Live vaccines: severe immunodeficiency, pregnancy	Prematurity, low birth weight
Pertussis vaccine: encephalopathy within 7 days of previous dose (give DT)	**Family history of convulsions/SIDS/adverse events**
Progressive/unstable neurological disorder – defer pertussis until stable	Malnutrition, breastfeeding, antibiotics (except oral typhoid/live cholera)
Rotavirus: history of intussusception, SCID, uncorrected GI malformation	Stable neurological conditions (CP, controlled epilepsy)
Moderate–severe acute illness – defer	Allergy to penicillin, non-specific allergies, asthma

True contraindications / precautions	NOT contraindications (vaccinate)
Previous GBS within 6 wk of influenza/tetanus vaccine (precaution)	Recent exposure to infection, convalescence

9.4 BCG

- **Live attenuated M. bovis – Danish 1331 strain** used in India (Copenhagen); freeze-dried, reconstituted with **normal saline**.
- Dose **0.05 mL in < 1 month (neonates)**, 0.1 mL after; **intradermal**, **left upper arm at the insertion of deltoid**; Tuberculin syringe, 26G needle.
- Course: papule in 2–3 weeks → pustule → ulcer → **scar by 6–12 weeks**. **No dressing/antiseptic.**
- Protects mainly against **severe childhood forms (TB meningitis, miliary TB) – 70–80%**; variable protection against adult pulmonary TB (0–80%).
- **No booster**; tuberculin test not needed before BCG in infants.
- Complications: **suppurative/non-suppurative lymphadenitis (MC – axillary)**, keloid, **BCG-osis** (disseminated in SCID/CGD), lupus vulgaris. **Reconstituted vaccine used within 4 hours**; protect from light.
- **Contraindicated in symptomatic HIV**, SCID, CGD, pregnancy.

9.5 Polio Vaccines

	OPV (Sabin) – bivalent (types 1 & 3)	IPV (Salk)
Type	Live attenuated	**Inactivated (formalin)**, trivalent
Route / dose	**2 drops oral**	Full dose **0.5 mL IM**; **fractional IPV (fIPV) 0.1 mL intradermal** (UIP – right upper arm)
Immunity	**Intestinal (mucosal IgA) + humoral → interrupts transmission**, herd effect; cheap, easy	Humoral only (poor gut immunity)
Problems	**VAPP** (1 per 2.4 million doses – most with **type 3**/first dose), **cVDPV**, **heat-labile (most heat-sensitive vaccine – stored at −20 °C)**, interference by enteroviruses	Costly, injection
Contraindication	**Immunodeficient child or household contact** → use IPV	—

- **Type 2 wild poliovirus eradicated (1999); type 3 (2019)** – only **type 1 WPV** circulates (Pakistan, Afghanistan).
- **AFP surveillance**: any child **< 15 years** with acute flaccid paralysis (or any age with suspected polio) → **2 stool samples 24–48 h apart within 14 days of onset**; 60-day follow-up for residual paralysis. Non-polio AFP rate target **≥ 2/100,000 < 15 y** (India); **MCC of AFP now – GBS**.
- **Pulse polio**: all children 0–5 years regardless of previous doses.

9.6 DPT & Combination Vaccines

- **Pentavalent** (UIP) = **DTwP + Hep B + Hib**, 0.5 mL IM anterolateral thigh; freeze-sensitive.
- **Whole-cell (DTwP)** more reactogenic but **more effective/longer-lasting** than acellular (DTaP); IAP accepts both.
- Pertussis component: **most reactogenic**; diphtheria & tetanus are **toxoids** with **aluminium phosphate/hydroxide adjuvant** – **shake test** for freezing damage; **thiomersal** preservative.
- **Adverse events of DTwP**: fever, local reaction (MC), **persistent inconsolable crying ≥ 3 h**, **hypotonic-hyporesponsive episode (HHE)**, febrile seizures, encephalopathy (rare).
- **Tdap/Td**: reduced diphtheria (d) & pertussis (ap) for **≥ 7 years** (adult-type).

- Tetanus-prone wound: < 5 y since last dose – nothing; 5–10 y – Td booster; > 10 y or unknown – **Td + TIG (250–500 IU)**.

9.7 Measles, MR & MMR

- Live attenuated (**Edmonston–Zagreb strain** in India); **0.5 mL SC right upper arm**; reconstituted with **distilled water/diluent**, keep cold, **use within 4 hours**, protect from light.
- **Seroconversion ~85% at 9 months, ~95% at 12 months** (maternal antibodies interfere earlier) → second dose essential.
- **Outbreak/high-risk**: extra dose from **6 months** (doesn't count; continue schedule).
- AEFI: **fever & rash 7–12 days post-vaccine**, **ITP (1 in 30,000)**, febrile seizures; **toxic shock syndrome** (programmatic error – contaminated reconstituted vial used beyond 4–6 h, deaths within hours).
- **Rubella (RA 27/3 strain)**; mumps (Jeryl Lynn / L-Zagreb) – aseptic meningitis with Urabe strain.
- **MR campaign 2017** (9 m–15 y); target – **measles & rubella elimination**.

9.8 Hepatitis B, Hib, Pneumococcal, Rotavirus

Vaccine	Key facts
Hepatitis B	Recombinant **HBsAg (yeast-derived)**; 0.5 mL (10 µg) IM in children < 10–19 y; adults 1 mL (20 µg); **birth dose within 24 h** prevents perinatal transmission; **HBIG 0.5 mL within 12 h** for babies of HBsAg+ mothers (different site); **most freeze-sensitive**; protective anti-HBs **≥ 10 mIU/mL**
Hib	**Conjugate** (PRP-T/PRP-OMP); polysaccharide alone is T-independent – poor < 2 y; protects against meningitis, epiglottitis, pneumonia
PCV	**Pneumosil (PCV10)** in UIP – **2 + 1 schedule (6 wk, 14 wk, 9 m)**; PCV13/PCV15/PCV20 private (IAP 3+1 or 2+1); **PPSV23 (polysaccharide) only > 2 y** in high-risk (asplenia, sickle cell, nephrotic, CSF leak, cochlear implant)
Rotavirus	**Rotavac (116E, monovalent, 5 drops – 0.5 mL)**, **Rotasiil (pentavalent bovine-human reassortant, 2.5 mL)** in UIP; Rotarix (RV1, 2 doses), RotaTeq (RV5); **first dose ≥ 6 weeks**, **max age for first dose 15 weeks (IAP)**, **complete by 8 months**; small **risk of intussusception** (~1–2/100,000) – contraindicated if past intussusception, SCID

9.9 Other Vaccines

Vaccine	Type & schedule
Typhoid conjugate (TCV – Vi-TT)	**Single 0.5 mL IM from 6 months** (IAP: 6–9 m); preferred over Vi-polysaccharide (only > 2 y, revaccinate 3-yearly); **Ty21a** – live oral capsules > 6 y (days 1, 3, 5) – avoid antibiotics
Hepatitis A	**Live attenuated (H2 strain) – single dose SC ≥ 12 m**; inactivated – 2 doses IM 6 months apart
Varicella	Live (Oka strain); 2 doses (15 m & 18–19 m / 4–6 y); post-exposure within 3–5 days
Influenza	Inactivated quadrivalent (IIV4); 6 m–8 y first time – **2 doses 4 weeks apart**, then yearly; **0.5 mL** at all ages; **annual pre-monsoon (southern hemisphere strain for South India)**
HPV	**Bivalent (16, 18), quadrivalent (6, 11, 16, 18 – Gardasil, Indian Cervavac)**, nonavalent (Gardasil 9); 9–14 y – 2 doses (0, 6–12 m); ≥ 15 y / immunocompromised – 3 doses
JE	**SA-14-14-2 live (Chinese)** – 0.5 mL SC; **JENVAC** (inactivated, Indian) – 2 doses; **Ixiaro** (inactivated); UIP – 9 m & 16–24 m in endemic districts; adult JE campaign in endemic areas
Meningococcal	Quadrivalent conjugate (MenACWY) – travel (Hajj, Umrah), outbreaks, asplenia, complement deficiency
Cholera (oral)	Inactivated whole-cell (Shanchol/Euvichol) – 2 doses 2 weeks apart, ≥ 1 year
Yellow fever	Live 17D; **single dose ≥ 9 months** for travel to Africa/S. America; valid for life (certificate 10 days after vaccination)

Vaccine	Type & schedule
COVID-19	Covaxin (≥ 6 y), Corbevax (≥ 5 y), ZyCoV-D (≥ 12 y) – used under national programme
Dengue	Dengvaxia (seropositive only, 9–45 y – not in India); **Qdenga (TAK-003)** – WHO-prequalified 6–16 y in high-burden settings
Malaria	**RTS,S/AS01** & **R21/Matrix-M** – WHO-recommended for children in endemic Africa from 5 months (4 doses)

9.10 Rabies Prophylaxis (NRCP 2019 / WHO)

Category	Exposure	Action
I	Touching/feeding animals, **licks on intact skin**	**Wash; no prophylaxis**
II	**Nibbling of uncovered skin, minor scratches/abrasions without bleeding**	**Wound care + vaccine**
III	**Single/multiple transdermal bites or scratches, licks on broken skin, contamination of mucous membrane with saliva, bat exposure**	**Wound care + RIG + vaccine**

POST-EXPOSURE REGIMENS

- **Wound washing with soap & running water for 15 minutes** (most important first step), apply povidone-iodine/alcohol; **avoid suturing** (if essential, delay & infiltrate RIG first).
- **IM (Essen) – 5 doses: days 0, 3, 7, 14, 28** (1 vial each, deltoid/anterolateral thigh).
- **ID (Updated Thai Red Cross) – 0.1 mL at 2 sites on days 0, 3, 7 and 28** ("2-2-2-0-2").
- **RIG**: **human RIG 20 IU/kg** or **equine RIG 40 IU/kg**, or **rabies monoclonal antibody (3.33 IU/kg)**; infiltrate maximum into and around the wound, **on day 0 (up to day 7 of first vaccine dose)**; never in same syringe/site as vaccine.
- **Previously vaccinated** (complete PrEP/PEP): **2 booster doses days 0 & 3 (IM) – no RIG**.
- **Pre-exposure prophylaxis**: days **0, 7 and 21 or 28** (WHO 2018: days 0 & 7) – vets, lab staff, children in high-risk areas.
- Observe dog/cat for **10 days** – if healthy, rabies excluded (but don't delay PEP).

9.11 Cold Chain & Vaccine Logistics

Level	Equipment / temperature
State/regional/district stores	**Walk-in freezers (−20 °C) for OPV**; walk-in coolers (+2 to +8 °C)
PHC / cold chain point	**Ice-lined refrigerator (ILR) +2 to +8 °C** (all vaccines incl. OPV); **deep freezer −15 to −25 °C** (ice packs, OPV)
Outreach session	**Vaccine carrier with 4 conditioned ice packs** (keeps cold ~12 h); **cold box** for transport/storage up to 5–7 days
Ice packs	**Conditioned** (sweating, water heard on shaking) to avoid freezing freeze-sensitive vaccines

- **Heat sensitivity (most → least)**: **OPV > measles/MR > BCG (reconstituted) > DPT/Penta, JE > Hep B, TT/Td. Reconstituted MR/BCG/JE are most heat- & light-sensitive.**
- **Freeze sensitivity (most → least)**: **Hep B > Hib (liquid)/Penta > DPT > DT > Td/TT > IPV, PCV, HPV. Never freeze**: anything with aluminium adjuvant (absorbed vaccines).
- **Shake test** – to detect freeze damage of adsorbed vaccines (frozen sample sediments faster with flakes).

- **Vaccine vial monitor (VVM)**: heat-sensitive label on vial; use if **inner square lighter than outer circle**; **discard when inner square matches or is darker than circle** (stage 3/4). VVM shows cumulative heat exposure, **not freezing**.
- **Open vial policy (India)**: opened multi-dose vials of **OPV, DPT, Td, Hep B, Pentavalent, PCV, fIPV** may be used in subsequent sessions **up to 28 days** if expiry not passed, VVM usable, cold chain maintained, no contamination. **Not applicable to reconstituted BCG, MR/measles, JE** – discard **4 hours** after reconstitution or at end of session (whichever earlier).
- **Diluent** must be cooled (stored with vaccine 24 h before use) to avoid thermal shock.

9.12 Adverse Events Following Immunisation (AEFI)

Category (WHO)	Example
Vaccine product-related	**VAPP** (OPV), ITP (MMR), intussusception (rotavirus), febrile seizure (DTwP, MMR)
Vaccine quality defect	Incompletely inactivated vaccine
Immunisation (programme) error	**Toxic shock syndrome** (reused reconstituted measles vial), abscess (non-sterile technique, wrong route – e.g. BCG given SC), sciatic nerve injury (gluteal injection)
Immunisation anxiety (stress)	Fainting (syncope) – adolescents
Coincidental	Unrelated illness

- **Serious AEFI** (death, hospitalisation, disability, cluster) → report within **24 h**, investigate within **7–10 days**; **anaphylaxis is the commonest serious acute reaction – keep adrenaline (AEFI kit)** at every session; observe **30 minutes** after vaccination.

Practice MCQs · Immunization

77 questions · correct option highlighted in green · cover the answer strip to self-test

National Immunization Schedule

1. Which vaccines are recommended at birth as per the universal immunization programme?

A. BCG
B. **BCG & Hepatitis B**
C. BCG & DPT
D. DT & OPV

Ans B Vaccines recommended at birth as per the universal immunization programme are: BCG, OPV zero dose & Hepatitis B vaccine.

2. Which is not given at the time of birth?

A. OPV
B. BCG
C. Hepatitis B
D. **Hib vaccine**

Ans D

3. Rabies pre-exposure prophylaxis includes how many doses of vaccine?

A. 1
B. 2
C. **3**
D. 4

Ans C

4. About Hepatitis A vaccine schedule, true is:

A. Recommended at age of 12 months
B. 2 dose of killed vaccine 6 months apart
C. 1 dose of live vaccine
D. **All are true**

Ans D As per IAP, Two doses of both killed and live hepatitis A vaccines were recommended in 201314; IAP ACVIP recommends initiating hepatitis A vaccine at the age of 1223 months & separate the 2 doses by 6 to 18 months

5. An un-immunized 13 months old child comes to you in OPD, according to the latest immunization schedule, what vaccines will you advise?

A. OPV 3 doses, 1 IPV, 3 Pentavalent
B. BCG, OPV 3 doses, 3 IPV, 3 Pentavalent and 1 measles
C. **OPV 3 doses, 1 IPV, 3 Pentavalent and 2 measles**
D. OPV 3 doses, 3 IPV, 3 DPT, 3 Hep B

Ans C Catch up vaccination with BCG is not recommended beyond 1 year age as per the national immunisation schedule.

BCG & Tuberculosis Vaccines

6. Which vaccine cannot be given to a child < 2 years age?

A. BCG
B. Pneumococcal conjugate
C. **Pneumococcal polysaccharide**
D. Haemophilus influenzae

Ans C

7. Active and passive immunity should be given together in all except:

A. Tetanus
B. Rabies
C. **Measles**
D. Hepatitis B

Ans C

8. BCG vaccine gives protection against:

A. CNS TB and pulmonary TB
B. **CNS TB and disseminated TB**
C. Pulmonary TB
D. Pulmonary TB and skeletal TB

Ans B CNSTB & and disseminated TB BCG is 50% e ective in preventing pulmonary tuberculosis in adults and children. The protective e ect for disseminated and meningeal tuberculosis appears to be slightly higher, with BCG preventing 50–80% of cases. ff ff

9. Which is true about BCG?

A. Distilled water is used as diluent
B. Site for injection is cleaned with spirit
C. Mantoux test positive in 6 weeks
D. **WHO recommends Danish 1331 for vaccine production**

Ans D Nelson's 20/e p 3212, Ghai, 8/e p 190-191 Discussing about the options one by one, Option a False; Sterile normal saline may be used as diluent for BCG Option b False; For BCG Vaccination, the selected site may be swabbed clean using sterile saline and local antiseptics should be avoided Option c False; Of BCG vaccine recipients whose initial tuberculin skin test is negative, 10%–25% will have a positive tuberculin skin test if they are retested within 1–4 weeks—the so-called "booster phenomenon" Option d True;

10. Strain used for BCG vaccine:

A. Edmonston Zagreb strain
B. Oka strain
C. **Danish 1331**
D. RA 27/3 strain POLIO VACCINE

Ans C Ghai, 8/e p 190-191

11. A 11-month-old child came for vaccination. He had received Polio vaccine & BCG at birth. What vaccines should be given to him now?

A. BCG, OPV, Hep B
B. **Measles, DPT, OPV, Hib, Hep B**
C. DPT, OPV, Hib, Hep B
D. DPT, OPV, Hep B

Ans B IAP 115 All the above vaccines should be given for catch up immunization now.

Polio Vaccines (OPV/IPV)

12. Active immunity is not acquired by:

A. Infection
B. Vaccination
C. Immunoglobulin transfer
D. Subclinical infection

Ans C Immunoglobulins give passive immunity & not active immunity.

13. Antisera is obtained from:

A. Guinea pig
B. Rabbit
C. Rat
D. Horse

Ans D Antisera are usually prepared in horses; they provide passive immunization; Antisera are used against diseases like Diphtheria, Tetanus, Rabies

14. Vaccine associated paralytic poliomyelitis is defined as those cases of acute flaccid paralysis:

A. Who have residual weakness 6 months after the onset of paralysis and from whose stool samples, vaccine related poliovirus but no wild virus is isolated.
B. Who have residual weakness 60 days after the onset of paralysis and from whose stool samples, vaccine related poliovirus but no wild virus is isolated.
C. Who have residual weakness 6 days after the onset of paralysis and from whose stool samples, vaccine related poliovirus but no wild virus is isolated.
D. Who have residual weakness 60 days after the onset of paralysis and from whose stool samples, wild polio virus is isolated.

Ans B Who have residual weakness 60 days after the onset of paralysis and from whose stool samples, vaccine related poliovirus but no wild virus is isolated Vaccine associated paralytic poliomyelitis (VAPP) is defined as those cases of AFP which have residual weakness 60 days after the onset of paralysis and from whose stool samples, vaccinerelated poliovirus but no wild poliovirus is isolated.

15. Which of the following serotypes of Polio virus is most commonly associated with vaccine associated paralytic poliomyelitis?

A. Serotype
B. Serotype
C. Serotype
D. Serotype 1 and

Ans C

16. A PHC medical officer wants to give pulse polio immunization. The following finding in the Vaccine Vial Monitor (VVM) indicates that the vaccine should not be used:

A. Inner square is white, outer circle is blue
B. Inner square is blue but lighter than outer circle
C. Inner square matches the color of the outer circle
D. Inner square is lighter than outer circle

Ans C Interpretation of the color change of VVM is as follows:
• Inner square is white, or lighter than outer circle: If the expiry date has not passed, vaccine can be used
• Inner square matches color of outer circle or is darker than outer circle: Vaccine should be discarded, regardless of the expiry date.

17. OPV Bivalent vaccine contain:

A. Oral Polio vaccine
B. BCG vaccine
C. DPT vaccine
D. Measles vaccine

Ans B P1 & P3 • Monovalent OPVs (mOPVs) & bivalent OPV (bOPV) are 2.5–3 times more efficacious than trivalent OPV
• In bivalent OPV, the type 2 component is removed

18. Zero dose of polio vaccine is given:

A. Before giving DPT
B. At birth
C. When child is having diarrhea
D. When child is having polio

Ans B OPV zero dose: At birth or as early as possible within the first 15 days.

19. The efficiency of cold chain system for oral polio vaccine as monitored by Vaccine Vial Monitor (VVM) depends on:

A. Change in the colour of vaccine
B. Temperature indicator of the system
C. Viral potency test
D. Change in colour of monitor DPT & TT VACCINE

Ans D Change in colour of monitor Vaccine Vial Monitor (VVM)
• It is a time and temperature sensitive colored label that provides an indication of cumulative heat to which the vial has been exposed
• Whether the OPV vial can be used or not depends upon the irreversible colour changes in vaccine vial monitor. DPT &

20. An 18-month-old child, who has received one dose of DTP and OPV at 2 months of age. What will be your next immunization plan?

A. When child is receiving 2 mg/kg of prednisolone for at least 2 weeks at present
B. When child has received 2 mg/kg of prednisolone for at least 2 weeks in the past one month
C. Child receiving 4 mg/kg of prednisolone for 4 weeks, prior to 2 weeks
D. Chronic asthmatic child receiving low dose inhaled steroid for 10 months

Ans B Measles, BCG, booster dose of DPT and OPV , IAP 115 For catch up immunisation, vaccines to be given on the first visit: 1 dose of all the vaccines that the child should have received till now, but did not receive, are to be given including Measles, OPV & DPT. BCG may be given with other live vaccines on the same day.

DTP & Combination Vaccines

21. Which of the following vaccines is given subcutaneously?

A. BCG
B. DPT
C. Hep B
D. **Measles**

Ans D

22. Which of the following is a live vaccine?

A. Salk polio vaccine
B. **Sabin polio vaccine**
C. Hepatitis B vaccine
D. Rabies vaccine

Ans B

23. Which of the following is true regarding whole cell pertussis vaccine?

A. Maternal antibody provides protection against pertussis
B. Effectiveness against pertussis is more than diphtheria/ tetanus
C. **Progressive neurological deficit is an absolute contra indication**
D. Not recommended for children more than 7 years of age

Ans C Progressive neurological deficit is an absolute contrai ndication Pregnant women should be given Tdap during every pregnancy to provide passive antibody protection to the infant until administration of DTaP. Optimal timing of maternal Tdap is 26 through 37 wk of gestation but Tdap can be given at any time during pregnancy. Progressive neurological deficit and anaplnglasis to previous doses is a contraindication for pertussis vaccination

24. Whole cell pertussis vaccine used to prevent infection can protect against?

A. Bortedella parapertussis only
B. Mild forms of Bordetella pertussis only
C. **Both Bordetella pertussis & parapertussis**
D. Only against severe Pertussis

Ans C Pertussis vaccine may induce crossimmunity & has been found to have 66%82% effectiveness against parapertussis. Controversial; Some sources say: As measles can depress cell mediated immunity & as BCG induces cell mediated immunity BCG should not be given along with measles or MMR; while some do not comment about it; So, there is not much recent evidence to substantiate the same.

25. Pertussis vaccine side effect:

A. Local pain
B. Excessive cry
C. Fever
D. **All of the above**

Ans D with Adverse effects of Pertussis vaccination are: Minor: like pain, swelling and redness at the local site, fever, fussiness, anorexia and vomiting Serious adverse effects are persistent crying (most common), fever > 40.5°C, hypotonic hyporesponsive episodes , seizures & encephalopathy.

26. All of the following statements are true about DPT vaccine except:

A. **It should be stored in deep freezer**
B. Exposure to direct sunlight when in use should be avoided
C. Stored stocks are needed for the months at PHC level
D. Half used vials should not be put back into the cold chain after the session

Ans A • Vaccines susceptible to damage by freezing include mainly all aluminium adjuvanted vaccines (DTwP, DTaP, TT, DT, Td, TT, hepatitis B, combination vaccines, hepatitis A, HPV, PCV 7) but also other vaccines including IPV, PPV 23, inactivated influenza vaccines, meningococcal vaccines, rotavirus vaccines, typhoid vaccines, & Hib and vaccines
• Vaccines that can be frozen without harm include OPV (vial must not be frozen and thawed repeatedly), and lyophilized

27. Which one of the following doses in Loffler ... Diphtheria Toxoid is incorporated in DPT?

A. 5
B. 15
C. **25**
D. 35

Ans C 25 Diphtheria toxoid 20–30 Lf & tetanus toxoid 5–25 Lf in DPT vaccine.

28. Adjuvant used in DPT is:

A. Silica
B. Magnesium
C. Manganese
D. **Aluminium HIB & PNEUMOCOCCAL VACCINE Questions**

Ans D Aluminium HIB &

Measles & MMR Vaccines

29. According to latest guidelines of vaccination, which of the following is applicable at the age of 5 years?

A. DT booster + Vitamin A
B. DT
C. DPT + OPV
D. **DPT + Vitamin A**

Ans D DPT booster & Vitamin A supplementation are recommended at 5 years age.

30. Which one of the following is a conjugated vaccine:

A. Hepatitis B
B. Rubella
C. **Haemophilus influenzae**
D. Pertussis

Ans C

31. True regarding MMR vaccine is:

A. May lead to local reaction in approx. 10% of vaccinated population
B. It is a live vaccine
C. It can be given at age 9 months
D. MMR can be given to immunocompromised but not to pregnant females
E. Jeryl Lynn strain has the lowest incidence of post vaccine aseptic meningitis

Ans B d. MMR can be given to immunocompromised but not to pregnant females & e. Jeryl Lynn strain has the lowest incidence of post vaccine aseptic meningitis Miller et al. Am J Epidemiol (2007) 165 (6): 704-709. Discussing about the options one by one, a. False; Adverse events from MMR vaccine include fever (usually 612 days following vaccination), rash in 5% of vaccinated persons, and, rarely, transient thrombocytopenia b True; MMR is a live vaccine c. False; 2 doses of MMR vaccine are recommended;

32. In measles outbreak, measles vaccine can be given within:

A. 23 months
B. 35 months
C. 27 months
D. 69 months

Ans D 6-9 months In case of an outbreak, the measles vaccine can be given to infants as young as 6 completed months of age.

33. Name of mumps vaccine is:

A. Jeryll Lynn
B. Edmonston
C. Danish 1331
D. OKA

Ans A Mumps vaccine virus strains include Leningrad-Zagreb, Leningrad-3, Jeryl Lynn strains and are grown in chick embryo/ human diploid cell cultures.

34. Which type of vaccine is MMR?

A. Live attenuated
B. Killed
C. Toxoid
D. Subunit RABIES IMMUNISATION

Ans A Commonly used live attenuated vaccines include BCG, oral polio, measles, MMR and chicken pox vaccines

Hepatitis B Vaccine

35. Killed vaccine is:

A. Hepatitis A
B. Measles
C. OPV
D. BCG

Ans A

36. Live vaccines are all except:

A. BCG
B. Measles
C. Polio
D. Rabies
E. Typhoid

Ans D Note: Both live & killed typhoid vaccines are available

37. For the prevention of human rabies, immediate flushing and washing the wound(s) in animal bite cases, with plenty of soap and water, under running tap should be carried out for how much time?

A. 11 years
B. 9 years
C. 15 years
D. 13 years

Ans C 15 minutes In case of animal bite wounds, the 1st step is thorough cleansing of the wound with soap & flushing under running water for 10 minutes; as 15 minutes is the option closest to 10 minutes, that is the best answer.

38. A child is hepatitis B immunized, the marker for seroconversion is?

A. HBs Ag
B. Anti HBc IgM
C. Anti HBs Ag
D. Anti HBc IgG

Ans C An anti-HBs concentration of 10 mIU/ml measured 1–3 months after administration of the last dose of the primary vaccination series is considered a reliable correlate of protection against infection.

Hib & Pneumococcal Vaccines

39. Which of the following is called first immunization of the baby?

A. Colostrum
B. Handing over the baby to mother
C. OPV
D. DPT + BCG

Ans A Colostrum is rich in immunoglobulins & other substances that protect the baby against infections; So, Colostrum is called the 'First immunization' of the baby.

40. Live vaccine are:

A. BCG
B. Oral polio Vaccine
C. Chickenpox vaccine
D. MMR
E. DPT

Ans A b. Oral polio Vaccine, c. Chickenpox vaccine, d. MMR

41. All are true regarding Hib vaccine except:

A. Protects against non capsulated H. influenza disease
B. Catch up vaccination before 5 years
C. Can be given in less than 6 months
D. Given 2 weeks before splenectomy

Ans A Hib conjugate vaccine It protects against encapsulated H. influenzae type B strain only. Routine vaccination:
• Minimum age: 6 weeks
• Primary series includes Hib conjugate vaccine at ages 6, 10, 14 weeks with a booster at age 12 through 18 months. Catchup vaccination:
• Catchup is recommended till 5 years of age.
• 6–12 months; 2 primary doses 4 weeks apart and 1 booster;
• 12–15 months: 1 primary dose and 1 booster;
• Above 15 months: single dose.

42. A child unimmunized previously with Hib vaccine, received 1st dose of Hib vaccine at 16 months, the next appropriate step is:

A. Give 3 doses of Hib at 2 months interval
B. **No further doses needed**
C. Give one booster dose
D. Give two booster doses at 4 weeks interval MEASLES & MMR VACCINE

Ans B No further doses needed MEASLES &

Rotavirus Vaccine

43. Not given in adolescent:

A. MMR vaccine
B. Tetanus vaccine
C. **Rotavirus vaccine**
D. HPV vaccine

Ans C • The maximum age for the first dose of Rotavirus vaccine is 14 weeks, 6 days, & final dose is 8 months
• Vaccination should not be initiated for infants aged 15 weeks or older with

44. Vaccine (s) not to be frozen is/are

A. BCG
B. OPV
C. **DPT**
D. Measles
E. TT

Ans C DPT, e. TT

45. Rotavirus vaccine- contraindication is:

A. SCID
B. Intussusception
C. Severe allergic reaction
D. **All of the above**

Ans D 2 doses of inactivated vaccine at least 6 months apart or only a single dose of live attenuated vaccine needs to be given.

46. Incorrect matches are:

A. **Measles: JerylLynn strain**
B. Chickenpox: OKAstrain
C. Oral polio: Sabin
D. Rubella: Edmonton Zagreb strain VACCINATION IN SPECIAL SITUATIONS

Ans A Measles: Jeryl-Lynn strain and d. Rubella: Edmonton Zagreb strain For Measles vaccine: Edmonston Zagreb strain is used; For Rubella vaccine: RA 27/3 vaccine strain is used.

HPV Vaccine

47. Mass vaccination is ineffective in:

A. Measles
B. Poliomyelitis
C. **Tetanus**
D. None of the above

Ans C 'Herd effect'
• It is defined as "reduction of infection or disease in unimmunized segment as a result of immunizing a proportion of population"
• Herd effect is due to reduced carriage of the causative microorganism by the vaccinated cohort and thus is seen only with vaccines against those diseases where humans are the only source
• An effective vaccine is a prerequisite for good herd effect; e.g. conjugated pneumococcal vaccine
• Tetanus and BCG vaccines have no herd effect So, mass vaccination is ineffective

48. Administration of which vaccine can result in paralysis in children?

A. Measles vaccine
B. **Sabin polio vaccine**
C. DT vaccine
D. DPT vaccine

Ans B

49. Which among the following is vaccine for cholera?

A. PPV
B. **Dukoral**
C. Ty 21 a vaccine
D. JEMB Nakayama

Ans B • The WC-rBS vaccine available internationally as Dukoral oral vaccine is widely used against Cholera
• It is a vaccine comprising of killed V. cholerae O1 with recombinant b subunit of cholera toxoid.

50. Which of the following Human papilloma virus subtypes are not covered by Quadrivalent Anti-cervical cancer vaccine?

A. Type
B. **Type**
C. Type
D. Type
E. Type

Ans B 7 /e p 200-201 Quadrivalent HPV vaccine contains serotypes 16, 18, 6 & 11 with aluminium containing adjuvant.

51. True regarding Cervical cancer vaccine is/are:

A. Bivalent and quadrivalent
B. Given to married women in 2045 years age group
C. Protects against subtypes 16,
D. Two doses given
E. Gives 100% protection against cervical cancer

Ans A c. Protects against subtypes 16, 18 About Cervical cancer vaccine or HPV vaccine:
• Either HPV4 (0, 2, 6 months) or HPV2 (0, 1, 6 months) is recommended for females aged 11 or 12 years
• The vaccine series can be started beginning at age 9 years & can be given upto 45 years if not previously vaccinated
• 3 doses of quadrivalent HPV vaccine at 0, 2 & 6 months have shown 99% efficacy against types 16, 18 related CIN- 2/3 99–100% efficacy was

52. HPV vaccine is:

A. Monovalent
B. Bivalent
C. Quadrivalent
D. Bivalent & quadrivalent

Ans D

Typhoid & Cholera Vaccines

53. False about vaccines:

A. Thiomersal is used as preservative in DPT vaccine
B. Kanamycin is used as preservative in measles vaccine
C. Neomycin is used as preservative in BCG vaccine
D. Magnesium chloride used to stabilize OPV

Ans C & b. Kanamycin is used as preservative in measles vaccine Additional substances present in different vaccines & their function: Vaccine Additional substance (function) DPT ThiomersalQ (Preservative); Aluminium hydroxideQ (adjuvant) Measles No Preservative BCG No preservativeQ OPV Magnesium chloride (Thermostabilizer)Q

54. All of the following are killed vaccines except:

A. Salk Polio
B. Japanese encephalitis
C. Rabies
D. Yellow fever BCG VACCINE

Ans D Yellow fever

55. Which of the following statement is not true about the following vaccines?

A. Rotavirus vaccine is a live vaccine
B. Vi capsular polysaccharide Typhoid vaccine can be administered in children at 18 months of age
C. The Hib vaccine which are routinely given these days are conjugated vaccine
D. IPV vaccine can be given at 6 weeks of life

Ans B IAP ACVIP recommends the administration of the currently available Vi polysaccharide Typhoid vaccine 0.5 ml IM every three years beginning at the age of 2 years.

56. A child presented at 18 months of age who has never been vaccinated before. Which vaccines will you administer?

A. DPT Booster and OPV
B. Pentavalent vaccine
C. BCG and OPV
D. Pentavalent, MMR, OPV

Ans D A measles containing vaccine should be given in an unimmunized child beyond 9 months age. OPV should be given at less than 5 years age.

Rabies & JE Vaccines

57. All are live vaccines except:

A. Measles
B. BCG
C. DT
D. DPT
E. Measles 200 Section 1: General Pediatrics 23. Which disease is prevented by giving booster dose to a 5-6 years old child?

Ans B Rabies /e p 202-203
• For Japanese Encephalitis, Polio & Typhoid, both live & killed vaccines are available
• New generation typhoid vaccines include oral live attenuated Ty21a vaccine, Parenteral Vipolysaccharide & Vi Polysaccharide Conjugate vaccines; For Rabies, only inactivated vaccines are used.

58. Salk vaccine is a:

A. Live vaccine
B. Live attenuated vaccine
C. Killed vaccine
D. Toxoid

Ans C Ghai, 8/e p 190- Polio vaccines were developed in the USA during 1950, first the inactivated polio vaccine (IPV) by Jonas Salk and later the live oral polio vaccine (OPV) by Albert Sabin. Remember: 'Salk' is a 'killed' vaccine.

59. Which one of the following vaccines is contraindi- cated in children with egg allergy?

A. MMR
B. Yellow Fever
C. DPT
D. BCG

Ans B Immediate severe hypersensitivity or anaphylactic reactions is seen with Yellow fever vaccine, most commonly in people with allergies to eggs or gelatine.

60. All of the following are contraindicated during preg- nancy except:

A. Measles vaccine
B. Mumps vaccine
C. HPV vaccine
D. Rabies vaccine

Ans D Because rabies is a lethal disease there are no contraindications for postexposure prophylaxis including infants, pregnant & lactating women.

Influenza Vaccine

61. Killed vaccine among the following is:

A. Oral Polio Vaccine
B. Hepatitis B
C. Yellow fever
D. Japanese encephalitis

Ans B • Hepatitis B is an inactivated (killed) vaccine, while OPV & Yellow fever are live vaccines
• For Japanese encephalitis, both live & killed vaccines are available.

62. Protective levels of Tetanus anti-toxin is:

A. >0.01 IU/mL
B. >0.5 IU/mL
C. >1.0 IU/mL
D. >5 IU/mL

Ans A Protective titer for diphtheria > 0.1 IU/ml and for tetanus > 0.01 IU/ml.

63. Which of the following vaccines has maximum efficacy after a single dose?

A. Tetanus Toxoid
B. DPT
C. Measles
D. Typhoid

Ans C Efficacy with single dose of:
• Rubella: 95% (maximum) • OPV: 6580%
• Measles: 85% • BCG: 50% (maximum)
• Tetanus toxoid: 70%, So, among the given options, Measles vaccine is the best answer.

64. A 9-month-old un-immunized child was brought to the Questions dispensary, which vaccination should be given to this baby at first visit?

A. OPV + BCG
B. OPV + DPT + Measles
C. OPV + BCG + DPT + Measles
D. OPV + DPT + BCG

Ans C Recommendations for Catch up immunisation
• Vaccination catch up regimens should preferably be individualized
• Any number of vaccines live/inactivated may be given on the same day either singly or as combination vaccines maintaining a gap of 5 cm between different vaccines
• Inactivated vaccines can be given at any time in relation to any other live/ inactivated vaccines
• If not given on the same day, a gap of 4 weeks should be maintained between two live injectable vaccines

COVID-19 Vaccines

65. Vaccine with maximum efficacy

A. OPV
B. Measles
C. BCG
D. TT

Ans B IAP guidebook of Immunisation 2013-14, p 222-223 Seroconversion rates after measles vaccination are around 60% at the age of 6 months, 80–85% at the age of 9 months and beyond 95% at the age of 12–15 months.

66. A full course of immunization with 3 doses of tetanus toxoid, confers immunity for how many years?

A. 5
B. 10
C. 15
D. 20

Ans B 10 In individuals who have completed primary & booster vaccination with DTP, TT boosters every 10 yrs provide sufficient protection.

67. Which of the following vaccines can result in Thrombocytopenia?

A. MMR vaccine
B. Typhoid vaccine
C. Influenza vaccine
D. HIB vaccine

Ans A With Measles vaccine, Thrombocytopenic purpura may occur at a frequency of 1/30,000 vaccinees. One of the components of MMR is the Measles vaccine; So MMR vaccine can cause thrombocytopenia.

68. Which vaccine(s) is/are not contraindicated in Pregnancy?

A. Rubella
B. Varicella
C. Hepatitis B
D. Measles
E. Rabies 204 Section 1: General Pediatrics

Ans C e. Rabies If the mother is known to be HBsAg negative, hepatitis B vaccine can be given in the 0–6 weeks–6 months schedule; Rabies vaccine can also be given during pregnancy. Advanced M What is being duoenset iino tnhis infant? Q 1 a. Conventional CPR b. Compression only CPR c. Airway is being secured d. Cervical spine is being stabilized M What is being suheoswtni oinn the picture Q below? 2 a. W-C clamp technique b. E-C clamp technique c. B-C clamp technique d. W-E clamp technique Latest Updates

Vaccine Contraindications & Adverse Events

69. The guidelines for immunization against infectious disease includes all of the following except:

A. Live vaccines are contraindicated in pregnant women
B. Live vaccines should not be administered within 3 months of giving human immunoglobulin
C. Inactivated vaccines should not be given if there has been significant reaction to previous dose
D. Hay fever, sickle cell anemia and tuberculosis are contraindications for vaccination

Ans D IAP guidebook of Immunisation 2013-14, p 70 True contraindications to vaccination are:
• Severe allergic reaction to a vaccine component or following a prior dose of a vaccine;
• Encephalopathy occurring within 7 days of pertussis vaccination;
• Severe combined immunodeficiency (SCID) as a contraindication to rotavirus vaccine

70. Osteitis may be seen as a side effect of:

A. Hepatitis B vaccine
B. **BCG**
C. Measles vaccine
D. IPV

Ans B Ghai, 8/e p 190-191 Side effects of BCG vaccine dna • BCG adenitis: Associated with subcutaneous administration of BCG
• Disseminated BCG infection is extremely unusual but may occur in children with cellular immunodeficiency
• Osteitis is a rare complication of BCG vaccination that appears to be related to certain strains of the vaccine
• Systemic complaints such as fever, convulsions, loss of appetite, & irritability are after BCG

71. Which vaccine is contraindicated in pregnancy?

A. **Rubella**
B. Diphtheria
C. Tetanus
D. Hepatitis B

Ans A Rubella infection during pregnancy can lead to severe consequences in the fetus.

Passive Immunization

72. Immunoglobulin is not used in prophylaxis of:

A. Hepatitis A
B. Varicella
C. **Influenza**
D. Measles

Ans C Following immunoglobulins are used for secondary prophylaxis: Tetanus, Hep A, Hep B, Rabies & Varicella Ig.

73. All of the following vaccines can prevent pneumonia, except:

A. Measles
B. **Rubella**
C. Pneumococcal
D. H. influenzae

Ans B Pneumonia is the most common cause of death in measles. It may manifest as giant cell pneumonia caused directly by the viral infection or as superimposed bacterial infection. The most common bacterial pathogens are Streptococcus pneumoniae, Haemophilus influenzae, and Staphylococcus aureus. So, vaccination against Measles, Pneumococcus & H.influenzae would give some protection against pneumonia.

74. Dose of Human Rabies Immunoglobulin is:

A. 10 IU/kg
B. **20 IU/kg**
C. 30 IU/kg
D. 40 IU/kg202 Section 1: General Pediatrics

Ans B • Human rabies immunoglobulin (HRIG— dose is 20 U/kg body weight, maximum dose 1500 IU); and
• Equine rabies immunoglobulin (ERIG—dose is 40 U/kg, maximum dose 3000 IU)

Vaccine Storage & Cold Chain

75. Most heat sensitive vaccine is:

A. BCG
B. **Polio**
C. Measles
D. DPT

Ans B

76. A child who has not been vaccinated with H. influenzae comes to the OPD at 14 months. How many doses, including booster would you like to give to this child?

A. 4 doses
B. 3 doses
C. **2 doses**
D. Only one dose

Ans C Haemophilus influenzae type b (Hib) conjugate vaccine schedule: Routine vaccination Catch-up vaccination
• Minimum age: 6 weeks • Catch-up is recommended till 5 years of age.
• Primary series includes Hib conjugate vaccine at ages 6, 10, 14
• 6–12 months; 2 primary doses 4 weeks apart & 1 booster; weeks with a booster at age 12 through 18 months. • 12–15 months: 1 primary dose and 1 booster;
• Above 15 months: Single dose

77. Typhoid Vi polysaccharide vaccine is usually administered in children above the age of:

A. 6 months
B. 1 year
C. **2 years**
D. 1 year 6 months

Ans C

10

CHAPTER 10

Pediatric Basic & Advanced Life Support (PALS)



WHAT THIS CHAPTER COVERS

- Basic Life Support (BLS)
- Advanced Life Support (ALS)
- Airway Management
- Cardiac Arrest & CPR
- Shock & Resuscitation
- Vascular Access
- Arrhythmias & Defibrillation
- Post-Resuscitation Care
- Drowning & Near-Drowning
- Poisoning & Toxicology
- Trauma & Injury

60 MCQs **11** subtopics

10.1 Paediatric Assessment & Normal Values

- **Paediatric Assessment Triangle (PAT)** – "from the doorway": **Appearance** (TICLS – Tone, Interactiveness, Consolability, Look/gaze, Speech/cry), **Work of breathing**, **Circulation to skin** (pallor, mottling, cyanosis).
- Then **ABCDE** primary assessment, secondary (SAMPLE history), tertiary (investigations).
- **Most common cause of cardiac arrest in children = progressive respiratory failure/shock → hypoxia → bradycardia → asystole/PEA** (not primary VF as in adults). Hence oxygenation & ventilation are key.

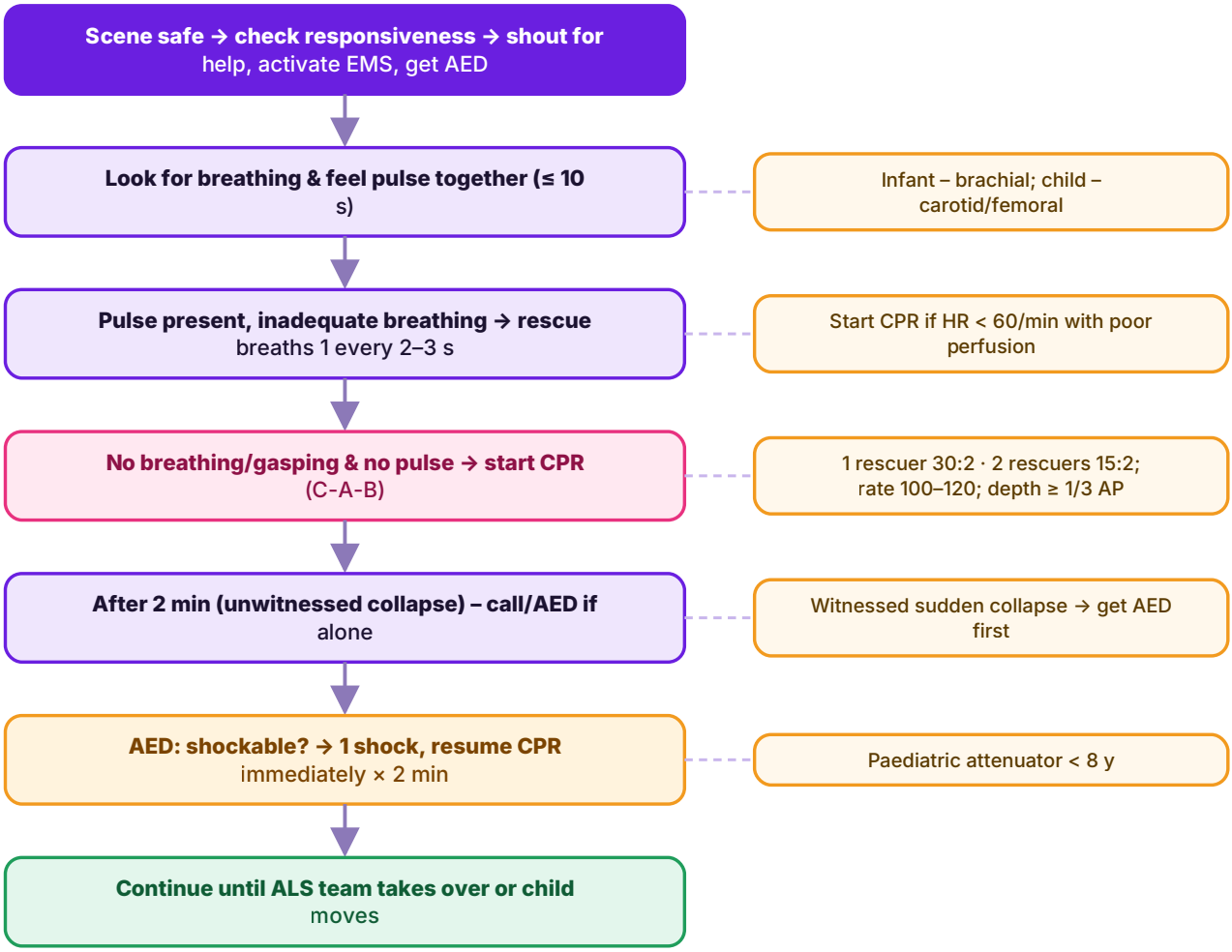
Age	HR (awake)	RR	SBP (minimum = hypotension)
Neonate	100–205	30–60	< 60 mmHg
Infant (1–12 m)	100–180	30–53	**< 70 mmHg**
1–10 years	70–140	22–37 (toddler) / 18–25 (school)	**< 70 + (2 × age in years)**
> 10 years	60–100	12–20	**< 90 mmHg**

- Normal SBP ≈ **90 + 2 × age (years)**; normal DBP ≈ 55 + 2 × age.
- **Weight estimate (APLS)**: 1–5 y: (age + 4) × 2; 6–12 y: 3 × age + 7. **Broselow tape** (length-based).
- **Endotracheal tube size** (uncuffed) = **age/4 + 4**; cuffed = **age/4 + 3.5**; **depth (lip) = 3 × ETT size** (or age/2 + 12). Neonates: 2.5 (< 1 kg), 3.0 (1–2 kg), 3.5 (2–3 kg), 3.5–4.0 (> 3 kg); depth = weight (kg) + 6 cm.
- Laryngoscope blade: **straight (Miller) blade in infants** (floppy epiglottis – lift directly); curved (Macintosh) in older children.

ANATOMICAL DIFFERENCES OF THE CHILD AIRWAY

Large occiput (neutral "sniffing" position; shoulder roll in infants), **large tongue**, **floppy U/omega-shaped epiglottis**, **higher & anterior larynx (C2–C3)**, **narrowest part historically at cricoid ring** (functional narrowing; cuffed tubes now acceptable), short trachea (endobronchial intubation risk), obligate nasal breathing in neonates, compliant chest wall, diaphragmatic breathing.

10.2 Paediatric Basic Life Support (AHA 2020)



Paediatric BLS for healthcare providers (AHA 2020)

Component	Infant (< 1 y)	Child (1 y – puberty)
Check responsiveness & breathing; pulse check	**Brachial** pulse (≤ 10 s)	**Carotid or femoral**
Sequence	**C-A-B** (compressions first)	C-A-B
Compression technique	AHA 2020: **1 rescuer – 2 fingers; 2 rescuers – 2-thumb encircling hands**. AHA 2025 update: **2-finger technique dropped – use 2-thumb encircling or heel of one hand**	**1 or 2 hands** (lower half of sternum)
Depth	**At least 1/3 AP diameter – ~4 cm (1.5 in)**	**≥ 1/3 AP – ~5 cm (2 in)**; adolescents 5–6 cm
Rate	**100–120/min**	100–120/min
Compression : ventilation (no advanced airway)	**30 : 2 (1 rescuer), 15 : 2 (2 rescuers)**	Same
With advanced airway	Continuous compressions + **1 breath every 2–3 s (20–30/min)**	Same
Rescue breathing (pulse present, no breathing)	**1 breath every 2–3 s (20–30/min)**	Same

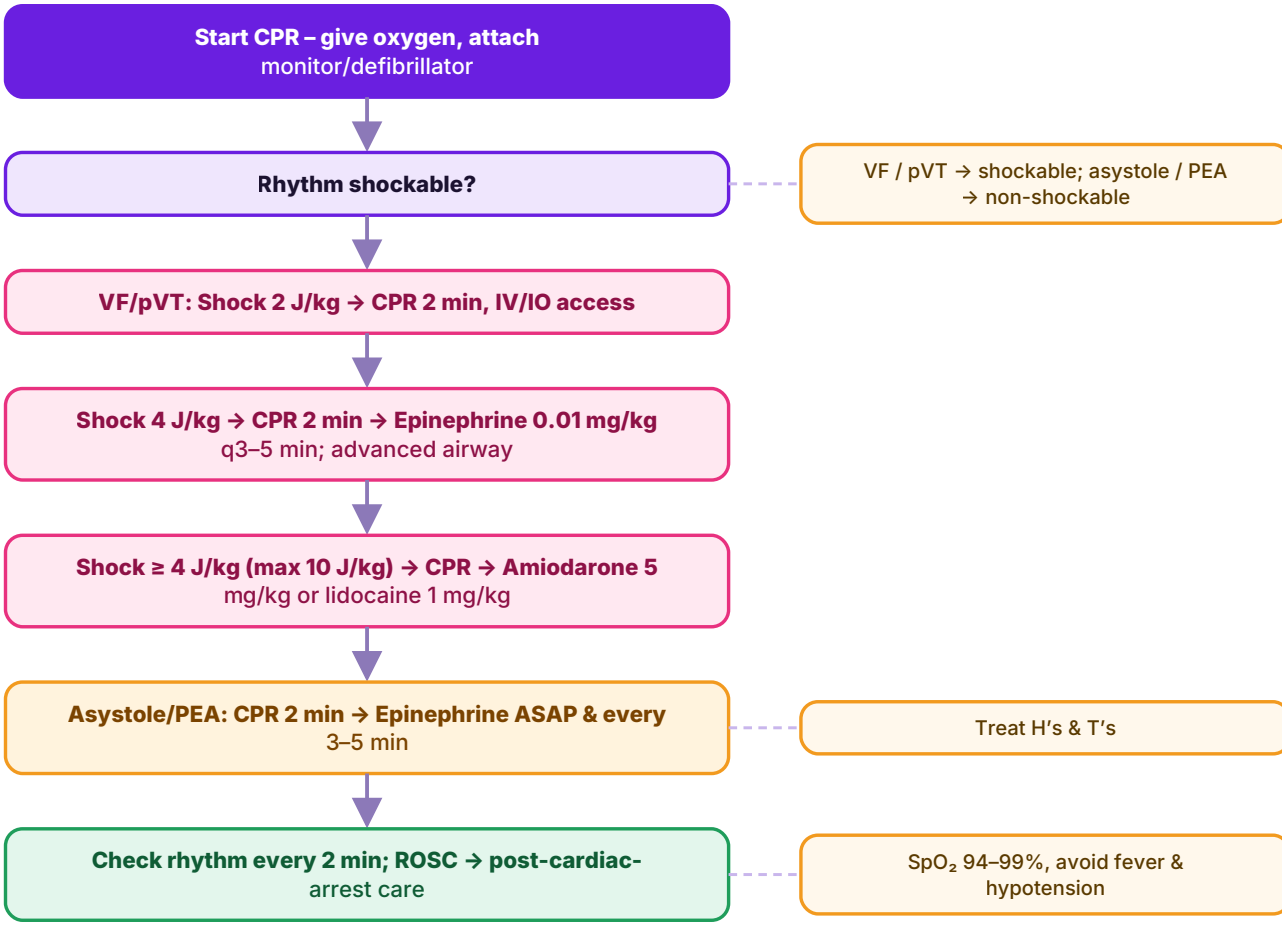
Component	Infant (< 1 y)	Child (1 y – puberty)
Start CPR if pulse present but	**HR < 60/min with poor perfusion** despite oxygenation/ventilation	Same

- **Lone rescuer, witnessed sudden collapse** → activate EMS & get AED first (likely arrhythmia). **Unwitnessed** → **2 minutes (5 cycles) of CPR first**, then call/AED.
- Allow **full chest recoil**, minimise interruptions (< 10 s), rotate compressors every 2 min, avoid excessive ventilation.
- **AED**: use **paediatric dose attenuator** for < 8 y; if unavailable, adult AED acceptable (even in infants).

Foreign body airway obstruction (choking)

Age	Conscious, ineffective cough
Infant	**5 back blows + 5 chest thrusts** (head down) – **no abdominal thrusts** (liver injury)
Child > 1 y	**Abdominal thrusts (Heimlich manoeuvre)**
Unconscious (any age)	Start CPR; look in mouth before breaths – **no blind finger sweeps**

10.3 Paediatric Advanced Life Support – Cardiac Arrest



PALS cardiac arrest algorithm (AHA 2020)

Rhythm	Management
Non-shockable: asystole / PEA (MC in children)	CPR → **epinephrine ASAP (within 5 min)** → **repeat every 3–5 min**; treat reversible causes

Rhythm	Management
Shockable: VF / pulseless VT	Shock **2 J/kg** → CPR 2 min → **4 J/kg** → CPR → **epinephrine** after 2nd shock → shock **≥ 4 J/kg (max 10 J/kg or adult dose)** → **amiodarone 5 mg/kg** (or lidocaine 1 mg/kg) after 3rd shock (repeat amiodarone up to 3 doses)

RESUSCITATION DRUGS (CHILD)

- **Epinephrine IV/IO 0.01 mg/kg = 0.1 mL/kg of 1:10,000 (0.1 mg/mL)**; max 1 mg; every 3–5 min. **ET dose 0.1 mg/kg (1:1000)**.
- **Amiodarone 5 mg/kg** bolus (max 300 mg), up to 15 mg/kg/day. **Lidocaine 1 mg/kg**.
- **Atropine 0.02 mg/kg** (min 0.1 mg, max 0.5 mg) – bradycardia due to ↑ vagal tone/AV block.
- **Adenosine 0.1 mg/kg rapid push (max 6 mg), 2nd dose 0.2 mg/kg (max 12 mg)** – SVT.
- **Calcium chloride 10% 20 mg/kg (0.2 mL/kg)** – hypocalcaemia, hyperkalaemia, Ca-channel blocker overdose.
- **Sodium bicarbonate 1 mEq/kg** – hyperkalaemia, TCA overdose, prolonged arrest.
- **Glucose 0.5–1 g/kg (2–4 mL/kg of 25% / 5–10 mL/kg of 10% dextrose)**.
- **Naloxone 0.1 mg/kg** (opioids).
- **Drugs via ET tube – "NAVEL"**: **N**aloxone, **A**tropine, **V**asopressin, **E**pinephrine, **L**idocaine (surfactant also given via ETT in neonates).

- **Reversible causes – 6 H's & 5 T's**: **H**ypovolaemia, **H**ypoxia, **H**ydrogen ion (acidosis), **H**ypo-/hyperkalaemia, **H**ypothermia, **H**ypoglycaemia; **T**ension pneumothorax, **T**amponade, **T**oxins, **T**hrombosis (pulmonary/coronary).
- **Quality**: **$ETCO_2$ ≥ 20 mmHg**; arterial diastolic BP ≥ 25 (infant) / ≥ 30 mmHg (child). Abrupt rise in $ETCO_2$ → ROSC.

10.4 Vascular Access

- **Intraosseous (IO) access** – if IV not obtained within **60–90 s** (or immediately in arrest); site: **proximal tibia (1–3 cm below tibial tuberosity, anteromedial flat surface)** – MC; distal tibia, distal femur, proximal humerus (older). **All drugs, fluids and blood can be given IO**; aspirate can be used for labs (not CBC after drugs). Complications: extravasation, compartment syndrome, osteomyelitis, fracture; avoid fractured bone.
- Others: peripheral IV, central venous (femoral safest during CPR), **umbilical venous catheter in neonates** (insert 2–4 cm until blood flow in emergency).

10.5 Shock in Children

Type	Causes	Features
Hypovolaemic (MC in children worldwide)	**Diarrhoea/dehydration**, haemorrhage, burns	Tachycardia, cold peripheries, ↑ CRT, low CVP
Distributive (septic – 2nd MC; anaphylactic, neurogenic)	Sepsis, anaphylaxis, spinal injury	Warm (vasodilated) or cold (vasoconstricted) shock; **neurogenic – hypotension with bradycardia**
Cardiogenic	Myocarditis, cardiomyopathy, CHD, arrhythmia	**Hepatomegaly, gallop, crepitations**, worsens with fluids
Obstructive	Tension pneumothorax, tamponade, **duct-dependent lesions closing (CoA, HLHS)**, PE	Distended neck veins, absent femoral pulses (duct)

- **Compensated shock**: normal SBP with signs of poor perfusion (**tachycardia earliest**, CRT > 2 s, cool skin, weak peripheral pulses, oliguria). **Hypotensive (decompensated)** = late, ominous.
- **Capillary refill time**: press on sternum/fingertip 5 s – **normal ≤ 2 s**.

SEPTIC SHOCK (SURVIVING SEPSIS CAMPAIGN PAEDIATRICS 2020)

- **Fluid bolus 10–20 mL/kg balanced crystalloid (RL/Plasma-Lyte) over 5–10 min**, reassess after each; up to **40–60 mL/kg in first hour** if ICU available & no fluid overload (**no bolus in non-hypotensive children where ICU unavailable** – FEAST trial).
- **Broad-spectrum antibiotics within 1 hour** (after blood culture).
- **Fluid-refractory shock → epinephrine or norepinephrine** (not dopamine) – can be started peripherally; **cold shock → epinephrine; warm shock → norepinephrine**.
- Catecholamine-resistant → **hydrocortisone** (stress dose 50–100 mg/m²/day).
- Targets: CRT ≤ 2 s, normal HR/BP, warm extremities, urine output > 1 mL/kg/h, normal sensorium, **lactate clearance**, $ScvO_2$ > 70%.
- **Cardiogenic shock**: small boluses (5–10 mL/kg) cautiously; inotropes (**milrinone**, dobutamine, epinephrine).

10.6 Arrhythmias

Rhythm	Recognition	Treatment
Sinus bradycardia with poor perfusion	HR < 60	Oxygenate/ventilate → CPR if < 60 with poor perfusion → **epinephrine**; atropine for vagal/AV block; pacing
Sinus tachycardia	**Infant < 220, child < 180**, variable, P waves present, history (fever, pain, hypovolaemia)	Treat cause
SVT (MC tachyarrhythmia in children; AVRT via accessory pathway – WPW)	**Infant ≥ 220, child ≥ 180/min**, fixed RR, **absent/abnormal P waves**, abrupt onset, narrow QRS (≤ 0.09 s)	**Stable: vagal manoeuvres (ice-bag to face in infants; no carotid massage/eyeball pressure) → adenosine 0.1 mg/kg → 0.2 mg/kg. Unstable: synchronised cardioversion 0.5–1 J/kg → 2 J/kg**. Prophylaxis – propranolol/digoxin (**avoid digoxin & verapamil in WPW; verapamil contraindicated < 1 y**)
VT with pulse	Wide QRS (> 0.09 s)	Stable – amiodarone/procainamide (expert); unstable – **synchronised cardioversion 0.5–1 → 2 J/kg**
VF / pulseless VT	Chaotic	**Defibrillation 2 → 4 J/kg**
Congenital complete heart block	Maternal **anti-Ro/La (SLE, Sjögren)**; HR 40–80	Pacemaker if symptomatic/HR < 55 (neonate)
Long QT (Jervell–Lange-Nielsen – AR, deafness; Romano–Ward – AD)	QTc > 0.46 s; **torsades**	β-blockers; **Mg sulfate 25–50 mg/kg** for torsades

10.7 Post-Resuscitation Care

- Avoid hypoxaemia & hyperoxia (**SpO_2 94–99%**), normocapnia, **avoid hypotension** (SBP > 5th centile), **targeted temperature management – avoid fever (≤ 37.5 °C)**; continuous EEG for seizures; glucose control; treat cause.
- Prognosis unreliable before 72 h.

10.8 Drowning

- **Drowning** – respiratory impairment from submersion/immersion (outcome: death/morbidity/no morbidity). "Near-drowning", "dry/wet" are obsolete terms.
- 2nd MCC accidental death in children (toddlers – bathtubs/buckets, adolescents – open water).
- **Hypoxia is the primary insult**; aspiration of fresh vs salt water clinically **similar** (surfactant washout → ARDS, non-cardiogenic pulmonary oedema).

- Management: **start with 5 rescue breaths** (drowning – A-B-C), then CPR 30:2/15:2; **C-spine immobilisation only if diving/trauma suspected**; no abdominal thrusts to drain water; remove wet clothes, rewarm; observe ≥ 6–8 h (delayed pulmonary oedema).
- **"Nobody is dead until warm and dead"** – continue resuscitation in hypothermic patients until core temp > 32–35 °C.
- Poor prognostic factors: **submersion > 5–10 min**, delayed CPR (> 10 min), **GCS ≤ 5/fixed dilated pupils on arrival**, asystole, pH < 7.1, need for CPR > 25 min.

10.9 Poisoning & Toxicology

Poison	Clinical clues	Antidote / specific Rx
Kerosene / hydrocarbons (MC poisoning in Indian children)	**Aspiration pneumonitis (MC complication)**, cough, fever, CNS depression	**No emesis, no gastric lavage**; oxygen, supportive; observe 6 h, CXR
Organophosphates	**Muscarinic (DUMBELS – Diarrhoea, Urination, Miosis, Bronchorrhoea/ bradycardia, Emesis, Lacrimation, Salivation), nicotinic (fasciculations, weakness), CNS**; ↓ RBC/plasma cholinesterase	**Atropine 0.02–0.05 mg/kg, double every 5 min until drying of secretions (endpoint – clear chest, HR > 80)**; **pralidoxime (2-PAM)** early; decontaminate skin
Paracetamol	Toxic dose **> 150 mg/kg** (> 200 in < 6 y); 4 stages – hepatotoxicity at 72–96 h; Rumack–Matthew nomogram (4-h level)	**N-acetylcysteine** (most effective within 8–10 h); IV 21-h regimen
Iron	**> 60 mg/kg elemental = serious**; vomiting, haematemesis, latent phase, shock/acidosis, hepatic failure, **pyloric stricture (late)**; radiopaque tablets on X-ray	**Desferrioxamine IV (vin rosé urine)**; whole-bowel irrigation; **charcoal doesn't bind iron**
Salicylates	**Respiratory alkalosis + high-AG metabolic acidosis**, tinnitus, hyperthermia, hypoglycaemia	Urinary **alkalinisation ($NaHCO_3$)**, haemodialysis
Tricyclic antidepressants	Anticholinergic, seizures, **wide QRS > 100 ms**, arrhythmias	**Sodium bicarbonate**
Opioids	**Pinpoint pupils, respiratory depression, coma**	**Naloxone 0.1 mg/kg**
Benzodiazepines	Sedation	Flumazenil (rarely; seizure risk)
Datura (anticholinergic)	**"Hot as a hare, dry as a bone, red as a beet, blind as a bat, mad as a hatter"** – mydriasis, dry skin, tachycardia, urinary retention, delirium	**Physostigmine**
Lead (chronic)	Pica, abdominal colic, anaemia (**basophilic stippling**), encephalopathy, **lead lines (metaphyseal) on X-ray**, Burton's line; ↑ blood lead (**≥ 3.5 µg/dL** CDC reference; chelate if ≥ 45)	**Succimer (DMSA) oral** (45–69 µg/dL); **BAL + $CaNa_2EDTA$** if ≥ 70 or encephalopathy
Snake bite – neurotoxic (cobra, krait)	Ptosis, ophthalmoplegia, respiratory paralysis (**krait – painless night bite, abdominal pain**)	**Polyvalent anti-snake venom (ASV) – 10 vials (same dose for children)**; **neostigmine + atropine** for cobra; ventilation
Snake bite – haemotoxic (viper)	Local swelling, **20-min whole blood clotting test (20WBCT) – non-clotting**, bleeding, AKI	ASV 10 vials, repeat every 6 h based on 20WBCT
Scorpion sting (Mesobuthus tamulus)	Autonomic storm – **hypertension, pulmonary oedema, myocarditis**, priapism, sweating	**Prazosin (α-blocker) – DOC**; scorpion antivenom

Poison	Clinical clues	Antidote / specific Rx
Carbon monoxide	Headache, **cherry-red** (late), SpO_2 falsely normal; ↑ COHb	**100% oxygen**, hyperbaric O_2
Cyanide	Bitter-almond smell, lactic acidosis	**Hydroxocobalamin**, sodium nitrite + thiosulfate
Digoxin	Vomiting, visual (yellow), arrhythmias, hyperkalaemia	**Digoxin-specific Fab**
Methaemoglobinaemia (nitrates, dapsone, local anaesthetics)	**Chocolate-brown blood, cyanosis unresponsive to O_2**, SpO_2 ~85%	**Methylene blue 1–2 mg/kg** (not in G6PD def. – use ascorbic acid/exchange)
Dhatura/Oleander (yellow)	Cardiac glycoside-like	Fab, atropine
Button battery ingestion	Oesophageal impaction → **liquefactive necrosis within 2 h** – "double-ring/halo" sign on X-ray	**Emergency endoscopic removal**; honey (≥ 1 y) en route

DECONTAMINATION RULES

Activated charcoal 1 g/kg within 1 hour for most ingestions; **does NOT bind: iron, lithium, alcohols, hydrocarbons, acids/alkalis, heavy metals (lead), potassium** ("PHAILS" – Pesticides(some), Hydrocarbons, Acids/Alkalis, Iron/Ions, Lithium, Solvents). **Gastric lavage & ipecac – no longer routine. Corrosives: no emesis, no lavage, no neutralisation**; endoscopy within 12–24 h.

10.10 Trauma & Injury

- **Injury is the leading cause of death in children > 1 year**; road traffic injuries, falls (MC non-fatal), drowning, burns.
- Primary survey **ABCDE** (catastrophic haemorrhage first), C-spine protection.
- **Children have greater physiological reserve** – hypotension appears only after **25–30% blood loss**; tachycardia is earliest sign.
- **Head injury** – MC cause of trauma death; **paediatric GCS** (verbal: coos/babbles = 5); CT head criteria (**PECARN**).
- **Abdominal trauma** – spleen MC organ injured; **non-operative management** for stable splenic/liver injuries.
- **Chest** – **pulmonary contusion without rib fractures** (compliant chest wall).
- **SCIWORA** (spinal cord injury without radiographic abnormality) – children < 8 y; MRI.
- **Burns**: **Lund–Browder chart** (head larger in infants – 19% at birth vs 9% adult; legs smaller); **Parkland formula: 4 mL × weight (kg) × % TBSA (RL) – half in first 8 h** from time of burn + maintenance fluids in children < 30 kg (with dextrose); fluid resuscitation if **> 10–15% TBSA** in children.

Child abuse (non-accidental injury) – red flags

- Injury inconsistent with history/developmental stage, delay in seeking care, changing story.
- **Metaphyseal corner/bucket-handle fractures**, **posterior rib fractures**, multiple fractures of different ages, spiral femur fracture in non-ambulant infant, skull fractures (complex).
- **Abusive head trauma (shaken baby)**: **subdural haematoma + retinal haemorrhages (multilayered) + encephalopathy**.
- Patterned bruises, bruises in non-mobile infants ("**those who don't cruise rarely bruise**"), **TEN-4-FACESp** (Torso, Ear, Neck bruising < 4 y; any bruise < 4 months), immersion burns (glove/stocking), cigarette burns.
- Differentials: **osteogenesis imperfecta**, Menkes, glutaric aciduria type 1, bleeding disorders, Mongolian spots.
- India: **POCSO Act 2012** – mandatory reporting of child sexual abuse.

Practice MCQs · Pediatric Basic & Advanced Life Support (PALS)

60 questions · correct option highlighted in green · cover the answer strip to self-test

Basic Life Support (BLS)

1. An infant brought to emergency, unresponsive, bradycardia, shallow breathing. Next step of management:

A. IV epinephrine
B. IV atropine
C. Stabilize the airway and assist breathing
D. **Cardiopulmonary resuscitation**

Ans D The sequence of resuscitation in infants & children is C-A-B (compressions, airway, breathing) rather than A-B-C i.e initiate CPR with chest compressions, rather than with rescue breaths

2. How are chest compressions given in a newborn?

A. Using palm on the lower third of sternum
B. Using two fingers on the middle third of sternum
C. **Using the two thumbs on the lower third of sternum**
D. Using three fingers on the lower third of sternum

Ans C The two thumb encircling technique is preferred for chest compressions in newborn

3. The MOST common pre-arrest rhythms in young children is

A. **bradyarrhythmia**
B. atrial flutter
C. ventricular fibrillation
D. supraventricular arrhythmia with WPW syndrome
E. atrioventricular block

Ans A

4. You were engaged in CPR for 3-year-old child found unresponsive in the ward. You were performing cycles of 15 chest compressions and 2 breathings alternatively with another medical personal. After 2 min of resuscitation you evaluate the pulse which shows PEA (pulseless electrical activity) status. The AED already is attached. Of the following, the correct NEXT action is to

A. give a shock of 0.5 joule/kg
B. give a shock of 1 joule/kg
C. ventricular fibrillation / pulseless VT
D. **give adrenaline 0.01mg/kg iv**
E. give adrenaline 0.1mg/kg iv

Ans D Pulseless electrical activity (PEA) is a clinical condition characterized by unresponsiveness and lack of palpable pulse in the presence of organized cardiac electrical activity. In this situation (and in asystole) adrenaline is indicated in cycles of 3-5 min, considering an advanced airways and reevaluation for the pulse status; if it became shockable give shock of 2 joules/kg. You need to look also for correctable causes (the 6H and 5T). If it became normal that goes for (ROSC) post-cardiac arrest care.

5. You were engaged in CPR for 3-year-old child found unresponsive in the ward. You were performing cycles of 15 chest compressions and 2 breathings alternatively with another medical personal. After 2 min of resuscitation the assistant told you that the pulse status is shockable. This means that the pulse is in status of

A. pulseless electrical activity PEA
B. asystole
C. **ventricular fibrillation / pulseless VT**

Ans C Shockable rhythms mean either VF or VT.

Advanced Life Support (ALS)

6. According to PALS 2010, all are components of initial impression of child EXCEPT:

A. Consciousness
B. Color
C. **Airway**
D. Breathing

Ans C PALS Systematic Approach Summary Initial Impression Your first quick (in a few seconds) "from the doorway" observation; It includes Consciousness Level of consciousness (e.g. unresponsive, irritable, alert) Breathing Increased work of breathing, decreased respiratory effort, or abnormal sounds heard Color Abnormal skin color, such as cyanosis, pallor, or mottling The purpose is to quickly identify a life-threatening problem.

7. In Pediatric advanced life support, intraosseous access is used for drug/fluid administration for pediatric age of?

A. < 1 year age
B. < 5 years age
C. < 6 years age
D. **Any age**

Ans D

8. In pediatrics advanced life support (PALS) curriculum, regarding the format of ABCDE, all the following are true EXCEPT

A. A refers to assessment of airways
B. B refers to assessment of breathing
C. C refers to assessment of circulation
D. **D refers to assessment of dehydration**
E. E refers to assessment of unidentified injuries

Ans D The D means Disability which refers to assessment of the child's neurologic function in terms of the level of consciousness and cortical function. It can be assessed quickly by checking the pupilary light respose and using Glasgow Coma scale, so the response D is only partially true.

9. The perfusion pressure of brain (cerebral perfusion pressure) equals to

A. intracranial pressure
B. systolic blood pressure minus intracranial pressure
C. mean blood pressure minus Intracranial pressure
D. mean blood pressure
E. mean blood pressure plus CSF pressure

Ans C The intracranial pressure (ICP) is derived from the volume of its components and the bony compliance. The perfusion pressure of the brain (cerebral perfusion pressure [CPP]) is equal to the pressure of blood entering the cranium (mean arterial pressure) minus the ICP.

10. An 11-year-old adolescent boy came to your clinic with a complaint of been collapsed while bathing with hot water after he felt dizzy. Father confirms that the boy was extremely pale when founded. He added that he toke few minutes to recover, also he had past history of similar condition one month ago when he was urinating. Of the following, the MOST likely explanation for this condition is

A. long QT syndrome
B. hypertrophic cardiomyopathy
C. neurocardiogenic syncope
D. drug abuse
E. seizure disorder

Ans C Typically, the patient with syncope will have been standing for a period of time, often on a hot day, or has gotten up suddenly from sleep or resting in a supine position. Occurrence in the shower is common, presumably caused by standing and vasodilation caused by hot water. For boys, the occurrence while urinating while standing is sometimes reported. The occurrence of syncope in girls while sitting or standing and having their hair brushed is common.

Airway Management

11. Torsades de pointes can be seen in:

A. Hyponatremia
B. Jervell and Lange-Nielsen syndrome
C. Hypomagnesemia
D. Hypokalemia
E. Hypocalcemia

Ans B c. Hypomagnesemia, d. Hypokalemia, e. Hypocalcemia It is a distinctive form of polymorphic ventricular tachycardia (VT) characterized by a gradual change in the amplitude and twisting of QRS complexes around the isoelectric line. Important Causes of Torsades De Pointes Cogenital long QT syndrome
• Romano-Ward syndrome
• Jervell and Lange-Nielsen syndrome Acquired disorders:
• Electrolyte disorders: Hypokalemia, Hypomagnesemia, Hypocalcemia
• Bradyarrhythmias
• Structural heart disease

12. The correct statement regarding the lower limit of systolic blood pressure for a neonate is

A. ≥40 mm Hg + (2 × age)
B. ≥50 mm Hg + (2 × age)
C. ≥60 mm Hg + (2 × age)
D. ≥70 mm Hg + (2 × age)
E. ≥80 mm Hg + (2 × age)

Ans C For a neonate ≥60 mm Hg, for an infant it is ≥70 mm Hg and for an older child it is ≥90 mm Hg, definitely the adult's value is inapplicable in children.

13. All the following are an absolute indications for endotracheal intubation EXCEPT

A. inability to protect the airway against aspiration
B. failing to maintain adequate oxygenation
C. complete airway obstruction
D. failing to control blood carbon dioxide levels
E. paralysis is required for a procedure

Ans C In settings of known complete airway obstruction, endotracheal intubation should be avoided, and emergency cricothyroidotomy performed instead.

14. Of the following, the mandatory test/study for all patients presenting for the first time with syncope is

A. electrocardiogram (ECG)
C. echocardiography
D. holter monitoring
E. complete blood count

Ans A All patients presenting with a first episode of syncope should have an electrocardiogram obtained, looking primarily for QT interval prolongation, preexcitation, ventricular hypertrophy, T-wave abnormalities, and conduction abnormalities. Other tests may be needed depending on the results of the initial evaluation.

15. All the following are controlled mode of mechanical ventilation EXCEPT

A. pressure support ventilation (PSV)
B. conventional mechanical ventilation (CMV)
C. intermittent mechanical ventilation (IMV)
D. synchronized Intermittent mechanical ventilation (SIMV)
E. synchronized Intermittent positive pressure ventilation (SIPPV)

Ans A Except the option A all other forms of mechanical ventilation variables are controlled either fully by the machine as in options B and C which need to paralyze the patient to get the synchrony or partially as in options D and E where there is patient trigger by an effort to respiration followed by the machine active work to do the preset ventilation values;

16. A 2-hour-old fullterm baby delivered to a mother with gestational diabetes by CS, he developed an increasing respiratory distress and grunting with blood gases values indicative of respiratory failure. Of the following, the BEST mode to be used in mechanical ventilation is

A. intermittent mechanical ventilation (IMV)
B. synchronized Intermittent mechanical ventilation (SIMV)
C. synchronized Intermittent positive pressure ventilation (SIPPV)
D. pressure-regulated volume control (PRVC)
E. pressure support ventilation (PSV)

Ans D IMV is not the choice because you need to paralyze the patient; options B and C both are good choice as they provide synchrony with the patient and can achieve the goal of ventilation by either controlling the pressure or volume values but in presence the option D in your ventilator, making it the best because it will reduces both baro and volutrauma.

17. In mechanical ventilation giving adequate PEEP can mainly reduces

A. volutrauma
B. barotrauma
C. **oxytrauma**
D. pneumonia
E. post-extubation airway obstruction

Ans C It can also reduce atelectotrauma.

18. All the following are recognized complications of mechanical ventilation which can result from use of high tidal volume and/or inspiratory pressure EXCEPT

A. volutrauma
B. barotrauma
C. oxytrauma
D. decrease surfactant production
E. **atelectotrauma**

Ans E It can result from low PEEP.

19. A 32-week-preterm baby on mechanical ventilation, he is recovering from RDS, his blood gas is satisfactory, and you reduced the ventilatory settings in process for weaning. Of the following, the MOST valuable indicator that the baby is ready for extubation is

A. frequency of respiration
B. fraction of inspired o2 0.55
C. peak expiratory end pressure
D. **inspiratory pressure**
E. inspiratory time 0.4 sec

Ans D In absence of tachypnea, increased work of breathing, hypoxemia, hypercapnia, acidosis, diaphoresis, tachycardia, and hypotension and when the ventilator rate is <5 breaths/min or (between 5-10 cm H2O in pressure controlled devices) indicate that the contribution of the ventilation to minute ventilation is minimal and the baby is ready to be extubated.

20. Prior to extubation and in order to reduce airway narrowing after extubation it is advised to use

A. **dexamethasone IV**
B. betamethasone IV
C. inhaled budesonide
D. nebulized budesonide
E. hydrocortisone IV

Ans A Administration of intravenous dexamethasone 0.5 mg/kg every 6 hr for 4 doses prior to extubation has been shown to minimize the incidence of postextubation airway obstruction. In patients in whom postextubation airway obstruction develops, the need for re-intubation may be obviated by administration of nebulized racemic epinephrine and heliox.

Cardiac Arrest & CPR

21. A 6-months-old child having severe dehydration comes to the casualty with weak pulse and unrecordable B.P. Repeated attempt in gaining IV access has failed. The next best step is:

A. Try again
B. Jugular Vein Catheterization
C. **Intraosseous IV fluids**
D. Venesection

Ans C

22. In pediatrics medical emergencies the survival rate can increase with good neurological outcome if rapid, effective, cardiopulmonary resuscitation (CPR) is done. The survival rate can reach up to

A. 10%
B. **30%**
C. 50%
D. 70%
E. 90%

Ans B

23. The features of high quality CPR include all the following EXCEPT

A. rate at least 100/min
B. **compression depth of 2/3 anterior-posterior diameter of the chest**
C. allow complete chest recoil after each compression
D. minimize interruptions in chest compressions
E. avoid excessive ventilation

Ans B The Depth of chest compression should be at least 1/3 anterior-posterior diameter of chest, about 1 1/2 inches (4 cm) in infants and 2 inches (5 cm) in children.

24. Ductus-dependent congenital heart lesions is a type of

A. distributive shock
B. **obstructive shock**
C. cardiogenic shock
D. hypovolemic shock
E. septic shock

Ans B It represent a decreased in cardiac output secondary to direct impediment to right- or left-heart outflow or restriction of all cardiac chambers.

25. You are treating a 6-year-old child who survived drowning after prolonged CPR, the mother is asking about the possibility of full neurological recovery of her child. Of the following, the BEST response is that it is possible if he regained full consciousness within

A. few hours
B. 12 hours
C. 24 hours
D. 48 hours
E. **72 hours**

Ans E Neurologic examination and progression during the 1st 24-72 hr are the best prognosticators of long-term CNS outcome. Children, who regain consciousness within 48-72 hr, even after prolonged resuscitation, are unlikely to have serious neurologic sequelae.

Shock & Resuscitation

26. An 8-month-old female child presented to emergency with a heart rate of 220/minute and features of congestive heart failure. Her heart rate comes down to normal after administering intravenous Adenosine. What is the most likely diagnosis?

A. Atrial fibrillation
B. Atrial flutter
C. **Paroxysmal supraventricular tachycardia**
D. Ventricular tachycardia Chapter 10: Pediatric Basic & Advanced Life Support (PALS) 219 Answers

Ans C M
• Most common cause of neonatal mortality in India are prematurity & low birth weight
• Neonatal infections, comprising pneumonia, neonatal sepsis and infections of CNS is the next most common cause of neonatal mortality Latest Updates Periviable birth refers to delivery occurring from 20 0/7 to 25 6/7 weeks, at the limit of viability (ACOG 2017) A neonate bourne stati on36 weeks gestation haQs a birth 1weight of 3.4 kg. What will be this neonate referred to as, based on the newborn birth weight chart provided?

27. The MOST common precipitating event for cardiac instability in infants and children is

A. electrolyte disturbances
B. trauma
C. **respiratory insufficiency**
D. poisoning
E. myocarditis

Ans C

28. All the following are recognized brain herniation due to increase intracranial pressure EXCEPT

A. cingulate
B. **cerebral tonsillar**
C. transcalvrial
D. uncal
E. upward cerebellar

Ans B In fact there are 6 types of brain herniation in addition to what have been mentioned in the question plus cerebellar tonsils and cerebral herniation through foramen magnum.

29. You are treating an 8-month-old baby in intensive care unit with cold shock. In the first 15 min he received adequate fluid therapy and a colleague had started dopamine for him. The baby condition is still unresponsive. Of the following, the BEST option now is to start

A. **epinephrine**
B. hydrocortisone
C. norepinephrine
D. terlipressin
E. angiotensin

Ans A After 15 min of treating shock, inotropes like dopamin need to be started, if the shock still unresponsive epinephrine for cold and norepinephrine for warm shock should be titrated. Hydrocortisone is considered after 60 min (Catecholamine resistant shock). Other options are used in warm shock and usually after other options are exhausted.

30. Septic shock usually shows a combination of hypovolemic and distributive shock in addition to

A. obstructive shock
B. **cardiogenic shock**
C. acute respiratory distress syndrome
D. disseminated intravascular coagulation
E. decreased steroid synthesis

Ans B Options C and D may be part of pathophysiology of septic shock, it is not a type.

31. All the following are component of cold water shock resulted from immersion in cold water EXCEPT

A. **hypoventilation**
B. decrease breath holding ability
C. hypertension
D. ectopics

Ans A Immersion in cold water has immediate respiratory and cardiovascular effects. Victims experience cold water shock, a dynamic series of cardiorespiratory physiologic responses that can cause drowning. In adults, immersion in icy water results in intense involuntary reflex hyperventilation and to a decrease in breath-holding ability to <10 sec, which leads to fluid aspiration.

Vascular Access

32. In a child with cardiopulmonary arrest, intraosseous access should be attempted, if venous access is not available within:

A. **1 minute**
B. 2 minutes
C. 3 minutes
D. 5 minutes

Ans A If venous access is not available within 1 minute in a child with cardiopulmonary arrest, an IO needle should be placed in anterior proximal tibia

33. Intraosseous line is placed in which bone in children?

A. Femur
B. **Tibia**
C. ASIS
D. Iliac crest

Ans B

34. All the following are signs of increased intracranial pressure (ICP) and impending brain herniation EXCEPT

A. pupillary dilation
B. **4th cranial nerve palsy**
C. systemic hypertension
D. bradycardia
E. extensor posturing

Ans B The development of increased ICP with impending herniation may be heralded by new onset or worsening headache, depressed level of consciousness, vital sign changes (hypertension, bradycardia, irregular respirations), and signs of 6th (lateral rectus palsy) or 3rd (anisocoria [dilated pupil], ptosis, down-and-out position of globe as a result of rectus muscle palsies) cranial nerve compression.

35. The percentage of Fio2 that can be delivered via the nasal cannula is up to

A. 30%
B. **40%**
C. 50%
D. 60%
E. 80%

Ans B The typical Fio2 value using this method is between 23% and 40%, and can be measured with following formula Fio2 %O2 delivered = 21 %+(nasal cannula flow (L/min) X3), and the flow rate should be <5 L/min.

Arrhythmias & Defibrillation

36. What is the drug of choice to control supraventricular tachycardia?

A. **Adenosine**
B. Propranolol
C. Verapamil
D. Digoxin

Ans A In stable patients with Supraventricular Tachycardia, Adenosine by rapid IV push is the treatment of choice because of its rapid onset of action and minimal effects on cardiac contractility.

37. Regarding Glasgow Coma scale in pediatrics, all the following are true EXCEPT

A. in modified type it uses 15 score points
B. it has 3 components
C. **valid as a prognostic scoring system**
D. score ≤8 require aggressive management
E. verbal response component has 5 possible points

Ans C Although the GCS has not been validated as a prognostic scoring system for infants and young children as it has been in adults, it is commonly used in the assessment of pediatric patients with an altered level of consciousness. The GCS is the most widely used method of evaluating a child's neurologic function and has 3 components. Individual scores for eye opening, verbal response, and motor response are added together, with a maximum of 15 points.

38. All the following factors are attributing in definition of clinically significant bradycardia EXCEPT

A. heart rate
B. **temperature**
C. perfusion
D. mental status
E. blood pressure

Ans B A clinically significant bradycardia occurs when the heart rate is slow and there are signs of systemic hypoperfusion (i.e., pallor, altered mental status, hypotension, and acidosis). Symptomatic bradycardia occurs most often in the setting of hypoxia but can also be caused by hypoglycemia, hypocalcemia, other electrolyte abnormalities, and intracranial hypertension.

39. You are evaluating a 3-year-old conscious child with bradycardia and poor perfusion in the emergency department. Of the following, the MOST important first step in treatment is

A. maintaining adequate perfusion by chest compression
B. **maintaining the airways and assisted breathing**
C. treating precipitating factors as hypoglycemia
D. epinephrine therapy
E. atropine therapy

Ans B Alleviating respiratory compromise and maintaining adequate breathing and oxygenation may correct clinically significant bradycardia. Start monitoring and obtain IV access. If bradycardia persist it is the time for chest compressions and if continues you need to start drug therapy.

40. Atropine is used in the treatment of bradyarrhythmia that is associated with

A. **raised intracranial pressure**
B. hypoxia
C. hypothermia
D. tension pneumothorax
E. hyperkalemia

Ans A Atropine is indicated in the case of increased vagal tone (e.g., in the setting of head injury with raised intracranial pressure) or primary atrioventricular block.

41. Factors in favour of SVT versus sinus tachycardia in pediatrics include the following

A. abrupt history
B. polymorphic P wave
C. absence of P wave
D. **poor perfusion**
E. constant RR

Ans D In sinus tachycardia, the history and onset are consistent with a known cause of tachycardia, such as fever or dehydration and P waves are consistently present, are of normal morphology, and occur at a rate that varies somewhat. In SVT, onset is often abrupt without prodrome and P waves are absent or polymorphic, and when present, their rate is often fairly steady at or above 220 beats/min. Poor perfusion can present in either causes as in dehydration with tachycardia and in late stage of SVT.

Post-Resuscitation Care

42. The MOST common reason for a sick child visit is

A. **fever**
B. altered mental status
C. vomiting
D. respiratory distress
E. abdominal pain

Ans A is the most common reason for a sick child visit. Most fevers are the result of self-limited viral infections. However, pediatricians need to be aware of the age- dependent potential for serious bacterial infections (e.g., urinary tract infections, sepsis, meningitis, pneumonia, dysentery, osteoarticular infection).

43. In secondary assessment of critically ill child; all the following are true EXCEPT

A. interviewing a wittiness by medical personnel that involved in resuscitation
B. taking focused history
C. performing a focused medical examination
D. enquire about allergies and medications
E. enquire about timing of the last meal

Ans A The components of a secondary assessment include a focused history and focused physical exam. The history should be targeted to information that could explain cardiorespiratory or neurologic dysfunction and should take the form of a SAMPLE history (Signs/symptoms, Allergies, Medications, past medical history, timing of Last meal, and Events leading to this situation). Medical personnel not engaged in resuscitative efforts can be dispatched to elicit history from witnesses or relatives.

44. All the following are regarded as first tier therapy of increased intracranial pressure

A. intubation
B. controlled mechanical ventilation
C. head of bed elevation
D. sedation and analgesia
E. barbiturate infusion

Ans E The first tier therapy also include neuromuscular blockade, CSF drainage, and the use of osmolar agents as hypertonic saline (3%) and mannitol. 2nd tier therapy include barbiturate infusion, decompressive craniectomy, mild hypothermia hyperventilation and lumbar CSF drainage.

45. The percentage of Fio2 that can be delivered via a mask is up to

A. 35%
B. 45%
C. 65%
D. 75%
E. 85%

Ans C Oxygen flow rates vary from 5-10 L/min, yielding typical Fio2 values between 0.30 and 0.65.

Drowning & Near-Drowning

46. The first response to unwitnessed unresponsive infant is to

A. activate emergency response system
B. check pulse
C. start rescue breathing
D. start chest compression
E. do endotracheal intubation

Ans A A child found unresponsive from an unwitnessedcollapse should be approached with a gentle touch and the verbal question, "Are you OK?" If there is no response, the caregiver should immediately shout for help and send someone to both activate the emergency response system (EMS) and locate an automated external defibrillator (AED).

47. All the following are part of tertiary assessment of critically ill child EXCEPT

A. coagulation profile
B. renal function
C. echocardiography
D. arterial and venous catheters
E. lumbar puncture

Ans E The tertiary assessment occurs in a hospital setting, where ancillary laboratory and radiographic assessments contribute to a thorough understanding of the child's condition. A basic blood chemistry profile, complete blood count, liver function tests, coagulation studies, and arterial blood gas analyses give fairly broad (but somewhat nonspecific) estimates of renal function, acid–base balance, cardiorespiratory function, and presence or absence of shock.

48. All the following are important to diagnose brain death status in a child EXCEPT

A. deep coma of unknown cause
B. apnea
C. absence of brainstem reflexes
D. absence of motor response
E. absence of confounding factors

Ans A

49. Rapid and deep breathing without other signs of respiratory distress may be caused by the following EXCEPT

A. diabetic ketoacidosis
B. renal tubular acidosis
C. encephalitis
D. CNS stimulants
E. heart failure

Ans E Causes rapid shallow breathing.

50. In surviving drowning patients; expecting brain edema can occur within

A. 5 min
B. 15 min
C. 30 min
D. 60 min
E. several hours

Ans E after cardiopulmonary arrest, cerebral edema may occur, although the mechanism is not entirely clear.

51. Of the following, the MOST common factor that increases the likelihood of drowning in adolescent (USA) is

A. epilepsy
B. long QT syndrome
C. alcohol
D. myocarditis
E. external trauma

Ans C The use of alcohol and drugs greatly increases the risk of drowning in teenagers and adults who die, 30-40% have positive blood alcohol level.

52. All the following mechanisms are contributing in pathophysiology of drowning

A. laryngospasm
B. surfactant washout
C. anoxic injury
D. pulmonary aspiration
E. **circulatory overload**

Ans E

Poisoning & Toxicology

53. After activation of emergency medical system, the next action to unresponsive child is to

A. asses the air ways
B. **check pulse**
C. give rescue breathings
D. start chest compression
E. attach automated external defibrillator (AED) to find shakable rhythm

Ans B After activation of EMS the next step is to check for pulse and find definite pulse within 10 seconds.

54. A 2-year-old toddler was playing with candies and sweets in playroom, he developed an attack of wheezing and shortness of breath, the parents brought the child to emergency room, RR was 68/min with subcostal retractions, and O2 saturation was 92%. Of the following, the MOST proper next action is to

A. **arrange for urgent bronchoscopy**
B. arrange for bedside CXR
C. arrange for CT chest
D. encourage coughing
E. reassure the parents

Ans A A history consistent with foreign-body aspiration is considered diagnostic. Any child in the proper setting with the sudden onset of choking, stridor, or wheezing has foreign body aspiration until proven otherwise. The choice A is the best answer as the child start to show signs of respiratory distress with borderline O2 sat. The choice B is the next appropriate action while encourage coughing may be appropriate at home when no respiratory distress is there.

55. All the following are red flags in evaluating a patient with syncope EXCEPT

A. syncope with exercise
B. family history of syncope
C. **presyncopal feeling of light headedness**
D. history of Kawasaki disease
E. injury with syncope

Ans C Typical prodromal symptoms prior to syncope include light headedness, dizziness, and nausea, sweating, and feeling hot or cold. Patients may report visual field changes and rushing in their ears.

56. You are ventilating a fullterm baby with meconium aspiration syndrome, he was fine on the machine, suddenly the monitor shows severe drop in O2 saturation. You checked the ventilator; it was working well with its connections. Of the following, the MOST likely cause is

A. O2 source supply
B. **endotracheal tube blockade**
C. self extubation
D. low inspiratory pressure value
E. low PEEP value

Ans B All the given options are true causes for sudden drop of O2. The possible causes of desaturating baby on ventilator are; Oxygen supply, machine and it is connections, settings, tube block, self extubation and the patient himself. In the given scenario, the most likely cause is B because of meconium aspiration. The best action to such scenario is to disconnect the baby from the machine and use Ambu bagging gently to check the air entry and rise of O2 saturation. If no rise you need to change the tube.

Trauma & Injury

57. A 12-month-old infant found unresponsive in kindergarten, you were there as health visitor, you asked for activation of EMS and to bring a nearby automated external defibrillator (AED) machine, you check the pulse; it was 60/min. Your next response is to

A. open the air ways
B. **give one breath every 3 seconds**
C. begin cycles of 30 compressions with 2 breaths
D. begin a cycles of 15 compressions with 2 breaths
E. attach and use AED

Ans B In summary, the plan of action for an unwitnessed infant found unresponsive as follows * Activate EMS and ask for AED machine. * Check for definite pulse, if found give one breath every 3 seconds. * Recheck pulse if it is < 60/min add chest compressions. * In case of no pulse found start directly chest compressions, for one rescuer give 30 compressions with 2 breaths and for 2 rescuers give 15 compressions and 2 breaths, reassess every 2 minutes.

58. You received a call from your neighbor, he had a 12-month-old baby boy who suddenly chock with a candy. When you arrive you find the baby unconscious. Of the following, the NEXT proper action is to

A. **open airway and give rescue breathing**
B. give 5 back blows
C. give 5 chest thrusts
D. a series of 5 back blows and chest thrusts
E. perform endotracheal intubation

Ans A In unconscious child, the child should be gently placed on the ground, supine, and then the provider should open the airway with the head-tilt/chin-lift maneuver and attempt mouth-to-mouth ventilation. If ventilation is unsuccessful, the airway is repositioned, and ventilation attempted again. If there is still no chest rise, attempts to remove a foreign body are indicated.

59. **A 6-year-old male child brought to emergency department after been hit by a car while riding his bike. GCS was 6 and brain CT was normal. However, immediate MRI demonstrates multiple areas of punctate hemorrhages consistent with diffuse axonal injury. Regarding the classification of traumatic brain injury (TBI), the child in this scenario has**

A. mild TBI
B. moderate TBI
C. **severe TBI**
D. pure axonal brain injury
E. parenchymal brain hemorrhage

Ans C The key for severity of TBI is GCS score, patient with GCS of 3-8 regarded as severe type while those between 9-12 regarded as moderate.

60. **The MOST effective strategy to reduce ventilator-associated pneumonia (VAP) is**

A. **reducing the period of ventilation**
B. elevation of the head of the bed to 30 degrees
C. use of a protocol for oral decontamination
D. liberal use of effective antibiotics
E. frequent endotracheal tube suction

Ans A The options B and C are a true technique, but the most effective strategy to minimize VAP is regular assessment of extubation readiness and liberation from mechanical ventilation as soon as clinically possible.

11

CHAPTER 11

Normal Neonate and Neonatal Resuscitation



WHAT THIS CHAPTER COVERS

- Neonatal Assessment & Examination
- Neonatal Reflexes
- Neonatal Physiology
- Neonatal Resuscitation (NRP)
- Apgar Score
- Neonatal Sepsis
- Neonatal Temperature Regulation
- Neonatal Feeding
- Common Neonatal Conditions

70 MCQs **9** subtopics

11.1 Definitions

Term	Definition
Term	**37⁰–41⁶ weeks** (early term 37–38⁶, full term 39–40⁶, late term 41–41⁶)
Preterm	**< 37 completed weeks** (late preterm 34–36⁶, moderate 32–33⁶, very preterm 28–31⁶, **extremely preterm < 28 wk**)
Post-term	**≥ 42 weeks**
Low birth weight (LBW)	**< 2500 g** (regardless of gestation)
Very LBW (VLBW)	**< 1500 g**
Extremely LBW (ELBW)	**< 1000 g**
SGA / AGA / LGA	Birth weight **< 10th / 10th–90th / > 90th centile** for gestational age
Neonatal period	First **28 days**; early neonatal = first 7 days
Perinatal period	**22 completed weeks of gestation to 7 days after birth** (WHO; India: 28 wk)
Neonatal mortality rate	Neonatal deaths per **1000 live births** (India SRS 2021: **19**; IMR 27)
Perinatal mortality rate	(Stillbirths + early neonatal deaths) / 1000 **total births**

- **India**: ~**1/3 of babies are LBW (mostly IUGR/SGA)** – in developed countries LBW is mainly due to prematurity.
- **MCC neonatal death (India)**: **prematurity/LBW complications** > birth asphyxia > neonatal sepsis > congenital anomalies.
- **Kangaroo mother care (KMC)**: continuous **skin-to-skin contact + exclusive breastfeeding + early discharge**; for stable LBW babies (WHO 2022 – immediate KMC for all < 2 kg).

11.2 Assessment of Gestational Age

- **New Ballard score** (most used): **6 neuromuscular** (posture, square window, arm recoil, popliteal angle, scarf sign, heel-to-ear) + **6 physical** (skin, lanugo, plantar creases, breast, eye/ear, genitals); accurate ± 2 weeks; extended to 20 weeks; best done **within 12–20 h of birth**.

Feature	Preterm	Term
Skin	Thin, gelatinous, red, visible veins	Thick, pale, desquamation, few veins
Lanugo	Abundant (peaks 28 wk)	Thinning/bald areas
Plantar creases	Only anterior transverse crease	**Creases over entire sole**
Breast nodule	< 5 mm (barely palpable)	**≥ 5–10 mm**
Ear	Flat, soft, slow recoil	Formed, firm, instant recoil
Male genitalia	Testes undescended, few rugae	**Testes descended, rugae well formed**
Female genitalia	**Prominent clitoris & labia minora**	**Labia majora cover minora & clitoris**
Posture	Extended (hypotonic)	**Fully flexed**
Scarf sign	Elbow crosses midline	Elbow doesn't reach midline

11.3 Normal Newborn Examination – Physiological Findings

Finding	Notes
Vernix caseosa	White cheesy coating (protective, thermal insulation) – **do not wipe off vigorously**; absent in post-term
Lanugo	Fine hair, preterm
Milia	**Tiny white papules on nose/chin** (retained sebum) – disappear spontaneously
Epstein pearls	Milia on **palate** (midline); **Bohn nodules** on gums
Erythema toxicum neonatorum	**MC rash**, day 2–3, erythematous macules with central papule/pustule; **eosinophils** on smear; benign, self-limiting
Transient neonatal pustular melanosis	Dark-skinned babies; pustules at birth → pigmented macules; **neutrophils**
Mongolian spots (dermal melanocytosis)	Bluish-grey patches over **sacrum/buttocks**; fade by 5–6 y; don't confuse with bruises
Stork bite (naevus simplex / salmon patch)	Nape, eyelids, glabella – fades (nape may persist)
Harlequin colour change	One half red, other pale – lying on side; autonomic immaturity
Cutis marmorata	Lacy mottling with cold – persistent → **Down, trisomy 18, hypothyroidism**
Acrocyanosis	Blue hands & feet in first 24–48 h – normal (central cyanosis is not)
Caput succedaneum	Oedema of scalp, **crosses suture lines**, present at birth, resolves in days
Cephalhaematoma	Subperiosteal bleed, **does NOT cross sutures** (usually parietal), appears hours after birth, resolves in weeks–months; jaundice, calcification
Subgaleal haemorrhage	Under aponeurosis, **crosses sutures, fluctuant, can cause shock** (vacuum delivery) – emergency
Physiological breast enlargement / witch's milk	Maternal oestrogen; don't squeeze (mastitis)
Pseudomenstruation	Vaginal bleeding/white discharge in girls in first week – oestrogen withdrawal
Physiological jaundice	Appears after 24 h – see Neonatal Jaundice
Physiological weight loss	≤ 10% in first week; regains by 10–14 days
Physiological phimosis, hydrocele	Normal
Umbilical cord	**2 arteries + 1 vein**; single umbilical artery → renal/cardiac anomalies; **cord falls off by 5–15 days**; delayed (> 3 weeks) → **LAD-1**, infection
Sneezing, hiccups, irregular breathing, startle	Normal

- **First meconium within 24 h** (99% of term babies by 48 h); delay → **Hirschsprung, meconium ileus (CF)**, hypothyroidism, anorectal malformation. **First urine within 24 h** (delay > 48 h → posterior urethral valves, renal agenesis).
- **Transitional stools** day 3–4 (greenish-yellow); breastfed babies – frequent golden stools, or once in several days.
- **Red reflex** (cataract, retinoblastoma), **hip examination (Ortolani, Barlow)**, femoral pulses (CoA), palate, spine, anus, SpO_2 screen for **CCHD** (**pre- & post-ductal SpO_2 after 24 h: < 90% fail; 90–94% or > 3% difference → repeat**).

11.4 Neonatal Reflexes (summary)

- Moro, rooting, sucking, palmar/plantar grasp, stepping, Galant, ATNR – see Development chapter. **Sucking & swallowing coordination at 32–34 weeks** → oral feeding possible.
- **Moro asymmetry → brachial plexus injury (Erb's) or clavicle fracture (MC fracture at birth)**.

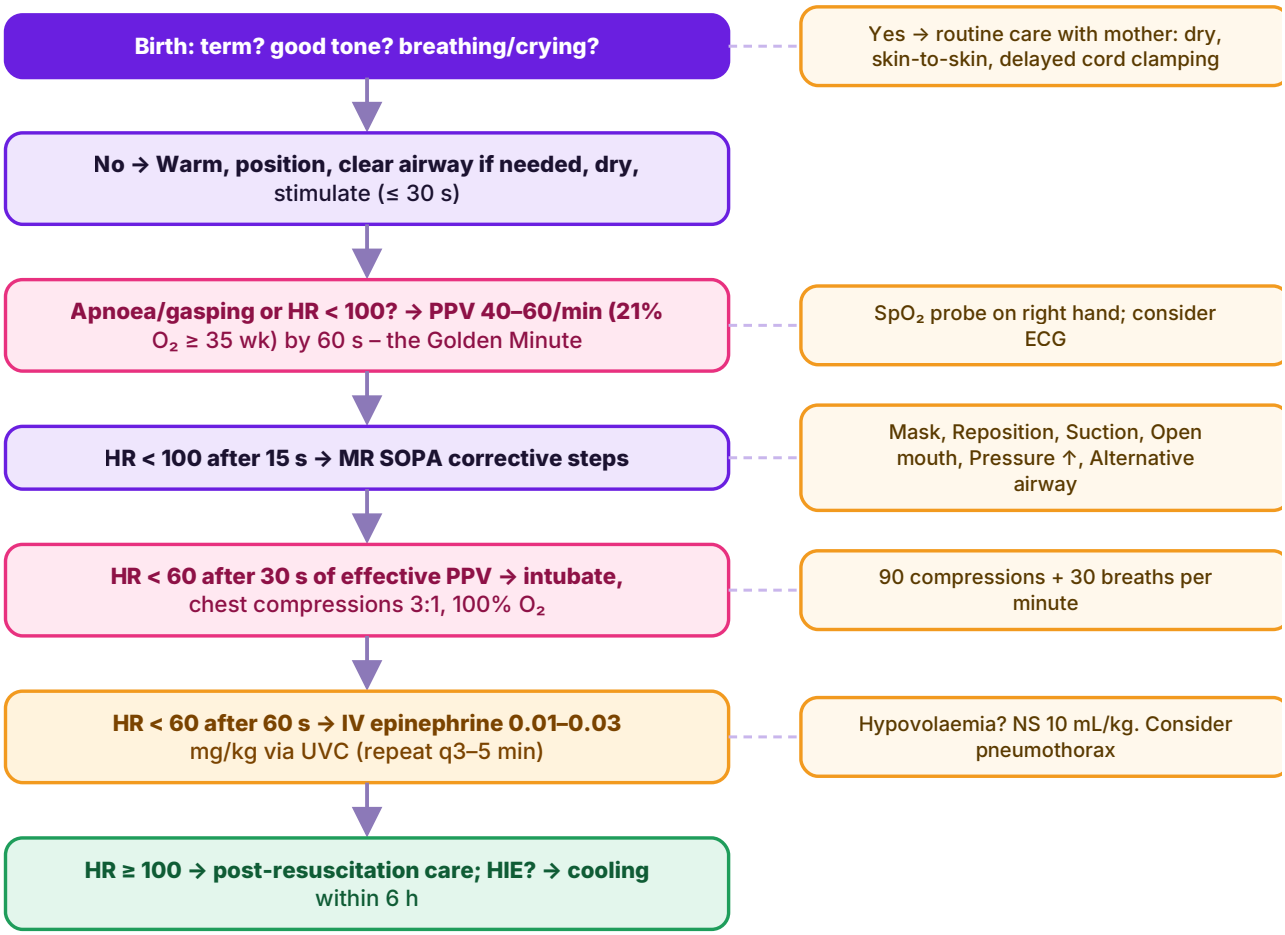
Brachial plexus palsy	Roots	Posture / features
Erb–Duchenne (MC)	**C5–C6** (± C7)	**"Waiter's tip"** – adduction & internal rotation of arm, extended elbow, pronated forearm, flexed wrist; **Moro absent on affected side, grasp present**
Klumpke	**C8–T1**	**Claw hand**, absent grasp; **Horner syndrome** (T1 sympathetic)
Total plexus	C5–T1	Flail arm

- Management: **physiotherapy, avoid immobilisation beyond 1–2 weeks; most recover in 3–6 months**; surgery (nerve grafting) if no biceps recovery by 3–6 months.

11.5 Neonatal Physiology – Key Points

- **Fetal Hb (HbF – $\alpha_2\gamma_2$)**: 70–80% at birth; higher O_2 affinity (↓ binding of 2,3-BPG); replaced by HbA by 6–12 months.
- Normal cord Hb **16.5–17 g/dL** (anaemia if < 13.5 at term); **physiological anaemia of infancy – nadir at 8–12 weeks (Hb ~9.5–11) in term**, **4–8 weeks (7–9 g/dL) in preterm** (↓ erythropoietin).
- Total blood volume 85 mL/kg (term), 90–100 mL/kg (preterm).
- **Delayed cord clamping (≥ 30–60 s; up to 3 min)** for vigorous term & preterm babies → ↑ iron stores, ↓ IVH/NEC in preterm; **cord milking** avoided < 28 wk.
- Kidney: low GFR, ↓ concentrating ability (max ~600–800 mOsm/L); **nephrogenesis complete at 34–36 weeks**.
- Liver: low UGT activity → physiological jaundice; low vitamin K-dependent factors.
- Thermoregulation: **brown fat (non-shivering thermogenesis)** – interscapular, perirenal, axillae; neonates can't shiver.
- Surfactant: **type II pneumocytes from 20–24 wk**, mature by **35 weeks**; **L/S ratio ≥ 2 = lung maturity** (**phosphatidylglycerol** most reliable in IDM); **antenatal corticosteroids** (betamethasone 12 mg IM × 2 doses 24 h apart / dexamethasone 6 mg × 4 doses 12-hourly) between **24–34 weeks** → ↓ RDS, IVH, NEC, death.

11.6 Neonatal Resuscitation (NRP 2020 / NNF)



Neonatal resuscitation algorithm – the Golden Minute

Initial questions at birth: **Term? Good tone? Breathing or crying?** → If yes – **routine care with mother** (skin-to-skin, dry, warm, delayed cord clamping, breastfeeding within 1 h).

If no → Initial steps (within 30 s): **warm (radiant warmer), position (neutral "sniffing", shoulder roll), clear airway if needed, dry, stimulate** (flick soles/rub back), reposition.

Situation	Action
Apnoea/gasping or HR < 100	**PPV with bag & mask at 40–60 breaths/min within the GOLDEN MINUTE** (by 60 s of birth); **SpO_2 monitor (right hand – pre-ductal)**, consider ECG
Laboured breathing/persistent cyanosis	Position, clear airway, SpO_2 monitor, supplemental O_2/CPAP
HR not rising	**MR. SOPA** ventilation corrective steps – **M**ask adjust, **R**eposition airway, **S**uction mouth & nose, **O**pen mouth, **P**ressure increase, **A**lternative airway (ETT/LMA)
HR < 60 after 30 s of effective PPV (that moves the chest)	**Intubate (if not already) + chest compressions** with **100% O_2**; **3 compressions : 1 breath (90 compressions + 30 breaths/min = 120 events)**, two-thumb technique, lower third of sternum, depth 1/3 AP
HR < 60 after 60 s of compressions + ventilation	**Epinephrine IV (UVC) 0.01–0.03 mg/kg (0.1–0.3 mL/kg of 1:10,000)**; ET 0.05–0.1 mg/kg while access obtained; repeat every 3–5 min; **volume (NS 10 mL/kg) if hypovolaemia suspected**

OXYGEN DURING NEONATAL RESUSCITATION

Start PPV with **21% O_2 (room air) in babies ≥ 35 weeks**, **21–30% in < 35 weeks**; titrate to **pre-ductal SpO_2 targets**: **1 min 60–65%, 2 min 65–70%, 3 min 70–75%, 4 min 75–80%, 5 min 80–85%, 10 min 85–95%**. **100% O_2 when chest compressions begin.** PPV pressure: **20–25 cm H_2O** (up to 30–40 in first breaths), PEEP 5 cm H_2O. **HR is the most important indicator** of effective resuscitation (auscultation; ECG most accurate).

MECONIUM-STAINED LIQUOR – CURRENT RULE

Routine intrapartum oro/nasopharyngeal suction and routine endotracheal suction of non-vigorous babies are NOT recommended (NRP 2015/2020). **Non-vigorous baby → initial steps + PPV within 1 minute**; intubation/suction only if airway obstructed. Vigorous baby → routine care.

- **Stop resuscitation**: no detectable HR after **all steps for ~20 minutes** (discuss with team/family).
- Do not initiate: < 22–23 wk / < 400 g, anencephaly, trisomy 13/18 (individualise).
- **Naloxone not recommended** in the delivery room for respiratory depression.
- **Sodium bicarbonate** not routinely used.
- **Therapeutic hypothermia** for moderate–severe HIE (≥ 36 wk) within **6 h**.
- **LMA** can be used ≥ 34 weeks / > 2 kg when mask & intubation fail.

Apgar score

Sign	0	1	2
Appearance (colour)	Blue/pale all over	Body pink, **extremities blue (acrocyanosis)**	Completely pink
Pulse (HR)	Absent	**< 100**	**≥ 100**
Grimace (reflex irritability)	No response	Grimace	Cry, cough, sneeze
Activity (tone)	Limp	Some flexion	Active motion
Respiration	Absent	**Slow, irregular, weak cry**	Good, strong cry

- Assessed at **1 and 5 minutes** (every 5 min up to 20 min if < 7 at 5 min). **7–10 normal; 4–6 moderately depressed; 0–3 severely depressed.**
- **Apgar is NOT used to decide when to start resuscitation** (resuscitation starts before 1-min score).
- Most important component: **heart rate**; first to disappear in asphyxia: **colour**; last – **tone/HR**.
- 1-min Apgar – need for resuscitation; **5-min Apgar – better predictor of outcome** (neurological outcome poorly predicted by Apgar alone).

11.7 Temperature Regulation

Category (WHO)	Axillary temperature
Normal	**36.5–37.5 °C**
Cold stress (mild hypothermia)	**36.0–36.4 °C**
Moderate hypothermia	**32.0–35.9 °C**
Severe hypothermia	**< 32 °C**
Hyperthermia	> 37.5 °C

- **Mechanisms of heat loss**: **evaporation (MC immediately after birth – wet baby)**, **radiation (MC in incubator/ warm environment)**, convection, conduction.
- **Warm chain (WHO 10 steps)**: warm delivery room (**25–28 °C**), **immediate drying**, skin-to-skin, breastfeeding, **bathing postponed (≥ 24 h; at least 6 h)**, appropriate clothing/bedding, mother & baby together, warm transport, warm resuscitation, training & awareness.
- Preterm < 32 wk: **polyethylene wrap/bag without drying**, cap, thermal mattress.

- **Neutral thermal environment**: temperature at which O_2 consumption is minimal (~**32–35 °C** for naked term/preterm in incubator).
- Hypothermia effects: hypoglycaemia, metabolic acidosis, ↑ O_2 consumption, apnoea, **sclerema**, PPHN, coagulopathy, **cold injury (red cheeks – deceptively healthy)**.
- **Rewarming**: moderate – skin-to-skin/radiant warmer; severe – rapid rewarming under radiant warmer/incubator (air temp 35–36 °C), treat hypoglycaemia & sepsis.
- **Hyperthermia**: dehydration fever (day 2–3, breastfed), overheating, sepsis.

11.8 Feeding the Newborn

- **Breastfeeding within 1 hour** for all stable babies.
- **Gestation-based feeding**:

Gestation / weight	Mode
≥ 34 weeks (> 1800 g)	**Direct breastfeeding** (coordinated suck-swallow-breathe)
30/32–34 weeks (1200–1800 g)	**Paladai / spoon / cup** feeding of expressed breast milk (EBM)
< 30–32 weeks (< 1200 g)	**Orogastric/nasogastric tube feeds** (+ IV fluids initially)
< 28 weeks / sick	**IV fluids ± TPN**, start trophic feeds (10–20 mL/kg/day) early

- Advance feeds by **20–30 mL/kg/day** in VLBW; full feeds 150–180 mL/kg/day.
- **Human milk fortifier** for < 1.5 kg / < 32 wk; **donor human milk** if mother's milk unavailable (preferred over formula – ↓ NEC).
- **Vitamin D 400 IU/day** from first week; **iron 2 mg/kg/day from 2 weeks (preterm)**, calcium-phosphorus for VLBW (metabolic bone disease).

11.9 Neonatal Sepsis (overview – details in Diseases of Newborn)

- Early onset (< 72 h) – maternal flora: **GBS, E. coli, Listeria**; late onset (> 72 h) – nosocomial/community: **Klebsiella (MC in India), Acinetobacter, S. aureus, CONS**.
- **Earliest feature: poor feeding/lethargy ("not doing well")**; hypothermia more common than fever.
- **Blood culture – gold standard**; sepsis screen (≥ 2 positive of: TLC < 5000, ANC low, **I:T ratio ≥ 0.2**, CRP > 1 mg/dL, micro-ESR > 15 mm 1st h).

11.10 Common Neonatal Conditions – Quick Table

Condition	Key facts
Oral thrush	White curdy plaques, don't scrape off; **nystatin/clotrimazole**; treat mother's nipples
Umbilical granuloma	Moist pink tissue after cord falls; **silver nitrate cautery / salt**
Omphalitis	Periumbilical redness; S. aureus; risk of portal vein thrombosis
Umbilical hernia	MC in **African, LBW, hypothyroid, Down** babies; **most close spontaneously by 4–5 y**; surgery if > 5 y or > 1.5–2 cm defect after 2 y, strangulation (rare)
Patent urachus	Urine leak from umbilicus
Persistent vitello-intestinal duct	Faecal/mucus discharge from umbilicus
Sticky eye / nasolacrimal duct obstruction	Watering from 2–4 weeks; **massage (Crigler) – 90% resolve by 1 y**; probing after 12 months
Ophthalmia neonatorum	See Eye chapter

Condition	Key facts
Neonatal mastitis	S. aureus; IV antibiotics
Physiological jaundice, dehydration fever, transitional stools	Normal phenomena
Positional deformities (talipes, plagiocephaly, torticollis)	Postural vs structural – check passive correction
Sternomastoid tumour (congenital muscular torticollis)	Firm mass in SCM at 2–4 weeks, head tilted to same side, chin to opposite side; **physiotherapy (stretching)** – > 90% resolve

Practice MCQs · Normal Neonate and Neonatal Resuscitation

70 questions · correct option highlighted in green · cover the answer strip to self-test

Neonatal Assessment & Examination

1. A full term baby, exclusively breast fed at the end of 1 week was passing golden yellow stools and was found to have adequate hydration with normal systemic examination. The weight of the baby was just same as it was at birth. The pediatrician should now advice:

A. Give oral solution with breastfeeding
B. Start top feeding
C. Investigate for lactic acidosis
D. **Reassure the mother that nothing is abnormal**

Ans D Term babies have physiological weight loss (upto 10% of their birth weight), that requires no treatment.

Neonatal Reflexes

2. Asymmetric tonic neck reflex (ATNR) disappears at:

A. **5–6 months**
B. 7–8 months
C. 9 months
D. 10 months

Ans A Asymmetric tonic neck reflex (ATNR) disappears at 5–6 months postnatal age & only then the baby learns to roll over;

3. Asymmetric Moro's reflex at birth is indicative of:

A. HIE
B. Brain damage
C. **Erb's palsy**
D. Kernicterus

Ans C

4. Grasp reflex is fully developed at:

A. 24 weeks
B. 28 weeks
C. **32 weeks**
D. 36 weeks

Ans C

5. Grasp reflex begins at:

A. 24 weeks
B. **28 weeks**
C. 32 weeks
D. 36 weeks

Ans B

6. Reflex which is not present in a child at birth is?

A. Moro's reflex
B. **Symmetric tonic neck reflex**
C. Crossed extensor reflex
D. Asymmetric tonic neck reflex

Ans B Important Neonatal reflexes appearing after birth are parachute reflex & symmetric Tonic Neck reflex.

7. Moro's reflex disappears at:

A. 5 months
B. 3 months
C. 7 months
D. **6 months NEONATAL RESUSCITATION**

Ans D 6 months This is a controversial question; Previously, Moro's reflex was believed to disappear at 3 months But as per latest Nelson's 20/e, Moro's reflex disappears at 5-6 months of life. Hence, 6 months is the best answer to this question, currently.

Neonatal Physiology

8. Color of transition stools:

A. **Green**
B. Brown
C. Yellow
D. Pale

Ans A Transitional stools are dark greenish yellow and loose, sometimes 'seedy' in texture. with

Neonatal Resuscitation (NRP)

9. A 32-week, 1400 g neonate is born to a primigravida. The baby did not require resuscitation and showed stable vitals. The baby was transferred to the NICU. How will you manage the feeding of the patient?

A. **Start total enteral feeding and IV nutrition not required**
B. Start IV nutrition with minimal enteral feeding
C. Start IV nutrition and introduce feeds on 2nd day of life
D. Start parenteral nutrition and institute oral feeding on 2nd day of life

Ans A Start total enteral feeding and IV feeding not required

10. Which of the following is false regarding neonatal resuscitation?

A. T-tube delivers free flow oxygen
B. **Self-inflating bag cannot be used without a reservoir**
C. Self-inflating bag can deliver free flow oxygen
D. Flow inflating bag delivers free flow oxygen

Ans B Self–inflating bag cannot be used without a reservoir Self-inflating bag can be used with/without a reservoir.

11. Correct order of suctioning during neonatal resuscitation is:

A. Trachea→nose→mouth
B. Nose→mouth
C. Mouth→nose→trachea
D. **Mouth→nose**

Ans D Mouth → nose If suction of mouth & oropharynx is not done prior to doing suction of nose, oropharyngeal secretions may get aspirated, when nasal suction of a neonate is done.

12. Resuscitation of a neonate with HR < 60/min includes all of the following except:

A. Chest compression
B. Inj Adrenaline
C. Endotracheal intubation
D. **None of the above**

Ans D Chest compression, Endotracheal intubation as well as Inj Adrenaline may be required in a neonate with HR <60/minute.

13. Which of the following is the best route of drug administration in a newborn baby during neonatal resuscitation?

A. Umbilical artery
B. **Umbilical vein**
C. Intraosseous
D. Intratracheal

Ans B • Umbilical venous catheter is the preferred method of obtaining emergency vascular access in delivery room;
• For treating hypovolemia, 0.9% NaCl (normal saline) or O Rh-negative blood is to be used

14. Dose of IV adrenaline in term infant during neonatal resuscitation:

A. 0.1–0.3mL/kg of 1:1000
B. 0.3–0.5 mL/kg of 1:1000
C. **0.1–0.3 mL/kg of 1:10,000**
D. 0.3–0.5mL/kg of 1:10,000

Ans C 0.1–0.3 mL/kg in 1:10,000 Epinephrine is preferably given by intravenous route at a dose of 0.01 to 0.03 mg/kg or 0.1-0.3 mL/kg of 1:10,000 concentration Endotracheal administration may be done if venous access not available, at a dose of 0.05 to 0.1 mg/kg

15. In newborn resuscitation, ratio of cardiac compressions to breaths is:

A. 15:1
B. 30:2
C. 15:2
D. **3:1**

Ans D

16. Components of neonatal resuscitation:

A. **Maintenance of temperature**
B. Maintenance of respiration
C. Maintenance of circulation
D. Chest compression
E. Feeding the baby

Ans A b. Maintenance of respiration, c. Maintenance of circulation, and d. Chest compression
• The most common indication for neonatal resuscitation is asphyxia followed by prematurity
• The main components of neonatal resuscitation are maintenance of temperature, respiration & circulation
• Chest compressions are required if heart rate is < 60/minute.Q

17. Bag and mask ventilation in newborn resuscitation is contraindicated in:

A. **Diaphragmatic hernia**
B. Pulmonary hypoplasia
C. Tracheo-oesophageal fistuala
D. Laryngomalacia

Ans A

Apgar Score

18. Normal Apgar Score with Acidosis in a neonate is seen in?

A. Spinal cord trauma
B. Choanal atresia
C. Diaphragmatic hernia
D. **High Fetal Catecholamine levels**

Ans D Important conditions in newborns with No acidosis but low apgar score Acidosis present with normal apgar score
• Prematurity; Acute cerebral trauma • Maternal acidosisQ
• Analgesics, narcotics, sedatives, Magnesium sulfate • High fetal catecholamine levelsQ
• Congenital myopathy or neuropathy or CNS anomaly
• Lung anomaly (CDH) or Airway obstruction (choanal atresia)

Neonatal Sepsis

19. How much time later should ROP screening be done for a 2-week-old baby being treated in NICU for sepsis, who was born at 28 wk gestation?

A. **2 weeks**
B. 4 weeks
C. 6 weeks
D. 8 weeks

Ans A Timing of the initial screening exam for ROP is based on the infant's age. dna According to AIIMS NICU protocol, 2014 Hardly any ROP is detected before 32 weeks of PMA. So, First screening examination should be carried out at 32 weeks of ost menstrual age (PMA) or 4 weeks of postnatal age, whichever is later In the given scenario, baby is born at 28 week gestation, so 1st screening examination should be done at 4 weeks postnatal age or 32 weeks post conceptional age;

Neonatal Temperature Regulation

20. Normal temperature of a newborn:

A. 34.5–35.5oC
B. 35.5–36.5oC
C. **36.5–37.5oC**
D. 37.5–38.5oC

Ans C 36.5–37.5°C Axillary temperature less than 36.5°C is called hypothermia in newborns.

Neonatal Feeding

21. In a stable 32 weeks neonate, the preferred mode of feeding is:

A. Direct breastfeeding
B. Nasogastric feed
C. **Katori spoon feed**
D. Parenteral feed

Ans C Katori spoon feed , AIIMS NICU Protocol

22. A neonate weighing 1500 gm is delivered at 33 wks. Which of the following would be most appropriate method of nutrition for the baby?

A. Breastfeeding
B. Orogastric feeding
C. Total parental nutrition
D. **Katori spoon feeding**

Ans D

Common Neonatal Conditions

23. Gynecomastia in neonate is seen due to:

A. Mother's estrogen
B. Mother's progesterone
C. GnRH
D. Gonadotropins

Ans A • Transient gynecomastia occurs in 60–90% of male newborns secondary to exposure to estrogens during pregnancy.
• Breast development may be asymmetrical and galactorrhea is seen in 5%.
• Most cases resolve within 4–8 wk of birth, but a few can last as long as 12 mo.

24. True about Cephalhematoma is:

A. Limited by sutures
B. Caused due to periosteal injury
C. Due to fluid accumulation below periosteum
D. Regresses in 24 hours
E. It is due to birth trauma

Ans A b. Caused due to periosteal injury; e. It is due to birth trauma Cephalohematoma is a subperiosteal hemorrhage, hence always limited to the surface of 1 cranial bone. It may take 2 wk to 3 months to disappear; e. Baby can have physiological weight loss upto 10% of birth weight About a False: In preterm baby elbow crosses xiphisternum (scarf sign) on flexion over chest, as shown in Ballard score below; About b False: In a term neonate, birth weight > 3.8 kg would be > 90th

25. True about term baby is:

A. Elbow crosses xiphisternum on flexion over chest
B. Weight > 3.8 kg is normal
C. Parachute reflex is not present after the age of 9 months
D. Jaundice is seen within first 24 hours of life
E. Baby can have physiological weight loss up to 10% of birth weight

Ans E Term neonates commonly show physiological weight loss of up to 10% of birth weight in the first week of life, regained by about 10-14 days.

26. Which of the following is true about Cephalhematoma?

A. Its size is maximum at birth
B. It resolves in 7-10 days
C. It is limited by periosteum
D. It is due to edema in the layers of scalp

Ans C

27. A hymenal tag in a newborn is best treated by:

A. Steroids
B. Surgery
C. Leaving it alone
D. None of the above

Ans C fo Hymenal tags around margins of hymen is a normal finding. weiveR 18. c. High larynx /e p 134-136 Differences between the airway of a young infant, child and adult include:
• Oral cavity is small in a newborn & totally filled by tongue due to slightly retracted lower jaw
• The soft palate & epiglottis are in contact at rest, providing an additional valve at the back of the oral cavity
• Infant larynx has a short funnel shaped lumen, that is disproportionately narrower than that of adult

28. Newborn babies are able to breathe and suck at the same time due to:

A. Wide short tongue
B. Short soft palate
C. High larynx
D. Short pharynx

Ans C The high position of the larynx in a newborn allows the epiglottis to interlock with the soft palate, letting the infant breathe and suck/swallow simultaneously.

29. The following skin changes are normal in a newborn except:

A. Infantile acropustulosis
B. Harlequin color change
C. Mongolian spots
D. Pustular melanosis

Ans A a. Infantile acropustulosis It is a recurrent, self-limited, pruritic, vesico-pustular eruption of palms & soles occurring in young children during first 2-3 years of life; Topical steroids & oral dapsone may be used to treat it. b. Harlequin color change It is a cutaneous condition seen in 10% of newborns, characterized by momentary red & pale colors of the two halves of body, sharply demarcated at the body's midline c.

30. About Caput Succedaneum all are true except:

A. Soft
B. Prolonged jaundice
C. Ill defined
D. None are true

Ans B Cephalhematoma predisposes to Neonatal Jaundice & not Caput succedaneum.

31. Early neonatal period is upto how many days of life?

A. 1 day
B. 7 days
C. 28 days
D. 14 days

Ans B

32. Normal HR in a newborn:

A. 80–100/min
B. 100–120/min
C. 120–160/min
D. 160–200/min

Ans C

33. Neonatal MCV is:

A. Glycosuria
B. Bacteriuria
C. WBCs in urine
D. Hyperbilirubinemia

Ans A 90–110 fL Normal Red Cell Indices in Infancy and Childhood Age MCV (fL) (mean) MCH (pg) (mean) MCHC (g/dL) (mean) Birth (cord blood) 108 34 33 1 month 104 34 33 1–2 years 78 27 33 2–6 years 81 27 34 6 – 12 years 77 29

34. Most common newborn rash which presents at 24–48 hours of life is:

A. Erythematous papular pustular lesions
B. Milia
C. Transient neonatal pustular melanosis
D. Hemangioma

Ans A In many neonates, small, white papules on an erythematous base develop 1-3 days after birth. This benign rash, erythema toxicum, persists for as long as 1 wk, contains eosinophils, and is usually distributed on the face, trunk, and extremities.

35. In a newborn, what is the normal respiratory rate?

A. 10–20 breaths/minutes
B. 30–40 breaths/minutes
C. 40–60 breaths/minutes
D. 60–80 breaths/minutes

Ans C 40–60 breaths/minutes Answers

36. Common sites for Mongolian spot are:

A. Face
B. Neck
C. Lumbosacral area
D. Leg
E. Thigh

Ans C

37. Skin changes in newborn that disappear spontaneously:

A. Harlequins skin change
B. Mongolian spots
C. Erythema toxicum
D. Lymphomia
E. Milia

Ans A b. Mongolian spots, c. Erythema toxicum and e. Milia

38. The appropriate approach to a neonate presenting with vaginal bleeding on day 4 of life is:

A. Administration of Vitamin K
B. Investigation for bleeding disorder
C. No specific therapy
D. Administration of 10 mL/kg of fresh frozen plasma over 4 hours

Ans C Vaginal bleeding is seen only in female neonates, usually seen after 3-5 days of birth; Occurs due to fall in the level of sex hormones after birth when baby is disconnected from the placenta.

39. Vomiting on the first day of baby's life may be caused by all of the following except:

A. Pyloric stenosis
B. Esophageal atresia
C. Aerophagy
D. Amniotic gastritis

Ans A Congenital hypertrophic pyloric stenosis usually presents with non-bilious projectile vomiting between 6-10 weeks age.

40. Normal finding in term neonate:

A. Erythema toxicum
B. Epstein's pearl
C. Bilateral cryptorchidism
D. Subconjunctival hemorrhages
E. Erythema nodosum PRETERM NEONATE

Ans A b. Epstein's pearl, and d. Subconjunctival hemorrhages • Subconjunctival & retinal hemorrhages are frequently seen in a neonate just after birth
• Probable cause: sudden increase in intrathoracic pressure during passage of the chest through the birth canal
• Three lesions resolve rapidly within the 1st 2 wk of life.

41. Which of the following is a component of kangaroo mother care?

A. Incubator
B. Heat therapy
C. Exclusive breastfeeding
D. Phototherapy

Ans C

42. Preterm VLBW infants ingesting unfortified breast milk need to be supplemented with what doses of calcium, and vitamin D?

A. Calcium (80–120 mg/kg/day) and vitamin D (200–400 IU/day)
B. Calcium (40–80 mg/kg/day) and vitamin D (400–1000 IU/day)
C. Calcium (500–1000 mg/kg/day) and vitamin D (200–400 IU/day)
D. Calcium (150–220 mg/kg/day) and vitamin D (200–400 IU/day)

Ans D As per the latest AAP Recommendations for Enteral Nutrition for VLBW Infants, a daily Calcium intake of 150–220 mg/kg, Phosphate 75–140 mg/kg & Vitamin D 200–400 IU should be ensured

43. As per the perinatal risk stratification of preterm neonates and developmental outcomes, which of the follow ing will constitute a moderate risk category?

A. Gestation 30–32 weeks, birth weight 1000–1500 gm, 1-3 days of hypoglycemia
B. Gestation < 28 weeks, birth weight 1251-1500 gm, 1–4 days of hypoglycemia
C. Gestation < 28 weeks, birth weight <1250 gm, 1-4 days of hypoglycemia
D. Gestation 30–32 weeks, birth weight 1251–1500 gm, more than 5 days of hypoglycemia

Ans A Gestation 30–32 weeks, birth weight 1000–1500 gm, 1–3 days of hypoglycemia NNF Recommendations for risk stratification of preterm neonates for follow-up: High Risk Moderate Risk Mild Risk
• Birth wt < 1 kg &/or gest <28 week • Birth weight 1000-1500 g
• Birth wt 1500 g - 2500 g
• Chronic lung disease / IVH / PVL • Gestational age < 33 weeks
• HIE grade I
• Severe HIE/ Seizures/ Meningitis • Twins/triplets • Transient hypoglycemia
• Surgical conditions, e.g. CDH, TEF • Moderate HIE • Suspected sepsis

44. Which one of the following is a strongly recommended intervention to reduce mortality & prevent future neuro-developmental impairment in preterm neonates:

A. IV IG administration
B. Use of Insulin for hyperglycemia
C. Avoiding aminoglycoside exposure for > 7 days
D. Use of antenatal steroid

Ans D Antenatal steroids reduce overall mortality, need for and duration of ventilator support and admission to a neonatal ICU, the incidence of severe IVH, necrotizing enterocolitis, and neurodevelopmental impairment in the neonate.

45. What is the recommended dose of steroids for attaining fetal lung maturity?

A. Inj Betamethasone 12 mg for 2 doses 12 hours apart
B. Inj Betamethasone 12 mg for 2 doses 24 hours apart
C. Inj Dexamethasone 6 mg for 4 doses 24 hours apart
D. Inj Dexamethasone 12 mg for 2 doses 12 hours apart

Ans B Inj Betamethasone 12 mg IM 2 doses, 24 hrs apart or Inj Dexamethasone 6 mg IM every 12 hrs for 4 doses can be used

46. All of the following are features of prematurity in a neonate, except:

A. No creases on sole
B. Abundant lanugo
C. Thick ear cartilage
D. Empty scrotum

Ans C The ears in a premature neonate are soft and flat with ear cartilage being deficient and pliant (and not thick)

47. Intracranial bleeds in premature infants is seen in the:

A. Germinal matrix
B. Corpus callosum
C. Basal ganglia
D. Choroid plexus

Ans A Intraventricular hemorrhage (IVH)
• Mainly seen in premature infants, Most commonly in the gelatinous sub-ependymal germinal matrix
• Immature blood vessels in this highly vascular region with poor tissue support predispose premature infants to hemorrhage
• Germinal matrix involutes in full-term neonates & vascular integrity improves; so, it is less common in term babies

48. Anticipated preterm delivery. Dose of dexamethasone given to mother is:

A. 12 mg 12 hrly 2 doses
B. 12 mg 24 hrly 4 doses
C. 6 mg 24 hrly 2 doses
D. 6 mg 12 hrly 4 doses

Ans D

49. Key features of kangaroo mother care are all the following except:

A. Skin to skin contact between mother and baby
B. Exclusive breastfeeding
C. Initiated in a facility and continued at home
D. Done for babies with cyanosis

Ans D Kangaroo mother care (KMC) is done for:
• All stable LBWQ babies are eligible for KMC; it can be started immediately after birth for birth wt > 1800 g
• Sick & very small babies (<1200 gm), KMC started after baby is hemodynamically stable a. Heart diseases, b. Respiratory distress syndrome, c. Necrotizing enterocolitis and e. Intraventricular hemorrhage

50. Preterm babies have increased chance of:

A. Heart disease
B. Respiratory distress syndrome
C. Necrotizing enterocolitis
D. Meconium aspiration syndrome
E. Intraventricular hemorrhage

Ans B from surfactant deficiency is the hallmark complication of prematurity (along with necrotizing enterocolitis and intraventricular hemorrhage, also increased in preterm infants).

51. Blood volume in preterm neonate is:

A. 90 mL/kg
B. 80 mL/kg
C. 70 mL/kg
D. 60 mL/kg

Ans A Blood volume is 80-100 mL/kg for term neonates & 90-105 mL/kg in preterm neonates (higher plasma volume) fo 49. b. 91-95% In preterm neonates, the currently recommended range of oxygen saturation target is 91–95%.

52. Preterm neonate target oxygen saturation is:

A. >95%
B. 91–95%
C. 85–90%
D. <80%

Ans B Current neonatal resuscitation guidance targets an oxygen saturation of about 91-95% in preterm infants to avoid the harms of both hypoxia and hyperoxia.

53. A pediatrician calls an ophthalmologist to examine baby if:

A. Baby born at 28 weeks gestation
B. Baby has respiratory distress
C. Birth weight of the baby is 2.3 kg
D. Jaundice

Ans A Indications of Screening for Retinopathy of prematurity (ROP):
• Birth weight <1,500 g or gestational age < 32 wk &
• Selected neonates with a birth weight between 1,500 and 2,000 g or gestational age of >32 wk with an unstable clinical course, including those requiring cardiorespiratory support.

54. Preterm babies have:

A. Extramedullary hematopoiesis
B. Greater risk of hypothermia
C. Greater risk of hypoglycemia
D. **All of above**

Ans D In a premature infant, hematopoiesis is frequently seen in the liver & occasionally in the spleen, lymph nodes or thymus.

55. A fetus born at 36 weeks, AGA, breast fed, should be adequately monitored for all of the following except:

A. Hyperbilirubinemia
B. Hypoglycemia
C. Hypocalcemia
D. **Bacterial sepsis**

Ans D Sepsis screen is done only when neonatal sepsis is suspected. Pre-emptive monitoring for sepsis is usually not done.

56. A woman delivered a baby of 2.2 kg weight. Her LMP is not known. To know the maturity of baby, which of the following are used:

A. **Sole crease**
B. Ear cartilage
C. Breast nodule
D. Anterior fontanel
E. Weight of the baby 11: Normal Neonate and Neonatal Resuscitation

Ans A b. Ear cartilage, and c. Breast nodule

57. Retinopathy of prematurity is caused by:

A. **Less gestation age**
B. Low birth weight
C. O toxicity
D. Carbohydrate excess 2 PRIMITIVE NEONATAL REFLEXES

Ans A Less gestational age > c. O toxicity 2 Retinopathy of prematurity (ROP) is characterized by abnormal proliferation of small retinal blood vessels in preterm neonate Risk factors for ROP are Hyperoxia (O toxicity), Shock, sepsis, hypoxia, acidosis, blood transfusion, PDA & mechanical ventilation

58. A newborn presents in emergency in a state of shock. Which is the ideal fluid to be given to the newborn?

A. **Normal saline**
B. Dextrose 5%
C. Ringer lactate
D. Dextrose 25%

Ans A For a newborn in shock, supportive treatment with type O Rh-negative blood or normal saline is indicated for hemorrhage or hypovolemia, respectively.

59. Definite contraindication for bag and mask ventilation is:

A. Cleft lip
B. Cleft palate
C. **Congenital diaphragmatic hernia**
D. Umbilical hernia

Ans C Bag & mask ventilation should be avoided in Congenital diaphragmatic hernia, because it enlarges the stomach and small bowel and with thus makes oxygenation more difficult.

60. Delayed clamping of Umbilical Cord at birth is associated with all of the following except:

A. Improved Iron status
B. Improved clinical anemia at 2–6 month of age
C. **Increase in symptomatic polycythemia among infants**
D. Increased chances of neonatal jaundice requiring phototherapy232 Section 2: Neonatology

Ans C NRP 2015 guidelines Delayed Cord Clamping
• It is associated with less intraventricular hemorrhage, higher blood pressure & blood volume, lesser anemia in infants upto 2-6 months age, lesser need for blood transfusion after birth, and less necrotizing enterocolitis; There is no evidence of decreased mortality.
• Late clamping may result in delivery of an extra 20-40 mL of blood and 30-35 mg of iron to the newborn.
• Polycythemia is a risk with delayed clamping but is often asymptomatic.

61. Special newborn care unit should comprise of:

A. 2:1
B. 3:1
C. **4:1**
D. 5:1

Ans C 12 beds 4 rooming in Newborn care facilities at various levels of public health services: Newborn Care Corner Newborn Stabilization Unit Special Newborn Care Unit Characteristics (NBCC) (NBSU) (SNCU) Location All points of childbirth CHC / FRU District/sub-district hospitals including PHC & subcentre Function To provide immediate To stabilize sick newborns be- Care for sick newborns < 1800 care at birth, feeding fore referral to higher centres; & gm (all types of care except

62. To provide immediate care to all newborns at birth, a health facility must have within its delivery room:

A. **Newborn care corner**
B. Newborn stabilization unit
C. Special newborn care unit
D. Home based newborn care unit

Ans A

63. Identify the device shown below:

A. Venturi mask
B. **Ambu bag**
C. Nasal canula
D. Laryngeal mask airway

Ans B This is an AMBU bag, that is a self-inflating bag used for ventilating neonates & children.

64. Most confirmatory sign of endotracheal intubation is?

A. Chest rise
B. Auscultation
C. Spirometry
D. **Capnography**

Ans D • Exhaled CO detection or Capnography is the recommended or confirmatory method to assess position of Endotracheal tube
• Other methods are: bilateral chest movements, presence of equal breath sounds bilaterally, condensation in endotracheal tube. b. ↓ pH; ↓ PaO ; ↑ PaCO 2 2 Hypoxia, hypercapnia & acidosis ⇒ Respiratory center in the brain stimulated ⇒ Breathing

65. **The initiation of the first breath in a newborn is due to:**

A. ↑pH; ↑PaO ; ↓PaCO
B. **↓pH; ↓PaO ; ↑PaCO 2 2 2**
C. ↓pH; ↓PaO , ↓PaCO
D. ↓pH; ↑PaO , ↓PaCO 2 2 2

Ans B The mild asphyxia of birth (falling pH and PaO2, rising PaCO2) stimulates chemoreceptors and is a key trigger of the newborn's first breath, along with thermal and tactile stimuli.

66. **A 32 week newborn baby with RR-86/min, grunting present with no nasal flaring, abdomen behind in movement than chest, minimum interocostals retraction and no xiphisternal retraction. What is the Silverman scoring?**

A. 1
B. **4**
C. 3
D. 6

Ans B Silverman-Anderson Score
• It is used to assess the degree of respiratory distress in preterms newborns with Respiratory Distress Syndrome.
• The higher the score, the greater the respiratory distress.
Components of Silverman score: Components 0 1 2 Score in given Q Upper chest retraction Chest & abdomen rise Lag of upper chest as abdo- Chest & abdomen move as 1 ogether men rises a "see-saw" Lower chest retraction Absent Minimal Marked 1 Xiphoid retraction Absent Minimal Marked 0

67. **Severe bradycardia in a newborn is HR: (TN PGMEE 2009**

A. <40/min
B. **<60/min**
C. <80/min
D. <100/min

Ans B Heart rate < 60/min is an indication for chest compressions along with positive pressure ventilation in a newborn.

68. **A baby is born with meconium stain liquor. All of the following are taken into account for terming a baby Vigorous, except:**

A. Tone
B. **Color**
C. HR
D. Respiration

Ans B A 'vigorous' neonate has Strong respiratory effortsQ, Good muscle toneQ & Heart rate > 100/minuteQ

69. **A non-ventilated preterm baby in incubator is under observation. Which is the best way to monitor the baby's breathing and detect apnea?**

A. Infrared throracic movement study
B. Capnography
C. Nasal digital temperature monitoring
D. **Impedance technique**

Ans D Respiratory monitoring is based on impedance technique, that measures changes in electrical resistance during breathing; The electrode is fixed on the chest wall to pick up signals which are displayed as respiratory rate.

70. **A newborn was given a drug in neonatal ICU, but then was found in respiratory distress. The likely drug responsible is:**

A. **Morphine**
B. Naloxone (AIPGMEE 07)
C. Salbutamol
D. Soda-bicarb Chapter 11: Normal Neonate and Neonatal Resuscitation 233 Answers

Ans A Causes of respiratory depression in neonate
• Intrapartum asphyxia (most common causeQ • Prematurity: CNS immaturity, surfactant deficiency
• Drugs: Morphine anesthetics • Respiratory: Diaphragmatic hernia, obstructive lesions
• Sepsis • CNS abnormalities: Malformation, trauma, myopathy 12 Chapter Diseases of IMPORTANT DISEASES OF NEWBORNS Diseases more common in: M Preterm Babies IUGR/SGA babies
• Neonatal sepsis • Polycythemia
• Respiratory: • Persistent pulmonary hypertension of newborn (PPHN)

12

CHAPTER 12

Diseases of Newborn



WHAT THIS CHAPTER COVERS

- IUGR & SGA/LGA
- Birth Asphyxia & HIE
- Neonatal Jaundice
- Neonatal Sepsis
- Respiratory Distress Syndrome
- Transient Tachypnea & Wet Lung
- Meconium Aspiration
- Persistent Pulmonary Hypertension
- Neonatal Convulsions
- Intraventricular Hemorrhage
- Periventricular Leukomalacia
- Neonatal Hypoglycemia
- Neonatal Hypocalcemia
- Necrotizing Enterocolitis
- Neonatal Anemia & Polycythemia
- Neonatal Thrombocytopenia
- Patent Ductus Arteriosus
- Neonatal Shock
- Neonatal Dermatology

249 MCQs **19** subtopics

12.1 IUGR / SGA, Preterm & LGA

	Symmetric IUGR (20–30%)	Asymmetric IUGR (70–80%)
Onset	**Early (1st/2nd trimester)**	**Late (3rd trimester)**
Cause	**Intrinsic** – chromosomal, TORCH, drugs, constitutional	**Extrinsic – placental insufficiency** (PIH, pre-eclampsia, anaemia, multiple gestation)
Proportion	Weight, length & HC all reduced proportionally	**Head sparing** – HC normal; weight ↓↓
Ponderal index [wt (g) × 100 / length (cm)^3]	**Normal (≥ 2.0)**	**Low (< 2.0)**
Cell number	↓ (hypoplastic)	Normal, cell size ↓
Prognosis	**Poor** (catch-up rare)	**Better – catch-up growth**

- **Problems of SGA/IUGR babies**: **hypoglycaemia (MC metabolic problem)**, hypothermia, **polycythaemia** (chronic hypoxia → EPO ↑), hypocalcaemia, **meconium aspiration**, birth asphyxia, PPHN, NEC; later – **metabolic syndrome (Barker hypothesis)**.
- **Preterm problems** (by system): **RDS**, apnoea of prematurity, BPD, **IVH, PVL**, PDA, **NEC**, ROP, jaundice (kernicterus at lower levels), hypothermia, hypoglycaemia, sepsis, anaemia, metabolic bone disease, feeding intolerance.
- **LGA / macrosomia** (> 4 kg): **infant of diabetic mother**, Beckwith–Wiedemann, Sotos, obese mother, post-term → birth injuries (shoulder dystocia, **Erb palsy, clavicle fracture**), hypoglycaemia.

Infant of diabetic mother (IDM)

- **Pedersen hypothesis**: maternal hyperglycaemia → fetal hyperglycaemia → **fetal hyperinsulinism** → macrosomia (except heart/brain/kidney).
- **Problems**: **hypoglycaemia (MC; first 1–6 h)**, **macrosomia**, birth trauma, **RDS (insulin delays surfactant maturation – PG is reliable maturity marker)**, **hypocalcaemia, hypomagnesaemia, polycythaemia & hyperviscosity**, **hyperbilirubinaemia**, **hypertrophic cardiomyopathy (septal; transient)**, **renal vein thrombosis**, **small left colon syndrome**, TTN.
- **Congenital anomalies (3–4× ↑, related to periconceptional HbA1c)**: **caudal regression syndrome / sacral agenesis – most specific (rare)**; **CHD – most common anomaly: VSD (MC)**, **TGA (most characteristic)**, HCM; NTD, anencephaly, holoprosencephaly, duplex kidney, small left colon.

12.2 Perinatal Asphyxia & Hypoxic-Ischaemic Encephalopathy (HIE)

- **Perinatal asphyxia** (AAP/ACOG criteria): **profound metabolic/mixed acidosis (cord pH < 7.0)**, **Apgar 0–3 beyond 5 min**, **neonatal encephalopathy (seizures, coma, hypotonia)** and **multi-organ dysfunction**.
- NNF/India practical: **no breathing/gasping at 1 minute** ("birth asphyxia" in NFHS context); moderate asphyxia – Apgar 4–6 at 1 min.
- **Organ involvement**: **kidney (MC – AKI/oliguria)**, CNS, heart (myocardial dysfunction, tricuspid regurgitation), lungs (PPHN, MAS), liver (↑ enzymes, coagulopathy), GI (NEC), haematological (DIC, thrombocytopenia), metabolic (hypoglycaemia, hypocalcaemia, **SIADH**).

Sarnat & Sarnat staging of HIE

Feature	Stage I (mild)	Stage II (moderate)	Stage III (severe)
Consciousness	**Hyperalert, irritable**	**Lethargic/obtunded**	**Stupor/coma**
Tone	Normal	**Mild hypotonia**	**Flaccid**

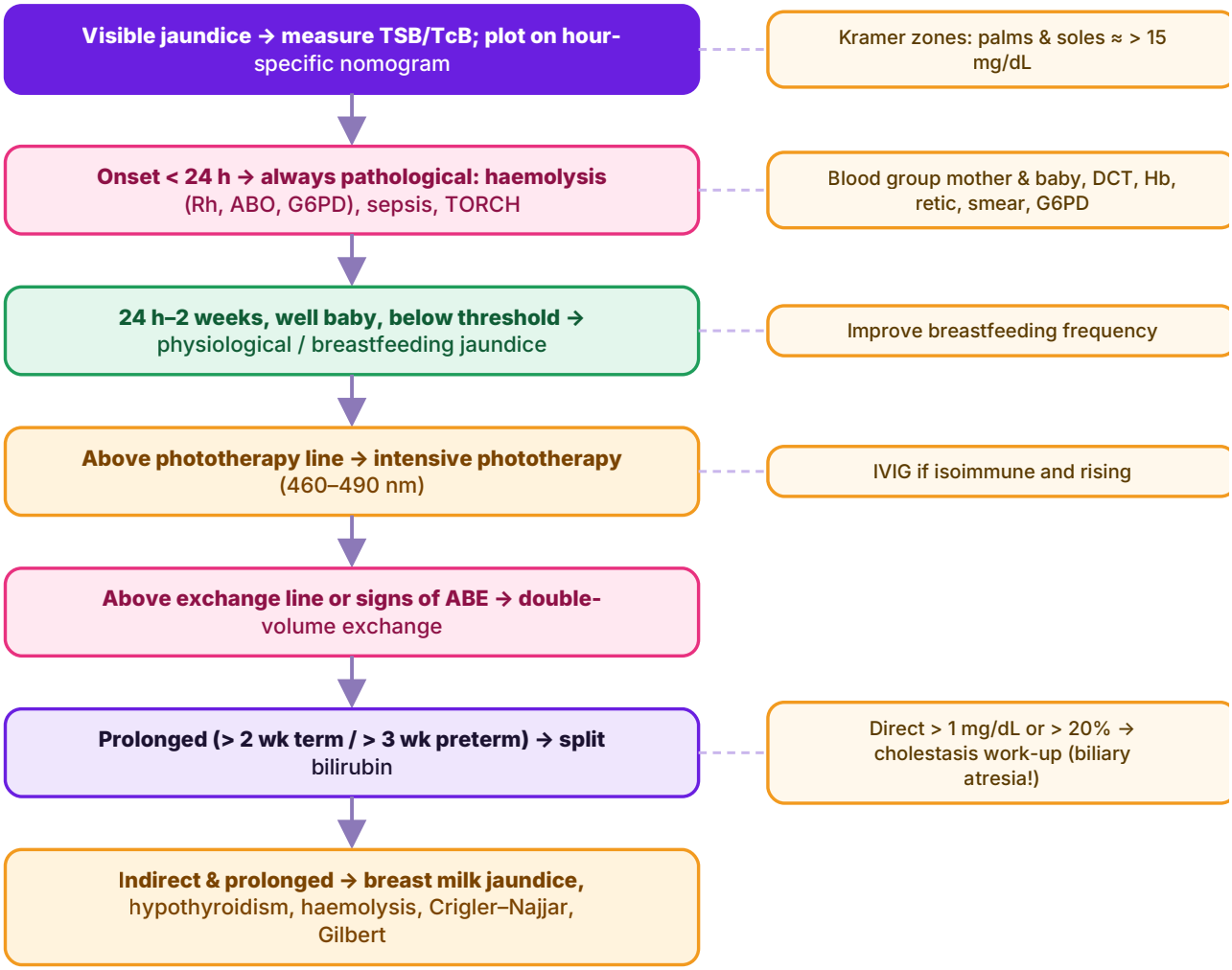
Feature	Stage I (mild)	Stage II (moderate)	Stage III (severe)
Posture	Mild distal flexion	Strong distal flexion	Intermittent decerebration
Reflexes	Hyperactive	Hyperactive	Decreased/absent
Moro	**Strong, exaggerated**	Weak, incomplete	Absent
Suck	Weak	Weak/absent	Absent
Pupils	**Mydriasis** (sympathetic)	**Miosis** (parasympathetic)	Variable, unequal, poor light reflex
Seizures	**None**	**Common (focal/multifocal)**	Uncommon (EEG suppression)
EEG	Normal	Low voltage, periodic	**Burst-suppression → isoelectric**
Duration	< 24 h	2–14 days	Hours–weeks
Outcome	**~100% normal**	80% normal; abnormal if > 5–7 days	**~50% die; survivors – severe sequelae (CP)**

HIE MANAGEMENT

Supportive: maintain **normal temperature (avoid hyperthermia)**, ventilation (avoid hypocapnia), perfusion (inotropes), **glucose 75–100 mg/dL**, calcium, **fluid restriction (SIADH/AKI) – 2/3 maintenance**, treat seizures (**phenobarbitone DOC 20 mg/kg loading**), avoid hyper-oxia. **Therapeutic hypothermia (whole-body cooling to 33.5 °C for 72 h, then rewarm over 6–12 h)** – **≥ 36 wk with moderate–severe HIE, started within 6 h of birth** – ↓ death/disability in **high-income NICUs**; HELIX trial (2021) showed **no benefit/↑ mortality in LMIC (incl. India)** – use only in tertiary NICUs with full intensive care.

- Neuroimaging: **MRI (diffusion-weighted) day 3–7 – IOC for HIE**; patterns – **basal ganglia-thalamus (acute profound/near-total)**, **watershed (prolonged partial)**. Cranial USG – PVL/IVH in preterm.
- Sequelae: **spastic quadriplegia, dyskinetic CP** (basal ganglia), ID, epilepsy, microcephaly, cortical visual impairment.
- **Preterm equivalent: periventricular leukomalacia (PVL) → spastic diplegia**.

12.3 Neonatal Jaundice



Approach to neonatal hyperbilirubinaemia

- Clinically visible at **> 5–7 mg/dL** in newborns; **cephalocaudal progression (Kramer's rule)**:

Kramer zone	Area	Approx. serum bilirubin
1	Head & neck	4–8 mg/dL (~6)
2	Upper trunk (above umbilicus)	5–12 (~9)
3	Lower trunk & thighs	8–16 (~12)
4	Arms & legs below knees	11–18 (~15)
5	**Palms & soles**	**> 15 mg/dL**

Physiological vs pathological jaundice

Feature	Physiological	Pathological (any one)
Onset	**After 24 h** (day 2–3)	**Within first 24 h**
Peak	Day 3–4 term (≤ 12–15 mg/dL); day 5–7 preterm	Exceeds phototherapy threshold for age/risk
Rate of rise	< 5 mg/dL/day	**> 5 mg/dL/day or > 0.2 mg/dL/h**
Duration	**< 2 weeks term (< 3 weeks preterm)**	**> 2 weeks term / > 3 weeks preterm (prolonged)**

Feature	Physiological	Pathological (any one)
Conjugated bilirubin	Normal	**Direct > 1 mg/dL (if TSB < 5) or > 20% of total**
Palms & soles	Not stained	**Stained**
Associated signs	Well baby	Pallor, hepatosplenomegaly, lethargy, poor feeding, **pale stools/ high-coloured urine**

- **Causes of physiological jaundice**: ↑ RBC mass & shorter RBC lifespan (70–90 days), **↓ UGT1A1 activity (1% of adult)**, ↑ enterohepatic circulation (β-glucuronidase, sterile gut), ↓ ligandin.

Causes by timing

< 24 h (always pathological)	24–72 h	> 72 h – 2 weeks	Prolonged (> 2–3 weeks)
Haemolytic disease – Rh, ABO, other minor group	Physiological	**Sepsis**, breastfeeding (inadequate intake) jaundice	**Breast milk jaundice**
G6PD deficiency, spherocytosis	Sepsis, polycythaemia, bruising/cephalhaematoma	Neonatal hepatitis, galactosemia, hypothyroidism	**Congenital hypothyroidism**
Congenital infection (TORCH), sepsis	G6PD, Crigler–Najjar, Gilbert		**Cholestasis – biliary atresia** (pale stools – urgent)

	Breastfeeding jaundice	Breast milk jaundice
Onset	**First week (day 2–5)**	**After first week (day 5–7), peaks 2–3 wk**
Cause	**Inadequate intake** → dehydration, ↑ enterohepatic circulation	**Substances in milk – β-glucuronidase, pregnanediol, NEFA** inhibit UGT
Management	**Increase frequency of breastfeeding (8–12/ day)**	**Continue breastfeeding**; rarely interrupt 24–48 h (diagnostic)
Prognosis	Good	Good; can last up to 2–3 months

Phototherapy & exchange transfusion

- Thresholds based on **gestational age, postnatal age (hours) and risk factors** (AAP 2022 nomograms / Bhutani nomogram for ≥ 35 wk). **NICE/AAP rough rule (≥ 35 wk, 72 h): phototherapy ~18–21 mg/dL; exchange ~25 mg/dL+**; for **< 35 weeks (NICE, after 72 h)**: phototherapy at **TSB (µmol/L) = gestational age (wk) × 10 – 100** and exchange at **GA × 10** (e.g. 30 wk → phototherapy ~200 µmol/L ≈ 11.7 mg/dL); **< 1000 g – phototherapy at ~5–6 mg/dL**. (1 mg/dL = 17.1 µmol/L.)
- **Mechanism**: **blue-green light 460–490 nm** → bilirubin converted to water-soluble isomers: **configurational (4Z,15E – fast, reversible)**, **structural isomerisation → lumirubin (most important for elimination – irreversible)**, photo-oxidation (minor).
- **Effective phototherapy**: irradiance **≥ 30 µW/cm²/nm (intensive)**, maximal skin exposure, distance 30–45 cm (LED closer), eyes covered, turn baby, continue feeds.
- **Side effects**: **insensible water loss ↑ (dehydration)**, loose green stools, skin rash, hyperthermia, **bronze baby syndrome (in cholestasis – contraindicated relatively)**, retinal damage (cover eyes), riboflavin deficiency, separation from mother, hypocalcaemia.
- **Exchange transfusion**: **double-volume (2 × 80–85 mL/kg = ~160–170 mL/kg)** removes ~85% of sensitised RBCs & ~50% of bilirubin; use **O-negative, cross-matched with mother**, fresh (< 5 days), irradiated; via umbilical vein (push-pull); complications – **hypocalcaemia (citrate)**, hypoglycaemia, hyperkalaemia, thrombocytopenia, NEC, portal vein thrombosis, infection, air embolism.
- **IVIG (0.5–1 g/kg)** in **isoimmune haemolysis** if TSB rising despite intensive phototherapy or within 2–3 mg/dL of exchange level.

KERNICTERUS (BILIRUBIN ENCEPHALOPATHY)

Unconjugated (indirect), unbound, lipid-soluble bilirubin crosses BBB → deposits in **basal ganglia (globus pallidus – MC site)**, subthalamic nuclei, hippocampus, cranial nerve nuclei (III, VIII), cerebellum. **Acute**: lethargy, hypotonia, poor suck → **hypertonia, retrocollis-opisthotonus, high-pitched cry, fever, seizures**. **Chronic (kernicterus)**: **choreoathetoid CP, sensorineural hearing loss (auditory neuropathy – most sensitive), upward gaze palsy (Parinaud), dental enamel dysplasia, ID (often spared)**. Risk ↑ with **prematurity, acidosis, hypoalbuminaemia, sepsis, hypoxia, drugs displacing bilirubin (sulfonamides, ceftriaxone, aspirin)**.

Inherited unconjugated hyperbilirubinaemias

	Gilbert	Crigler–Najjar type I	Crigler–Najjar type II (Arias)
UGT1A1 activity	~30%	**Absent**	< 10%
Inheritance	AR (promoter TATA repeat)	AR	AR
Bilirubin	< 4–5 mg/dL, with fasting/stress	**20–50 mg/dL – kernicterus**	6–20 mg/dL
Phenobarbitone response	Yes	**No**	**Yes (↓ > 30%)**
Rx	None	**Phototherapy (lifelong, 12 h/day), liver transplant**	Phenobarbitone

- **Lucey–Driscoll** – transient familial (maternal serum inhibitor). **Dubin–Johnson** (MRP2/ABCC2 – conjugated, **black liver**), **Rotor** (conjugated, normal liver colour) – benign.

12.4 Haemolytic Disease of the Newborn (Erythroblastosis Fetalis)

	Rh isoimmunisation	ABO incompatibility
Frequency	Less common (prophylaxis)	**MC cause of HDN**
Mother / baby	**Rh-negative mother, Rh-positive baby**	**Mother O, baby A or B** (anti-A/B IgG)
First pregnancy	**Spared (sensitisation needs prior exposure)**	**Can affect first pregnancy**
Severity	**Severe – hydrops fetalis, stillbirth**, severe anaemia, early jaundice	**Mild** – jaundice in first 24 h, mild anaemia
Direct Coombs (DAT)	**Strongly positive**	**Weakly positive/negative**
Smear	Erythroblasts (nRBCs), reticulocytosis	**Microspherocytes**
Prevention	**Anti-D Ig**	None

- **Anti-D immunoglobulin**: **300 µg (1500 IU) IM within 72 h of delivery** of Rh+ baby to unsensitised Rh– mother (covers 30 mL fetal blood/15 mL RBCs); **antenatal prophylaxis at 28 weeks**; after abortion/ectopic/amniocentesis/APH – 50 µg if < 12 wk, 300 µg if > 12 wk. **Kleihauer–Betke test** quantifies fetomaternal haemorrhage.
- Fetal monitoring: **indirect Coombs (ICT) titre ≥ 1:16 (critical)** → **MCA peak systolic velocity Doppler (> 1.5 MoM = fetal anaemia; non-invasive, replaced amniocentesis ΔOD450 – Liley chart)** → **intrauterine transfusion (IUT) via cordocentesis** (O-negative irradiated).
- **Hydrops fetalis**: fluid in ≥ 2 compartments (skin oedema, ascites, pleural/pericardial effusion); **immune (Rh)** vs **non-immune (now MC – cardiac/arrhythmia, chromosomal – Turner, α-thalassaemia major (Hb Bart's – MC in SE Asia), parvovirus B19, twin-twin transfusion, CMV, syphilis)**.
- **Late anaemia** in Rh disease (weeks after exchange/IUT) – follow Hb.

12.5 Neonatal Sepsis

	Early-onset sepsis (EOS)	Late-onset sepsis (LOS)
Onset	**< 72 h** (NNF)	**≥ 72 h**
Source	**Maternal genital tract (vertical)**	**Nosocomial / community (horizontal)**
Organisms (India)	**Klebsiella, E. coli, Acinetobacter** (GBS less common)	**Klebsiella (MC), Acinetobacter, S. aureus, CONS, Pseudomonas, Candida**
Organisms (West)	**GBS (MC)**, E. coli, Listeria	CONS (MC – VLBW), S. aureus
Presentation	**Pneumonia (MC)**, fulminant, multisystem	Meningitis/bacteraemia, osteomyelitis

- **Risk factors for EOS**: **PROM > 18 h**, maternal fever ≥ 38 °C (chorioamnionitis), foul-smelling liquor, **> 3 vaginal examinations in labour**, prolonged labour, preterm, LBW, perinatal asphyxia.
- **Clinical**: **refusal/poor feeding & lethargy (earliest)**, **hypothermia (more common than fever)**, apnoea, respiratory distress, poor perfusion (CRT > 3 s), jaundice, abdominal distension, sclerema, seizures, bulging fontanelle, **hyper/hypoglycaemia**, bleeding.
- **IMNCI danger signs (possible serious bacterial infection – PSBI)**: not feeding well, convulsions, fast breathing (≥ 60), severe chest indrawing, temp ≥ 37.5 °C or < 35.5 °C, movement only on stimulation/no movement.

INVESTIGATIONS

Blood culture – gold standard (≥ 1 mL). **Sepsis screen (positive if ≥ 2)**: **TLC < 5000/mm³**, **ANC below Manroe/Mouzinho charts**, **immature : total neutrophil ratio ≥ 0.2**, **micro-ESR > 15 mm in 1st hour** (or age in days + 3), **CRP > 1 mg/dL (10 mg/L)**. **Procalcitonin** rises earlier than CRP. **LP in all culture-positive / clinically septic babies** (meningitis in up to 1/3 of LOS).

ANTIBIOTICS (NNF / AIIMS PROTOCOL)

- **Community-acquired / EOS**: **ampicillin (or penicillin) + gentamicin**.
- **Nosocomial LOS**: **piperacillin–tazobactam/cefotaxime + amikacin** → based on unit antibiogram; **meropenem** (ESBL/Acinetobacter), **vancomycin** (MRSA), **colistin** (CRE); **fluconazole/amphotericin** for fungal.
- **Duration**: culture-negative sepsis 5–7 days; **culture-positive sepsis 10–14 days**; **meningitis 14–21 days** (gram-negative 21 d); osteomyelitis/septic arthritis 4–6 weeks.
- **WHO simplified OPD regimen (when referral not possible) for PSBI**: **IM gentamicin daily + oral amoxicillin × 7 days**.
- Supportive: thermal care, glucose, fluids, oxygen, inotropes; **IVIG not recommended** (INIS trial).

12.6 Respiratory Distress in the Newborn

- **Signs**: **tachypnoea (RR > 60)**, **retractions**, **grunting (expiratory – auto-PEEP to prevent alveolar collapse)**, nasal flaring, cyanosis.
- **Silverman–Andersen score** (preterm; 0 best – 10 worst: upper chest retraction, lower chest retraction, xiphoid retraction, nasal flaring, expiratory grunt) – **higher = worse**. **Downes score** (term/any; RR, retractions, cyanosis, air entry, grunting): **< 4 mild, 4–7 moderate, > 7 impending respiratory failure (ABG)**.

Disease	Baby	Features	X-ray
RDS / hyaline membrane disease	**Preterm (< 34 wk), IDM, male, C-section without labour, 2nd twin, asphyxia**	Distress **within 6 h of birth (usually immediately)**, worsens over 48–72 h, grunting; **surfactant deficiency** (↓ phosphatidylcholine/DPPC)	**Ground-glass/ reticulogranular opacities, air bronchograms, low lung volume → "white-out"**

Disease	Baby	Features	X-ray
Transient tachypnoea of newborn (TTN, wet lung, RDS type II)	**Term/near-term, elective C-section (no labour)**, IDM	**Tachypnoea (up to 100–120), minimal retractions, good air entry; resolves in 24–72 h**; delayed clearance of lung fluid (ENaC)	**Prominent perihilar streaking (vascular markings), fluid in horizontal fissure, hyperinflation, "wet silhouette"**
Meconium aspiration syndrome (MAS)	**Post-term, SGA, asphyxiated, term**	Meconium-stained liquor & nails/cord, **barrel chest**, PPHN, **air leaks (pneumothorax)**	**Coarse patchy/streaky infiltrates, hyperinflation (flat diaphragm), atelectasis + emphysema**
Congenital pneumonia	PROM, chorioamnionitis	Sepsis features	Non-specific/**GBS mimics RDS**
Pneumothorax	Ventilated, MAS, RDS	Sudden deterioration, shift	Translucent hemithorax; **transillumination** bedside
Congenital diaphragmatic hernia	Term	**Scaphoid abdomen**, bowel sounds in chest, shifted heart sounds	Bowel loops in chest – see Surgery
PPHN	Term/post-term, MAS, asphyxia, CDH, sepsis	**Labile hypoxaemia, pre-post ductal SpO_2 gradient > 10%**	Clear/oligaemic lungs

RDS MANAGEMENT

- **Antenatal steroids** (24–34 wk) – most effective preventive measure.
- **CPAP (5–8 cm H_2O) from birth** in spontaneously breathing preterms – first-line.
- **Surfactant** (poractant alfa 200 mg/kg, beractant/Survanta 100 mg/kg, Neosurf) **via ETT – INSURE (Intubate-SURfactant-Extubate) or LISA/MIST (less invasive)** if **FiO_2 > 30–40% on CPAP**; repeat dose if needed. Natural (animal-derived) surfactants better than synthetic.
- Complications: air leaks, **PDA**, IVH, BPD, ROP, NEC, pulmonary haemorrhage (post-surfactant).

PPHN (persistent fetal circulation)

- Failure of normal fall in PVR → **right-to-left shunting across PDA & PFO**; **pre-ductal (right hand) SpO_2 > post-ductal (foot) by ≥ 10%** (if shunt at ductal level).
- Echo – **IOC** (TR jet, R→L shunt, structurally normal heart).
- Rx: **gentle ventilation, avoid hypoxia/acidosis, sedation, surfactant (if MAS/RDS), inotropes (milrinone), inhaled nitric oxide (iNO) – selective pulmonary vasodilator (DOC)**; sildenafil; **ECMO** for refractory.

Apnoea of prematurity

- **Pause in breathing ≥ 20 s**, or **shorter with bradycardia (< 100) or desaturation**; **onset day 2–7**, inversely related to gestation (almost all < 28 wk); **central/obstructive/mixed (MC)**.
- **Periodic breathing**: pauses 5–10 s followed by 10–15 s of rapid breathing, **no bradycardia/desaturation** – benign.
- Exclude secondary causes (**sepsis, hypoglycaemia, hypothermia, IVH, seizures, anaemia, PDA, GER, drugs**) – especially if apnoea on **day 1 or after 2 weeks**.
- Rx: **caffeine citrate – DOC (loading 20 mg/kg, maintenance 5–10 mg/kg/day)** – also ↓ BPD & CP (CAP trial); CPAP/NIPPV; tactile stimulation. Usually resolves by **36–37 weeks PMA**. Prophylactic caffeine for all < 28–30 wk.

Bronchopulmonary dysplasia (BPD)

- **Need for O_2 at 36 weeks PMA** (in babies < 32 wk) after ≥ 28 days of O_2 (NICHD); "new BPD" – arrested alveolar development in ELBW.
- Risk: **prematurity (most important)**, oxygen toxicity, **barotrauma/volutrauma**, infection, PDA. Prevention: **caffeine, vitamin A (IM), CPAP over ventilation, less invasive surfactant, postnatal dexamethasone (DART – selective, low-dose after 7 days; early dexa → CP)**.
- CXR: hyperinflation with cystic & streaky areas.

12.7 Neonatal Seizures

Cause	Timing
HIE – MCC overall (first 24–72 h, within 12–24 h)	Day 1
Intracranial haemorrhage (IVH in preterm, SAH, subdural)	Day 1–3
Metabolic – hypoglycaemia, hypocalcaemia (early/late), hypomagnesaemia, hypo/hypernatraemia	Day 1–3; late hypocalcaemia day 5–10
Infection – meningitis, TORCH	Any (day 3+)
Pyridoxine dependency, IEM, NKH	Day 1–2, refractory
Neonatal abstinence (drug withdrawal)	Day 1–3
Perinatal arterial ischaemic stroke	Focal clonic, day 1–3
Benign familial neonatal epilepsy (KCNQ2) / "fifth-day fits" (benign neonatal convulsions)	Day 2–3 / **day 4–6**
Kernicterus	Day 3–7

Seizure type	Features
Subtle (MC – esp. preterm)	**Lip smacking, chewing, ocular (tonic horizontal deviation, staring, blinking), apnoea, "bicycling/pedalling/swimming" limb movements**
Clonic (focal/multifocal)	Rhythmic jerks 1–3/s; **focal clonic – best prognosis** (stroke); EEG correlate usually +
Tonic (generalised)	Preterm with IVH – poor prognosis
Myoclonic	Rare; **worst prognosis** (IEM, severe brain injury)
Spasms	Flexor/extensor

- **Jitteriness vs seizure**: jitteriness is **stimulus-sensitive, stops on gentle restraint/flexion, rhythmic tremor (equal amplitude), no ocular/autonomic changes**; seizures – not suppressed, ocular deviation, autonomic changes, EEG changes.
- **EEG/amplitude-integrated EEG (aEEG)** – electroclinical dissociation common (**electrographic-only seizures**).

TREATMENT SEQUENCE

1. **Glucose**: hypoglycaemia → **10% dextrose 2 mL/kg IV bolus** then infusion (GIR 6–8 mg/kg/min).
2. **Calcium gluconate 10% 2 mL/kg IV** slowly (if hypocalcaemia), **$MgSO_4$ 50% 0.2 mL/kg** (if hypomagnesaemia).
3. **Phenobarbitone 20 mg/kg IV loading – DOC**; additional 10 mg/kg × 2 (max 40 mg/kg); maintenance 3–5 mg/kg/day.
4. **Phenytoin/fosphenytoin 20 mg/kg** or **levetiracetam 40–60 mg/kg** (increasingly used, fewer side effects); then **midazolam infusion**, lidocaine.
5. **Pyridoxine 100 mg IV** trial (refractory seizures, under EEG).

12.8 Neonatal Hypoglycaemia

- **Operational threshold: blood glucose < 45–47 mg/dL (2.6 mmol/L)** in any newborn (NNF/AIIMS: < 45 mg/dL) – intervene; **< 40 mg/dL in first 4 h (AAP) / < 45 thereafter**.
- **At-risk babies (screen at 2, 6, 12, 24, 48, 72 h)**: **IDM, LGA, SGA/IUGR, preterm, perinatal asphyxia, sepsis, hypothermia, polycythaemia, Rh disease, Beckwith–Wiedemann, maternal β-blockers/terbutaline**.
- Symptoms: **jitteriness (MC), tremors, lethargy, poor feeding, apnoea, cyanosis, seizures, hypothermia**, high-pitched cry; many asymptomatic.

MANAGEMENT

- **Asymptomatic, glucose 25–45**: **feed (breast/EBM)**; buccal **dextrose gel 40% (200 mg/kg)**; recheck in 30–60 min; IV if persists.
- **Symptomatic or < 25 mg/dL (or persistent)**: **IV 10% dextrose 2 mL/kg bolus over 1 min, then infusion GIR 6–8 mg/kg/min** (≈ 100 mL/kg/day of 10% D gives GIR ~7); increase by 2 mg/kg/min every 15–30 min; **max via peripheral line 12.5% dextrose**.
- **GIR (mg/kg/min) = % dextrose × rate (mL/h) / (6 × weight in kg)** (or % dextrose × mL/kg/day / 144).
- **Refractory (GIR > 12 mg/kg/min) → hyperinsulinism**: **hydrocortisone**, **diazoxide** (KATP opener – DOC for hyperinsulinism), octreotide, glucagon (0.5–1 mg IM in emergency), partial pancreatectomy (focal – 18F-DOPA PET).

- **Persistent hypoglycaemia**: **congenital hyperinsulinism (ABCC8/KCNJ11 – MC)**, panhypopituitarism (with **micropenis, midline defects, cholestasis**), cortisol deficiency, GSD, galactosemia, FAO defects, Beckwith–Wiedemann.

12.9 Intraventricular Haemorrhage (IVH) & Periventricular Leukomalacia (PVL)

- **IVH**: preterm **< 32 weeks / < 1500 g**; origin – **germinal matrix (subependymal, caudothalamic groove)**; **90% within first 72 h (50% day 1)**; risk ↑ with fluctuating cerebral blood flow, RDS, pneumothorax, hypercapnia, rapid volume/bicarbonate, asphyxia.
- **Screening: cranial ultrasound (IOC) through anterior fontanelle** – at day 3–7 & at 36 weeks/discharge in all < 32 wk.

Papile grade	Description
I	**Germinal matrix haemorrhage** only
II	IVH **without ventricular dilatation** (< 50% ventricle)
III	IVH **with ventricular dilatation** (> 50%)
IV	**Periventricular haemorrhagic infarction (venous) – parenchymal**

- Complications: **post-haemorrhagic hydrocephalus (grade III/IV)**, CP, ID. Prevention – **antenatal steroids, delayed cord clamping**, avoid BP swings, gentle handling, indomethacin prophylaxis (selected).
- **PVL**: ischaemic necrosis of periventricular white matter (watershed, **dorsolateral to lateral ventricles**) in preterm (28–32 wk); **cystic PVL on USG at 2–6 weeks**; → **spastic diplegia** (lower limb fibres – corticospinal tracts nearest ventricles); visual impairment.

12.10 Necrotising Enterocolitis (NEC)

- **MC GI emergency in preterm neonates**; **> 90% in preterm (< 32 wk, VLBW)**; onset **2nd–3rd week** (inversely with gestation).
- **Risk factors**: **prematurity (most important)**, **formula feeding** (breast milk protective), rapid feed advancement, **asphyxia/hypoxia**, polycythaemia, exchange transfusion, **umbilical catheters**, PDA/indomethacin, H2 blockers, cocaine, CHD; term – CHD, polycythaemia.
- **Site**: **terminal ileum & proximal colon (MC)**.
- **Features**: **feeding intolerance (↑ gastric residuals), abdominal distension (earliest/most common), bloody stools**, bilious vomiting, abdominal wall erythema, apnoea, shock, DIC, thrombocytopenia.
- **X-ray**: **pneumatosis intestinalis (intramural gas – pathognomonic)**, **portal venous gas**, fixed dilated loop, **pneumoperitoneum – "football sign", Rigler sign (perforation)**.

Modified Bell's stage	Features	Management
I (suspected)	Non-specific – residuals, mild distension, occult blood; X-ray normal/ileus	NPO 3 days, antibiotics, observe
II (definite)	+ absent bowel sounds, tenderness; **pneumatosis ± portal venous gas**	**NPO + gastric decompression + IV antibiotics (ampicillin + gentamicin + metronidazole / pip-tazo) 7–14 days**
III (advanced)	Shock, DIC, peritonitis; **IIIB – pneumoperitoneum (perforation)**	**Surgery – laparotomy/resection or primary peritoneal drainage**

- Prevention: **exclusive breast milk (mother's/donor) – most effective**, probiotics, standardised slow feeding protocols. Late complication: **intestinal stricture (MC – colon, left side)**, short-bowel syndrome.

12.11 Polycythaemia, Birth Injuries & Other Problems

- **Neonatal polycythaemia**: **venous Hct > 65%**; causes – **delayed cord clamping, IDM, SGA, twin-twin (recipient), maternal hypertension, Down, Beckwith–Wiedemann, high altitude**; hyperviscosity → plethora, lethargy, hypoglycaemia, jaundice, NEC, **renal vein thrombosis**, seizures; Rx **partial exchange transfusion with NS** if symptomatic or Hct > 70–75% asymptomatic. Volume (mL) = blood volume × weight × (observed Hct – desired Hct) / observed Hct.
- **Birth injuries**: **clavicle fracture (MC fracture)** – crepitus, asymmetric Moro, callus by 7–10 days, no treatment needed; **facial nerve palsy** (forceps/pressure) – inability to close eye, mouth drawn to normal side on crying, most recover; **sternomastoid tumour**; **cephalhaematoma, subgaleal**; humerus/femur fracture; **phrenic nerve palsy (C3–C5, with Erb palsy) – elevated hemidiaphragm**; liver/spleen rupture; spinal cord injury (breech).
- **Neonatal abstinence syndrome** (maternal opioids): high-pitched cry, tremors, poor sleep, hyperactive reflexes, sneezing, yawning, diarrhoea, **seizures**; **Finnegan score**; Rx **non-pharmacological first ("eat-sleep-console")**, morphine/methadone; phenobarbitone for polysubstance.
- **Neonatal hypocalcaemia** – see Fluids chapter. **Late hypocalcaemia (day 5–10)** – high phosphate cow's milk, hypoparathyroidism (DiGeorge), maternal hyperparathyroidism.
- **Neonatal thrombocytopenia**: sepsis (MC in sick neonates), **neonatal alloimmune thrombocytopenia (NAIT – anti-HPA-1a, first pregnancy affected, ICH risk; Rx washed maternal/HPA-compatible platelets, IVIG)**, maternal ITP (autoimmune – milder), TAR, Kasabach–Merritt, congenital infections.
- **Retinopathy of prematurity** – see Eye chapter (screen < 34 wk / < 1750 g at 2–4 weeks – India).

Congenital lung malformations

Lesion	Key features
Congenital pulmonary airway malformation (CPAM / CCAM)	MC congenital lung lesion; multicystic mass, respiratory distress/hydrops; Rx resection
Pulmonary sequestration	Non-functioning lung with **systemic arterial supply (from aorta)**; intralobar (MC, **left lower lobe**, pulmonary venous drainage) vs extralobar (systemic venous drainage, associated anomalies)
Congenital lobar emphysema	**Left upper lobe (MC)** overinflation – bronchial cartilage deficiency; mediastinal shift; lobectomy
Pulmonary hypoplasia	**CDH, oligohydramnios (Potter), thoracic dystrophy**, neuromuscular disease
Bronchogenic cyst	Mediastinal/subcarinal

Practice MCQs · Diseases of Newborn

249 questions · correct option highlighted in green · cover the answer strip to self-test

IUGR & SGA/LGA

1. Which of the following statements is NOT true regarding IUGR?

A. Elevated Head circumference/ Abdominal circumf erence (HC/AC) ratio
B. **Ponderal index above 10th percentile**
C. Increased diastolic velocity in Middle cerebral artery (MCA) Doppler
D. Femur length/Abdominal circumference (FL/AC) ratio is >23.5

Ans B Discussing about the options one by one, About option a. True, because, a fetus with asymmetric IUGR has a normal head dimension but a small abdominal circumference (due to decreased liver size); so HC/AC ratio increased About option b. False, as Ponderal index should be less than 10th percentile & not more in IUGR About option c. True, Increased diastolic velocity in Middle cerebral artery (MCA) doppler is seen in IUGR About option d. True, Femur length/ Abdominal circumference (FL/AC) ratio is >23.5 in IUGR

2. In asymmetrical IUGR which organ is not affected?

A. Subcutaneous fat
B. Muscle
C. Liver
D. **Brain**

Ans D In Asymmetric IUGR, Brain growth (head circumference) is usually spared & it is usually due to Medical/obstetric problem of mother in late trimesters like poor nutrition, hypertension, placental insufficiency

3. IUGR is caused by all except:

A. **Diabetes**
B. Alcohol
C. Smoking
D. Chronic renal failure

Ans A Maternal Diabetes usually leads to a LFD baby & not IUGR

4. Infants of diabetic mothers are at high risk of which of the following anomalies?

A. A baby born to mother treated with beta blockers
B. Infant of diabetic mothers
C. **Appropriate for gestational age babies**
D. IUGR babies

Ans C Transposition of great arteries

5. Twin within 1-3 days of zygote formation:

A. **Diamniotic dichorionic**
B. Diamniotic monochorionic
C. Monoamniotic monochorionic
D. None of the above

Ans A Time interval between ovulation and Nature of membrances cleavage of egg 0-72 hr Diamniotic, dichorionic 4-8 days Diamniotic, monochorionic 9-12 days Monoamniotic, monochorionic

6. When mother complains that baby is more comfortable and breaths better when held against shoulder. What important clinical problem do you suspect?

A. Nose block 182.
B. Psychological anxiety or insecurity
C. Aerophagy with abdominal fullness
D. **Orthopnea**

Ans D b. Multicystic dysplastic kidney Multicystic dysplastic kidneyQ is the most common cause of abdominal mass in a neonate.

7. The following drugs can be given with caution to breast-feeding mother

A. **psychotropic drugs**
B. amphetamines
C. bromocriptine
D. chloramphenicol
E. methimazole

Ans A Amphetamines, bromocriptine, chloramphenicol, and methimazole are contraindicated in breast-feeding mother.

8. The incidence of all the following are increased in large for gestational age newborn EXCEPT

A. cephalohematoma
B. **hypocalcemia**
C. hypoglycemia
D. congenital heart disease
E. developmental retardation

Ans B Large for gestational age LGA infants, regardless of their gestational age, have a higher incidence of birth injuries, such as cervical and brachial plexus injuries, phrenic nerve damage with paralysis of the diaphragm, fractured clavicles, cephalohematomas, subdural hematomas, and ecchymoses of the head and face. LGA infants are also at increased risk for hypoglycemia and polycythemia.

9. The blood smear of infant with hemolytic disease of the newborn (erythroblastosis fetalis) typically shows the following EXCEPT

A. polychromasia
B. increasedreticulocyte count
C. a marked increase in nucleated RBCs
D. thrombocytopenia
E. **low white blood cell count**

Ans E The white blood cell count is usually normal but may be elevated.

10. The following definitions are true EXCEPT

A. a syndrome is defined as a pattern of multiple abnormalities that are related by pathophysiology and result from a common defined etiology
B. dysplasia sequence is a poor organization of cells into tissues or organs
C. an association refers to a nonrandom collection of malformations in which there is an unclear relationship among the malformations
D. **disruption sequence is a mechanical (uterine) forces that alter structure of intrinsically normal tissue**
E. malformation sequence is a single, local tissue morphogenesis abnormality that produces a chain of subsequent defects

Ans D Disruption sequence is in utero tissue destruction after a period of normal morphogenesis while deformation sequence is a mechanical (uterine) force that alters structure of intrinsically normal tissue.

11. Which common sugar does the clinitest screen not detect?

A. glucose
B. fructose
C. galactose
D. **sucrose**
E. lactose

Ans D Reducing sugars (e.g., glucose, fructose, galactose, pentoses, and lactose) are detected, but sucrose is not a reducing sugar.

Birth Asphyxia & HIE

12. Asymmetrical fetal growth restriction is associated with:

A. Chromosomal aberration
B. Viral infection
C. Idiopathic
D. **Placental insufficiency**

Ans D

13. Which of the following is the principal mode of heat exchange in an infant incubator?

A. Radiation
B. Evaporation
C. **Convection**
D. Conduction

Ans C

14. In Sarnat and Sarnat classification of hypoxic ischemic encephalopathy, seizures occur in which stage?

A. Hypoxic ischemic encephalopathy
B. **Meconium aspiration syndrome**
C. Kernicterus
D. Necrotizing enterocolitis

Ans B Stage

15. About Hypoxic Ischemic encephalopathy true is:

A. Lower limbs affected more than upper limbs
B. Prox. Muscle > distal muscles
C. **Seizure**
D. Trunk involved

Ans C > b. Prox. Muscle > distal muscles This is a controversial question: Discussing about each option, a. In term babies, upper limbs are more affected than lower while in preterms, lower limbs are more affected b. Proximal muscle weakness is seen in parasagittal injury, commonly seen in term babies c. Seizures are seen in 20-50% of cases (in both, term & preterm neonates) d. Limbs are mainly involved; Hence, seizures can be seen in both term & preterm babies with HIE

16. All of the following therapies may be required in one-hour-old infant with severe birth asphyxia except:

A. Glucose
B. **Dexamethasone**
C. Calcium gluconate
D. Normal saline NEONATAL SEIZURES

Ans B The management protocol of babies with perinatal asphyxia includes:
• Oxygen supplementation, thermal control, monitor blood glucose & maintain euglycemia
• Fluid balance: Give dextrose containing IV fluids at 2/3 maintenance. For shock: Normal saline boluses Inotropes
• Prevent/control convulsions: PhenobarbitoneQ is the drug of choice of neonatal seizures
• Treat hypocalcemia, if it occurs calcium gluconate administration.

17. Which mechanism in phototherapy is chiefly responsible for reduction in serum bilirubin:

A. Photo-oxidation
B. Photo-isomerization
C. **Structural isomerization**
D. Conjugation

Ans C

18. Of the following, the condition which is associated with polyhydramnios is

A. renal agenesis (Potter syndrome)
B. Prune-belly syndrome
C. pulmonary hypoplasia
D. intestinal pseudo-obstruction
E. **diaphragmatic hernia**

Ans E Renal agenesis (Potter syndrome), Prune-belly syndrome, pulmonaryhypoplasia, and intestinal pseudo-obstruction are associated with oligohydramnios.

19. After severe birth asphyxia, infants may have motor automatisms characterized by

A. abscence of oral-buccal-lingual movements
B. time-synchronized electroencephalographic discharges
C. significant cortical epileptic activity
D. good response to anticonvulsant therapy
E. a poor prognosis

Ans E After severe birth asphyxia, infants may have motor automatisms characterized by oral-buccal-lingual movements, rotary limb activities (rowing, pedaling, and swimming), tonic posturing, or myoclonus. These motor activities are not usually accompanied by time-synchronized electroencephalographic discharges, may not signify cortical epileptic activity, respond poorly to anticonvulsant therapy, and are associated with a poor prognosis.

20. One of the following is FALSE in hemolytic disease of the newborn caused by blood group A and B incompatibility

A. ABO incompatibility occurs in 20-25% of pregnancies
B. A1 is more antigenic than A2
C. natural antibodies against A and B factors are usually IgM antibodies that do not cross the placenta
D. isoimmune hemolytic disease may be found in first-born infants irrespective of infant blood group
E. most cases are mild

Ans D Although antibodies against A and B factors occur without previous immunization ("natural" antibodies), they are usually IgM antibodies that do not cross the placenta. However, IgG antibodies to A antigen may be present and these do cross the placenta, so A-O isoimmune hemolytic disease may be found in first-born infants.

21. Chromosomal causes of congenital malformations include

A. Prader-Willi syndrome
B. X-linked hydrocephalus
C. Achondroplasia
D. Treacher Collins syndrome
E. Apert disease

Ans A X-linked hydrocephalus, Achondroplasia, Treacher Collins syndrome, and Apert disease are monogenic causes of congenital malformations.

22. The antenatal administration of magnesium sulfate before preterm delivery has been suggested to

A. reduce rates of necrotizing enterocolitis
B. decrease the risk of cerebral palsy in surviving infants
C. improve overall survival
D. decrease respiratory distress syndrome in premature infants
E. reduce rates of intraventricular hemorrhage

Ans B The antenatal administration of magnesium sulfate before preterm delivery has been suggested to decrease the risk of cerebral palsy in surviving infants but not improve overall survival. Prospective multicenter randomized trials are currently examining this potential benefit.

Neonatal Jaundice

23. What is the ponderal index of a neonate with weight 2 kg and height 50 cm:

A. 1.6
B. 3.6
C. 2.2
D. 2.6

Ans A Ponderal Index (PI) in Neonate = Weight (gm)/ Length (cm)3 × 100 ⇒ In this case, PI = 2000/(50)3 × 100 = 1.6

24. Hypothermia in neonate is characterized by:

A. Hyperactivity
B. Hypoglycemia
C. Apnea
D. Increased urinary output
E. Hyperglycemia PERINATAL ASPHYXIA

Ans B and c. Apnea Ghai 8/e p 144-146

25. Lungs do not collapse during expiration because of the presence of?

A. Hyaline membrane
B. Dipalmitoyl phosphatidyl choline
C. Macrophages
D. Interstitial fluid

Ans B Dipalmitoyl phosphatidyl choline is an important constituent of Surfactant, which prevent alveolar collapse during expiration.

26. All are features of infant born to diabetic mothers except:

A. Obesity
B. Learning disability
C. Ketotic hypoglycemia
D. Future diabetes mellitus

Ans B

27. Jaundice at birth or within 24 hours of birth is most commonly due to:

A. Erythroblastosis fetalis
B. Congenital hyperbilirubinemia
C. Biliary atresia
D. Physiological jaundice of newborn

Ans A Jaundice, consisting of either indirect or direct bilirubin, that is present at birth or appears within the 1st 24 hr after birth requires immediate attention and may be due to erythroblastosis fetalis, concealed hemorrhage, sepsis, or congenital infections, including syphilis, cytomegalovirus, rubella, and toxoplasmosis.

28. MC cause of cholestatic jaundice in newborn is:

A. Hypoplasia of biliary tract
B. Neonatal hepatitis
C. Choledochal cyst
D. Physiological

Ans B Neonatal hepatits Biliary atresia and neonatal hepatitis are the most common causes of cholestasis in early infancy.

29. A 4-day-old breastfed, otherwise well, term neonate presented with jaundice, on testing the bilirubin level was found to be 18 mg/dl. Which of the following is the best step of management?

A. Start phototherapy and continue breastfeeding
B. Start iv fluids and given phototherapy
C. Initiate exchange transfusion
D. Stop breastfeeding and do phototherapy

Ans A AAP nomogram for phototherapy in hospitalized infants of > 35 weeks gestation. AAP nomogram for exchange transfusion in hospitalized infants of ≥> 35 weeks gestation Out of the given options, start phototherapy and continue breastfeeding is the best answer.

30. A 3 days old neonate is brought by the mother to the pediatrician and is found to have a bilirubin of 18 g/dl. What is the next step?

A. Stop breastfeeding
B. Start phototherapy and continue breastfeeding
C. Start phototherapy and stop breastfeeding
D. Exchange transfusion

Ans B First line of treatment for pathological neonatal jaundice is phototherapy; breastfeeding should be continued;

31. All of the following are true about breast milk jaundice except?

A. Bilirubin is 10-15 mg/dl
B. Occurs between day 5-10 of life
C. Phototherapy is the treatment of choice
D. Staining of Diaper by urine is present
E. Breastfeeding should be stopped to treat it

Ans D e. Breastfeeding should be stopped to treat it • Breast milk jaundice: It is unconjugated hyperbilirubinemia, so staining of diaper is not present
• Seen in 2% of breastfed term infants after the 7th day, with maximal bilirubin 10-30 mg/dL reached in 2nd-3rd wk.
• Breastfeeding should be continued unless bilirubin level becomes > 20 mg/dL; Phototherapy may be of benefit.

32. A newborn presents with jaundice within 1st 24 hours. His mother's blood group is O +ve. Next line of management is?

A. Wait for serum bilirubin level before starting treatment
B. Phototherapy
C. Exchange transfusion
D. Stop breastfeeding

Ans B Any clinical jaundice appearing in the 1st 24 hours of life is always pathological, so the treatment (phototherapy) should start immediately without waiting for the reports. Exchange transfusion is not the answer here as the bilirubin level is not mentioned & the baby does not have any clinical evidence of bilirubin encephalopathy (previously called kernicterus), So, Phototherapy is the best answer here.

33. Which of the following is/are true regarding conjugated bilirubin in newborn?

A. Direct bilirubin fraction > 2 mg/dL at any time is pathological
B. Neonatal cholestasis is elevation of conjugated bilirubin beyond the 1st 14 days of life
C. Biliary atresia is cause of raised levels of conjugated bilirubin
D. Unconjugated hyperbilirubinemia can be physiologic
E. Acholuric stools indicates conjugated hyperbilirubi nemia

Ans A b. Neonatal cholestasis is elevation of conjugated bilirubin beyond the 1st 14 days of life, c. Biliary atresia is cause of raised levels of conjugated bilirubin, d. Unconjugated hyperbilirubinemia can be physiologic, e. Acholuric stools indicates conjugated hyperbilirubinemia; p 873-875;

34. For Double volume exchange transfusion, which of the following is to be used?

A. Plasma alone
B. Packed RBCs
C. RBCs in plasma
D. None of these

Ans C Fresh packed leukoreduced & irradiated O –ve RBCs reconstituted with AB group fresh-frozen plasma, whose cells are compatible with both infant's & mother's sera, is usually used for double volume exchange transfusion in Erythroblastosis fetalis

35. A 4-day-old newborn baby is having icterus involving sole; Approximate bilirubin level (mg/dl) is:

A. 12
B. 15
C. 8
D. 6262 Section 2: Neonatology

Ans B If palms & soles are stained yellow, bilirubin levels are 15-20 mg/dl or even more than that.

36. Cause of jaundice in day 1 of a baby are all except:

A. Rh incompatibility
B. ABO incompatibility
C. Prematurity
D. Breast milk jaundice

Ans D

37. Phototherapy is used in the treatment of?

A. Necrotising enterocolitis
B. Polycythemia
C. Neonatal hypocalcemia
D. Neonatal jaundice

Ans D Neonatal Jaundice Answers

38. Conjugated hyperbilirubinemia is seen in

A. Alpha1 AT deficiency
B. Cong hepatic fibrosis
C. Extrahepatic biliary atresia
D. Ductal plate defect
E. Crigler-Najjar syndrome 127. 1 month old newborn presents with conjugated Hyper- bilirubinemia, intrahepatic cholestasis and high alkaline phosphatase. Eosinophilic, PAS positive cytoplasmic granules are seen in liver biopsy specimen. Diagnosis is?

Ans A Rotors syndrome & b. Dubin Johnson syndrome

39. Which of the following characterizes pathological jaundice?

A. Jaundice on 3rd day
B. Jaundice upto the trunk
C. High colored urine
D. Jaundice accompanied by yellow stools

Ans C

40. Which of the following causes neonatal jaundice?

A. Sickle cell anemia
B. β–Thalassemia
C. Meningitis
D. G6PD deficiency
E. Rh incompatibility

Ans D and e. Rh incompatibility Sickle cell anemia & β thalassemia major usually becomes symptomatic beyond 3-6 months age (not in neonatal period)

41. Phototherapy is used in:

A. Neonatal jaundice
B. Neonatal meningitis
C. Neonatal tetanus
D. HIE

Ans A

42. All affect effectiveness of phototherapy except:

A. Skin pigmentation
B. Initial level of serum bilirubin
C. Spectral irradiance
D. Type of lamp used

Ans A Factors affecting the effectiveness of phototherapy are:
• Type of lamps used: LED lamps are better than fluorescent • Surface area of exposed skin
• Distance between the lights and the infant • Rate of hemolysis

43. A 32 weeks premature infant, 900 gm weight presents on the third day with jaundice. The serum bilirubin is 15 mg%. The treatment of choice is:

A. Exchange transfusion
B. Phototherapy
C. Wait and watch therapy
D. Pharmacologic therapy

Ans A

44. One gm of Hb liberates how many mg of bilirubin?

A. 40
B. 34
C. 15
D. 55

Ans B 34 Bilirubin is the end product of catabolism of hemoglobin; Hemoglobin (1 g) → Heme → Biliverdin → Bilirubin (34 mg)

45. In Rh Isoimmunization, exchange transfusion is indicated in all of the following except:

A. Cord blood hemoglobin < 10 g/dl
B. Cord bilirubin is more than 5 mg/dl
C. DCT positive & History of previous sibling affected
D. Hydrops fetalis

Ans D Indications of Exchange transfusion in a neonate with Rh incompatibility are:
• Cord hemoglobin < 10 g/dL or Cord bilirubin >5 mg/dl
• Direct coomb's test positive & H/o kernicterus or severe erythroblastosis in a sibling.

46. If Neonatal Jaundice appears for the first time in the 2nd week. Not a cause is:

A. Galactosemia
B. Rh Incompatibility
C. Hypothyroidism
D. Breast milk Jaundice

Ans B In Rh incompatibility, Jaundice usually presents at birth or within 24 hours of life

47. Medical management of kernicterus is:

A. Sulfonamide
B. Erythromycin
C. IVIg
D. Phenytoin

Ans C

48. Conjugated hyperbilirubinemia in infancy is seen in:

A. Gilbert syndrome
B. Crigler-Najjar syndrome
C. Dubin Johnson syndrome
D. Rotor syndrome
E. Neonatal hepatitis

Ans C d. Rotor syndrome & e. Neonatal hepatitis

49. A case of jaundice with 50% direct bilirubin, other LFTs normal. Diagnosis is:

A. Rotor syndrome
B. Gilbert syndrome
C. Glucuronyl transferase deficiency
D. Primary biliary cirrhosis

Ans A • Amongst the given options, Rotor syndrome and Primary biliary cirrhosis cause conjugated hyperbilirubinemia
• In Primary biliary cirrhosis, LFTs are abnormal.

50. The late features of kernicterus include all except:

A. Hypotonia
B. Sensorineural hearing loss
C. Choreoathetosis
D. Upward gaze palsy

Ans A Hypotonia is an early feature of acute bilirubin encephalopathy; For details

51. Unconjugated hyperbilirubinemia is seen in:

A. Physiological jaundice
B. Breast milk jaundice
C. Gilbert syndrome
D. Biliary atresia
E. Rotor syndrome

Ans A b. Breast milk jaundice, c. Gilbert syndrome

52. A term neonate with unconjugated hyperbilirubinemia of 18 mg/dl on 20th day. All are common causes except:

A. Breast milk jaundice (AIIMS May 07)
B. Congential cholangiopathy
C. G 6PD deficiency
D. Hypothyroidism

Ans B Ghai 8/e p 172-176 All options except 'b' are causes of unconjugated hyperbilirubinemia.

53. A 50-hour old full-term breast-fed newborn boy weighing 3100 g presents with clinically evident jaundice. Physical examination is otherwise normal. The total bilirubin is 11.0 mg/dl with a direct bilirubin of 0.4 mg/dl. What would be the correct treatment?

A. Continue breastfeeds and review after 24 hours
B. Stop breastfeeds and review after 24 hours
C. Continue breastfeeds and start blue-light phototherapy
D. Often become constipated

Ans A /e p 172-176 At 48-72 hrs of life, phototherapy cut off for a term neonate is total bilirubin > 18 mg/dl.

54. A newborn presented with jaundice. Most diagnostic investigation of choice is:

A. Ceftriaxone
B. Phenobarbitone
C. Ampicillin
D. Sulphonamide

Ans A Total and Direct bilirubin Measurement of total & direct bilirubin will help to classify jaundice into its respective type and aid in diagnosis. d. Sulphonamide Sulphonamides displace unconjugated bilirubin from albumin → ↑ level of free unconjugated bilirubin → ↑ risk of kernicterus

55. Hyperbilirubinemia in a child can be due to all of the following except:

A. Breast milk jaundice
B. Cystic fibrosis
C. Fanconi's syndrome
D. a-1 antitrypsin deficiency

Ans C

56. A term baby developed jaundice on 3rd day upto the thigh with normal stool and urine. Mother's blood group is 'O' –ve and that of babies 'A' +ve. The cause of jaundice is

A. Rh incompatibility
B. Physiological jaundice
C. Extrahepatic biliary atresia
D. Sepsis
E. Glucose-6 phosphate dehydrogenase deficiency

Ans A b. Physiological jaundice The given clinical scenario points towards unconjugated hyperbilirubinemia in the neonate, which is not very severe (upto thigh); so, both, physiological Jaundice & Rh incompatibility are possible causes.

57. Conjugated hyperbilirubinemia in infancy seen in:

A. Choledochal cyst
B. Extrahepatic biliary atresia
C. Crigler-Najjar disease
D. Gilbert disease
E. Rotor syndrome

Ans A b. Extrahepatic biliary atresia, e. Rotor syndrome

58. True about physiological Jaundice:

A. Jaundice appear within first 24 hour
B. Jaundice disappear in 3rd week
C. Sudden rise of bilirubin
D. Jaundice may be seen on day 5 of life
E. Breastfeeding should be stopped

Ans B and d. Jaundice may be seen on day 5 of life 879 Criteria for physiological jaundice
• Clinical jaundice appears after 24 hours of age & it is unconjugated hyperbilirubinemia
• Total bilirubin rises by less than 5 mg/dl per day (no sudden rise)
• Peak bilirubin occurs at 3–5 days of age, with a total bilirubin of no more than 15 mg/dl; (soles not stained)
• Clinical jaundice is usually resolved by 1 week in term & 2 weeks in preterm infants; so in 3rd week, there is no clinical jaundice. fo 152. d.

59. A child has total bilirubin of 4 mg/dl. Conjugated bilirubin and alkaline phosphatase are normal, bile salts and bile in urine are absent. However urobilinogen in urine is raised. What is the likely diagnosis?

A. Obstructive jaundice
B. Rotor's syndrome
C. Biliary cholestasis
D. Hemolytic jaundice

Ans D Normal conjugated bilirubin and alkaline phosphatase with absent bile in the urine but raised urobilinogen point to increased bilirubin production (hemolysis) rather than an obstructive process.

60. True about physiological jaundice in neonates:

A. Occurs in first 6 hours of delivery
B. Unconjugated hyperbilirubinemia
C. Neurological sequelae are common
D. Best treated by phototherapy
E. Starts on 2nd day of life NECROTIZING ENTEROCOLITIS

Ans B e. Starts on 2nd day of life

61. The following factors are associated with decreased risk of significant jaundice

A. gestational age ≥41 wk
B. **male gender**
C. exclusive bottle-feeding
D. black race
E. discharge from hospital after 72 hr

Ans B Male gender is a minor risk factor.

62. Jaundice, consisting of either indirect or direct bilirubin that is present at birth or appears within the 1st 24 hr of life may be due to all the following EXCEPT

A. erythroblastosis fetalis
B. **Crigler-Najjar syndrome**
C. concealed hemorrhage
D. sepsis
E. congenital infections

Ans B Jaundice that first appears on the 2nd or 3rd day is usually physiologic but may represent a more severe form. Familial nonhemolytic icterus (Crigler-Najjar syndrome) and early-onset breast-feeding jaundice are seen initially on the 2nd or 3rd day.

63. A search to determine the cause of jaundice should be made in all the following conditions EXCEPT

A. if it appears in the first 24-36 hr of life
B. if serum bilirubin is rising at a rate faster than 5 mg/dL/24 hr
C. if serum bilirubin is >12 mg/dL in a full-term infant
D. if jaundice persists after 10-14 days of life
E. **if direct bilirubin fraction is >1 mg/dL at any time**

Ans E Direct bilirubin fraction is >2 mg/dL at any time.

64. Regarding breast milk jaundice, the following are true EXCEPT

A. it develops in an estimated 2% of breast-fed term infants
B. maximal unconjugated bilirubin concentrations as high as 10-30 mg/dL reached during the 2nd-3rd week
C. jaundice may persist for 3-10 wk
D. phototherapy may be of benefit
E. **kernicterus never occur**

Ans E Although uncommon, kernicterus can occur in patients with breast milk jaundice.

65. The therapeutic effect of phototherapy depends on the following factors EXCEPT

A. distance between the lights and the infant
B. **skin color**
C. surface area of exposed skin
D. rate of hemolysis
E. in vivo metabolism and excretion of bilirubin

Ans B Dark skin does not reduce the efficacy of phototherapy.

66. The following factors suggest hemolytic disease as a cause of jaundice in the newborn EXCEPT

A. bilirubin rise of >0.5 mg/dL/h
B. **reticulocytosis >5% at birth**
C. onset of jaundice before 24 hours of age
D. significant decrease in hemoglobin
E. failure of phototherapy to lower serum bilirubin levels

Ans B Reticulocytosis (>8% at birth, >5% during first 2-3 days, >2% after first week).

Neonatal Sepsis

67. Not seen in small for date babies?

A. Hypoglycemia
B. Polycythemia
C. **Intracranial bleed**
D. Hypocalcemia

Ans C

68. Which is the most common cause of neonatal sepsis in India?

A. **Klebsiella pneumoniae**
B. Mycoplasma pneumonia
C. Group B Streptococcus
D. Listeria monocytogenes

Ans A Acinetobactes is not there in options. Klebsiella, the second most common organism is the answer here.

69. Most common cause of Neonatal sepsis in India:

A. **Acinetobacter**
B. Klebsiella
C. Staphylococcus
D. Streptococcus

Ans A As per the latest evidence, most common cause of Neonatal sepsis in India is Acinetobacter

70. The most common manifestation of late onset neonatal sepsis is:

A. **Meningitis**
B. Disseminated Intravascular coagulation
C. Pneumonia
D. Focal infections of bone and joints

Ans A

71. Sepsis screen includes all the following except?

A. Total leucocyte count
B. Absolute neutrophil count
C. **Band cell count**
D. IT ratio

Ans C

72. Most common cause of neonatal sepsis in India is:

A. Staphylococcus
B. Escherichia
C. Streptococcus
D. **Klebsiella**

Ans D Among the given options, the best ans is Klebsiella, but as per a recent multicentric study from India (Lancet, 2016), Most common organism responsible for Neonatal sepsis in India is Acinetobacter

73. Which one of the following statements is TRUE regarding neonatal sepsis?

A. Refusal to suck, poor cry and lethargy are common features
B. Neutrophilia is the hallmark of neonatal sepsis
C. Sepsis occurs on the 4th day and is mostly due to microbes from maternal genitalia
D. Antibiotics are withheld till the causative bacterium & drug sensitivity are known through culture-sensitivity testing

Ans A Discussing the options one by one, a. True; These are the earliest and very common features of neonatal sepsis b. False, Neutropenia & not neutrophilia is a feature of neonatal sepsis c. False, Sepsis can occur anytime in the neonatal period fo d. False, Empirical antibiotics should be started if neonatal sepsis is suspected, even before culture- sensitivity reports are available weiveR 17. a. Klebsiella

74. The most common cause of neonatal sepsis in India is:

A. Klebsiella
B. E. coli
C. Group B streptococci
D. Staph aureus

Ans A species are commonly cited as the leading cause of neonatal sepsis in Indian NICU studies, in contrast to Group B streptococcus, which predominates in Western countries.

75. Most common cause of neonatal sepsis in hospital in India is:

A. Streptococcus pyogenes
B. Streptococcus agalactiae
C. Peptostreptococci
D. Enterococcus faecium

Ans B Klebsiella

76. True about neonatal sepsis:

A. Meningitis commonly occur lately
B. Jaundice predisposes
C. Fever is a common feature
D. Jaundice is a common feature
E. Leukocytosis seen HYPOTHERMIA

Ans A and d. Jaundice is a common feature Hypothermia is more common than fever in neonatal sepsis; Leukopenia is seen in neonatal sepsis & not leukocytosis. HYPOTHERMIA

77. Agents that commonly cause nosocomialneonatal infection are

A. group B streptococci GBS
B. coagulase-negative staphylococci
C. proteus
D. gonococci
E. chlamydiae

Ans B Agents that commonly cause nosocomial infection are coagulase-negative staphylococci, gram-negative bacilli (E. coli, Klebsiella pneumoniae, Salmonella, Enterobacter, Citrobacter, Pseudomonas aeruginosa, Serratia), enterococci, S. aureus, and Candida. Viruses contributing to nosocomial neonatal infection include enteroviruses, CMV, hepatitis A, adenoviruses, influenza, respiratory syncytial virus (RSV), rhinovirus, parainfluenza, HSV, and rotavirus.

78. In a cohort of 6,215 VLBW infants in the NICHD Neonatal Research Network, gram- positive agents were associated with which percent of cases of late-onset sepsis

A. 90%
B. 70%
C. 50%
D. 30%
E. 10%

Ans B In a cohort of 6,215 VLBW infants in the NICHD Neonatal Research Network, gram-positive agents were associated with 70%, gram-negative with 18%, and fungi with 12% of cases of late-onset sepsis.

79. Neonatal sepsis case fatality rate is highest for

A. Staphylococcus—coagulase negative
B. Group B streptococcus
C. Escherichia coli
D. Pseudomonas
E. Candida albicans

Ans D Staphylococcus—coagulase negative (9.1%), Group B streptococcus (21.9%), Escherichia coli (34.0%), Pseudomonas (74.4%), and Candida albicans (43.9%).

Respiratory Distress Syndrome

80. Full term, small for date babies are at high risk of:

A. Hypoglycemia
B. IVH
C. BPD
D. Hyperthermia NEONATAL SEPSIS

Ans A

81. If APGAR score is 6, at 5 minutes of life, then what is the interpretation?

A. Child is normal
B. Moderate asphyxia
C. Severe birth asphyxia
D. It requires immediate resuscitation

Ans B Apgar score Interpretation 0-3 Severe birth asphyxia 4-7 Moderate birth asphyxia > 7 Normal

82. A 3 kg term baby delivered by caesarean section develops respiratory distress soon after birth. The liquor was meconium stained. Breathing rate is 90/minute; Which of the following are correct statements:

A. Transient tachypnea of newborn
B. Meconium aspiration syndrome
C. Reticulonodular shadows in X-ray chest
D. Surfactant production is defective
E. Oral feeding should be started immediately RESPIRATORY DISTRESS SYNDROME (RDS)

Ans A & b. Meconium aspiration syndrome , Meconium stained liquor with respiratory distress soon after birth suggests the diagnosis of meconium aspiration syndrome Transient tachypnea of newborn is also a possibility as the baby was delivered by caesarean section. In a baby with severe respiratory distress, oral feeding should be witheld till the respiratory distress improves; RESPIRATORY DISTRESS SYNDROME (RDS)

83. A preterm neonate with hyaline membrane disease needs to be treated with:

A. Dexamethasone
B. Betamethasone
C. Hydrocortisone
D. **Surfactant**

Ans D Systemic or inhaled corticosteroids have been used, mainly in those infants with RDS, who continue to require respiratory support & with in whom bronchopulmonary dysplasia (BPD) develops. Routine use of corticosteroids for the prevention or treatment of BPD is not recommended by the American Academy of Pediatrics.

84. Surfactant appear in amniotic fluid at:

A. 20 wk
B. **28 wk**
C. 32 wk
D. 4 wk

Ans B Surfactant synthesis begins in fetal lungs at 20 week & appears in amniotic fluid between 28-32 weeks.

85. Which of the following is NOT a feature of acute respiratory distress syndrome?

A. Interstitial inflammatory infiltrates
B. Interstitial edema
C. **Mucus plugs in the bronchioles**
D. Hyaline membranes lining the alveoli

Ans C

86. A 3.5 kg newborn born by full-term normal vaginal delivery presented with respiratory distress not responding to surfactant therapy. There is a history of previous sibling's death at one month of age due to respiratory distress. What diagnosis should you suspect?

A. **Pulmonary alveolar proteinosis**
B. Hyaline membrane disease
C. Meconium aspiration syndrome
D. Persistent pulmonary hypertension of newborn

Ans A Pulmonary alveolar proteinosis For details,

87. Respiratory distress syndrome–true is:

A. Not related to premature birth
B. L:S ratio >
C. Easily treatable and has low mortality
D. **Amniotic fluid phosphatidyl choline levels is lower than normal**

Ans D Respiratory distress syndrome is due to deficiency of surfactant, of which phosphatidyl choline is an important constituent.

88.
A term gestation newborn developed respiratory distress. Which of the following would favour respiratory distress syndrome (HMD)?

A. History of receiving antenatal corticosteroids
B. **Air bronchogram on chest X-ray**
C. Onset of distress after 6 hours of birth
D. Term birth

Ans B /e p 169-170 Chest X-ray findings in RDS include atelectasis (lung collapse), diffuse reticulogranular or reticulonodular pattern commonly referred to as "ground glass", presence of air bronchograms & complete "white out" (severe disease cases)

89. Surfactant is produced by:

A. **Type 2 pneumocytes**
B. Type 1 pneumocytes
C. Clara cells
D. Endothelium

Ans A

90. Hyaline membrane deposition is seen in:

A. ARDS
B. **RDS**
C. Pulmonary hemorrhage
D. Interstitial lung disease

Ans B

91. Hyaline membrane disease of lungs is characterized by:

A. **FRC is smaller than closing volume**
B. FRC is greater than closing volume
C. FRC is equal to closing volume
D. FRC is independent of closing volume

Ans A HMD reduces lung compliance & causes a fall in FRC below the normal closing volume of lung, so that the alveoli close.

92. Which of the following will favour the diagnosis of RDS in newborn:

A. Receipt of antenatal steroids
B. **Air bronchogram in chest X-ray**
C. Manifests after 6 hrs
D. Occurs after term gestation

Ans B Discussing about the options one by one, a. Receipt of antenatal steroids No Antenatal steroids protect against RDS b. Air bronchogram in chest X-ray Yes Air bronchogram on Chest X-ray is a feature of RDS c. Manifests after 6 hrs No RDS manifests soon after birth & not after 6 hours d. Occurs after term gestation No It usually occurs in preterm neonates

93. The dose of betamethasone in prenatal to prevent respiratory distress syndrome is:

A. Pulmonary edema
B. **Collapse of alveoli**
C. Elastic recoil of lungs
D. All

Ans B 12 mg every 24 hours • Dose of Inj Betamethasone is 12 mg IM 2 doses, 24 hrs apart
• Antenatal steroids should be given to all mother at risk of preterm delivery between 24 & 34 weeks of gestation. d. All Surfactant deficiency is the primary cause of RDS; Alveolar atelectasis, hyaline membrane formation & interstitial edema are seen in RDS.

94. A newborn weighing 1000 g is born at gestational age of 30 weeks with respiratory distress after 2–3 hrs of birth. What are the diagnostic possibilities:

A. **L/S ratio**
B. Bilirubin content of amniotic fluid
C. Ultrasound
D. X-ray
E. Pulmonary hemorrhage 87. Fetal lung maturity is assessed by: M

Ans A Diaphragmatic hernia, b. Congenital Bronchopulmonary cysts, d. HMD and e. Pulmonary hemorrhage 20/e p 850-857, Ghai 8/e p 168 Note: Bronchopulmonary dysplasia (BPD) occurs commonly in neonate of HMD who have been treated with O & mechanical ventilation & it usually takes weeks to develop. So, BPD does not present 2–3 hrs after birth.

95. Incidence of respiratory distress syndrome in a 28 weeks baby weighing 1500 g:

A. 10%
B. 20%
C. 40%
D. **80%**

Ans D Gestation age < 28 wk 32-36 wk > 37 wk Incidence of RDS 60-80% 15-30% Rare

96. With reference to RDS, all of the following statements are true except:

A. Usually occurs in infants born before 34 weeks of gestation
B. Is more common in babies born to diabetic mothers
C. Leads to cyanosis
D. **Is treated by administering 100% oxygen**

Ans D Discussing about the options, Option a. Yes, RDS is seen mainly in preterm neonates Option b. Yes, RDS is also common in babies born to diabetic mother due to delayed maturity of surfactant Option c. Yes, RDS can cause hypoxia & cyanosis Option d. CPAP ± Surfactant is the treatment of choice; Oxygen may be supplemented to treat hypoxia & titrated according to O saturation; 100% O is avoided to prevent Retinopathy of prematurity 2

97. A 26-year-old third gravida mother delivered a male baby weighing 4.2 kg at 37 weeks of gestation through an emergency caesarean section, for obstructed labour. The child developed respiratory distress one hour after birth. He was kept nil per orally (NPO) and given intravenous fluids. He maintained oxygen saturation on room air. No antibiotics were given. Chest radiograph revealed fluid in interlobar fissure. Respiratory distress settled by 24 hours of life. What is the most likely diagnosis?

A. **Transient tachypnea of the newborn**
B. Meconium aspiration syndrome
C. Persistent fetal circulation
D. Hyaline membrane disease

Ans A Respiratory distress, which resolves within 24 hours without any respiratory support and fluid in interlobar fissure on chest X-ray suggest the diagnosis of Transient tachypnea of newborn.

98. The risk for development of respiratory distress syndrome RDS increases with the following conditions EXCEPT

A. **prolonged rupture of membranes**
B. maternal diabetes
C. multiple births
D. precipitous delivery
E. cold stress

Ans A The risk of RDS is reduced in pregnancies with chronic or pregnancy-associated hypertension, maternal heroin use, prolonged rupture of membranes, and antenatal corticosteroid prophylaxis.

99. The following are true regarding respiratory distress syndrome (RDS) EXCEPT

A. signs of RDS usually appear within minutes of birth
B. **in most cases, the symptoms and signs reach a peak within 7days**
C. apnea and irregular respirations are ominous signs
D. improvement is often heralded by spontaneous diuresis
E. mixed respiratory-metabolic acidosis may be seen

Ans B In most cases, the symptoms and signs reach a peak within 3 days, after which improvement is gradual.

100. One of the following conditions can mimic RDS both clinically and radiographically

A. persistent pulmonary hypertension
B. meconium aspiration syndrome
C. **total anomalous pulmonary venous return**
D. pulmonary lymphangiectasia
E. lobar emphysema

Ans C In the differential diagnosis, early-onset sepsis may be indistinguishable from RDS. In pneumonia manifested at birth, the chest roentgenogram may be identical to that for RDS. Maternal group B streptococcal colonization, identification of organisms on gram staining of gastric or tracheal aspirates or a buffy coat smear, and/or the presence of marked neutropenia may suggest the diagnosis of early-onset sepsis.

101. **The basic defect requiring treatment in RDS is**

A. metabolic acidosis
B. circulatory insufficiency
C. hypothermia
D. **inadequate pulmonary exchange of oxygen and carbon dioxide**
E. electrolytes disturbance

Ans D The basic defect requiring treatment in RDS is inadequate pulmonary exchange of oxygen and carbon dioxide; metabolic acidosis and circulatory insufficiency are secondary manifestations.

102. **Prophylactic and rescue administrations of synthetic and natural surfactants have the following advantages EXCEPT**

A. reduces adverse outcomes
B. reduces neonatal mortality
C. decreses the risk for pneumothorax
D. decreses the risk for pulmonary interstitial emphysema
E. **reduces bronco-pulmonary dysplasia BPD rates**

Ans E The lack of reduction in BPD rates following surfactant replacement is probably, in part, due to the survival of infants with severe RDS who would have died without surfactant administration.

103. **Alkali therapy for the treatment of metabolic acidosis in RDS may result in all the following EXCEPT**

A. skin slough from infiltration
B. increased serum osmolarity
C. hypernatremia
D. hypocalcemia
E. **hyperkalemia**

Ans E Alkali therapy may result in skin slough from infiltration, increased serum osmolarity, hypernatremia, hypocalcemia, hypokalemia, and liver injury when concentrated solutions are administered rapidly through an umbilical vein catheter wedged in the liver.

104. **The normal cord at term is 55 cm long. Abnormally short cords are associated with the following EXCEPT**

A. fetal hypotonia
B. **wrapping around fetal parts**
C. uterine constraint
D. oligohydramnios
E. increased risk for complications of labor and delivery for both mother and infant

Ans B Long cords (>70 cm) increase risk for true knots, wrapping around fetal parts (neck, arm), and/or prolapse. Straight untwisted cords are associated with fetal distress, anomalies, and intrauterine fetal demise.

Transient Tachypnea & Wet Lung

105. **A forceps delivered premature baby develops fever, abnormal behavior, bulging fontanelle 5 days after birth. What is the likely causative organism?**

A. Neisseria meningitidis
B. **Listeria monocytogenes**
C. Staph aureus
D. Pneumococcus

Ans B Most common cause of Acute Bacterial Meningitis in: Age group Organism Neonates Gr B Streptococcus, E. coli, Listeria Infants and Children S. pneumoniae, N. meningitidis, H. influenzae Adolescents and Young Adults S. pneumoniae, N. meningitidis Neonatal Listeriosis Two clinical presentations are recognized: Early onset (<5 days) Late onset (≥5 days) Positive result of maternal listeria culture Negative results of maternal listeria culture Obstetric complications Uncomplicated pregnancy

106. **APGAR acronym stands for?**

A. Activity, pulse pressure, grimace, appearance, rate of respiration
B. Appearance, pressure, grimace, MAP, heart rate
C. Appearance, pressure, grimace, appearance, rate of heart beat
D. **Appearance, pulse, grimace, activity, respiration**

Ans D

107. **Lecithin/Sphingomyelin ratio is done to assess the Questions maturity of:**

A. **Lung**
B. Fetal circulation
C. Brain
D. Gonad

Ans A Lecithin/Sphingomyelin ratio of > 2 suggests lung surfactant maturity.

108. **Transient tachypnea of newborn (TTN) is commonly seen in which of the following situations?**

A. Term delivery requiring forceps
B. Term requiring ventouse
C. **Elective cesarean section**
D. Normal vaginal delivery APNEA OF NEWBORN

Ans C Delivery by caesarian section is a risk factor for Transient tachypnea of NewbornQ.

109. **Which of the following does not describe a “high risk" infant?**

A. **Third baby**
B. Birth weight less than 2.5 kg
C. Premature neonate
D. Twins

Ans A Factors that define an infant as being ‘high risk’: Demographic social factors Previous pregnancy Present pregnancy Maternal age <16 or >40 yr Intrauterine fetal demise Vaginal bleeding (abruptio, placenta previa) Illicit drug, alcohol, cigarette use Neonatal death Sexually transmitted ds (e.g. syphilis, HIV) Past medical history Prematurity Multiple gestation with Genetic disorders Intrauterine growth restriction Preeclampsia Diabetes mellitus Neonate Premature rupture of membranes

110. **A neonate at 48 hours of birth with a history of non- 184. passage of meconium. Next step in evaluation will be?**

A. Lower GI study
B. Manometry
C. Sweat chloride levels
D. CFTR mutation analysis

Ans A Lower GI study will give clue about underlying etiology like Hirschsprung disease, meconium ileus & other anorectal malformations.

111. **Low maternal serum α-fetoprotein (MSAFP) is associated with**

A. open neural tube defects
B. trisomy
C. gastroschisis
D. omphalocele
E. congenital nephrosis

Ans B Second-trimester screening (15-18 wk) of maternal serum α-fetoprotein (MSAFP) values is used to screen for open neural tube defects. About 90% of affected pregnancies can be detected by an elevated MSAFP value. Gastroschisis, omphalocele, congenital nephrosis, twins, and other abnormal conditions can also be identified. Low MSAFP is associated with incorrect gestational age estimates, trisomy 18 or 21, and intrauterine growth restriction.

112. **The following are true regarding apnea EXCEPT**

A. apnea is a common problem in preterm infants
B. in term infants, apnea is always worrisome
C. obstructive apnea is characterized by absence of airflow but persistent chest wall motion
D. serious apnea is defined as cessation of breathing for longer than 20 sec
E. bradycardia follows the apnea by 1-2 sec in more than 95% of cases and is most often nodal

Ans E Bradycardia follows the apnea by 1-2 sec in more than 95% of cases and is most often sinus, but on occasion it can be nodal. Vagal responses and, rarely, heart block are causes of bradycardia without apnea.

113. **One of the following is FALSE in the chest radiograph of transient tachypnea of the newborn (TTN)**

A. prominent pulmonary vascular markings
B. fluid in the intralobar fissures
C. overaeration
D. flat diaphragms
E. diffuse reticulogranular pattern

Ans E The distinctive features of transient tachypnea are rapid recovery of the infant and the absence of radiographic findings for RDS (hypoaeration, diffuse reticulogranular pattern, air bronchograms) and other lung disorders.

114. **One of the following definitions is FALSE**

A. brachydactyly=A condition of having short digits
B. postaxial polydactyly=Extra finger or toe present on the medial side of the hand or foot
C. clinodactyly=A medial or lateral curving of the fingers
D. camptodactyly=Permanent flexion of one or more fingers
E. syndactyly=Incomplete separation of the fingers

Ans B Postaxial polydactyly= Extra finger or toe present on the lateral side of the hand or foot. Preaxial polydactyly=Extra finger or toe present on the medial side of the hand or foot.

Meconium Aspiration

115. **What is the most common cause of Neonatal mortality in India?**

A. Severe anemia
B. Sepsis
C. Prematurity
D. Hypothermia

Ans C

116. **Effects of hypoxemic ischemia in a neonate include?**

A. Neurological damage
B. Subcutaneous fat necrosis
C. Pulmonary hypertension
D. Hyperglycemia
E. Hypercalcemia

Ans A b. Subcutaneous fat necrosis, c. Pulmonary hypertension
Effects of Asphyxia in a neonate:
• CNS: HIE, infarction, intracranial hemorrhage, seizures, tone abnormalities;
• Cardiovascular: Myocardial ischemia, poor contractility, hypotension
• Pulmonary: Pulmonary hypertension, pulmonary hemorrhage, RDS
• Renal: Acute tubular or cortical necrosis
• Adrenal: Adrenal hemorrhage
• Gastrointestinal: Perforation, ulceration with hemorrhage, necrosis

117. **An infant is born with meconium stained liquor. Which of the following is false regarding the management?**

A. Intrapartum suctioning is not recommended
B. Suctioning in vigorous and non-vigorous babies
C. Risk of cardiac arrhythmias in nasopharyngeal suctioning
D. Tracheal suctioning can be done using ET tube

Ans B As per the latest guidelines, suctioning of trachea is not required for all babies with Meconium stained liquor (

118. **A baby is born with meconium stained liquor which of the following is taken account of in terming a baby vigorous except:**

A. Tone
B. Colour
C. Heart Rate
D. Respiration

Ans B

119. **Meconium aspiration is done for 3 times but no breathing occurs. Next step in resuscitation would be:**

A. Chest compression
B. O2 inhalation
C. Bag and mask ventilation
D. Tickling of sole

Ans C

120. **A 3-week-old child presents with an abdominal mass. What is the most common cause of the presentation?**

A. Neuroblastoma
B. Wilms tumor
C. **Multicystic dysplastic kidney**
D. Distended bladder

Ans C Multicystic dysplastic kidney is the most common cause of an abdominal mass in the newborn, but the majority are nonpalpable at birth.

121. **Most common cause of abdominal mass in neonates is?**

A. Neuroblastoma
B. **Multicystic dysplastic kidney**
C. Polycystic kidney
D. Umbilical hernia

Ans B Renal masses account for the majority of neonatal abdominal masses, with multicystic dysplastic kidney classically cited as one of the most common causes.

122. **Administration of antenatal corticosteroids to women between 24 and 34 wk of gestation significantly reduces the following EXCEPT**

A. incidence and mortality of RDS
B. **postnatal growth**
C. the overall neonatal mortality
D. need for and duration of ventilatory support
E. incidence of severe intraventricular hemorrhage

Ans B is not adversely affected. Antenatal steroids do not increase the risk of maternal death, chorioamnionitis, or puerperal sepsis.

123. **The following are true regarding meconium aspiration syndrome (MAS) EXCEPT**

A. (MAS) develops in 5% of meconium-stained infants
B. 30% require mechanical ventilation
C. 3-5% die
D. overdistention of the chest may be prominent
E. **usually occurs in preterm or near-term infants**

Ans E Meconium-stained amniotic fluid is found in 10-15% of births and usually occurs in term or post-term infants.

124. **Intrauterine transplacental infections of significance to the fetus and/or newborn include the following EXCEPT**

A. **hepatitis B virus (HBV)**
B. rubella
C. toxoplasmosis
D. Parvovirus B19
E. varicella

Ans A Although HSV, HIV, hepatitis B virus (HBV), hepatitis C virus, and tuberculosis (TB) can each result in transplacental infection, the most common mode of transmission for these agents is intrapartum, during labor and delivery with passage through an infected birth canal (HIV, HSV, HBV), or postpartum, from contact with an infected mother or caretaker (TB) or with infected breast milk (HIV).

Persistent Pulmonary Hypertension

125. **A 6-day-old neonate weighing 2800 gm (Birth weight 3200 gm), was brought with c/o fever, poor feeding and poor activity. No history of vomiting or diarrhea. Axillary temperature was 39oC, there was depressed fontanelle, sunken eyes, decreased urine output and decreased skin turgor. Her mother has a history of decreased milk production. What is your diagnosis?**

A. **Neonatal sepsis**
B. Galactosemia
C. Dehydration fever
D. Acute renal failure

Ans A In the given scenario, the baby has excessive weight loss (> 10% of birth weight), features of dehydration (like depressed fontanelle, sunken eyes, decreased urine output and decreased skin turgor) along with poor feeding & poor activity. Failure to feed properly is seen in most sick newborn infants and should lead to a careful search for infection (sepsis), a central or peripheral nervous system disorder, intestinal obstruction, and other abnormal conditions.

126. **What is the interpretation of Apgar score of 3?**

A. Moderately depressed baby
B. **Severely depressed baby**
C. Normal
D. Mild birth asphyxia

Ans B Apgar score of 0-3 indicates a severely depressed neonate. with

127. **Which one of the following medical disorder in the mother leads to delayed fetal lung maturity:**

A. Heart disease
B. **Diabetes**
C. Thalassemia minor
D. Epilepsy

Ans B

128. **Infants of diabetic mother manifests which of the following?**

A. Hyperglycemia
B. **Hypoglycemia**
C. Hypocalcemia
D. Increased fetal defect
E. Hyperbilirubinemia

Ans B c. Hypocalcemia, d. Increased fetal defect, e. Hyperbilirubinemia

129. **Which is not a cause of non-immune hydrops fetalis:**

A. a-thalassemia
B. Congenital rubella
C. **Rh incompatibility**
D. Cystic hygroma

Ans C Rh incompatibility is a cause of immune hydrops, not non-immune

130. **Non-immune fetal hydrops caused by all except:**

A. Parvovirus B19 infection
B. **Rh incompatibility**
C. Congenital heart block

Ans B Causes of Hydrops fetalis dna System involved Underlying cause Immune Rh incompatibilityQ Anemia α-thalassemia, G-6PD deficiency Cardiac Arrhythmias: Supraventricular tachycardia, AF, Congenital heart blockQ Structural: Hypoplastic left heart, endocardial cushion defect, Cardiomyopathy Vascular Twin-twin transfusionQ, thrombosis of renal or Umblical vein Lymphatic Lymphangiectasia, Cystic HygromaQ CNS Encephalocele, Intracranial hemorrhage Thoracic Mediastinal teratoma, Diaphragmatic hernia

131. **A delay in fetal pulmonary maturation may be associated with**

A. **hydrops fetalis**
B. severe premature separation of the placenta
C. premature rupture of the fetal membranes
D. narcotic addiction
E. maternal hypertensive and renal vascular disease

Ans A Earlier lung maturation may occur in the presence of severe premature separation of the placenta, premature rupture of the fetal membranes, narcotic addiction, or maternal hypertensive and renal vascular disease. A delay in pulmonary maturation may be associated with hydrops fetalis or maternal diabetes without vascular disease.

132. **Regarding CPAP, the following are true EXCEPT**

A. its prevents collapse of surfactant-deficient alveoli
B. **its indicated, if oxygen saturation cannot be kept > 95%**
C. its improves functional residual capacity FRC
D. its improves ventilation-perfusion matching
E. its reduces ventilatory needs

Ans B Warm humidified oxygen should be provided at a concentration initially sufficient to keep arterial oxygen pressure between 50 and 70 mm Hg (91-95% saturation) in order to maintain normal tissue oxygenation while minimizing the risk of oxygen toxicity. If oxygen saturation cannot be kept >90% at inspired oxygen concentrations of 40-70% or greater, applying CPAP at a pressure of 5-10 cm H2O via nasal prongs is indicated and usually produces a rapid improvement in oxygenation.

133. **The following are predisposing factors for persistent pulmonary hypertension of the newborn (PPHN) EXCEPT**

A. **anemia**
B. meconium aspiration syndrome
C. early-onset sepsis
D. hypoglycemia
E. birth asphyxia

Ans A Persistent pulmonary hypertension of the newborn (PPHN) occurs in term and post-term infants. Predisposing factors include birth asphyxia, MAS, early-onset sepsis, RDS, hypoglycemia, polycythemia, maternal use of nonsteroidal anti-inflammatory drugs with in utero constriction of the ductus arteriosus, maternal late trimester use of selective serotonin reuptake inhibitors, and pulmonary hypoplasia due to diaphragmatic hernia, amniotic fluid leak, oligohydramnios, or pleural effusions. PPHN is often idiopathic.

134. **One of the following is FALSE regarding intrapartum antibiotics**

A. reduce vertical transmission of GBS
B. lessen neonatal morbidity after preterm rupture of membranes
C. prevent perinatal transmission of GBS
D. **reduce the rates of late-onset GBS disease**
E. has no effect on the rates of infection with non-GBS pathogens

Ans D Intrapartum chemoprophylaxis does not reduce the rates of late-onset GBS disease and has no effect on the rates of infection with non-GBS pathogens. Of concern is a possible increase in gram-negative infections (especially E. coli) in VLBW and possibly term infants in spite of a reduction in early GBS sepsis by intrapartum antibiotics.

Neonatal Convulsions

135. **Which of the following streptococci cause neonatal meningitis?**

A. **S. agalactiae**
B. S. pyogenes
C. S. bovis
D. S. mutans

Ans A Group B Streptococcus (S. agalactiae) cause neonatal sepsis & meningitis.

136. **Apgar score < 3 at 5 minutes of life is a predictor of:**

A. Poor neurological outcome
B. **Increased risk of neonatal mortality**
C. Risk of cerebral palsy
D. Neonatal depression

Ans B Apgar score is normal in most patients in whom cerebral palsy subsequently develops. Low Apgar scores (0-3 at 5 min) is a predictor of neonatal death, in both term and preterm neonates

137. **The type of neonatal seizures with worst prognosis is?**

A. Subtle
B. Focal clonic
C. Atonic
D. **Myoclonic**

Ans D

138. **Commonest type of seizure in newborn is?**

A. Clonic
B. Tonic
C. **Subtle**
D. Myoclonic

Ans C • Neonatal Seizures • Type with best prognosis: Focal clonic seizures
• Most common type is Subtle seizures • Neonatal seizures with worst prognosis: Myoclonic seizures
• Least common type is Myoclonic seizures • CNS imaging of choice: Transcranial ultrasound
• Most common cause is Hypoxia • Drug of choice: Phenobarbitone a. Transcranial ultrasound

139.A 2-day-old neonate in the neonatal ICU develops seizures. Which of the following would be the best investigation for the child:

A. Transcranial ultrasound
B. Skull X-ray
C. CT scan
D. MRI

Ans A Cranial (transcranial) ultrasound is the preferred first-line bedside investigation for a neonate with seizures, as it is readily available, avoids radiation, and can detect hemorrhage or major structural lesions.

140.Best prognosis of neonatal seizures is in?

A. Myoclonic
B. Tonic clonic
C. Focal clonic
D. Opsoclonus

Ans C

141. A preterm infant with poor respiration at birth starts throwing seizures at 10 hours after birth. Antiepileptic of choice shall be:

A. Levetiracetam
B. Phenytoin
C. Phenobarbitone
D. Lorazepam

Ans C

142.Which vitamin deficiency is responsible for neonatal seizure:

A. Pyridoxine
B. Vitamin C
C. Thiamine
D. Cobalanum

Ans A Pyridoxine deficiency is one of the causes of refractory seizures in the neonatal period & infancy.

143.Most common cause of seizure in newborn is:

A. Hypoxia induced ischemic encephalopathy
B. Hypocalcemia
C. Metabolic abnormality
D. Sepsis

Ans A

144.Commonest type of seizure in newborn:

A. Clonic
B. Tonic
C. Subtle
D. Myoclonic

Ans C

145.Jitteriness can be distinguished from seizures by all of the following except:

A. Sensitivity to stimulus
B. Frequency of movement
C. Abnormality of gaze
D. Autonomic disturbance MECONIUM ASPIRATION SYNDROME

Ans B Differences between jitteriness & seizures: Feature Jitteriness Seizures Ocular deviation Not seen Ocular deviation seen Stimulus sensitive Yes, It stops with passive movement of limb It is not stimulus sensitive Autonomic changes No autonomic changes seen Usually seen

146.Infant of diabetic mother with weight 3.8 Kg presented with seizures after 16 hours of birth. What is the most probable cause?

A. Hypoglycemia
B. Hypocalcemia
C. Birth asphyxia
D. Intraventricular hemorrhage

Ans A Hypoglycemia is seen in 25–50% of Infants of diabetic mothers & 15–25% of infants of mother with gestational diabetes Hyperinsulinism is the main cause of hypoglycemia, which manifests as jitteriness, tremulousness & hyperexcitablity during the 1st 3 days after birth, although hypotonia, lethargy, seizures and poor sucking may also occur: Early appearance of these signs is more likely to be related to hypoglycemia, and their later appearance to hypocalcemia

147. Seizures beginning in the delivery room or shortly thereafter may be due to

A. hypoxic-ischemic encephalopathy
B. unintentional injection of maternal local anesthetic into the fetus
C. intracranial hemorrhage
D. cerebral anomaly
E. hypoglycemia

Ans B Seizures beginning in the delivery room or shortly thereafter may be due to the unintentional injection of maternal local anesthetic into the fetus. Convulsions may also result from hyponatremia and water intoxication in the infant after the administration of large amounts of hypotonic fluid to the mother shortly before and during delivery.

Intraventricular Hemorrhage

148.The most common fungal infection in the neonates transmitted by caregiver's hands is?

A. Candida albicans
B. Candida tropicalis
D. Candida parapsilosis

Ans D

149.APGAR stands for?

A. Appearance, Pressure, Grimace, Activity, respiration
B. Appearance, Pulse, Grimace, Activity, Respiration
C. Appearance, Pressure, Grimace, Activity, Respiration
D. Awareness, Pulse, Grimace, Activity, Respiration

Ans B APGAR score is based on 5 parameters: Appearance, Pulse, Grimace, Activity, Respiration

150.Nile blue sulfatase test is to detect fetal:
A. TAPVC
B. Meconium aspiration
C. Neonatal pulmonary alveolar proteinosis
D. **Difuse herpes simplex infection**

Ans D Lung maturity

151. All of the following are the complications in the new born of a diabetic mother except:
A. Hyperbilirubinemia
B. **Hyperglycemia**
C. Hypocalcemia
D. Hypomagnesemia

Ans B

152.Which one of the following does NOT present with a large anterior fontanelle?
A. **Crouzon syndrome**
B. Hypophosphatasia
C. Achondroplasia
D. Congenital rubella syndrome

Ans A Crouzon syndrome is a cause of craniosynostosis (premature fusion of sutures), so the fontanelle are small & close early.

153.Which sinus will be absent in a newborn?
A. **Frontal sinus**
B. Maxillary
C. Anterior ethmoidal
D. Posterior ethmoidal 187.

Ans A High Yielding Points about Development of Paranasal sinuses:
• Ethmoidal and maxillary sinuses are present at birth, but only the ethmoidal sinuses are pneumatized.
• Sphenoidal sinuses are present by 5 yr of age
• Frontal sinuses begin development at age 7-8 yr and are not completely developed until adolescence.

154.True umbilical cord knots are seen in approximately 1% of births and are associated with the following conditions EXCEPT
A. **short cord**
B. small fetal size
C. polyhydramnios
D. monoamniotic twinning
E. fetal demise

Ans A s and noncoiled cords occur with chromosome abnormalities and omphalocele.

155.Regarding intraventricular hemorrhage (IVH) of prematurity, the following are true
A. it usually develops spontaneously
B. **MRI is the preferred imaging technique for screening IVH**
C. prophylactic administration of low-dose indomethacin reduces the incidence of severe IVH
D. majority of patients with IVH have no clinical symptoms
E. it may rarely manifest at birth

Ans B Ultrasonography is the preferred imaging technique for screening because it is noninvasive, portable, reproducible, and sensitive and specific for detection of IVH.

156.One of the following is FALSE in hemolytic disease of the newborn caused by Rh incompatibility
A. infant is Rh Positive (D, sometimes C)
B. **never occur in first-born infant**
C. maternal antibody titers can help predict the severity of fetal disease
D. severe anemia is frequent
E. stillbirth/hydrops is frequent

Ans B 5% of first-born infants may be affected.

157. Osteitis may be a feature of one of the following transplacental infections
A. cytomegalovirus
B. herpes simplex virus
C. varicella-zoster virus
D. **rubella**
E. T. gondii

Ans D Osteitis may be a feature of rubella and T. pallidum.

158.When screening for intraventricular hemorrhage IVH, the best time to perform an ultrasound is
A. first day of life
B. second day of life
C. third day of life
D. **fourth day of life**
E. fifth day of life

Ans D In a series of infants studied by ultrasonography, approximately 50% had the onset of hemorrhage on the first day of life, 25% on the second day, and 15% on the third day. Thus, a single scan on the fourth day of life would be expected to detect >90% of IVHs.

Periventricular Leukomalacia

159.Neonatal meningitis is caused by all except:
A. **N. meningitidis**
B. E. coli
C. Listeria
D. Group B streptococci

Ans A

160.What is the 1 minute APGAR score of a baby who is gasping, completely pale, is lying totally limp & showing no movements, has heart rate 40/min and grimaces on doing oropharyngeal suction?
A. 0
B. 1
C. 2
D. **3**

Ans D 3 Appearance: completely pale, so score of 0, Pulse rate 40/minute → score 1, Grimaces → 1, Activity → 0 (as no movements), Respiratory effort → 1 (as gasping); So, the APGAR score for this baby is: 0 + 1 + 1 + 0 + 1 =

161. **A 1.5 kg child born at 32 weeks by LSCS presents with moderate respiratory difficulty (RR 70/minutes). Which of the following is the most appropriate management:**

A. CPAP
B. Mechanical ventilation
C. Warm oxygen
D. Surfactant and ventilation

Ans A In at-risk neonates for respiratory distress syndrome, CPAP could be started at the earliest sign of respiratory distress; If one of the options would have been surfactant or surfactant with CPAP, that would have been a better choice... but the option in this question is surfactant & ventilation, that is why D is not the correct ans..! about CPAP (Continuous positive airway pressure)

162. **Which of the following malformation in a newborn is specific for maternal insulin dependent diabetes mellitus?**

A. Transposition of great arteries
B. Caudal regression
C. Holoprosencephaly
D. Meningmyelocele

Ans B

163. **A neonate while suckling milk can respire without difficulty due to**

A. Stout soft palate
B. High larynx
C. Small tongue
D. Small arytenoides

Ans B

164. **Best screening test to evaluate hearing in a neonate:**

A. Neonatal sepsis
B. Cerebral palsy
C. Neonatal tetanus
D. Hypoxic ischemic encephalopathy Chapter 12: Diseases of Newborn 265 Answers

Ans D Brainstem evoked auditory response Screening Tests for hearing evaluation in a neonate: Name of the method Characteristics Clinical Evaluation Not reliable Transient Evoked Otoacoustic Emissions Records acoustic feedback from cochlea following a click stimulus It evaluates the functioning of peripheral auditory system Unreliable due to false positive results & lack of gestation related norms Auditory Brainstem Responses It can be used in neonates after 34 weeks of post conceptional maturity

165. **Regarding multiple gestation pregnancies, the following are true EXCEPT**

A. the reported incidence of spontaneous twinning is lowest in the Asian races
B. triplets are estimated to occur in 1 in 862 pregnancies in the USA
C. the incidence of monozygotic twins is unaffected by racial or familial factors
D. the overall incidence of multifetal gestation is unchanged
E. polyovular pregnancies are more frequent beyond the 2nd pregnancy

Ans D Although the incidence of spontaneous multifetal gestation has been stable over the years, the overall incidence of multifetal gestation is increasing as a result of treatment of infertility with ovarian stimulants (clomiphene, gonadotropins) and in vitro fertilization. Twins account for about 2.5% of births but about 20% of very low birthweight (VLBW) infants.

166. **Vitamin A supplementation given largely to infants < 1,000 g resulted in all the following EXCEPT**

A. a decrease in death
B. a decrease in bronco-pulmonary dysplasia BPD at 36 wk
C. less nosocomial sepsis
D. less retinopathy of prematurity
E. decreases the need for extracorporeal membrane oxygenation (ECMO)

Ans E Inhaled nitric oxide (iNO) decreases the need for extracorporeal membrane oxygenation (ECMO) in term and near-term infants with hypoxic respiratory failure or persistent pulmonary hypertension of the neonate.

167. **Regarding hemorrhagic disease of the newborn, all the following are true EXCEPT**

A. age of onset is usually in 2-7 days old infants
B. more frequent in breast-fed than in formula-fed infants
C. bleeding time is normal
D. oral vitamin K is as effective as intramuscular route in the prevention and treatment of the disease
E. prothrombin time (PT), and partial thromboplastin time (PTT) are prolonged

Ans D Although oral vitamin K (birth, discharge, 3-4 wk: 1-2 mg) has been suggested as an alternative, oral vitamin K is less effective in preventing the late onset of bleeding due to vitamin K deficiency and thus cannot be recommended for routine therapy. The intramuscular route remains the method of choice.

168. **Intracranial calcification may be a feature of one of the following transplacental infections**

A. cytomegalovirus
B. herpes simplex virus
C. varicella-zoster virus
D. syphilis
E. rubella

Ans A Intracranial calcification may be a feature of CMV, HIV, toxoplasmosis, and T. cruzi.

Neonatal Hypoglycemia

169. **A newborn loses maximum heat from:**

A. Head
B. Abdomen
C. Palms & soles
D. Neck

Ans A

170. **What is the APGAR core for a baby with HR 120/ min, RR-40/min, strong respiratory efforts, peripheral cyanosis, grimace while suctioning and having slightly flexed posture but no active movements of limbs?**

A. 6
B. 7
C. 8
D. 9

Ans B 7 Appearance: peripheral cyanosis, so score of 1, Pulse rate 120/minute → score 2, Grimaces → 1, Activity → 1 (flexed posture, but no movements), Respiratory effort → 2 (as strong respiratory efforts present); So, the APGAR score for this baby is: 1 + 2 + 1 + 1 + 2 =

171. Ventilator settings are FiO of 70, and rate of 50/ 2 minute. Next step to be taken should be?

A. Increase rate to 60 per minute
B. **Increase FiO to 80**
C. Continue ventilation with the same settings
D. Start weaning off ventilator TRANSIENT TACHYPNEA OF NEWBORN

Ans B Diabetes

172. After the delivery of an infant of diabetic mother, glucose of the infant was 60 mg/dL. Which other investigation does the sister expects that the physician would ask her to do?

A. Serum potassium
B. CBC
C. **Serum calcium**
D. Serum chloride

Ans C with
• Hypoglycemia seen in 25-50% of infants of diabetic mothers.
• The nadir of blood glucose is usually reached at 1-3 hr;
• Presents with jitteriness, tremulousness & hyperexcitability during the 1st 3 days after birth • Early appearance of these signs is more likely to be related to hypoglycemia, and their later appearance to hypocalcemia; these abnormalities may also occur together.

173. The cut-off for neonatal hypoglycemia is blood glucose less than _____ mg/dl:

A. 30
B. 35
C. 45
D. **40**

Ans D

174. Hypoglycemia in an infant is defined as blood glucose level less than:

A. **40 mg/dl**
B. 60 mg/dl
C. 10 mg/dl
D. 20 mg/dl

Ans A

175. Administration of glucose solution is prescribed for all of the following situations except:

A. **A normal term neonates**
B. Infant of a diabetic mother
C. History of unconsciousness
D. History of hypoglycemia

Ans A Discussing the options one by one, a. In a normal term neonate exclusive breastfeeding is usually sufficient to prevent hypoglycemia. Administration of glucose or dextrose is not routinely required for all neonates b. These babies are prone to develop hypoglycemia; may require glucose administration, if symptomatic c. Hypoglycemia can be one of the underlying causes for unconsciousness; so glucose administration may be required d.

176. A 4 kg baby born to a diabetic mother was found to have hypoglycemia on routine blood glucose monitoring. Which of the following is to be done?

A. Reassess the baby again after 2 hours
B. Give 10% dextrose IV
C. **Start oral feeding**
D. Give injection insulin
E. Give injection glucagon

Ans C Feeding is the initial treatment for hypoglycemia in infant of diabetic mother

177. All of the following groups of newborns are at an increased risk of hypoglycemia except:

A. Birth asphyxia (AIIMS Nov 02)
B. Respiratory distress syndrome
C. Maternal diabetes
D. **Postterm infant NEONATAL HYPERBILIRUBINEMIA Questions**

Ans D Post term infant Common causes of Hypoglycemia in newborns: Inadequate substrate Hyperinsulinism
• Prematurity • Respiratory distress syndrome • Infant of diabetic Mother • Perinatal asphyxia
• SGA • Infant of toxemic mother • Infant with erythroblastosis fetalis

178. Hyperglycemia in Neonate is blood glucose above?

A. 150 mg/dL
B. **125 mg/dL**
C. 180 mg/dL
D. 100 mg/dL

Ans B Although the definition of Neonatal hyperglycemia varies, a blood glucose concentration greater than 125 mg/dL (6.9 mmol/L) or a plasma or serum glucose concentration greater than 150 mg/dL (8.3 mmol/L) is often used.

Neonatal Hypocalcemia

179. The range of environmental temperature at which the BMR (Basal Metabolic rate) of the baby is at a minimum, oxygen consumption is least and the baby maintains its normal body temperature is called as:

A. **Thermoneutral environment**
B. Thermal comfort zone
C. Ambient environmental temperature
D. Ideal thermal environment

Ans A

180. APGAR score -include all except:

A. 0
B. **1**
C. 2
D. 3

Ans B Respiratory rate Respiratory effort is included in Apgar score & not respiratory rate; For details,

181. **Respiratory rate per minute in a 1 month old baby, to label it tachypnea should be more than:**

A. 40
B. 50
C. **60**
D. 70

Ans C

182. **Infants of diabetic mother have which of the following:**

A. **Macrosomia**
B. Neural tube defect
C. Hyperglycemia
D. Hypocalcemia
E. Hypoglycemia

Ans A b. Neural tube defect and d. Hypocalcemia, e. Hypoglycemia

183. **Normal level of Hb in a newborn (at 3 days):**

A. **16.5 g/dL**
B. 18.5 g/dL
C. 11 g/dL
D. 10 g/dL

Ans A

184. **Major cause of neonatal mortality in full-term newborn is**

A. respiratory distress syndrome
B. necrotizing enterocolitis
C. bronchopulmonary dysplasia (BPD)
D. **congenital anomalies**
E. intraventricular hemorrhage

Ans D Severe immaturity, respiratory distress syndrome, intraventricular hemorrhage, infection, necrotizing enterocolitis, and bronchopulmonary dysplasia (BPD) are major causes of mortality in preterms.

185. **One of the following is a common neonatal respiratory problem associated with premature infants**

A. bronchopulmonary dysplasia
B. **apnea**
C. congenital pneumonia
D. pneumothorax
E. pneumomediastinum

Ans B Respiratory distress syndrome (hyaline membrane disease) and apnea are common neonatal respiratory problem associated with premature infants.

186. **Contributary factors for the development of broncho-pulmonary dysplasia (BPD) include the following EXCEPT**

A. immaturity
B. **dehydration during the 1st days of life**
C. chorioamnionitis
D. symptomatic PDA
E. malnutrition

Ans B Overhydration during the 1st days of life may also contribute to the development of BPD. Vitamin A supplementation (5,000 IU intramuscularly 3 times/wk for 4 wk) in VLBW infants reduces the risk of BPD.

187. **Apt test is useful for diagnosis of**

A. disseminated intravascular coagulopathy
B. **swallowed blood syndrome**
C. hemorrhagic disease of the newborn
D. neonatal thrombocytopenic purpura
E. congenital deficiency of factor VII

Ans B Apt devised the following test for differentiation of fetal from maternal blood: (1) Rinse a blood-stained diaper or some grossly bloody (red) stool with a suitable amount of water to obtain a distinctly pink supernatant hemoglobin solution; (2) centrifuge the mixture and decant the supernatant solution; (3) add 1 part of 0.25 N (1%) sodium hydroxide to 5 parts of the supernatant fluid. Within 1-2 min, a color reaction takes place: A yellow-brown color indicates that the blood is maternal in origin;

188. **Limb hypoplasia may be a feature of the following transplacental infections**

A. cytomegalovirus
B. herpes simplex virus
C. **varicella-zoster virus**
E. rubella

Ans C

189. **The following are manifestations of hypocalcemia in the neonate EXCEPT**

A. jitteriness
B. seizures
C. **carpopedal spasm**
D. high-pitched cry
E. laryngospasm

Ans C Chvostek's sign (facial muscle twitching on tapping), and Trousseau's sign (carpopedal spasm) may be present, but more commonly these are absent during the neonatal period.

Necrotizing Enterocolitis

190. **What is the body temperature range to categorise a neonate as having cold stress?**

A. 35.4 to 36.0 degoC
B. 33.4 to 34. degoC
C. 34.4 to 35.4 degoC
D. **36 to 36.4 degoC**

Ans D 36 to 36.4 deg C

191. **All the following factors affect APGAR score except:**

A. Prematurity
B. Maternal drugs
C. Neurological condition of the newborn
D. **Mode of delivery**

Ans D A low Apgar score may be the result of fetal distress, prematurity & drugs given to the mother during labor.

192. **Characteristic radiological feature of transient tachypnoea of newborn is:**

A. Reticulogranular appearance
B. Low volume lungs
C. **Prominent horizontal fissure**
D. Air bronchogram

Ans C Chest X-ray in transient tachypnea of newborn shows prominent pulmonary vascular markings, fluid in the interlobar fissure & prominent inter lobar fissure; Occasionally pleural fluid is also seen.

193. **Abnormality in Crigler-Najjar syndrome:**

A. Uptake of bilirubin
B. **Defect in hepatic conjugation**
C. Canalicular Release
D. Reduced bile excretion

Ans B Crigler-Najjar syndrome is due to glucuronyl transferase deficiency → defective conjugation of bilirubin

194. **Indication for surgery in Necrotizing Enterocolitis is?**

A. Pneumatosis intestinalis
B. **Pneumoperitoneum**
C. Portal vein gas
D. Peritonitis

Ans B

195. **A very preterm baby on 30 mL/kg of enteral feeding developed sudden severe abdominal distension with visible bowel loops on day 6 of life. The baby also showed temperature instability and lethargy. X-ray of the abdomen showed portal venous gas. The staging of NEC is?**

A. I
B. 2a
C. **2b**
D. 3a

Ans C 2b Stage of NEC Radiological feature I Normal/mild intestinal dilatation II IIa: Pneumatosis intestinalis; IIb: Portal vein gas III IIIa: Peritonitis / Ascites; IIIb: Pneumoperitoneum

196. **One of the following drugs may cause pyloric stenosis if administered to a premature infant**

A. intravenous vitamin E
B. indomethacin
C. enteric gentamicin
D. **prostaglandins**
E. dexamethasone

Ans D and erythromycin may cause pyloric stenosis if administered to a premature infant.

197. **Methylxanthines for the treatment of broncho-pulmonary dysplasia (BPD) have the following effects EXCEPT**

A. increase respiratory drive
B. decrease apnea
C. improve diaphragmatic contractility
D. **increase pulmonary vascular resistance PVR**
E. diuretic effects

Ans D Methylxanthines may also decrease PVR and increase lung compliance in infants with BPD, probably through direct smooth muscle relaxation.

198. **All the following are true, regarding pathology of neonatal necrotizing enterocolitis (NEC) EXCEPT**

A. in fatal cases, gangrene may extend from the stomach to the rectum
B. the greatest risk factor for NEC is prematurity
C. **in most situations, a pathogen is identified**
D. NEC is much less common in infants fed human milk
E. NEC in term infants is often a "secondary" disease

Ans C Clustering of cases suggests a primary role for an infectious agent. Various bacterial and viral agents, including Escherichia coli, Klebsiella, Clostridium perfringens, Staphylococcus epidermidis, astrovirus, norovirus, and rotavirus, have been recovered from cultures. Nonetheless, in most situations, no pathogen is identified.

199. **Absolute indication for surgery in neonatal necrotizing enterocolitis (NEC) include**

A. **positive result of abdominal paracentesis**
B. failure of medical management
C. a single fixed bowel loop on radiographs
D. abdominal wall erythema
E. a palpable mass

Ans A A surgeon should be consulted early in the course of treatment. Indications for surgery include evidence of perforation on abdominal roentgenograms (pneumoperitoneum) or positive result of abdominal paracentesis (stool or organism on Gram stain preparation from peritoneal fluid). Failure of medical management, a single fixed bowel loop on radiographs, abdominal wall erythema, and a palpable mass are relative indications for exploratory laparotomy.

200. **The MOST important risk factor for necrotizing enterocolitis (NEC) in preterm infants is**

A. delivery by cesarean section
B. breast feeding
C. apgar score
D. exposure to glucocorticoids during the first week of life
E. **gestational age and birth weight**

Ans E In an analysis of 15,072 neonates born at 98 centers over a 2-year period, the most important variables associated with NEC were gestational age and birth weight.

Neonatal Anemia & Polycythemia

201. **What should be the temperature of the delivery room for the neonate to be kept in warmer?**

A. 37-39°C
B. 34-36°C
C. **22-26°C**
D. 30-32°C

Ans C 22-26°C

202. **Failure to initiate and maintain spontaneous respiration following birth is clinically known as:**

A. **Birth asphyxia**
B. RDS—Respiratory distress syndrome
C. Respiratory failure
D. Pulmonary edema

Ans A

203.In a neonate, cessation of breathing for 10 second with bradycardia is:

A. Apnea
B. Dyspnea
C. Cheyne Stokes respiration
D. None

Ans A Apnea in a newborn is defined as: cessation of respiration for 20 seconds with or without bradycardia and cyanosis or cessation of respiration for less than 20 seconds if it is associated with bradycardia or cyanosis.

204.Peripheral smear of neonate with ABO incompatibility will show:

A. Microspherocytes
B. Elliptocytes
C. Fragmented RBCs
D. Polychromasia

Ans A about ABO incompatibility
• ABO incompatibility is the most common cause of hemolytic disease of the newborn
• Usually, the mother is type O and the infant is type A or B; most commonly seen in babies with A, blood group
• Peripheral smear in these patients show Microspherocytes, Reticulocytosis & Polychromasia Note: Polychromasia may be seen but is not very prominent in ABO incompatibility, as the hemolysis is usually mild, so the compensatory erythroid response from the bone marrow too would be mild.

205.Monochorionic-Monoamniotic twin occurs if division occurs?

A. < 24 hrs
B. 1-4 days
C. 4-8 days
D. > 8 days

Ans D Hacker & Moore's Essentials of Obstetrics and Gynecology The relationship between the timing of cleavage & nature of membranes in twin gestations: Time interval between ovulation and cleavage of egg Nature of membranes 0-72 hr Diamniotic, dichorionic 4-8 days Diamniotic, monochorionic 9-12 days Monoamniotic, monochorionic

206.Generalized edema of the newborn may occur in the following conditions EXCEPT

A. prematurity
B. nonimmune hydrops
C. Turner syndrome
D. congenital nephrosis
E. Hurler syndrome

Ans C Localized edema suggests a congenital malformation of the lymphatic system; when confined to one or more extremities of a female infant; it may be the initial sign of Turner syndrome.

207.The following factors pose a risk for poor academic performance of premature infants EXCEPT

A. birthweight below 750 g
B. periventricular leukomalacia
C. antenatal exposure to magnesium sulfate
D. bronchopulmonary dysplasia
E. posthemorrhagic hydrocephalus

Ans C may have neuroprotective effects and may reduce the incidence of cerebral palsy in high-risk neonates.

208.Associated anomalies have been reported in up to 30% of congenital diaphragmatic hernia cases; these include the following EXCEPT

A. pulmonary hypoplasia
B. CNS lesions
C. esophageal atresia
D. omphalocele
E. cardiovascular lesions

Ans A and malrotation of the intestine are part of the lesion, not associated anomalies.

209.One of the following is FALSE in plethora in the newborn infant (polycythemia)

A. polycythemia is defined as a central Hct of 65% or higher
B. peripheral (heelstick) Hct and Coulter counter results values are higher than central values
C. the incidence of neonatal polycythemia is 8% in small for gestational age (SGA) infants
D. many affected infants are asymptomatic
E. neonatal RBCs have decreased deformability and filterability, which predispose to stasis in the microcirculation

Ans B Peripheral (heelstick) Hct values are higher than central values, whereas Coulter counter results are lower than Hct values determined by microcentrifugation.

210.Common respiratory tract manifestations of neonatal bacterial infections is

A. ethmoiditis
B. otitis media
C. mastoiditis
D. retropharyngeal cellulitis
E. empyema

Ans E Pneumonia and empyema are common respiratory tract manifestations of neonatal bacterial infections.

Neonatal Thrombocytopenia

211. Brown fat is absent in?

A. Scapula
B. Mesentery
C. Subcutaneous tissue
D. Adrenal cortex

Ans B Care of the Newborn by David E. Hertz, pg 43
• Neonates are incapable of shivering & rely on non-shivering thermogenesis, due to brown fat for heat.
• Brown fat is located around great vessels, kidneys, adrenals, axillae, nape of neck, & between scapulae.

212.Meconium passage in utero leads to which of the following:

A. Listeriosis
B. Obstructive emphysema
C. Pathological jaundice
D. Meconium ileus

Ans B

213.Neonatal apnea is seen in all except:

A. Prematurity
B. **Hyperglycemia**
C. Hypoglycemia
D. Hypocalcemia
E. Hypothermia NEONATAL HYPOGLYCEMIA & INFANT OF DIABETIC MOTHER

Ans B For details about neonatal apnea, NEONATAL HYPOGLYCEMIA & INFANT OF DIABETIC MOTHER Answers

214.Bronze baby syndrome is caused by:

A. **Phototherapy**
B. Phenobarbitone
C. Hemosiderosis
D. Chloramphenicol

Ans A Bronze baby syndrome: is a dark, grayish brown skin discoloration mainly seen in infants with elevated conjugated bilirubin, as an adverse effect of phototherapy.

215.Two substances are needed for a developing fetus to grow male genitalia?

A. Wolffian factor and Mullerian inhibitory factor
B. **Testosterone and Mullerian inhibitory factor**
C. Wolffian factor and testosterone
D. Mullerian factor and testosterone

Ans B Development of genitalia in males:
• SRY gene produces SRY protein that directs the development of gonad into testes
• Male development can only occur when fetal testis secretes Anti-Müllerian hormone (AMH) & testosterone
• Anti-Müllerian hormone causes the paramesonephric ducts to regress
• Testosterone converts the mesonephric ducts into male accessory structures, including the epididymis, vas deferens, and seminal vesicle & also controls the descent of the testes.

216.Soft areas in the occipital region suggest the irregular calcification and wormian bone formation usually associated with the following conditions EXCEPT

A. osteogenesis imperfecta
B. **craniosynostosis**
C. cleidocranial dysostosis
D. cretinism
E. Down syndrome

Ans B Persistently small fontanels suggest microcephaly, craniosynostosis, and congenital hyperthyroidism.

217. Sudden onset of hypotension in a very low birthweight (VLBW) infant suggests

A. **pneumothorax**
B. necrotizing enterocolitis
C. bacterial sepsis
D. myocarditis
E. hypoglycemia

Ans A Sudden onset of hypotension in a very low birthweight (VLBW) infant suggests pneumothorax, intraventricular hemorrhage, or subcapsular hepatic hematoma.

218.Congenital diaphragmatic hernia (CDH) can be diagnosed on prenatal ultrasonography (between 16 and 24 wk of gestation) in > 50% of cases. Findings on ultrasonography may include the following EXCEPT

A. chest mass
B. mediastinal shift
C. gastric bubble
D. **olighydramnios**
E. a liver in the thoracic cavity

Ans D Polyhydramnios

219.Surgical repair of umbilical hernia is advised in the following conditions EXCEPT

A. if hernia persists to the age of 4-5 yr
B. causes symptoms
C. **hernias that appear before the age of 6 mo**
D. becomes strangulated
E. becomes progressively larger after the age of 1-2 yr

Ans C Most umbilical hernias that appear before the age of 6 mo disappear spontaneously by 1 yr of age. Even large hernias (5-6 cm in all dimensions) have been known to disappear spontaneously by 5-6 yr of age.

220.Common skin and soft tissue manifestations of neonatal bacterial infections are

A. facial cellulitis
B. scalp abscess
C. fasciitis
D. **breast abscess**
E. omphalitis

Ans D and impetigo are common skin and soft tissue manifestations of neonatal bacterial infections.

Patent Ductus Arteriosus

221.Temperature of NICU is:

A. 20–22°C
B. **22–26°C**
C. 26–30°C
D. 30–35°C

Ans B 22–26°C The nursery temperature should be kept between 22-26oC (72-78°F).

222.Calculate the Silverman score for a neonate with paradoxical breathing, marked chest retractions, but minimal nasal flare and expiratory grunt audible only with stethoscope (video given in exam):

A. 4
B. 5
C. 7
D. **8**

Ans D Ref: Silverman scoring is calculated using the following 5 parameters, each of which can be given a score of 0, 1, 2. In the given scenario, score for paradoxical breathing (2), marked lower chest & xiphisternal retractions (2+ 2), minimal nasal flare (1), expiratory grunt audible only with stethoscope (1), give a total score of 8. Score > 6 indicate impending respiratory failure.

223.What is the most probable diagnosis in this neonate shown below?

A. Infant of diabetic mother
B. Beckwith Wiedemann syndrome
C. Congenital hypothyroidism
D. IUGR baby

Ans A Large for date baby with hairy pinna suggests a diagnosis of Infant of diabetic mother.

224.Gas in portal veins is seen in

A. Necrotizing enterocolitis
B. Clostridium sepsis
C. Gas gangrene
D. Septicemia

Ans A

225.Most common type of phagus in twins?

A. Amino acid
B. Carbohydrate
C. Lipid
D. Minerals

Ans C Thorachophagus Conjoined twins (Also called Siamese twins) Result from relatively late monovular separation; Types depending on the site of connections are: thoraco-omphalopagus (28% of conjoined twins), thoracopagus (18%), omphalopagus (10%), craniopagus (6%) & incomplete duplication (10%)

226.The following disorders are associated with a large anterior fontanel EXCEPT

A. congenital rubella syndrome
B. hypophosphatasia
C. Apert syndrome
D. Russell-Silver syndrome
E. vitamin A deficiency

Ans E Vitamin D deficiency rickets is associated with a large anterior fontanel.

227.Vomiting in the neonatal period is usually due to

A. pyloric stenosis
B. milk allergy
C. overfeeding
D. stress ulcer
E. an inborn error of metabolism

Ans C Vomiting during the 1st day of life suggests obstruction in the upper digestive tract or increased intracranial pressure. Roentgenographic studies are indicated when obstruction is suspected. Vomiting may also be a nonspecific symptom of an illness such as septicemia.

228.A poor prognostic sign of congenital diaphragmatic hernia (CDH) is

A. grunting
B. use of accessory muscles
C. early respiratory distress, within 6 hr of life
D. cyanosis
E. scaphoid abdomen

Ans C is thought to be a poor prognostic sign.

229.Generalized edema may be seen in the neonatal period with the following conditions EXCEPT

A. congenital nephrosis
B. Turner syndrome
C. Hurler syndrome
D. hydrops fetalis
E. offspring of diabetic mothers

Ans B Persistent edema of 1 or more extremities may represent congenital lymphedema (Milroy disease) or, in females, Turner syndrome.

230.One of the following is not included in the definition of the systemic inflammatory response (SIRS) in neonates and pediatric patients

A. temperature instability
B. abnormal white blood cell (WBC) count
C. respiratory dysfunction
D. cardiac dysfunction
E. perfusion abnormalities

Ans B or an increase in immature forms is included in the definition of the systemic inflammatory response SIRS in adults.

Neonatal Shock

231.All of the following can occur in a neonate for heat production except:

A. Shivering
B. Breakdown of brown fat with adrenaline secretion
C. Universal flexion like a fetus
D. Cutaneous vasoconstriction

Ans A

232.A baby is born prematurely at 29 weeks gestation by caesarean delivery done for fetal distress. Neonate develops tachypnea, flaring, subcostal and intercostal retractions immediately after birth. Chest radiography shows bilateral, diffuse, ground glass appearance, air bronchograms and poor lung expansion. What is the best treatment plan in the present case?

A. Oxygen therapy
B. Surfactant administration and respiratory support
C. Emergency surgical repair of tracheoesophageal fistula
D. Only observation at this point

Ans B The given scenario indicates a diagnosis of moderate – severe respiratory distress syndrome. So, the treatment of choice is: surfactant administration and respiratory support

233.Infant of diabetic mother have the following except:

A. Macrosomia
B. Neural tube defect
C. Hyperglycemia
D. Hypocalcemia

Ans C

234.A preterm baby presents with feed intolerance, abdominal distension and bloody stool on day 4 of life. Most likely diagnosis is:

A. Intususception
B. **Necrotising enterocolitis Questions**
C. Volvulus
D. Hirschsprung disease MISCELLANEOUS

Ans B Necrotising enterocolitis MISCELLANEOUS

235.A neonate presents with bilious vomiting. What is the first investigation?

A. Chest X-ray
B. **Baby –gram**
C. CT
D. USG

Ans B Ghai 8/e Baby –gram: An X-ray of the entire body of an infant; To look for the underlying cause of obstruction, this is the best initial investigation, with the radiation exposure much less than a CT scan. It is also called an 'Infantogram'

236.Leukokoria (white pupillary reflex) in newborn infant suggests the following disorders EXCEPT

A. cataracts
B. tumor
C. **congenital glaucoma**
D. chorioretinitis
E. retinopathy of prematurity

Ans C A cornea >1cm in diameter in a term infant (with photophobia and tearing) suggests congenital glaucoma and requires prompt ophthalmologic consultation.

237.Regarding cephalohematoma, all the following are true EXCEPT

A. is a subperiosteal hemorrhage
B. **it may extend across the midline and across suture lines**
C. occur in 1-2% of live births
D. no discoloration of the overlying scalp
E. an underlying skull fracture may be associated with 10-25% of cases

Ans B Cephalohematoma is a subperiosteal hemorrhage, hence always limited to the surface of one cranial bone, while caput succedaneum is a diffuse, sometimes ecchymotic, edematous swelling of the soft tissues of the scalp involving the area presenting during vertex delivery. It may extend across the midline and across suture lines.

238.Eventration of the diaphragm may be associated with the following EXCEPT

A. **pulmonary hypoplasia**
B. pulmonary sequestration
C. congenital heart disease
D. chromosomal trisomies
E. recurrent infections

Ans A Congenital eventration may affect lung development, but it has not been associated with pulmonary hypoplasia.

239.Diabetic mothers have a high incidence of all the following EXCEPT

A. **oligohydramnios**
B. preeclampsia
C. pyelonephritis
D. preterm labor
E. chronic hypertension

Ans A Diabetic mothers have a high incidence of polyhydramnios, preeclampsia, pyelonephritis, preterm labor, and chronic hypertension; their fetal mortality rate is greater than that of nondiabetic mothers, especially after 32 wk of gestation. Fetal loss throughout pregnancy is associated with poorly controlled maternal diabetes (especially ketoacidosis) and congenital anomalies.

240.Persistence of the umbilical cord beyond which time should prompt consideration of an underlying abnormality

A. 10 days
B. 20 days
C. **30 days**
D. 40 days
E. 50 days

Ans C The umbilical cord generally dries up and sloughs by 2 weeks of life. Delayed separation can be normal up to 45 days. However, because neutrophilic and/or monocytic infiltration appears to play a major role in autodigestion, persistence of the cord beyond 30 days should prompt consideration of an underlying functional abnormality of neutrophils (leukocyte adhesion deficiency) or neutropenia.

Neonatal Dermatology

241.The different manifestations of hypothermia are:

A. **Apnea**
B. Hypoglycemia
C. Hyperglycemia
D. Tachycardia
E. Hypoxia

Ans A b. Hypoglycemia, e. Hypoxia

242.A 3 hour old preterm baby develops respiratory grunt and X-ray shows reticulonodular pattern. Diagnosis is:

A. **Hyaline membrane disease**
B. Transient tachypnea of newborn
C. Meconium aspiration syndrome
D. All of above

Ans A

243.Which is the most common congenital abnormality in a baby of diabetic women?

A. **Ventricular Septal defect**
B. Sacral agenesis
C. Meningomyelocele
D. Anencephaly\

Ans A Ventricular Septal defect For details,

244.Craniopagus is defined as fusion of:

A. Head and spine
B. Head only
C. Thorax and spine
D. Thorax only

Ans B Conjoined twins (Siamese twins—incidence 1/50,000) Result from relatively late monovular separation. Prognosis for conjoined twins depends on the extent to which vital organs are shared. Site of connections varies:
• Thoraco-omphalopagus (28% of conjoined twins),
• Thoracopagus (18%)
• Omphalopagus (10%)
• Craniopagus (6%): joined at head

245.A single umbilical artery in a newborn infant increases the risk for

A. meningomyelocele
B. occult renal anomaly
C. omphalocele
D. gastroschisis
E. omphalitis

Ans B A single umbilical artery increases the risk for an occult renal anomaly.

246.Early postnatal exposure to dexamethasone, within the 1st wk of life, in VLBW infants, is associated with the following EXCEPT

A. metabolic derangements
B. poor growth
C. increased risk for sepsis
D. increased risk of spontaneous bowel perforation
E. cerebral palsy

Ans E Infants exposed to postnatal steroids after the 1st wk of life have an increased risk of cerebral palsy and developmental delay.

247.Early ultrasonographic signs of hydrops include

A. double–bowel wall sign (bowel edema)
B. polyhydramnios
C. ascites
D. pleural effusions
E. scalp edema

Ans A Early ultrasonographic signs of hydrops include organomegaly (liver, spleen, heart), the double–bowel wall sign (bowel edema), and placental thickening. Progression to polyhydramnios, ascites, pleural or pericardial effusions, and skin or scalp edema may then follow.

248.In Infants of diabetic mothers, the following are true EXCEPT

A. hypoglycemia develops in about 25-50% of infants of diabetic mothers
B. increased weight of the placenta and all infant organs
C. heart failure occurs in 5-10% of infants of diabetic mothers
D. the incidence of congenital anomalies is increased threefold in infants of diabetic mothers
E. lumbosacral agenesis is one of most common congenital anomalies

Ans B The probable pathogenic sequence is that maternal hyperglycemia causes fetal hyperglycemia, and the fetal pancreatic response leads to fetal hyperinsulinemia; fetal hyperinsulinemia and hyperglycemia then cause increased hepatic glucose uptake and glycogen synthesis, accelerated lipogenesis, and augmented protein synthesis.

249.Pneumatosis intestinalis is pathognomonic for

A. Hirschsprung's disease
B. necrotizing enterocolitis (NEC)
C. pseudomembranous enterocolitis
D. neonatal ulcerative colitis
E. meconium ileus

Ans E Pneumatosis intestinalis can be seen in various other conditions, including Hirschsprung's disease, pseudomembranous enterocolitis, neonatal ulcerative colitis, and ischemic bowel disease. However, it is a characteristic finding in 85% of patients with NEC. Dark, concentric rings within the bowel wall represent hydrogen as a byproduct of bacterial metabolism

13

CHAPTER 13

Pediatric Cardiology

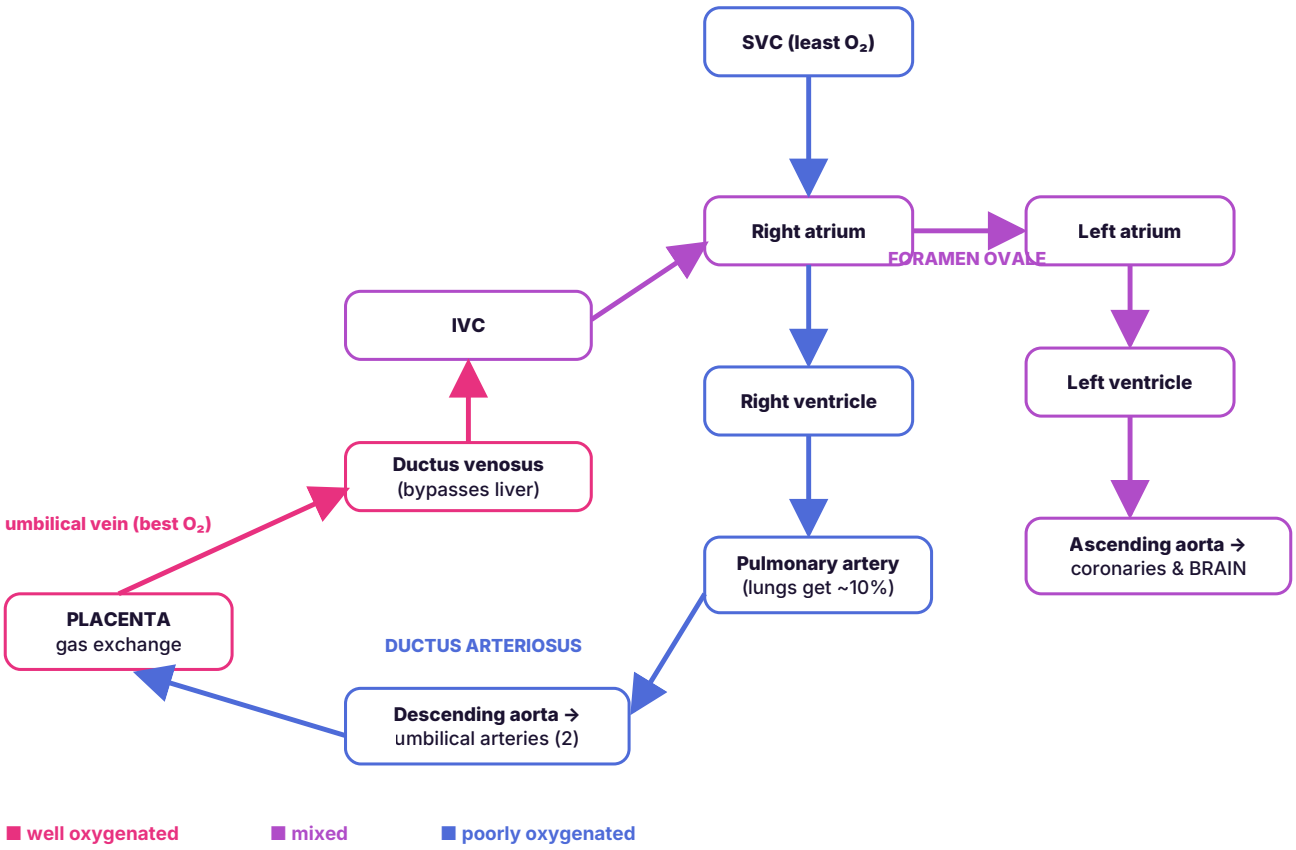


WHAT THIS CHAPTER COVERS

- Fetal & Neonatal Circulation
- Congenital Heart Disease - General
- Acyanotic CHD (L-R Shunts)
- Cyanotic CHD (R-L Shunts)
- ASD, VSD & PDA
- Tetralogy of Fallot
- Transposition of Great Arteries
- Coarctation of Aorta
- Pulmonary Stenosis & Atresia
- Heart Failure
- Infective Endocarditis
- Rheumatic Fever & RHD
- Myocarditis & Cardiomyopathy
- Arrhythmias
- Hypertension
- Kawasaki Disease
- Pericardial Diseases
- Cardiac Examination & Murmur

212 MCQs **18** subtopics

13.1 Fetal & Transitional Circulation



Fetal circulation – three shunts and the oxygen pathway

- **Placenta = organ of gas exchange**; **umbilical vein** carries the **most oxygenated blood (PO_2 ~30–35 mmHg, SpO_2 ~80%)**.
- **Three shunts**:
  1. **Ductus venosus** – umbilical vein → IVC (bypasses liver) → closes functionally within minutes–hours, anatomically by **2–3 weeks** → **ligamentum venosum**.
  2. **Foramen ovale** – IVC blood (oxygenated) directed by **crista dividens/Eustachian valve** → RA → LA → LV → **ascending aorta → coronaries & brain (best-oxygenated blood)**; functional closure at birth (LA pressure > RA), anatomical by ~3 months–1 y (patent in 25% adults) → **fossa ovalis**.
  3. **Ductus arteriosus** – pulmonary artery → descending aorta (**90% of RV output bypasses lungs**); kept open by **low PO_2 & PGE_2**; **functional closure 12–24 h (by 72 h in term)**, anatomical 2–3 weeks → **ligamentum arteriosum**.
- **Umbilical arteries** (2) → **medial umbilical ligaments**; umbilical vein → **ligamentum teres**.
- **Least oxygenated blood**: SVC; most – umbilical vein > IVC (ductus venosus) > LV/ascending aorta.
- **Fetal cardiac output ~450 mL/kg/min (combined)**, RV dominant (RV > LV, ~55–60%); neonatal CO ~350 mL/kg/min.
- At birth: lung expansion → **↓ PVR**, clamping cord → **↑ SVR** → LA pressure ↑ → foramen ovale closes; ↑ PO_2 & ↓ PGE_2 → ductus constricts.

KEEPING THE DUCTUS OPEN / CLOSING IT

Open (duct-dependent lesions): PGE_1 (alprostadil) 0.05–0.1 µg/kg/min – side effect **apnoea (MC)**, fever, flushing, cortical hyperostosis. **Close (preterm PDA): indomethacin, ibuprofen (preferred – fewer renal effects), paracetamol** (prostaglandin synthesis inhibitors); surgical ligation/device. **NSAIDs ineffective in term PDA.**

Duct-dependent lesions

Duct-dependent pulmonary circulation (cyanosis worsens as duct closes)	Duct-dependent systemic circulation (shock as duct closes)	Mixing lesion
Pulmonary atresia, critical PS, tricuspid atresia, severe TOF, Ebstein	**HLHS, critical coarctation, interrupted aortic arch, critical AS**	**TGA with intact septum** (also needs atrial septostomy)

13.2 Congenital Heart Disease – General

- Incidence **~8–10 per 1000 live births**; **MC CHD overall – VSD** (bicuspid aortic valve is commonest if included); **MC cyanotic CHD – TOF** (beyond infancy); **MC cyanotic CHD presenting in the newborn period – TGA**.
- Aetiology: multifactorial (90%); chromosomal (Down, Turner, 22q11), maternal (**diabetes, rubella, SLE, PKU, alcohol, lithium, phenytoin, retinoids, valproate**).

Syndromes & their typical heart lesions

Condition	Lesion
Down syndrome	**AVSD (endocardial cushion defect) – most characteristic**; VSD, ASD, TOF
Turner	**Bicuspid aortic valve (MC)**, **coarctation**
Noonan	**Pulmonary stenosis (dysplastic)**, HCM
Williams	**Supravalvular aortic stenosis**, peripheral PS
DiGeorge (22q11)	**Interrupted aortic arch type B, truncus arteriosus**, TOF
Marfan	**MVP, aortic root dilatation**
Congenital rubella	**PDA, peripheral pulmonary artery stenosis**
Infant of diabetic mother	**VSD (MC), TGA (most specific), HCM**
Holt–Oram	**ASD (secundum)** + upper-limb defects
Ellis–van Creveld	**Common atrium** (single atrium)
Edwards (T18) / Patau (T13)	VSD, PDA / ASD, VSD
Alagille	**Peripheral pulmonary stenosis**
Friedreich ataxia	HCM
Maternal lithium	**Ebstein anomaly**
Maternal SLE (anti-Ro)	**Congenital complete heart block**
Tuberous sclerosis	**Cardiac rhabdomyoma**
Kartagener	Dextrocardia/situs inversus

Classification (with pulmonary blood flow)

	Increased PBF	Normal / decreased PBF
Acyanotic	**L→R shunts: VSD, ASD, PDA, AVSD**, PAPVC	**Obstructive: PS, AS, CoA**
Cyanotic	**TGA, TAPVC, truncus arteriosus, single ventricle, HLHS** ("admixture lesions")	**TOF, tricuspid atresia, pulmonary atresia, Ebstein**, critical PS

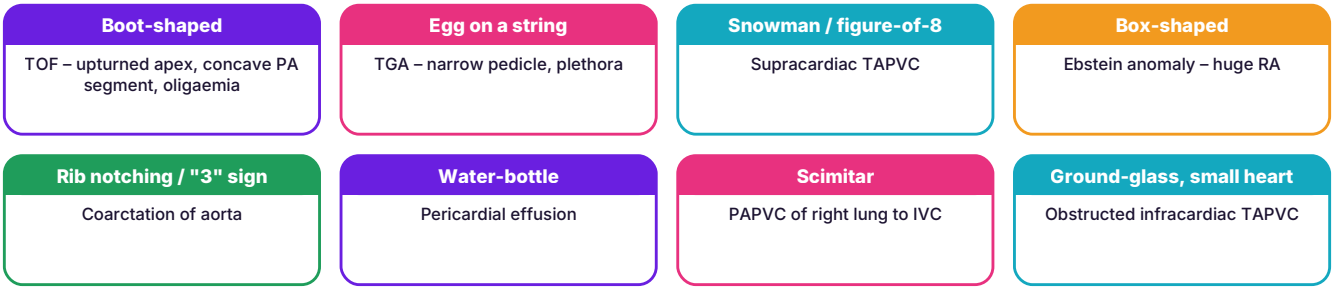
5 T'S OF CYANOTIC CHD (+ HLHS, EBSTEIN)

Tetralogy of Fallot, **T**ransposition (TGA), **T**runcus arteriosus, **T**ricuspid atresia, **T**otal anomalous pulmonary venous connection (TAPVC).

Approach clues

- **Hyperoxia (nitrogen washout) test**: 100% O_2 for 10 min → **PaO_2 > 150–200 mmHg → lung disease**; **< 100 (esp. < 50) → cyanotic CHD**.
- **Differential cyanosis** (upper pink, lower blue) → **PPHN or preductal CoA/interrupted arch with PDA (R→L)**. **Reverse differential cyanosis** (upper blue, lower pink) → **TGA with PDA + PPHN/CoA**.
- **Heart failure in first week** → obstructive left-sided (HLHS, critical AS/CoA), **large AV fistula (vein of Galen)**, arrhythmia; **L→R shunts present at 6–8 weeks** (when PVR falls).
- **Eisenmenger syndrome**: long-standing L→R shunt → irreversible pulmonary vascular disease → shunt reversal (R→L) → cyanosis, clubbing; **loud P2**, murmur disappears; earliest in **AVSD/large VSD/PDA (1–2 y)**, ASD (adulthood). **Inoperable** (heart-lung transplant). **PDA with Eisenmenger – differential cyanosis (toes blue & clubbed, fingers spared)**.

Chest X-ray signs



Classic cardiac silhouettes on chest X-ray

Sign	Lesion
Boot-shaped heart (coeur en sabot) – uplifted apex, concave pulmonary bay, oligaemic lungs	**TOF**
Egg on a string / egg on side – narrow superior mediastinum	**TGA**
Snowman / figure-of-8	**Supracardiac TAPVC**
Box-shaped / "wall-to-wall" heart, oligaemia	**Ebstein anomaly**
Rib notching (3rd–8th ribs), "3 sign" (reverse 3/E sign on barium)	**Coarctation of aorta** (> 5–8 y)
Water-bottle / flask-shaped heart	**Pericardial effusion**
Scimitar sign	**Scimitar syndrome – PAPVC of right lung to IVC**
Sitting duck heart	Persistent truncus arteriosus
Ground-glass lungs with normal heart size	**Obstructed (infracardiac) TAPVC**
"Shmoo" heart, LV prominence	AR/LV enlargement
Jug-handle appearance	Primary pulmonary hypertension

13.3 Acyanotic CHD – Left-to-Right Shunts

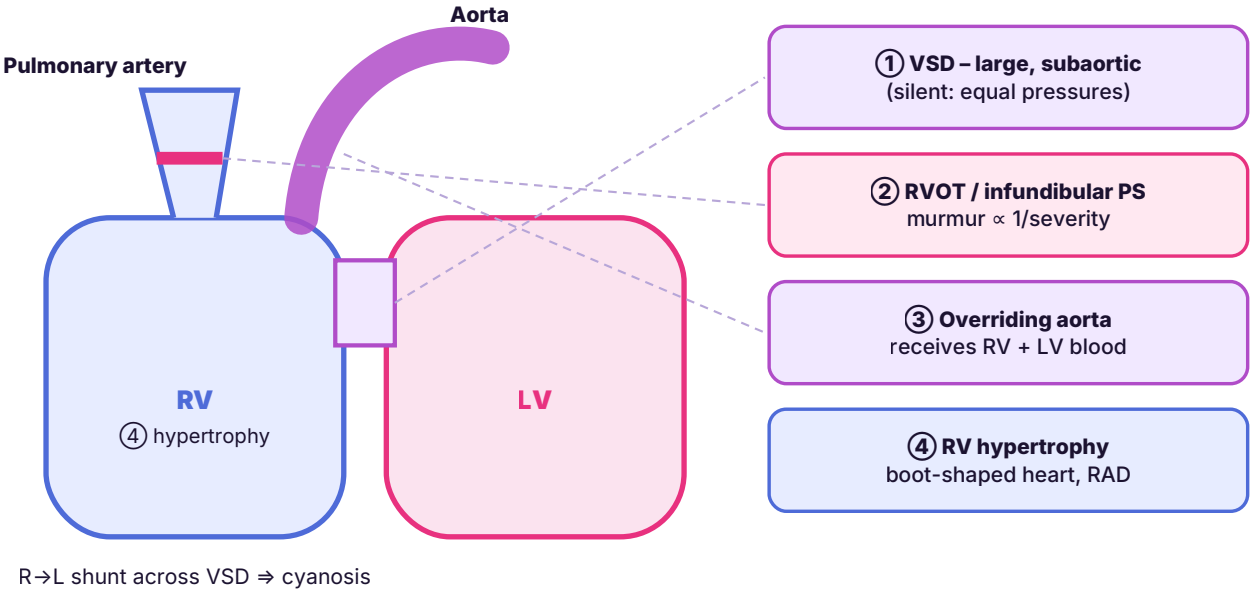
Feature	VSD	ASD (secundum)	PDA
Frequency	**MC CHD (25–30%)**; **perimembranous MC type**	6–10%; **F > M**; **ostium secundum MC**	5–10%; **preterm, rubella, high altitude**, F > M
Presentation	Small – asymptomatic, loud murmur; large – **CHF at 6–8 weeks**, FTT, recurrent LRTI	Usually **asymptomatic in childhood**; exercise intolerance, arrhythmia (AF) in adults	Small – asymptomatic; large – CHF, **bounding pulses, wide pulse pressure**

Feature	VSD	ASD (secundum)	PDA
Murmur	**Pansystolic (holosystolic), LLSB (3rd–4th ICS)**, ± thrill; large VSD – mid-diastolic at apex (↑ mitral flow)	**Ejection systolic at ULSB (pulmonary flow murmur – not across ASD)** + **mid-diastolic (tricuspid flow) at LLSB** (large shunt)	**Continuous "machinery" murmur, left infraclavicular / 2nd ICS (Gibson murmur)**; preterm – often systolic only
S2	Normal split; loud P2 if PAH	**Wide, FIXED split S2 (hallmark)**	**Paradoxical split** (large); masked
ECG	LVH (moderate), **biventricular hypertrophy (large – Katz–Wachtel)**	**RAD, rsR' in V1 (incomplete RBBB), RVH**; **primum ASD → left axis deviation**	LVH, LAE
CXR	Cardiomegaly (LA, LV), plethora	**RA, RV enlargement, prominent PA, plethora**; **LA normal**	LA, LV enlargement, prominent aorta
Natural history	**30–50% small (muscular) VSDs close spontaneously (by 2 y)**; risk – IE, AR (supracristal), Eisenmenger (by 1–2 y in large)	**Small (< 6–8 mm) may close in infancy**; rarely Eisenmenger (late)	Term PDA rarely closes; **IE (endarteritis)**, Eisenmenger
Treatment	CHF – diuretics, ACEi; **surgical closure if large/uncontrolled CHF/ PAH by 3–6 m**; device for muscular	**Closure at 2–5 years (device – secundum; surgery – primum/ sinus venosus)**	**Preterm: ibuprofen/ indomethacin/paracetamol; term – device (coil/Amplatzer) or surgical ligation**

- **Chamber enlargement rule**: **VSD & PDA → LA + LV**; **ASD → RA + RV** (LA not enlarged – decompresses). In VSD, **aorta is not enlarged**; in **PDA, aorta enlarged**.
- **Types of ASD**: secundum (MC, fossa ovalis), **primum** (part of AVSD – **cleft mitral valve, LAD**, Down), **sinus venosus** (SVC type – with **PAPVC of right upper pulmonary vein**), coronary sinus.
- **AVSD (complete)**: **Down syndrome**, **superior QRS axis (left axis/northwest)**, early CHF & PAH → **surgical repair by 3–6 months**.
- **Infective endocarditis prophylaxis** is NOT indicated for isolated ASD/VSD/PDA (only prosthetic valves, prior IE, unrepaired cyanotic CHD, repaired CHD with prosthetic material in first 6 months, residual defects).

13.4 Tetralogy of Fallot (TOF)

- **MC cyanotic CHD after infancy (overall)**; basic defect – **anterior & cephalad deviation of the infundibular (outlet) septum**.
- **4 components**: **(1) VSD (large, non-restrictive, subaortic), (2) RVOT obstruction – infundibular pulmonary stenosis (determines severity), (3) overriding aorta, (4) RV hypertrophy**. **Pentalogy = TOF + ASD**. **Right-sided aortic arch in ~25%**.



Tetralogy of Fallot – anatomy and haemodynamics

- **Cyanosis appears by 3–6 months** (progressive infundibular stenosis), **clubbing**, **squatting** (↑ SVR → ↓ R→L shunt; ↑ venous return), polycythaemia.
- **Auscultation**: **single S2 (soft/absent P2)**; **ejection systolic murmur at ULSB/LSB due to pulmonary stenosis (NOT the VSD)** – **murmur intensity inversely proportional to severity** (shortens/disappears during spell). **Large VSD is silent** (equal ventricular pressures).
- **ECG – RAD + RVH**; **CXR – boot-shaped heart, oligaemic lung fields**, normal heart size, concave pulmonary bay.
- **Echo – IOC**; cardiac catheterisation/CT for coronary anatomy (anomalous LAD crossing RVOT).

Hypercyanotic (Tet / blue) spells

- Peak **2–4 months** (rare after 2 y), **morning after waking, crying, feeding, defecation, dehydration, fever**; **hyperpnoea, ↑ cyanosis, limpness, loss/decrease of murmur**, syncope, seizures, death.
- Mechanism: **infundibular spasm** + ↓ SVR → ↑ R→L shunt → hypoxia → acidosis → hyperpnoea (vicious cycle).

MANAGEMENT OF A TET SPELL

1. **Knee–chest position** (↑ SVR) – first step; calm the child (on mother's shoulder).
2. **Oxygen** (limited benefit, but give).
3. **Morphine 0.1–0.2 mg/kg SC/IM/IV** (sedation, ↓ hyperpnoea).
4. **IV fluids (NS bolus 10–20 mL/kg)**; correct acidosis – **sodium bicarbonate 1–2 mEq/kg**.
5. **IV propranolol (0.01–0.25 mg/kg) / esmolol** – relaxes infundibular spasm; **phenylephrine (α-agonist) 5–20 µg/kg** – ↑ SVR; ketamine.
6. Refractory → emergency **modified Blalock–Taussig shunt** / RVOT stenting. - **Avoid: digoxin, inotropes (isoprenaline/dobutamine), vasodilators. Oral propranolol 0.5–1.5 mg/kg 6-hourly** to prevent spells; **treat iron-deficiency anaemia (relative anaemia precipitates spells)**.

- **Surgery**: **total (complete) repair at 3–12 months** (VSD closure + RVOT widening/transannular patch); palliative **modified Blalock–Taussig shunt (subclavian → pulmonary artery – Gore-Tex)** in small/unfavourable anatomy.
- **Complications**: **cerebral thrombosis (< 2 y, polycythaemia + dehydration)**, **brain abscess (> 2 y – R→L shunt bypasses lung filter)**, **paradoxical embolism**, IE, **relative iron-deficiency anaemia**, CHF (rare – only with PS absent valve/after BT shunt), late – **pulmonary regurgitation, RV dilatation, ventricular arrhythmia, RBBB** post-repair.

TOF DOES NOT USUALLY CAUSE HEART FAILURE

Unrepaired TOF presents with **cyanosis, not CHF** (RV decompresses into aorta through the VSD). CHF in TOF suggests **TOF with absent pulmonary valve**, anaemia, IE or a large BT shunt. **"Pink TOF"** (acyanotic) = mild RVOT obstruction with L→R shunt.

13.5 Other Cyanotic CHD

Lesion	Key features	Management
TGA (D-transposition)	**MC cyanotic CHD in newborn**; aorta from RV, PA from LV → **parallel circulations** – survival depends on **mixing (PFO/ASD, PDA, VSD)**; **male, IDM, macrosomic**; **severe cyanosis from day 1, no/soft murmur**, single loud S2; **CXR – egg-on-string, plethoric lungs**	**PGE_1 + balloon atrial septostomy (Rashkind)**; **arterial switch operation (Jatene) within 2–3 weeks – TOC** (before LV deconditions); Senning/ Mustard (atrial switch – historic)
Congenitally corrected TGA (L-TGA)	Ventricular inversion; acyanotic unless associated VSD/PS; **complete heart block**	
TAPVC	Pulmonary veins drain to systemic veins; **supracardiac (MC – vertical vein → innominate → SVC; snowman)**, cardiac (coronary sinus), **infracardiac (portal vein/IVC – always obstructed)**, mixed; **obligatory ASD**; **obstructed TAPVC = surgical emergency** (cyanosis, pulmonary oedema, "ground-glass" lungs, normal heart size – mimics RDS/PPHN)	Early surgical repair
Tricuspid atresia	**Cyanosis + LAD + LVH on ECG** (unique among cyanotic CHD – **"left axis + cyanosis = tricuspid atresia"**; also AVSD); obligatory ASD & VSD; hypoplastic RV	PGE_1, BT shunt → **Glenn (bidirectional cavopulmonary, 4–6 m) → Fontan (total cavopulmonary connection, 2–4 y)**
Truncus arteriosus	Single arterial trunk over VSD; **DiGeorge (22q11)**, right arch; truncal valve regurgitation; plethora, CHF, **bounding pulses**	Repair in neonatal period (Rastelli)
Ebstein anomaly	**Apical displacement of septal & posterior tricuspid leaflets → atrialised RV**, TR; **maternal lithium**; cyanosis (R→L at ASD), **massive cardiomegaly (box-shaped)**, **WPW syndrome (SVT) – 20%**, **"Himalayan" P waves**, RBBB; triple/ quadruple rhythm	Medical; surgery (cone repair)
HLHS	Hypoplastic LV, mitral & aortic atresia; **systemic circulation duct-dependent → shock, metabolic acidosis, weak pulses when duct closes (day 1–3)**	PGE_1; **Norwood → Glenn → Fontan**, or transplant
Pulmonary atresia with intact septum	Hypoplastic RV, RV-coronary fistulae; duct-dependent	PGE_1, RVOT perforation/shunt

13.6 Obstructive Lesions

	Coarctation of aorta (CoA)	Aortic stenosis	Pulmonary stenosis
Site/type	**Juxtaductal – just distal to left subclavian artery** (MC); preductal (infantile, severe) / postductal (adult)	Valvular (MC – **bicuspid**), supravalvular (**Williams**), subvalvular	**Valvular (MC – dome-shaped)**; dysplastic (**Noonan**); infundibular (TOF)
Association	**Turner (MC), bicuspid aortic valve (70%)**, berry aneurysms, VSD, PDA, Shone complex	Turner, Williams	Noonan, rubella, Alagille, Williams

	Coarctation of aorta (CoA)	Aortic stenosis	Pulmonary stenosis
Clinical	**Upper-limb hypertension**, **weak/delayed femoral pulses (radio-femoral delay)**, **BP arm > leg by ≥ 20 mmHg**, claudication; neonatal – **shock when duct closes**; ejection systolic murmur at back (interscapular), collaterals	**Ejection click, ESM at right 2nd ICS → neck (carotids)**, thrill in suprasternal notch, **narrow pulse pressure**, **slow-rising pulse**, syncope/sudden death on exertion	**Ejection click (↓ on inspiration – the only right-sided sound that decreases with inspiration)**, ESM at ULSB, **wide split S2 with soft P2**, RVH
X-ray	**Rib notching (posterior, 3rd–9th ribs; > 5 y)**, **"3" sign**, reverse-3 (E) on barium	Post-stenotic aortic dilatation	**Post-stenotic dilatation of main/left PA**
Rx	Neonate – **PGE_1** then surgery (resection + end-to-end anastomosis); older – **balloon angioplasty/stent**; **residual hypertension** (lifelong follow-up)	**Balloon valvotomy** (gradient > 50 mmHg)	**Balloon pulmonary valvuloplasty – TOC** (gradient > 40 mmHg)

13.7 Heart Failure in Children

Age	Common causes
First week	**HLHS**, critical AS/CoA, TAPVC (obstructed), large AV malformation, **arrhythmia (SVT)**, myocarditis, birth asphyxia, sepsis, severe anaemia, hypoglycaemia/hypocalcaemia
1–6 weeks → 6 months	**Large L→R shunts – VSD (MC), PDA, AVSD**, ALCAPA (anomalous left coronary from PA – infarction pattern at 2–3 m), CoA
Older children	**Rheumatic heart disease (MC in India)**, myocarditis, dilated cardiomyopathy, IE, hypertension (AGN), severe anaemia, thyrotoxicosis, beriberi

- **Features in infants**: **feeding difficulty (sweating & breathlessness during feeds – earliest)**, FTT, tachypnoea, tachycardia, **hepatomegaly (most reliable sign of right failure)**, gallop rhythm, cardiomegaly; **pedal oedema & raised JVP uncommon in infants** (facial/eyelid puffiness).
- **Ross classification** (infants) – modified NYHA.

TREATMENT OF CHF

Diuretics (furosemide 1–2 mg/kg) – first-line for congestion; spironolactone; **ACE inhibitors (enalapril/captopril)** – afterload reduction in L→R shunts & DCM; **β-blockers (carvedilol)** in DCM; **digoxin** (inotrope; less used, toxicity – vomiting, arrhythmias, **hypokalaemia potentiates toxicity**); IV inotropes (**milrinone, dobutamine**); **calorie-dense feeds (120–150 kcal/kg/day)**, treat anaemia & infection; definitive – surgical correction.

13.8 Infective Endocarditis (IE)

- Predisposing: **VSD (MC CHD with IE), TOF, PDA, bicuspid AV**, RHD, prosthetic valves, central lines (neonates); **not ASD secundum**.
- Organisms: **Viridans streptococci (MC – native valves, after dental procedures)**, **S. aureus (acute, IV drug use, central lines – MC overall in some series)**, enterococci, CONS (prosthetic), **HACEK** (culture-negative), Candida (neonates).
- **Modified Duke criteria** – major: **positive blood cultures (typical organism in 2 separate cultures)**, **echo evidence (vegetation, abscess, new regurgitation)**; minor: predisposition, fever ≥ 38 °C, vascular phenomena (emboli, **Janeway lesions**), immunological (**Osler nodes, Roth spots, GN**, RF), positive culture not meeting major. **Definite: 2 major or 1 major + 3 minor or 5 minor**.
- **Blood cultures – 3 sets from different sites before antibiotics** (most important investigation); **TEE more sensitive than TTE**.
- Rx: **penicillin/ceftriaxone + gentamicin (viridans) 4 weeks**; S. aureus – cloxacillin/vancomycin 6 weeks; surgery for CHF, abscess, large vegetations, fungal.

- **Prophylaxis (AHA)**: **amoxicillin 50 mg/kg (max 2 g) 30–60 min before dental procedures involving gingival manipulation** – only in highest-risk patients.

13.9 Acute Rheumatic Fever (ARF) & Rheumatic Heart Disease

- **Group A β-haemolytic streptococcal pharyngitis** (not skin) → **molecular mimicry (M protein)**; latent period **2–4 weeks**; age **5–15 years**; India – MC cause of acquired heart disease in children.
- **Revised Jones criteria (AHA 2015)**: evidence of **preceding GAS infection** (**ASO titre/anti-DNase B ↑, throat culture/rapid antigen**) **+ 2 major, or 1 major + 2 minor** (initial); recurrent – 2 major / 1 major + 2 minor / 3 minor.

	Low-risk population	Moderate/high-risk (India)
Major	**Carditis (clinical/subclinical – echo)**, **polyarthritis**, **chorea**, **erythema marginatum**, **subcutaneous nodules**	Carditis, **monoarthritis or polyarthritis or polyarthralgia**, chorea, EM, SC nodules
Minor	Polyarthralgia, **fever ≥ 38.5 °C**, **ESR ≥ 60 mm/h and/or CRP ≥ 3 mg/dL**, **prolonged PR interval** (if no carditis)	**Monoarthralgia**, **fever ≥ 38 °C**, **ESR ≥ 30 and/or CRP ≥ 3**, prolonged PR

- **Chorea (Sydenham's)** and **indolent carditis** alone are sufficient for diagnosis (no need for strep evidence).
- **Arthritis (MC major manifestation, 75%)**: **migratory/fleeting, large joints (knee, ankle, elbow, wrist), very painful, dramatic response to aspirin within 48 h, non-deforming**.
- **Carditis (50–60%) – pancarditis**; **mitral regurgitation – MC murmur (apical pansystolic)**; **Carey Coombs murmur** (mid-diastolic, apex – valvulitis); AR; pericarditis (rub); CHF. **Only manifestation causing permanent damage**.
- **Sydenham chorea (St Vitus' dance)**: **late (1–6 months after infection)**, **girls**, purposeless movements, emotional lability, **milkmaid's grip, pronator sign, spooning, darting tongue (jack-in-the-box)**, disappears in sleep; may be the only manifestation.
- **Erythema marginatum**: evanescent pink rings with **clear centre, serpiginous margins**, trunk & proximal limbs, **non-itchy, never on face**, ↑ with heat.
- **Subcutaneous nodules**: painless, pea-sized, over **extensor surfaces/bony prominences (elbows, knees, occiput, spine)**, appear weeks after onset, **associated with severe carditis**.

TREATMENT OF ARF

- **Eradicate strep**: **benzathine penicillin G single IM dose** (6 lakh U < 27 kg; 12 lakh U ≥ 27 kg) or oral penicillin V/amoxicillin × 10 days; erythromycin/azithromycin if allergic.
- **Arthritis/mild carditis**: **aspirin 50–100 mg/kg/day** (or naproxen) × 2 weeks, taper over 2–4 weeks.
- **Moderate–severe carditis/CHF**: **prednisolone 2 mg/kg/day** × 2–3 weeks then taper (+ aspirin while tapering).
- **Chorea**: **haloperidol**, valproate, carbamazepine; steroids/IVIG in severe.
- CHF – diuretics, ACEi, bed rest.

SECONDARY PROPHYLAXIS (MOST IMPORTANT FOR PREVENTING RHD)

Benzathine penicillin G 1.2 million U IM every 3 weeks (India/WHO; 4-weekly in low-risk); oral penicillin V 250 mg BD or erythromycin if allergic. **Duration**: ARF **without carditis – 5 years or until 21 y** (whichever longer); **carditis without residual disease – 10 years or until 21 y**; **carditis with residual heart disease (persistent valve disease) – 10 years or until 40 y, sometimes lifelong**.

- **Rheumatic heart disease**: **mitral valve MC (MR in children; MS in adolescents/adults – juvenile MS in India < 20 y)**; aortic next; tricuspid; pulmonary rarely. **"M > A > T"**. Echo – leaflet thickening, restricted motion; balloon mitral valvotomy for MS.
- **Post-streptococcal reactive arthritis** – non-migratory, poor aspirin response, shorter latency.
- **PANDAS** – OCD/tics after GAS.

13.10 Myocardial & Pericardial Disease, Hypertension

Condition	Key facts
Viral myocarditis	**Coxsackie B (MC), adenovirus**, parvovirus B19, HHV-6, SARS-CoV-2; tachycardia out of proportion to fever, CHF, arrhythmias, ↑ troponin; **cardiac MRI** / endomyocardial biopsy (Dallas criteria); Rx supportive, **IVIG** (debated), inotropes; diphtheritic myocarditis
Dilated cardiomyopathy	MC cardiomyopathy in children; idiopathic, post-myocarditis, **DMD/Becker**, carnitine deficiency, **selenium (Keshan)**, anthracyclines (doxorubicin), tachycardia-induced
Hypertrophic cardiomyopathy	AD (MYH7, MYBPC3); **asymmetric septal hypertrophy**, LVOT obstruction (murmur ↑ with Valsalva/ standing, ↓ squatting), **MCC sudden cardiac death in young athletes**; IDM (transient), Pompe, Noonan, Friedreich; Rx **β-blockers**, avoid competitive sports, ICD; **avoid digoxin/inotropes/ vasodilators**
Restrictive cardiomyopathy	Rare; endomyocardial fibrosis (tropical)
Acute pericarditis	Viral (MC), bacterial (S. aureus – purulent), TB (constrictive), uraemic, post-pericardiotomy, SLE; **pericardial rub**, chest pain relieved leaning forward, **diffuse ST elevation (concave), PR depression**; tamponade – **Beck triad, pulsus paradoxus (> 10 mmHg)**, electrical alternans
Hypertension in children	**≥ 95th centile for age, sex & height on ≥ 3 occasions** (AAP 2017; **≥ 130/80 for ≥ 13 y**); **secondary HTN commoner in young children – renal parenchymal (MCC: reflux nephropathy, GN, CKD)**, renovascular (**Takayasu, FMD**), CoA, endocrine (pheochromocytoma, Cushing, CAH 11β-OH), drugs; adolescents – primary (obesity)
Hypertensive emergency	**IV labetalol, nicardipine, sodium nitroprusside**, esmolol; reduce BP by ≤ 25% in first 8 h

Practice MCQs · Pediatric Cardiology

212 questions · correct option highlighted in green · cover the answer strip to self-test

Fetal & Neonatal Circulation

1. The heart lesion not found in congenital rubella infection is:

A. ASD
B. VSD
C. PDA
D. PS

Ans A Cardiac anomalies in congenital rubella are PDA (most common) > PS > VSD > ASD (least associated).

2. Which one of the following does not produce cyanosis in the first year of life?

A. Atrial septal defect
B. Hypoplastic left heart syndrome
C. Truncus arteriosus
D. Double outlet right ventricle

Ans A Apart from ASD, rest of the 3 heart diseases are cyanotic heart diseases.

3. A child is admitted on 7th days of life with severe respiratory distress and shock. He was discharged 2 days back healthy. What could be the probable diagnosis?

A. VSD large
B. Hypoplastic left heart syndrome
C. Ebstein anomaly
D. AP window defect

Ans B High yield points about hypoplastic left heart syndrome (HLHS)
• It usually presents in the first few days of life with Cyanosis and heart failure
• It is the most common congenital heart disease causing mortality in 1st week of life

4. Pulmonary plethora is seen in all except:

A. VSD
B. ASD
C. Fallot's tetralogy
D. PDA

Ans C

5. One of the following cardiac lesions have been identified more in male

A. transposition of the great arteries
B. atrial septal defect
E. pulmonic stenosis

Ans A Gender differences in the occurrence of specific cardiac lesions have been identified. Transposition of the great arteries and left-sided obstructive lesions are slightly more common in boys (≈65%), whereas atrial septal defect, VSD, PDA, and pulmonic stenosis are more common in girls.

6. Tall (>2.5 mm), narrow, and spiked P waves are seen in

A. Ebstein anomaly
B. ventricular septal defect [VSD]
C. patent ductus arteriosus [PDA]
D. severe mitral stenosis
E. mitral regurgitation

Ans A Tall (>2.5 mm), narrow, and spiked P waves are indicative of right atrial enlargement and are seen in congenital pulmonary stenosis, Ebstein anomaly of the tricuspid valve, tricuspid atresia, and sometimes cor pulmonale. These abnormal waves are most obvious in leads II, V3R, and V1. Similar waves are sometimes seen in thyrotoxicosis. Broad P waves, commonly bifid and sometimes biphasic, are indicative of left atrial enlargement.

7. For one of the following, total repair really achieved, with no requirement for long- term follow-up

A. atrial septal defects
B. ventricular septal defects
C. valvar pulmonic stenosis
D. uncomplicated tetralogy of Fallot
E. isolated patent ductus arteriosis

Ans E Many argue that only for isolated patent ductus arteriosus is total repair really achieved, with no requirement for long-term follow-up.

8. Obstructed total anomalous pulmonary venous return (TAPVR) is a pediatric cardiac surgical emergency because

A. prostaglandin therapy is usually not effective
B. severe pulmonary congestion develop
C. left atrium may be small
D. left ventricle may be small
E. pulmonary hypertension develop

Ans A Obstructed TAPVR is a pediatric cardiac surgical emergency because prostaglandin therapy is usually not effective.

9. The vast majority of tumors originating from the heart are benign. Of the following, the MOST common pediatric cardiac tumors are

A. fibromas
B. rhabdomyomas
C. myxomas
D. hemangiomas
E. papillomas

Ans B are the most common pediatric cardiac tumors and are associated with tuberous sclerosis in 70-95% of cases.

Congenital Heart Disease - General

10. Which of the following is NOT a major NADA's criteria for clinical diagnosis of congenital heart disease:

A. Diastolic murmur
B. Cyanosis
C. Congestive cardiac failure
D. **Abnormal X-ray**

Ans D /e p 406; 14. a PGE1 PGE1 or Alprostadil can be used to keep the ductus arteriosus open in Ductus dependent heart diseases.

11. Commonest type of congenital heart disease is:

A. ASD
B. **VSD**
C. TOF
D. PDA

Ans B

12. The most common congenital heart disease is:

A. ASD
B. **VSD**
C. PDA
D. TOF

Ans B

13. Which of the following syndromes is best associated with congenital heart disease?

A. Lesch-Nyhan syndrome
B. Rasmussen syndrome
C. **Holt-Oram syndrome**
D. Leopard syndrome

Ans C Discussing about the options one by one, Name of the syndrome Clinical features a. Lesch-Nyhan syn- Hyperuricemia, intellectual disability, dystonia, choreoathetosis, spasticity, dysarthric speech and compulsive drome self-biting, usually beginning with eruption of teeth b. Rasumussen syn- It is an immune mediated disease associated with intractable unilateral seizures, progressive hemiparesis and drome intellectual dysfunction c.

14. All of the following are acyanotic congenital heart diseases except:

A. VSD
B. PDA
C. ASD
D. **Tetralogy of Fallot**

Ans D

15. Cyanotic congenital heart disease with LA enlargement and left axis deviation?

A. **Tricuspid atresia**
B. TOF
C. TAPVC
D. Ebstein anomaly

Ans A For all practical purposes, Cyanotic heart disease with left axis deviation is Tricuspid atresia. with

16. Ductus arteriosus dependent blood flow is mandatory in all of the following congenital heart diseases except:

A. **Truncus arteriosus**
B. Hypoplastic left heart syndrome
C. TGA with Intact ventricular septum
D. Obliterated aortic arch

Ans A In Truncus arteriosus, both the aorta and pulmonary artery arise from a common large blood vessel. So there is already a complete mixing of blood of systemic and pulmonary circulation, with no obstructive lesion in the pulmonary or systemic circulatiosssn; So ductus dependent blood flow is not mandatory.

17. Which one of the following is a cyanotic congenital heart disease?

A. Recurrent chest infection
B. Clubbing of hands and feet
C. Squatting
D. **Cyanotic spells**
E. CHF not seen

Ans D Tetralogy of Fallot

18. Congenital heart disease associated with pre-excitation is:

A. Atrial septal defect
B. Bicuspid aorta valve
C. **Ebstein anomaly**
D. Patent ductus arteriosus

Ans C Ebstein's anomaly Diagnosis of Congenital Heart Disease By M Satpathy, p 245 Pre-excitation refers to early activation of ventricles due to impulses bypassing the AV node via an accessory pathway; Accessory pathways, also known as bypass tracts, are abnormal conduction pathways formed during cardiac development and are seen in conditions like WPW syndrome; ECG findings of Ebstein anomaly:
• Normal or tall and broad ('himalayan') P waves, and a normal or prolonged P-R interval
• Right bundle-branch block

19. The most common congenital cardiac defect associated with sudden death in children is:

A. Valvular aortic stenosis
B. **Origin of the left coronary artery from the right sinus of Valsalva**
C. Hypertrophic cardiomyopathy
D. Congenital mitral regurgitation300 Section 3: Systemic Pediatrics 163.

Ans B Anomalous origin of the left coronary artery from the right sinus of Valsalva (or from the pulmonary artery) is a well-recognized cause of sudden cardiac death in children, along with hypertrophic cardiomyopathy.

20. A 3-month-old child has a typical mewing cry and a congenital heart disease. The most probable chromo- somal abnormality is:

A. 5q-
B. **5p-**
C. 4p-
D. 9p-

Ans B Cat cry with congenital heart disease in an infant suggests Cri du chat syndrome

21. Congenital heart disease causing cyanosis without respiratory distress include the following EXCEPT

A. tricuspid atresia
B. **aortic stenosis**
C. Ebstein anomaly
D. pulmonary atresia
E. tetralogy of Fallot

Ans B

22. The MOST common cause of death from cardiac defects in the first month of life is

A. d-Transposition without associated lesions
B. **hypoplastic left heart syndrome**
C. pulmonary atresia
D. truncus arteriosus
E. complex single ventricle

Ans B accounts for 1% of all congenital heart defects but is the most common cause of death from cardiac defects in the first month of life.

23. The chest x-ray may help to differentiate the types of congenital heart defects. Increased pulmonary markings (increased pulmonary blood flow) is seen in

A. pulmonary atresia
B. **truncus arteriosus**
C. tetralogy of Fallot
D. tricuspid atresia
E. Ebstein anomaly

Ans B Decreased pulmonary markings (diminished pulmonary blood flow) Pulmonary atresia or severe stenosis Tetralogy of Fallot Tricuspid atresia Ebstein anomaly Increased pulmonary markings (increased pulmonary blood flow) Transposition of great arteries Total anomalous pulmonary venous return Truncus arteriosus

24. Careful evaluation of the character of the pulses is an important early step in the physical diagnosis of congenital heart disease. A wide pulse pressure with bounding pulses may suggest the following EXCEPT

A. patent ductus arteriosus
B. aortic insufficiency
C. **cardiomyopathy**
D. anemia
E. anxiety

Ans C The presence of diminished pulses in all extremities is associated with pericardial tamponade, left ventricular outflow obstruction, or cardiomyopathy.

25. D-transposition of the great vessels, a common cyanotic congenital anomaly, accounts for ≈5% of all congenital heart disease. In this anomaly, all the following are true EXCEPT

A. the systemic veins return to the right atrium
B. the pulmonary veins return to the left atrium
C. the connections between the atria and ventricles are normal
D. **the aorta is posterior and to the right of the pulmonary artery**
E. the aorta is anterior and to the right of the pulmonary artery

Ans D In normally related great vessels, the aorta is posterior and to the right of the pulmonary artery; in d-transposition of the great arteries (d-TGA), the aorta is anterior and to the right of the pulmonary artery (the d indicates a dextropositioned aorta, transposition indicates that it arises from the anterior right ventricle).

Acyanotic CHD (L-R Shunts)

26. A child after 4 weeks of birth is acyanotic, but a systolic murmur is detected. Causes are/is:

A. **VSD**
B. PDA
C. TOF
D. Coarctation of aorta
E. Tricuspid stenosis

Ans A b. PDA, d. Coarctation of aorta, e. Tricuspid stenosis A systolic murmur can be heard in all the above heart diseases; PDA usually produces a continuous murmur, but may sometimes present with a systolic murmur in the initial period of life. Out of the above diseases, TOF is a cyanotic heart disease.

27. Definitive treatment of TGV:

A. **Arterial switch**
B. Atrial switch
C. Atrial septostomy
D. Medical management RHEUMATIC HEART DISEASE

Ans A Transposition of Great vessels or arteries (TGV or TGA) is a condition characterized by aorta arising from right ventricle and pulmonary artery from left ventricle; Definite treatment for TGA is Arterial switch operation or Jatene's repair;

28. Regarding mitral valve prolapse; all the following are true EXCEPT

A. it is predominantly congenital
B. it is usually sporadic
C. it is more common in girls
D. the dominant abnormal signs are auscultatory
E. **antibiotic prophylaxis is recommended during surgery and dental procedures**

Ans E This lesion is not progressive in childhood, and specific therapy is not indicated. Antibiotic prophylaxis is no longer recommended during surgery and dental procedures.

29. Of the following, the cardiac lesion resulting in increased volume load is

A. valvular pulmonic stenosis
B. **patent ductus arteriosus**
C. valvular aortic stenosis
D. coarctation of the aorta
E. mitral stenosis

Ans B The most common lesions in this group are those that cause left-to-right shunting: atrial septal defect, ventricular septal defect (VSD), AV septal defects (AV canal), and patent ductus arteriosus.

30. Pregnancy is contraindicated in mothers with

A. **severe pulmonary hypertension**
B. ventricular septal defects
C. prosthetic valves
D. atrial septal defects
E. unoperated cyanotic congenital heart disease

Ans A Pregnancy is contraindicated in mothers with severe pulmonary hypertension, severe obstructive lesions, and Marfan syndrome, aortic root >40 mm.

31. In neonates with total anomalous pulmonary venous return and marked pulmonary venous obstruction, the chest x-ray demonstrates

A. **small heart**
B. enlarged heart
C. prominent pulmonary artery
D. prominent right ventricle
E. subaortic stenosis

Ans A In neonates with marked pulmonary venous obstruction, the chest x-ray demonstrates a very dramatic perihilar pattern of pulmonary edema and a small heart.

32. Adverse reactions to angiotensin-converting enzyme inhibitors (ACEIs) include the following EXCEPT

A. hypotension
B. **hypokalemia**
C. maculopapular pruritic rash
D. renal toxicity
E. chronic cough

Ans B Adverse reactions to ACEIs include hypotension and its sequelae (weakness, dizziness, syncope) and hyperkalemia.

Cyanotic CHD (R-L Shunts)

33. True about ostium primum defect:

A. **It is found in ASD**
B. More commonly associated with ASD than VSD
C. May be associated with Down syndrome
D. Ostium primum ASD is more common than ostium secundum ASD
E. Right axis deviation is seen on ECG

Ans A b. More commonly associated with ASD than VSD & c. May be associated with Down syndrome Left axis deviation is seen in ostium primum ASD due to RV conduction defect. Ostium primum type ASD
• It is situated in the lower portion of atrial septum and overlies mitral and tricuspid valves.
• In most instances, a cleft in anterior leaflet of mitral valve is also noted; Ventricular septum is intact.
• Tricuspid valve is usually functionally normal, but anatomic abnormality of septal leaflet is present.

34. A 4-year-old girl always had to wear warm socks even in summer season. On physical examination, it was noticed that she had high blood pressure and her femoral pulse was weak as compared to radial and carotid pulse, a chest radiography showed remarkable notching of ribs along with their lower borders. This was due to:

A. Femoral artery thrombosis (AIIMS Nov 02)
B. **Coarctation of aorta**
C. Raynaud's disease
D. Takayasu arteritis

Ans B Hypertension, Weak femoral pulses and notching of inferior margin of ribs point towards Coarctation of aorta.

35. In a child with tetralogy of Fallot with fever and diarrhoea, which of the following is the surest sign of a cyanotic spell?

A. Arterial oxygen saturation of less than 75%
B. S3 gallop rhythm
C. **Absence of murmur**
D. Hepatomegaly

Ans C weiveR During cyanotic spell, temporary disappearance or a decrease in intensity of systolic murmur is usual as flow across the right ventricular outflow tract diminishes.

36. A child comes with cyanotic spells and chest X-ray was as shown below; What is the most probable diagnosis?

A. **Tetralogy of Fallot**
B. Tricuspid atresia
C. Pulmonary atresia with intact ventricular septum
D. TAPVC

Ans A The given history along with chest X-ray finding of Boot shaped heart suggests a diagnosis of Tetralogy of Fallot.

37. Most common cyanotic heart disease:

A. **TOF**
B. VSD
C. TAPVC
D. Ebstein anomaly

Ans A • Overall the most common cyanotic heart disease is TOF.

38. A newborn baby develops cyanosis on day three of life. On auscultation, there is a systolic murmur. Echocardiography reveals a cyanotic heart disease in 101. the baby. Which one of the following drugs can be administered to prolong the life of the baby pending intervention?

A. Indomethacin
B. Ibuprofen
C. **Prostaglandin E1**

Ans C Most common cyanotic heart disease presenting in the 1st week of life is TGA. In TGA, which is a ductus dependent heart disease, PGE1 may be administered or balloon atrial septostomy may be done for initial stabilization of the patient. Definite surgery for TGA is arterial switch operation or Jatene repair.

39. A five-day-old, full term male infant was severely cyanotic at birth. Prostaglandin E1 was administered initially and later balloon atrial septosomy was done which showed improvement in oxygenation. The most likely diagnosis of this infant is:

A. Tetralogy of Fallot
B. Transposition of great vessels
C. Truncus arteriosus
D. Tricuspid atresia

Ans B Balloon atrial septostomy is helpful in a baby with TGA with intact ventricular septum for initial stabilization, as by creating a communication between the left and right atria, it helps in mixing blood of systemic and pulmonary circulations.

40. The MOST common cyanotic cardiac lesion to present in the newborn period is

A. dextroposed transposition of the great arteries
B. hypoplastic left heart syndrome
C. tetralogy of Fallot
D. truncus arteriosus
E. pulmonary atresia

Ans A Although dextroposed transposition of the great arteries represents only about 5% of congenital heart defects, it is the most common cyanotic lesion to present in the newborn period.

41. Of the following, the cyanotic cardiac lesion with decreased pulmonary blood flow is

A. transposition of the great vessels
B. single ventricle
C. truncus arteriosus
D. tricuspid atresia
E. total anomalous pulmonary venous return without obstruction

Ans D This group of congenital heart lesions include (tetralogy of Fallot, pulmonary atresia with an intact septum, tricuspid atresia, total anomalous pulmonary venous return with obstruction).

42. Paroxysmal hypercyanotic attacks (hypoxic, "blue," or "tet" spells) are a particular problem during the 1st 2 yr of life. They are characterized by

A. early evening occurrence
B. an increase in intensity of the systolic murmur
C. unpredictable onset
D. metabolic alkalosis
E. more freguent spells in patients with marked cyanosis at rest

Ans C The spells occur most frequently in the morning on initially awakening or after episodes of vigorous crying. Temporary disappearance or a decrease in intensity of the systolic murmur is usual as flow across the right ventricular outflow tract diminishes. The onset is usually spontaneous and unpredictable. Spells are associated with reduction of an already compromised pulmonary blood flow, which, when prolonged, results in severe systemic hypoxia and metabolic acidosis.

43. Depending on the frequency and severity of hypercyanotic attacks in tetralogy of Fallot, all the following procedures are true EXCEPT

A. placement of the infant in the knee-chest position
B. administration of oxygen
C. injection of morphine subcutaneously
D. rapid correction with intravenous sodium bicarbonate irrespective of the spell severity
E. intravenous administration of propranolol

Ans D Because metabolic acidosis develops when arterial Po2 is <40 mm Hg, rapid correction (within several minutes) with intravenous administration of sodium bicarbonate is necessary if the spell is unusually severe and the child shows a lack of response to the foregoing therapy. 54 (C). Concavity in the region of the main pulmonary artery.

ASD, VSD & PDA

44. Which of the following are true about ASD?

A. Ostium primum is the most common defect
B. Children are always growth retarded
C. Surgery is electively done at 1-3 year age for large defects
D. Commonly affected by Infective endocarditis
E. It is the commonest congenital heart disease

Ans C The timing for elective closure of ASD is usually after the 1st yr and before entry into school.

45. Drug used to keep PDA open:

A. PGE1
B. PGI2
C. PGE2
D. PGH2

Ans A Prostaglandin E1 (PGE1) is used to maintain patency of the ductus arteriosus in duct-dependent congenital heart disease.

46. Which type of ASD is commonest?

A. Patent foramen ovale
B. Ostium primum
C. Ostium secundum
D. Sinus venosus

Ans C

47. Large PDA leads to:

A. Endocardial valvulitis
B. Essenmenger syndrome
C. Heart failure
D. All of the above

Ans D All of above Complications of large PDA are Cardiac failure; Infective endarteritis, Pulmonary or systemic emboli, Pulmonary hypertension (Eisenmenger syndrome). dna 23. a. PGE1

48. ASD is associated with all except:

A. Infective endocarditis
B. Stroke
C. Arrhythmia
D. Pulmonary hypertension

Ans A Infective endocarditis is usually not seen in ASD, as the gradient between right and left atria is very small in ASD.

49. **In patent ductus arteriosus, connection is between:**

A. Aorta and coronary artery
B. **Aorta and pulmonary artery**
C. Aorta and subclavian artery
D. Pulmonary artery and subclavian artery

Ans B Ductus arteriosus is a connection between the aorta and pulmonary artery.

50. **In a preterm baby with patent ductus arteriosus all are true except?**

A. **Narrow pulse pressure**
B. Necrotising enterocolitis
C. Continuous murmur
D. Congestive heart failure

Ans A In PDA, there is shunting of blood from systemic circulation to pulmonary circulation, which leads to: congestive cardiac failure, "wide pulse pressure", "bounding pulses" Overloading of pulmonary vasculature pulmonary edema/ hemorrhage and CO2 retention Blood flow to kidney & GIT is compromised → acute renal failure; necrotizing enterocolitis & metabolic acidosis.

51. **A child is having perimembranous VSD, CCF develops due to:**

A. Aortic regurgitation
B. Spontaneous closure
C. Changes in pulmonary vasculature
D. **Infective endocarditis**

Ans D

52. **In ostium secundum ASD, maximally affected is:**

A. **Right atrium → right ventricle → left atrium**
B. Right ventricle → right atrium → left atrium
C. Left atrium → right atrium → right ventricle
D. Left ventricle → left atrium → right atrium

Ans A

53. **Patient with VSD develops pulmonary hypertension. Which of the following will be present?**

A. Cyanosis
B. **Initial resolution of symptoms**
C. Clubbing
D. Ejection systolic murmur in pulmonary area

Ans B • Initial resolution of symptoms can be seen in a child with VSD, when pulmonary hypertension develops;
• This is so because pulmonary hypertension poses resistance to blood flowing to lungs → lesser blood goes to lungs;
• Ultimately, when Eissenmenger syndrome develops, cyanosis and clubbing appears.

54. **Child with PDA will not have:**

A. Bounding pulses
B. Pulmonary hemorrhage
C. Necrotising enterocolitis
D. **CO washout**

Ans D

55. **Which of the following statements is/are FALSE about ostium secundum ASD?**

A. Fixed splitting of 2nd heart sound
B. **Narrow splitting of 2nd heart sound**
C. Lt axis deviation in ECG
D. Shunt murmur prominent
E. Rt axis deviation

Ans B c. Lt axis deviation in ECG, d. Shunt murmur prominent • In ASD S2 is fixed and wide split (not narrow split); Ostium secundum ASD is associated with Right axis deviation
• Pressure difference between 2 atria is small, blood passes at a narrow pressure difference → So, no shunt murmur.

56. **In ostium secundum ASD, which of the following is maximally affected:**

A. Right ventricle
B. Left ventricle
C. Left atrium
D. **Right atrium**

Ans D

57. **Most common type of atrial septal defect is:**

A. Ostium primum
B. **Ostium secondum**
C. Endocardial cushion defect
D. Endocardial hypertrophy

Ans B Ostium secondum Answers

58. **A premature infant is born with a patent ductus arteriosus. Its closure can be stimulated by adminis- tration of:**

A. Prostaglandin analogue
B. Estrogen
C. Anti-estrogen compounds
D. **Prostaglandin inhibitors**

Ans D

59. **A child with large perimembranous VSD has congestive heart failure. What may be the cause of improvement of cardiac failure in the patient?**

A. Aortic regurgitation
B. **Vascular changes in pulmonary circulation**
C. Infective endocarditis
D. Closure of VSD spontaneously

Ans B Spontaneous closure of a large membranous VSD is unlikely; Vascular changes in pulmonary circulation & pulmonary hypertension can lead to improvement of cardiac failure, but may subsequently lead to cyanosis & clubbing.

60. **ASD is seen in all except:**

A. **Turner's syndrome**
B. Ellis-Van Creveld syndrome
C. Down's syndrome
D. Holt-Oram syndrome
E. TAR syndrome

Ans A For Mnemonic on syndromes associated with ASD,

61. All of the following are true about ASD except:

A. Right atrial hypertrophy
B. Left atrial hypertrophy
C. Right ventricular hypertrophy
D. Pulmonary hypertension

Ans B

62. The following statements are true about patent ductus arteriosus (PDA) except:

A. Spontaneous closure occurs in some term infants
B. Pulmonary hypertension develops
C. Bacterial endocarditis is more frequent with small PDA
D. Recurrent chest infection and congestive failure may develop
E. Anatomic existence of PDA is an indication for surgery

Ans E Discussing about the options one by one a. True, Spontaneous closure can occur in term as well as preterm babies with PDA b. True, as pulmonary blood flow increased in PDA, pulmonary hypertension & Elssenmenger syndrome can develop c. True, smaller the PDA, more forceful will be the jet of blood flow, causing more turbulence and erosion of endocardium, predisposing to infective endocarditis d. True, because of increased pulmonary blood flow, recurrent chest infection and congestive failure may develop e.

63. All of the following about PDA are true except:

A. More common in males
B. Common heart lesion in rubella
C. Treatment is closure of defect by ligation and division of 108. ductus
D. Hypoxia and immaturity are important in maintaining the patency

Ans A M neMonic: in Boys—'Baaya' Meaning Left; so, Left sided oBstructive Lesions
• Congenital heart diseases more common in girls are ASD, PDA, VSD, Pulmonic stenosis
• Congenital heart diseases more common in boys are TGA, Left sided obstructive lesions like coarctation of aorta a. Valvular stenosis • In TOF, obstruction to pulmonary arterial blood flow is usually both at the right ventricular infundibulum (subpulmonic area) and the pulmonary valve

64. A child with VSD presents with development of cyanosis because of Eisenmenger physiology. What is the correct sequence of events which leads to this change?

A. Left to right shunt, pulmonary hypertension, right ventricular hypertrophy, right to left shunt.
B. Left to right shunt, right ventricular hypertrophy, pulmonary hypertension, right to left shunt.
C. Pulmonary hypertension, right to left shunt, right ventricular hypertrophy, left to right shunt.
D. Left to right shunt, right ventricular hypertrophy, right to left shunt, pulmonary hypertension.

Ans A Left to right shunt, pulmonary hypertension, right ventricular hypertrophy, right to left shunt. 2243 Sequence of changes in Eisenmenger syndrome is: Left to right shunt (VSD or PDA) → Increased blood flow to lungs → Pulmonary with hypertension → Right ventricular hypertrophy → Right to left shunt → cyanosis and clubbing.

65. A very large ventricular septal defect should be operated:

A. In school going age
B. Only if CHF is uncontrolled
C. During infancy
D. Soon after birth

Ans C Indications for surgical closure of a VSD include:
• Patients at any age with large defects in whom clinical symptoms and failure to thrive cannot be controlled medically;
• Infants 6 – 12 months age with large defects & pulmonary hypertension, even if symptoms are controlled by medication;
• Patients older than 24 months with a Qp : Qs ratio greater than 2 : 1.
• Patients with a supracristal VSD of any size (because of the high risk for aortic valve regurgitation) a.

66. B. Continuous machinery murmur 2. ASD C. Muffled heart sounds 3. PDA D. Wide variable split of S2 4. Pericard. effusion Code: A B C D

A. 2 4 3
B. 2 3 4
C. 1 4 3
D. 1 3 4 2 169.

Ans A Renal artery stenosis, b. Coarctation of aorta, c. Glomerulonephritis, e. Pheochromocytoma

67. In MOST patients with an ASD, the characteristic physical finding is

A. a right ventricular systolic lift
B. a fixed splitted 2nd heart sound
C. a systolic ejection murmur
D. a short, rumbling mid-diastolic murmur
E. a mild left precordial bulge

Ans B In most patients with an ASD, the characteristic finding is that the 2nd heart sound is widely split and fixed in its splitting during all phases of respiration.

68. Indications for surgical closure of a VSD include the following EXCEPT

A. patients in whom clinical symptoms and failure to thrive cannot be controlled medically
B. infants between 6 and 12 mo of age with large defects associated with pulmonary hypertension
C. patients older than 24 mo with a Qp : Qs ratio greater than 2 : 1.
D. patients with a supracristal VSD of any size
E. severe pulmonary vascular disease nonresponsive to pulmonary vasodilators

Ans E

69. Regarding supracristal ventricular septal defect, the following are true EXCEPT

A. a supracristal VSD is complicated by aortic insufficiency in 50-90% of patients
B. the incidence is higher in Asian children
C. aortic insufficiency is most often not recognized until late in the 1st decade of life
D. the murmur of a supracristal VSD is usually heard at the mid to upper left sternal border
E. closure of supracristal ventricular VSDs at the time of diagnosis is not recommended in an asymptomatic child

Ans E Closure of all supracristal ventricular VSDs at the time of diagnosis is commonly recommended to prevent the development of aortic regurgitation, even in an asymptomatic child.

70. Surgical repair of VSD at the time of diagnosis should be considered in

A. small supracristal VSD
B. nonrestrictive VSD
C. hemodynamically significant VSD
D. restrictive VSD
E. VSD with heart failure

Ans A The exception to this rule is patients with small supracristal or perimembranous VSD with associated prolapse of the aortic cusp into the defect resulting in progressive aortic regurgitation. These patients should be considered for surgical repair at the time of diagnosis to prevent progressive aortic valve damage.

71. Patients with a small PDA have the following criteria EXCEPT

A. continuous murmur heard best at the left upper sternal border
B. normal peripheral pulses
C. normal pulmonary artery pressure by echocardiography
D. asmptomatic and live a normal life expectancy
E. risk for endocarditis is extremely low

Ans E A silent PDA is a tiny defect that cannot be heard by auscultation and is only detected by other nonclinical means such as echocardiography. Life expectancy is always normal in this population and the risk for endocarditis is extremely low. Patients with a small PDA have an audible long-ejection or continuous murmur heard best at the left upper sternal border that radiates to the back. In addition, they have normal peripheral pulses.

72. All patients with clinical evidence of a PDA are at increased risk for endocarditis. As result, PDA should be considered for closure EXCEPT

A. PDA with severe irreversible pulmonary hypertension
B. small, hemodynamically insignificant PDA
C. small, hemodynamically significant PDA
D. moderate size PDA
E. large PDA

Ans A All PDAs except for small silent PDAs and those patients with severe irreversible pulmonary hypertension should be considered for closure.

Tetralogy of Fallot

73. In Ductus dependent heart diseases, drug used is

A. PGE1
B. PGI2
C. PGE2
D. PGH2294 Section 3: Systemic Pediatrics

Ans A is the drug of choice to keep the ductus arteriosus open in duct-dependent cardiac lesions until definitive surgery or catheter intervention.

74. True statement about ductus arteriosus is:

A. It undergoes anatomic closure within 24 hours of birth
B. Forms the ligamentum venosum in later life
C. It is induced to close by high levels of prostaglandins
D. May cause a machinery murmur by its patency.

Ans D Closure of ductus arteriosus
• Physiological closure: occurs almost immediately after birth by contraction of muscular wall, mediated by bradykinin
• Anatomic closure: occurs by proliferation of intima in 10-21 days; After obliteration, it forms ligamentum arteriosum. Note: Murmur due to PDA is classically referred to as a continuous machinery murmur.

75. All of the following statements are true regarding TOF, except:

A. Right ventricular hypertrophy
B. Atrial septal defect
C. Overriding of aorta
D. Subpulmonic stenosis

Ans B Components of Tetralogy of fallot include ventricular septal defect, infundibular pulmonic stenosis, right ventricular hypertrophy & overriding of aorta.

76. What is the treatment for sudden worsening of TOF?

A. Morphine
B. O therapy
C. Propranolol
D. All of the above

Ans D All of the above a. Supracardiac

77. All are seen in tetralogy of Fallot except?

A. VSD
B. RVH
C. ASD
D. PS

Ans C 4 components of TOF are: Large VSD, infundibular pulmonary stenosis, overriding of Aorta, Right ventricular hypertrophy.

78. Which of the following represents the additional component in pentalogy of Fallot?

A. Atrial Septal Defect (ASD)
B. Ventricular Septal Defect (VSD)
C. Right ventricular Hypertrophy (RVH)
D. Pulmonic Stenosis

Ans A Pentalogy of Fallot = Tetralogy of Fallot + Atrial Septal defect.

79. All are characteristic of Tetralogy of Fallot except:

A. Infundibular stenosis
B. VSD
C. Overriding of aorta
D. **Left ventricular hypertrophy**

Ans D

80. Which of the following is a component of pentalogy of Fallot:

A. **Atrial Septal Defect (ASD)**
B. Patent Ductus Arteriosus (PDA)
C. Coarctation of Aorta (COA)
D. Left Ventricular Hypertrophy (LVH)

Ans A

81. A child with tetralogy of Fallot uses which of the following positions?

A. Supine
B. Prone
C. **Squatting**
D. Leaning forwards

Ans C • The patients of TOF assume a sitting posture (squatting) as soon as they get dyspneic
• TOF is the commonest congenital lesion in which squatting is noted. Squatting helps by:
• Increasing systemic vascular resistance, so that more blood goes to lungs for oxygenation
• Trapping deoxygenated venous blood in lower limbs.

82. Systolic murmur in TOF is due to?

A. Fallot's tetralogy 106.
B. **Transposition of great vessels**
C. Ebstein's anomaly
D. Ventricular septal defect

Ans B Pulmonary stenosis Ejection systolic murmur in TOF is due to pulmonary stenosis. The intensity and duration of this systolic murmur is inversely proportional to the severity of TOF.

83. Essential criteria for TOF includes all except:

A. **Valvular stenosis**
B. Infundibular stenosis
C. Over riding of aorta
D. RVH

Ans A The essential anatomic features of tetralogy of Fallot are infundibular (subpulmonary) stenosis, overriding aorta, VSD, and right ventricular hypertrophy — the pulmonary valve itself may be stenotic but valvular stenosis is not a core defining criterion.

84. Which of the following are true about Tetralogy of Fallot?

A. **VSD**
B. Lt. Ventricular hypertrophy
C. Lt. Axis deviation
D. Taussig-Blalock shunt is between pulmonary and subclavian artery
E. Morphine is given for cyanotic spell

Ans A d. Taussig-Blalock shunt is between pulmonary and subclavian artery, e. Morphine is given for cyanotic spell Treatment of cyanotic spell involves use of knee chest position (or squatting), oxygen, morphine, sodium bicarbonate, beta blockers (propranolol) and alpha agonists (phenylephrine)

85. The typical radiologic configuration in tetralogy of Fallot as seen in the anteroposterior view consists of all the following EXCEPT

A. **normal heart size**
B. some elevation of the cardiac apex
C. convexity in the region of the main pulmonary artery
D. right-sided aortic arch
E. diminished pulmonary vascularity

Ans A The classic 'boot-shaped heart' (coeur en sabot) of tetralogy of Fallot shows a normal-to-small heart size, upturned apex, concave main pulmonary artery segment, right aortic arch (in ~25%), and diminished pulmonary vascular markings — the heart size itself is typically normal, not enlarged.

Transposition of Great Arteries

86. Holt-Oram syndrome is characterized by?

A. **RA + RV**
B. LA + LV
C. RA + LA
D. LA

Ans A ASD Holt-Oram syndrome is characterized by skeletal abnormalities of hands and arms (upper limbs) and heart problems like ASD; Autosomal dominant inheritance seen.

87. True about Pott shunt:

A. Ascending aorta and right pulmonary artery
B. Left pulmonary artery with descending aorta
C. **Subclavian artery and pulmonary artery**
D. Superior vena cava and right pulmonary artery296 Section 3: Systemic Pediatrics

Ans C The classic Potts shunt anastomoses the descending aorta to the left pulmonary artery (now rarely used due to difficulty controlling shunt size).

88. Modified Jones's criteria includes all the following as major criteria except:

A. Subcutaneous nodules
B. Arthritis
C. **Fever**
D. Carditis

Ans C Fever For Mnemonic,

89. CHARGE association is characterized by the following cardiac feature

A. endocardial cushion defect
B. supravalvular aortic stenosis
C. **tetralogy of Fallot**
D. patent ductus arteriosus
E. anomalous pulmonary venous return

Ans C CHARGE association (coloboma, heart, atresia choanae, retardation, genital and ear anomalies) is associated with TOF, aortic arch and conotruncal anomalies.

90. The following drugs may cause long Q-T syndromes (LQTS) EXCEPT

A. trimethoprim/sulfamethoxazole
B. erythromycin
C. imipramine
D. **pentostam**
E. risperidone

Ans D The other antiprotozoal agent, pentamidine isethionate may cause long Q-T syndrome (LQTS).

91. The following are common side effects of sildenafil EXCEPT

A. flushing
B. **elevated liver function tests**
C. headache
D. diarrhea
E. myalgia

Ans B Flushing, headache, diarrhea, hypotension, fluid retention, exacerbation of heart failure, anemia, elevated liver function tests, and palpitations are common side effects of bosentan.

92. The MOST common long-term complication for those patients who underwent early repair of complete atrioventricular canal before the development of pulmonary vascular disease is

A. **left AV valve regurgitation**
B. subaortic stenosis
C. residual ventricular level shunts
D. complete heart block
E. endocarditis

Ans A Overall, for those patients who underwent early repair before the development of pulmonary vascular disease, the long-term prognosis is good. The most common long-term complication is left AV valve regurgitation, with approximately 5-10% of patients requiring surgical revision for left AV valve repair or replacement during follow-up.

93. In truncus arteriosus, a single arterial trunk arises from the heart and supplies the systemic, pulmonary, and coronary circulations. All the following are true EXCEPT

A. VSD is always present
B. the truncus overriding the defect
C. both ventricles are at systemic pressure
D. **heart failure ensues immediately after birth**
E. clinical cyanosis is usually mild

Ans D When pulmonary vascular resistance is relatively highimmediately after birth, pulmonary blood flow may be normal; as pulmonary resistance drops in the 1st mo of life, blood flow to the lungs is greatly increased and heart failure ensues.

Coarctation of Aorta

94. Inferior Rib notching is seen in?

A. **Coarctation of aorta**
B. Marfan's syndrome
C. Rickets
D. SLE

Ans A Notching of the inferior border of 3rd-9th ribs from pressure erosion by enlarged collateral vessels, is usually seen beyond the age of 3 years, in a child with coarctation of aorta.

95. Coarctation of aorta is most commonly associated with?

A. ASD
B. PDA
C. **Bicuspid aortic valve**
D. VSD

Ans C Coarctation of aorta is associated with a bicuspid aortic valve in more than 70% of patients

96. Coarctation of aorta is associated with?

A. VSD
B. PDA
C. **Bicuspid aortic valve**
D. None of above

Ans C Bicuspid aortic valve Ref. Nelson's 20/621

97. The clinical features associated with coarctation of aorta in older children are the following except:

A. Upper body hypertension (UPSC-II 08)
B. Prominent pulsation in neck
C. Fatiguability and tiredness in legs
D. **Absence of flow murmurs over scapular region**

Ans D

98. All of the following cause death in coarctation of aorta except:

A. Infective endocarditis
B. Heart failure
C. Intracranial hemorrhage
D. Anterior MI
E. **Hypotension**

Ans E The causes of mortality in coarctation of aorta are:
• Hypertension • Premature atherosclerosis • Left ventricular failure
• Aortic dissection & rupture • Cerebral hemorrhage • Infective endocarditis

99. Coarctation of the aorta is common in which syndrome?

A. Down's
B. **Turner's**
C. Klinefelter's
D. Noonan's CYANOTIC CONGENITAL HEART DISEASES

Ans B

100. Of the following, the MOST common clinical sign of coarctation of the aorta in older children is

A. cardiac enlargement
B. notching of the inferior border of the ribs
C. a systolic ejection click or thrill in the suprasternal notch
D. **differential blood pressure: arms > legs**
E. diminished or absent femoral or lower-extremity pulses

Ans D The most common clinical sign of coarctation of the aorta in older children is differential blood pressure: arms > legs (100%).

101. **All the following are signs of coarctation of the aorta EXCEPT**

A. femoral pulses are weak or absent
B. bounding pulses of the arms
C. **femoral pulse occurs slightly before the radial pulse**
D. blood pressure in the legs is lower than that in the arms
E. precordial impulse and heart sounds are usually normal

Ans C The radial and femoral pulses should always be palpated simultaneously for the presence of a radial-femoral delay. Normally, the femoral pulse occurs slightly before the radial pulse. A radial-femoral delay occurs when blood flow to the descending aorta is dependent on collaterals, in which case the femoral pulse is felt after the radial pulse.

102. **All the following are true regarding the diagnosis of coarctation of the aorta EXCEPT**

A. cardiac enlargement are noted with severe coarctation
B. notching of the ribs is common by late childhood
C. doppler is useful for demonstrating the specific site of the obstruction
D. CT and MRI are valuable noninvasive tools for evaluation of coarctation when the echocardiogram is equivocal
E. **diagnostic catheterization is mandatory before surgery**

Ans E In cases that are well defined by echocardiography, CT, or MRI, diagnostic catheterization is not usually required before surgery.

Pulmonary Stenosis & Atresia

103. **Pick up the correct one: Total fetoplacental blood volume at term is estimated to be:**

A. 150 ml/kg body wt
B. 250 ml/kg body wt
C. **350 ml/kg body wt**
D. 450 ml/kg body wt

Ans C fo • Total fetoplacental blood volume at term will be equivalent to cardiac output of newborn
• Cardiac output of a term newborn and the blood going through systemic circulation is 350 ml/kg/minute.

104. **Most common type of TAPVC:**

A. **Supracardiac**
B. Cardiac
C. Infracardiac
D. Mixed

Ans A The supracardiac type, where the pulmonary veins drain via a vertical vein into the innominate vein and superior vena cava, is the most common form of total anomalous pulmonary venous connection.

105. **Right aortic arch is most commonly associated with:**

A. Tetralogy of Fallot
B. **Truncus arteriosus**
C. Corrected TGA 105.
D. Total anomalous pulmonary venous connection

Ans B In Right aortic arch, aorta curves to right, instead of left; Right aortic arch is associated with 30% cases of Truncus arteriosus & 20% cases of TOF.

106. **Most common cause of acquired heart disease in children:**

A. Kawasaki disease
B. Ischaemic heart disease
C. Viral myocarditis
D. **Acute rheumatic fever**

Ans D

107. **The following are characters of venous hum EXCEPT**

A. continuous murmur
B. heard in infraclavicular region
C. grades I-III/VI
D. **louder with patient in supine position**
E. changes with compression of jugular vein

Ans D Louder with patient in upright position.

108. **The MOST common cause of death in competitive athletes is**

A. aortic stenosis
B. **hypertrophic cardiomyopathy**
C. coronary artery abnormalities
D. myocarditis
E. long Q-T syndrome (LQTS)

Ans B The most common cause of death in competitive athletes is hypertrophic cardiomyopathy, with or without obstruction to left ventricular outflow.

109. **In utero heart failure, often with fetal pleural and pericardial effusions and generalized ascites (non-immune hydrops fetalis) may occur in**

A. ventricular septal defect
B. coarctation of aorta
C. d-Transposition of great arteries
D. **Ebstein anomaly**
E. single ventricle

Ans D One notable exception is the case of severe regurgitant lesions, most commonly of the tricuspid valve. In these lesions (e.g., Ebstein anomaly or severe right ventricular outflow obstruction, the parallel fetal circulation cannot compensate for the volume load imposed on the right side of the heart. In utero heart failure, often with fetal pleural and pericardial effusions and generalized ascites (nonimmune hydrops fetalis) may occur.

110. **Severe pulmonary valve stenosis defined as a peak gradient of**

A. >40 mm Hg
B. >45 mm Hg
C. **>50 mm Hg**
D. >55 mm Hg
E. >60 mm Hg

Ans C For those patients with severe stenosis (defined as a peak gradient of >50 mm Hg), the majority ultimately require an intervention, either surgery or balloon valvuloplasty by age 25 yr.

111. Clinical manifestations of hypoplastic left-heart syndrome include

A. cyanosis always obvious in the 1st 48 hr of life
B. deep-blue color skin
C. bounding peripheral pulses
D. **occasional association with Turner syndrome**
E. autosomal recessive inheritance is usual

Ans D Although cyanosis may not always be obvious in the 1st 48 hr of life, a grayish- blue color of the skin is soon apparent and denotes a mix of cyanosis and poor perfusion. All of the peripheral pulses may be weak or absent. This lesion may be isolated or associated in 5-15% of patients with known genetic syndromes, such as Turner syndrome, trisomy 13, 18, or 21, Jacobsen syndrome (11q deletion), Holt-Oram syndrome, andRubinstein-Taybi syndrome.

Heart Failure

112. Most common cardiac anomaly in Turner's syndrome is?

A. Coarctation of aorta
B. **Bicuspid aortic valve**
C. Ventricular septal defect
D. Atrial septal defect

Ans B Most common heart defects in Turner syndrome are bicuspid aortic valves, coarctation of aorta and aortic stenosis.

113. Uncommon finding in congestive cardiac failure in newborn:

A. Congenital aortic stenosis (AIPGMEE 06)
B. Coarctation of aorta
C. Patent ductus arteriosus
D. **Congenital aortoiliac disease**

Ans D Pedal edema

114. A 29-day-old child presents with features of congestive cardiac failure and left ventricular hypertrophy. Auscul- tation shows a short systolic murmur. Most likely diagnosis is:

A. Rheumatic fever
B. Tetralogy of Fallot
C. Transposition of great arteries
D. **Ventricular septal defect**

Ans D Rheumatic fever usually presents between 5-15 years; TOF and TGA are cyanotic heart diseases. VSD patients usually become symptomatic around 6 to 10 weeks age, but a large VSD may manifest at an earlier age.

115. Commonest cause of enlarged cardiac shadow in X-ray of a child is:

A. PDA
B. Coarctation of aorta
C. Pericarditis
D. **Rheumatic carditis HEART FAILURE Questions**

Ans D Rheumatic carditis • Cardiac failure causes cardiomegaly • MC cause of cardiac failure in children (older than infants) is rheumatic fever and rheumatic heart disease. Heart Failure

116. In infants, CHF (congestion heart failure) is diagnosed by:

A. Raised JVP
B. Pedal edema
C. **Tender hepatomegaly**
D. Basal crepts

Ans C Clinical features of heart failure in infants:
• Tachypnea, feeding difficulties, poor weight gain, excessive perspiration, irritability, labored respiration
• Tachycardia, gallop rhythm, wheezing are frequently seen
• Hepatomegaly and cardiomegaly seen; palpation of an enlarged liver is a more reliable sign
• Assessment of jugular venous pressure may be difficult because of shortness of the neck in infants
• Edema may be generalized and usually involves eyelids as well as sacrum and less often the legs and feet

117. Causes of heart failure in newborn are all except:

A. **ASD**
B. TGA with large VSD
C. Hypoplastic left ventricle
D. Baby of diabetic mother

Ans A Discussing the options one by one: a. ASD • A child with an ostium secundum ASD is most often asymptomatic;
• Even an extremely large secundum ASD rarely produces clinically evident heart failure in childhood. b. TGA with large VSD When the VSD is large and not restrictive to ventricular ejection, significant mixing of oxygenated and deoxygenated blood usually occurs and clinical manifestations of cardiac failure are seen c.

118. Commonest cause of heart failure in infancy is:

A. Myocarditis
B. Rheumatic fever
C. Cardiomyopathy
D. **Congenital heart disease OTHER HEART DISEASES**

Ans D Congenital heart disease • The most common cause of Congestive heart failure (CHF) in infants— Congenital heart disease
• The most common cause of CHF in the older children— Rheumatic fever and rheumatic heart disease.

119. An infant presents with cardiac failure. Examination reveals a weaker femoral pulse when compared to the 171. radial pulse. What is the probable diagnosis?

A. Ventricular septal defect with aortic stenosis
B. **Ruptured sinus of valsalva aneurysm**
C. Atrial septal defect with mitral stenosis
D. Complete common atrioventricular canal Chapter 13: Pediatric Cardiology 301 Answers
E. Pheochromocytoma Children born to mothers with systemic lupus erythema- tosus are likely to have one of the following anomalies: (Karnataka 03)

Ans B Coarctation of aorta

120. All the following are causes of heart failure in full-term neonate EXCEPT

A. asphyxial cardiomyopathy
B. coarctation of aorta
C. hypoplastic left heart
D. transposition of great arteries
E. **ventricular septal defect**

Ans E Fluid overload, PDA, VSD, and Cor pulmonale (BPD) are causes of heart failure in premature neonate.

121. One of the following is a sign of right-sided heart failure
A. edema
B. tachypnea
C. orthopnea
D. wheezing
E. pulmonary edema

Ans A If left-sided failure is predominant, tachypnea, orthopnea, wheezing, and pulmonary edema are seen. Hepatomegaly, edema, and distended neck veins are signs of right-sided failure.

122. The following are causes of congestive heart failure and cardiomegaly during the newborn period but no murmur EXCEPT
A. endocardial fibroelastosis
B. asphyxia
C. glycogen storage disease (Pompe disease)
D. paroxysmal supraventricular tachycardia
E. sepsis

Ans A is a cause after the neonatal period.

123. In urgent situations of supraventricular tachycardia (SVT) when symptoms of severe heart failure have already occurred. Of the following, the initial management is
A. adenosine by rapid intravenous push
B. DC cardioversion (0.5-2 J/kg)
C. verapamil
D. digoxin
E. amiodarone

Ans B In urgent situations when symptoms of severe heart failure have already occurred, synchronized DC cardioversion (0.5-2 J/kg) is recommended as the initial management.

124. Afterload reducers are not indicated in children with heart failure secondary to
A. cardiomyopathy
B. severe mitral insufficiency
C. aortic insufficiency
D. ventricular septal defect
E. aortic stenosis

Ans E ACEIs and ARBs are not generally used in the presence of stenotic lesions of the left ventricular outflow tract because of concern over coronary perfusion.

Infective Endocarditis

125. Infective endocarditis is most commonly seen in?
A. ASD
B. VSD
C. PDA
D. Pulmonary Stenosis

Ans B Most common congenital heart disease is & Most common congenital heart disease affected by infective endocarditis is are VSD

126. Patent truncus arteriosus leads to:
A. Both ventricular hypertrophy
B. Pulmonary oligemia
C. Ductus dependent flow
D. CCF at birth

Ans A • In truncus arteriosus, both ventricles are at systemic pressure & both eject blood into the truncus.
• When pulmonary vascular resistance is relatively high immediately after birth, pulmonary blood flow may be normal;
• As pulmonary resistance drops in 1st month of life, blood flow to the lungs is greatly increased → heart failure
• Cardiac enlargement develops over 1st several weeks of life, as a result of prominence of both ventricles.

127. Max limit of cardiothoracic ratio in children:
A. 0.40
B. 0.45
C. 0.50
D. 0.55

Ans C When maximal cardiac width is > half the maximal chest width (cardiothoracic ratio >50%), the heart is usually enlarged.

128. Infective endocarditis is rarely seen in:
A. VSD
B. ASD
C. PDA
D. RHD

Ans B Risk of infective endocarditis in various lesions: High risk Moderate risk Low risk
• Prosthetic heart valve • AS • Mitral valve prolapse • PS • Mitral valve prolapse
• Tetralogy of Fallot • VSD with MR • MS • ASD
• PDA • MR • TS
• AR • Coarctation of aorta • TR

129. The following cardiac lesions are at increased risk for bacterial endocarditis EXCEPT
A. mitral insufficiency
B. aortic stenosis
C. atrial septal defect seccundum
D. coarctation of the aorta
E. patent ductus arteriosus

Ans C

130. The leading causative agents for endocarditis in pediatric patients are
A. group D enterococci
B. viridans-type streptococci
C. Pseudomonas aeruginosa
D. fungal organisms
E. Serratia marcescens

Ans B (α-hemolytic streptococci) and Staphylococcus aureus remain the leading causative agents for endocarditis in pediatric patients.

131. The following is a major Duke criterion for the diagnosis of endocarditis

A. new valve regurgitant flow by echocardiography
B. Osler nodes
C. single positive blood culture
D. serologic evidence of infection
E. high erythrocyte sedimentation rate

Ans A The Duke criteria help in the diagnosis of endocarditis. Major criteria include (1) positive blood cultures (2 separate cultures for a usual pathogen, 2 or more for less typical pathogens), and (2) evidence of endocarditis on echocardiography (intracardiac mass on a valve or other site, regurgitant flow near a prosthesis, abscess, partial dehiscence of prosthetic valves, or new valve regurgitant flow).

132. Polysplenia (left isomerism) is characterized by

A. right-sided stomach
B. decreased pulmonary blood flow
C. absent gallbladder
D. howell-jolly bodies
E. severe cyanosis

Ans C All other options are features of asplenia (right isomerism).

133. In 90% of cases of endocarditis, the causative agent is recovered from the first 2 blood cultures. Antimicrobial pretreatment of the patient reduces the yield of blood cultures to

A. 10%
B. 30%
C. 40%
D. 60%
E. 80%

Ans D Antimicrobial pretreatment of the patient reduces the yield of blood cultures to 50-60%. The microbiology laboratoryshould be notified if the patient has received antibiotics so thatmore sophisticated methods can be used to recover the offending agent.

134. Despite the use of antibiotic agents, mortality remains high, in the range of 20-25%. Serious morbidity occurs in 50-60% of children with documented infective endocarditis. Of the following, the MOST common morbidity is

A. heart failure
B. pulmonary emboli
C. mycotic aneurysms
D. acquired ventricular septal defect
E. heart block

Ans A The most common is heart failure caused by vegetations involving the aortic or mitral valve. Myocardial abscesses and toxic myocarditis may also lead to heart failure without characteristic changes in auscultatory findings and, occasionally, to life- threatening arrhythmias.

Rheumatic Fever & RHD

135. Down syndrome is most commonly associated with?

A. ASD with ostium secundum
B. ASD with ostium primum
C. VSD
D. TOF

Ans B Down Syndrome by Mark Selikowitz p 87 It is a controversial question; There is no doubt about the most common cardiac lesion in Down syndrome, confusion is about the 2nd most common disease... this is what most references say (but obviously, not very clearly).. Most common heart disease in Down syndrome is: Atrioventricular septal defect > Ostium primum ASD (partial AVSD) > VSD > Ostium Secundum ASD

136. Differential cyanosis occurs in which disease?

A. Severe coarctation
B. Interrupted aortic arch
C. PPHN
D. All of the above

Ans D In severe coarctation and interrupted aortic arch, lesser amount of blood goes to lower limbs, as compared to upper limbs. In Persistent pulmonary hypertension of newborn (PPHN), deoxygenated blood flows from pulmonary artery to descending aorta. So in all these 3 conditions, cyanosis may be seen in lower limbs, but not in upper limbs (differential cyanosis), as upper limbs get adequate amount of oxygenated blood.

137. In which of the following a 'Coeur en Sabot' shape of the heart is seen:

A. Sodium bicarbonate
B. Propranolol
C. Phenylephrine
D. Isoprenaline

Ans D Tetralogy of Fallot

138. All of the following are true regarding the jones criteria as revised in 2015 by the American heart association, except:

A. Describes 5 Major criteria, 4 minor criteria and a requirement of evidence of recent group A streptococcal infection
B. Diagnosis of recurrent of acute rheumatic fever in Moderate/high risk population requires 2 minor criteria and evidence of preceding GAS infection
C. Has separate criteria for low risk and moderate/high risk population
D. Acute rheumatic fever is diagnosed in the presence of 2 major or 1 major and 2 minor criteria with evidence of preceding GAS infection

Ans B Diagnosis of recurrent of acute rheumatic fever in Moderate/high risk population requires 2 minor criteria and evidence of preceding GAS infection Changes in 2015 AHA revision of modified Jone's criteria include:
• Separate criteria for Low-Risk populations (defined as those with incidence ≤ 2 per 100,000 school-age children per year or all-age rheumatic heart disease prevalence of ≤1 per 1000 population) and Moderate/High-Risk populations (defined as those with higher incidence or prevalence rates).

139. **Which of the following is a major criteria in the diagnosis of rheumatic fever?**

A. Erythema nodosum
B. Erythema multiforme
C. Erythema toxicum
D. **Erythema marginatum**

Ans D

140. **A girl child comes with complaints of involuntary movements and is diagnosed as a case of Sydenham chorea, suggestive of a diagnosis of acute rheumatic fever. There is no complaint of carditis or arthritis. Throat culture is negative. Which of the following will most likely suggest recent streptococcal infection?**

A. Antistreptolysin S
B. **ASLO**
C. PCR for M protein
D. Antihyaluronidase

Ans B

141. **To prevent acute rheumatic fever, acute pharyngitis due to group A streptococci should be treated with antibiotics before:**

A. 7 days of illness
B. 8 days of illness
C. **9 days of illness**
D. 10 days of illness

Ans C Antibiotic therapy for patients with GAS pharyngitis can prevent acute rheumatic fever, shorten the clinical course of the illness, reduce transmission of the infection to others, and prevent suppurative complications. When therapy is instituted within 9 days of onset of symptoms and continued for the full course, acute rheumatic fever is almost always prevented.

142. **All are minor Jones criteria except:**

A. Arthralgia
B. Fever
C. Prolonged PR interval
D. **Chorea**

Ans D

143. **Major criteria for rheumatic fever are all except:**

A. Carditis
B. Erythema marginatum
C. Migratory polyarthrits
D. **Increased CRP**

Ans D

144. **Which is not a major criteria of Jones in rheumatic fever?**

A. Pancarditis
B. Arthritis
C. Chorea
D. **Elevated ESR**

Ans D

145. **A 7-year-old child with rheumatic heart disease presents with pallor, fever and a palpable spleen. The following investigations would be needed to arrive at a diagnosis except:**

A. **Electrocardiogram**
B. Echocardiogram
C. Blood culture
D. Urine examination

Ans A Infective endocarditis should be suspected in presence of pallor, fever and splenomegaly in a known case of RHD
• Urine examination shows albuminuria and microscopic hematuria
• Blood culture should be done for diagnosis of the causative agent
• Echocardiography is done to detect the cardiac changes like vegetations.

146. **Not a minor criteria for acute rheumatic fever:**

A. Arthralgia
B. Increased ESR
C. Increased PR interval
D. **ASO titer**

Ans D ASO titre

147. **Major criteria for Rheumatic fever:**

A. Fever
B. **Erythema marginatum**
C. ECG changes
D. Positive throat culture

Ans B

148. **Which of the following is a minor criteria for diagnosis of rheumatic fever (RF) according to modified Jones criteria:**

A. ASO titer
B. Past history of rheumatic fever
C. **Fever**
D. Subcutaneous nodules

Ans C

149. **Drug of choice for rheumatic fever prophylaxis in penicillin allergic patient:**

A. **Erythromycin**
B. Clindamycin
C. Vancomycin
D. Gentamycin

Ans A For a patient who is allergic to both penicillin and sulphonamides, a macrolide (erythromycin or clarithromycin or azithromycin) may be used. Recommendations of American heart association for the duration of secondary prophylaxis Category: Rheumatic fever Duration of secondary prophylaxis Without carditis 5 years or until 21 years of age, whichever is longer With carditis but without residual heart disease 10 years or until 21 years, whichever is longer

150.In a patient of rheumatic carditis, full dose of steroid is given for:

A. 3 weeks
B. 6 weeks
C. 9 weeks
D. 12 weeks

Ans A about corticosteroids in rheumatic carditis
• Indication: Patients with carditis and more than minimal cardiomegaly and/or congestive heart failure
• Usual dose: Prednisone 2 mg/kg/day in 4 divided doses for 2-3 wk followed by half the dose for 2-3 wk and then tapering of the dose by 5 mg/24 hr every 2-3 days.
• When prednisone is being tapered, aspirin should be started at 50 mg/kg/day in 4 divided doses for 6 wk fo 141. a. Polyarthritis, d.

151. True statement about rheumatic fever in children:

A. Polyarthritis (PGI Dec 03)
B. Caused by alpha hemolytic streptococci
C. Erythema marginatum is most common manifestation
D. MC valve involvement is Mitral
E. Erythema marginatum is common in face

Ans D The mitral valve is the most commonly affected valve in rheumatic fever/rheumatic heart disease in children, typically causing mitral regurgitation.

152.Which of the following symptoms of rheumatic fever may be the only isolated manifestation?

A. Carditis
B. Arthritis
C. Chorea
D. Subcutaneous nodules

Ans C 3 circumstances in which strict application of diagnostic criteria of Rheumatic fever is not needed are: Chorea, Indolent carditis and Recurrence of Rheumatic fever.

Myocarditis & Cardiomyopathy

153.Most common syndrome associated with AV canal defect?

A. Down syndrome
B. Klinefelter syndrome
C. Turner syndrome
D. Marfan syndrome

Ans A

154.Pulmonary plethora is not seen in:

A. VSD
B. TOF
C. Truncus arteriosus
D. TAPVC

Ans B In TOF, pulmonary oligemia i.e. decreased pulmonary vascularity is seen and not pulmonary plethora.

155.Cyanosis is seen in:

A. Patent ductus arteriosus
B. Tricuspid atresia
C. Ostium primum ASD
D. Eisenmenger complex
E. Tetralogy of Fallot

Ans B d. Eisenmenger complex, e. Tetralogy of Fallot

156.The V wave of jugular venous pulses is produced due to:

A. Atrial systole preceding tricuspid valve closure
B. Descent of the tricuspid valve ring
C. Closure of tricuspid valve
D. Passive filing of atrium during ventricular systole

Ans D Passive filling of atrium during ventricular systole Normal waves in jugular venous pulse Name of Underlying cause wave with a wave Atrial contraction c wave Transmission of carotid arterial impulse through jugular veins or by bulging of tricuspid valve cusps into right atrium in early systole. v wave Filling up of Right atrium in late systole and early diastole. x wave Atrium relaxes and tricuspid valve moves downward y wave Filling of ventricle after tricuspid opening

157. Which of the following drugs is not given in dilated cardiomyopathy?

A. Beta blocker
B. Calcium channel blocker
C. Spironolactone
D. ACE inhibitors

Ans B Treatment of Dilated Cardiomyopathy:
• Drugs used include diuretics, ACE inhibitors, angiotensin receptor blockers, digitalis, β-blockers (carvedilol or metoprolol)
• In patients presenting with extreme degrees of heart failure or circulatory collapse, intensive care measures are often required, including IV inotropes, diuretics, mechanical ventilatory support ± circulatory support, which may include ventricular assist devices, extracorporeal membrane oxygenation (ECMO), and ultimately cardiac transplantation.

158.Cardiomyopathy is not a feature of?

A. Duchenne muscular dystrophy
B. Friedreich's ataxia
C. Pompe disease
D. Lowe's syndrome

Ans D

159.Hypertrophic cardiomyopathy is a recognized association with

A. infant of diabetic mother
B. Marfan syndrome
C. William syndrome
D. trisomy 21 (Down syndrome)
E. XO (Turner syndrome)

Ans A Marfan syndrome is associated with dilated and dissecting aorta, aortic valve regurgitation, mitral valve prolapsed, William syndrome with supravalvular aortic stenosis, peripheral pulmonary stenosis, Trisomy 21 (Down syndrome) with endocardial cushion defect, VSD, ASD, PDA, while XO (Turner syndrome) associated with coarctation of aorta, aortic stenosis.

160.Symptoms in patients with aortic stenosis depend on the severity of the obstruction. Critical aortic stenosisis associated with the following EXCEPT

A. cardiomegaly
B. pulmonary edema
C. weak pulses in all extremities
D. pale skin
E. maximum intensity of the murmur

Ans E If cardiac output is significantly decreased, the intensity of the murmur at the right upper sternal border may be minimal.

161. **Pentalogy of Cantrell consists of the following EXCEPT**

A. ectopia cordis
B. midline supra umbilical abdominal defect
C. deficiency of the anterior diaphragm
D. **defect of the upper sternum**
E. intracardiac defect

Ans D Defect of the lower sternum.

162. **Anthracycline cardiotoxicity (doxorubicin [Adriamycin]) on rare occasion causes acute inflammatory myocardial injury, but more classically results in dilated cardiomyopathy (DCM) and occurs in up to 30% of patients given a cumulative dose of doxorubicin exceeding**

A. 250 mg/m2
B. 350 mg/m2
C. 450 mg/m2
D. **550 mg/m2**
E. 650 mg/m2

Ans D Exceeding 550 mg/m2. The risk of toxicity appears to be exacerbated by concomitant radiation therapy.

163. **Restrictive cardiomyopathy (RCM) accounts for <5% of cardiomyopathy cases. RCM is characterized by all the following EXCEPT**

A. normal ventricular chamber dimensions
B. normal myocardial wall thickness
C. preserved systolic function
D. **normal atrial chamber dimensions**
E. high ventricular diastolic pressure

Ans D Dramatic atrial dilation can occur as a result of the abnormal myocardial compliance and high ventricular diastolic pressure.

164. **In Asia, the following hepatitis virus appears to be a significant cause of viral myocarditis**

A. hepatitis A virus
B. hepatitis B virus
C. **hepatitis C virus**
D. hepatitis D virus
E. hepatitis E virus

Ans C Coxsackievirus and other enteroviruses, adenovirus, parvovirus, Epstein-Barr virus, parechovirus, influenza virus, and cytomegalovirus are the most common causative agents in children, though most known viral agents have been reported. In Asia, hepatitis C virus appears to be significant as well.

Arrhythmias

165. **Wide fixed S2 is seen in:**

A. **ASD**
B. VSD
C. PDA
D. All of above

Ans A Auscultatory findings in ASD are Wide fixed splitting of S2, Shunt remains silent, Ejection systolic murmur in pulmonary area, Mid-diastolic murmur in tricuspid area.

166. **WPW syndrome is associated with:**

A. **Ebstein anomaly**
B. TOF
C. VSD
D. TAPVC

Ans A Wolff-Parkinson White syndrome may be present in patients with Ebstein anomaly and these patients may have episodes of supraventricular tachycardia.

167. **Pott's shunt is:**

A. Rt subclavian artery to right pulmonary artery
B. **Descending aorta to left pulmonary artery**
C. Left subclavian to left pulmonary artery
D. Ascending aorta to right pulmonary artery

Ans B

168. **What is the approximate Total fetal cardiac output?**

A. 300 ml/kg/min
B. **450 ml/kg/min**
C. 600 ml/kg/min
D. 150 ml/kg/min

Ans B Total fetal cardiac output, i.e. combined output of both left and right ventricles—is ≈450 mL/kg/min. 65% of descending aortic blood flow returns to the placenta and remaining 35% perfuses the fetal organs and tissues.

169. **Chest pain in the pediatric patient often generates a significant amount of patient and parental concern. Of the following, the MOST common cause is**

A. myocarditis
B. pericarditis
C. pleurisy
D. pneumothorax
E. **pneumonia**

Ans E Common causes are costochondritis, Trauma or muscle overuse/strain, asthma (often exercise-induced), severe cough, pneumonia, reflux esophagitis, anxiety, hyperventilation, precordial catch syndrome (Texidor's twinge), sickle cell vasoocclusive crisis, and idiopathic.

170. **SVT in children differ from physiologic sinus tachycardia by all the following EXCEPT**

A. sudden onset and termination
B. persistent ventricular rate of >180 bpm
C. fixed RR interval on ECG
D. abnormal P-wave shape or axis or absent P waves
E. **evident change in heart rate with activity**

Ans E Little change in heart rate with activity, crying, or breath holding.

171. **Sinus bradycardia is due to slow discharge of impulses from the sinus node, the heart's natural pacemaker. Sinus bradycardia in neonates is considered if sinus rate is**

A. <100 beats/min
B. **<90 beats/min**
C. <80 beats/min
D. <70 beats/min
E. <60 beats/min

Ans B Sinus rate <90 beats/min in neonates and <60 beats/min in older children is considered to be sinus bradycardia.

172. **The following procedures may abort an attack of SVT EXCEPT**

A. placing of the face in ice water
B. straining
C. breath holding
D. standing on head
E. **ocular pressure**

Ans E must never be performed, and carotid sinus massage is very rarely effective.

173. **Phenytoin is used for treatment of**

A. supraventricular tachycardia
B. atrial fibrillation
C. atrial flutter
D. ventricular tachycardia
E. **digitalis intoxication**

Ans E Other options are treated by procainamide.

174. **Ventricular tachycardia (VT) is defined as at least 3 premature ventricular contractions PVCs at >120 beats/min. Hemodynamically unstable patients with VT should be immediately treated with**

A. catheter ablation
B. ICD implantation
C. **DC cardioversion**
D. amiodarone
E. procainamide

Ans C

Hypertension

175. **A ten-year-old boy presents to the pediatric emergency unit with seizures. Blood pressure in the upper extremity measured as 200/140 mm Hg. Femoral pulses were not palpable. The most likely diagnosis amongst the following is:**

A. Takayasu aortoarteritis
B. Renal parenchymal disease
C. Grand mal seizures
D. **Coarctation of aorta**

Ans D Isolated upper extremity hypertension (200/140 mmHg), together with absent/diminished femoral pulses is a characteristic feature of coarctation of aorta.

176. **Reversal of shunt is not possible in natural history of?**

A. ASD
B. VSD
C. **TOF**
D. PDA

Ans C • Reversal of shunt or Eisenmenger syndrome is seen in– VSD, ASD, AV canal defect, PDA.

177. **A patient with cyanosis presents with LVH and pulmonary complications. ECG, shows left axis deviation. Most likely diagnosis is:**

A. TOF
B. **Tricuspid atresia**
C. TAPVC
D. VSD

Ans B

178. **The ECG shows left ventricular hypertrophy and a superior QRS axis (between 0° and –90°) in**

A. **tricuspid atresia**
B. Ebstein anomaly
C. pulmonary atresia
D. pulmonary stenosis
E. tetralogy of Fallot

Ans A

179. **All the following are causes of chronic pulmonary venous hypertension EXCEPT**

A. congenital mitral stenosis
B. total anomalous pulmonary venous return with obstruction
C. **peripheral pulmonary stenosis**
D. left atrial myxomas
E. cor triatriatum

Ans C A variety of lesions may give rise to chronic pulmonary venous hypertension, which when extreme may result in pulmonary arterial hypertension and right-sided heart failure. These lesions include congenital mitral stenosis, mitral insufficiency, total anomalous pulmonary venous return with obstruction, left atrial myxomas, cor triatriatum (stenosis of a common pulmonary vein), individual pulmonary vein stenosis, and supravalvular mitral rings.

180. **The predominant manifestations of primary pulmonary hypertension include the following EXCEPT**

A. exercise intolerance
B. precordial chest pain
C. syncope
D. **low arterial oxygen saturation**
E. dizziness

Ans D Arterial oxygen saturation is usually normal unless there is an associated intracardiac shunt.

181. **Postoperative (cardiac surgery) pulmonary hypertension can be managed with**

A. **hyperventilation and inhaled nitric oxide**
B. catecholamines
C. phosphodiesterase inhibitors
D. nitroprusside
E. diuretics

Ans A

182. **The MOST common cause of pulmonary hypertension in pediatric patients is**

A. **idiopathic pulmonary arterial hypertension**
B. pulmonary venoocclusive disease
C. left-sided valvular heart disease
D. chronic obstructive pulmonary disease
E. interstitial lung disease

Ans A Idiopathic or familial disease is the most common in pediatric patients (~55%), followed by pulmonary hypertension secondary to congenital heart disease (~35%) and chronic respiratory disorders (~15%).

183. **The definition of hypertension in children is**

A. average systolic blood pressure (SBP) and/or diastolic BP that is ≥95th percentile for age, sex, on ≥3 occasions
B. average systolic blood pressure (SBP) and/or diastolic BP that is ≥95th percentile for age, sex, and height on ≥2 occasions
C. average systolic blood pressure (SBP) and/or diastolic BP that is ≥95th percentile for age, sex, and height on ≥3 occasions
D. average systolic blood pressure (SBP) and/or diastolic BP that is ≥90th percentile for age, sex, and height on ≥3 occasions
E. average systolic blood pressure (SBP) and/or diastolic BP that is ≥99th percentile for age, sex, and height on ≥3 occasions

Ans C The National High Blood Pressure Education Program Working Group on High Blood Pressure in Children and Adolescents published the Fourth Report on the Diagnosis, Evaluation, and Treatment of High Blood Pressure in Children and Adolescents (Fourth Report) in 2004. The Fourth Report defined hypertension as average systolic blood pressure (SBP) and/or diastolic BP that is ≥95th percentile for age, sex, and height on ≥3 occasions.

184. **Renal childhood diseases responsible for chronic hypertension include**

A. hemolytic-uremic syndrome
B. acute tubular necrosis
C. congenital dysplastic kidney
D. pyelonephritis
E. renal trauma

Ans C Other options are responsible for acute/ intermittent hypertension.

185. **Measuring serum potassium is essential in a child with confirmed hypertension because hyperkalemia may be seen in**

A. Gordon syndrome
B. Liddle syndrome
C. glucocorticoid remedial aldosteronism
D. apparent mineralcorticoid excess
E. pheochromocytoma

Ans A Measuring serum potassium is essential because hypokalemia may be present in Liddle syndrome, glucocorticoidremedial aldosteronism, and apparent mineralcorticoid excesssyndrome, while hyperkalemia may be seen in Gordon syndrome.

Kawasaki Disease

186. **Ductus arteriosus closes in response to:**

A. Decrease in peripheral oxygen saturation
B. Indomethacin therapy
C. Prostaglandin E1
D. Increase in pulmonary vascular resistance

Ans B Indomethacin, a Prostaglandin inhibitor is used in medical closure of Ductus arteriosus.

187. **Ebstein anomaly is due to maternal intake of:**

A. Lithium
B. Copper
C. Cobalt
D. Nickel

Ans A Maternal drug Congenital heart disease Phenytoin VSD, ASD, coarctation of aorta, PDA Valproate Coarctation of aorta, aortic stenosis, pulmonary atresia, VSD Lithium Ebstein anomaly, Tricuspid atresia Alcohol ASD, VSD

188. **A newborn presents with cyanosis. The X-ray chest reveals oligemic lung fields and normal sized heart. Most likely diagnosis is:**

A. Ebstein's anomaly
B. Pulmonary atresia
C. Transposition of great arteries
D. Tetralogy of Fallot

Ans B In Pulmonary atresia, child is cyanotic at birth and there is pulmonary oligemia.

189. **What is the minimal internal diameter of Giant coronary aneurysm in children with Kawasaki disease which pose highest risk for ruptures, thrombosis. Stenosis and myocardial infarction?**

A. 8 mm
B. 10 mm
C. 12 mm
D. 15 mm

Ans A Giant coronary artery aneurysms (classic definition of > 8 mm internal diameter) pose the greatest risk for rupture, thrombosis or stenosis, and myocardial infarction

190. **The MOST common cause of syncope in children is**

A. Wolff-Parkinson-White syndrome
B. prolonged QT syndrome
C. atrioventricular block
D. neurocardiogenic syncope
E. myocarditis

Ans D In otherwise healthy children, neurocardiogenic syncope is most common. This entity goes by a number of terms, including vasovagal syncope, neurally mediated syncope, and autonomic syncope.

191. **The following factors are most strongly associated with the development of coronary artery disease in patients with Kawasaki disease EXCEPT**

A. duration of fever of >16 days
B. first-degree heart block
C. cardiomegaly
D. male gender
E. age of <1 year

Ans B Dysrhythmias (other than first-degree heart block) is an associated factor.

192. **Patients who have undergone surgery entailing the use of cardiopulmonary bypass, should be watched carefully for**

A. epilepsy
B. **learning disabilities**
C. anemia
D. pulmonary hypertension
E. arrhythmias

Ans B Patients who have undergone surgery entailing the use of cardiopulmonary bypass, especially in the newborn period, should be watched carefully during their early school years for signs of mild to moderate learning disabilities, which are often amenable to early remedial intervention. The risk is higher in patients who have undergone repair using hypothermic total circulatory arrest than in those where systemic blood flow is maintained using cardiopulmonary bypass.

193. **Shone complex consist of the following EXCEPT**

A. coarctation of the aorta
B. subvalvar aortic stenosis
C. valvar aortic stenosis
D. **pulmonic stenosis**
E. mitral stenosis

Ans D Congenital mitral stenosis is a rare anomaly that can be isolated or associated with other defects, the most common being subvalvar and valvar aortic stenosis and coarctation of the aorta (Shone complex).

194. **Side effects of propranolol may include**

A. hypothyroidism
B. elevated triglycerides
C. **school performance problems**
D. hepatic toxicity
E. pulmonary fibrosis

Ans C Other options are side effects of amiodarone.

Pericardial Diseases

195. **In which of the following conditions left atrium is not enlarged?**

A. Ventricular septal defect
B. **Atrial septal defect**
C. Aortopulmonary window
D. Patent ductus arteriosus

Ans B with
• ASD is associated with an enlarged right atrium and right ventricle to accommodate large volume of blood
• Left atrium is not enlarged in ASD, because it decompresses itself by shunting its blood to right atrium.

196. **Double arch of aorta is seen in:**

A. CATCH22
B. Di George syndrome
C. Velo cardiofacial syndrome
D. **All of the above**

Ans D All of above Double aortic arch
• It is associated with a chromosome 22q11 deletion in approximately 20% of patients
• 22q11 deletion is responsible for DiGeorge, velocardiofacial, and conotruncal anomaly face syndromes, which are often referred to using the unified terms CATCH-22 syndrome
• It is also associated with VACTERL (vertebral, anal, cardiac, tracheal, esophageal, renal, and limb abnormalities) or CHARGE (posterior coloboma, heart defect, choanal atresia, retardation, genital, and ear) associations.

197. **A five-year-old child presents with left ventricular hypertrophy and central cyanosis; What is the most probable diagnosis:**

A. **Tricuspid atresia**
B. Eisenmenger syndrome
C. Tetrology of Fallot
D. Total anomalous pulmonary venous drainage

Ans A The combination of cyanosis and left axis deviation is highly suggestive of tricuspid atresia.

198. **Least common cause of pericarditis/pericardial effusion in children:**

A. Rheumatic fever
B. **Sarcoidosis**
C. Rheumatoid arthritis
D. SLE

Ans B Important causes of pericarditis/pericardial effusion in children: Infectious
• Viral (Coxsackie virus B, EBV, Influenza, Adenovirus); Fungal (Histoplasmosis, Actinomycosis)
• Bacterial (TB, Streptococcus, Pneumococcus, Staphylococcus, Meningococcus, Mycoplasma, Tularemia, Listeria, Leptospirosis)
• Immune complex (Meningococcus, Hemophilus influenzae)
• Parasitic (Toxoplasmosis, Echinococcosis) Connective tissue diseases Metabolic-endocrine Rheumatoid arthritis Systemic sclerosis Uremia

199. **The following are major side effects of PGE1 EXCEPT**

A. apnea
B. fever
C. cutaneous flushing
D. seizures
E. **hypertension**

Ans E Apnea, fever, cutaneous flushing, seizures, hypotension, and bradycardia/tachycardia.

200. **The lower limit of pulse rate in neonate at rest is**

A. 50/min
B. 60/min
C. **70/min**
D. 80/min
E. 90/min

Ans C Lower limit of normal is 70/min, average 125/min, and upper limit is 190/min.

201.The postpericardiotomy syndrome is characterized by all the following EXCEPT

A. fever
B. decreased appetite
C. listlessness
D. vomiting
E. chest pain

Ans E The postpericardiotomy syndrome may occur toward the end of the 1st postoperative week or may sometimes be delayed until weeks or months after surgery. This febrile illness is characterized by fever, decreased appetite, listlessness, nausea, and vomiting. Chest pain is not always present, so a high index of suspicion should be maintained in any recently postoperative patient.

202.The severity of symptoms of Ebstein anomaly and the degree of cyanosis are highly variable and depend on the extent of

A. cardiac dysrhythmias
B. displacement of the tricuspid valve
C. atrial right-to-left shunt
D. pulmonary vascular resistance
E. polycythemia

Ans B The severity of symptoms and the degree of cyanosis are highly variable and depend on the extent of displacement of the tricuspid valve and the severity of right ventricular outflow tract obstruction.

203.The following criteria are indications for further investigation of premature ventricular contractions (PVCs) that could require suppressive therapy EXCEPT

A. multiform PVCs
B. disappear during exercise
C. R-on-T phenomenon
D. extreme frequency of beats
E. presence of underlying heart disease

Ans B It is important to distinguish PVCs that are benign from those that are likely to lead to more severe arrhythmias. The former usually disappear during the tachycardia of exercise. If they persist or become more frequent during exercise, the arrhythmia may have greater significance.

Cardiac Examination & Murmur

204.A young female presents with history of dyspnea on exertion. On examination, she has wide, fixed split S2 with ejection systolic murmur (III/VI) in left second intercostal space. Her ECG shows left axis deviation. The most probable diagnosis is:

A. Total anomalous pulmonary venous drainage
B. Tricuspid atresia
C. Ostium primum atrial septal defect
D. Ventricular septal defect with pulmonary arterial hyper-tension

Ans C • Wide fixed splitting of S2 in association with ejection systolic murmur suggests the diagnosis of atrial septal defect
• The presence of left axis deviation in A.S.D., suggests ostium primum type of defect.

205.A neonate has recurrent attacks of restlessness, irritability Questions and diaphoresis on feeding. Cardiac auscult ation reveals a nonspecific murmur. He is believed to be at risk for M.I. Likely diagnosis here is:

A. ASD
B. VSD
C. TOF
D. Anomalous coronary artery

Ans D Heart failure in a neonate with recurrent attacks of discomfort, restlessness, irritability, sweating, dyspnea and pallor and increased risk for Myocardial infarction is seen in Anomalous origin of Left Coronary Artery from Pulmonary Artery (ALCAPA);

206.Single second heart sound (S2) is heard in:

A. Transposition of great vessels
B. Ebstein's anomaly
C. Tetralogy of Fallot
D. Eisenmenger's syndrome

Ans C In Tetralogy of Fallot, P2 is soft & inaudible, giving rise to single second heart sound. dna 67. b. Left pulmonary artery with descending aorta

207.A 2 weeks baby with central cyanosis has grade II systolic murmur, normal S1, single S2, plethoric lung. What is the diagnosis?

A. TGA
B. TAPVC
C. TOF
D. Pulmonary atresia

Ans A • The given clinical picture suggests a diagnosis of TGA (Transposition of great arteries)
• In TOF and Pulmonary atresia, pulmonary vascularity is decreased
• In TAPVC, S2 is not single, rather there is a wide fixed split of the 2nd heart sound.

208.A two-month-old infant is brought to the hospital emergency with marked respiratory distress. On examination, the infant has cyanosis and bilateral crepitations. Heart rate is 180/min, respiratory rate 56/min and the liver span 7.5 cm. The child has had repeated episodes of fever, cough and respiratory distress since the time of birth. 103. Cardiovascular examination reveals a grade III ejection systolic murmur in left parasternal area and the chest X-ray reveals cardiomegaly with a narrow base and plethoric lung fields. What is the most likely diagnosis?

A. Congenital methemoglobinemia (UPSC-1 09)
B. Transposition of great arteries

Ans B In the given case scenario, an infant has features of heart failure, cyanosis with repeated episodes of pneumonia suggesting increased pulmonary blood flow. These clinical features along with the chest X ray findings are suggestive of Transposition of great arteries.

209.dusky. On examination, there was central cyanosis and clubbing. There was no pallor, oedema or respiratory distress. The heart was normal sized with a parasternal heave. A systolic thrill was palpable over the left middle sternal border. First heart sound was normal and only the aortic component was audible in the second heart sound. Liver was not enlarged. What would be the likely Questions diagnosis?

A. VSD
B. PDA
C. **ASD**
D. Tetralogy of Fallot
E. Ebstein anomaly The following features are true for tetralogy of Fallot, except: (AIPGMEE 06)

Ans C Truncus arteriosus

210.A neonate has central cyanosis and short systolic murmur on the 2nd day of birth. The diagnosis is:

A. Tetralogy of Fallot (AIIMS May 01)
B. **Transposition of great vessels**
C. Atrial septal defect
D. Ventricular septal defect

Ans B ASD and VSD are acyanotic heart diseases and TOF usually does not present with cyanosis in neonatal period.

211. Causes of wide fixed splitting of second heart sound include all except:

A. Atrial septal defect
B. **Ebstein anomaly**
C. Total anomalous pulmonary venous return
D. Right bundle branch block

Ans B Wide fixed splitting of second heart sound is seen in Atrial septal defect, Total anomalous pulmonary venous return (TAPVC) and Right bundle branch block (RBBB)

212.Diastolic dysfunction and normal systolic function are recognized features of

A. dilated cardiomyopathy
B. hypertrophic cardiomyopathy
C. left ventricular noncompaction
D. **restrictive cardiomyopathy**
E. arrhythmogenic right ventricular cardiomyopathy

Ans D

14

CHAPTER 14

Pediatric Hematology



WHAT THIS CHAPTER COVERS

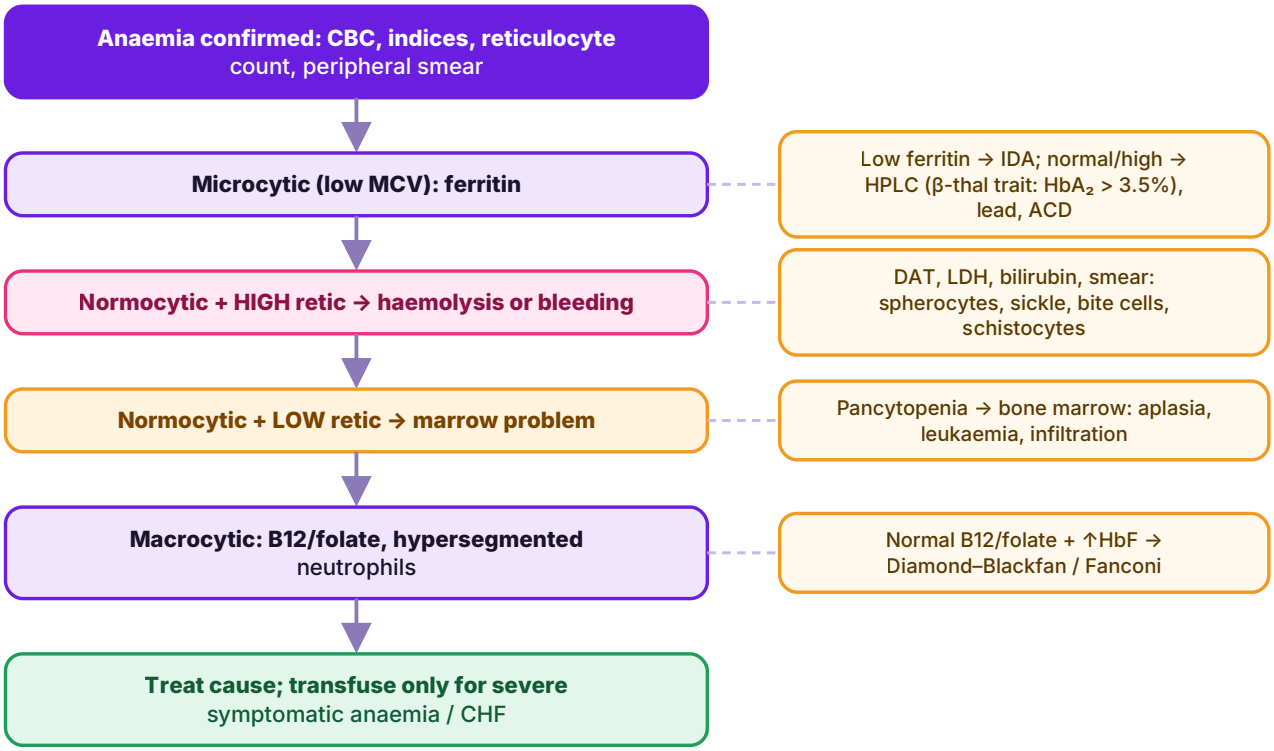
- Anemia - General & Classification
- Iron Deficiency Anemia
- Megaloblastic Anemia
- Hemolytic Anemia - General
- Sickle Cell Disease
- Thalassemia
- G6PD Deficiency
- Hereditary Spherocytosis
- Autoimmune Hemolytic Anemia
- Aplastic Anemia
- Hemophilia & Coagulation Disorders
- Von Willebrand Disease
- ITP & Platelet Disorders
- DIC & Coagulopathy
- Leukemia
- Lymphoma
- Neutropenia
- Bone Marrow & Stem Cell Transplant
- Blood Transfusion

251 MCQs **19** subtopics

14.1 Anaemia – Definition & Classification

Age	Hb cut-off for anaemia (WHO)
6–59 months	**< 11 g/dL**
5–11 years	< 11.5 g/dL
12–14 years	< 12 g/dL
Neonate (term)	**< 13.5 g/dL** (cord); preterm lower

- **WHO severity (6–59 m)**: **mild 10–10.9, moderate 7–9.9, severe < 7 g/dL**.
- **Physiological nadir**: term **8–12 weeks (~9.5–11)**; preterm 4–8 weeks (7–9) – "anaemia of prematurity" (↓ EPO).
- **Reticulocyte count** (normal 0.5–2%; corrected retic = retic × patient Hct/45) separates **production failure (low retic)** from **destruction/loss (high retic)**.



MCV-based approach to childhood anaemia

Microcytic (MCV < 70–80 fL)	Normocytic	Macrocytic (MCV > 100)
Iron deficiency (MC), **thalassaemia trait**, anaemia of chronic disease (late), **lead poisoning**, sideroblastic anaemia (B6, Pearson) – "**TAILS**"	Acute blood loss, **haemolysis**, chronic disease, CKD, **aplastic anaemia**, leukaemia/marrow infiltration, red-cell aplasia	**B12/folate deficiency (megaloblastic)**, **Diamond–Blackfan**, **Fanconi anaemia**, liver disease, hypothyroidism, reticulocytosis, drugs (valproate, AZT), Down syndrome, normal newborn

14.2 Iron Deficiency Anaemia (IDA)

- **MC nutritional deficiency & MCC anaemia in children** worldwide/India (NFHS-5: ~67% of 6–59-month-olds anaemic).

- **Peak ages**: **6–24 months** (rapid growth, cow's-milk feeding, delayed complementary feeds) and **adolescence**; LBW/preterm, delayed cord clamping not done, hookworm, blood loss (cow's milk enteropathy, Meckel's), celiac, Helicobacter.
- Features: pallor, fatigue, **pica (pagophagia – ice)**, irritability, **breath-holding spells**, ↓ attention & **cognitive impairment (may be irreversible if in infancy)**, **koilonychia**, angular stomatitis, glossitis, systolic flow murmur, CHF in severe; **Plummer–Vinson** (rare in children); febrile seizures risk ↑.

Sequence of changes in iron deficiency

↓ Serum ferritin (earliest; stores depleted) → ↓ serum iron & ↑ TIBC, ↓ transferrin saturation (< 16%), ↑ free erythrocyte protoporphyrin & ↑ sTfR → ↓ reticulocyte Hb (CHr) → microcytosis (↑ RDW – earliest index) → ↓ Hb (anaemia – last).

Test	IDA	β-thal trait	Anaemia of chronic disease	Lead poisoning	Sideroblastic
Serum ferritin	**↓↓**	N/↑	**N/↑**	N	↑
Serum iron	↓	N/↑	↓	N	↑
TIBC	**↑**	N	**↓**	N	N/↓
Transferrin saturation	↓	N	↓/N	N	↑
RDW	**↑ (high)**	**Normal**	Normal	N	↑
RBC count	↓	**↑ (> 5 million)**	↓	↓	↓
Mentzer index (MCV/RBC)	**> 13**	**< 13**	> 13		
HbA_2	N/↓	**↑ (> 3.5%)**	N	N	N
Smear	Microcytic hypochromic, **pencil cells**, anisocytosis	**Target cells, basophilic stippling**		**Basophilic stippling**	**Ring sideroblasts** (marrow)

TREATMENT OF IDA

Oral elemental iron 3 (–6) mg/kg/day (ferrous sulfate/fumarate, on empty stomach or between meals, with vitamin C; avoid with milk/tea) – **continue for 2–3 months after Hb normalises** (to replenish stores). Response: **reticulocytosis in 5–7 days (earliest – peak 7–10 days)**; Hb rises ~1 g/dL per 2–3 weeks (≥ 1 g/dL in 4 weeks confirms diagnosis). **Limit cow's milk to < 500 mL/day. Parenteral iron** (iron sucrose/ferric carboxymaltose) – malabsorption, non-compliance, intolerance. **Blood transfusion only if Hb < 4–5 g/dL or CHF** (packed cells 5–10 mL/kg slowly, ± furosemide). Deworm with albendazole.

- **Iron poisoning** – see PALS chapter (> 60 mg/kg elemental is serious).

14.3 Megaloblastic Anaemia

	Vitamin B12 deficiency	Folate deficiency
Causes	**Vegetarian/vegan diet & exclusive breastfeeding by B12-deficient mother (MC in India)**, pernicious anaemia (juvenile), **ileal resection, Crohn's, fish tapeworm**, **transcobalamin II deficiency**, Imerslund–Gräsbeck (cubilin), intrinsic factor deficiency	Goat's milk feeding, malnutrition, **haemolytic anaemia (↑ demand)**, drugs (**phenytoin, methotrexate, trimethoprim, pyrimethamine**), malabsorption (celiac), pregnancy
Stores	Last **3–5 years** (adults)	Last **3–4 months**
Neurological	**Present – developmental regression, hypotonia, tremors (ITS), SACD, neuropathy, dementia**	**Absent** (but NTDs in fetus)

	Vitamin B12 deficiency	Folate deficiency
Skin	**Knuckle hyperpigmentation**, hair depigmentation	
Labs	**↑ methylmalonic acid + ↑ homocysteine**	**Normal MMA**, ↑ homocysteine

- **Common to both**: macrocytosis, **hypersegmented neutrophils (≥ 5% with ≥ 5 lobes / one with 6 – earliest peripheral smear sign)**, **macro-ovalocytes**, pancytopenia, **ineffective erythropoiesis → ↑ LDH, ↑ indirect bilirubin**, megaloblastic marrow (nuclear–cytoplasmic asynchrony), glossitis.
- Rx: **B12 – IM cyanocobalamin/hydroxocobalamin 1000 µg daily × 1 week → weekly → monthly**; folic acid 1–5 mg/day. **Never give folate alone in B12 deficiency** (worsens neurological damage). Hypokalaemia can occur on treatment.

14.4 Haemolytic Anaemias – General

- **Clues**: pallor + **jaundice (unconjugated)** + **splenomegaly**, dark urine, gallstones (pigment), **reticulocytosis**, ↑ LDH, **↓ haptoglobin**, ↑ indirect bilirubin, urinary urobilinogen ↑; **extramedullary haematopoiesis → frontal bossing, "hair-on-end" skull X-ray, maxillary hyperplasia (chipmunk facies)** in chronic severe haemolysis.
- **Intravascular** (haemoglobinuria, haemosiderinuria): **G6PD, PNH, transfusion reaction, malaria, microangiopathic (HUS, DIC), burns**. **Extravascular** (spleen): spherocytosis, AIHA (warm), thalassaemia, sickle cell.

Intrinsic (corpuscular – mostly inherited)	Extrinsic (acquired)
Membrane: hereditary spherocytosis, elliptocytosis, PNH (acquired)	**Immune**: AIHA (warm IgG, cold IgM), isoimmune (HDN), drug-induced
Enzyme: G6PD (HMP shunt), **pyruvate kinase** (glycolysis – MC enzyme defect of Embden–Meyerhof pathway)	**Microangiopathic**: HUS, TTP, DIC, Kasabach–Merritt
Haemoglobin: sickle cell, thalassaemia, unstable Hb	Infections (malaria), toxins (lead, snake venom), hypersplenism, burns

14.5 Thalassaemia

- **β-thalassaemia**: AR, chr 11; **point/splice mutations** (India MC – **IVS 1-5 G→C**); carrier frequency India ~3–4% (higher in Sindhi, Punjabi, Gujarati, Bengali, Parsi communities).
- **α-thalassaemia**: chr 16; **gene deletions**; 4 genes: 1 deleted – silent carrier; 2 – trait (α-thal minor; cis deletion (– –/αα) in SE Asians → Hb Bart's risk); 3 – **HbH disease (β_4)**; 4 – **Hb Bart's (γ_4) hydrops fetalis** (incompatible with life).

Type	Genotype	Clinical
β-thal minor (trait)	β/β^+ or β/β^0	Asymptomatic, mild microcytic anaemia, **HbA_2 > 3.5% (diagnostic)**, Mentzer < 13, **no iron therapy needed**
β-thal intermedia / NTDT	β^+/β^+ etc., HbE-β	Moderate anaemia (Hb 7–10), presents later (> 2 y), occasional transfusions, extramedullary haematopoiesis, iron overload from gut absorption
β-thal major (Cooley's anaemia)	β^0/β^0, β^0/β^+	**Presents at 6–12 months (after HbF → HbA switch)**: severe anaemia, **massive hepatosplenomegaly**, jaundice, FTT, **thalassaemic facies (frontal bossing, maxillary prominence, depressed nasal bridge, malocclusion)**, **hair-on-end skull**, transfusion dependence

- **Investigations (thal major)**: **Hb 3–7 g/dL, microcytic hypochromic, target cells, nucleated RBCs, basophilic stippling**, ↑ retic; **HbF ↑↑ (up to 90–100% in β^0/β^0), HbA_2 variable**; **Hb electrophoresis/HPLC – IOC (HPLC preferred)**; **NESTROFT** (naked-eye single-tube red cell osmotic fragility – **screening for carriers – positive (↓ fragility)** in thal trait); DNA analysis for prenatal diagnosis (**CVS at 10–12 wk**).

MANAGEMENT OF THALASSAEMIA MAJOR

- **Hypertransfusion regimen**: **packed RBCs 10–15 mL/kg every 2–4 weeks to keep pre-transfusion Hb 9.5–10.5 g/dL** (suppresses ineffective erythropoiesis, normal growth, prevents bone changes & splenomegaly); leucodepleted, extended-phenotype-matched; **Hep B vaccine** before starting.
- **Iron chelation** – start after **10–20 transfusions or ferritin > 1000 ng/mL** (usually ~2–3 y):
  - **Desferrioxamine** – SC infusion 8–12 h/night, 5–7 nights/week (20–40 mg/kg) – oto/ocular toxicity, growth retardation, **Yersinia** infection.
  - **Deferasirox** (oral, once daily, ≥ 2 y) – most used; renal & hepatic toxicity, GI upset.
  - **Deferiprone** (oral, 3 times/day) – **agranulocytosis (weekly CBC)**, arthropathy; best for **cardiac iron**.
- **Monitoring**: ferritin 3-monthly, **cardiac T2* MRI (< 20 ms = cardiac iron overload; MCC death – cardiac failure/arrhythmia)**, liver iron (MRI R2/T2*), endocrine (growth, puberty – hypogonadism (MC endocrine complication), diabetes, hypothyroidism, hypoparathyroidism), bone (osteoporosis).
- **Splenectomy** if transfusion requirement > **200–250 mL/kg/year** (hypersplenism) – **after 5 years**, with pneumococcal, Hib, meningococcal vaccines + penicillin prophylaxis.
- **Folic acid**; avoid iron & vitamin C (except with DFO).
- **Curative: allogeneic HSCT (HLA-identical sibling) – best before liver fibrosis (Pesaro class I)**; **gene therapy (betibeglogene)**; **luspatercept** reduces transfusion need; **hydroxyurea** in some intermedia/HbE-β.
- Prevention: **carrier screening (premarital/antenatal), genetic counselling, prenatal diagnosis**.

14.6 Sickle Cell Disease

- AR, β-globin **Glu6Val (GAG → GTG)**; HbS polymerises when deoxygenated → sickling → **vaso-occlusion + haemolysis**. India – tribal belts of **central India (MP, Chhattisgarh, Maharashtra, Odisha, Gujarat)**; **National Sickle Cell Anaemia Elimination Mission 2023** (target 2047).
- **Protective factor: high HbF** (Arab-Indian haplotype – milder disease in India); **sickle trait protects against falciparum malaria**.
- Presents after **6 months** (HbF falls).

Crisis / complication	Key features
Dactylitis (hand-foot syndrome)	**Earliest manifestation (6 m–2 y)** – painful symmetric swelling of hands & feet
Vaso-occlusive (painful) crisis	**MC manifestation** – bones, chest, abdomen; precipitated by cold, dehydration, infection, hypoxia
Acute chest syndrome	**MCC death in adults & older children**; new infiltrate + chest pain/fever/hypoxia; Rx O_2, antibiotics (cephalosporin + macrolide), **exchange transfusion**
Splenic sequestration crisis	Young children (< 5 y): **sudden enlargement of spleen + acute fall in Hb + shock**; Rx **urgent transfusion**
Aplastic crisis	**Parvovirus B19** – reticulocytopenia, sudden fall in Hb
Hyperhaemolytic crisis	Haemolysis ↑ (G6PD coexistent, infection)
Infections	**Functional asplenia (autosplenectomy by 5 y; Howell–Jolly bodies) → S. pneumoniae (MC – sepsis/meningitis), Hib**, **Salmonella osteomyelitis** (S. aureus remains overall MC)
Stroke	**Peak 2–9 y**; screening by **transcranial Doppler (TCD) annually 2–16 y – velocity > 200 cm/s → chronic transfusion programme**
Others	Priapism, **avascular necrosis of femoral head**, leg ulcers, retinopathy, **renal papillary necrosis, hyposthenuria (enuresis)** (also in trait), pigment gallstones, growth delay, pulmonary hypertension

- **Diagnosis**: **sickling test (sodium metabisulfite)/solubility test** – screening (positive in trait too); **Hb electrophoresis/HPLC – confirmatory**; **point-of-care tests** (HemoTypeSC, Sickle SCAN); smear – sickle cells, target cells, Howell–Jolly bodies; newborn screening.

- **Management**: hydration, **analgesia (NSAIDs, morphine)**, oxygen if hypoxic, warmth; **hydroxyurea (↑ HbF – from 9 months of age for all HbSS; ↓ crises, ACS, transfusion, mortality)**; **penicillin prophylaxis from 2–3 months to 5 years**; **pneumococcal (PCV + PPSV23), Hib, meningococcal, influenza vaccines**; **folic acid**; transfusion (simple/exchange); **HSCT – curative**; **voxelotor**, **crizanlizumab**, L-glutamine; **gene therapy (exagamglogene – CRISPR, lovotibeglogene)**.

14.7 G6PD Deficiency

- **X-linked recessive** – **MC RBC enzyme defect worldwide**; ↓ **NADPH → ↓ reduced glutathione** → oxidant damage → **Heinz bodies (denatured Hb – supravital stain)** → **bite cells (degmacytes)**, blister cells; **intravascular haemolysis** (haemoglobinuria – "cola-coloured urine" 24–72 h after trigger).
- **Triggers**: **drugs – primaquine, dapsone, nitrofurantoin, sulfonamides (co-trimoxazole), quinolones, rasburicase, methylene blue, high-dose aspirin, vitamin K (synthetic), chloramphenicol**; **fava beans (favism)**; **infections (MC trigger)**; naphthalene (mothballs); DKA.
- **Neonatal jaundice** (may need exchange) – a common cause in India.
- **Diagnosis**: **G6PD enzyme assay (quantitative)** – **falsely normal during acute haemolysis** (young RBCs have more enzyme) → **repeat after 2–3 months**; fluorescent spot test (screening).
- Rx: avoid triggers, transfusion if severe, hydration (renal protection); usually self-limiting.
- WHO class I (chronic non-spherocytic haemolytic anaemia) – most severe; **Mediterranean (class II) – common in India**.

14.8 Hereditary Spherocytosis & Other Membrane Defects

- **AD (75%)**; defects in **ankyrin (MC), spectrin, band 3, protein 4.2** → loss of membrane → spherocytes → **extravascular (splenic) haemolysis**; MC in Northern Europeans.
- Features: **anaemia, jaundice, splenomegaly (triad)**, **pigment gallstones** (even in childhood), **aplastic crisis (parvovirus B19)**, neonatal jaundice; leg ulcers (rare).
- Labs: **spherocytes (no central pallor), ↑ MCHC (> 36 g/dL – only anaemia with high MCHC)**, ↑ retic, **DAT negative** (vs AIHA – DAT positive).
- **Diagnosis**: **EMA (eosin-5-maleimide) binding test by flow cytometry – test of choice (most sensitive & specific)**; **incubated osmotic fragility test – increased fragility** (classic); acidified glycerol lysis test.
- Rx: folic acid; transfusion as needed; **splenectomy (curative for anaemia) – delay until ≥ 6 years** (partial splenectomy earlier) with vaccines & penicillin prophylaxis; **cholecystectomy** if gallstones.
- **Hereditary elliptocytosis** (spectrin, AD) – mostly asymptomatic; **hereditary pyropoikilocytosis** – severe, heat-sensitive RBCs; **Southeast Asian ovalocytosis** – malaria protection.
- **Pyruvate kinase deficiency** (AR) – **echinocytes (burr cells/prickle cells)**, chronic haemolysis, **↑ 2,3-BPG (better O_2 delivery – tolerate anaemia)**; Rx transfusion, splenectomy, **mitapivat**.

14.9 Autoimmune Haemolytic Anaemia (AIHA)

	Warm AIHA	Cold agglutinin disease	Paroxysmal cold haemoglobinuria
Antibody	**IgG** (37 °C)	**IgM** (anti-I)	**Donath–Landsteiner IgG** (biphasic, anti-P)
Haemolysis	Extravascular (spleen)	Intravascular/liver	**Intravascular**
Associations	Idiopathic, **SLE**, Evans syndrome (+ ITP), ALPS, lymphoma, drugs, CVID	**Mycoplasma pneumoniae (anti-I), EBV (anti-i)**	**Post-viral (children)**, congenital syphilis (historic)
DAT	**IgG ± C3**	**C3 only**	C3

	Warm AIHA	Cold agglutinin disease	Paroxysmal cold haemoglobinuria
Rx	**Steroids (prednisolone 2 mg/kg) – first line**, IVIG, **rituximab**, splenectomy	Keep warm, rituximab; steroids ineffective	Self-limiting, keep warm

- **Evans syndrome** – AIHA + ITP (± neutropenia).

14.10 Aplastic Anaemia & Bone Marrow Failure

Disorder	Key features
Acquired aplastic anaemia	Idiopathic (MC), post-hepatitis (**seronegative hepatitis**), drugs (**chloramphenicol** – idiosyncratic), benzene, radiation, viruses (EBV, parvovirus, HIV), PNH; **pancytopenia + hypocellular marrow (< 25%)**, no splenomegaly; **severe AA: ANC < 500, platelets < 20,000, retic < 20,000 (or < 1%)**; Rx **HLA-matched sibling HSCT (TOC)**; **ATG + ciclosporin + eltrombopag** (immunosuppressive therapy)
Fanconi anaemia (MC inherited marrow failure)	**AR (FANC genes – DNA repair)**; presents **5–10 y** with pancytopenia; **short stature, café-au-lait spots/hyperpigmentation, absent/hypoplastic thumbs & radii**, microcephaly, renal anomalies (horseshoe), hypogonadism, microphthalmia; **macrocytosis, ↑ HbF**; **chromosomal breakage test with DEB (diepoxybutane) / mitomycin C – diagnostic**; ↑ **AML, MDS, squamous cancers**; Rx HSCT (reduced intensity), androgens (oxymetholone)
Diamond–Blackfan anaemia (congenital pure red-cell aplasia)	**Ribosomal protein genes (RPS19)**; **presents < 1 y (often by 2–3 months)**; **macrocytic anaemia, reticulocytopenia, ↑ HbF, ↑ erythrocyte adenosine deaminase (eADA)**, **triphalangeal thumbs**, cleft palate, short stature; marrow – absent erythroid precursors; Rx **steroids (80% respond)**, transfusion, HSCT
Transient erythroblastopenia of childhood (TEC)	**1–4 y**, post-viral, **normocytic**, retic ↓, **normal HbF & eADA**, no anomalies; **spontaneous recovery in 1–2 months**
Dyskeratosis congenita	Telomere defect (DKC1 – X-linked): **triad – reticulate skin pigmentation, nail dystrophy, oral leukoplakia** + marrow failure, pulmonary fibrosis
Shwachman–Diamond	Neutropenia + exocrine pancreatic insufficiency + metaphyseal dysostosis
Amegakaryocytic thrombocytopenia / TAR	TAR – **thrombocytopenia + absent radius with thumbs present**; improves after 1 y

14.11 Platelet Disorders

Immune thrombocytopenic purpura (ITP)

- **MCC of acute thrombocytopenia in a well child**; **2–5 y**, **1–4 weeks after viral infection/vaccination (MMR)**; antibodies against **GPIIb/IIIa**.
- **Sudden petechiae/purpura, bruising, mucosal (epistaxis, gum) bleeding** in an otherwise **well child**; **no hepatosplenomegaly/lymphadenopathy**; ICH rare (< 0.5%).
- **Isolated thrombocytopenia (often < 20,000)**, normal Hb (unless bleeding) & WBC, **large platelets (↑ MPV)**; **bone marrow – normal/↑ megakaryocytes** (**BM not routinely needed** – do if atypical features/before steroids where leukaemia not excluded).
- Classification: **newly diagnosed (< 3 m), persistent (3–12 m), chronic (> 12 months)**; **~80% resolve within 6 months** (children); chronic more in adolescents (adolescent girls → SLE).
- **Management – based on bleeding, not count**: **observation for skin-only bleeding** (ASH 2019); **severe/mucosal bleeding → IVIG 0.8–1 g/kg (fastest rise), short-course steroids (prednisolone 2–4 mg/kg/day × 5–7 d), anti-D (Rh+ non-splenectomised)**; life-threatening (ICH) → **platelet transfusion + IVIG + IV methylprednisolone**; chronic → **TPO-receptor agonists (eltrombopag, romiplostim)**, rituximab, splenectomy (> 5 y, rarely). Avoid NSAIDs/aspirin, contact sports.

Other platelet disorders

Disorder	Features
Bernard–Soulier (AR)	**GPIb-IX-V deficiency** (vWF receptor) → defective adhesion; **giant platelets + thrombocytopenia**; **absent aggregation with ristocetin (not corrected by normal plasma)**
Glanzmann thrombasthenia (AR)	**GPIIb/IIIa deficiency** → defective aggregation; **normal platelet count & size**; **no aggregation with ADP, collagen, epinephrine (normal with ristocetin)**; ↑ bleeding time; Rx platelet transfusion, rFVIIa
Wiskott–Aldrich	Thrombocytopenia with **small platelets**, eczema, immunodeficiency
Neonatal alloimmune thrombocytopenia	Maternal anti-HPA-1a; severe; first pregnancy
Kasabach–Merritt phenomenon	**Kaposiform haemangioendothelioma/tufted angioma** (not common infantile haemangioma) → platelet trapping, consumptive coagulopathy; Rx **sirolimus**, vincristine, steroids
Grey platelet syndrome	Absent α-granules
HUS / TTP	See Renal; TTP – ADAMTS13 deficiency (Upshaw–Schulman congenital)

14.12 Coagulation Disorders

Test	Pathway / use	Prolonged in
PT (INR)	**Extrinsic (VII) + common**	**Vitamin K deficiency (first – factor VII shortest t½)**, liver disease, warfarin, DIC, factor VII def.
aPTT	**Intrinsic (XII, XI, IX, VIII) + common**	**Haemophilia A, B**, vWD (if FVIII low), heparin, lupus anticoagulant, factor XI/XII def.
Bleeding time / PFA-100	Platelet function	vWD, Glanzmann, Bernard–Soulier, thrombocytopenia, aspirin
Thrombin time	Fibrinogen → fibrin	Hypo/dysfibrinogenaemia, heparin
Factor XIII	Clot stabilisation	**All routine tests normal** – **delayed umbilical stump bleeding**, ICH, poor wound healing; **urea clot solubility test**

Haemophilia

	Haemophilia A	Haemophilia B (Christmas disease)
Factor	**VIII** (80–85%)	**IX**
Inheritance	**X-linked recessive** (30% new mutations); **intron 22 inversion (MC in severe A)**	X-linked recessive
Lab	**↑ aPTT, normal PT, platelet count & BT**	Same

- **Severity**: **severe < 1% factor activity** (spontaneous bleeds), **moderate 1–5%** (bleeding after minor trauma), **mild 5–40%** (bleeding after surgery/major trauma).
- **Haemarthrosis – hallmark (MC site: knee > elbow > ankle)** → **target joint → chronic synovitis → haemophilic arthropathy**; muscle haematomas (**iliopsoas – femoral nerve palsy**, compartment syndrome), **ICH (MCC death)**, circumcision bleeding (often first sign), **bleeding at birth is uncommon (cephalhaematoma)**.
- **Treatment**: **factor VIII replacement – 1 IU/kg raises plasma level by 2% (t½ 8–12 h)**; **factor IX – 1 IU/kg raises by 1% (t½ 18–24 h)**. Target levels: minor bleed 30–40%, **joint 40–60%**, **life-threatening/ICH/surgery 80–100%**.
  - Dose FVIII (IU) = **weight (kg) × desired rise (%) × 0.5**; FIX = weight × desired rise × 1.
- **Prophylaxis** (from early childhood, before 2nd joint bleed) – **standard of care**; **emicizumab** (bispecific antibody mimicking FVIIIa – SC weekly/monthly) – also for inhibitors.

- **Inhibitors (alloantibodies)** – **~30% of severe haemophilia A** (5% B); measured in **Bethesda units**; Rx **bypassing agents – rFVIIa, aPCC (FEIBA)**, **immune tolerance induction**, emicizumab.
- **Adjuncts**: **desmopressin (DDAVP) – mild haemophilia A & vWD type 1** (releases FVIII/vWF from endothelium; ineffective in haemophilia B); **tranexamic acid/EACA** (mucosal bleeds, dental; **not in haematuria**); **RICE** for joint bleeds; physiotherapy; **avoid IM injections, aspirin/NSAIDs** (give vaccines SC/IM with fine needle & pressure); **gene therapy** (valoctocogene, etranacogene).

Von Willebrand disease (vWD)

- **MC inherited bleeding disorder** (1%); **AD (types 1, 2)**; type 3 AR (severe).
- vWF – platelet adhesion (GPIb) + **carrier of factor VIII** (protects from degradation).
- **Mucocutaneous bleeding** – epistaxis (MC), easy bruising, **menorrhagia**, post-dental extraction bleeding; haemarthrosis only in type 3.
- Labs: **↑ bleeding time/PFA**, ↑/N aPTT (low FVIII), normal PT & platelet count; **↓ vWF antigen, ↓ ristocetin cofactor activity (vWF:RCo)**, ↓ FVIII; **ristocetin aggregation ↓ (corrected by normal plasma)**; **type 2B – ↑ response to low-dose ristocetin + thrombocytopenia**.
- Rx: **desmopressin (type 1 – DOC)**, tranexamic acid, **vWF-containing FVIII concentrates (type 2, 3)**, cryoprecipitate (if nothing else); **DDAVP contraindicated in type 2B**; OCPs for menorrhagia.

Feature	Platelet / vascular (ITP, vWD)	Coagulation factor (haemophilia)
Site	**Skin & mucous membranes** – petechiae, purpura, epistaxis, gum bleeding, menorrhagia	**Deep tissues – joints, muscles**
Onset after injury	Immediate	**Delayed**
Petechiae	Present	Absent
Ecchymoses	Small, multiple	Large, palpable

DIC

- Sepsis (MC), asphyxia, shock, malignancy (APL), snake bite, NEC, purpura fulminans; **↑ PT, ↑ aPTT, ↓ fibrinogen, ↑ D-dimer/FDP, ↓ platelets, schistocytes**; Rx treat cause, FFP, platelets, cryoprecipitate (fibrinogen < 100 mg/dL).
- **Purpura fulminans**: neonatal – **homozygous protein C/S deficiency**; post-infectious (varicella – protein S antibodies), meningococcaemia; Rx FFP, protein C concentrate.

Thrombosis in children

- Rare; **central venous catheters (MC risk factor)**, neonates, nephrotic syndrome, cyanotic CHD, sepsis, malignancy (asparaginase); inherited – **factor V Leiden (MC inherited thrombophilia), prothrombin G20210A**, protein C/S/antithrombin deficiency, ↑ lipoprotein(a), homocystinuria; **antiphospholipid antibodies**. Rx LMWH (enoxaparin), UFH, warfarin, DOACs (rivaroxaban/dabigatran approved in children).

14.13 Blood Transfusion – Paediatric Points

- **PRBC 10–15 mL/kg raises Hb by 2–3 g/dL** (rule: 4 mL/kg ↑ Hb by 1 g/dL; whole blood 6 mL/kg ↑ 1 g/dL).
- **Platelets 10–15 mL/kg (or 1 unit/10 kg)** ↑ by 50,000–1 lakh; prophylactic threshold **< 10,000** (well), < 20,000 (fever/sepsis), < 50,000 (bleeding/procedure), neonates < 25,000.
- **FFP 10–15 mL/kg**; **cryoprecipitate** (fibrinogen, FVIII, vWF, FXIII) 1 unit/5–10 kg.
- **Irradiated products (prevent TA-GvHD)**: **IUT, exchange transfusion, preterm**, SCID/DiGeorge, HSCT recipients, Hodgkin, **related-donor blood**, fludarabine.
- **Leucodepleted**: thalassaemia/chronic transfusion (↓ febrile reactions, HLA alloimmunisation, CMV).
- Reactions: **febrile non-haemolytic (MC)**, allergic/urticarial, **acute haemolytic (ABO – clerical error; MCC fatal)**, TRALI, TACO, delayed haemolytic, bacterial contamination (platelets), iron overload.

Practice MCQs · Pediatric Hematology

251 questions · correct option highlighted in green · cover the answer strip to self-test

Anemia - General & Classification

1. Transfer of Iron from enterocyte to plasma is inhibited by:

A. **Hepcidin**
B. DMT-1
C. Ferroportin
D. Hepestin

Ans A Hepcidin: principal iron regulatory hormone that negatively regulates ferroportin on basolateral surface of enterocyte.

2. Hemoglobin H disease is caused by deletion:

A. Single α globin chain
B. Two α globin chains
C. **Three α globin chains**
D. All α globin chain

Ans C

3. Which of the following drugs is not associated with pure red cell aplasia?

A. Phenytoin
B. Isoniazid
C. **Erythropoietin**
D. Chloramphenicol

Ans C Classification of pure red cell aplasia Acquired
• Self-limited: Transient erythroblastopenia of childhood • Thymoma
• Hereditary: Congenital pure red cell aplasia (Diamond- • Lymphomas: Hodgkin's, or Non Hodgkin's, large granular Lymphoma Blackfan syndrome)Q • Paraneoplastic to solid tumors
• Connective tissue disorders: SLE, JRA, RA with
• Virus: Parvovirus B19Q, hepatitis, HTLV, EBV
• Drugs: phenytoin, azathioprine, chloramphenicol, isoniazid
• Post ABO incompatible transplantQ

4. Reed Sternberg cells are found in:

A. **Hodgkin's disease**
B. Sickle cell anemia
C. Thalassemia
D. CML

Ans A Diagnostic Hallmark of Hodgkin Lymphoma are Reed-Sternberg cells, which are binucleate cell or having a single nucleus with multiple nuclear lobes.

5. A 17-year-old boy presented with TLC of 138 × 109/L with 80% blasts on the peripheral smear. Chest X-ray demonstrated a large mediastinal mass. Immunophenotyping of this patient's blasts would most likely demonstrate:

A. No surface antigen (Null phenotype)
B. **An immature T-cell phenotype (Tdt/CD34/CD3)**
C. Myeloid markers, such as CD13, CD33 and CD15
D. B-cell markers such as CD19, CD20 and CD22

Ans B • Anterior mediastinal mass can be seen due to thymic infiltration in case of T-cell ALL
• Tdt and CD34 are markers of Blasts while CD3 is a marker of T cell.

6. True about hematological disorder:

A. In Hemophilia B cryoprecipitate is useful
B. **Both PT and aPTT are increased in DIC**
C. IVIg is useful in ITP
D. Desmopressin has no role in vWD
E. Hemorrhagic disease of newborn is due to vit C deficiency

Ans B & (c) IV Ig is useful in ITP Cryoprecipitate is not useful for treating hemophilia B, as cryoprecipitate contains a high concentration of coagulation factor VIII, coagulation factor XIII, and fibrinogen (but not factor IX)

7. Anemia and reticulocytopenia that occur in the 2nd half of infancy period is LEAST likely due to

A. congenital hypoplastic anemia (Diamond-Blackfan anemia)
B. transient erythroblastopenia of childhood
C. **a protracted, prolonged course of the anemia of hemolytic disease of the newborn**
D. aplastic crises complicating various types of chronic hemolytic anemias
E. Fanconi anemia

Ans C The anemia of hemolytic disease of the newborn can have a protracted course, but this usually terminates spontaneously at 5-8 wk of age. Aplastic anemic crises, frequently caused by parvovirus B19 infections, may complicate various types of chronic hemolytic disease, but usually after the first several months of life.

8. An extremely high RBC distribution width (RDW) is MOST likely consistent with

A. iron deficiency anemia
B. megaloblastic anemia
C. **sideroblastic anemia**
D. thalassemia
E. sickle cell anemia

Ans C RDW is usually high in IDA, megaloblastic anemia, and sideroblastic anemia, but it is extremely high in the last.

9. You are evaluating a 7-year-old male child with cervical lymphadenopathy; the MOST likely finding that raise your suspicion of malignancy is

A. matted nodes
B. fluctuant nodes
C. **fixed nodes**
D. presence of systemic symptoms
E. no regression in the size of the nodes to its normal within 2 weeks

Ans C A firm fixed node to the surrounding tissue should always raise the question of malignancy, regardless of the presence or absence of systemic symptoms or other abnormal physical findings. Fluctuance suggests abscess formation. Tuberculous nodes may be matted.

Iron Deficiency Anemia

10. True about iron deficiency anemia is/are:

A. Normochromic, normocytic hypoproliferative anemia
B. Normochromic, normocytic hyperproliferative anemia
C. **Plasma transferrin saturation decreases**
D. Marked poikilocytosis
E. Blood transfusion is indicated at Hb < 4.0g/dL

Ans C and e.B lood transfusion is indicated at Hb < 4.0g/dL In Iron deficiency Anemia, peripheral blood smear reveals hypochromic, microcytic red cells with anisocytosis. Elliptocytic or cigar-shaped red cells are often seen Parameter MCV and RBC RDW Reticulocyte Hb Serum Ferritin TIBC Transferrin Transferrin FEP count conc saturation Receptor In Iron def. Decreased Increased Decreased Decreased Increased Decreased Increased Increased Anemia

11. Iron stores are best indicated by:

A. **S. ferritin**
B. S. Iron
C. S. Transferrin
D. TIBC

Ans A Serum ferritin reflects the storage of Iron which is decreased even in the pre-latent stage of Iron deficiency Anemia and is the fo most sensitive marker. weiveR 15. a. Hepcidin Ferroportin which regulates the Iron release from tissue store house is regulated by hepcidin hormone released by liver.

12. Release of ferroportin store is controlled by:

A. **Hepcidin**
B. Transferrin
C. Ferritin
D. Hepoxin

Ans A the master iron-regulatory hormone produced by the liver, binds ferroportin and triggers its internalization/ degradation, controlling the release of iron from stores into plasma.

13. True about iron deficiency anemia in children:

A. Iron absorption from terminal ileum (PGI June 09)
B. Cow milk contains less iron than breast milk
C. **Serum ferritin depletes first**
D. Decreased alertness
E. Decreased Red cell distribution width (RDW)

Ans C and d. Decreased alertness Discussing about the options one by one, a. Iron is absorbed from proximal small intestine mainly from duodenum b. Cow milk has more iron than breast milk. But, bioavailability of iron of breast milk is much more than cow milk. c. S. ferritin depletes first: Next, serum iron level decreases, iron binding capacity of serum (serum transferrin)increases, and percent saturation (transferrin saturation) is reduced d.

14. Which of the following findings is diagnostic of iron deficiency anemia?

A. **Increased TIBC, decreased serum ferritin**
B. Decreased TIBC, decreased serum ferritin
C. Increased TIBC, increased serum ferritin
D. Decreased TIBC, increased serum ferritin

Ans A

15. The earliest indicator of response after starting iron in a 6-year-old girl with iron deficiency is:

A. **Increased reticulocyte count**
B. Increased hemoglobin
C. Increased ferritin
D. Increased serum iron

Ans A Increased reticulocyte count 21 above

16. True about iron deficiency anemia:

A. **Microcytic hypochromic anemia**
B. Decreased TIBC
C. Increased ferritin
D. Bone marrow iron decreased earlier than serum iron
E. Decreased transferrin saturation

Ans A d. Bone marrow iron decreased earlier than serum iron, e. Decreased transferrin saturation

17. Which of the following is true about oral therapy for iron deficiency anemia?

A. In 300 mg elemental iron given, 100 mg gets absorbed
B. Reticulocytosis appears in one to 2 weeks and then peaks in 3–4 weeks
C. Response to treatment seen in 4 weeks
D. **Decrease in absorption occurs with improvement of symptoms**
E. Stop the treatment after normalising the Hb

Ans D Discussing about the options one by one, a. Only 10% of the iron given by oral route gets absorbed b. Reticulocytosis appears in 48-72 hours, peaking at 5–7 days c. Clinical response to treatment appears within 24 hours d. The normalisation of body iron store relatively decreases the iron absorption

18. Most sensitive and specific test for diagnosis of iron deficiency is:

A. Serum iron level
B. **Serum ferritin levels**
C. Serum transferrin receptor populations
D. Transferrin saturation

Ans B Low serum ferritin level invariably indicates iron deficiency and hence it is the most specific and sensitive test for iron deficiency anemia.

19. Which is not seen in iron deficiency anemia?

A. **Hypersegmented neutrophils**
B. Microcytosis precedes hypochromia
C. MCHC < 50%
D. Commonest cause of anemia in India

Ans A • Hypersegmented neutrophils are seen in megaloblastic anemia.

20. A 5 year old child presented with microcytic hypochromic anaemia. Serum ferritin is low. Which of the following is least likely?

A. Celiac disease
B. **Thalassemia major**
C. Nutritional anemia
D. Paroxysmal nocturnal hemoglobinuria

Ans B S. ferritin is normal or increased in Thalassemia major

21. The valuable diagnostic feature that differentiate anemia of chronic disorder from iron deficiency anemia is

A. low serum iron
B. **low or normal serum transferrin**
C. normal bone marrow cellularity
D. normochromic normocytic RBC
E. leukocytosis

Ans B The serum iron level in anemia of chronic disorder is low, but without the increase in total iron-binding capacity (serum transferrin) that occurs in iron deficiency. This pattern of low serum iron and low to normal iron-binding protein (serum transferrin) is a regular and valuable diagnostic feature.

22. The first laboratory marker in progressive iron deficiency anemia is

A. **depletion of bone marrow hemosiderin**
B. falling of serum ferritin
C. decrease of serum iron and increase of the iron-binding capacity
D. decrease hemoglobin synthesis
E. hypochromic microcytic anemia

Ans A In progressive iron deficiency, a sequence of biochemical and hematologic events occurs. First, the tissue iron stores represented by bone marrow hemosiderin disappear.

23. A 9-month-old child with a hemoglobin concentration of 10 gm/dL and marked microcytosis; serum iron and total iron binding capacity are within normal limits; serum ferritin and hemoglobin electrophoresis are also normal. Of the following, the MOST likely diagnosis is

A. iron deficiency anemia
B. sideroblastic anemia
C. ß-thalassemia minor
D. **α-thalassemia trait**
E. anemia due to chronic disease

Ans D The α-thalassemia traits present as a microcytic anemia that can be mistaken for iron-deficiency anemia. The hemoglobin electrophoresis is normal. The diagnosis is confirmed by DNA testing.

24. All the following conditions may be associated with iron deficiency EXCEPT

A. hookworm infestation
B. prolonged intravascular hemolysis
C. celiac disease
D. congenital heart disease with right to left shunt
E. **prolonged use of isoniazid (INH)**

Ans E Secondary iron deficiency may complicate the intravascular hemolysis because of urinary hemoglobin and hemosiderin iron loss. Cardiovascular defects involving right-to-left shunts interfere with proper oxygenation leading to secondary polycythemia which may cause iron deficiency due to increased RBC demands production. INH therapy may cause secondary sideroblastic anemia with high serum iron and increased transferrin saturation.

Megaloblastic Anemia

25. Best test for assessment of iron status is:

A. Transferrin
B. **Ferritin**
C. Serum iron
D. Hemoglobin

Ans B Serum ferritin level indicates the amount of iron present in both liver and reticuloendothelial system and hence is the best assessment of iron status.

26. Megaloblastic anemia due to folic acid deficiency is commonly due to:

A. **Inadequate dietary intake**
B. Defective intestinal absorption
C. Absence of folic acid binding protein in serum
D. Absence of glutamic acid in the intestine

Ans A • Folic acid deficiency is primarily due to insufficient intake and increased demand with
• Vitamin B12 deficiency is primarily due to malabsorption.

27. Dry tap is a feature of:

A. Anemia of chronic disease
B. Megaloblastic anemia
C. **Aplastic anemia**
D. Sickle cell anemia

Ans C

28. Bimodal distribution is seen in:

A. ALL
B. DLBCL
C. **Hodgkin's Lymphoma**
D. CML

Ans C

29. Transient myeloproliferative disorder of the newborn is seen in association with:

A. Turner syndrome
B. **Down syndrome**
C. Neurofibromatosis
D. Ataxia telangiectasia

Ans B 'Neonates and infants with Down Syndrome may have a condition known as 'transient myeloproliferative syndrome which mimics congenital leukemia'.

30. Low molecular weight heparins are used very frequently to prevent propagation of thrombus in children with stroke. What is the laboratory test used in blood to monitor the LMW Heparins?

A. **Factor V**
B. Factor VI
C. Protein C
D. Protein S

Ans A Anti factor X a Low Molecular Weight Heparin (LMWH) use in children:
• Because of ease of dosing and need for less monitoring, LMWH is being used more frequently in pediatric patients.
• Unlike unfractionated Heparin, which is monitored using the aPTT, LMWH is monitored via anti-Xa activity.
• LMWH formulation that has been used most often in pediatric patients is enoxaparin
• Recommended standard starting dose of enoxaparin for infants <2 mo is 1.5 mg/kg/dose subcutaneously every 12 hr; for patients >2 mo it

31. The main stay of treatment for congenital hypoplastic anemia is

A. **corticosteroids**
B. androgen
C. antithymocyte globulin (ATG)
D. fully matched-related stem cell transplantation
E. fully matched-unrelated stem cell transplantation

Ans A Corticosteroid therapy is beneficial in three fourths of patients; the mechanism of its effect is unknown. Stem cell transplantation from a related histocompatible donor has a role in children who do not respond to corticosteroids and who have demonstrated a several-year need for RBC transfusions; The survival results for matched-related donors have been very encouraging, but the responses have been much inferior with the use of partially mismatched siblings or matched unrelated donors.

32. Folic acid deficiency may develop in the following children EXCEPT

A. a term infant fed on goat`s milk
B. an infant fed on unfortified powdered milk
C. **an infant fed on pasteurized cow's milk**
D. a child with chronic use of phenytoin anticonvulsant
E. a child with trimethoprim prophylaxis for recurrent urinary tract infection

Ans C Goat's milk and powdered milk are deficient with folic acid. Human breast milk, pasteurized cow's milk and infant formulas provide adequate amounts of folic acid. Anticonvulsants may decrease folate absorption. Trimethoprim may affect folate metabolism.

33. A 1-year-old child has folic acid deficiency since the age of 4 months, the best indicator of this deficiency is

A. significant increment of mean corpuscular volume MCV
B. significant fall of reticulocytes count
C. high level of lactate dehydrogenase LDH
D. **decrease level of RBC folate**
E. increase number of hypersegmented neutrophils

Ans D

34. A healthy 5-mo-old boy appears pale. Examination is unremarkable. Lab findings include: Hb 8.1 g/dl; WBC 4,800/mm3; platelets 144,000/mm3; MCV, 111 fl; blood film showed hypersegmented neutrophils; serum B12, 65 pg/mL (low). The infant is vigorously breast fed. Of the following, the NEXT step of the management is

A. performing Schilling test
B. **check serum B12 of the mother**
C. parenteral administration of vitamin B12
D. administration of both folic acid and vitamin B12
E. transfuse packed RBCs

Ans B Older children and adults have sufficient vitamin B12 stores to last 3-5 yr. However, in young infants born to mothers with low vitamin B12 stores (breast fed infants of vegans mothers or having pernicious anemia), clinical signs of cobalamin deficiency can become apparent in the first 4-5 mo of life.

Hemolytic Anemia - General

35. Iron metabolism and regulation are important for RBC precursor cell. Which of the following helps in regulation of iron metabolism but is not specific for iron?

A. Hepcidin
B. **DMT-1**
C. Ferroportin
D. Ferritin

Ans B DMT1 has been shown to transport a variety of divalent metal ions, including Mn2+, Co2+, Cu2+, Zn2+, Cd2+, and Pb2+. (Hence non-specific).

36. Which of the following is ABSENT in hemolytic anemia?

A. Increased indirect bilirubin
B. **Increased direct bilirubin**
C. Increased reticulocyte count
D. Jaundice
E. Increased Hematocrit

Ans B and e. Increased Hematocrit

37. Direct Coomb's test is positive in hemolytic anemia due to:

A. Paraxysmal cold hemoglobinuria
B. Paroxysmal nocturnal hemoglobinuria
C. Idiopathic thrombocytopenic purpura
D. Hemolytic uremic syndrome

Ans A 340 Direct globulin test is positive in Paroxysmal cold hemoglobinuria, as anti P antibody is formed.

38. Hemolysis in G6PD (glucose 6 phosphate dehydro- Questions genase) enzyme deficiency may occur with all of the following drugs except:

A. Primaquine
B. Phenacetin
C. Probenecid
D. Penicillin

Ans D

39. In G6PD deficiency, which cells are more prone for hemolysis?

A. Older red cells
B. Young red cells
C. Reticulocytes
D. All are susceptible

Ans A Because mature red cells do not synthesize new proteins, G6PD enzyme activity falls quickly to levels inadequate to protect against oxidant stress as red cells age. Thus, older red cells are much more prone to hemolysis than younger ones.

40. Drug which may lead to hemolysis in a child with G6PD deficiency is?

A. Penicillin
B. Primaquine
C. Ceftriaxone
D. Erythromycin

Ans B

41. Which of the following is not associated with a high reticulocyte count?

A. Acute bleed
B. Hemolytic anemia
C. Megaloblastic anemia
D. Response to treatment in nutrition deficiency anemia

Ans C is characterized by ineffective erythropoiesis and a low (not high) reticulocyte count despite anemia, unlike hemolysis, acute blood loss, or response to treatment of a nutritional deficiency.

42. All the following may cause autoimmune hemolytic anemia EXCEPT

A. systemic lupus erythematosus (SLE)
B. immunodeficiency
C. mycoplasma pneumoniae infection
D. lymphoproliferative disorder
E. penicillin drug administration

Ans E Penicillin may cause hemolysis via the "hapten" mechanism (immune but not autoimmune) by binding tightly to the RBC membrane. Antibodies to the drug, either newly or previously formed, bind to the drug molecules on RBCs, mediating their destruction in the spleen.

43. Fragmentation hemolysis by mechanical injury may be seen in all the following

A. extensive burns
B. Kasabach-Merritt syndrome
C. after cardiac surgery for prosthetic heart valve replacement
D. thrombotic thrombocytopenic purpura (TTP)
E. hemolytic uremic syndrome (HUS)

Ans A may directly damage the RBCs and result in hemolysis that results in the formation of spherocytes. Blood loss and marrow suppression may contribute to anemia and require blood transfusion; others cause RBC destruction by mechanical injury.

44. A previously healthy 10-month-old boy develops pallor over 2 weeks. Examination is unremarkable except for pallor. Lab tests includes: hemoglobin, 4.9 g/dL; WBC, 6700/mm3; platelets count, 180000/mm3; MCV, 79 fL; reticulocyte count 0.8%; hemoglobin electrophoresis, normal. Of the following, the MOST likely cause for these findings is

A. pure red cell aplasia
B. transient erythroblastopenia of childhood
C. thalassemia minor
D. aplastic anemia
E. parvovirus infection

Ans B Normal hemoglobin electrophoresis usually excludes thalassemia and Diamond- Blackfan syndrome. Anemia due to parvovirus infection usually occurs in children with hemolytic anemia or immunodeficiency. In aplastic anemia, the anemia is usually macrocytic rather than normocytic.

Sickle Cell Disease

45. Iron absorption is increased by:

A. Phytates
B. Tannates
C. Plant food
D. Ascorbic acid

Ans D Iron Absorption
• Site of iron absorption: duodenum and upper jejunum; Ferrous form (Fe2+) of Iron is absorbedQ
• Transport of Fe2+ into enterocyte occurs via DMT1Q Increased by Decreased by
• Acids e.g. ascorbic acid (Vitamin C)Q • Alkalies • Tetracycline
• Amino acid containing SH-group • Phosphates and PhytatesQ
• Presence of other food in stomach

46. Which of the following can result in dactylitis?

A. Hemophilia
B. Von Willebrand disease
C. Thalassemia
D. Sickle cell anemia

Ans D Dactylitis, referred to as hand-foot syndrome, is often the first manifestation of pain in infants and young children with sickle cell anemia Occurring in 50% of children with sickle cell disease by their 2nd yr of life Dactylitis often manifests with symmetric or unilateral swelling of the hands and/or feet.

47. Best treatment for Sickle cell anemia is:

A. Hydroxyurea
B. Sulphonamide
C. Iron Injection
D. Blood transfusion

Ans A Hydroxyurea increases HbF levels and so is used for treatment of sickle cell anemia.

48. **Mutation in Sickle cell anemia is:**
A. Frameshift
B. Non sense mutation
C. **Non conservative missense**
D. Complete

Ans C

49. **In sickle cell anemia, all are true except:**
A. Sickle cells
B. Target cells
C. Howell Jolly bodies
D. **Ringed sideroblast**

Ans D Ringed sideroblast is seen in cases of Iron overload and not in sickle cell anemia.

50. **Person having heterozygous sickle cell trait is protected from infection of:**
A. **Plasmodium falciparum**
B. P. vivax
C. Pneumococcus
D. Salmonella

Ans A

51. **True about sickle cell anemia are all except:**
A. **Sequestration is common in older children**
B. Infection is major cause of mortality
C. Glutamic acid is replaced by valine
D. Common infections are by encapsulated bacteria

Ans A Acute splenic sequestration is a life-threatening complication occurring primarily in infants and young children with sickle cell anemia. Sequestration can occur as early as 5 wk of age, but most often occurs in children between 6 mo and 2 yr.

52. **All of the following statements are true about sickle cell disease except:**
A. Patient may require frequent blood transfusions
B. Acute infection is the most common cause of mortality before 3 years of age
C. There is positive correlation between concentration of. HbS and polymerisation of HbS
D. **Patient present early in life before 6 months of life**

Ans D • Patients rarely present before 6 months due to abundance of HbF which inhibits polymerisation of Hb S.

53. **The primary defect which leads to sickle cell anemia is:**
A. An abnormality in porphyrin part of hemoglobin
B. **Replacement of glutamate by valine in β-chain of HbA**
C. A nonsense mutation in the β-chain of HbA
D. Substitution of valine by glutamate in the β-chain of HbA.

Ans B

54. **Which of the following is not seen on hemoglobin electrophoresis in sickle cell anemia?**
A. **Hb A**
B. Hb A2
C. Hb F
D. Hb S

Ans A • Hb electrophoresis shows 80–90% of HbS and the rest is HbF and HbA2, however HbA is absent.

55. **Sickle cell trait patient do not have manifestations as that of sickle cell disease because:**
A. 50% HbS is required for occurrence of sickling
B. HbA prevents sickling
C. 50% sickles
D. **HbA prevents polymerisation of HbS**

Ans D • Sickling depends on multiple factors like pH, ionic strength, PaO level and not only on HbS%
• Increased HbF and HbA (Hb F > Hb A) concentration prevents polymerisation of HbS.

56. **All the following may occur as a renal manifestation in a child with sickle cell anemia EXCEPT**
A. polyuria
B. hyposthenuria
C. nephrotic syndrome
D. hematuria
E. **acute renal injury**

Ans E Renal function is progressively impaired by diffuse glomerular and tubular fibrosis leading to chronic, rather than acute, renal shutdown.

57. **The least common infection that may occur in a 6-year-old child with sickle cell anemia is**
A. Pneumococcus sepsis
B. Haemophilus influenzae meningitis
C. Salmonella osteomyelitis
D. **E.coli urinary tract infection**
E. Parvovirus B19 aplastic episode

Ans D There is an increased incidence of urinary tract infection with bacteremia in young women with sickle cell anemia.

58. **Blood transfusion therapy in a child with sickle cell anemia is indicated in all the following conditions EXCEPT**
A. acute chest syndrome
B. stroke
C. chronic intolerable pain syndrome
D. **avascular necrosis of the hip joint**
E. splenic sequestration

Ans D Blood transfusions usually do not reverse ischemic damage. Even while on chronic transfusion therapy children can still have transient cerebral ischemia as well as avascular necrosis of the hips and shoulders.

Thalassemia

59. **Microcytic anemia is not seen in which of the following condition?**
A. Osteomyelitis
B. **Leukopenia**
C. Papillary necrosis
D. Rheumatoid arthritis

Ans B Differential diagnosis of microcytic hypochromic RBCs include
• Iron Deficiency Anemia • Thalassemia
• Anemia of Inflammation/chronic disease • Sideroblastic Anemia

60. A child with Thalassemia major has a history of treatment for cardiac arrhythmia due to iron overload. Now the child came for regular transfusion and while transfusion the child became anxious and developed complaints of backpain. What is the next management?

A. ECG
B. **Stop transfusion and look for clerical errors**
C. Continue transfusion while monitoring for vitals
D. Check for reddish discolouration of urine

Ans B Transfusion reactions: Category 1: Mild reactions Signs Symptoms Possible cause Immediate management Urticaria/ Pruritis (itching) Allergic • Stop transfusion rash • Assess patient • An antihistamine may be required • Transfusion may be restarted if no other signs/symptoms are present • If signs/symptoms worsen treat as Category 2. Category 2: Moderately severe reactions Signs Symptoms Possible cause Immediate management

61. In beta thalassemia, the most common gene mutation in India is:

A. **IVS1 G C**
B. Intron
C. 619 bp deletion
D. 3.7 bp deletion

Ans A IVS1 G → C Most common mutations in β thalassemia in India are IVS-1, position 5 (G → C) followed by 619-bp deletion

62. Screening test for thalassemia:

A. Alkali denaturation test
B. Kleihauer test
C. Hb electrophoresis
D. **NESTROFT**

Ans D Screening test for Thalassemia trait: NESTROFT (Naked Eye Single Tube Red cell Osmotic Fragility Test)Q Assesses osmotic fragility of red cells at a single concentration of buffered saline (0.36% in single tube).

63. Thalassemia gives protection against:

A. Streptococcus pneumoniae
B. H. influenza-B
C. **Myco tuberculosis**
D. All of the above

Ans C Malaria Thalassemia gives a reduced parasite invasion/ growth and an increased susceptibility to phagocytosis of the infected RBC as a malaria-protective effect.

64. In β-thalassemia there is:

A. Excess α-chain
B. Reduced α-chain
C. Excess β-chain
D. **Reduced β-chain**

Ans D In beta-thalassemia there is reduced β chain and relatively excess α chain.

65. The following are the features of thalassemia major except:

A. Bone marrow hyperplasia
B. Hair-on-end appearance
C. Splenomegaly
D. **Increased osmotic fragility**

Ans D c. Megaloblastic anemia - 2321 Megaloblastic anemia causes a low reticulocyte count because it's a nutritional anemia, others being hemolytic anemia have a high reticulocyte count.

66. All of the following are true of thalassemia major, except:

A. Splenomegaly
B. Target cells on peripheral smear
C. Microcytic hypochromic anemia
D. **Increased osmotic fragility**

Ans D Cells that are more spherical, have a limited capacity to expand in hypotonic solutons lyse at a higher concentration of NaCl than do normal biconcave cells → increased osmotic fragility, as in hereditary spherocytosis Cells that are flatter have a greater capacity to expand in hypotonic solutions, lyse at lower concentration, are said to have decreased osmotic fragility e.g. iron deficiency, thalassemia, liver disease

67. Which of the following hemoglobin (Hb) estimation will be diagnostically helpful in a case of beta thalassemia trait?

A. Hb-F
B. HbA
C. **HbA2**
D. Hb-H

Ans C Hb electrophoresis findings in Thalassemia: Type of Hb β thalassemia major β-Thalassemia trait HbA Decreased 90–96% HbA2 5–7% 3.5–7% HbF 10–100% 1–5%

68. Diagnosis of beta thalassemia is established by:

A. NESTROFT Test (AIPGMEE 05)
B. HbA1 c estimation
C. **Hb electrophoresis**
D. Target cells in peripheral smear

Ans C

69. True about β-thalassemia trait is:

A. Increased HbF
B. **Increased HbA2**
C. Microcytosis
D. Severe anemia
E. Hydrops fetalis seen

Ans B Increased HbA2, c. Microcytosis

70. In beta thalassemia, there is:

A. Increase in beta chain, decrease in alpha chain
B. **Decrease in beta chain, increase in alpha chain**
C. Decrease in beta chain, decrease in alpha chain
D. Increase in beta chain, increase in alpha chain

Ans B

71. Thalassemia occurs due to which mutations?

A. Missense
B. **Splicing**
C. Transition
D. Frame-shift
E. Truncation APLASTIC ANEMIA

Ans B and d. Frame-shift Molecular defects in pathogenesis of thalassemia: β-thalassemia:
• Most common type of genetic abnormality in β-thalassemia is point mutation, i.e. nonsense
• Some may also occur due to deletion or insertion i.e. frameshift mutations Mutations leading to aberrant splicing are the most common cause of β-thalassemia α-thalassemia
• The most common cause of reduced α-chain synthesis is deletion of α-globin genes (rarely non sense mutation)

72. Children with Thalassemia and Iron overload are at an increased risk for infection with:

A. **Yersinia Enterocolitica**
B. Campylobacter jejuni
C. Escherichia coli
D. Vibrio cholera

Ans A Iron therapy may increase the virulence of malaria and certain, gram-negative bacteria, particularly in developing countries. Iron overload is associated with Yersinia infection.

73. The best assessment of iron overload for patients with thalassemia major is achieved by

A. **liver MRI**
B. bone marrow biopsy
C. serum iron
D. serum ferritin
E. total iron binding capacity

Ans A Quantitative liver iron by approved MRI technology is the best indicator of total- body iron stores and should be obtained in chronically transfused patients after chronic transfusion therapy has initiated.

74. You are evaluating a 4-year-old child with ß-thalassemia major; he is on chronic transfusion therapy since the age of 1 year; you suspect transfusion-induced hemosiderosis. Of the following, the organ that is LEAST likely to be affected by iron deposition at this time is

A. liver
B. pancreas
C. **heart**
D. pituitary gland
E. thyroid gland

Ans C Iron is initially deposited in the liver. Liver hemosiderosis develops after 1 yr of chronic transfusion therapy and is followed by iron deposition in the endocrine system. This leads to a high rateof hypothyroidism, hypogonadotropic gonadism, growth hormone deficiency, hypoparathyroidism, and diabetes mellitus. After 10 yr of transfusion, cardiac dysfunction secondary to hemosiderosis begins.

G6PD Deficiency

75. True statement about anemia of chronic disease is:

A. **IL-1**
B. IL-6
C. TNF alpha
D. IFN-Y

Ans A Increased serum ferritin

76. Drug that is safe in G6PD deficiency:

A. Primaquine
B. Acetanilide
C. **Quinidine**
D. Dapsone

Ans C Quinidine does not carry risk of hemolysis in persons with G-6-PD deficiency.

77. What is the definitive finding of G6PD?

A. **Bite cells**
B. Intravascular hemolysis
C. Splenomegaly
D. Hemoglobinuria

Ans A Bite cells are characteristic finding of G6PD deficiency while others can be seen in any intravascular hemolysis.

78. Regarding G6PD deficiency true are:

A. Autosomal dominant
B. **Bite cells seen**
C. Protects against kala azar
D. Enzyme level directly proportional to age of RBC
E. Sex preponderance

Ans B and e. Sex preponderance G6PD deficiency is an X-linked inherited disorder which is seen more frequently in boys;

79. Non-specific esterase is present in:

A. Megakaryocytic leukemia
B. Lymphocytic leukemia
C. Erythroleukemia
D. **AML M4**
E. AML M5

Ans D e. AML M5 Monocytic stain is non-specific esterase (NSE) is positive in monocytic leukemia like AML M4 and M5.

80. The best diagnostic test for paroxysmal nocturnal hemoglobinuria (PNH) is

A. acidified serum hemolysis (HAM) test
B. sucrose lysis test
C. complement assay
D. **flow cytometry**
E. bone marrow study

Ans D is the best diagnostic test for PNH. With the use of anti-CD59 for RBCs and anti-CD55 and anti-CD59 for granulocytes, flow cytometry is more sensitive than the classic RBC lysis tests in detecting the reduced glycolipid-bound membrane proteins.

81. In which of the following conditions you should use a plasma that is frozen within 24 hours after collection?

A. factor V deficiency
B. factor VII deficiency
C. factor IX deficiency
D. factor X deficiency
E. factor XI deficiency

Ans A Factor V is lost rapidly from FFP. It is therefore important to use FFP that is less than 2 mo old. Because the plasma half-life of factor VII is 2-4 hr, therapy with FFP is difficult and often complicated by fluid overload.

82. Of the following, the MOST accurate reflection of active erythropoiesis of the bone marrow is

A. hemoglobin concentration
B. packed cell volume (PCV)
C. reticulocyte count
D. mean corpuscular volume (MCV)
E. reticulocyte index

Ans E The Reticulocyte production index (RPI, also called a corrected reticulocyte count) is a calculated value used in the diagnosis of anemia. This calculation is necessary because the raw reticulocyte count is misleading in anemic patients. Reticulocyte index = reticulocyte count× (observed hematocrit/ normal hematocrit).

Hereditary Spherocytosis

83. To diagnose anemia in age group 2 years to 6 years, level of hemoglobin should be:

A. Hb 14g/dL
B. Hb 11g/dL
C. Hb 12g/dL
D. Hb 13g/dL

Ans B

84. A nine-month-old boy of Sindhi parents presented to you with complaints of progressive lethargy, irritability and pallor since 6 months of age. Examination revealed severe pallor. Investigation showed Hb-3.8 g%; MCV- 58 fL, MCH-19.4 pg/ cell. Osmotic fragility is normal; Blood film shows target cells and normoblasts. X-ray skull shows expansion of erythroid marrow. Which of the following is the most likely diagnosis?

A. Iron deficiency anemia
B. Acute lymphoblastic anemia
C. Hemoglobin D disease
D. Hereditary spherocytosis

Ans C HbD disease a. Iron deficiency anemia So severe anemia and expansion of erythroid marrow are usually not seen b. Acute lymphoid Leukemia Low MCV, no obvious involvement of other cell lines, and expansion of erythroid marrow are incompatible with diagnosis of ALL c. Hemoglobin D disease Low MCV and MCH with target cells suggest abnormality of hemoglobin like HbD disease d. Hereditary spherocytosis Normal osmotic fragility goes against hereditary spherocytosis

85. Most common mutation in hereditary spherocytosis:

A. Spectrin
B. Ankyrin
C. Glycophorin A
D. Band

Ans B Most common mutation in Hereditary Spherocytosis are AnkyrinQ > band 3> Spectrin and band 4.2.

86. A 6 years old child belonging to Punjabi family with past history of blood transfusions presented with hemoglobin–3.5 gm/dL, MCV–30 fL. Peripheral smear findings of microcytic hypochromic anemia with target cell and reduced osmotic fragility. The probable diagnosis of patient:

A. Alpha thalassemia
B. Beta thalassemia
C. Sickle cell anemia
D. G6Pd deficiency

Ans B Peripheral smear findings of microcytic hypochromic anemia with target cell and reduced osmotic fragility with severe transfusion requiring anemia is suggestive of Beta thalassemia.

87. In Langerhans Cell Histiocytosis, the characteristic abnormality seen is:

A. Birbecks granules
B. Macrophages
C. Plasma cell
D. Giant cell

Ans A X-bodies called Birbeck granules are characteristically seen in LCH.

88. In Glanzman's thrombasthenia, there is defect of:

A. Gp IIb/IIa
B. Gp IIb/IIIa
C. Gp Ib/IX
D. Gp IIb/IX 193.

Ans B M neMonic Mnemonic "B comes before G and Ib comes before IIb" Disease Deficient Gp Qualitative defect in Bernard Soulier syndrome Ib/IX Platelet adhesion (Bernard-defective adhesion) Glanzmann thrombasthenia IIb/IIIa Platelet aggregation

89. The laboratory finding that is MOST likely consistent with the diagnosis of spherocytosis is

A. high reticulocytes count of more than 10%
B. normal mean corpuscular volume MCV
C. low mean corpuscular hemoglobin MCH
D. high mean corpuscular hemoglobin concentration MCHC
E. indirect hyperbilirubinemia

Ans D The MCHC is a measure of cellular hydration status. A high value (36-38 g/ dL) is characteristic of spherocytosis and a low value is commonly associated with iron deficiency.

90. In hereditary spherocytosis, all the following will resolve postsplenectomy EXCEPT

A. osmotic fragility
B. anemia
C. reticulocytosis
D. hyperbilirubinemia
E. hypoplastic/aplastic crises

Ans A After splenectomy, osmotic fragility often improves because of diminished splenic conditioning and less RBC membrane loss, but not resolved.

91. Increased incubated osmotic fragility test that is not corrected by the addition of glucose is MOST likely suggestive of

A. hereditary spherocytosis
B. hereditary elliptocytosis
C. hereditary stomatocytosis
D. glucose-6-phosphate dehydrogenase deficiency
E. pyruvate kinase deficiency

Ans E Autohemolysis is moderately or markedly increased in pyruvate kinase deficiency, but the addition of glucose does not regularly correct the abnormality as it does in hereditary spherocytosis.

Autoimmune Hemolytic Anemia

92. First change of improvement noted after iron therapy is initiated:

A. Decreased irritability
B. Reticulocytosis
C. Increase in serum iron levels
D. Replenishment of iron stores

Ans A Rapid subjective improvement, with decrease in fatigue & irritability occurs before improvement in anemia

93. Schistocytes are characteristic feature of:

A. Hereditary spherocytosis
B. Microangiopathic hemolytic anemia
C. G6PD deficiency
D. Thalassemias

Ans B Schistocytes are fragmented RBCs, also called 'helmet' cells, found in microangiopathic hemolytic anemia.

94. Prominent reticulocytosis is a feature of:

A. Aplastic Anemia
B. Hemolytic Anemia
C. Nutritional Anemia
D. Anemia of chronic disease

Ans B

95. In infant, Bone Marrow biopsy is done from?

A. Tibia
B. Sternum
C. Posterior superior Iliac spine
D. Iliac crest

Ans A Age group Site of bone marrow sampling Younger children Anterior medial tibialQ area, below tibial tuberosity Adult Anterior or posterior iliac crest area

96. Langerhans cell shows which of these?

A. Badminton racquet appearance
B. CD100a
C. MPO +
D. Birbeck's granules

Ans D Tennis racquet and not badminton racquet appearance is seen in EM of LCH; LCH is positive for CD1a and S100.

97. Affinity of fetal Hemoglobin to oxygen is more than adult hemoglobin due to:

A. 2, 3 BPG in blood
B. Decreased pH
C. Increased pCO
D. Right shift 194.

Ans A • Greater affinity of fetal Hb for oxygen is explained by lack of fetal Hb's interaction with 2,3-bisphosphoglycerate (2,3-BPG or 2,3-DPG); In adult red blood cells, 2,3 DPG decreases the affinity of hemoglobin for oxygen
• 2,3-BPG interacts less efficiently with fetal hemoglobin than adult hemoglobin, due to a change in a single amino acid (residue 143) found in the 2,3-BPG 'binding pocket': from histidine to serine, leading to greater oxygen affinity.

98. The best treatment for spinal cord compression at the vertebral canal with neurological symptoms caused by extramedullary hematopoiesis in thalassemic patients is

A. blood transfusion
B. splenectomy
C. iron chelation therapy
D. local radiotherapy
E. vitamin D supplementation

Ans D Extramedullary hematopoiesis can occur in the vertebral canal, compressing the spinal cord and causing neurologic symptoms; the latter is a medical emergency requiring immediate local radiation therapy to halt erythropoiesis.

99. In severe liver disease with prolongation of the bleeding time, the best effective treatment before doing liver biopsy is

A. intravenous vitamin K
B. intramuscular vitamin K
C. oral vitamin K
D. fresh frozen plasma
E. intravenous DDAVP

Ans E DDAVP (0.3 µg/kg IV) has been found to be effective in shortening the bleeding time and has been used effectively to augment hemostasis prior to liver biopsy.

100. Which of the following is not a feature of Wiskott–Aldrich Syndrome (WAS)

A. thrombocytopenia
B. eczema
C. recurrent otitis media and pneumonia
D. propensity to develop autoimmune disorders
E. giant platelets by blood smear

Ans E

Aplastic Anemia

101. A 6 months old infant with Hb of 6 gm% has peripheral blood showing target cells. Family history of similar complaints is Positive. Diagnosis can be best done with which of the following?

A. Hb electrophoresis
B. ELISA
C. Coomb's Test
D. Osmotic fragility

Ans A • Presence of: 1. Anemia (Hb < 6%), 2. Target cells, 3. Increased reticulocytes suggest the diagnosis of thalassemia
• Out of the investigations given in the options, Hb electrophoresis will help the most in diagnosing Thalassemia.

102. **Fanconi anemia can lead to:**
A. B12 deficiency
B. Folate deficiency
C. Iron deficiency
D. **Aplastic anemia**

Ans D Fanconi anemia can lead to Pancytopenia with Hypocellular Bone Marrow.

103. **Which of the following conditions does not cause pancytopenia?**
A. Hypersplenism
B. Aplastic anemia
C. Cancer infiltrating the bone-marrow
D. **Hemolysis from G6PD enzyme deficiency**

Ans D

104. **Pancytopenia with hypocellular bone marrow seen in:**
A. **Fanconi anemia**
B. Dyskeratosis congenita
C. Tuberous sclerosis
D. Osteogenesis imperfecta

Ans A Fanconi's anemia Fanconi's anemia shows pancytopenia with hypocellular bone marrow while others in the options cause hypercellular bone marrow.

105. **Pancytopenia with cellular bone marrow is seen in all except:**
A. Megaloblastic Anemia
B. MDS
C. PNH
D. **G6PD deficiency**

Ans D G6PD deficiency causes hemolytic anemia and not pancytopenia.

106. **Aplastic anemia can progress to all except:**
A. AML
B. Myelodysplastic anemia
C. **Pure red cell aplasia**
D. Paroxysmal nocturnal hemoglobinuria

Ans C Aplastic anemia can progress to AML, PNH and Myelodysplastic syndrome.

107. **Constitutional pancytopenia can be seen in following except:**
A. Fanconi anemia
B. **Diamond-Blackfan syndrome**
C. Dyskeratosis congenita
D. Schwachman Diamond syndrome

Ans B Diamond-Blackfan syndrome causes PRCA and not aplastic anemia.

108. **Parvo virus B19 infection may cause transient pancytopenia in**
A. thalassemia major
B. **sickle cell anemia**
C. hereditary spherocytosis
D. hereditary elliptocytosis
E. pyruvate kinase deficiency

Ans B Parvovirus B19 is classically associated with isolated red cell aplasia, but in patients with sickle cell disease or immunodeficiencies it can result in transient pancytopenia.

109. **A 5-year-old child has multiple bruises on her extremities and oral mucosal bleeding of 3 days duration; she had a mild respiratory tract infection 2 weeks before; physical examination was normal apart from multiple ecchymoses and petechiae. Of the following, the NEXT diagnostic step is**
A. **complete blood count**
B. prothrombin time
C. bleeding time
D. partial thromboplastin time
E. antinuclear antibody titer

Ans A

Hemophilia & Coagulation Disorders

110. **Iron requirement is determined from the equation:**
A. 3 × wt(kg) × Hb deficit (gm/dL) (AMU 05)
B. 3.3 × wt(kg) × Hb deficit (gm/dL)
C. **4 × wt(kg) × Hb deficit (gm/dL)**
D. 4.3 × wt(kg) × Hb deficit (gm/dL) HEMOLYTIC ANEMIA

Ans C 4 × wt (kg) × Hb deficit (gM/dL) • Blood volume averages 75 to 80 mL/kg. Each gram of hemoglobin contains 3.4 mg of iron. 50% more iron should be given to replenish iron stores. Therefore iron requirements are determined from the equation. Iron requirement (mg) = wt (kg) × Hb deficit (g/dL) × 80/100 × 3.4 × 1.5. = wt (kg) × Hb deficit (g/dL) × 4.

111. **Best treatment of atypical HUS is:**
A. **Plasmapheresis**
B. Antibiotics
C. Ivlg
D. Dialysis

Ans A In HUS, Fresh frozen plasma can induce remission by replenishing the missing Complement Factor H or I or removing the antibodies.

112. **Kostmann's syndrome—treatment is:**
A. Antithymocyte globulin + cyclosporin
B. Antithymocyte globulin + cyclosporine + GM-CSF
C. **G-CSF**
D. GM-CSF NON-NEOPLASTIC DISORDERS OF WBC

Ans C Kostman Syndrome (Severe congenital neutropenia)
• Arrest in myeloid maturation at promyelocyte stage in bone marrow resulting in consistent ANCs < 200/μL
• This disorder occurs sporadically or with autosomal dominant (ELA2 mutation) or recessive (HAXI) inheritance
• Monocytosis, eosinophilia, recurrent, severe pyogenic infections, and anemia of chronic inflammation are usually present
• 20% of patients develop AML or myelodysplasia associated with monosomy 7 Treatment: Usually respond to administration of G-CSF;

113. A 10-year-child presented with bilateral cervical lymphadenopathy. Lymph node biopsy was performed, which showed cells as given in the figure. Which of the following is true regarding this condition?

A. Hodgkin lymphoma; EBV and embryo cell
B. Non Hodgkin lymphoma; HIV and Giant B cell
C. TB, Mycobacteria and tiny granuloma
D. **Hodgkin lymphoma: EBV and Reed Sternberg cell**

Ans D The given cell shows Reed Sternberg cell seen in Hodgkin lymphoma and caused by EBV.

114. CD marker of histiocytosisis:

A. **Letterer-Siwe disease**
B. Pulmonary Langerhans cell histiocytosis
C. Hand-Schuller-Christian disease
D. Eosinophilic granuloma

Ans A CD 1a Immunohistochemistry shows: CD1a, S-100 or Langerin (CD 207) in Histiocytosis.

115. Deficiency of factor VIII leads to:

A. **Hemophilia**
B. Christmas disease
C. Von Willebrand disease
D. Bernard Soulier syndrome

Ans A

116. Characteristic lab findings of hemophilia A are:

A. Codominant
B. **Autosomal recessive**
C. Autosomal dominant
D. X-linked recessive Chapter 14: Pediatric Hematology 337 Answers
E. Normal aPTT The coagulation profile in a 13 year old girl with Menorrhagia having von Willebrands disease is:

Ans B Increased aPTT, c. X-linked recessive

117. Transfusion of fresh frozen plasma (FFP) is efficacious for the treatment of deficiency of all the following coagulation factors EXCEPT

A. factor V
B. factor X
C. factor XI
D. **factor XIII**
E. protein C

Ans D Transfusion of FFP is efficacious for the treatment of deficiencies of clotting factors II, V, X, and XI. Factor XIII and fibrinogen deficiencies are treated with cryoprecipitate. FFP also contains several anticoagulant proteins (antithrombin III, protein C, and protein S), whose deficiencies have been associated with thrombosis.

118. In hemophilia A, factor VIII level activity should be increased to 100% in

A. epistaxis
B. **iliopsoas bleeding**
C. tooth extraction
D. hematuria
E. gum bleeding

Ans B

119. A 5-year-old child, with hemophilia A of severe type, presents to the emergency unit with a groin pain after a minor trauma to his back; his blood pressure is 60/30 mm Hg; his pulse rate is 180/min; he holds his right hip in a flexion position with internal rotation. Of the following, the NEXT step in the management of this child is

A. **factor VIII replacement therapy**
B. intravenous 1-deamino-8-d-arginine vasopressin (DDAVP)
C. factor VIII assay
D. abdominal ultrasonography
E. abdominal computed tomography (CT) scan

Ans A Iliopsoas bleeding is life threatening that needs immediate factor replacement therapy.

120. In a child with a recent diagnosis of hemophilia A who developed moderate epistaxis, the aim of the treatment is to increase the level of factor VIII to

A. 5-20%
B. **35-50%**
C. 55-60%
D. 65-70%
E. 80-100%

Ans B When mild to moderate bleeding occurs, level of factor VIII or IX must be raised to a hemostatic level, in the range of 35-50%.

121. The standard care for MOST children with severe hemophilia is

A. **prevention by F VIII replacement therapy to prevent spontaneous bleeding and early joint deformities**
B. aggressive treatment by F VIII replacement therapy when significant bleeding occurs
C. avoid trauma
D. avoid aspirin and other NSAID
E. avoid violent contact sports

Ans A

Von Willebrand Disease

122. A child aged 2 years presents with non-specific symptoms suggestive of anemia. On examination, hepato- spleno megaly is present. Peripheral smear shows target cells. He has microcytic hypochromic picture & Hb of 6 gm%. There is positive family history. Next investigation is:

A. NESTROFT test
B. HbA1c estimation
C. **Hb electrophoresis**
D. Osmotic fragility test

Ans C Microcytic hypochromia anemia, in a child with hepatosplenomegaly & positive family history, suggest a diagnosis of Thalassemia. Hb electrophoresis or Hb HPLC can be done for diagnosing Thalassemia. NESTROFT test (Naked Eye Single Tube Red Cell Osmotic Fragility Test)
• Normally, red cells put in saline solution begin to lyse at a saline concentration of 0.4-0.5% and lysis is complete at 0.32%.

123.Salmonellosis is most common in:

A. Hb-F
B. Hb -C
C. Hb-A2
D. Hb-H

Ans A Sickle cell anemia and 2339, Ghai 8/e p
• Children with sickle cell disease are at increased risk for Salmonella septicemia and osteomyelitis.

124.Serious infections can occur when absolute neutrophil count decreases to?

A. Less than 500/uL
B. Less than 800/uL
C. Less than 1000/uL
D. Less than 1500/uL

Ans A Less than 500/mL Serious infections are most likely when the neutrophil count falls below 500 per mm3.

125.A 10-year-old boy presents with mass in the abdomen. On imaging, the paraaortic LN is enlarged. On biopsy, starry sky appearance is seen. What is the underlying abnormality?

A. p53 gene mutation
B. RB gene mutation
C. Translocation involving BCR-ABL genes
D. Translocation involving MYC gene

Ans D /e p 600
• Starry sky appearance is a feature of Burkitt lymphoma
• The genetic hallmark of Burkitt is translocation involving MYC gene on chr 8.

126.A 2-year-old child presents with scattered lytic lesions in the skull. Biopsy revealed Langerhans giant cells. The most commonly associated marker with this condition will be:

A. CD Ia
B. CD57
C. CD3
D. CD68

Ans A

127. The main reason behind gamma irradiation of the blood products is to prevent

A. transfusion-related bacterial infections
B. transfusion-related viral infections
C. graft-versus-host disease (GVHD)
D. allergic reactions
E. transfusion-related acute lung injury (TRALI)

Ans C

128.Which of the following is FALSE about factor V Leiden mutation?

A. it is the most common inherited risk factor for thrombosis
B. heterozygous individuals have less risk for thrombosis than homozygotes
C. heterozygotes have an increased risk of arterial thrombosis
D. there is an increased frequency of thrombosis while receiving oral contraceptive agents
E. there is an increased risk of recurrent abortions

Ans C Individuals who are heterozygous have a 5-7–fold increase in risk of venous thrombosis

129.Of the following, the LEAST likely feature of Bernard-Soulier syndrome is

A. autosomal recessive inheritance
B. severe thrombocytopenia
C. giant Platelets
D. abnormal ristocetin test
E. platelet transfusion is the only reliable therapy

Ans B Bernard-Soulier Syndromeis characterized by moderate, rather than severe, thrombocytopenia.

ITP & Platelet Disorders

130.Hair on end appearance is seen in:

A. Dermoid cyst
B. Thalassemia
C. Kwashiorkar
D. Tinea capitis

Ans B

131. HbA2 is increased in:

A. Beta thalassemia trait
B. α-thalassemia
C. alpha-thalassemia trait
D. All of above

Ans A HbA2 is increased (>3.5%) in beta thalassemia trait.

132.Eosinophilia is found in infection with:

A. Cryptococcus
B. HPV
C. Strongyloides
D. Typhoid

Ans C Parasitic infestations: Ascariasis, Hookworm, Strongyloides, Filariasis, Trichinosis can cause eosinophillia.

133.Common childhood tumors are:

A. Acute lymphoblastic leukemia
B. Chronic lymphoblastic leukemia
C. Chronic myeloid leukemia
D. Brain tumor
E. Wilms' tumor

Ans A d. Brain tumor, e. Wilms' tumor Common Childhood Tumors are: Leukemia (ALL) > Brain tumors > Lymphoma > Neuroblastoma > Wilm's tumor. about childhood tumors
• Most common childhood leukemia is ALL • Most common tumor in infancy is Neuroblastoma
• Most common solid tumor of childhood is Brain tumor • Most common abdominal tumor in child is Neuroblastoma

134.Which of the following drugs is used for the treatment of refractory histiocytosis?

A. High dose methotrexate
B. High dose cytarabine
C. Cladribine
D. Fludrabine

Ans C Treatment of LCH
• Single system disease: spontaneous remission seen; curettage or local radiotherapy;
• Multisystem disease: Multiagent chemotherapy, which may include vinblastine, etoposide, prednisolone
• For unresponsive disease: Cyclosporine/antithymocyte globulin, Imatinib, Cladribine or Stem cell transplantation.

135.In Alloimmune thrombocytopenia antigen against which antibody is found?

A. HPA 1a
B. HPA 1b
C. HPA 2a
D. HPA 2b

Ans A • Neonatal alloimmune thrombocytopenic purpura (NATP) is caused by development of maternal antibodies against antigens present on fetal platelets that are shared with father and recognized as foreign by maternal immune system.
• The most common cause is incompatibility for the platelet alloantigen HPA-1a.

136.True about acute ITP:

A. More common in females
B. Specific anti platelet antibodies detected
C. Viral infection predisposes, as seen after vaccination
D. 80% cases transform to chronic
E. More common in children

Ans C Viral infection predisposes and e. More common in children 2407 a. False; In childhood, males and females are equally affected b. False; Platelet antibody testing is seldom useful in acute ITP. c. True; A recent history of viral illness is described in 50-65% of cases of childhood ITP d. False; 20% of children (and 50% of adolescents) who present with acute ITP go on to have chronic ITP. e. True; The peak age is 1-4 yr, although the age ranges from early in infancy to the elderly. a.

137. Quinine induced thrombocytopenia is:

A. Antibody mediated
B. Dose related toxicity
C. Idiosyncratic reaction
D. Inhibits production of platelets

Ans A Quinine-induced thrombocytopenia is an antibody-mediated (drug-dependent antibody) immune reaction against platelets.

138.Which of the following is generally not seen in idiopathic thrombocytopenic purpura (ITP)?

A. More common in females
B. Petechiae, ecchymosis and bleeding
C. Palpable splenomegaly
D. Increased megakaryocytes in bone marrow

Ans C a. Chronic ITP occurs most commonly in adult women, younger than age 40 years (Female: Male = 4:1) b. Cutaneous bleeding occurs in the form of pinpoint hemorrhages, petechiae or ecchymoses. Often there will be history of easy bruising, nose bleeds, bleeding from gums and hemorrhage in soft tissues from relatively minor trauma. c. Despite the increased destruction of platelets in spleen, spleen size usually remains normal in ITP. d. Bone marrow reveals a modestly increased number of megakaryocytes.

139.Platelet function defect is seen in:

A. Glanzmann syndrome
B. Bernard-Soulier syndrome
C. Wiskott-Aldrich syndrome
D. von Willebrand disease
E. Weber Christian disease

Ans A b. Bernard-Soulier syndrome, c. Wiskott-Aldrich syndrome Platelet function defect can be seen in:
• Glanzmann thrombasthenia • Hermansky-Pudlak syndrome • Wiskott-Aldrich syndrome
• Bernard-Soulier syndrome • Chediak-Higashi syndrome • Grey platelet syndrome.

140.Platelet function is assessed by:

A. Platelet adhesion
B. BT
C. CT
D. PTT
E. PFA-100

Ans A b. BT, e. PFA-100 Platelet function assessment: weiveR • PFA-100—This is a test to measure the platelet adhesion-aggregation in response to agonist combination like collagen-epinephrine or collagen-ADP.
• Bleeding time—The usual bleeding time is 4–8 minutes. Bleeding for a longer time suggests platelet defects.

141. The following laboratory determinant is abnormally prolonged in ITP:

A. APTT
B. Prothrombin time
C. Bleeding time
D. Clotting time COAGULATION DISORDERS

Ans C • ITP is characterized by thrombocytopenia which prolongs the bleeding time.

142.Immune destruction of platelet is seen in:

A. SLE
B. DIC

Ans A SLE Answers

143.Which of the following is a qualitative defect of platelets?

A. Immune thrombocytopenic purpura
B. Bernard Soulier syndrome
C. Systemic Lupus erythematosus
D. Von Willebrand disease

Ans B

144.Low platelets count may be encountered in all the following EXCEPT

A. Von Willebrand disease (VWD) type 2 B
B. VWD type 2 N
C. Platelet type pseudo VWD
D. Bernard Soulier syndrome
E. Wiskott Aldrich syndrome

Ans B Type 2N VWD is caused by the reduction of factor VIII binding by VWF. This disorder has also been termed autosomal hemophilia. With this variant, platelet interaction with VWF is normal but the 2N VWF binds weakly (or not at all) to factor VIII, resulting in rapid clearance of factor VIII that is weakly complexed to VWF.

145. **A healthy 5-day-old male neonate develops bruising and melena. The pregnancy, and delivery course were unremarkable. The mother received heparin therapy for recurrent abortions secondary to lupus anticoagulants. The infant is vigorously breastfeeding. Examination reveals only multiple bruises. Lab tests reveals: hemoglobin, 8.4 g/dL; WBC, 8600/mm3; platelets count, 200000/mm3; PTT, 74 seconds; PT, 28 seconds; serum fibrinogen is normal. Of the following, the MOST likely cause of this bleeding is**

A. disseminated intravascular coagulopathy (DIC)
B. hemophilia
C. **vitamin K deficiency**
D. heparin therapy
E. lupus anticoagulants

Ans C Heparin does not cross the placenta, and so it does not affect the baby. DIC causes prolonged PTT, PT, thrombocytopenia and low serum fibrinogen. Hemophilia causes prolonged PTT only. Lupus anticoagulants may cause thrombosis rather than bleeding and usually associated with prolonged PTT because of the presence of antiphospholipids that affect the phospholipid reagent.

146. **You are evaluating a 5-year-old male child with multiple ecchymoses distributed all over the extremities and trunk; examination reveal a palpable spleen 3 cm below the left costal margin. Lab findings include: hemoglobin, 11.3 g/dL; white blood cell count, 8700/mm3; platelets count, 21000/mm3. Of the following, the most proper NEXT step is**

A. bleeding time
B. coagulation profile
C. platelets aggregation test
D. abdominal ultrasonography
E. **bone marrow study**

Ans E Although spleen may be palpable in 10% of children with ITP, bone marrow examination is mandatory when there is an unusual presentation.

147. **Both quantitative and qualitative platelets defects are seen in**

A. generalized vasculitis
B. **Bernard-Soulier syndrome**
C. Glanzmann's Thrombasthenia
D. type I von Willebrand disease
E. uremia

Ans B In Bernard-Soulier syndrome there is moderate thrombocytopenia and abnormal ristocetin test.

148. **Which of the following viruses is more likely to be associated with chronic immune thrombocytopenia?**

A. cytomegalovirus
B. parvovirus
C. influenza virus
D. parainfluenza virus
E. **human immune deficiency virus**

Ans E

149. **Which of the following is least likely to be associated with prolonged thrombocytopenia in children?**

A. **immune thrombocytopenia**
B. systemic lupus erythematosus (SLE)
C. HIV infection
D. type 2B von Willebrand disease
E. Wiskott-Aldrich syndrome (WAS)

Ans A In 70-80% of children who present with acute ITP, spontaneous resolution of their ITP will occur within 6 mo.

150. **Which of the following drugs is least likely to cause thrombocytopenia in children?**

A. valproic acid
B. phenytoin
C. sulfonamides
D. trimethoprim-sulfamethoxazole
E. **heparin**

Ans E Common drugs used in pediatrics that cause thrombocytopenia include valproic acid, phenytoin, sulfonamides, and trimethoprim-sulfamethoxazole. Heparin-induced thrombocytopenia is seldom seen in pediatrics.

151. **Thrombocytopenia that improves with age is MOST likely a feature of**

A. congenital amegakaryocytic thrombocytopenia
B. Bernard-Soulier syndrome
C. Wiskott-Aldrich syndrome
D. **thrombocytopenia absent radius syndrome (TAR)**
E. aplastic anemia

Ans D Thrombocytopenia of TAR syndrome frequently remits over the first few years of life.

152. **You are going to explain the role of anti-D therapy to the parents of a 4-year-old boy with acute immune thrombocytopenia (ITP) who has several ecchymotic lesions located over extremities and trunk. There is no mucosal bleeding. An important statement that should be included during the discussion is**

A. anti-D therapy is more effective than IVIG therapy in acute ITP
B. anti-D therapy is very effective to all children with ITP
C. **when anti-D therapy is given, monitoring of hemoglobin concentration is recommended**
D. anti-D therapy is used in acute ITP only
E. anti-D therapy can be used as initial treatment for children with acute ITP

Ans C The role of IV anti-D in initial therapy of acute ITP is under investigation; the response appears to be somewhat slower than after IVIG; it is effective for Rh+ children; it can be used in acute or chronic ITP.

153. **Each of the following may cause thrombocytopenia EXCEPT**

A. heparin therapy
B. **aspirin ingestion**
C. disseminated intravascular coagulation (DIC)
D. systemic lupus erythematosus (SLE)
E. hemolytic-uremic syndrome (HUS)

Ans B Aspirin may affect platelets function rather than the count.

154. Which of the following children with acute immune thrombocytopenia is MOST likely to have a benefit from splenectomy?

A. an 8-year-old girl with multiple bruises and platelets count of 10,000/ml
B. a 6-year-old boy with mild epistaxis and platelets count of 20,000/ml
C. a 4-year-old boy with subdural hematoma and platelets count of 100,000/ml after receiving IVIG
D. an 11-year-old boy with severe rectal bleeding and platelets count of 20000/ml not responding to medical treatment
E. a 13-year-old female adolescent with moderate menstrual bleeding and platelets count of 50000/ml

Ans D Severe life-threatening bleeding not responding to all modalities of medical treatment is an indication for emergent splenectomy.

155. Of the following, the LEAST likely feature of acute immune thrombocytopenia (ITP) is

A. a preceding viral infection
B. isolated thrombocytopenia
C. mucocutaneous bleeding
D. isolated splenomegaly
E. increased number of megakaryocytes in the bone marrow

Ans D Splenomegaly is present in only 10% of cases of acute ITP.

156. Inherited thrombocytopenias include all the following EXCEPT

A. congenital Amegakaryocytic Thrombocytopenia
B. thrombocytopenia with Absent Radii (TAR) Syndrome
C. Glanzmann's Thrombasthenia
D. Bernard-Soulier Syndrome
E. Wiskott–Aldrich Syndrome (WAS)

Ans C is characterized by an abnormal platelets function and normal platelets count.

157. Which of the following is the best treatment for a 5-year-old girl with acute immune thrombocytopenia (ITP) and platelets count of 30,000/ml and mild petechial rash all over her body?

A. intravenous immunoglobulin (IVIG)
B. anti-D gamma globulin
C. corticosteroids
D. platelets transfusion
E. no treatment

Ans E Spontaneous recovery occurs in the majority of children with ITP; treatment is required when the platelets count is less than 20,000/ml.

DIC & Coagulopathy

158. Beta globin missense gene mutation seen in:

A. Thalassemia
B. Sickle cell anemia
C. BART
D. HbH

Ans B Point mutation (missense) in 6th codon of β-globin gene that leads to replacement of glutamate by valine residue.

159. NESTROFT test is a screening test for:

A. β-thalassemia
B. Hereditary spherocytosis
C. Autoimmune hemolytic anemia
D. Megaloblastic anemia

Ans A NESTROFT: Naked Eye Single Tube Red cell Osmotic Fragility TestQ Assesses osmotic fragility of RBCs at a single concentration of buffered saline (0.36% in single tube).Q

160. Pan T cell marker is

A. CD3
B. CD8
C. CD19
D. CD20

Ans A CD3 is a marker present on all T cells and so is Pan T cell marker. Hematological cells Immunophenotyping markers (on Flow cytometry) Blasts CD34, Megakaryocytic marker CD41,Q CD61 WBC's CD 45Q (Leukocyte Common Antigen)Q Myeloid cells Anti- MPO,Q CD13, CD33, CD14, CD117 B –cells CD19,Q CD20, CD22, FMC7, CD23, CD79 a, CD79 b, S Ig, IgM T-cells CD3,Q CD2, CD5, CD7, CD8, TCR-α/β, TCR-g/d NK cells CD16,Q CD56,Q CD57 Plasma cells CD38,Q CD138,Q kappa and Lambda light chains

161. The following parameter in ALL indicates poor prognosis:

A. Age >10 years
B. WBC count <50000/mm3 at diagnosis
C. Hyperdiploidy
D. Early pre-B phenotype

Ans A

162. Poor prognostic indicator of ALL is:

A. Female sex
B. Leukocyte count < 50,000/µL
C. Age greater than 1 year
D. Hypoploidy MYELOID NEOPLASMS

Ans D Hypoploidy 115

163. The gold standard Lab test for diagnosis of PNH 198. (Paroxysmal Nocturnal Hemoglobinuria) is:

A. Ham test
B. Haptoglobin
C. Flow cytometry
D. Sucrose Lysis test

Ans C Flow cytometry of RBC showing absence of CD55 and CD59 is gold standard diagnosis of PNH. with

164. **Fresh frozen plasma (FFP) transfusion/replacement is indicated in all the following conditions EXCEPT**

A. a 9-year-old child with chronic liver disease with prolonged clotting times who needs a liver biopsy
B. a 6-year-old child with protein S deficiency and significant deep venous thrombosis
C. an 11-year-old child with thrombotic thrombocytopenic purpura (TTP) post allogenic bone marrow transplantation
D. **a 2-day-old preterm neonate with clotting deficiencies to prevent intraventricular hemorrhage**
E. a 1-week-old term neonate with hemorrhage secondary to vitamin K deficiency

Ans D In neonates, Clotting times are prolonged, owing to developmental deficiency of clotting proteins, and FFP should be transfused only after reference to normal values expected for the birthweight and age of the infant. The use of prophylactic FFP transfusions to prevent intraventricular hemorrhage in premature infants is not recommended.

165. **Arterial thrombosis is the least likely cause of**

A. stroke
B. a cold and pulseless lower extremity
C. renal infarction
D. myocardial infarction
E. **pulmonary embolism**

Ans E is a feature of venous thrombosis.

166. **You are explaining the risk of sepsis after splenectomy to medical students. An important statement that should be included during this discussion is that risk is especially high in children**

A. older than 5 yr at the time of surgery
B. with hereditary spherocytosis
C. with chronic immune thrombocytopenia (ITP)
D. after trauma
E. **with Hodgkin lymphoma**

Ans E This risk of sepsis is especially high in children younger than 5 yr at the time of surgery. The risk of sepsis is less after splenectomies performed for trauma, RBC membrane defects, and immune thrombocytopenia than when there is pre-existing immune deficiency (Wiskott-Aldrich syndrome, Hodgkin disease) or reticuloendothelial blockade (storage diseases, severe hemolytic anemias).

167. **Hematologic disease with upper limb orthopedic congenital abnormalities with presence of thumbs is MOST likely seen in**

A. Fanconi anemia
B. **thrombocytopenia with Absent Radii (TAR) Syndrome**
C. congenital Amegakaryocytic Thrombocytopenia
D. Glanzmann Thrombasthenia
E. Bernard-Soulier Syndrome

Ans B TAR is characterized by absent radii with presence of thumbs; this distinguishes from Fanconi anemia in which thumbs are absent.

Leukemia

168. **True about Hereditary Hemochromatosis are all except:**

A. Mutations in the HFE gene
B. Autosomal recessive disorder
C. Excess iron affects organ function
D. **Diagnosed by serum ferritin**

Ans D Hereditary hemochromatosis (Autosomal recessive) is caused due to mutations of HFE gene on chr 6p21.3 and diagnosed on the basis of tissue ferritin; Staining for Iron is done on buccal scraping smear.

169. **All of the following cause microcytic hypochromic anemia, except:**

A. Lead poisoning
B. Thalassemia
C. Iron deficiency
D. **Fanconi's anemia**

Ans D Fanconi's anemia presents as pancytopenia and normocytic normochromic anemia.

170. **A 5-year-old girl came with history of progressively increasing pallor since birth and hepatosplenomegaly. Which of the following is the most relevant test for achieving diagnosis?**

A. **Hb electrophoresis**
B. Peripheral smear examination
C. Osmotic fragility test
D. Bone marrow examination

Ans A • Progressively increasing pallor and hepatosplenomegaly suggest the diagnosis of thalassemia
• Hb electrophoresis is the diagnostic test of choice for thalassemia.

171. **A 6-month-old baby with severe pallor and hepatosplenomegaly. Similar history is present in a sibling. Investigation of choice:**

A. Bone marrow biopsy
B. **Hb electrophoresis**
C. Hb estimation
D. Platelet count

Ans B • Presence of pallor and hepatosplenomegaly at 6 months of age with a positive family history indicates towards thalassemia major, which can be diagnosed by Hb electrophoresis.

172. **All the following features precipitate sickling of HbS except:**

A. Hypoxia
B. Dehydration
C. Infections
D. **Alkalosis**

Ans D Acidosis rather than alkalosis precipitate sickling of HbS.

173. **All are features of hemolytic uremic syndrome, except:**

A. Hyperkalemia
B. Anemia
C. Renal microthrombi
D. **Neuro-psychiatric disturbances**

Ans D Neuro-psychiatric disturbances are not a feature of hemolytic uremic syndrome, but may be seen in TTP.

174. **A child died soon after birth. On examination there was hepatosplenomegaly and edema all over body. Most probable diagnosis in:**

A. Crossover mutation
B. Frame shift
C. Deletion
D. Non dysjunction
E. Point mutation

Ans A α-thalassemia This case history suggests NON IMMUNE HYDROPS which is seen in severe variety of alpha thalassemia (HbH or Hb Barts). ABO incompatibility usually leads to mild hemolysis, which is not severe enough to lead to hydrops fetalis.

175. **Pure red cell aplasia is associated with all except:**

A. ABO incompatibility after renal transplant
B. 5q- syndrome
C. Drugs
D. Large granular lymphocytic leukemia

Ans B

176. **All are true regarding Hypereosinophilic syndrome in children except:**

A. Eosinophilia of > 1500 cells/uL
B. Persistent eosinophilia for 6 months
C. Absence of any specific organ involvement
D. Corticosteroids are first line therapy 332 Section 3: Systemic Pediatrics LYMPHOID NEOPLASMS

Ans C • Hypereosinophilic syndrome is characterized by sustained overproduction of eosinophils;
• Diagnostic criteria include: eosinophilia >1,500 cells/L persisting for >6 months, signs and symptoms of organ involvement and absence of another diagnosis to explain the eosinophilia;
• Most serious & life-threatening complication is cardiac disease due to endomyocardial thrombosis & fibrosis.
• Therapy is initiated with corticosteroids; Imatinib mesylate, Hydroxyurea or Interferon may be beneficial.

177. **Good prognosis factors of ALL are all except:**

A. Hyperdiploidy
B. WBC count < 5000/µL
C. Age > 10 years
D. t(12;21) translocation

Ans C

178. **Which of the following statements is true regarding juvenile chronic myeloid leukemia?**

A. Philadelphia chromosome is negative
B. Thrombocytopaenia is uncommon
C. The prognosis is better than the adult form of chronic myeloid leukemia
D. Single agent chemotherapy with busulfan or hydroxyurea can achieve remission

Ans A

179. **Which one is the best prognostic factor for ALL?**

A. Hyperploidy
B. Orgnomegaly
C. TLC more than 50,000/uL
D. Response to treatment

Ans D Discussing options one by one: a. Hyperploidy Intermediate prognosis; can be considered good b. Organomegaly L. Node, liver, spleen enlargement, Testicular enlargement → poor prognosis c. TLC > 50,000/uL Poor prognosis; TLC <10,000/uL has good prognosis d. Response to treatment Early response to treatment is a good prognostic factor for ALL

180. **True about ALL:**

A. tdT positive
B. Gamma globulins
C. t(8,14)
D. Insidious onset

Ans A tdt is a marker of lymphoblasts and so seen in ALL.

181. **Drug that is not used in the treatment of ALL:**

A. Rituximab
B. Methotrexate
C. Vincristine
D. Daunorubicin

Ans A

182. **Most common type of ALL in children:**

A. Pre-B cell ALL
B. Mature B cell ALL
C. Pre-T cell ALL
D. Mature T cell ALL

Ans A

183. **In a 6-year-child with ALL relapse, most common site:**

A. Bone marrow
B. Liver
C. Testes
D. Lung

Ans A • Infants with ALL, and patients with t(4;11), have a higher risk of relapse despite intensive therapy
• Higher levels of MRD (>0.01%) present at end of induction suggest a poorer prognosis
• Relapse occurs in the bone marrow in 15-20% of patients with ALL and carries the most serious implications

184. **All of the following are good prognostic factors for childhood ALL except:**

A. Hyperdiploidy
B. Female sex
C. Pre B cell ALL
D. t(12:21) translocation

Ans C Pre B cell ALL

185. **ALL L morphology is a malignancy arising from which 3 cell lineage?**

A. Mature B-cell
B. Precursor B-cell
C. Immature T-cell
D. Mixed B-cell and T-cell

Ans A • ALL L3 is Burkitt Lymphoma with spill over, so it arises from mature B cell
• ALL L1 and L2 are Acute Leukemia, so it arises from immature B cell.

186. **All of the following are associated with good prognosis in childhood leukemia except:**

A. Hyperdiploidy
B. Female sex
C. Precursor B-cell ALL
D. t(12; 21) translocation

Ans C Precursor B-cell ALL, B-cell ALL and T-cell ALL has poor prognosis.

187. **A 5-year-old child presents with history of fever off- and-on for past 2 weeks and petechial spots all over the body and increasing pallor for past 1 month. Examination reveals splenomegaly of 2 cm below costal margin. The most likely diagnosis is:**

A. Acute leukemia
B. Idiopathic thrombocytopenic purpura
C. Hypersplenism
D. Aplastic anemia

Ans A Pallor (anemia), petechial spot (thrombocytopenia), fever (infection due to neutropenia) with splenomegaly suggest the diagnosis of acute leukemia. Splenomegaly eliminates aplastic anemia; presence of anemia and recurrent infections, go against ITP; Hypersplenism presents with massive splenomegaly.

188. **Treatment of ALL includes use of:**

A. Hydroxyurea
B. All transretinoic acid
C. Prednisolone
D. L-asparginase
E. Vincristine

Ans C d. L-asparginase, e. Vincristine Anticancer therapy with prednisolone, vincristine, daunorubic, L-asparginase, 6-mercaptopurine and methotrexate are used for induction and consolidation in ALL.

189. **In AML best prognosis is seen with:**

A. Acute megakaryocytic leukemia
B. Acute monocytic leukemia
C. Acute promyelocytic lukemia (M3)
E. Erythroleukemia

Ans C Acute Promyelocytic Leukemia (APML), characterized by a gene rearrangement involving retinoic acid receptor [t(15;17); PML-RARA], is very responsive to all-transretinoic acid (ATRA, tretinoin) combined with anthracyclines and cytarabine. The success of this therapy makes marrow transplantation in 1st remission unnecessary for patients with this disease. Arsenic trioxide is also an effective non-cytotoxic therapy for APML b. t(15:17)

190. **Most common translocation in acute promyelocytic leukemia:**

A. t(8:14)
B. t(15:17)
C. t(9:22)
D. t(8:11)
E. t(11:14)

Ans B Acute promyelocytic leukemia (APL, AML-M3) is characterized by the t(15;17) translocation involving the PML-RARA genes.

191. **Most common type of AML in Down's syndrome:**

A. M2
B. M3
C. M6
D. M7

Ans D M7 AML with megakaryocytic maturation is the most common Acute Leukemia in Down syndrome. dna 141. b. Philadelphia chromosome JMML is a childhood mixed MDS/MPD that includes childhood leukemias previously classified as CMML, juvenile CML, and infantile monosomy 7 syndrome. Remember: presence of bcr/abl fusion (Philadelphia chromosome) gene is not compatible with a diagnosis of juvenile myelomonocytic leukemia (JMML).

192. **Juvenile CML is characterized by all except:**

A. Increased HbF
B. Philadelphia chromosome
C. Immature granulocyte
D. Nucleated RBC

Ans B Juvenile myelomonocytic leukemia (juvenile CML) is Philadelphia-chromosome NEGATIVE, distinguishing it from adult-type chronic myeloid leukemia.

193. **A child with acute myeloid leukemia presents with hyper-leukocytosis. Treatment includes all of the following except:**

A. IV fluid
B. Allopurinol
C. Alkalinization
D. Immediately start induction chemotherapy

Ans D Hyperleukocytosis weiveR • Hyperleukocytosis refers to leukocyte count usually greater than 100,000/µL
• It is a medical emergency most commonly seen in with AML or CML in blast crisis
• There is sludging of blasts in microcirculation → obstruction of small vessels esp in brain & lungs
• Treatment includes: IV fluids (hydration), Alkalinisation, Allopurinol (to prevent tumor lysis syndrome) and Hydroxyurea (for leukoreduction)

194. **Non-specific esterase is positive in all the categories of AML except:**

A. M
B. M 3
C. M
D. M 5

Ans D M 6 FAB classification of AML is based on degree of maturation (M0 to M3) & lineage of leukemic blast (M4 to M7).
• Nonspecific esterase is positive in monoblasts (AML M and M) and Promyelocytes (AML M). 4 5

195. **AML with worst prognosis:**

A. 8/21 translocation
B. Inversion
C. Normal cytogenetics
D. **Monosomy**

Ans D 7 • Good prognostic factors in AML are t(8;21), inv16, t(15;17), CEBPa mutation, NPM mutation, Down syndrome
• Bad prognostic factors in AML are monosomy 5 or 7, 5q-, FLT3 gene duplication and abnormal 11q

196. **A 15-year-old boy presented with 1 day history of blee- Questions ding gums, subconjunctival bleed and purpuric rash. Invest ig ations revealed: Hb 6.4 gm/dL; TLC-26,500/ mm3; prothrombin time-20 sec with a control of 13 sec; partial thromboplastin time-50 sec; and fibrinogen 10 mg/dL. Peripheral smear was suggestive of acute myeloblastic leukemia. Which of the following is the most likely?**

A. Myeloblastic leukemia without maturation
B. Myeloblastic leukemia with maturation
C. **Promyelocytic leukemia**
D. Myelomonocytic leukemia

Ans C • This patient has complaints of bleeding along with thrombocytopenia, decreased fibrinogen & increased PT & APTT. This is consistent with DIC; DIC is most commonly seen with M AML

197. **Genetic risk factors for leukemia are all except:**

A. Down's syndrome
B. Bloom syndrome
C. Ataxia telangiectasia
D. **Turner's syndrome**

Ans D Genetic conditions predisposing to leukemia
• Down syndrome • Klinefelter syndrome • Bloom syndrome
• Diamond-Blackfan anemia • Neurofibromatosis type 1 • Schwachman syndrome
• Turner syndrome (least associated) • Ataxia - telangiectasia • Paroxysmal nocturnal hemoglobinuria
• Fanconi syndrome • Severe combined immune deficiency. • Li-Fraumani syndrome Note: All the given conditions are risk factors for leukemia. Out of them Turner syndrome is least associated with leukemia.

198. **A 2-year-old boy is suffering from leukemia; following X-ray findings are seen:**

A. **Osteolytic lesion in flat bones**
B. Metaphyseal osteoporosis
C. Periosteal new bone formation
D. Osteosclerosis of long bones
E. Transverse line of dark band below the growth plate

Ans A b. Metaphyseal osteoporosis, c. Periosteal new bone formation, e. Transverse line of dark band below the growth plate Features seen on X-ray in a child with Acute Leukemia:
• Reduced bone density • Collapsed vertebra
• Metaphyseal lucent band • Widening of sutures
• Metaphyseal cortical bone erosions • Periosteal reactions & lytic bone lesions

199. **X-ray features of leukemia in a 2-year-old child is/ are:**

A. **Osteolytic lesions in flat bones**
B. Subperiosteal erosions
C. Osteoporosis
D. Thick luscent line just below growth plate
E. Metaphyseal lucent bands

Ans A b. Subperiosteal erosions, c. Osteoporosis, d. Thick luscent line just below growth plate and e. Metaphyseal lucent bands

200. **All of the following are features of juvenile CML except:**

A. Thrombocytopenia
B. Fetal Hb is increased
C. **Philadelphia chromosome is positive**
D. Lymphadenopathy LANGERHANS CELL HISTIOCYTOSIS

Ans C 127 LANGERHANS CELL HISTIOCYTOSIS with

201. **About Langerhans cell histiocytosis, true are all except:**

A. Peak incidence less than 3 years of age
B. Radiosensitive
C. Diffuse form is known as Letterer-Siwe-disease
D. **Gonadal involvement occurs**

Ans D a. Peak incidence less than 3 years of age: True; More common and more severe in younger children b. Radiosensitive: True, as Bone lesions and Pituitary lesions, causing DI, respond well to radiotherapy c. Diffuse form is known as Letterer-Siwe disease: True, 154 above d. Gonadal involvement occurs: FALSE; Gonadal involvement is usually not seen in LCH.

202. **A child presented in the OPD with multiple permeating lesions involving all the bones of the body which of the following is the most probable diagnosis?**

A. Neuroblastoma
B. Metastasis from osteosarcoma
C. **Histiocytosis X**
D. Metastasis from Wilms' tumor BLEEDING DISORDERS

Ans C Osteolytic lesions in a child points towards the diagnosis of Histiocytosis X; Most common is skull.

203. **True about Kasabach-Merritt syndrome, are all except:**

A. Platelet sequestration
B. Infantile hemangioma
C. Consumption coagulopathy
D. **Portwine hemangioma**
E. Thrombocytosis

Ans D and e. Thrombocytosis Kasabach Meritt syndrome:
• Association of giant hemangioma with localized intravascular coagulation causing thrombocytopenia and hypofi brinogenemia is Kasabach-Merritt syndrome; Heart failure can be seen due to AV malformations
• Treatment: Drugs (corticosteroids, alpha interferon and vincristine), laser photocoagulation and radiation
• Surgery is usually not indicated due to risk of hemorrhage

204. **Cryoprecipitate contains all except:**

A. Factor VIII
B. **Factor IX**
C. Fibrinogen
D. Von- Willebrand factor 195.

Ans B Cryoprecipitate contains, Factor VIII, Fibrinogen, and Von-Willebrand factor.

205. **The hematologic features of congenital hypoplastic anemia (Diamond-Blackfan Anemia) include all the following EXCEPT**

A. normocytic anemia
B. reticulocytopenia
C. deficiency or absence of red blood cell bone marrow precursors
D. elevated fetal hemoglobin (Hb F)
E. elevated serum iron levels

Ans A The RBCs in Diamond-Blackfan anemia are usually macrocytic for age, but there is no hypersegmentation of neutrophils.

206. **All the following are true regarding Transient Erythroblastopenia of Childhood (TEC)**

A. it is more common than congenital hypoplastic (Diamond-Blackfan) anemia
B. most of the affected children are older than 12 mo at onset
C. Parvovirus B19 infections is a common causative agent
D. virtually all children recover within 1-2 months
E. corticosteroid therapy is of no value

Ans C In rare instances, a prolonged case of apparent TEC may be caused by parvovirus- induced RBC aplasia, occurring in children with hemolytic anemia or congenital or acquired immunodeficiencies.

207. **All the following are true regarding physiologic anemia of infancy EXCEPT**

A. it is exaggerated in premature infants
B. the hemoglobin concentration range is between 9-11 g/dL
C. the hemoglobin concentration rarely falls below 10 g/dL in healthy infants
D. vitamin E deficiency does not play a role
E. iron deficiency is a contributing factor

Ans E The iron previously stored from the degraded fetal RBCs can be used for hemoglobin synthesis. The supply of stored iron is sufficient for hemoglobin synthesis, even in the absence of dietary iron intake, until approximately 20 wk of age. Unless there has been significant blood loss, iron stores should be sufficient to maintain erythropoiesis early on.

208. **Splenectomy is recommended in all the following conditions EXCEPT**

A. a 6-year-old child with hereditary spherocytosis and significant hemolysis
B. a 7-year-old child with hereditary elliptocytosis and a hemoglobin level of 7 g/dL and corrected reticulocytes count of more than 15%
C. an 8-year-old child with hereditary stomatocytosis with hemolysis
D. a 6-year-old child with thalassemia major with splenomegaly and frequent blood transfusion requirement
E. a 12-year-old child with chronic immune thrombocytopenia not responding to all modalities of medical treatment

Ans C Hemolytic anemia may be associated with hereditary stomatocytosis, but splenectomy is not a recommended treatment. Persistent symptomatic thrombocytosis may follow splenectomy if the hemolysis is not eliminated or markedly decreased. Patients have developed a life-threatening tendency toward in situ thrombosis after splenectomy in association with the abnormal adherence of the stomatocytic RBC to vascular endothelium in conjunction with the thrombocytosis.

209. **The criteria for successful bone marrow transplantation in thalassemic patients include all the following EXCEPT**

A. age younger than 15-year-old
B. no hepatomegaly
C. no bone deformities
D. no iron overload
E. full HLA-matched sibling

Ans C

210. **Hereditary intrinsic factor deficiency (HIFD), formerly called congenital pernicious anemia, differs from the typical adult pernicious anemia by all the following EXCEPT**

A. it is prominent at around 3 mo of age
B. stomach secretes acid normally
C. stomach is histologically normal
D. there are no antibodies to parietal cells
E. there are no associated endocrine disorders

Ans A In HIFD, symptoms become prominent at an early age (6-24 mo), consistent with exhaustion of vitamin B12 stores acquired in utero.

211. **All the following are correct regarding treatment of cold agglutinin disease EXCEPT**

A. patient should avoid exposure to cold
B. patient should be treated for underlying disease
C. plasmapheresis is a modality of treatment
D. glucocorticoids treatment is a modality of treatment
E. patient can be treated by splenectomy when there is no response to other modalities of treatment

Ans E Splenectomy is not useful in cold agglutinin disease.

212. **A 9-year-old male child is about to do elective tonsillectomy; preoperative investigations are all normal except for a significant prolonged partial thromboplastin time PTT; there was no previous bleeding after circumcision and emergency appendectomy. Of the following, the MOST likely diagnosis is**

A. hemophilia A
B. hemophilia B
C. hemophilia C
D. prekallikrein deficiency
E. factor XIII deficiency

Ans D Deficiencies of the contact factors prolong the PTT but are not causes of clinical bleeding. These factors include factor XII, prekallikrein, and high molecular weight kininogen.

213. **All the following are true regarding factor XIII EXCEPT**

A. the half-life is 5-7 days
B. PT, PTT, and thrombin time (TT) are prolonged when the factor is deficient
C. clot solubility test with 5m urea is a screening test for factor XIII deficiency
D. factor assay is a specific test to detect its level
E. the hemostatic level is only 2-3%

Ans B The usual screening tests for hemostasis are normal in patients with factor XIII deficiency, antiplasmin deficiency, and plasminogen activator inhibitor deficiency.

214. **Vitamin K-dependent clotting factors include all the following EXCEPT**

A. **factor I**
B. factor II
C. factor VII
D. factor IX
E. protein S

Ans A Vitamin K-dependent clotting factors include factors II, VII, IX, X, protein C, and protein S.

215. **A 16-year-old female is found to have multiple bruising and hypochromic microcytic anemia; during her first pregnancy she noticed disappearance of the bruises; she has a past history of uncomplicated emergency appendectomy at the age of 10 year. Of the following, the drug MOST likely beneficial for alleviation of her symptoms is**

A. ferrous sulphate
B. **desmopressin acetate (DDAVP)**
C. recombinant factor VIII
D. recombinant factor IX
E. aminocaproic acid

Ans B Von Willebrand disease (VWD) is the most likely cause for this case; menstruating female may present with iron deficiency due to menorrhagia. Because VWF is an acute-phase protein, stress will increase its level; thus, patients may not bleed during appendectomy and childbirth. Bruising symptoms may diminish during pregnancy, because the VWF levels may double or triple during pregnancy.

216. **Lupus anticoagulants usually affect the following laboratory test**

A. bleeding time (BT)
B. **partial thromboplastin time (PTT)**
C. prothrombin time (PT)
D. thrombin time (TT)
E. clot solubility test

Ans B The lupus anticoagulant causes a prolongation of the PTT that fails to correct on 1:1 mixing with normal plasma. The antibody is directed against the phospholipid used as a reagent in the PTT.

217. **All the following are true regarding low molecular weight heparin (LMWH) in comparison with unfractionated heparin (UFH) EXCEPT**

A. it is more stable
B. it is ease to titrate
C. **it is monitored by activated PTT**
D. it can be used subcutaneously
E. it can be used as an outpatient

Ans C Unlike UFH, which is monitored using the aPTT, LMWH is monitored via the anti- Xa activity.

218. **A 3-year-old male child develops hematoma and bruising of his right hand next day after falling on the ground; the mother stated that her child has a poor wound healing and a history of delayed umbilical separation during the neonatal period. Of the following, the MOST valuable test for this case is**

A. bleeding time
B. partial thromboplastin time
C. prothrombin time
D. thrombin time
E. **clot solubility test**

Ans E The most likely diagnosis is factor XIII deficiency.

219. **Allogeneic bone marrow stem cell transplantation is least likely to be required in**

A. Fanconi anemia
B. **thrombocytopenia with Absent Radii (TAR) Syndrome**
C. congenital Amegakaryocytic Thrombocytopenia (CAT)
D. Wiskott –Aldrich Syndrome (WAS)
E. severe combined immune deficiency

Ans B In TAR, thrombocytopenia frequently remits but some cases may require allogenic bone marrow transplantation.

Lymphoma

220. **Autosplenectomy occurs in:**

A. Thalassemia
B. **Sickle cell anemia**
C. HbH disease
D. Hydrops fetalis

Ans B Patients of sickle cell anemia may undergo auto-splenectomyQ due to multiple infarcts in splenic artery, that causes spleen to be reduced to fibrous tissue (splenic atrophy → spleen may become non-palpable).

221. **A neonate has ABO incompatibility. Peripheral smear will show:**

A. **Microspherocytes**
B. Elliptocytes
C. Fragmented RBCs
D. Polychromophilia

Ans A ABO incompatibility causes immune hemolytic anemia which is characterized by microspherocytes on peripheral smear.

222. **Dohle bodies are seen in:**

A. **Neutrophils**
B. Macrophages
C. Plasma cells
D. Histiocytes

Ans A fo In sepsis or severe inflammatory disorders, morphologic changes can be seen in neutrophils, like cytoplasmic vacuoles, Toxic granules (coarser and darker than normal neutrophilic granules) and Döhle bodies, which are patches of dilated endoplasmic reticulum that appear as sky-blue cytoplasmic “puddles.”

223.True about Hodgkin's lymphoma is/are:

A. Often localized to single axial group of lymph nodes
B. Hepatomegaly is always present
C. Contiguous spread of lymph nodes
D. Can be cured by Chemotherapy and Radiotherapy
E. Commonly presents with painless lymphadenopathy

Ans A c. Contiguous spread of lymph nodes, d. Can be cured by Chemotherapy and Radiotherapy, e. Commonly presents with painless lymphadenopathy Harrison's 19/e p 708-709 HL appears to arise in lymphoid tissue and spread to adjacent lymph node areas in a relatively orderly fashion. Patients commonly present with painless, non-tender, firm, rubbery, cervical or supraclavicular lymphadenopathy and usually some degree of mediastinal involvement. Chemotherapy & radiation therapy are both effective in treatment of HL. with

224.A 2-year-old child comes with ear discharge, seborrheic dermatitis, polyuria and hepatosplenomegaly. Which of the following is the most likely diagnosis?

A. Leukemia
B. Lymphoma
C. Langerhan's cell histiocytosis
D. Germ cell tumor

Ans C Seborrheic dermatitis, polyuria, hepatosplenomegaly and ear discharge suggests diagnosis of LCH.

225.Characteristic cell in Hodgkin lymphoma is:

A. Reed Sternberg
B. Gaucher cell
C. Erythrophagocytes
D. Langhans cell

Ans A

226.A prolonged thrombin time with normal reptilase time is consistent with

A. dysfibrinogenemia
B. afibrinogenemia
C. factor XIII deficiency
D. disseminated intravascular coagulopathy (DIC) with high level of fibrin split products
E. heparin overdose

Ans E Unlike the thrombin time, the reptilase time is not sensitive to heparin and is prolonged only by reduced or dysfunctional fibrinogen and fibrin split products.

227.Which of the following is FALSE regarding standard unfractionated heparin therapy?

A. it acts by enhancing the antithrombin III activity
B. it can be used safely during pregnancy
C. it should not be given for a child with bacterial endocarditis
D. it can be given intramuscularly
E. it needs monitoring by PTT test

Ans D

Neutropenia

228.Most common mutation in hereditary elliptocytosis:

A. Spectrin
B. Ankyrin
C. Glycophorin A
D. Band 4.2

Ans A In Spectrin mutations, the most common RBC abnormality is Hereditary Elliptocytosis > spherocytosis.

229.A 10-year-old boy presents with mucosal bleeding of 1 week duration. The investigation of choice that will be most useful in him is:

A. Prothrombin time
B. Clotting time
C. Partial thromboplastin time
D. Platelet count

Ans D Mucosal bleeding can be a result of thrombocytopenia or any platelet abnormality. So platelet count needs to be done .

230.A 20 months old female child is brought for routine check-up. Complete blood count (CBC) shows moderate neutropenia. Child looks healthy & eats well. Family history is unremarkable. CBC after 1 and 2 weeks shows same results. Bone marrow examination is normal. Next step:

A. Corticosteroid administration
B. Multivitamin administration
C. Watch and wait strategy
D. Antibiotic to prevent infection

Ans C Cyclic neutropenia
• It is a rare autosomal dominant disorder where neutrophil elastase gene, ELANE is affected)
• Regular, periodic oscillations, with the ANC ranging from normal to <200/µL, mirrored by reciprocal cycling of monocytes seen.
• Mean oscillatory period of the cycle is 21 days (± 4 days).
• Affected patients with severe neutropenia may be treated with filgrastim (G-CSF)

231.Auer rods are specific for:

A. Acute myeloid leukemia
B. Acute lymphocytic leukemia
C. Chronic lymphocytic leukemia
D. Hodgkin's lymphoma

Ans A • Auer rods are Needle-like azurophilic fusiform inclusions in cytoplasm of myeloblasts
• Auer rods are most reliable morphological feature of AML; Myeloblasts Stain +ve with MPO and Sudan Black B

232.A one-year-old boy presented with hepatosplenomegaly and delayed milestones. The liver biopsy and bone marrow biopsy revealed presence of histiocytes with PAS posit ive cytoplasm. Electron microscopic examination of these histiocytes is most likely to reveal the presence of:

A. Birbeck granules in the cytoplasm (AIIMS 03)
B. Myelin figures in the cytoplasm
C. Parallel rays of tubular structures in lysosomes
D. Electron dense deposit in the mitochondria

Ans C Given scenario suggests Gaucher disease, in which glucocerebroside in lysosomes seen.

233.vWF protects:

A. Factor V
B. Factor VI
C. Factor VII
D. **Factor VIII 200.**

Ans D Factor VIII 2 major functions of vWF are: Platelet adhesion via Gp Ib/IX and stabilization of factor VIII in circulation.

234.Spontaneous intracranial hemorrhage is more likely to occur in

A. **factor VII deficiency**
B. factor VIII deficiency
C. factor IX deficiency
D. factor XI deficiency
E. factor XIII deficiency

Ans A

235.Microthrombocytes are MOST likely to be seen in

A. immune thrombocytopenia (ITP)
B. Bernard-Soulier syndrome
C. **Wiskott-Aldrich syndrome (WAS)**
D. Glanzmann's Thrombasthenia
E. type 2B von Willebrand disease

Ans C WAS is characterized by thrombocytopenia with tiny platelets, eczema, and recurrent infections due to immune deficiency.

Bone Marrow & Stem Cell Transplant

236.Kleihaeur Bethke test is done for:

A. Cephalopelvic disproportion
B. **Fetomaternal hemorrhage**
C. Determining karyotype of normal fetus
D. Diagnosing fetal infections

Ans B A common approach to evaluating Fetomaternal Hemorrhage (FMH) is the rosette test to screen for the presence of fetal cells followed by the Kleihauer-Betke acid elution method to quantify the magnitude of feto-maternal bleed. with

237.Bart's hydrops fetalis is lethal because:

A. Hb Bart's cannot bind oxygen
B. The excess α-globin form insoluble precipitates
C. **Hb Bart's cannot release oxygen to fetal tissues**
D. Microcytic red cells become trapped in the placental

Ans C As Hb Bart's consists of 4y chains, which have very high oxygen affinity, it cannot release oxygen to fetal tissues.

238.A 6-year-old boy with lymphoreticular malignancy has come for a cycle of chemotherapy. Which of the following investigations need to be done for next 4 hours to diagnose tumour lysis syndrome?

A. Urea, creatinine, chloride, Ca2+ and K+
B. **Urea, creatinine, phosphate, Ca2+ and K+**
C. Urea, creatinine, magnesium, Ca2+ and K+
D. Urea, creatinine, sodium and phosphate

Ans B Tumor lysis syndrome (TLS)
• Uric acid, phosphates & potassium are released in circulation in large quantities from death of tumor cells.
• Hyperuricemia can lead to impairment of renal function, which further exacerbates the metabolic abnormalities.
• Hypocalcemia is a consequence of acute hyperphosphatemia with subsequent precipitation of calcium phosphate in soft tissues.
• TLS can occur before therapy in patients with a large tumor burden (e.g., Burkitt lymphoma, Hyperleukocytosis)

239.Increased fetal hemoglobin is seen in:

A. **Juvenile CML**
B. Congenital red cell aplasia
C. Hereditary spherocytosis
D. AML

Ans A Increased hemoglobin F for age is a feature of JMML or Juvenile CML.

240.True about Langerhan's histocytosis 'X':

A. **Can be associated with diabetes insipidus**
B. X-ray shows pathognomonic osteosclerotic lesions
C. Birbeck's granules in Langerhan's cell
D. Proliferation of antigen presenting cells
E. Associated with HLA DR

Ans A c. Birbeck's granules in langerhan's cell, d. Proliferation of antigen presenting cells, e. Associated with specific

241.Factor Xa is necessary for conversion of prothrombin to thrombin in which pathway?

A. Factor X
B. Factor XI (AIIMS May 06)
C. **Factor XII**
D. Factor XIII

Ans C As part of both extrinsic and intrinsic pathways /e p Factor Xa is necessary for conversion of prothrombin to thrombin in the common pathway, that means as as part of both extrinsic and intrinsic pathways.

242.Systemic amyloidosis may be associated with deficiency of

A. factor VII
B. factor VIII
C. factor IX
D. **factor X**
E. factor XI

Ans D Systemic amyloidosis may be associated with factor X deficiency owing to the adsorption of factor X on the amyloid protein. In the setting of amyloidosis, transfusion therapy is often not successful because of the rapid clearance of factor X.

243.A 9-month-old boy appears pale. Examination reveals a palpable liver 3 cm below the right costal margin and palpable spleen 4 cm below the left costal margin. Lab findings include: hemoglobin concentration, 6.4 g/dl; mean corpuscular volume (MCV), 67 fL; and WBC, 15,000/mm3. Of the following, The MOST valuable test to confirm the diagnosis is

A. blood smear
B. serum iron, total iron binding capacity, and serum ferritin
C. **hemoglobin electrophoresis**
D. bone marrow study
E. abdominal ultrasonography

Ans C ß-thalassemia major is the most likely diagnosis; hemoglobin electrophoresis is quite helpful to confirm the diagnosis. Blood smear usually shows hypochromic microcytic RBCs but it is difficult to differentiate between iron deficiency anemia and thalassemia.

Blood Transfusion

244.Bart hemoglobin is tetramer of:

A. α chain
B. β chain
C. **γ chain**
D. δ chain

Ans C

245.A child aged 2 years presents with non-specific symptoms suggestive of anemia. On peripheral blood smear, target cells are seen. He has hypochromic microcytic picture and Hb of 6 g%. He also has a positive family history. Next investigation of choice is:

A. **Hb electrophoresis**
B. Coomb's test
C. Liver function tests
D. Osmatic fragility test

Ans A • A low Hb of 6 g%, microcytic and hypochromic RBCs and target cell points towards thalassemia major
• Thalassemia major is diagnosed by elevated HbF in Hb electrophoresis.

246.MC malignancy in children is:

A. Retinoblastoma
B. **Leukemia**
C. Wilms tumor
D. Neuroblastoma

Ans B Most frequent childhood cancers are: (in order of decreasing frequency)
• Leukemia (Most common is ALL)
• Brain Tumors
• Neuroblastoma
• Wilms tumor

247.Arsenic is used in the management of:

A. **Acute promyelocytic leukemia**
B. ALL
C. CML
D. Transient myeloproliferative disorder

Ans A • Arsenic trioxide can be used for treatment of resistant cases of Acute promyelocytic leukemia (AML M3).

248.A 5 years old boy comes with appearance of petechial spots overnight. 2 weeks back he had history of abdo- minal pain and no hepatosplenomegaly. Diagnosis is:

A. Acute lymphatic leukemia
B. Aplastic anemia
C. **Idiopathic thrombocytopenic purpura**
D. Acute viral infection

Ans C

249.The MOST specific test that is helpful to differentiate Diamond-Blackfan anemia from transient erythroblastopenia of childhood is

A. hemoglobin electrophoresis
B. reticulocytes count
C. **erythrocyte adenosine deaminase (ADA) enzyme level assay**
D. bone marrow examination
E. mean corpuscular volume (MCV)

Ans C Erythrocyte adenosine deaminase (ADA) activity is increased in most patients with congenital hypoplastic anemia, a finding that helps distinguish this disorder from acquired transient erythroblastopenia of childhood. Also, because elevated ADA activity is not a fetal RBC feature, measurement of this enzyme is helpful in diagnosing Diamond-Blackfan anemia in very young infants. Hemoglobin F may be increased in 20% of cases with transient erythroblastopenia of childhood at the time of diagnosis.

250.Fresh frozen plasma (FFP) transfusion is the best treatment for which of the following conditions

A. hemophilia a
B. hemophilia b
C. factor vii deficiency
D. immunoglobulin replacement therapy
E. **bleeding due to warfarin therapy**

Ans E Transfusion of FFP is no longer recommended for treatment of patients with severe hemophilia A or B or for factor VII deficiency because safer factor VIII, IX, and VII concentrates are available; also it is not indicated for correction of hypovolemia or as immunoglobulin replacement therapy, because safer alternatives exist;

251.CMV-transmitted infection is least likely occurring in transfusion of which of the following blood products?

A. whole blood
B. packed RBCs
C. granulocytes
D. platelets
E. **fresh frozen plasma**

Ans E CMV infections may be transmitted by leukocyte containing blood component transfusions; FFP does not transmit such infection because it is acellular component.

15

CHAPTER 15

Tumors of Infancy and Childhood



WHAT THIS CHAPTER COVERS

- Retinoblastoma
- Neuroblastoma
- Wilms Tumor
- Hepatoblastoma
- Rhabdomyosarcoma
- Osteosarcoma & Ewing Sarcoma
- Brain Tumors
- Leukemia & Lymphoma
- Langerhans Cell Histiocytosis
- Germ Cell Tumors
- Hodgkin & Non-Hodgkin Lymphoma
- Soft Tissue Sarcomas
- Tumor Markers & Staging
- Oncology Emergencies

152 MCQs **14** subtopics

15.1 Childhood Cancer – Overview

- **Most common childhood cancer: leukaemia (ALL)** (~30%) > **CNS tumours** (most common **solid** tumour) > lymphomas > neuroblastoma > Wilms > soft-tissue sarcomas > bone tumours > retinoblastoma.
- **Most common solid extracranial tumour: neuroblastoma**; **most common malignancy in infancy (< 1 y): neuroblastoma**; **most common abdominal malignancy (overall children): neuroblastoma (< 2 y) / Wilms (2–5 y)**.
- **Most common tumour of newborn: sacrococcygeal teratoma**; **MC benign tumour of infancy: infantile haemangioma**.
- India: ~50,000 new childhood cancers/year; overall cure rates in high-income countries > 80%.
- **Warning signs (St Siluan / SIOP list)**: persistent unexplained fever, pallor, bone pain/limping, **leukocoria**, abdominal mass, **headache with early morning vomiting**, unexplained lymphadenopathy, bruising/bleeding, weight loss.

High-yield "most commons"

Category	Most common
Brain tumour (overall)	**Pilocytic astrocytoma (low-grade glioma)**
Malignant brain tumour	**Medulloblastoma**
Brain tumour < 1 year	Supratentorial (teratoma, choroid plexus papilloma, ATRT)
Intraocular malignancy	**Retinoblastoma**
Orbital malignancy	**Rhabdomyosarcoma**
Soft-tissue sarcoma	**Rhabdomyosarcoma (embryonal)**
Bone tumour (malignant)	**Osteosarcoma** (> 10 y); Ewing (< 10 y – common in younger)
Liver malignancy	**Hepatoblastoma** (< 3 y); HCC (adolescents, HBV, tyrosinaemia)
Benign liver tumour	**Infantile hepatic haemangioma (haemangioendothelioma)**
Testicular tumour (prepubertal)	**Yolk-sac tumour (endodermal sinus) – ↑ AFP**; teratoma
Ovarian tumour	**Germ-cell (mature cystic teratoma/dermoid)**
Bladder/prostate tumour	**Rhabdomyosarcoma (botryoid)**
Cardiac tumour	**Rhabdomyoma** (tuberous sclerosis)
Lung tumour (benign)	**Hamartoma**
Cause of osteolytic bone metastases	**Neuroblastoma**
Thyroid cancer	Papillary
Mediastinal mass – anterior	**Lymphoma (T-ALL/NHL), teratoma, thymoma** ("4 Ts")
Mediastinal mass – posterior	**Neuroblastoma (neurogenic tumours)**

15.2 Leukaemia

Acute lymphoblastic leukaemia (ALL)

- **75–80% of childhood leukaemia**; peak **2–5 years**; **precursor B-ALL (85%)**; T-ALL (adolescent boys, **mediastinal mass**, high WBC).
- Risk factors: **Down syndrome (10–20×)**, Fanconi, Bloom, ataxia telangiectasia, NF-1, ionising radiation.

- Features: **pallor, fever, bleeding (marrow failure)**, **bone pain/limp (MC presentation in some – periosteal/marrow infiltration; night pain)**, hepatosplenomegaly, lymphadenopathy; **testicular enlargement**, **CNS** (headache, cranial nerve palsy – VII), **SVC syndrome (T-ALL)**.
- Labs: anaemia, thrombocytopenia, WBC low/normal/high, **blasts on smear**; **bone marrow ≥ 20% (WHO) / ≥ 25% lymphoblasts**; **flow cytometry – IOC for lineage (CD10/CALLA, CD19, CD22 – B; CD3, CD7 – T)**, **TdT positive**, PAS block positivity; cytogenetics; **CSF cytology**.
- **Tumour lysis syndrome** (hyperuricaemia, **hyperkalaemia, hyperphosphataemia → hypocalcaemia**, AKI) – hyperhydration, **allopurinol/rasburicase**.

Good prognosis	Poor prognosis
Age 1–10 years	**< 1 year (infant – KMT2A/MLL rearrangement t(4;11)) or > 10 years**
WBC < 50,000	**WBC ≥ 50,000**
Hyperdiploidy (> 50 chromosomes), DNA index > 1.16, trisomies 4, 10	**Hypodiploidy (< 44)**
t(12;21) ETV6-RUNX1 (TEL-AML1) – MC & best	**t(9;22) Philadelphia (BCR-ABL)** (now improved with **imatinib/dasatinib**), Ph-like, t(4;11), iAMP21, TCF3-HLF
Early B-precursor, CD10+	**T-cell (historically)**, mature B (Burkitt-type)
Rapid early response – MRD negative at end of induction (most important prognostic factor)	MRD positive, CNS/testicular disease at diagnosis
Female	Male, black/Hispanic

- Treatment (~2–3 years): **induction (vincristine, steroid, L-asparaginase ± anthracycline) → consolidation/CNS-directed therapy (intrathecal methotrexate, high-dose MTX) → interim maintenance → delayed intensification → maintenance (6-MP + MTX)**; **~90% cure** in high-income settings. **Blinatumomab, inotuzumab, CAR-T (tisagenlecleucel)** for relapse.
- **MC site of relapse: bone marrow**; extramedullary – **CNS, testes** ("sanctuary sites").
- Drug toxicities: **vincristine – neuropathy, constipation, SIADH, jaw pain**; **L-asparaginase – pancreatitis, thrombosis, hypersensitivity, hyperglycaemia**; **anthracyclines – cardiotoxicity (cumulative)**; **6-MP – TPMT/NUDT15 deficiency → severe myelosuppression**; methotrexate – mucositis, leucoencephalopathy; **cyclophosphamide – haemorrhagic cystitis (mesna)**.

Acute myeloid leukaemia (AML)

- 15–20% of childhood leukaemia; **chloroma (granulocytic sarcoma – orbit, skin)**, **gum hypertrophy (M4/M5 – monocytic)**, **DIC in APL (M3, t(15;17), PML-RARA – Rx ATRA + arsenic)**; **Auer rods**, MPO positive.
- **Down syndrome**: **transient abnormal myelopoiesis (TAM) in newborn (GATA1 mutation, resolves spontaneously)** → 20–30% develop **AML-M7 (megakaryoblastic) by 4 y – excellent prognosis**.
- Good prognosis: **t(8;21), inv(16), t(15;17)**, NPM1, CEBPA; poor: monosomy 7, FLT3-ITD.

Chronic & other

- **CML (Ph+)**: rare in children; Rx imatinib.
- **Juvenile myelomonocytic leukaemia (JMML)**: < 4 y, **NF-1, Noonan (PTPN11)**, **↑ HbF, monocytosis, hepatosplenomegaly, skin rash**; Rx HSCT.
- **Leukaemoid reaction vs leukaemia**: high LAP score in leukaemoid reaction, no blasts beyond myelocytes.

15.3 Lymphomas

Feature	Hodgkin lymphoma (HL)	Non-Hodgkin lymphoma (NHL)
Age	**Bimodal – adolescents** (15–30) & > 50; < 5 y rare (India – younger, mixed cellularity, EBV)	**Younger children (< 15 y)**, boys

Feature	Hodgkin lymphoma (HL)	Non-Hodgkin lymphoma (NHL)
Cell	**Reed–Sternberg cell (owl-eye, CD15+, CD30+)**	Burkitt (MC in children), DLBCL, lymphoblastic, ALCL
Spread	**Contiguous lymph-node spread**	**Non-contiguous, extranodal** (abdomen, mediastinum, Waldeyer ring)
Presentation	**Painless cervical/supraclavicular lymphadenopathy (rubbery)**, mediastinal mass, **B symptoms** (fever > 38 °C, night sweats, weight loss > 10% in 6 m), **pruritus, alcohol-induced pain**	Rapidly growing mass, abdominal (ileocaecal – intussusception), mediastinal (T-lymphoblastic – SVC syndrome, airway compression)
Subtypes	**Nodular sclerosis (MC overall/adolescents)**; **mixed cellularity (MC in India/young children, EBV)**; lymphocyte-rich (best); lymphocyte-depleted (worst); NLPHL (popcorn cells, CD20+)	**Burkitt (t(8;14), c-MYC – "starry-sky", fastest-growing human tumour; endemic African – jaw, EBV; sporadic – abdomen)**, lymphoblastic (T-cell, mediastinum), DLBCL, ALCL (ALK+, t(2;5))
Staging	**Ann Arbor/Lugano** + PET-CT	**St Jude/Murphy**
Treatment	**ABVD**/OEPA-COPDAC, response-adapted RT; **> 90% cure**	Intensive multi-agent chemo (Burkitt – short intense; rituximab)

15.4 Neuroblastoma

- **Neural crest** origin (sympathetic chain); **MC extracranial solid tumour of childhood**, **MC malignancy in infants**; median age **~18 months–2 y**; **adrenal medulla (MC site, 40%)** > paraspinal ganglia (abdomen, chest, pelvis, neck).
- **Presentation**: **abdominal mass – firm, irregular, nodular, crosses midline** (vs Wilms), **calcification on X-ray (85–90%)**; **metastases at diagnosis in ~50%** – **bone (skull, orbit – "raccoon eyes"/periorbital ecchymosis & proptosis)**, **bone marrow**, liver (**Pepper syndrome – massive hepatomegaly in stage 4S infant**), skin (**"blueberry muffin" nodules**), lymph nodes.
- Paraneoplastic/other: **opsoclonus–myoclonus–ataxia ("dancing eyes, dancing feet") – associated with good prognosis tumour**; **VIP secretion → watery diarrhoea (Kerner–Morrison)**, **Horner syndrome (cervical/thoracic)**, **spinal cord compression (dumbbell tumour)**, hypertension, heterochromia iridis.
- **Diagnosis**: **urinary catecholamine metabolites – ↑ VMA (vanillylmandelic acid) & HVA (homovanillic acid) in > 90%**; **MIBG (metaiodobenzylguanidine) scan – staging/metastases**; CT/MRI (calcification); biopsy – **small round blue cells, Homer-Wright pseudorosettes**, neuropil; bone marrow aspirate/biopsy; **N-MYC amplification**.

Favourable	Unfavourable
Age < 18 months (esp. < 1 y)	Age > 18 months
Stage L1, L2, **MS (4S: < 18 m with metastases limited to skin, liver, marrow < 10%) – spontaneous regression**	**Stage M (metastatic), age > 18 m**
No N-MYC amplification	**N-MYC amplification (most important adverse genetic factor)**
Hyperdiploidy (DNA index > 1)	Diploid, **1p deletion, 11q aberration**, ALK mutation
Favourable histology (Shimada), **TrkA expression**	Unfavourable histology, high MKI

- Treatment: low-risk – surgery/observation; high-risk – **induction chemo → surgery → myeloablative chemo with autologous SCT → radiotherapy → anti-GD2 antibody (dinutuximab) + isotretinoin (13-cis-retinoic acid, differentiation)**; 131I-MIBG therapy.

15.5 Wilms Tumour (Nephroblastoma)

- **MC renal malignancy in children**; age **2–5 years (peak 3–4 y)**; **bilateral in 5–7%** (younger, syndromic).

- **Associations**:
  - **WAGR syndrome** (WT1 deletion, 11p13): **W**ilms, **A**niridia, **G**enitourinary anomalies, **R**etardation (ID).
  - **Denys–Drash** (WT1 point mutation): Wilms + **early-onset nephrotic syndrome (diffuse mesangial sclerosis)** + **male pseudohermaphroditism/gonadal dysgenesis**.
  - **Beckwith–Wiedemann (WT2, 11p15)**: macroglossia, hemihypertrophy, organomegaly, omphalocele → screen with **3-monthly abdominal USG till 8 years** (+ AFP for hepatoblastoma till 4 y).
  - **Isolated hemihypertrophy**, Perlman, Frasier (WT1 – late nephrotic syndrome + gonadal dysgenesis), Sotos, trisomy 18, Bloom.
- **Presentation**: **asymptomatic smooth, firm flank mass noticed by parents while bathing (MC)**, **does NOT cross midline (usually)**, abdominal pain, **haematuria (25%)**, **hypertension (25% – renin)**, fever; **varicocele (left – renal vein thrombus)**; rarely acquired vWD.
- **Imaging**: **USG → CECT/MRI abdomen** – intrarenal mass with **claw sign**, **calcification uncommon (10%, curvilinear)**, **tumour thrombus in renal vein/IVC**; **CT chest** (lung – **MC site of metastasis**), liver.
- **Pathology**: **triphasic – blastemal, epithelial (abortive tubules/glomeruli), stromal**; **anaplasia = unfavourable histology** (TP53).
- Do **not biopsy** routinely (NWTS/COG) – risk of upstaging; SIOP – preoperative chemo without biopsy.

Wilms	Neuroblastoma
2–5 y	< 2 y
Smooth, flank mass; **rarely crosses midline**	Irregular, nodular; **crosses midline**
Intrarenal (claw sign), displaces collecting system	Extrarenal – displaces kidney **downward & laterally ("drooping lily")**, encases vessels
Calcification rare (curvilinear)	**Calcification common (stippled/amorphous)**
Child well; haematuria, hypertension	Child ill – fever, bone pain, raccoon eyes
Urinary VMA normal	**↑ VMA/HVA**
Lung metastasis	**Bone, marrow, liver, skin** metastasis
Excellent prognosis (~90% cure)	Variable – poor in high-risk

- **Treatment**: **COG (North America) – primary nephrectomy (radical, transabdominal; inspect contralateral kidney, sample lymph nodes) → chemo**; **SIOP (Europe/India) – preoperative chemo (vincristine + actinomycin D) → nephrectomy → chemo ± RT**. **Most chemosensitive: actinomycin D + vincristine (± doxorubicin)**. Overall survival **> 90%** (stage I favourable 98%).

15.6 Retinoblastoma

- **MC primary intraocular malignancy of childhood**; **RB1 tumour-suppressor gene (chr 13q14)** – **Knudson's "two-hit" hypothesis**.
- **Heritable (40%)** – germline mutation (AD with **90% penetrance**), **bilateral/multifocal**, earlier (< 1 y), **risk of second cancers – osteosarcoma (MC; esp. after radiotherapy)**, soft-tissue sarcomas, melanoma; **trilateral retinoblastoma (pineoblastoma)**. **Non-heritable (60%)** – unilateral, unifocal, later (2 y).
- **Presentation**: **leukocoria (white pupillary reflex – MC, "cat's-eye reflex")** > **strabismus (2nd MC)** > red painful eye (glaucoma), poor vision, proptosis (orbital spread), **pseudohypopyon**, heterochromia.
- **Diagnosis**: **fundus examination under anaesthesia (EUA)**; **USG B-scan & CT – intraocular calcification (most characteristic)**; **MRI – optic nerve/CNS extension (preferred, avoids radiation)**; **biopsy contraindicated (seeding)**. Histology – **Flexner–Wintersteiner rosettes (specific)**, Homer-Wright rosettes, fleurettes.
- **Spread**: **optic nerve → CNS (MC route of death)**; haematogenous (bone, marrow).
- **Treatment**: by stage (ICRB A–E) – focal (**laser photocoagulation, cryotherapy, thermotherapy**), **chemoreduction (vincristine, etoposide, carboplatin)**, **intra-arterial chemotherapy (ophthalmic artery – melphalan)**, intravitreal chemo, plaque brachytherapy, EBRT (avoid in heritable), **enucleation (with long optic nerve stump ≥ 10 mm) for group E/advanced**.
- **Screening of siblings & offspring**: genetic testing; fundus exams from birth.

DIFFERENTIAL DIAGNOSIS OF LEUKOCORIA

Retinoblastoma (must exclude first), congenital cataract, **retinopathy of prematurity (stage 5)**, **Coats disease (exudative retinal telangiectasia – boys, unilateral, no calcification)**, **persistent hyperplastic primary vitreous (PFV – microphthalmic eye)**, **toxocariasis**, coloboma, retinal detachment, Norrie disease.

15.7 Other Solid Tumours

Tumour	Key points
Hepatoblastoma	**< 3 years**, **↑↑ AFP** (> 90%), associated with **Beckwith–Wiedemann, FAP (APC), extreme prematurity/VLBW**, trisomy 18; **asymptomatic RUQ mass**, osteopenia, **precocious puberty (β-hCG)**; Rx **cisplatin chemo + resection/liver transplant** (PRETEXT staging)
Hepatocellular carcinoma	Older children; HBV, tyrosinaemia, GSD I, biliary atresia; **fibrolamellar variant** (adolescents, normal AFP)
Rhabdomyosarcoma	**MC soft-tissue sarcoma**; **embryonal (MC, < 10 y; head & neck – orbit, parameningeal; GU – bladder/vagina – botryoid "bunch of grapes" sarcoma)** – better prognosis; **alveolar (adolescents, extremities, trunk; t(2;13) PAX3-FOXO1) – worse**; **desmin, myogenin, MyoD1** positive; associations – **Li-Fraumeni (p53), NF-1, Beckwith–Wiedemann, Costello, Noonan**; Rx VAC chemo + surgery/RT
Osteosarcoma	**MC malignant bone tumour**; **adolescents during growth spurt**; **metaphysis of long bones – distal femur > proximal tibia > proximal humerus ("near the knee, far from the elbow")**; **sunburst appearance, Codman's triangle**, osteoid; ↑ ALP; associated with **hereditary retinoblastoma, Li-Fraumeni, Paget's, radiation**; **lung metastases (MC)**; Rx **neoadjuvant chemo (MAP – methotrexate, doxorubicin, cisplatin) → limb-salvage surgery → adjuvant**
Ewing sarcoma	**5–15 y (younger than osteosarcoma)**; **diaphysis of long bones & flat bones (pelvis)**; **t(11;22) EWS-FLI1**, **CD99 (MIC2) +**, small round blue cells, PAS + (glycogen); **onion-peel periosteal reaction**, moth-eaten lytic lesion; **fever, ↑ ESR, leucocytosis – mimics osteomyelitis**; **radiosensitive**; Rx chemo + surgery/RT
Langerhans cell histiocytosis (LCH)	Clonal dendritic-cell proliferation (**BRAF V600E**); **CD1a, S-100, CD207 (langerin) +**; **Birbeck granules (tennis-racket) on EM**; **skull (MC bone) – "punched-out" lytic lesion ("hole within hole", bevelled edge)**, **vertebra plana**, **floating teeth**; **Hand–Schüller–Christian triad: skull lesions + exophthalmos + diabetes insipidus (MC endocrine)**; Letterer–Siwe – infants, multisystem (skin seborrhoea-like rash, hepatosplenomegaly, marrow); risk organs – liver, spleen, marrow; Rx single bone – curettage/observe; multisystem – **vinblastine + prednisolone**
HLH (haemophagocytic lymphohistiocytosis)	Primary (perforin – PRF1, familial) / secondary (EBV, malignancy, MAS in sJIA); **fever, splenomegaly, cytopenias, ↑↑ ferritin, ↑ TG, ↓ fibrinogen, haemophagocytosis**, ↓ NK activity, ↑ sCD25; Rx **HLH-94/2004 (dexamethasone + etoposide ± ciclosporin)**, HSCT
Germ-cell tumours	**Sacrococcygeal teratoma – MC neonatal tumour, girls (4:1)**, Altman types I–IV (type I external – best); malignancy risk ↑ if excision delayed > 2 months – **resect with coccyx early**; follow AFP; **yolk-sac – AFP, Schiller–Duval bodies**; choriocarcinoma – β-hCG
Infantile haemangioma	**MC tumour of infancy**; girls, preterm; **appears in first weeks, proliferates till 6–9 m, involutes (50% by 5 y, 90% by 9 y)**; **GLUT-1 positive**; complications – ulceration, airway ("beard" distribution), amblyopia (periorbital), **PHACE syndrome** (large facial), **LUMBAR**; Rx **oral propranolol 1–3 mg/kg/day – DOC**, topical timolol
Brain tumours	See Neurology (15.8)

15.8 Brain Tumours in Children

- **2nd MC childhood cancer & MC solid tumour**; **infratentorial (posterior fossa) predominance in 1–10 y**; supratentorial in infants & adolescents.
- **Features**: **raised ICP – morning headache with vomiting**, papilloedema, **head tilt, ataxia (cerebellar), cranial nerve palsies (VI)**, macrocephaly in infants, **"setting-sun" sign**, endocrine (craniopharyngioma), diencephalic syndrome (emaciation with euphoria – hypothalamic glioma), Parinaud syndrome (pineal).

Tumour	Location & features
Pilocytic astrocytoma	**MC brain tumour of children**; **cerebellar hemisphere (MC)**; **cystic with mural nodule**, **Rosenthal fibres**, BRAF fusion; **excellent prognosis after resection**; optic pathway glioma – **NF-1**
Medulloblastoma	**MC malignant brain tumour**; **cerebellar vermis/roof of 4th ventricle**, boys 5–9 y; **truncal ataxia**, hydrocephalus; **drop metastases via CSF (spinal MRI + CSF cytology)**; **Homer-Wright rosettes**; molecular groups – **WNT (best)**, SHH, group 3 (worst, MYC), group 4; Rx surgery + **craniospinal RT (> 3 y)** + chemo
Ependymoma	**Floor of 4th ventricle** (children); hydrocephalus, cranial nerve palsies; **perivascular pseudorosettes**; extends through foramina (**"toothpaste" tumour**)
Diffuse midline glioma, H3 K27M-altered (DIPG – brainstem)	**Pons**; **cranial nerve palsies (VI, VII) + long-tract signs + ataxia**; diagnosis by MRI (no biopsy needed classically); **worst prognosis – median survival < 1 y**; RT palliative
Craniopharyngioma	**MC supratentorial tumour in children**; suprasellar; **Rathke's pouch** remnant; **bitemporal hemianopia, growth failure (GH deficiency), diabetes insipidus, hypothyroidism, obesity**; **calcified cystic mass ("motor-oil" fluid, cholesterol crystals)** on CT; adamantinomatous type (CTNNB1)
Germinoma (pineal/ suprasellar)	Parinaud syndrome, **DI**, precocious puberty (β-hCG); **radiosensitive**
ATRT (atypical teratoid/ rhabdoid tumour)	< 3 y, **SMARCB1 (INI1) loss** – aggressive
Choroid plexus papilloma	Infants, **lateral ventricle**, hydrocephalus (CSF overproduction)
SEGA	Tuberous sclerosis – foramen of Monro; **everolimus**

Practice MCQs · Tumors of Infancy and Childhood

152 questions · correct option highlighted in green · cover the answer strip to self-test

Retinoblastoma

1. All are true about Retinoblastoma gene except:

A. Autosomal dominant inheritance
B. Needs both mutation of both copies of gene–two hit
C. Chr 13p14
D. Tumor suppressor gene

Ans C Autosomal dominant inheritance is seen in familial retinoblastoma Rb gene is a tumor suppressor gene on Chr 13q14.

2. Retinoblastoma spreads to CNS through:

A. Direct
B. Lymphatic
C. Arterial
D. Vitreous

Ans A Spread of Retinoblastoma:
• By direct extension to choroid; Along the optic nerve beyond lamina cribrosa to the CNS, or
• By hematogenous or lymphatic spread to distant sites, including bones, bone marrow, and lungs.

3. Flexner Wintersteiner rosettes are seen in:

A. Hepatoblastoma
B. Neuroblastoma
C. Nephroblastoma
D. Retinoblastoma

Ans D Rosette type Associated tumors Homer Wright rosetteQ Neuroblastoma, medulloblastoma, primitive neuroectodermal tumor, pineoblastoma Flexner-Wintersteiner rosetteQ Retinoblastoma, pineoblastoma, medulloepithelioma Perivascular pseudorosette Ependymoma, medulloblastoma, primitive neuroectodermal tomor, central neurocytoma, glioblas- toma, monomorphous pilomyxoid astrocytomas

4. Children with germline retinoblastoma are more likely to develop other primary malignancies in their later lifetime course. Which of the following can occur in such patients?

A. Osteosarcoma of lower limbs and soft tissue sarcoma
B. Thyroid carcinoma
C. Seminoma
D. Squamous cell carcinoma

Ans A Most common secondary tumor in a child with Retinoblastoma are Osteosarcoma, soft tissue sarcoma and malignant melanoma

5. True about retinoblastoma are all except:

A. AD
B. Most common intraocular tumor of childhood
C. Females are more affected than males
D. Bilateral involvement not rare

Ans C Discussing options one by one: a. AD: true; because of the loss of heterozygosity it behaves as autosomal dominant Let's try to understand it, in the hereditary form of retinoblastoma, the first mutation in RB gene is inherited through germinal cells and a 2nd mutation occurs subsequently in somatic retinal cells. 2nd mutations that lead to retinoblastoma often result in loss of the normal allele & concomitant loss of heterozygosity. b. True;

6. Retinoblastoma gene is located on:

A. Chromosome
B. Chromosome
C. Chromosome
D. Chromosome 16 NEUROBLASTOMA

Ans C 13 Retinoblastoma (RB) gene
• It is located on chr 13q14; its mutation or deletion is responsible for hereditary form of Retinoblastoma
• It encodes the retinoblastoma protein, a tumor suppressor protein that controls cell-cycle phase transition and has roles in apoptosis and cell differentiation with NEUROBLASTOMA

7. During a routine examination of a 10-mo-old male infant, you find a white pupillary reflex of right eye; the eye movements are normal. You suspect retinoblastoma. Of the following, the BEST confirmatory diagnostic evaluation of this infant is

A. indirect ophthalmoscopy with slit-lamp examination
B. examination under general anesthesia by an experienced ophthalmologist
C. retinal biopsy
D. orbital ultrasonography
E. brain MRI

Ans B Imaging studies are not diagnostic, and biopsies are contraindicated. Indirect ophthalmoscopy with slit-lamp evaluation can detect retinoblastoma tumors, but a complete evaluation requires an examination under general anesthesia by an experienced ophthalmologist to obtain complete visualization of both eyes, which also facilitates photographing and mapping of the tumors.

8. A 9-mo-old infant is recently diagnosed with bilateral retinoblastoma; examination under anesthesia reveals bilateral multifocal involvement of the retina. An important step in the management is

A. radiotherapy of both eyes
B. retinal examination of 1st degree relatives
C. orbital ultrasonography
D. bilateral bone marrow examination
E. bilateral enucleation

Ans B Retinoblastoma can be either hereditary or sporadic. Hereditary cases usually are diagnosed at a younger age and are multifocal and bilateral, whereas sporadic cases are usually diagnosed in older children who tend to have unilateral, unifocal involvement. All 1st-degree relatives of children with known or suspected hereditary retinoblastoma should have retinal examinations to identify retinomas or retinal scars.

Neuroblastoma

9. Highest cure rate is of:

A. Wilms' tumor
B. Retinoblastoma
C. Rhabdomyosarcoma
D. ALL

Ans D Discussing about the options one by one, a. Wilms' Tumor: Overall survival of children with WT approaches 90%, with some prognostic factors (low stage, favorable histology, young age, low tumor weight) conferring even better outcomes. b. Retinoblastoma: 95% of children in US with retinoblastoma are cured with modern treatment. Although survival in developed countries is high, retinoblastoma progresses to metastatic disease & death in > 50% of children worldwide; c.

10. All of the following are true about neuroblastoma except:

A. Diarrhea
B. Bone pain
C. Cafe-au-lait spots
D. Opsomyoclonus

Ans C Cafeaulait spots are associated with neurofibromatosis.

11. All are good prognostic factors in neuroblastoma except:

A. Stage 4S
B. Trk A expression
C. Trk B expression
D. Age <18 months

Ans C

12. Most common site of metastases in neuroblastoma:

A. Lung
B. Skull
C. Liver
D. Vertebrae

Ans B Metastasis in Neuroblastoma
• Metastatic spread is more common in children >1 yr of age at diagnosis
• Most common organs involved are lymph nodes, long bones, skull, bone marrow, liver, & skin
• Lung & brain metastases are rare, occurring in less than 3% of cases

13. True regarding stage IV-S of neuroblastoma are all except:

A. Limited to infants < 1 year
B. Primary localized tumor
C. Dissemination to bone
D. Good prognosis

Ans C International Neuroblastoma Staging System (INSS) stage 4S refers to neuroblastoma in children <1 yr age with dissemination to liver, skin, and/or bone marrow without bone involvement and with a primary tumor that would otherwise be staged as INSS stage 1 or 2.

14. Opsoclonus is associated with:

A. Neuroblastoma
B. Nephroblastoma
C. Retinoblastoma
D. Hypernephroma

Ans A 'Opsoclonus–myoclonus–ataxia syndrome'
• Paraneoplastic syndrome of autoimmune origin associated with Neuroblastoma
• Rapid, uncontrollable jerking eye & body movements, poor coordination, cognitive dysfunction scan

15. Which of the following statements about neuroblastoma is not true?

A. Most common extracranial solid tumor in childhood
B. > 50% present with metastasis at the time of diagnosis
C. Lung metastasis are common
D. Often encase aorta and its branches at the time of diagnosis

Ans C

16. Neuroblastoma originates from:

A. Adrenals
B. Mediastinum
C. Chest wall
D. Peripheral nerves
E. Neck

Ans A b. Mediastinum and e. Neck Origin of neuroblastoma:
• Adrenal medullaQ (Most common site) f/b Along the sympathetic chain in the paravertebral region of abdomen (25%)
• Posterior mediastinumQ (15%); Other sites: pelvis, neck, and brain (cerebral neuroblastomas)

17. To which of the following events is 'good' outcome in neuroblastoma associated?

A. Diploidy
B. N-myc amplification
C. Chromosome I p deletion
D. Trk A expression

Ans D Trk A expression (Trk'A' = 'A'chaa)

18. Neuroblastoma differs from Wilms' tumor radiologically by all except:

A. Calcification
B. Aorta and IVC are not eroded but pushed aside
C. Location
D. Intraspinal extension of tumor

Ans A Calcification 36 below;

19. **Neuroblastoma—good prognostic factor is:**

A. N-myc amplification
B. RAS oncogene
C. Hyperdiploidy
D. Translocation
E. Hypodiploidy

Ans C

20. **Which statement(s) is/are true about neuroblastoma with respect to Wilms' tumor?**

A. Neuroblastoma causes displacement of kidney infero-laterally without distortion of collecting system
B. Stippled calcification is present in Wilms' tumor
C. Aortic and IVC invasion by neuroblastoma
D. Neuroblastoma crosses midline
E. Intraspinal extension by Wilms' tumor

Ans A Neuroblastoma causes displacement of kidney inferolaterally without distortion of collecting system, b. Stippled calcification is present in Wilms' tumor and d. Neuroblastoma crosses midline Comparison between Wilms' tumor & Neuroblastoma: Wilms' Tumor Neuroblastoma Abdominal mass Yes Yes Primary origin IntrarenalQ Extrarenal; Arises from adrenal gland or paravertebral sympathetic ganglia Physical examination Displacing mass, mainly confined to the flank Non-mobile mass. more likely to cross the midlineQ

21. **A 9-mo-old infant develops a left adrenal mass; histological examination reveals a significant degree of undifferentiated round blue cells consistent with neuroblastoma. Which of the following genetic characteristics of the tumor cells carry a better outcome?**

A. amplification of the MYCN (N-myc) proto-oncogene
B. hyperdiploidy
C. loss of heterozygosity of 1p chromosome
D. loss of heterozygosity of 11q chromosome
E. gain of 17q chromosome

Ans B confers better prognosis if the child is younger than 1 yr of age at diagnosis. Amplification of MYCN is strongly associated with advanced tumor stage and poor outcomes. Other chromosomal abnormalities, including loss of heterozygosity of 1p, 11q, and 14q, and gain of 17q, are commonly found in neuroblastoma tumors and are also associated with worse outcomes.

22. **A 1-year-old child develops right eye ptosis, myosis, and loss of sweating; you suspect neuroblastoma. Of the following, the MOST valuable investigation to confirm the diagnosis is**

A. Computerized tomography (CT) scan of the neck and chest
B. CT scan of the abdomen
C. CT scan of the brain
D. magnetic resonance imaging (MRI) of the brain
E. X-ray the chest

Ans A Horner syndrome, characterized by unilateral ptosis, myosis, and anhidrosis, is associated with a thoracic or cervical primary tumor (e.g., superior cervical ganglion). Symptoms do not resolve with tumor resection.

23.
Neuroblastoma is a highly malignant childhood neoplasm. Metastatic spread can occur via local invasion or distant hematogenous or lymphatic routes. Of the following, the LEAST common site of metastases in neuroblastoma is

A. long bones
B. bone marrow
C. lung
D. skin
E. liver

Ans C The most common sites of metastasis are the regional or distant lymph nodes, long bones and skull, bone marrow, liver, and skin. Lung and brain metastases are rare, occurring in >3% of cases.

24. **Neuroblastoma can be associated with paraneoplastic syndrome. All the following are paraneoplastic features of neuroblastoma EXCEPT**

A. uncontrollable jerking eye and body movements
B. cerebellar ataxia and poor coordination
C. increased sweating and hypertension
D. unilateral ptosis, myosis, and anhidrosis
E. profound secretory diarrhea

Ans D Horner syndrome results from direct invasion of the neural foramina by tumor, causing nerve root compression. opsoclonus– myoclonus–ataxia syndrome is of paraneoplastic nature of autoimmune origin. Neuroblastoma can produce catecholamines that can cause increased sweating and hypertension; it may release vasoactive intestinal peptide, causing a profound secretory diarrhea.

25. **You are evaluating a 6-mo-old girl with a firm abdominal mass. Radiography reveals a calcified small right suprarenal mass. The tumor is completely resected and histologically is confirmed as neuroblastoma. Metastatic work-up shows massive liver involvement, 10% bone marrow involvement by tumor cells, and multiple subcutaneous nodules involvement. There is no bony involvement. The N-myc oncogene from the tumor is not amplified. According to the international neuroblastoma staging system, the infant is stratified as**

A. stage I
B. stage II A
C. stage III
D. stage IV
E. stage IV S

Ans E Infants younger than 1 yr of age also can present in unique fashion, termed stage 4S, with widespread subcutaneous tumor nodules, massive liver involvement, limited bone marrow disease, and a small primary tumor without bone involvement or other metastases.

26. **A 10-mo-old boy has a left suprarenal mass. Surgery is accomplished with complete surgical removal of the mass as well as the non-adherent ipsilateral lymph nodes; multiple contralateral lymph nodes biopsies are taken during surgery. The histology reveals poorly differentiated neuroblastoma with microscopic involvement of the ipsilateral lymph nodes; the contralateral lymph nodes are negative for tumor cells. Of the following, the BEST therapeutic approach for this infant is**

A. chemotherapy
B. radiotherapy
C. concomitant chemo-radiotherapy
D. chemotherapy followed by radiotherapy
E. close observation

Ans E The infant has stage 2A and is stratified as low risk. The usual treatment for children with low-risk neuroblastoma is surgery for stages 1 and 2 and observation for stage 4S with cure rates generally >90% without further therapy.

27. **Children with high-risk neuroblastoma have poor survival. Current treatment consists of all the following EXCEPT**

A. surgery
B. intensive chemotherapy
C. radiation
D. allogenic bone marrow transplantation
E. differentiating agents (13-cis-retinoic acid)

Ans D High-dose chemotherapy with autologous stem cell rescue, and not allogenic bone marrow transplantation, is one of the treatment modality for high risk patients.

Wilms Tumor

28. **Most common inherited childhood tumor is:**

A. Leukemia
B. Neuroblastoma
C. Retinoblastoma
D. Wilms' tumor

Ans C

29. **Which of the following are true about Wilms' tumor?**

A. In stage 2 tumor extend into inferior vena cava
B. Most common presentation is asymptomatic abdominal mass
C. Hypertension is seen in >80% patients
D. Patient with stage 1 requires radiation therapy after chemotherapy
E. Prognosis depends upon histology and staging

Ans B e. Prognosis depends upon histology and staging Extension to Inferior vena cava occurs in stage 3 of Wilms' tumor; Hypertension is present in ~25% of tumors at presentation & has been attributed to increased renin activity. a. A s asymptomatic abdominal mass • Most common initial clinical presentation for WT is incidental discovery of an asymptomatic abdominal mass by parents while bathing or clothing an affected child or by a physician during a routine physical examination;

30. **The most common presentation of a child with Wilms' tumor is:**

A. As asymptomatic abdominal mass
B. Hematuria
C. Hypertension
D. Hemoptysis due to pulmonary secondary

Ans A Wilms' tumor most commonly presents as an asymptomatic abdominal mass discovered incidentally by a parent or on examination.

31. **All are true about Wilms' tumor except:**

A. Triphasic morphology
B. MC renal malignancy in children
C. Associated with hemihypertrophy
D. Does not respond to chemotherapy and radiotherapy

Ans D Patients with favorable histologic findings on Wilms' tumor biopsy, have a good outcome, as it is sensitive to chemotherapy & radiotherapy.

32. **All are true about Wilms' tumor except:**

A. Most commonly presents as abdominal mass in children
B. Locally invasive
C. Chloramphenicol is used
D. All are true

Ans C • Chloramphenicol is not used for treatment of Wilms' tumor.

33. **Wilms' tumor is associated with the following except:**

A. Aniridia
B. Horse-shoe kidney
C. Hemihypertrophy
D. Opsoclonus

Ans D Opsoclonus is seen in Neuroblastoma & not Wilms' tumor

34. **Which of the following is post-chemotherapy based staging system in Wilms' tumor?**

A. National Wilms' tumor staging system (NWTS)
B. International society of pediatric oncology (ISPO)
C. AJCC TNM
D. Chadwick

Ans A • The two major groups which have tremendous contributions in the management of Wilms' tumor are National Wilms' Tumor Study (NWTS) and the Societe Internationale D'oncologie Pediatrique (SIOP).
• In 2001, NWTS merged with several other pediatric oncology cooperative groups to create the Children's Oncology Group (COG). However, the NWTS is still active in name today completing follow-up of the late effects of treatment for patients previously enrolled in its trials.

35. **Wilms' tumor most commonly presents as:**

A. Fever
B. Hematuria
C. Abdominal mass
D. Pain in abdomen

Ans C

36. Which of the following are associated with Wilms' tumor?

A. Aniridia
B. Genitourinary abnormalities
C. Cataract
D. Hemihypertrophy
E. Macroglossia

Ans A b. Genitourinary abnormalities, d. Hemihypertrophy and e. Macroglossia Aniridia & Genitourinary abnormalities are seen in WAGR syndrome; Hemihypertrophy & macroglossia are features of Beckwith Wiedemann syndrome; Both these syndromes are associated with Wilms' tumor.

37. The ideal timing of radiotherapy for Wilms' tumor after surgery is:

A. Within 10 days
B. Within 2 weeks
C. Within 2 months
D. Any time after surgery

Ans B with Radiation therapy for Wilms' tumor:
• Dose for favorable histology Wilms' tumor is 1080 cGy for the abdomen & 1200 cGy for the lung.
• Postoperative radiotherapy is started within 14 days of nephrectomy.

38. The most important determinant of prognosis in Wilms' tumor:

A. Stage of disease
B. Loss of heterozygosity of chromsome I p
C. Histology
D. Age less than one year at presentation

Ans C • Histology plays a major role in risk stratification of Wilms' tumor; Absence of anaplasia is considered favorable.
• Other prognostic factors are: age, stage, tumor weight, & loss of heterozygosity at chromosomes 1p & 16q.

39. Constellation of aniridia and hemihypertrophy is strongly associated with increased risk of which of the following tumors?

A. rhabdomyosarcoma
B. hepatoblastoma
C. Wilms` tumor
D. neuroblastoma
E. medulloblastoma

Ans C Beckwith-Wiedemann syndrome, an overgrowth syndrome characterized by macrosomia, macroglossia, hemihypertrophy, omphalocele, and renal anomalies, is associated with an increased risk of Wilms` tumor, hepatoblastoma, rhabdomyosarcoma, neuroblastoma, and adrenal cortical carcinoma, while aniridia is associated with Wilms` tumor.

40. A 2-year-old male child has an asymptomatic right flank mass discovered incidentally by the mother while bathing. Suspicion of Wilms` tumor is raised. Of the following, the LEAST likely investigation to be performed in this child is

A. plain abdominal radiography
B. computerized tomography (CT) scan of the abdomen
C. CT scan of the chest
D. Biopsy of the mass
E. ultrasonography with Doppler imaging of the abdominal blood vessels

Ans D Although biopsy is a reliable diagnostic tool, it is discouraged as it results in disease upstaging. A core needle biopsy obtained via a posterior approach should be performed in cases of unusual presentation (older age, signs of infection, inflammation) or unusual imaging findings (significant adenopathy, no renal parenchyma seen, intratumoral calcification).

41. Biopsy for children with Wilms` tumor is not usually performed as it may upstage the disease. All the following are indications of accomplishing renal biopsy for a child with suspicion of Wilms` tumor EXCEPT

A. age of 2-3 year at presentation
B. signs of infection and inflammation around the tumor
C. significant abdominal lymph nodes enlargement radiologically
D. absence of renal parenchyma radiologically
E. radiological appearance of intratumoral calcification

Ans A Older age, more than 5 years, is an indication to perform renal biopsy.

42. A 2-year-old child is being evaluated for a right flank mass; radiological appearance is consistent with right renal Wilms` tumor. During radical nephrectomy there is intra-abdominal tumor rupture stated by the surgeon. Of the following, the BEST therapeutic approach for this child is

A. chemotherapy
B. radiotherapy
C. concomitant chemotherapy-radiotherapy
D. another surgery
E. observation

Ans C The child is stratified as stage III; so he needs chemo-radiotherapy.

43. Although children with Wilms` tumor have a favorable prognosis, there are some adverse factors that affect outcome. All the following are poor prognostic factors in children with Wilms` tumor EXCEPT

A. young age at diagnosis
B. advanced stage
C. large tumors
D. anaplastic histology
E. loss of heterozygosity at chromosome 1p and 16q

Ans A Prognostic factors for risk-adapted therapy include age, stage, tumor weight, and loss of heterozygosity at chromosomes 1p and 16q. Histology plays a major role in risk stratification of WT. Absence of anaplasia is considered a favorable histologic finding.

44. Although it is not common, about 15% of children with Wilms` tumor suffer relapse, most relapses occur early (within 2 yr of diagnosis). Factors associated with a favorable outcome after relapse include all the following

A. low stage (I/II) at diagnosis
B. no prior radiotherapy
C. **anaplastic histology**
D. relapse to lung only
E. late relapse more than 12 mo from nephrectomy

Ans C Factors associated with a favorable outcome after relapse include low stage (I/II) at diagnosis, treatment with vincristine and actinomycin D only, no prior radiotherapy, favorable histology, relapse to lung only, and interval from nephrectomy to relapse 12 mo or longer.

Hepatoblastoma

45. Childhood malignancy producing proptosis are:

A. **Neuroblastoma**
B. Hepatoma
C. Retinoblastoma
D. Germ cell tumor
E. Nephroblastoma

Ans A c. Retinoblastoma Proptosis Definition Protrusion of the eye; it is a common indicator of orbital disease Pathophysiology It may be caused by: shallowness of the orbits, as in many craniofacial malformations, or by increased tissue mass within the orbit, as with neoplastic, vascular, and inflammatory disorders Tumors of Benign tumors: Most common are vascular lesions (principally hemangiomas) & dermoids orbit causing Malignant neoplasms: Rhabdomyosarcoma, Lymphosarcoma & Metastatic Neuroblastoma are the most

46. Deletion of chromosome 11 leads to:

A. **As asymptomatic abdominal mass**
B. Hematuria
C. Hypertension
D. Hemoptysis due to pulmonary secondary CNS TUMORS

Ans A Wilms' tumor

47. The most common malignancy in childhood resulting to death is:

A. Leukemia
B. Wilms' tumor
C. Retinoblastoma
D. **Neuroblastoma**

Ans D Tumor Leukemia Wilms tumor Retinoblastoma Neuroblastoma 5 year survival 80-90% 90% 97% 78% This shows the most common cause of death (malignancy associated) is Neuroblastoma

48. Of the following, the genetic syndrome MOST likely associated with increased risk of optic glioma is

A. Down syndrome
B. **neurofibromatosis**
C. monosomy
D. Bloom's syndrome
E. ataxia-telangiectasia

Ans B 1 is strongly associated with optic glioma. Down syndrome and Monosomy 7 are associated with increased risk of AML. Bloom's syndrome and ataxia-telangiectasia are associated with increased risk of ALL.

49. Chemotherapy has a major role in many childhood CNS tumors. In which of the following tumors chemotherapy is not effective?

A. medulloblastoma
B. pilocytic astrocytoma
C. **craniopharyngioma**
D. pineoblastoma
E. supratentorial primitive neuroectodermal tumors (SPNETs)

Ans C There is no role for chemotherapy in craniopharyngioma.

50. Hepatoblastoma occurs predominantly in children younger than 3 yr of age. Although etiology is unknown, there are many associated risk factors. All the following are associated risk factors for development of hepatoblastoma EXCEPT

A. familial adenomatous polyposis
B. Beckwith-Wiedemann syndrome
C. prematurity
D. low birth weight
E. **hepatitis C infection**

Ans E Hepatitis B or C infection is often associated with hepatocellular carcinoma.

51. Hepatoblastoma is the most common neoplasm of the liver; it arises from the precursors of hepatocytes. It is of different histological classification which has a direct correlation with clinical outcome. Of the following, the histological type that predict the MOST favorable outcome is

A. **epithelial type of pure fetal histology**
B. epithelial type of pure embryonal histology
C. epithelial type of mixed histology (fetal and embryonal)
D. mixed type of epithelial and mesenchymal elements
E. undifferentiated type

Ans A The pure fetal histology subtype predicts a more favorable outcome and the small cell undifferentiated subtype is associated with normal AFP levels and predicts a worse outcome.

Rhabdomyosarcoma

52. A malignant tumor of childhood that frequently metastasizes to bone marrow most often is:

A. Wilms' tumor
B. **Neuroblastoma**
C. Adrenal gland tumors
D. Granulosa cell tumor of ovary

Ans B The malignancies of prostate, breast, lungs, kidney & thyroid in adults and neuroblastoma and rhabdomyosarcoma in children are the primary tumors which frequently involve bone marrow.

53. MC optic nerve tumor in children causing blindness:

A. **Glioma**
B. Meningioma
C. Craniopharyngioma
D. Astrocytoma

Ans A about Optic Glioma
• Optic nerve glioma is the most frequent tumor of optic nerve in childhood; it is a benign hamartoma;
• Principal clinical manifestations are unilateral loss of vision, proptosis, deviation of the eye & optic atrophy

54. Rhabdomyosarcoma is treated by:

A. Chemotherapy
B. Radiation
C. Surgery
D. All of the above

Ans D Treatment of Rhabdomyosarcoma: for the staging of medullo- of 5-7 yr. exchanges. syndrome with uveitis of eyebrows & eyelashes cancers Pheochromocytoma
• It is based on risk classification of the tumor, which is determined by the stage of tumor, the tumor histology & the amount of Answers tumor that was surgically resected prior to chemotherapy: – For low-risk: vincristine & actinomycin – For higher-risk disease: vincristine, actinomycin & cyclophosphamide along with radiation.

55. Which is not a tumor of the first decade of life? (DPG 10, AIIMS 92) a Rhabdomyosarcoma

B. Neuroblastoma
C. Ameloblastoma
D. Retinoblastoma

Ans C

56. Alveolar type of rhabdomyosarcoma (RMS) accounts for approximately 1/3 of all cases of pediatric RMS and carries the poorest prognosis. Of the following, the MOST common site of involvement by alveolar type RMS is

A. orbit
B. middle ear
C. extremities
D. vagina
E. bladder

Ans C Alveolar tumors occur most often in the trunk and extremities.

57. In pediatric rhabdomyosarcoma, stages are dependent on primary site whether favorable or unfavorable. Which of the following sites of involvement is considered unfavorable?

A. vagina
B. uterus
C. testis
D. orbit
E. paranasal sinuses

Ans E Favorable sites include female genital, paratesticular, and head and neck (nonparameningeal) regions; all other sites are considered unfavorable. Involvement of paranasal sinuses by rhabdomyosarcoma is considered as unfavorable site.

Osteosarcoma & Ewing Sarcoma

58. Which SOLID Tumor causes bilateral proptosis?

A. Rhadbomyosarcoma
B. Retinoblastoma
C. Neuroblastoma
D. PNET

Ans C • Retinoblastoma is an intraocular malignancy; Rhabdomyosarcoma presents as a unilateral proptosis.
• In Neuroblastoma, orbital metastases may be bilateral & present with abrupt onset proptosis & lid ecchymosis (Raccoon eyes).

59. Which of the following childhood tumors show extracranial metastasis?

A. Glioblastoma multiforme
B. Medulloblastoma
C. Choroid plexus carcinoma
D. Ependymoblastoma

Ans B Among the given options, Medulloblastoma is the most malignant brain tumor seen in children. Up to 30% of patients with medulloblastoma present with neuroimaging evidence of leptomeningeal spread. b. Medulloblastoma Chang Staging System: was published in 1969 and was the most widespread classification system blastomas. However, it is no longer used regularly in clinical practice.

60. Uncommon in children:

A. Ewing's sarcoma
B. Neuroblastoma
C. Osteosarcoma
D. None of above

Ans D • Osteosarcoma is most common primary malignant bone tumor in children followed by Ewing sarcoma.
• In children younger than 10 yrs of age, Ewing sarcoma is more common than osteosarcoma.
• Both tumor types are most likely to occur in the second decade of life, i.e. in adolescent age group

61. During the routine exam of an infant, the parents state a strong family history of adenomatous polyposis. The statement that should be included during the discussion is

A. the infant is at increased risk for colonic adenocarcinoma
B. the infant is at increased risk of acute lymphocytic leukemia
C. the infant is at increased risk of intestinal Burkitt lymphoma
D. the infant is at increased risk of hepatoblastoma
E. the infant is at increased risk of germ cell tumor

Ans D Beckwith-Wiedemann syndrome, hemihypertrophy, Gardner syndrome, and family history of adenomatous polyposis are associated with increased risk of hepatoblastoma.

62. Osteosarcoma is the most common primary malignant bone tumor in children and adolescents; it has multiple subtypes and requires different modalities of treatment including surgery and chemotherapy. Of the following, the subtype of osteosarcoma which is treated by surgery alone is

A. fibroblastic
B. chondroblastic
C. telangiectatic
D. parosteal
E. periosteal

Ans D osteosarcoma is a low-grade, well-differentiated tumor that does not invade the medullary cavity and most commonly is found in the posterior aspect of the distal femur. Surgical resection alone often is curative in this lesion, which has a low propensity for metastatic spread.

63. Of the following, the WORST prognostic factor in pediatric osteosarcoma is

A. primary pelvic bone tumor
B. poor histologic response to treatment
C. bony metastases at the time of diagnosis
D. lung metastases at the time of diagnosis
E. chondroblastic subtype

Ans C All the mentioned factors carry poor prognosis, but multiple bony metastases at the time of diagnosis is the worst.

Brain Tumors

64. A 4-year-old child having palpable abdominal mass and hypertension with sweating and diarrhea is due to:

A. **Neuroblastoma**
B. Nephroblastoma
C. PCKD (Polycystic kidney disease)
D. All of the above

Ans A Sometimes neuroblastoma produce catecholamines that can cause increased sweating and hypertension, and some release vasoactive intestinal peptide, causing a profound secretory diarrhea.

65. Medulloblastoma is:

A. Malignant tumor in adults
B. **Malignant tumor in children**
C. Benign brain tumor
D. Malignant cerebellar tumor in adults

Ans B Medulloblastoma
• WHO grade IV tumor;
• It is an embryonal tumor or PNET
• Occurs predominantly in children & exclusively in Cerebellum
• Dissemination through the CSF is a common complication—giving rise to nodular masses at some distance from the primary tumor called as "drop metastases."
• Treatment: Exquisitely radiosensitive.

66. Syndromes related to child brain tumor include:

A. **Tuberous sclerosis**
B. Neurofibromatosis
C. Cockayne syndrome
D. Cowden syndrome
E. Pierre Robin syndrome

Ans A b. Neurofibromatosis & d. Cowden syndrome Cockayne syndrome is a rare autosomal recessive disorder, characterized by facial erythema in a butterfly distribution after sun exposure. Patients with Cockayne syndrome do not have increased risk of cancers.

67. All of the following are true about Medulloblastoma except:

A. **It arises from Medulla**
B. It is an embryonal tumor
C. It is a small round blue cell tumor
D. Chang staging system used

Ans A Medulloblastoma is a cerebellar tumor occurring predominantly in males & at a median age of 5–7 years.

68. A 1.5-year-old female is brought to the clinic with complaints of excessive enlargement of head, intolerance to feeds and severe malnourishment. MRI imaging was suggestive of a medulloblastoma causing obstructive hydroc ephalus. Which of the following is an example of irrational management of the patient?

A. Craniotomy and sub-total excision of the tumor. Surgeon leaves the layer of the tumor adherent with colliculus
B. First ventriculoperitoneal shunt was done
C. CCNU and vincristine were given as chemotherapy
D. **Radiotherapy 35–40 Gy was given to the whole cranio spinal axis**

Ans D Radiotherapy 35–40 Gy was given to the whole craniospinal axis In children < 3 years age, use of radiation therapy is not preferred, as it results in severe late neurologic sequelae, including microcephaly, learning disabilities, cognitive impairment, neuroendocrine dysfunction (growth failure, hypothyroidism, hypogonadism, and absence/delay of puberty) &/or second malignancies. Treatment of Medulloblastoma:

69. About Pilocytic astrocytoma, false is:

A. Spreads to posterior fossa
B. **Seen at eighty years of age**
C. Best prognosis among all intracranial neoplasms.
D. Surgery and radiation therapy used for treatment

Ans B Pilocytic Astrocytoma usually occurs in children & young adults.

70. Which of the following statements about cerebellar astrocytomas in pediatric age group is False?

A. These are usually low grade tumors
B. These tumors have a good prognosis
C. These are more commonly seen in 1st & 2nd decades
D. **These tumors are more common in females**

Ans D dna 54. b. Craniopharyngioma Craniopharyngioma
• It is a solid tumor with cysts and calcification located in the suprasellar region.
• It presents with (short stature), visual field defects & hydrocephalus.

71. Medulloblastoma exclusively occurs in the:

A. Medulla
B. **Cerebellum**
C. Cerebral hemispheres
D. Spinal cord

Ans B • Medulloblastoma is a cerebellar tumor occurring predominantly in males and at a median age fo • CT & MRI show a solid, homogeneous, contrast enhancing 4th ventricular obstruction & hydrocephalus. weiveR 56. d. Midline posterior fossa tumor

72. True statement regarding brain tumor in children is:

A. **Mostly is infratentorial**
B. Papilledema is rare
C. Is the most common tumor in children
D. Hydrocephalus is rare OTHER CHILDHOOD TUMORS

Ans A Remember, the most common tumor in children is acute leukemia.

73. **A 6-year-old male child presented with a cerebellar mass; after undergoing complete surgical resection of the mass, the histology of the mass reveals pilocytic astrocytoma. Of the following, the MOST appropriate next step in the management is**

A. radiotherapy
B. chemotherapy
C. **observation**
D. concomitant chemo-radiotherapy
E. radiotherapy followed by chemotherapy

Ans C With complete surgical resection the overall survival approaches 80-100%.

74. **You are discussing the risk of radiotherapy with the parents of a child with medulloblastoma; the mother has a concern about the late neurological complications post radiotherapy. The statement that should be included in the discussion that late neurological sequelae post radiotherapy is more severe with**

A. focal radiotherapy rather than craniospinal irradiation
B. **children with an age of less than 3 year**
C. concomitant chemo-radiotherapy
D. low grade tumors rather than high grade tumors
E. infratentorial tumors rather than supratentorial tumors

Ans B

75. **The least common late neurologic sequelae that may be encountered post craniospinal irradiation for a 9-year-old child with medulloblastoma is**

A. **microcephaly**
B. learning disabilities
C. cognitive impairment
D. neuroendocrine dysfunction
E. second malignancy

Ans A Craniospinal radiation in children younger than 3 yr of age results in severe late neurologic sequelae, including microcephaly, learning disabilities, cognitive impairment, and neuroendocrine dysfunction. Similarly, in older children, late sequelae, such as learning disabilities, neuroendocrine dysfunction, and/or second malignancies, can occur.

76. **Extraneural metastasis from primary brain tumors is less likely to occur. Which of the following primary brain tumor is MOST commonly metastasizing extraneurally?**

A. **medulloblastoma**
B. primitive neuroectodermal tumor (PNET)
C. ependymoma
D. malignant glioma
E. choroid plexus carcinoma

Ans A

77. **Childhood primary brain tumors are less likely to metastasize extraneurally. Which of the following is MOST likely considered a risk factor for extraneural metastasis?**

A. supratentorial tumor
B. age of the patient of less than 10 year
C. female gender
D. **ventriculoperitoneal (VP) shunt insertion**
E. glial tumors

Ans D Ventriculoperitoneal shunts have been known to allow extraneural metastases, primarily within the peritoneal cavity but also systemically.

Leukemia & Lymphoma

78. **Catecholamines are secreted by:**

A. Retinoblastoma
B. Nephroblastoma
C. **Neuroblastoma**
D. Glioblastoma

Ans C

79. **Wilm's tumor is associated with all the following except:**

A. Aniridia
B. Beckwith syndrome
C. **Polycystic kidney**
D. Hemi-hypertrophy

Ans C Wilms' tumor is associated with WAGR syndrome, Denys Drash Syndrome & Beckwith Wiedemann Syndrome (hemih ypertrophy).

80. **A 6-year-old boy has been complaining of headache, ignoring to see objects on the sides for four months. On examination, he is not mentally retarded, his grades at school are good, and visual acuity is diminished in both eyes. Visual charting showed significant field defect. CT scan of head showed suprasellar mass with calcification. Which of the following is the most probable diagnosis?**

A. Astrocytoma
B. **Craniopharyngioma**
C. Pituitary adenoma
D. Meningioma

Ans B A suprasellar calcified mass with bitemporal visual field defect and preserved intellect in a school-age child is classic for craniopharyngioma.

81. **All are associated with malignancy except:**

A. Down's syndrome
B. **Fragile X-syndrome**
C. Bloom syndrome
D. Fanconi's anemia

Ans B Genetic diseases associated with cancer Disorder Tumor/ Cancer Down syndrome AML M7, ALL, Transient leukemoid reaction Fanconi anemia Leukemia, Myelodysplastic syndrome, Gastrointestinal and genitourinary Bloom syndrome Leukemia, Lymphoma, Solid tumors Neurofibromatosis type 1 Neurofibroma, Optic glioma, Acoustic neuroma, Astrocytoma, Meningioma, Neurofibromatosis type 2 Bilateral acoustic neuromas, Meningiomas Tuberous sclerosis Fibroangiomatous nevi, Myocardial rhabdomyoma 60 a.

82. **All of the following are small round blue cell tumors except:**

A. Fibrosarcoma
B. **Sarcoma botryoides**
C. Leiomyosarcoma
D. Inflammatory polyp

Ans B Osteosarcoma Small round blue cell tumors are tumors with similar histology of small round cell.

83. **A child presents with seborrheic dermatitis, sinusitis and chronically draining ears. On examination child has failure to thrive, with hepatosplenomegaly and exophthalmos, probable diagnosis is:**

A. Histiocytosis X
B. Wegener's granulomatosis
C. Retinoblastoma
D. Chediak-Higashi syndrome

Ans A The given clinical picture prints to a diagnosis Langerhans cell; Histiocytosis of previously called Histiocytosis X.

84. **Germ cell tumor(s) of pediatric age group includes all Questions except:**

A. Pure yolk sac tumor
B. Leydig cell tumor
C. Choriocarcinoma
D. Embryonal cell carcinoma e Endodermal sinus tumor

Ans B Testicular Tumors Germ cell tumors (95%) Non-germinal tumors (5%) a. Seminomatous b. Non-seminomatous • Sertoli cell tumor
• Seminoma • Embryonal carcinoma • Leydig cell tumor
• Spermatocytic seminoma • Choriocarcinoma
• Yolk sac tumor
• Endodermal sinus tumor
• Teratoma
• Most malignant tumors of the gonads in children are Germ cell tumors
• Testicular GCTs occur predominantly before age 4 yr and after puberty.

85. **True about childhood tumor are all except:**

A. Wilms' tumor
B. Neuroblastoma
C. Retinoblastoma
D. Embryonal rhabdomyoma
E. Pleomorphic rhabdomyosarcoma

Ans E Pleomorphic rhabdomyosarcoma arises in the deep soft tissue of adults; It is a rare tumor with poor prognosis.

86. **In PML, all of the following are seen except:**

A. Retinoic acid is used in treatment
B. 15/17 translocation may be seen
C. CD 15/34 both seen in same cell
D. Associated with disseminated intravascular coagulation

Ans C Acute Promyelocytic Leukemia is negative for CD34

87. **A 9-year-old boy develops acute myelogenous leukemia (AML) one year after completion of therapy for soft tissue sarcoma at his right thigh. Which of the following chemotherapeutic agents is MOST likely the cause of secondary acute myelogenous leukemia AML in this boy?**

A. cyclophosphamide
B. vincristine
C. etoposide
D. doxorubicin
E. cisplatin

Ans C Epipodophyllotoxins like etoposide may cause secondary AML early within two years after administration; alkylating agents like cyclophosphamide may cause secondary AML as far as 5 years after therapy.

88. **Epstein-Barr virus (EBV) infection is more likely to be associated with all the following malignancies EXCEPT**

A. Burkitt lymphoma
B. nasopharyngeal carcinoma
C. T-cell lymphoma
D. Hodgkin lymphoma
E. hepatocellular carcinoma

Ans E is associated with hepatitis B and C infection.

89. **Nearly all cervical carcinomas contain human papillomavirus (HPV). Which of the following types is never associated with such malignancy?**

A. type
B. type
C. type
D. type
E. type

Ans A High-risk HPVs include types 16 and 18 but also types 31, 33, 35, 45, and 56, which are also commonly found in women without lesions. The low-risk HPVs, including 6 and 11 that are commonly found in genital warts, are almost never associated with malignancies.

90. **Which of the following chromosomal abnormalities in acute lymphoblastic leukemia of childhood carry a favorable outcome?**

A. t(12;21)
B. t(4;11)
C. t(9;22)
D. hypodiploidy
E. 11q23

Ans A

91. **You are meeting with parents of 10-year-old child who recently develops acute lymphoblastic leukemia (ALL); the mother has a concern about risk of CNS relapse. Which of the following is LEAST likely to increase the risk of CNS relapse in children with ALL?**

A. first traumatic lumbar puncture (LP)
B. T-cell leukemia
C. cranial nerve involvement at the time of diagnosis
D. presence of lymphoblast in the CSF at any time during treatment
E. age of more than 10 year at the time of the diagnosis

Ans E

92. **Children with ALL who carry poor outcome include all the following EXCEPT**

A. age younger than 1 year and older than 10 year
B. T-cell immunophenotype
C. hyperdiploidy chromosomal abnormality
D. initial leukocyte count of > 50,000
E. poor response to initial therapy

Ans C More favorable characteristics include a rapid response to therapy, hyperdiploidy, trisomy of specific chromosomes (4, 10, and 17), and rearrangements of the ETV6-RUNX1 (formerly TEL-AML1) genes.

93. Which of the following chromosomal abnormalities of childhood ALL carries the highest risk of relapse despite intensive chemotherapy?

A. t(9;22)
B. **t(4;11)**
C. hypodiploidy
D. t(1;19)
E. t(12;21)

Ans B Infants with ALL, along with patients who present with specific chromosomal abnormalities, such as t(4; 11), have an even higher risk of relapse despite intensive therapy. However, the poor outcome of Philadelphia chromosome positive ALL with t(9;22) has dramatically changed by the addition of imatinib to an intensive chemotherapy backbone.

94. You are meeting the parents of a child with ALL who want to go for a trip with their child. The MOST nontoxic phase of the treatment of ALL that allow the family to have a safe trip is

A. before starting treatment
B. remission induction
C. consolidation
D. delayed intensification
E. **interim maintenance**

Ans E

95. You are evaluating a 6-year-old child with ALL on interim maintenance phase who has frequent mucositis and myelosuppression that needs frequent discontinuation of his treatment. Of the following, the MOST valuable test for this child is

A. complete blood count
B. bone marrow study
C. **pharmacogenetic testing of the thiopurine s-methyltransferase (tpmt) gene**
D. lumbar puncture
E. renal function test

Ans C which encodes one of the metabolizing enzymes of mercaptopurine, can identify patients who are wild type (normal TPMT enzyme activity), heterozygous (slightly decreased TPMT enzyme activity), or homozygous (low or absent enzyme activity). Decreased TPMT enzyme activity results in an accumulation of a toxic metabolite of mercaptopurine and results in severe myelosuppression, requiring dose reductions of the chemotherapy.

96. You are evaluating a 9-year-old boy child with ALL who recently develops testicular relapse; an important statement that should be mentioned to his parents is

A. testicular relapse occurs in the majority of boys with ALL
B. it usually occurs during the course of therapy
C. such relapse occurs as painful swelling of oneor both testes
D. the diagnosis is confirmed by ultrasonography of the affected testis
E. **the majority of affected boys can be successfully retreated, and the survival rate is good**

Ans E Testicular relapse occurs in less than 2% of boys with ALL, usually after completion of therapy. Such relapse occurs as painless swelling of one or both testes. The diagnosis is confirmed by biopsy of the affected testis. Treatment includes systemic chemotherapy and possibly local irradiation. A high proportion of boys with a testicular relapse can be successfully retreated, and the survival rate of these patients is good.

97. Which of the following types of translocation of childhood AML that typically associated with granulocytic sarcoma mass?

A. t(15:17)
B. **t(8:21)**
C. inv(16)
D. t(6:9)
E. inv(3)

Ans B Discrete masses, known as chloromas or granulocytic sarcomas, can occur in the absence of apparent bone marrow involvement and typically are associated with a t(8;21) translocation. Chloromas also may be seen in the orbit and epidural space.

98. You are explaining the risk of leukemia in children with Down syndrome to medical students; your discussion will include all the following statements EXCEPT

A. acute leukemia occurs more frequently in children with Down syndrome than in the general population
B. AML is more common in children with Down syndrome in the 1st 3 yr of life than ALL as compared with the ratio of ALL/AML in general population
C. **in children with Down syndrome who have AML, the expected outcome of treatment is slightly inferior to that for other children**
D. patients with Down syndrome who develops acute leukemia demonstrate a remarkable sensitivity to antimetabolites
E. neonates with Down syndrome may develop a transient leukemia or myeloproliferative disorder

Ans C In AML, patients with Down syndrome have much better outcomes than non– Down syndrome children, with a >80% long-term survival rate.

99. A healthy 20-day-old male neonate with Down syndrome appears pale; examination reveals a palpable liver 6 cm below the right costal margin and palpable spleen 3 cm below the left costal margin; lab findings include: hemoglobin, 8.8 g/dl; platelets count, 55000/mm3; white blood cell count, 18700/mm3 with 10% blast cells; bone marrow examination is consistent with acute leukemia. Of the following, the BEST approach for the management is

A. intensive chemotherapy
B. low dose chemotherapy
C. pulses of chemotherapy
D. bone marrow transplantation
E. **close follow up**

Ans E Approximately 10% of neonates with Down syndrome develop a transient leukemia or myeloproliferative disorder which usually resolves within the 1st 3 mo of life. Although these neonates can require temporary transfusion support, they do not require chemotherapy unless there is evidence of life-threatening complications. However, those patients require close follow-up, because 20-30% will develop typical leukemia by 3 yr of life.

100. **Which of the following types of leukemia is MOST likely to develop in a 2-year-old child with Down syndrome who has been developed transient myeloproliferative disorder in the neonatal period?**

C. AML M1
D. AML M6
E. AML M7

Ans E Twenty-thirty percent of patients who have Down syndrome and who develop transient leukemia or myeloproliferative disorder will develop typical leukemia (often acute megakaryocytic leukemia) by 3 yr of life (mean onset, 16 mo).

101. **Which of the following chromosomal abnormalities is often characteristic of infantile ALL?**

A. t(4;11)
B. t(12;21)
C. t(9;22)
D. t(10;14)
E. t(1;19)

Ans A More than 80% of the cases of infantile ALL demonstrate rearrangements of the MLL gene, found at the site of the 11q23 band translocation, the majority of which are the t(4;11). This subset of patients largely accounts for the very high relapse rate.

102. **Hodgkin lymphoma in children usually has favorable outcome; however, the prognosis may be grim in some cases. Poor prognostic factors in Hodgkin Lymphoma include all the following EXCEPT**

A. age of more than 15 year at the time of the diagnosis
B. stage IV disease
C. presence of "B" symptoms
D. poor response to therapy manifested by positron emission tomography (PET) scan positivity
E. presence of bulky mediastinal mass

Ans A

103. **Metabolic derangement secondary to tumor lysis syndrome is commonly encountered after starting chemotherapy in children with malignancy; it include all the following EXCEPT**

A. hyperuricemia
B. hypernatremia
C. hyperkalemia
D. hyperphosphatemia
E. hypocalcemia

Ans B

104. **Childhood primary brain stem tumors are heterogenous group of tumors; the outcome usually depends on tumor location. Which of the following brain stem tumors, depending on the site of tumor, carry the worst prognosis?**

A. focal
B. dorsally exophytic
C. cervicomedullary
D. diffuse intrinsic
E. none of the above

Ans D Surgical resection is the primary treatment approach for focal and dorsally exophytic tumors and leads to a favorable outcome. Histologically, these 2 groups usually are low-grade gliomas. Cervicomedullary tumors, owing to their location, may not be amenable to surgical resection but are sensitive to radiation therapy. Diffuse intrinsic tumors, characterized by the diffuse infiltrating pontine glioma, are associated with a very poor outcome independent of histologic diagnosis.

105. **Small round blue cell tumors refer to tumors that have round blue cells histologically. They include all the following EXCEPT**

A. neuroblastoma
B. non-Hodgkin lymphoma
C. osteosarcoma
D. Ewing sarcoma
E. rhabdomyosarcoma

Ans C

106. **A 2-mo-old male infant has a left flank mass discovered incidentally by the mother; radiological imaging reveals a left renal mass. Of the following, the MOST likely diagnosis is**

A. nephroblastoma
B. neuroblastoma
C. mesoblastic nephroma
D. clear cell sarcoma
E. rhabdoid tumor of the kidney

Ans C is the most common solid renal tumor identified in the neonatal period. Most of the patients are diagnosed before 3 mo of age, whereas Wilms` tumor is rarely diagnosed before 6 mo of age. Radical nephrectomy is the treatment of choice and may be sufficient by itself.

107. **Renal cell carcinoma (RCC) is rare in children, accounting for <5% of all renal tumors of childhood. All the following are true regarding RCC in children EXCEPT**

A. patients may present with frank hematuria, flank pain, and/or a palpable mass
B. it can be asymptomatic and detected incidentally
C. it has a propensity to metastasize to the lungs, bone, liver, and brain
D. it can be associated with von hippel–lindau disease
E. local lymph node involvement has an adverse outcome

Ans E Unlike the case for adult RCC, local lymph node involvement is not a poor prognostic indicator in pediatric RCC. Nephrectomy alone may be adequate for early- stage RCC.

108. **A 12-year-old male adolescent has pain of the right upper thigh that gradually becomes more severe and most often at night; it usually relieved by taking salicylates medication. Examination reveals limping, atrophy, and weakness of the right lower extremity. Plain radiography shows a round lucent lesion at the diaphysis of the right upper femur, about 0.5 cm in diameter, surrounded by sclerotic cortical bone formation. Of the following, the MOST likely diagnosis is**

A. Ewing sarcoma
B. osteosarcoma
C. **osteoid osteoma**
D. osteoblastoma
E. nonossifying fibroma

Ans C s are diagnosed between 5 and 20 yr of age. The clinical pattern is characteristic, consisting of unremitting and gradually increasing pain that often is worst at night and is relieved by aspirin. Boys are more affected than girls. Radiographs are distinctive, showing a round or oval metaphyseal or diaphyseal lucency (nidus) surrounded by sclerotic bone.

109. **The serum α-fetoprotein (AFP) level is elevated with some malignant germ cell tumors (GCTs) especially endodermal sinus tumors; it can be used as a measure of response to treatment and during follow up after completion of chemotherapy. However, it is normally elevated during infancy. At which age does AFP physiologically fall to normal adult level?**

A. three month
B. **eight month**
C. one year
D. three year
E. ten year

Ans B Infants normally have higher levels of AFP, which fall to normal adult levels by about age 8 mo; consequently, high AFP levels must be interpreted with caution in this age group.

110. **The β subunit of human chorionic gonadotropin (HCG) is a useful surface marker in some malignant germ cell tumors (GCTs); it is secreted by syncytiotrophoblasts. In which of following GCTs this surface marker is characteristically elevated?**

A. teratoma
B. endodermal sinus tumor
C. **germinoma**
D. gonadoblastoma
E. embryonal carcinoma

Ans C Elevation of the β subunit of human chorionic gonadotropin, which is secreted by syncytiotrophoblasts, is seen with choriocarcinoma and germinomas.

111. **Kasabach-Merritt syndrome is a benign vascular tumor which may be life- threatening; it is characterized by all the following EXCEPT**

A. rapidly enlarging lesion
B. thrombocytopenia
C. microangiopathic hemolytic anemia
D. coagulopathy
E. **association with infantile hemangiomas**

Ans E All the features of Kasabach-Merritt syndrome results from platelet and red blood cell trapping and activation of the clotting system within the vasculature of the hemangioma. This syndrome is associated with kaposiform hemangioendotheliomas or tufted angiomas but not with infantile hemangiomas.

112. **You are examining a 12-year-old female adolescent with a small nevus in the left thigh; her mother is concerned regarding development of melanoma in future. All the following findings raise the suspicion of melanoma EXCEPT**

A. rapidly enlarging nevus
B. nevus with changing colors
C. nevus with irregular margins
D. easily bleeds nevus
E. **Spitz nevus**

Ans E is a benign harmless skin lesion. However, making the distinction from diagnosis of melanoma can be difficult.

113. **Although melanoma is relatively rare in children, some risk factors may increase its incidence. All the following are risk factors for development of melanoma EXCEPT**

A. positive family history of melanoma
B. **dark skinned child**
C. giant hairy nevus
D. dysplastic nevus syndrome
E. xeroderma pigmentosum

Ans B Patients with fair skin are at particularly high risk for development of melanoma.

114. **In Langerhans cell histiocytosis (LCH), systemic manifestations are more likely to occur in patients with multisystem disease who are at high risk of mortality (i.e., "risk organ"-positive patients). All the following are risk organs in LCH EXCEPT**

A. **bone**
B. liver
C. spleen
D. lung
E. hematopoietic system

Ans A

Langerhans Cell Histiocytosis

115. **A malignant tumor of childhood, that metastasizes to bones most often, is:**

A. Malignant teratoma
B. **Neuroblastoma**
C. Wilms' tumor
D. Hepatoblastoma

Ans B

116. **A 10-year-old child presented with headache, vomiting, gait instability and diplopia. On examination he had papilledema and gait ataxia. The most probable diagnosis is:**

A. Hydrocephalus
B. Brainstem tumor
C. Suprasellar tumor
D. **Midline posterior fossa tumor**

Ans D Headache, vomiting, gait ataxia, diplopia, and papilledema in a child point to a midline posterior fossa tumor (e.g., medulloblastoma) causing obstructive hydrocephalus.

117. **Which of the following is the most common malignant tumor of infancy?**

A. **Neuroblastoma**
B. Wilm's tumor
C. Polycystic kidney disease
D. Renal cell carcinoma

Ans A Both Neuroblastoma & Wilms' tumor can present as a solid abdominal mass with calcifications. As the child in the given question is an infant, Neuroblastoma is the best answer.

118. **Most common site of histiocytosis is:**

A. **Bone**
B. Skin
C. Lung
D. Liver

Ans A (especially the skull) is the most common site of involvement in Langerhans cell histiocytosis.

119. **Radiotherapy is an effective modality of treatment in variable pediatric solid tumors. Of the following, the LEAST responsive tumor to radiotherapy is**

A. rhabdomyosarcoma
B. neuroblastoma
C. nephroblastoma
D. **osteosarcoma**
E. Ewing sarcoma

Ans D is a radio-resistant tumor.

120. **A 3-day-old neonate has a large soft, painless mass involving the right lower part of the head and neck that transilluminate; computerized tomography reveals a cystic mass infiltrating the neck and intrathoracic region mostly the mediastinum. Of the following, the BEST modality of treatment for this neonate is**

A. aspiration of mass
B. surgical resection
C. **Injection of sclerosing agent**
D. laser therapy
E. systemic interferon

Ans C Localized cystic hygroma lesions may be surgically resected, but this can be difficult, owing to their infiltrative nature. Recurrence is common with incompletely resected lesions. Aspiration can provide temporary relief in an emergency and reaccumulation will occur. Treatment by injection of sclerosing agents (by streptococcal immunotherapeutic agent OK-432) is the treatment of choice; its use will prevent the need for surgery in most cases. Laser therapy is not used in such age.

121. **You are evaluating a 6-year-old male child with Langerhans cell histiocytosis (LCH); you states to the parents that skeletal survey and bone scan are important investigations as bones are the most common sites of involvement. Of the following, the MOST common site of bone involvement by LCH is**

A. **skull**
B. mandible
C. vertebra
D. pelvis
E. femur

Ans A LCH has an extremely variable presentation. The skeleton is involved in 80% of patients and may be the only affected site, especially in children older than 5 yr of age. Bone lesions may be single or multiple and are seen most commonly in the skull.

122. **Langerhans cell histiocytosis (LCH) has an extremely variable presentation. Single or multiple organs may be affected by the disease. Of the following, the LEAST likely organ affected by LCH is**

A. skin
B. bone
C. middle ear
D. **lung**
E. lymph nodes

Ans D Bone is the most common site of involvement by LCH. About 50% of patients experience skin involvement at some time during the course of disease. Localized or disseminated lymphadenopathy is present in approximately 33% of patients. Otitis media is present in 30-40% of patients. In 10-15% of patients, pulmonary infiltrates are found on radiography. The lesions may range from diffuse fibrosis and disseminated nodular infiltrates to diffuse cystic changes.

Germ Cell Tumors

123. **A 1-year-old child presented with abdominal mass with calcification, possibilities are:**

A. **Neuroblastoma**
B. Wilms' tumor
C. Nephronophthisis
D. Pheochromocytoma
E. Hydronephrosis WILMS' TUMOR

Ans A b. Wilms' tumor 36; Both neuroblastoma & Wilms' tumor can present as abdominal mass with calcification. WILMS' Tumor

124.A child with pigment dilution of skin and eyes, hemorrhagic diathesis, ceroid deposition within the reticuloendothelial system has:

A. Bloom syndrome
B. **Hermansky Pudlak syndrome**
C. Alezzandrini syndrome
D. Vogt-Koyanagi syndrome

Ans B Bloom syndrome
• AR mutation in BLM/RECQL3 gene, encoding a DNA helicase.
• Patients are sensitive to UV radiation with increased rates of chromosomal breaks & sister chromatid
• Erythema & telangiectasia on face after exposure to sunlight.
• Café-au-lait spots & hypopigmented macules may be present.
• Immunodeficiency, GI malabsorption & hypogonadism are common.
• Increased tendency to develop solid tumors & lymphoreticular malignancies.
• Sister chromatid exchange analysis used for diagnosis.

125.Which of the following does not require a lumbar puncture in children?

A. ALL
B. **HL**
C. NHL
D. AML

Ans B HL Discussing the options one by one, a. ALL CNS infiltration is common in ALL, so diagnostic lumbar puncture & CSF cytopathology to look for blasts is done; LP is also done to administer intrathecal methotrexate b. HL Usually Lumbar puncture is not required c. NHL Hematogenous spread may cause CNS spread, so LP done d. AML CNS spread though rare can be seen, which will require lumbar puncture for diagnosis as well as treatment

126.Undescended testis is a risk factor for the development of which of the following tumors?

A. rhabdomyosarcoma
B. leukemia
C. **yolk sac tumor**
D. lymphoma
E. neuroblastoma

Ans C

127. You are discussing with medical students the role of chemotherapy in malignant germ cell tumors (GCTs); you state that GCTs are sensitive to some types of chemotherapy. Of the following, the MOST effective chemotherapeutic agent in GCTs is

A. vincristine
B. cyclophosphamide
C. **cisplatin**
D. methotrexate
E. cytosine Arabinoside

Ans C based chemotherapy regimens usually are curative in GCTs that cannot be complete5ly resected, even if metastases are present.

128.You are meeting with parents of 12-year-old girl who recently diagnosed with papillary thyroid carcinoma (PTC). The statement that should be included in your discussion is

A. PTC has a grim overall prognosis
B. chemotherapy is the primary therapy
C. children with PTC do not require radioactive iodine therapy (131I)
D. **supraphysiologic levothyroxine is required during the course of treatment**
E. long-term follow-up involves monitoring of calcitonin/ carcinogenic embryonic antigen tumor markers

Ans D PTC has an excellent overall prognosis even in the presence of metastatic disease. The primary therapy is a total thyroidectomy and lymph node dissection. 131I is used postoperatively to treat distant metastasis and unresectable residual neck disease. The TSH level is suppressed by giving supraphysiologic levothyroxine as TSH stimulates tumor growth. Long-term follow-up involves monitoring of thyroglobulin tumor markers.

Hodgkin & Non-Hodgkin Lymphoma

129.Most common tumor of kidney in children is:

A. **Wilms' tumor**
B. Neuroblastoma
C. Polycystic kidney disease
D. Renal cell carcinoma
E. Lymphoma

Ans A Wilms' tumor dna • Wilms' tumor (WT), also known as nephroblastoma, is the most common primary malignant renal tumor of childhood;
• It is the second most common malignant abdominal tumor in childhood.
• The most common sites of metastases from Wilms tumor are the lungs, regional lymph nodes, and liver.

130.Most common tumor in infant:

A. **Hemangioma**
B. Brain tumor
C. Leukemia
D. Ewing sarcoma

Ans A Infantile hemangioma is the most common tumor of infancy overall.

131. Most common benign tumors during infancy is:

A. Lymphangioma
B. **Hemangioma**
C. Cystic hygroma
D. Lipoma

Ans B Hemangioma a. Bone

132.Which of the following malignancies is least likely to occur in a 10-month-old infant?

A. neuroblastoma
B. nephroblastoma
C. retinoblastoma
D. hepatoblastoma
E. **Ewing sarcoma**

Ans E During the 1st yr of life, embryonal tumors such as neuroblastoma, nephroblastoma (Wilms tumor), retinoblastoma, rhabdomyosarcoma, hepatoblastoma, and medulloblastoma are most common. As children age, bone malignancies, Hodgkin disease, gonadal germ cell malignancies (testicular and ovarian carcinomas), and other carcinomas increase in incidence.

133.A 12-year-old male adolescent, recently diagnosed with Hodgkin lymphoma; the PET-CT scan shows an involvement of the left cervical and supraclavicular lymph nodes; he has a history of drenching night sweating and high grade fever. Based on Ann Arbor Staging Classification for Hodgkin Lymphoma, the patient is classified as

A. stage IA
B. stage IB
C. stage IIA
D. **stage IIB**
E. stage IIIB

Ans D Stage II includes involvement of 2 or more lymph node regions on the same side of the diaphragm (II) or localized involvement of an extralymphatic organ or site and 1 or more lymph node regions on the same side of the diaphragm (IIE). HL is subclassified into A or B categories: A is used to identify asymptomatic patients and B is for patients who exhibit any B symptoms.

134.Which of the following manifestations is more specific to occur in anaplastic large cell lymphoma (ALCL) than other types of non-Hodgkin Lymphoma (NHL)?

A. primary intestinal involvement
B. bone marrow involvement
C. CNS involvement
D. **primary cutaneous involvement**
E. primary mediastinal involvement

Ans D The primary site of tumor involvement and metastasis pattern varies by pathologic subtype. Lymphoblastic lymphoma (LBL) commonly manifests as an intrathoracic or mediastinal supradiaphragmatic mass and also has a predilection for spreading to the bone marrow and CNS. Burkitt lymphoma (BL) commonly manifests as abdominal (sporadic type) or head and neck (endemic type) tumor and can metastasize to the bone marrow or CNS.

135.A 5-year-old child, recently diagnosed with ileocecal Burkitt lymphoma; lab investigations reveal: serum uric acid 12 mg/dl, serum sodium, 145 meq/dl; serum potassium, 4.5 meq/dl; serum phosphate 4.4 meq/dl; serum calcium, 8.9 mg/dl; blood urea, 22 mg/dl; serum creatinine, 0.8 mg/dl. Of the following, the MOST effective treatment is

A. excessive hydration
B. sodium bicarbonate
C. xanthine oxidase inhibitor
D. **recombinant urate oxidase**
E. no treatment

Ans D is preferred in patients with a high risk of tumor lysis. Frequently, only a single dose is needed; however, repeat doses can be given if a subsequent rise in uric acid is seen.

Soft Tissue Sarcomas

136.Most common abdominal mass in children:

A. Hydronephrosis
B. Wilms' tumor
C. **Neuroblastoma**
D. Rhabdomyosarcoma

Ans C • Neuroblastoma is the most common extracranial solid tumor in children
• Localized Neuroblastoma can manifest as an asymptomatic mass or can cause symptoms because of the mass itself, including weiveR spinal cord compression, bowel obstruction & superior vena cava syndrome. Note: Both Neuroblastoma & Wilms' tumor can present as abdominal mass in children but Neuroblastoma is more common than Wilms' tumor, so it is the better answer to this question

137. Most common cancer in children less than 10 years:

A. **Leukemia**
B. Neuroblastoma
C. Brain tumor
D. Wilm's tumor

Ans A with Most frequent childhood cancers are: (in order of decreasing frequency) Leukemia (Most common is ALL)Q > Neuroblastoma > Wilms' tumor > Hepatoblastoma > Retinoblastoma

138.A 7-year-old boy with left renal mass had bone pain and was detected to have bone metastatic deposits. The most likely renal tumor is:

A. **Wilms' tumor**
B. Renal cell carcinoma
C. Clear cell sarcoma
D. Rhabdoid tumor

Ans A

139.Hepatitis C virus infection is a risk factor for which of the following malignancy?

A. hepatoblastoma
B. **splenic lymphoma**
C. Hodgkin lymphoma
D. nasopharyngeal carcinoma
E. cervical carcinoma

Ans B Hepatitis C virus infection is a risk factor for hepatocellular carcinoma and is also associated with splenic lymphoma.

140.Granulocytic sarcoma masses are commonly seen in which type of the following malignancies?

A. neuroblastoma
B. rhabdomyosarcoma
C. soft tissue sarcoma
E. **acute myeloid leukemia**

Ans E

141. **A healthy 4-year-old child has a left flank mass. Computerized tomography (CT) scan of the abdomen shows a localized left renal mass. Radical nephrectomy is performed; the histology shows clear cell sarcoma of the kidney. Of the following, the MOST appropriate next step in the management is**

A. CT scan of the chest
B. CT scan of the brain
C. **bone scan**
D. magnetic resonance imaging (mri) of the brain
E. skeletal survey

Ans C Bone is the most common site of distant metastasis in clear cell sarcoma of the kidney followed by lung, abdomen, retroperitoneum, brain, and liver.

Tumor Markers & Staging

142. **Most common intra-abdominal solid tumor in children:**

A. Wilms' tumor (Nephroblastoma)
B. **Neuroblastoma**
C. Rhabdomyosarcoma
D. Hodgkin lymphoma

Ans B • Most common intra-abdominal solid tumor in children is Neuroblastoma, followed by Wilms' tumor.

143. **Chang staging is used for:**

A. Retinoblastoma
B. **Medulloblastoma**
C. Ewing's sarcoma
D. Rhabdomyosarcoma

Ans B The Chang staging system is specifically used for medulloblastoma.

144. **Which of the following can present as mass in posterior mediastinum in children?**

A. Rhabdosarcoma
B. **Duplication cyst of esophagus**
C. Lymphoma
D. Neuroblastoma
E. Thymoma 362 Section 3: Systemic Pediatrics

Ans B c. Lymphoma & d. Neuroblastoma Important causes of Mediastinal Mass Superior mediastinum Anterior mediastinum Posterior
• Lymphoma • Thymoma (MC) • Neurogenic
• Thymoma • Teratoma (schwannoma,
• Thyroid lesions • Lymphoma • Lymphoma
• Metastatic carcinoma • Thyroid lesions • Gastroenteric
• Parathyroid tumors • Parathyroid tumors • Duplication mediastinum Middle mediastinum tumors (MC) • Bronchogenic cyst neurofibroma) • Pericardial cyst
• Lymphoma hernia cyst of esophagus 16 Chapter Pediatric Gastrointestinology

145. **A 6-year-old child complains of bilateral thigh pain, motor weakness and some sensory deficits of both lower limbs; he has a history of bladder dysfunction over the past 2 weeks; MRI of the spinal cord shows a mass arising from the filum terminale and conus medullaris causing some pressure effect. A CNS tumor is suspected. Which of the following tumor is MOST likely arising in such site?**

A. medulloblastoma
B. anaplastic astrocytoma
C. **myxopapillary ependymoma**
D. choroid plexus carcinoma
E. oligodendroglioma

Ans C (WHO grade I) is a slow-growing tumor arising from the filum terminale and conus medullaris; it carries good prognosis.

146. **Hemangiomas are the most common benign tumors of infancy, occurring more often in the head and neck region. Of the following, the infant at higher risk of development of hemangioma is**

A. full term infant
B. male gender infant
C. **infant of woman who had chorionic villus sampling**
D. infant delivered by cesarean section
E. infant of diabetic mother

Ans C The risk of hemangioma is 3-5 times higher in girls than boys. The risk is doubled in premature infants and 10 times higher in offspring of women who had chorionic villus sampling.

147. **Nasopharyngeal carcinoma is one of the most common nasopharyngeal tumors in pediatric patients. Many factors may affect the prognosis. Which of the following carries the worst outcome?**

A. elevated lactate dehydrogenase (LDH) level
B. advanced local disease
C. extensive cervical lymph nodes involvement
D. Epstein-Barr virus (EBV) DNA levels
E. **evidence of distant metastases**

Ans E Most pediatric patients present with advanced locoregional disease. LDH is elevated but it is not specific. Epstein-Barr virus DNA levels correlate with disease stage, have prognostic valueand can be used to monitor for recurrence.The outcome depends on the extent of disease; patients with distant metastases have a very poor prognosis.

Oncology Emergencies

148. **A 2-year-old male child presents with a lump in the right side of the abdomen. Ultrasound revealed it to be a solid mass. On examination, his right arm and leg were found to be longer. The most likely diagnosis is:**

A. **Wilms' tumor**
B. Neuroblastoma
C. Angiomyolipoma
D. Nephroblastoma

Ans A • The finding of right arm and leg to be longer suggests hemihypertrophy.
• This is a feature of Beckwith Wiedemann syndrome associated with Wilms' tumor.

149. **The following tumor is not common in the first decade:**

A. **Ameloblastoma**
B. Retinoblastoma
C. Neuroblastoma
D. Rhabdomyosarcoma

Ans A

150. **Which of the following malignant tumors is least likely to occur in adults in comparison with children?**

A. acute Lymphoblastic Leukemia (ALL)
B. osteosarcoma
C. medulloblastoma
D. **retinoblastoma**
E. Hodgkin lymphoma

Ans D is an embryonal tumor, usually occurs in childhood.

151. **Tumor lysis syndrome (TLS) is a common complication during the treatment of pediatric malignancies, which of the following malignancies is more likely to manifest such complication?**

A. acute myelogenous leukemia AML
B. nephroblastoma
C. neuroblastoma
D. **Burkitt lymphoma**
E. Rhabdomyosarcoma

Ans D TLS can occur from rapid cell turnover, which is especially common in Burkitt lymphoma.

152. **You are examining an infant with multiple cutaneous hemangiomas; you suspect an involvement of internal organs. Of the following, the MOST common site of visceral involvement by hemangiomas is**

A. brain
B. heart
C. lung
D. **liver**
E. pancreas

Ans D An ultrasonographic scan or MRI of the liver should be performed if multiple cutaneous lesions are present.

16

CHAPTER 16

Pediatric Gastrointestinology



WHAT THIS CHAPTER COVERS

- Vomiting & GERD
- Pyloric Stenosis
- Intussusception
- Malrotation & Volvulus
- Hirschsprung Disease
- Anorectal Malformations
- Meckel Diverticulum
- Appendicitis
- Constipation & Encopresis
- Diarrhea & Acute Gastroenteritis
- Chronic Diarrhea
- Celiac Disease
- Inflammatory Bowel Disease
- Peptic Ulcer Disease
- Acute Abdomen
- Hernias
- Biliary Atresia
- Pancreatic Disorders

166 MCQs **18** subtopics

16.1 Vomiting & Gastro-oesophageal Reflux

Clue	Think of
Bilious vomiting in a neonate	**Surgical emergency until proved otherwise – malrotation with volvulus** (also duodenal atresia distal to ampulla, jejunoileal atresia, meconium ileus, Hirschsprung)
Non-bilious projectile vomiting at 3–6 weeks, hungry baby	**Hypertrophic pyloric stenosis**
Vomiting + **double bubble**	Duodenal atresia (Down syndrome)
Early-morning vomiting + headache	Raised ICP
Vomiting + polyuria/polydipsia	DKA
Cyclical stereotyped episodes, well between	**Cyclic vomiting syndrome** (migraine equivalent)
Vomiting + seizures/encephalopathy in neonate	**IEM**, CAH (salt-wasting – hyperkalaemia), sepsis
Recurrent vomiting with blood	Mallory–Weiss, oesophagitis, gastritis

- **Physiological GER ("happy spitter")**: peaks at **4 months**, resolves by **12–18 months** in > 90%; thriving baby → **reassurance, smaller frequent feeds, upright after feeds, thickened feeds**; **do not prescribe acid suppressants routinely**.
- **GERD** (complications): FTT, oesophagitis (irritability, feeding refusal), **Sandifer syndrome (torticollis/dystonic posturing)**, aspiration pneumonia, apnoea/ALTE (BRUE), reactive airway disease, dental erosion.
- **Investigations**: clinical diagnosis; **24-h pH-impedance monitoring – gold standard** (quantifies reflux, symptom association); endoscopy + biopsy (oesophagitis, **eosinophilic oesophagitis ≥ 15 eos/HPF**); barium study for anatomy (not GER).
- **Rx**: positioning (**supine for sleep – SIDS prevention**), thickened feeds, 2–4-week trial of **extensively hydrolysed formula (CMPA mimics GERD)**; **PPIs (omeprazole)** for erosive oesophagitis; **fundoplication (Nissen)** for refractory/neurologically impaired.

16.2 Hypertrophic Pyloric Stenosis (HPS)

- **First-born males (4–5:1)**, family history (multifactorial; mother affected → higher risk), **erythromycin/azithromycin in first 2 weeks**, blood group B/O; onset **3–6 weeks** (2–8 weeks).
- **Non-bilious projectile vomiting after feeds**, **hungry after vomiting ("hungry vomiter")**, weight loss, dehydration, constipation; **visible gastric peristalsis (left to right)**; **palpable "olive" in RUQ/epigastrium** (best felt after vomiting, relaxed baby); **jaundice (unconjugated, 2–5% – ↓ UGT from starvation)**.
- **Metabolic: hypochloraemic hypokalaemic metabolic alkalosis with paradoxical aciduria** (late).
- **Investigation of choice: ultrasound** – **pyloric muscle thickness ≥ 3–4 mm, pyloric channel length ≥ 15–16 mm**, diameter ≥ 14 mm; "**target/doughnut sign**". Barium meal (if USG unavailable): **string sign, double-track sign, shoulder sign, beak sign, mushroom sign**.
- **Treatment**: **correct dehydration & electrolytes first (NS + 5% dextrose + KCl) – surgery is NOT an emergency**; then **Ramstedt pyloromyotomy** (open/laparoscopic) – feeds within hours; atropine (non-surgical, less used).

16.3 Intussusception

- **MCC of intestinal obstruction in children 3 months–3 years** (peak **5–9 months**, boys); **ileocolic (MC – 90%)**; idiopathic (**hypertrophied Peyer's patches after viral infection – adenovirus, rotavirus**); **pathological lead**

points (> 2 y) – **Meckel's diverticulum (MC lead point)**, polyp, lymphoma (> 5 y), HSP (ileo-ileal), duplication cyst, CF.

- **Classic triad (only in 1/3)**: **colicky abdominal pain (intermittent screaming, drawing up legs), vomiting (bilious later), "red-currant jelly" stools (late sign – mucosal ischaemia)**; **sausage-shaped mass (RUQ/epigastrium, concave towards umbilicus) with emptiness in right iliac fossa (Dance's sign)**; lethargy between attacks (may mimic encephalitis); rectal exam – blood.
- **Ultrasound – IOC (sensitivity ~100%)**: **"target/doughnut" sign (transverse)**, **"pseudo-kidney" sign (longitudinal)**. Contrast enema – **claw/meniscus sign, coiled-spring appearance**.
- **Treatment**: resuscitation, NG decompression → **non-operative reduction – hydrostatic (saline under USG) or pneumatic (air) enema – success 80–90%** (contraindicated if **peritonitis, perforation, shock**); recurrence ~10% (usually within 72 h); **surgery** (manual reduction – "milking/squeezing" not pulling; resection if gangrenous/lead point) when enema fails or contraindicated.

16.4 Malrotation & Volvulus

- Abnormal rotation of midgut (normal **270° counter-clockwise around SMA**); narrow mesenteric base → **midgut volvulus (clockwise twist)**; **Ladd's bands** cross duodenum → obstruction.
- **Presentation**: **sudden bilious vomiting in first month (75% in first month)**, abdominal distension (may be absent – high obstruction), bloody stools, shock; **gangrene of entire midgut in hours**.
- **Upper GI contrast study – IOC**: **duodenojejunal (DJ) flexure to right of midline/below pylorus; "corkscrew" sign (volvulus)**; **"bird-beak"**; USG – **whirlpool sign**, **inversion of SMA/SMV relationship (SMV left of SMA)**.
- **Treatment: emergency Ladd's procedure** – counter-clockwise detorsion, division of Ladd's bands, broadening mesentery, appendicectomy, placing small bowel on right & colon on left.

16.5 Hirschsprung Disease

- **Congenital aganglionic megacolon**: absence of ganglion cells in **Meissner (submucosal) & Auerbach (myenteric) plexuses** – failure of **cranio-caudal migration of neural crest cells**; **always involves the rectum**, extends proximally (**rectosigmoid – 75–80%**, short segment); total colonic aganglionosis (5%).
- Male : female **4:1**; **Down syndrome (5–10%)**, **RET proto-oncogene (MC)**, EDNRB, MEN-2A, Waardenburg–Shah, congenital central hypoventilation (Haddad), Smith–Lemli–Opitz.
- **Presentation**: **delayed passage of meconium > 48 h (90% of affected)**, abdominal distension, bilious vomiting; **older child – chronic constipation since birth, ribbon-like stools, FTT, distended abdomen, EMPTY rectum on PR exam with "squirt/blast sign" (explosive gush of stool & gas after withdrawal of finger)**; **enterocolitis – MCC death** (fever, explosive foul diarrhoea, distension).
- **Investigations**: **plain X-ray** – distal obstruction; **barium enema – transition zone (narrow aganglionic distal segment → dilated proximal colon), recto-sigmoid index < 1, delayed evacuation (24-h film)** – do **without** prior rectal washout; **anorectal manometry – absent recto-anal inhibitory reflex (RAIR)**; **rectal suction biopsy – GOLD STANDARD (diagnostic)**: **absence of ganglion cells, hypertrophied nerve trunks, ↑ acetylcholinesterase staining, absent calretinin**.
- **Treatment**: rectal washouts/colostomy initially; **definitive pull-through** – **Swenson, Duhamel, Soave (endorectal)**, transanal endorectal pull-through (TERPT).

Feature	Hirschsprung	Functional constipation
Onset	**Since birth** (delayed meconium)	After 2 y (toilet training)
Encopresis/soiling	**Rare**	**Common (overflow)**
Rectum on PR	**Empty, tight**	**Full of faeces (loaded)**
Stool size	Small, ribbon-like	**Large, painful**
FTT, enterocolitis	Common	Rare

16.6 Anorectal Malformations (ARM)

- **Imperforate anus** – boys: **rectourethral (bulbar/prostatic) fistula (MC)**, rectoperineal (low); girls: **rectovestibular fistula (MC)**, rectoperineal; **persistent cloaca** (single perineal opening – girls).
- Associations: **VACTERL (MC)**, Down, Currarino triad (ARM + sacral defect + presacral mass), **tethered cord**, GU anomalies (vesicoureteric reflux).
- Assessment: perineal inspection after **24 h** (meconium in urine/perineum); **invertogram (Wangensteen–Rice) – historic**; **cross-table lateral prone X-ray at 24 h** – distance of rectal gas from perineum **< 1 cm → low (perineal anoplasty)**; **> 1 cm/high → colostomy → posterior sagittal anorectoplasty (PSARP – Peña)** later.

16.7 Meckel's Diverticulum

- **Persistent vitellointestinal (omphalomesenteric) duct**; **true diverticulum (all layers)** on **antimesenteric border of ileum**.
- **Rule of 2s**: **2% of population, 2 feet (60 cm) from ileocaecal valve, 2 inches long, 2 types of ectopic mucosa (gastric – MC, pancreatic), presents before 2 years, 2:1 male**.
- **Presentation**: **painless massive lower GI bleeding (brick-red/maroon) in a child < 2 y – MC presentation (ulcer in adjacent ileal mucosa from acid)**; **intestinal obstruction (MC in adults) – intussusception lead point, volvulus around fibrous band**; **Meckel's diverticulitis (mimics appendicitis)**; Littre hernia.
- **Investigation of choice: 99mTc-pertechnetate scan (Meckel scan)** – taken up by ectopic gastric mucosa; sensitivity ↑ with **pentagastrin, H2 blockers (cimetidine), glucagon**.
- Rx: **diverticulectomy/segmental resection**.

16.8 Appendicitis

- **MC surgical emergency in children** (> 5 y; rare < 2 y – **high perforation rate in young children** due to delayed diagnosis & thin omentum).
- **Periumbilical pain → shifts to RIF (McBurney's point)**, anorexia (most consistent), vomiting **after** pain, low-grade fever; **Rovsing, psoas, obturator signs**; rebound tenderness; **Pediatric Appendicitis Score (PAS) / Alvarado score**.
- **USG – first imaging (non-compressible tubular structure > 6 mm, target sign, appendicolith)**; CT/MRI if equivocal.
- Rx: **appendicectomy (laparoscopic)**; perforated with abscess – antibiotics ± drainage → interval appendicectomy. Non-operative antibiotics for selected uncomplicated cases.

16.9 Constipation & Encopresis

- **Functional constipation (> 95%)** – **Rome IV**: ≥ 2 of – ≤ 2 defecations/week, ≥ 1 episode of faecal incontinence/week (after toilet training), retentive posturing, painful/hard bowel movements, large faecal mass in rectum, large-diameter stools that may obstruct toilet – for **1 month (infants/toddlers) / ≥ 2 months (≥ 4 y)**.
- Triggers: **toilet training, school entry, painful defecation (anal fissure – MC cause of bright-red blood in infants/toddlers)**, cow's milk excess, low fibre.
- **Red flags (organic)**: **delayed meconium > 48 h**, onset < 1 month, FTT, ribbon stools, bilious vomiting, abnormal spine/sacrum (tethered cord), empty rectum, abnormal neurology, hypothyroidism, lead, hypercalcaemia, celiac.
- **Encopresis** (faecal incontinence > 4 y): **retentive (overflow – 80%)**; non-retentive (psychological).
- **Rx**: **disimpaction – oral PEG 1–1.5 g/kg/day × 3–6 days** (or enemas) → **maintenance PEG 0.4–0.8 g/kg/day (DOC)**, lactulose; **behavioural – toilet sitting after meals (gastrocolic reflex), star chart/diary**, fibre & fluids, continue for months after normal pattern.

16.10 Acute Diarrhoea & Gastroenteritis

- **MCC of acute diarrhoea in children < 5 y: rotavirus** (↓ after vaccine; now norovirus in vaccinated populations); bacterial – **E. coli (ETEC MC bacterial cause in developing countries)**, **Shigella (MCC dysentery)**, Campylobacter, Salmonella, V. cholerae.

E. coli type	Mechanism / disease
ETEC	**Heat-labile (LT – cAMP) & heat-stable (ST – cGMP) toxins** – **travellers' diarrhoea**, infant diarrhoea in developing countries
EPEC	Attaching & effacing lesions – **infant diarrhoea (nurseries)**
EHEC (O157:H7)	**Shiga-like toxin** – bloody diarrhoea → **HUS** (avoid antibiotics)
EIEC	Invasive – dysentery (Shigella-like)
EAEC	Aggregative – persistent diarrhoea, HIV

- **Definitions**: **acute diarrhoea < 14 days; persistent ≥ 14 days; chronic > 4 weeks (non-infective)**; **dysentery = visible blood in stool**.
- **Cholera**: **rice-water stools**, no fever, severe dehydration; **Rx doxycycline single dose (≥ 8 y), azithromycin (children)**; hanging-drop (darting motility), TCBS agar.
- **Dysentery Rx (IMNCI/WHO)**: **ciprofloxacin 15 mg/kg BD × 3 days** (WHO) – but in India with resistance, **IAP: ceftriaxone (sick) or azithromycin/cefixime**; amoebic only if trophozoites seen.
- **Management of dehydration**: Plans A, B, C (see Fluids chapter), **low-osmolarity ORS, zinc × 14 days**, continued feeding.
- **Persistent diarrhoea**: often secondary lactose intolerance/malnutrition; Rx nutritional rehabilitation, **reduced-lactose diet (curd, rice-lentil)**, micronutrients, treat infection.
- **Lactose intolerance (secondary)**: **frothy, explosive, acidic stools (pH < 5.5), perianal excoriation, reducing substances +**; **hydrogen breath test** diagnostic; Rx lactose-free/reduced feeds temporarily.

16.11 Chronic Diarrhoea & Malabsorption

Disorder	Key points
Celiac disease	Gluten (gliadin) sensitive enteropathy; **HLA-DQ2 (90%)/DQ8**; onset **6 m–2 y after introducing wheat** (or later – atypical: short stature, anaemia, delayed puberty); **chronic diarrhoea, abdominal distension, wasting of buttocks, irritability, FTT**; **iron-deficiency anaemia refractory to oral iron (MC extraintestinal)**, **dermatitis herpetiformis**, dental enamel defects, osteopenia; associated – **T1DM, Down, Turner, Williams, IgA deficiency, autoimmune thyroiditis**. **Screening: IgA anti-tTG (tissue transglutaminase) + total IgA** (IgG-based tests if IgA deficient); **anti-endomysial antibody** (most specific); **duodenal biopsy (≥ 4 from D2 + 1–2 bulb) – gold standard**: **villous atrophy, crypt hyperplasia, intraepithelial lymphocytes (Marsh–Oberhuber)**; ESPGHAN 2020 – **no biopsy if tTG > 10× ULN + positive EMA in second sample**. Rx **lifelong gluten-free diet (no wheat, barley, rye; oats if uncontaminated)**; rice, maize, millets, potatoes allowed. Untreated → **enteropathy-associated T-cell lymphoma**
Cow's milk protein allergy	Infants; enteropathy, proctocolitis; Rx elimination (EHF)
Cystic fibrosis	Pancreatic insufficiency – steatorrhoea; ↓ faecal elastase
Shwachman–Diamond	Pancreatic insufficiency + neutropenia
Giardiasis	Steatorrhoea, FTT
Intestinal lymphangiectasia	**Protein-losing enteropathy** – hypoalbuminaemia, oedema, **lymphopenia**, hypogammaglobulinaemia, ↑ faecal α1-antitrypsin; Rx **MCT-rich, low-LCT diet**

Disorder	Key points
Abetalipoproteinaemia (MTP gene, AR)	**Acanthocytes**, fat malabsorption, **very low cholesterol/TG, absent apo-B lipoproteins**, retinitis pigmentosa, spinocerebellar degeneration (vitamin E deficiency); Rx vitamin E, low-fat diet
Congenital chloride diarrhoea (SLC26A3)	Polyhydramnios, watery diarrhoea, **metabolic alkalosis**, high stool Cl
Congenital sucrase–isomaltase deficiency	Diarrhoea on introduction of sucrose/fruits
Microvillous inclusion disease / tufting enteropathy	Intractable neonatal secretory diarrhoea – intestinal failure
Inflammatory bowel disease	**Crohn's** (growth failure – often first sign, perianal disease, terminal ileum, skip lesions, granulomas) vs **UC** (bloody diarrhoea, continuous from rectum, pancolitis common in children); **VEO-IBD < 6 y (IL-10R defects)**; **faecal calprotectin** screen; Rx **exclusive enteral nutrition (Crohn's first-line in children)**, steroids, 5-ASA (UC), azathioprine, anti-TNF (infliximab, adalimumab)
Toddler's diarrhoea (functional)	1–3 y, thriving, loose stools with undigested food; excess juice; reassure, ↑ fat, ↓ juice

- **Approach**: osmotic vs secretory – **stool osmotic gap = 290 − 2 (stool Na + K)**: **> 125 mOsm/kg → osmotic (stops on fasting – carbohydrate malabsorption)**; **< 50 → secretory (continues on fasting – congenital chloride/sodium diarrhoea, toxins, VIPoma)**.
- Fat malabsorption: **72-h faecal fat**, Sudan III stain, **D-xylose test** (mucosal vs pancreatic: abnormal in mucosal disease, normal in pancreatic insufficiency).

16.12 GI Bleeding & Other Disorders

Age	Upper GI bleed	Lower GI bleed
Neonate	**Swallowed maternal blood (Apt test – alkali denaturation: fetal Hb resists, stays pink)**, VKDB, stress gastritis	**NEC**, malrotation-volvulus, **allergic proctocolitis**, anal fissure, Hirschsprung enterocolitis
Infant/toddler (1 m–2 y)	Oesophagitis, gastritis, Mallory–Weiss	**Anal fissure (MC)**, **intussusception**, **Meckel's**, infectious colitis, **juvenile polyp**
Preschool (2–5 y)	Varices (**portal vein thrombosis – EHPVO MC in India**), peptic ulcer	**Juvenile polyp (MCC painless rectal bleeding 2–5 y)**, Meckel's, HSP, HUS, infectious
School/ adolescent	**Varices, peptic ulcer (H. pylori, NSAIDs)**	**IBD**, polyps, infectious colitis, haemorrhoids (rare)

- **Juvenile (retention/inflammatory) polyp**: **MC GI polyp in children**, **rectosigmoid**, **hamartomatous, no malignant potential (solitary)**; painless bright-red bleeding, prolapse; Rx colonoscopic polypectomy. **Juvenile polyposis syndrome (≥ 5 polyps; SMAD4/BMPR1A)** – cancer risk.
- **Peutz–Jeghers (STK11, AD)**: **hamartomatous polyps (small bowel) + mucocutaneous pigmentation (lips, buccal)**; intussusception; ↑ cancer risk.
- **Familial adenomatous polyposis (APC)**: hundreds of adenomas, CHRPE, **Gardner (osteomas, desmoids)**, **Turcot (medulloblastoma)**; prophylactic colectomy.
- **Peptic ulcer/H. pylori**: Rx PPI + amoxicillin + clarithromycin/metronidazole × 14 days.
- **Eosinophilic oesophagitis**: food impaction, dysphagia (older), feeding refusal (young); **≥ 15 eosinophils/HPF**, rings/furrows; Rx PPI, swallowed budesonide/fluticasone, elimination diet, dupilumab.
- **Achalasia**: rare in children; **bird-beak** on barium; Rx pneumatic dilatation, Heller myotomy, POEM. **Triple A (Allgrove)**: achalasia, alacrimia, adrenal insufficiency.

- **Foreign body ingestion**: **coin – MC**; oesophageal impaction at **cricopharyngeus (MC)**; **button battery & multiple magnets – emergency removal**; coin in oesophagus seen **en face on AP X-ray** (trachea – sagittal/edge-on).
- **Corrosive ingestion**: **alkali → liquefactive necrosis (oesophagus)**, acid → coagulative (stomach); **no emesis/lavage/neutralisation**; endoscopy in 12–24 h (Zargar grading); late stricture.
- **Recurrent abdominal pain / functional abdominal pain** (Apley): ≥ 3 episodes over 3 months interfering with activities; MC **functional (95%)**; red flags – weight loss, GI bleeding, night waking, pain away from umbilicus, family history of IBD.
- **Infantile colic**: **rule of 3s – crying > 3 h/day, > 3 days/week, > 3 weeks** in a thriving baby (Rome IV: ≥ 3 h/day, ≥ 3 days in any 7 days) – 2 weeks to 4 months; reassurance, soothing, probiotics (L. reuteri – breastfed).

Practice MCQs · Pediatric Gastrointestinology

166 questions · correct option highlighted in green · cover the answer strip to self-test

Vomiting & GERD

1. The Gold standard investigation for Gastro-esophag eal reflux disease is:

A. Endoscopy
B. **24 hour pH recording**
C. Oesophageal manometry
D. Measurement of length of lower oesophageal sphincter

Ans B Extended esophageal pH monitoring of distal esophagus, provides a quantitative sensitive documentation of acidic reflux episodes, the most important type of reflux episodes for pathologic reflux

2. Which of the following is the earliest indicator of patholo gical gastroesophageal reflux in infants (GERD)?

A. **Respiratory symptoms**
B. Postprandial regurgitation
C. Upper GI bleed
D. Stricture esophagus

Ans A • Presence of Respiratory symptoms point towards pathological GERD
• Stricture esophagus and Upper GI bleeding are complications due to severe GERD.

3. A 6-year-old drowsy child came in emergency with history of vomiting, loose motion for 3 days. On examination he had sunken eyes, hypothermia, skin on pinching was taking 5 seconds to revert. What is your diagnosis?

A. Norwalk virus
B. Rotavirus
C. Adenovirus
D. **Cholera**

Ans D Severe dehydration In this case, sunken eyes and skin "goes back very slowly" (in 5 seconds) in a "lethargic" (drowsy) child, so it is a case of severe dehydration.

4. A 4-month-old child has 10 episodes of vomiting and 2–3 episodes of loose stools and crying since the last 24 hours. Best line of management will be:

A. **Intravenous fluids**
B. ORS
C. Breastfeeding only
D. Antibiotics

Ans A Risks associated with severe dehydration that might necessitate intravenous resuscitation include: age <6 mo; prematurity; chronic illness; high fever; bloody diarrhea; persistent emesis; poor urine output; sunken eyes and a depressed level of consciousness. In this case, as the child is < 6 months old and is having persistent emesis, intravenous fluid is indicated.

5. A 9-month-old girl is brought with PR bleed, vomiting, mass in right lumbar region with masked liver dullness. She is in shock like condition. Management should include:

A. Barium enema
B. Saline reduction
C. **Give O**
D. Nasogastric tube
E. IV fluid

Ans C d. Nasogastric tube, e. IV fluid 2
• In patients with prolonged intussusception with signs of shock, peritoneal irritation, intestinal perforation or pneumatosis intestinalis, reduction (with barium, saline or air) should not be attempted
• In such cases, urgent laprotomy should be planned
• But the first priority is urgent resuscitation ie A, B, C (airway, breathing, circulation). Hence in case of shock, and as the child in question is also vomiting, she should be made NPO and Nasogastric tube insertion should

6. The commonest cause of vomiting in a one-month old-Questions infant is:

A. **Pyloric stenosis**
B. Cardiac chalasia
C. Aerophagy
D. Gastro-esophageal reflux

Ans A

7. A newborn with recurrent vomiting, cyanosis after each feed is likely to be suffering from:

A. **Tracheo-esophageal fistula**
B. Tetralogy of Fallot
C. Congenital hypertrophic pyloric stenosis
D. ARDS

Ans A 69. Ans. b. Bad bowel habits Megacolon
• Megacolon is an abnormal dilatation of colon that is not caused by mechanical obstruction
• Two main types of megacolon are: – Congenital megacolon->Hirschsprung disease – Acquired megacolon: Poor bowel habit, Infections (Chagas disease), Idiopathic, antipsychotic drugs.

8. All the following criteria must be met for the consensus definition of cyclical vomiting syndrome EXCEPT

A. at least 5 attacks in any interval
B. recurrent episodes of intense vomiting and nausea lasting 1 hr to 10 days and occurring at least 1 wk apart
C. vomiting during episodes occurs ≥4 times/hr for ≥1 hr
D. return to baseline health between episodes
E. **usually attributed to another disorder**

Ans E Not attributed to another disorder.

9. Gastroesophageal reflux disease (GERD) is the most common esophageal disorder in children of all ages. Of the following, the MOST common symptom in infants is

A. excessive crying
B. **regurgitation**
C. failure to thrive
D. abnormal posturing
E. sleeping disturbances

Ans B Infantile refluxmanifests more often with regurgitation(especially postprandially), signs of esophagitis (irritability, arching, choking, gagging, feeding aversion), and resulting failure to thrive; symptoms resolve spontaneously in the majority of infants by 12-24 mo.

10. The following procedures satisfactorily improve gastroesophageal reflux disease (GERD) symptoms in infants EXCEPT

A. modified feeding volumes
B. hydrolyzed infant formulas
C. prone position, when the infant is awake and observed
D. avoidance of smoke exposure
E. **seated position**

Ans E worsens infant reflux and should be avoided in infants with

11. Erosive esophagitis is found in approximately 12% of children with gastroesophageal reflux disease (GERD) symptoms and is more common in the following EXCEPT

A. **girls**
B. older children
C. neurologically abnormal children
D. children with severe chronic respiratory disease
E. children with hiatal hernia

Ans A More common in boys.

12. Appendicitis-like symptoms (diarrhea, vomiting, fever, and abdominal pain) may be seen in the following foodborne bacterial illness

A. Vibrio vulnificus
B. **Yersinia enterocolitica**
C. Shigella spp.
D. Staphylococcus aureus (preformed enterotoxin)
E. Listeria monocytogenes

Ans B Signs and symptoms of Yersinia enterocolitica and Yersinia pseudotuberculosis are, appendicitis-like symptoms (diarrhea and vomiting, fever, abdominal pain) occur primarily in older children and young adults; might have a scarlatiniform rash or erythema nodosum with Y.Pseudotuberculosis.

13. Ondansetron is an effective and less-toxic antiemetic agent and can be given as a useful adjunct to the treatment of vomiting in ambulatory settings, it is usually given

A. intravenously
B. intramuscularly
C. subcutaneously
D. **sublingually**
E. orally

Ans D Because persistent vomiting can limit oral rehydration therapy, a single sublingual dose of an oral dissolvable tablet of ondansetron (4 mg 4-11 yr and 8 mg for children older than 11 yr [generally 0.2 mg/kg]) may be given.

Pyloric Stenosis

14. Which one of the following life-threatening congen ital anomalies in the newborn presents with polyhydram nios, aspiration pneumonia, excessive salivation and difficulty in passing a nasogastric tube?

A. Gastroschisis
B. Diaphragmatic hernia
C. **Tracheo-esophageal fistula**
D. Choanal atresia

Ans C • Presence of polyhydramnios, aspiration pneumonia, excessive salivation and difficulty in passing a nasogastric tube all point towards a diagnosis of tracheoesophageal fistula, which is usually accompanied by esophageal atresia. Esophageal atresia (EA) and TEF (Tracheoesophageal fistula): Important • EA is the most frequent congenital anomaly of esophagus; 90% of EA babies have TEF features • Most common type is upper esophagus ends in a blind pouch and TEF connected to distal esophagus

15. Low osmolar ORS composition?

A. Na 90 + 311 mOsmol
B. **Na 75 + 245 mOsmol**
C. Na 60 + 245 mOsmol
D. Na 60 + 240 mOsmol

Ans B

16. False statement on FAP:

A. Most common genetic polyposis syndrome
B. >100 polyps may be present in colon
C. Risk of developing colon cancer is 90–100%
D. **Colon cancer usually develops around 20 years of age**

Ans D Familial adenomatous polyposis (FAP)
• It is the most common genetic polyposis syndrome
• Characterized by numerous adenomatous polyps (100s to 1000s) throughout colon; stomach & duodenum may be involved
• Extraintestinal manifestations include hypertrophy of retinal pigment epithelium, desmoid tumors, epidermoid cysts, osteomas, fibromas, and lipomas.
• Caused by mutations in APC gene, a tumor suppressor gene (chr 5q21).
• Polyps generally develop in adolescence (mean age of presentation is 16 year).

17. Double bubble sign in children is seen in all except:

A. Ladds band
B. Annular pancreas
C. Pancreatic pseudocyst
D. **Diaphragmatic hernia**
E. Duodenal atresia

Ans D • Obstruction of 2nd part of duodenum cause 'double bubble' sign on plain abdominal radiograph
• It may be due to- Annular pancreas, Pancreatic pseudocyst, Ladds band a Tumor in head of pancreas.

18. One of the following cause osmotic diarrhea

A. toxigenic Escherichiacoli
B. carcinoid
C. neuroblastoma
D. **glucose-galactose malabsorption**
E. congenital chloride diarrhea

Ans D Lactase deficiency, glucose-galactose malabsorption,lactulose, and laxative abuse are causes of osmotic diarrhea.

19. Hypertrophic pyloric stenosis has been associated with the following EXCEPT

A. eosinophilic gastroenteritis
B. Apert syndrome
C. Zellweger syndrome
D. **trisomy**
E. Cornelia de Lange syndrome

Ans D Trisomy 18.

20. Oral and intravenous atropine sulfate (pyloric muscle relaxant) has been described when surgical treatment is not available for hypertrophic pyloric stenosis with a success rate of

A. 20%
B. 40%
C. 60%
D. **80%**
E. 90%

Ans D

21. The risk of colon cancer is 100% in the following condition

A. Peutz-Jeghers syndrome
B. Cowden syndrome
C. **Gardner syndrome**
D. Juvenile polyposis
E. Bannayan-Riley- Ruvalcaba syndrome

Ans C 100% in Familial adenomatous polyposis (FAP) and Gardner syndrome.

22. Regarding epidemiology of HBV, the following are true EXCEPT

A. intrauterine infection occurs in 2.5%
B. antigenemia appear 1-3 mo after birth
C. HBsAg is inconsistently recovered in human milk of infected mothers
D. **breastfeeding of nonimmunized infants by infected mothers confer a greater risk of hepatitis**
E. HBV has 8 genotypes (A-H)

Ans D Breastfeeding of nonimmunized infants by infected mothers does not confer a greater risk of hepatitis than does formula feeding.

Intussusception

23. A coin is loaded in the esophagus of an asymptomatic 3 years female child, Treatment is:

A. **Endoscopic removal within 24 hours**
B. Immediate endoscopic removal
C. Wait for 48 hours
D. Dislodge the coin to the stomach by inserting Ryle's tube

Ans A Asymptomatic blunt objects and coins can be observed for up to 24 hr anticipating passage into stomach; If they still remain lodged in the esophagus, endoscopic removal is warranted.

24. All of the following are true about Peutz-Jeghers syndrome except:

A. **It is the most common genetic polyposis syndrome**
B. Oral mucocutaneous pigmentation seen
C. Polyps most commonly involve small intestine
D. Autosomal dominant inheritance

Ans A Familial adenomatous polyposis is the most common genetic polyposis syndrome.

25. Which one of the following is most suggestive of neonatal small bowel obstruction:

A. Generalized abdominal distension
B. Failure to pass meconium in the first 24 hours
C. **Bilious vomiting**
D. Refusal of feeds

Ans C Bilious vomiting is the most common symptom of Intestinal obstruction in newborn; Presentation depends on the site of obstruction:
• Proximal obstruction weiveR – A history of polyhydramnios is common; Abdominal distension and constipation will be less prominent – The higher the obstruction in the intestine the earlier the infant will develop vomiting which may be bile stained
• Distal obstruction, Abdominal distension and constipation are more common, while vomiting is less common. Note: In

26. Advanced dentition for age and sex is seen in

A. nutritional disturbances
B. hypopituitarism
C. cleidocranial dysplasia
D. **hyperthyroidism**
E. trisomy

Ans D Delayed eruption of the 20 primary teeth can be familial or indicate systemic or nutritional disturbances such as hypopituitarism, hypothyroidism, cleidocranial dysplasia, trisomy 21, and multiple syndromes.

27. The classic triad of intussusception, (pain, a palpable sausage-shaped abdominal mass, and bloody or currant jelly stool) is seen in

A. ≤20% of patients
B. **≤30% of patients**
C. ≤40% of patients
D. ≤50% of patients
E. ≤60% of patients

Ans B The classic triad of pain, a palpable sausage-shapedabdominal mass, and bloody or currant jelly stool is seen in <30% of patients with intussusception.

28. Direct person-to-person contact outbreaks of gastroenteritis are usually caused by

A. Shigella
B. Salmonella
C. Rotavirus
D. Giardia
E. Clostridium difficile

Ans A Direct person-to-person contact outbreaks of gastroenteritis are usually caused by norovirus and Shigella species. Unknown agents are seen in 30-40%; other pathogens include Salmonella, rotavirus, Giardia, Cryptosporidium, Clostridium difficile, and C. jejuni.

29. Hamartomatous polyps, which represent the most common intestinal tumors of childhood have the following features EXCEPT

A. occurring in 1-2% of children
B. present in the 1st decade
C. most often at ages 2-5 yr
D. most commonly in the colon
E. often multiple

Ans E Polyps may be found anywhere in the gastrointestinal (GI) tract, most commonly in the colon or rectum; they are often solitary but may be multiple.

30. Routine screening for HBV infection requires assay of multiple serologic markers, all the following are true EXCEPT

A. HBsAg is the first serologic marker of infection to appear
B. Anti-HBc is a valuable serologic marker of acute HBV infection
C. anti-HBs and anti- HBc are detected in persons immunized with hepatitis B vaccine
D. HBeAg is a marker of infectivity
E. HBsAg levels fall before symptoms wane

Ans C Only anti-HBs is present in persons immunized with hepatitis B vaccine, whereas both anti-HBs and anti- HBc are detected in persons with resolved infection.

Malrotation & Volvulus

31. The commonest type of Congenital atresia is?

A. Proximal end blind, distal end communicating with trachea
B. Distal end blind, proximal end communicating with trachea
C. Proximal and distal ends open and communicating with trachea
D. Both ends blind

Ans A

32. Enzyme deficiency that occurs after viral gastroenter itis:

A. Lipase
B. Amylase
C. Lactase
D. Enterokinase

Ans C Post infectious diarrhea Definition Chronic diarrhea following infectious enteritis, regardless of the nature of the pathogen Epidemiology Usually seen in infants and very young toddlers Pathogenesis May be related to secondary lactase deficiency, food protein allergy, antibiotic associated colitis (including pseudo- membranous colitis caused by Clostridium difficile toxin), or a combination of these. Treatment Supportive; may include a lactose free diet in the presence of secondary lactase deficiency

33. A mother comes with her 3 month child asking the physician, if she can give cereals to her child. What problem can this lead to her child?

A. Allergy due to the food content
B. Retarded oralmotor development
C. Contaminated food leading to reflux
D. Risk of gastrointestinal infection

Ans A Nelson latest Ed says "Exclusive breastfeeding for the first 4-6 months of life may reduce allergic disorders in first few years of life. Potentially allergenic foods (eggs, milk, wheat, soy, peanut and fish) should be introduced after this period of exclusive breastfeeding to decrease chances of food allergy;" Hence, Increased food allergy is the best answer.

34. A 6-month-old baby presents with history of bloody diarrhea of 2 days duration with abdominal distension and on examination the baby screams, diagnosis is:

A. Chronic appendicitis
B. Chronic pancreatitis
C. Crohn's disease
D. Bulimia 380 Section 3: Systemic Pediatrics

Ans A Intussusception Discussing the options one by one: a. Intussusception The typical age group with short history of bloody diarrhea and extreme tenderness on examination point towards intussusception b. HUS In HUS, the typical clinical picture is of sudden onset pallor, oliguria, edema, petechiae and lethargy; Usually there is history of severe gastroenteritis with blood stained stools but no severe pain abdomen; c. Appendicitis Typically presents with right lower quadrant pain with/ without gastroenteritis

35. Mottled enamel (permanent teeth) may be seen in

A. renal failure
B. immunosuppression
C. neutrophil chemotactic deficiency
D. maternal infections
E. uncontrolled juvenile diabetes

Ans A Mottled enamel (permanent teeth) is seen in kidney failure and cystic fibrosis.

36. The following infections may cause malabsorption in immunocompromised children

A. Shigella
B. Salmonella
C. enteropathogenic
D. Giardia
E. rotavirus

Ans C Malabsorption is mainly seen after infection with Campylobacter, Shigella, Salmonella, Giardia, cryptosporidium, coccidioidosis, and rotavirus. These infectious causes of malabsorption are more common in immunocompromisedchildren.

37. Guillain-Barré syndrome can be a sequel in the following foodborne bacterial illness

A. Bacillus anthracis
B. Bacillus cereus
C. Brucella abortus
D. **Campylobacter jejuni**
E. Clostridium botulinum

Ans D Treatment of Campylobacter jejuni is supportive care.For severe cases, antibiotics, such as azithromycin and quinolones, may be indicated early in the diarrheal diseaseGuillain-Barré syndrome can be a sequel.

38. The features of the Johanson-Blizzard syndrome include the following EXCEPT

A. exocrine pancreatic deficiency
B. hypoplasia of the alae nasi
C. congenital deafness
D. **hyperthyroidism**
E. ectodermal scalp defects

Ans D The features of the Johanson-Blizzard syndrome include exocrine pancreatic deficiency, aplasia or hypoplasia of the alae nasi, congenital deafness, hypothyroidism, developmental delay, short stature, ectodermal scalp defects, and absence of permanent teeth, urogenital malformations, and imperforate anus.

39. HBV is usually spread by

A. breastfeeding
B. kissing
C. hugging
D. sharing utensils
E. **sharing needles**

Ans E HBV is not spread by breastfeeding, kissing, hugging, orsharing water or utensils.

Hirschsprung Disease

40. Gasless abdomen on X-ray is seen in what type of trachea-esophageal fistula

A. Isolated TEF
B. **EA with proximal TEF**
C. EA with distal TEF
D. EA with double TEF

Ans B • In EA with proximal TEF, as there is no connection of the distal esophagus with the trachea or proximal esophagus, gasless abdomen will be seen on X-ray.

41. Profuse watery diarrhea in an immuno-compromised child is due to:

A. **Cryptosporidium**
B. Ameba
C. Giardia
D. Lactose intolerance

Ans A Opportunistic microorganisms induce diarrhea exclusively, more severely or for more prolonged periods, in specific opulations, such as immunocompromised children. Cryptosporidium induces severe and protracted diarrhea in AIDS patients.

42. Carbohydrates malabsorption is detected by:

A. **Breath hydrogen test**
B. Nitrogen breath test
C. Fecal nitrogen
D. Rapid urease test

Ans A Breath hydrogen test
• It is used to identify the specific carbohydrate (lactose, sucrose, fructose, or glucose) that is malabsorbed.
• After an overnight fast, the suspected sugar is administered as an oral solution (carbohydrate 1-2 g/kg, maximum 50 g).
• In malabsorption the sugar is not digested in the small bowel, passes on to colon & is metabolized by colonic bacteria flora
• Note: child should not be on antibiotics at the time of test, because colonic flora is essential for fermenting the sugar

43. All the following are causes oropharyngeal dysphagia EXCEPT

A. cerebral palsy
B. **esophagitis**
C. brain tumors
D. cerebrovascular accidents
E. hyperthyroidism

Ans B is a cause of esophageal dysphagia.

44. Pierre Robin syndrome consists of the following EXCEPT

A. micrognathia
B. high arched palate
C. cleft palate
D. **small tongue**
E. foreshortened floor of the mouth

Ans D The tongue is usually of normal size, but the floor of the mouth is foreshortened. The air passages can become obstructed, particularly on inspiration, usually requiring treatment to prevent suffocation.

45. Hirschsprung disease has been seen in association with the following EXCEPT

A. microcephaly
B. mental retardation
C. autism
D. **cleft lip**
E. hydrocephalus

Ans D Cleft palate

46. Antibiotics are not indicated and may be detrimental in the following foodborne bacterial illness

A. Enterotoxigenic
B. **Enterohemorrhagic Escherichia coli O157:H7**
C. Campylobacter jejuni
D. Brucella abortus
E. Bacillus anthracis

Ans B Treatment of Enterohemorrhagic Escherichia coli(EHEC) E. coli O157:H7 include supportive care, monitor renalfunction, hemoglobin, and platelets closely. E. coli O157:H7infection is also associated with hemolytic uremic syndrome(HUS), which can cause lifelong complications. Studies indicate that antibiotics might promote the development of HUS. Antidiarrheal agents like Imodium may also increase the risk of developing HUS.

47. For patients with advanced liver disease, hepatic transplantation has a success rate >85%. If the operation is technically feasible, it will prolong life and might correct the metabolic error in diseases such as

A. tyrosinemia
B. galactosemia
C. hereditary fructose intolerance
D. Zellweger syndrome
E. Crigler-Najjar

Ans A It will prolong life and might correct the metabolic error in diseases such as α1- antitrypsin deficiency, tyrosinemia, and Wilson disease.

48. Total parenteral nutrition (TPN) can cause a variety of liver diseases. Of the following, the MOST severe complication is

A. hepatic steatosis
B. gallbladder damage
C. bile duct damage
D. cholestasis
E. sclerosing cholangitis

Ans D is the most severe complication and can lead to progressive fibrosis and cirrhosis. It is the major factor limiting effective long-term use of TPN in children and adults. Risk factors for TPN-associated cholestasis include prolonged duration of TPN, prematurity, low birthweight, sepsis, necrotizing enterocolitis, and short bowel syndrome.

Anorectal Malformations

49. A one-day-old baby presents with excessive frothing from mouth and baby was cyanosed:

A. Esophageal atresia
B. Diaphragmatic hernia
C. Congenital lung cyst
D. Lung hypoplasia

Ans A Congenital disease Mode of presentation Cong diaphragmatic hernia New born with cyanosis, respiratory distress, shift of mediastinum and scaphoid abdomen Bilateral choanal atresia New born with feeding dificulty, cyanosis on closing mouth, which disappears on crying Pure esophageal atresia Neonate with frothing and bubbling at mouth and nose, with coughing, cyanosis and respiratory distress; X-ray shows coiled feeding tube in esophageal pouch and absent gastric air shadow

50. In a 12-month-old child with Diarrhea, the dose of zinc is:

A. 1 mg/day for 10–14 days
B. 10 mg/day for 10–14 days
C. 15 mg/day for 10–14 days
D. 20 mg/day for 10–14 days

Ans D Dose of Zinc for children older than 6 months with diarrhea is 20 mg/day for 10–14 days.

51. All the following are True regarding Familial Adenomatous Polyposis except?

A. > 100 colorectal polyps are present
B. Congenital Hypertrophy of Retinal Pigment Epithelium is seen in up to 50% patients
C. It is due to a mutation of APC gene on chromosome
D. It is an Autosomal Dominant disorder

Ans C It is due to a mutation of APC gene in chromosome 15 General Features of Inherited Colorectal Cancer Syndromes Polyp Polyp Age of Risk of Genetic Syndrome histology distribution onset colon cancer lesion Clinical manifestations Familial Adenoma Large intestine 16 years 100% 5q (APC gene) Rectal bleeding, abdominal pain, adenomatous (often >100) (range 8–34 years) bowel obstruction polyposis Gardner Adenoma Large and small 16 year (range 100% 5q (APC gene) Rectal bleeding, abdominal pain,

52. All the following are common causes of emesis in childhood EXCEPT

A. gastroenteritis
B. toxic ingestion
C. pertussis syndrome
D. peptic ulcer
E. otitis media

Ans D is a rare cause of emesis in childhood.

53. The following are features of achalasia EXCEPT

A. dysphagia for solids and liquids
B. may be accompanied by undernutrition
C. may be accompanied by chronic cough
D. may be misdiagnosed as asthma
E. usually diagnosed before school age

Ans E It is uncommon before school age.

54. Vitamin B12 and bile salts are only absorbed in

A. distal ileum
B. duodenum
C. proximal jejunum
D. colon
E. proximal ileum

Ans A Vitamin B12 and bile salts are only absorbed in the distal ileum.

55. The first-line treatment for children with Vibrio cholera is

A. azithromycin
B. ciprofloxacin
C. doxycycline
D. ampicillin
E. TMP-SMX

Ans A Treatment of Vibrio cholerae is supportive care with aggressive oral and intravenous rehydration.Doxycycline is recommended as first-line treatment for adults, whereas azithromycin is recommended as first-line treatment for children and pregnant women. Ciprofloxacin and doxycycline recommended as second-line drugs for children.

56. There are many anomalies associated with anorectal malformations. Of the following, the MOST common are

A. kidney anomalies
B. cardiac anomalies
C. esophageal atresia
D. spina bifida
E. tethered cord

Ans A There are many anomalies associated with anorectal malformations. The most common are anomalies of the kidneys and urinary tract in conjunction with abnormalities of the sacrum. This complex is often referred to as the caudal regression syndrome.

57. Autoimmune hepatitis is a clinical diagnosis based on certain diagnostic criteria; no single test will make this diagnosis. Important positive features include the following

A. female gender
B. primary elevation in alkaline phosphatase
C. elevated γ-globulin levels
D. presence of autoantibodies
E. characteristic histologic findings

Ans B Primary elevation in transaminases and not alkaline phosphatase.

Meckel Diverticulum

58. Esophageal atresia may occur as a part of VACTER group of anomalies. What does 'TE' stand for?

A. Tetralogy of Fallot
B. Thoracic empyema
C. Tracheo-esophageal fistula
D. Talipes equinovarus

Ans C • VACTERL association (also called VATER association) refers to the non-random co-occurrence of birth defects including Vertebral anomalies, Anal atresia, Cardiac defects, Tracheoesophageal fistula and/or Esophageal atresia, Renal and Radial anomalies and Limb defects.

59. The most common bacterial cause for diarrhea in Questions children in India is:

A. Enterotoxigenic E. coli (ETEC)
B. Enteropathogenic E. coli (EPEC)
C. Enterohemorrhagic E. coli (EHEC)
D. V. cholerae

Ans A • E. coli is the most common bacteria causing diarrhea in children in developing countries
• Rotavirus is the most common viral agent causing diarrhea
• Shigella, ETEC and EHEC can cause dysentery; EHEC is a cause of hemolytic uremic syndrome.

60. A preterm baby on 30 mL/kg of enteral feeding developed sudden severe abdominal distension with visible bowel loops on day 6 of life. He also showed temperature instability and lethargy. X-ray of the abdomen showed portal venous gas. The staging of NEC is:

A. Lower GI study
B. Manometry
C. Sweat Chloride levels
D. CFTR mutation analysis

Ans C IIb with Stage of NEC Radiological feature I Normal/mild intestinal dilatation IIa Pneumatosis intestinalis IIb Portal vein gas IIIa Peritonitis/Ascites IIIb Pneumoperitoneum

61. Secretory diarrhea can be caused by

A. neuroblastoma
B. laxative abuse
C. lactase deficiency
D. irritable bowel syndrome
E. thyrotoxicosis

Ans A Laxative abuse and lactase deficiency are causes of osmotic diarrhea while irritable bowel syndrome and thyrotoxicosis are caused by increased bowel motility.

62. Meckel diverticulum has been conveniently referred to by the "rule of 2s," which explains the classic presentation of this congenital anomaly. One of the following is FALSE in this rule

A. found in approximately 2% of the general population
B. usually located 2 feet proximal to the ileocecal valve
C. approximately 2 inches in length
D. contain 2 types of ectopic tissue (pancreatic or gastric)
E. found twice as commonly in males

Ans E Meckel diverticulum is found twice as commonly in females.

63. Confirmation of a Meckel diverticulum can be difficult. Of the following, the MOST sensitive study is

A. plain abdominal radiographs
B. barium studies
C. radionuclide technetium-99m scans
D. abdominal ultrasound
E. abdominal CT scan

Ans C The most sensitive study is a Meckel radionuclide scan, which is performed after intravenous infusion of technetium-99m pertechnetate. The mucus-secreting cells of the ectopic gastric mucosa take up pertechnetate, permitting visualizationof the Meckel diverticulum. The uptake can be enhancedwith various agents, including cimetidine, ranitidine, glucagon, and pentagastrin. The sensitivity of the enhanced scan is approximately 85%, with a specificity of approximately 95%.

64. Persistence of unconjugated hyperbilirubinemia at levels >20 mg/dL after the 1st wk of life in the absence of hemolysis should suggest

A. galactosemia
B. tyrosinemia
C. α1-Antitrypsin deficiency
D. Crigler-Najjar type I
E. Maple serum urine disease

Ans D The diagnosis of CN type I is based on the early age of onset and the extreme level of bilirubin elevation in the absence of hemolysis. In the bile, bilirubin concentration is <10 mg/dL compared with normal concentrations of 50-100 mg/dL; there is no bilirubin glucuronide. Definitive diagnosis is established by measuring hepatic glucuronyl transferaseactivity in a liver specimen obtained by a closed biopsy.

65. **The gallbladder is congenitally absent in approximately 0.1% of the population. Hypoplasia or absence of the gallbladder can be associated with**

A. biliary dyskinesia
B. **cystic fibrosis**
C. cirrhosis
D. cholestasis
E. Wilson disease

Ans B Hypoplasia or absence of the gallbladder can be associated with extrahepatic biliary atresia or cystic fibrosis.

Appendicitis

66. **A newborn baby had normal APGAR score at birth and developed excessive frothing and choking on attempted feeds. The investigation of choice is:**

A. Esophagoscopy
B. Bronchoscopy
C. MRI chest
D. **X-ray chest and abdomen with the red rubber catheter passed per orally into esophagus**

Ans D

67. **Most common bacteria causing diarrhea in children in India is:**

A. **Enterotoxigenic E. coli**
B. Enteroinvasive E. coli
C. Enterohemorrhagie E. coli
D. Enteropathogenic E. coli

Ans A

68. **True about Peutz-Jegher's syndrome:**

A. **Pigmentary changes in skin and mucous membrane around mouth**
B. Adenomatous polyp in intestine
C. Most common of pattern inheritance is autosomal recessive
D. 20-30% premalignant
E. May presents as anemia in children

Ans A e May present as anemia in children 20/e p 1902, 3134 Peutz Jegher Syndrome What is it? Characterized by mucocutaneous pigmentation and extensive GI hamartomatous polyposis Epidemiology Median age of 11 years; GI cancer in 2-3% of patients Genetics Autosomal dominant; Heterozygous loss-of-function of STK11Q Site Jejunum is the most common site Symptoms • Intussusception; Hematochezia can be present in 14% cases – can present as anemia

69. **All the following suggest a potentially serious organic etiology of abdominal pain**

A. **age >5 yr**
B. fever
C. flank pain
D. awakening from sleep
E. referred pain to shoulder

Ans A Age less than 5 year may suggest serious organic etiology.

70. **In healthy full-term infants, meconium is passed within 48 hr of birth in**

A. **99%**
B. 90%
C. 80%
D. 70%
E. 60%

Ans A In 99% of healthy full-term infants, meconium is passed within 48 hr of birth.

71. **Management of the diarrhea in chronically malnourished children is based on**

A. intravenous therapy
B. standard osmolarity oral rehydration solutions
C. **reduced osmolarity oral rehydration solutions**
D. slow resumption of feeds
E. nothing by mouth for 24 hour

Ans C Management of the diarrhea in chronically malnourished children is based on 3 principles: oral rehydration to correct dehydration, rapid resumption of feeds with avoidance of periods of nothing by mouth, and treating the etiology of the diarrhea.

72. **The low-osmolality World Health Organization (WHO) oral rehydration solution (ORS) has the following per liter EXCEPT**

A. 75 mEq of sodium
B. **50 mEq of chloride**
C. 20 mEq of potassium
D. 75 mmol of glucose
E. total osmolarity of 245 mOsm

Ans B The low-osmolality World Health Organization (WHO) oral rehydration solution (ORS) containing 75 mEq of sodium, 64 mEq of chloride, 20 mEq of potassium, and 75 mmol of glucose per liter, with total osmolarity of 245 mOsm/L, isnow the global standard of care and more effective than home fluids, including decarbonated soda beverages, fruit juices, and tea. These are not suitable for rehydration or maintenance therapy because they have inappropriately high osmolalities and low sodium concentrations.

73. **A variety of laboratory tests have been used in the evaluation of children with suspected appendicitis. Of the following, the MOST sensitive and specific one is**

A. leukocyte count
B. urinalysis
C. electrolytes
D. C-reactive protein
E. **serum amyloid A protein**

Ans E is consistently elevated in patients with acute appendicitis with a sensitivity and specificity of 86% and 83%, respectively.

74. **Plain abdominal radiographs may be helpful in selected cases of abdominal pain/suspected appendicitis. Plain abdominal x-rays can demonstrate the following findings in acute appendicitis EXCEPT**

A. sentinel loops of bowel and localized ileus
B. scoliosis from psoas muscle spasm
C. a colonic air–fluid level above the right iliac fossa
D. a RLQ soft-tissue mass
E. **a calcified appendicolith (50% of cases)**

Ans E A calcified appendicolith (5-10% of cases),

75. Findings that suggest advanced appendicitis on ultrasound include the following

A. asymmetric wall thickening
B. **increased local tenderness to compression**
C. abscess formation
D. associated free intraperitoneal fluid
E. surrounding tissue edema

Ans B Decreased local tenderness to compression.

76. The clinical presentation of one of the following closely mimics appendicitis

A. **Meckel diverticulitis**
B. mesenteric adenitis
C. sickle cell disease
D. pancreatitis
E. ovarian torsion

Ans A is an infrequent condition, butthe clinical presentation closely mimics appendicitis and the diagnosis is usually made at surgery.

Constipation & Encopresis

77. A newborn has dribbling after feeds. He has respiratory distress and froths at the mouth. Diagnosis is:

A. **Tracheoesophageal fistula**
B. Tetralogy of Fallot
C. Respiratory distress syndrome
D. None of the above DISORDERS OF INTESTINE: DIARRHEA

Ans A DISORDERS

78. Chronic constipation in children is seen in all except:

A. Hirschsprung disease
B. **Jejunal polyp**
C. Hypothyroidism
D. Stricture

Ans B Important Causes of constipation in children: Nonorganic (Functional)—Retentive Psychiatric—Anorexia nervosa Anatomic Abnormal Musculature
• Anal stenosis, atresia with fistula • Prune-Belly syndrome
• Imperforate anus and other malformations • Gastroschisis
• Intestinal stricture • Down syndrome Intestinal Nerve or Muscle Abnormalities Drugs
• Hirschsprung disease • Anticholinergics, Narcotics, Lead
• Spinal cord defects including Tethered cord • Chemotherapeutic agents (vincristine)

79. A neonate is suspected to be suffering from necrotizing enterocolitis (NEC). On further examination and investigation, he is diagnosed to be Bell's stage I NEC. The management of choice would be:

A. Laparotomy and proceed
B. Insertion of bilateral pelvic drains
C. **Conservative management with IV fluids and antibiotics**
D. Initial conservative management and laparotomy after 24 hours

Ans C

80. Organic causes of constipation in infant are all except:

A. **High fibre diet**
B. Cystic fibrosis
C. Hypothyroidism
D. Hyperthyroidism

Ans A

81. All the following metabolic disorders can cause constipation EXCEPT

A. hypercalcemia
B. **hyperkalemia**
C. hypothyroidism
D. diabetes mellitus
E. diabetes insipidus

Ans B Hypokalemia causes constipation.

82. In the blue diaper syndrome, symptoms can include all the following EXCEPT

A. vomiting
B. **diarrhea**
C. failure to thrive
D. nephrocalcinosis
E. ocular abnormalities

Ans B Symptoms can include digestive disturbances such as vomiting, constipation, poor appetite, failure to thrive, hypercalcemia, nephrocalcinosis, fever, irritability, and ocular abnormalities.

83. Immunoglobulin A (IgA) nephropathy is an extraintestinal manifestation of the following enteric infection

A. Salmonella
B. Shigella
C. Yersinia
D. **Campylobacter**
E. Cryptosporidium

Ans D Characterized by recurrent episodes of blood in the urine, this condition results from deposits of the protein IgA in the glomeruli. IgA nephropathy can progress for years with no noticeable symptoms. Men seem more likely to develop this disorder than women.

84. Liver histology demonstrates normal architecture, but hepatocytes contain black pigment similar to melanin in

A. **Dubin-Johnson syndrome**
B. Rotor syndrome
C. Gilbert syndrome
D. Crigler-Najjar syndrome
E. Zellweger syndrome

Ans A Liver histology demonstrates normal architecture, but hepatocytes contain black pigment similar to melanin. Liver function is normal and prognosis is excellent. The most commonly reported symptoms are abdominal pain and fatigue, jaundice, dark urine, and slight enlargement of the liver. Jaundice fluctuates in intensity and is aggravated by intercurrent disease.

85. Biliary sludge or cholelithiasis can be detected in >40% of children who are treated with prolonged course of

A. cefotaxime
B. meropenem
C. vancomycin
D. erythromycin
E. **ceftriaxone**

Ans E Prolonged use of high-dose ceftriaxone, a third-generation cephalosporin, has been associated with the formation of calcium-ceftriaxone salt precipitates (biliary pseudolithiasis) in the gallbladder. Biliary sludge or cholelithiasis can be detected in >40% of children who are treated with ceftriaxone for at least 10 days. In rare cases, children become jaundiced and develop abdominal pain; precipitates usually resolve spontaneously within several months after discontinuation of the drug.

Diarrhea & Acute Gastroenteritis

86. Low osmolarity ORS has (as compared to WHO-ORS):

A. Low glucose and high sodium
B. **Low glucose and low sodium**
C. Low glucose and high potassium

Ans B Composition of oral rehydration solutions Composition Reduced-osmolarity ORS Standard WHO-ORS Glucose (mmol/L) 75 111 Sodium (mmol/L) 75 90 Potassium (mmol/L) 20 20 with Chloride (mmol/L) 65 80 Citrate (mmol/L) 10 10 Osmolarity (mmol/L) 245

87. A child presents with acute diarrhea with signs of dehydration; On investigation, Na+ 125 mEq/L, K+ 3 mEq/L, HCO3- 16 mEq/L and pH is 7.23. IV Fluid to be given is?

A. **Normal saline**
B. 3% saline
C. N/3 saline + 10% dextrose
D. N/3 saline + 5% dextrose

Ans A • Hyponatremic dehydration is treated by correction of intravascular volume with isotonic fluid (NS or LR).
• An overly rapid (>12 mEq/L over the first 24 hr) or overcorrection of serum sodium concentration (>135 mEq/L) is associated with an increased risk of central pontine myelinolysis.

88. A 6-month-old infant presents to the diarrhea clinic unit with some dehydration. The most likely organism causing diarrhea is:

A. E. histolytica
B. **Rotavirus**
C. Giardia lamblia
D. Shigella

Ans B

89. Acute diarrhea in infancy is commonly caused by

A. primary disaccharidase deficiency
B. **overfeeding**
C. Hirschsprung toxic colitis
D. adrenogenital syndrome
E. neonatal opiate withdrawal

Ans B Rare causes of acute diarrhea in infancy include 🅱️ Primary disaccharidase deficiency 🅱️ Hirschsprung toxic colitis 🅱️ Adrenogenital syndrome 🅱️ Neonatal opiate withdrawal

90. Risks associated with severe dehydration that might necessitate intravenous resuscitation include

A. age < 9 mo
B. postmaturity
C. fever >38°C if 3-36 mo of age
D. secretary diarrhea
E. **depressed level of consciousness**

Ans E Risks associated with severe dehydration that might necessitate intravenous resuscitation include: age <6 mo; prematurity; chronic illness; fever >38°C (100.4°F) if younger than 3 mo or >39°C (102.2°F) if 3-36 mo of age; bloody diarrhea; persistent emesis; poor urine output; sunken eyes;and a depressed level of consciousness.

91. Kayser-Fleischer (K-F) ring is a brown discoloration at the outer margin of the cornea; the following are true EXCEPT

A. might not be present in younger children
B. requires a slit-lamp examination
C. **is permenant even after trearment**
D. is present in 95% of patients with neurologic symptoms
E. is deposition of copper in Descemet's membrane

Ans C After adequate treatment, Kayser-Fleischer rings resolve.

92. The MOST common presentation of portal hypertension is

A. jaundice
B. splenomegaly
C. **esophageal hemorrhage**
D. ascites
E. dilated cutaneous collateral vessels in the periumbilical region

Ans C Bleeding from esophageal varices is the most common presentation. Splenomegaly, sometimes with hypersplenism, is the next most common presenting feature in portal vein obstruction and may be discovered first on routine physical examination.

Chronic Diarrhea

93. Baby with diarrhea presented with restlessness but was able to drink water. Skin turgor goes back in 2 sec. Best management is:

A. Plan A
B. **Plan B**
C. Plan C
D. Plan D

Ans B The given scenario indicates a child with acute gastroenteritis with some dehydration. So treatment plan B is to be used.

94. Concentration of K+ in ORS is ___ mEq/L:

A. **20**
B. 40
C. 90
D. 10

Ans A

95. A 2-yr-old boy presented with abdominal distension, chronic diarrhea, severe anemia and failure to thrive. Which of the following is the investigation of choice?

A. Anti milk protein antibody
B. **Anti endomysial antibody**
C. Antinuclear antibody
D. Intestinal biopsy INFLAMMATORY BOWEL DISEASE

Ans B The given clinical features suggest a diagnosis of Celiac disease Anti Endomysial antibody (anti EMA) & Anti tissue transglutaminase (anti TTG) antibody help in diagnosis of Celiac disease.

96. All the following are common causes of gastrointestinal bleeding in infancy EXCEPT

A. bacterial enteritis
B. intussusception
C. **Meckel diverticulum**
D. anal fissure
E. lymphonodular hyperplasia

Ans C Meckel diverticulum is a rare cause.

97. Ileus is the failure of intestinal peristalsis caused by loss of coordinated gut motility without evidence of mechanical obstruction. In children, ileus accompanies the following metabolic abnormalities EXCEPT

A. uremia
B. hypokalemia
C. **hypocalcemia**
D. hypermagnesemia
E. acidosis

Ans C In children, it is most often associated with abdominal surgery or infection (gastroenteritis, pneumonia, peritonitis). Ileus also accompanies metabolic abnormalities (e.g., uremia, hypokalemia, hypercalcemia, hypermagnesemia, and acidosis) or administration of certain drugs, such as opiates, vincristine, and antimotility agents such as loperamide when used during gastroenteritis.

98. In infants and very young toddlers chronic diarrhea can appear following infectious enteritis. The pathogenesis of the diarrhea is not always clear and may be related to

A. **food protein allergy**
B. bacterial overgrowth
C. giardiasis
D. Strongyloides stercoralis
E. eosinophilic gastroenteropathy

Ans A The pathogenesis of the diarrhea is not always clear and may be related to secondary lactase deficiency, food protein allergy, antibiotic-associated colitis (including pseudomembranous colitis caused by Clostridiumdifficile toxin), or a combination of these.

99. Continued enteral feeding in diarrhea aids in recovery from the episode, but the following should be avoided

A. rice
B. potatoes
C. lean meats
D. fruits
E. **juices**

Ans E Fatty foods or foods high in simple sugars (juices, carbonated sodas) should be avoided.

100. A major attempt in Wilson disease should be made to restrict dietary copper intake to <1 mg/day. Of the following, the food that should be avoided is

A. **liver**
B. meat
C. egg
D. chicken
E. fish

Ans A A major attempt should be made to restrict dietary copper intake to <1 mg/day. Foods such as liver, shellfish, nuts, and chocolate should be avoided.

101. The MOST common indication for liver transplantation in children is

A. Alagille syndrome
B. congenital hepatic fibrosis
C. hepatocellular carcinoma
D. **biliary atresia**
E. Wilson disease

Ans D is the most common indication for liver transplantation in children, followed by metabolic and inborn disorders, autoimmune and familial cholestatic disorders, and acute hepatic necrosis.

Celiac Disease

102. A 5-year-old child comes to hospital with history of loose stools but no history of fever or blood in stools. Mother says he is irritable and drinks water if given. On examination eyes sunken and on Skin pinch test—the skin retracted within two seconds but not immediately. What will be the appropriate management?

A. Administer the first dose of IV antibiotic and immediately refer to higher centre
B. Give oral antibiotics and ORS and ask the mother to continue the same and visit again next day
C. Consider severe dehydration start IV fluids, IV antibiotics and refer to higher centre
D. **ORS with zinc for 14 days**

Ans D This child has acute diarrhea with some dehydration. So, the treatment of choice is ORS with zinc for 14 days.

103. A two-year-old child presents with persistent diarrhea, acidic stools and presence of one percent of reducing substance in the fresh stools. What is the most probable diagnosis?

A. Cystic fibrosis
B. **Lactose intolerance**
C. Rotavirus induced diarrhea
D. Intestinal tuberculosis

Ans B Presence of acidic stools with reducing substance positive, suggests a diagnosis of lactose intolerance.

104. The following tests can be used for diagnosing celiac disease except:

A. Anti-gliadin antibodies
B. **Anti-nuclear antibodies**
C. Anti-endomysial antibodies
D. Anti-tissue transglutaminase antibodies

Ans B

105. **The IgA anti TTG (Tissue Transglutaminase) is a sensit ive marker for diagnosis of:**

A. Celiac Disease
B. Eosinophilic Gastroenteritis
C. Primary Immunodeficiency
D. Crohn's Disease

Ans A Celiac disease
• It is an immune mediated disorder elicited by wheat, rye and barley in genetically susceptible individuals
• It is characterized by presence of–specific antibodies, HLADQ2 or DQ8 haplotypes, and enteropathy
• Celiac disease–specific antibodies comprise autoantibodies against TG2 (Tissue Transglutaminase), Endomysial antibodies (EMA), and antibodies against deamidated forms of gliadin peptides

106. **The following cereals should be avoided in patient with celiac disease, except:**

A. Wheat
B. Barley
C. Maize
D. Rye

Ans C

107. **Gluten sensitive enteropathy is most strongly associated with:**

A. HLA-DQ2
B. HLA-DR4
C. HLA-DQ3
D. Blood group 'B'

Ans A

108. **The histological features of celiac disease include all of the following except:**

A. Crypt hyperplasia
B. Increase in thickness of the mucosa
C. Increase in intraepithelial lymphocytes
D. Increase in inflammatory cells in lamina propria

Ans B • In celiac disease proximal intestine is commonly involved while in tropical sprue whole of intestine is involved. Biopsy of intestine in celiac sprue demonstrates (overall mucosal thickness remains same) – Villous atrophy, loss of microvilli brush border, with crypt hyperplasia – Plasma cells, lymphocytes, macrophages, eosinophils, mast cells are present in lamina propria

109. **The MOST common extraintestinal manifestation of celiac disease is**

A. osteoporosis
B. short stature
C. arthritis
D. iron-deficiency anemia
E. aphthous stomatitis

Ans D The most common extraintestinal manifestation of celiac disease is iron- deficiency anemia, unresponsive to iron therapy. Osteoporosis may be present; in contrast to the situation in adults, it can be reversed by a gluten-free diet, with restoration of normal peak bone densitometric values.

110. **The following neurologic manifestations are seen in celiac disease EXCEPT**

A. myopathy
B. epilepsy
C. irritability
D. cerebral calcifications
E. cerebellar ataxia

Ans A Peripheral neuropathy.

111. **The following conditions can be associated with celiac disease EXCEPT**

A. Williams syndrome
B. Down syndrome
C. Turner syndrome
D. Klinefilter syndrome
E. Sjogren syndrome

Ans D Some diseases, many with an autoimmune pathogenesis, are found with a higher-than-normal incidence in celiac disease patients. Among these are type 1 diabetes, autoimmune thyroid disease, Addison disease, Sj ̧gren syndrome, autoimmune cholangitis, autoimmune hepatitis, primary biliary cirrhosis. Such associations have been interpreted as a consequence of the sharing of identical HLA haplotypes. The relation between celiac disease and other autoimmune diseases is poorly defined;

Inflammatory Bowel Disease

112. **A 10-year-old child presents with fever since 24 hours. History reveals 3 episodes of chest infection and passage of bulky, foul smelling stools. The most probable diagnosis is?**

A. Cystic fibrosis
B. Maple syrup urine disease
C. Bilirubin conjugation defect
D. Crigler-Najjar syndrome

Ans A Recurrent chest infections & bulky, foul smelling stools point to a diagnosis of cystic fibrosis

113. **All are true regarding osmotic diarrhea except:**

A. Due to highly osmotic substances in lumen
B. Fasting may improve symptoms
C. Stool reducing substances are present
D. Can be mediated by bacterial toxins

Ans D None ≥ Can be mediated by bacterial toxins Osmotic diarrhea is caused by non-absorbed nutrients in intestinal lumen as a result of:
• Intestinal damage (e.g., enteric infection) or Reduced absorptive surface area (e.g., active celiac disease)
• Defective digestive enzyme or nutrient carrier (e.g., lactase deficiency)
• Decreased intestinal transit time (e.g., functional diarrhea); and
• Nutrient overload, exceeding the digestive capacity (e.g., overfeeding, sorbitol in fruit juice).

114. **True about Crohn disease is:**

A. Loss of haustration
B. **Growth failure**
C. Cobblestone colon
D. String sign of Kantor
E. Perianal fistula formation

Ans B c. Cobblestone colon, d. String sign of Kantor & e. Perianal fistula formation Discussing about the options one by one, a. Loss of haustrations is a feature of ulcerative colitis b. Growth failure can even be the only manifestation of Crohn disease. c. Linear ulcers can give a cobblestone appearance to the mucosal surface in Crohn disease d. 'String sign of Kantor' refers to the string-like appearance of a contrast-filled bowel loop caused by severe narrowing of a bowel loop.

115. **Ulcerative Colitis and Crohn's diseases are associated with all except?**

A. Wiskott Aldrich Syndrome
B. Turner's syndrome
C. **Glycogen storage diseases Type III**
D. Hermansky Pudlak Syndrome

Ans C Genetic disorders that have been associated with IBD include Turner syndrome, the Hermansky-Pudlak syndrome, glycogen storage disease type Ib, Wiskott Aldrich syndrome, SCID and Agammaglobulinemia.

116. **In Crohn's disease all are true except:**

A. Can occur anywhere in GIT
B. Fistula formation is common
C. **Crypt abscess are common**
D. Full thickness bowel involvement

Ans C Crypt abscesses are common in Ulcerative colitis, but less common in Crohn's disease

117. **Which of the following are inflammatory bowel disease in children?**

A. Celiac disease
B. Tropical sprue
C. **Regional ileitis (Crohn's disease)**
D. Cystic fibrosis
E. Ulcerative colitis

Ans C Regional ileitis (Crohn's disease) and e. Ulcerative colitis

118. **One of the following features is MORE common in ulcerative colitis than in Crohn disease**

A. abdominal pain
B. growth failure
C. **rectal bleeding**
D. mouth ulceration
E. strictures

Ans C diarrhea, mucus, pus, rectal involvement, toxic megacolon, sclerosing cholangitis, and crypt abscesses are more common in ulcerative colitis.

119. **Neonatal iron storage disease (NISD), also known as neonatal hemochromatosis is a rapidly fatal, progressive illness characterized by the following EXCEPT**

A. hypoprothrombinemia
B. hypoalbuminemia
C. hyperferritinemia
D. hyperbilirubinemia
E. **hyperglycemia**

Ans E NISD is a rapidly fatal, progressive illness characterized by hepatomegaly, hypoglycemia, hypoprothrombinemia, hypoalbuminemia, hyperferritinemia, and hyperbilirubinemia. The coagulopathy is refractory to therapy with vitamin K. Liver pathology demonstrates severe liver injury with acute and chronic inflammation, fibrosis, and cirrhosis.

120. **The MOST common hepatobiliary disease associated with inflammatory bowel disease (IBD) is**

A. hepatic abscess
B. portal vein thrombosis
C. **sclerosing cholangitis**
D. autoimmune hepatitis
E. biliary carcinoma

Ans C is the most common hepatobiliary disease associated with IBD, occurring in 2-8% of adult patients with ulcerative colitis and less often in Crohn disease.

Peptic Ulcer Disease

121. **The most common finding in children with severe viral enteritis:**

A. **Isotonic dehydration with Acidosis**
B. Isotonic dehydration with Alkalosis
C. Hypotonic dehydration with Acidosis
D. Hypotonic dehydration with Alkalosis

Ans A Isotonic dehydration with acidosis is the most common finding in children with severe viral enteritis.

122. **One of the intestinal enzymes that is generally deficient in children following an attack of severe infectious enteritis is:**

A. **Lactase**
B. Trypsin
C. Lipase
D. Amylase

Ans A Because Lactase activity in mucosa is marginal, it is likely to be depleted secondary to diffuse mucosal disease Causes of Acquired Lactase deficiency
• Acute Infective enteritis: Viral or Bacterial gastroenteritis • Cow's milk protein intolerance
• Cystic fibrosis • Prolonged use of drugs like Neomycin
• Protein energy malnutrition

123.A newborn suffering from perforated necrotizing enterocolitis is having very poor general condition. He is currently stabilized on ventilator. Which of the following should be done in the management of this patient?

A. Conservative treatment
B. Resection and anastomosis
C. Stabilization with membrane oxygenator and defer surgery
D. **Peritoneal drainage by putting drains in the flanks**

Ans D Management of necrotizing enterocolitis (NEC): Medical treatment of NEC include Surgical interventions in NEC
• Cessation of feeding and Nasogastric decompression. • Evidence of perforation is an indication for laparotomy
• Administration of IV fluids. and resection of necrotic bowel.
• Systemic broad spectrum antibiotics • Peritoneal drainage in unstable patients with stage III From this discussion, we choose the answer to be peritoneal drainage because; Patients is on ventilator (and unstable).

124.Delayed eruption of the primary teeth can be due to the following EXCEPT

A. familial
B. hypopituitarism
C. **hyperthyroidism**
D. cleidocranial dysplasia
E. trisomy

Ans C Hypothyroidism is a cause of delayed eruption.

125.Regarding foreign bodies in the stomach and intestine, all the following are true

A. coins are the most commonly ingested foreign body
B. perforation is estimated to be <1% of all objects ingested.
C. most objects pass though the intestine in 4-6 days
D. cathartics should be avoided
E. **ingestion of batteries usually leads to problems**

Ans E Ingestion of batteries rarely leads to problems, but symptoms can arise from leakage of alkali or heavy metal (mercury) from battery degradation in the gastrointestinal tract. Batteries can also generate electrical current and thereby cause low-voltage electrical burns to the intestine.

126.The usual presenting symptoms of peptic ulcer disease in infants and younger children are the following EXCEPT

A. feeding difficulty
B. vomiting
C. **epigastric pain**
D. crying episodes
E. hematemesis

Ans C School-age children and adolescents more commonly present with epigastric pain and nausea, presentations generally seen in adults.

127. Zinc supplementation in children with diarrhea in developing countries leads to the following EXCEPT

A. **decreased use of ORS**
B. reduced duration of diarrhea
C. reduced severity of diarrhea
D. reduced hospital admission
E. improving diarrhea recovery rates

Ans A Administration of zinc in community settings leads to increased use of ORS and reduction in the inappropriate use of antimicrobials.

128.HBV is present in high concentrations in

A. **serous exudates**
B. saliva
C. vaginal fluid
D. semen
E. urine

Ans A HBV is present in high concentrations in blood, serum, and serous exudates and in moderate concentrations in saliva, vaginal fluid, and semen.

129.Ascites is the pathologic accumulation of fluid within the peritoneal cavity. Of the following, the MOST common cause in children is

A. neoplasia
B. **hepatic**
C. cardiac
D. trauma
E. infection

Ans B In children, hepatic and renal diseases are the most common causes.

Acute Abdomen

130.A child presents with malnutrition and persistent diarrhea. Apart from antibiotics what would you like to add in your prescription?

A. Zinc, Vit A, Iron
B. Zinc, Iron
C. **Zinc, Vit A**
D. B complex, Zinc

Ans C Management of Persistent diarrhea involves:
• Correction of dehydration, electrolytes and hypoglycemia; Evaluation for infections and their management
• Nutritional therapy, including vitamin A and Zinc supplementation.

131. True about diarrhea:

A. Defined as passage of 2–3 formed stools/day
B. **Blood mixed with mucus stool is defined as dysentery**
C. Rotavirus is the most common organism in children
D. Persistent diarrhea is defined if duration is more than 21 days
E. Enterotoxigenic E. coli is the most common bacteria causing diarrhea in children

Ans B c. Rotavirus is the most common organism in children, e. Enterotoxigenic E. coli is the most common bacteria causing diarrhea in children

132.True about Peutz-Jegher syndrome

A. **Most common site is small intestine**
B. Polypectomy and Colonoscopy is treatment of choice
C. Hamartomatous polyp
D. Mucosal involvement present
E. Have no malignant potential

Ans A i.e. Most common site is small intestine; b. Polypectomy and Colonoscopy is treatment of choice; c. Hamartomatous polyp; d. Mucosal involvement present

133. **All the following are features of Stickler syndrome EXCEPT**

A. prominent joints
B. **autosomal recessive inheritance**
C. hypotonia
D. mitral valve prolapse
E. retinal detachment

Ans B Stickler syndrome, an autosomal dominant condition that includes other findings such as prominent joints, arthritis, hypotonia, hyperextensible joints, mitral valve prolapse, and ocular problems (retinal detachment, myopia, cataracts).

134. **A bezoar is an accumulation of exogenous matter in the stomach or intestine. Regarding bezoar, all the following are true EXCEPT**

A. trichobezoars are composed of the patient's own hair
B. lactobezoars can be attributed to the high casein or calcium content of some premature formulas.
C. phytobezoars are composed of a combination of plant and animal material
D. lactobezoars usually resolve when feedings are withheld for 24-48 hr
E. **sunflower seed bezoars are reported to cause small bowel obstruction**

Ans E Sunflower seed bezoars are reported to cause rectal pain and constipation as a result of the seed shells being associated with fecal impaction. Endoscopic removal is indicated, as these bezoars are refractory to enema or lavage management.

135. **Features of Tangier disease include the following EXCEPT**

A. orange tonsils
B. hepatosplenomegaly
C. relapsing neuropathy
D. orange-brown spots on the colon and ileum
E. **increased plasma cholesterol**

Ans E Features of Tangier disease include orange tonsils, hepatosplenomegaly, relapsing neuropathy, orange-brown spots on the colon and ileum, diarrhea in association with decreased plasma cholesterol levels (apolipoprotein A-I and A-II), and normal or elevated triglyceride levels. Specific therapy for Tangier disease has not yet been established.

136. **Nitazoxanide is an anti-infective agent effective in the treatment of a wide variety of pathogens including the following EXCEPT**

A. Giardia lamblia
B.
C. **difficile**
D.
E. Rotavirus

Ans C Nitazoxanide, an antiinfective agent, is effective in the treatment of a wide variety of pathogens, including C. parvum, G. lamblia, E. histolytica, Blastocystis hominis, C. difficile, and rotavirus.

137. **Risk factors for HBV infection in children and adolescents include acquisition by the following EXCEPT**

A. intravenous drug
B. contaminated needles
C. sexual contact
D. intimate contact with carriers
E. **sharing toys**

Ans E HBV is not thought to be transmitted via indirect exposure, such as sharing toys.

138. **Primary peritonitis usually refers to bacterial infection of the peritoneal cavity without a demonstrable intraabdominal source. Of the following, the MOST common isolated bacteria are**

A. group A streptococci
B. Staphylococci
C. Escherichia coli
D. Mycobacterium tuberculosis
E. **Pneumococci**

Ans E (most common), group A streptococci, enterococci, staphylococci, and Gram-negative enteric bacteria, especially Escherichia coli and Klebsiella pneumoniae, are most commonly found. Mycobacterium tuberculosis, Neisseria meningitidis, and Mycobacterium bovis are rare causes.

139. **Acute secondary peritonitis most often results from entry of enteric bacteria into the peritoneal cavity through a necrotic defect in the wall of the intestines or other viscus. It MOST commonly follows**

A. **perforation of the appendix**
B. rupture of a Meckel diverticulum
C. intussusception
D. peptic ulceration
E. necrotizing enterocolitis

Ans A It most commonly follows perforation of the appendix. Other causes include incarcerated hernias, rupture of a Meckel diverticulum, midgut volvulus, intussusception, hemolytic uremic syndrome, peptic ulceration, inflammatory bowel disease, necrotizing cholecystitis, necrotizing enterocolitis, typhlitis, and traumatic perforation.

Hernias

140. **Fluid of choice in an infant with diarrhea is:**

A. **ORS**
B. Dextrose
C. Salt water
D. Sugar water

Ans A ORS is now the global standard of care and more effective than home fluids, including decarbonated soda beverages, fruit juices, and tea for fluid replacement in diarrhea.

141. **Intractable diarrhea in children is caused by all except:**

A. Cystic fibrosis
B. **Giardiasis**
C. Secreting tumors
D. Milk allergy 378 Section 3: Systemic Pediatrics CELIAC DISEASE

Ans B Intractable diarrhea Definition Severe, protracted diarrhea for which the common pharmacologic and dietetic treatment had been unsuccessful Pathophysiology It is often the result of a permanent defect in the structure or function of the enterocyte, leading to progressive, potentially irreversible intestinal failure Etiology • Structural enterocyte defects: e.g. Microvillus inclusion disease, Tufting enteropathy with • Disorders of intestinal motility: e.g. Hirschsprung disease, intestinal pseudoobstruction

142. **Acquired megacolon in children most commonly occurs due to:**

A. **Psychological problems**
B. Bad bowel habit
C. Chagas disease
D. Hirschsprung's disease

Ans A Unlike congenital Hirschsprung disease, acquired megacolon in children is most often functional/psychogenic, resulting from chronic stool withholding and fecal retention rather than an organic cause.

143. **Periodontitis is often associated with the following conditions EXCEPT**

A. leukocyte adhesion defects
B. hypophosphatasia
C. leukemia
D. **vitamin D resistant rickets**
E. histiocytosis X

Ans D Periodontitis in children before puberty is a rare disease that often begins between the time of eruption of the primary teeth and the age of 4 or 5 yr. The disease occurs in localized and generalized forms. There is rapid bone loss, often leading to premature loss of primary teeth. It is often associated with systemic problems, including neutropenia, leukocyte adhesion or migration defects, hypophosphatasia, Papillon-Lefvre syndrome, leukemia, and histiocytosis X.

144. **H. pylori are among the most common bacterial infections in humans. All the following are true EXCEPT**

A. H. pylori is a Gram-negative, S-shaped rod
B. H. pylori infection can manifest with abdominal pain or vomiting
C. H. pylori is classified by the World Health Organization as a group I carcinogen
D. **If H. pylori is identified in a child with no symptoms, no therapy should be offered**
E. H. pylori can be associated, though rarely, with chronic autoimmune thrombocytopenia

Ans D If H. pylori is identified, even in a child with no symptoms, eradication therapy should be offered

145. **Newborns with congenital chloride diarrhea present with severe life-threatening secretory diarrhea during the 1st few wk of life. Of the following, the MOST common laboratory finding is**

A. **metabolic alkalosis**
B. hyperchloremia
C. hyperkalemia
D. hypernatremia
E. hypercalcemia

Ans A Laboratory findings are metabolic alkalosis, hypochloremia, hypokalemia, and hyponatremia (with high plasma renin and aldosterone activities).

146. **Ciprofloxacin is the drug of choice for the treatment of**

A. Salmonella
B. **Shigella**
C. Campylobacter jejuni
D. Clostridium difficile
E. Cyclospora spp.

Ans B The drug of choice for shigella is Ciprofloxacin, ampicillin, ceftriaxone, azithromycin, or TMP-SMX.

147. **Inguinal hernias are one of the most common conditions seen in pediatric practice and the most common surgical procedure performed in pediatric surgical practice. Regarding the incidence of congenital indirect inguinal hernia, all the following are true**

A. approaches 30% in very-low birth weight infants
B. more common in boys
C. approximately 60% of inguinal hernias occur on the right side
D. **bilateral hernias is higher in boys**
E. an increased incidence in twins

Ans D The incidence of bilateral hernias is higher in girls and appears to be 20-40%.

Biliary Atresia

148. **Fasting provides relief in?**

A. **Osmotic diarrhea**
B. Secretory diarrhea
C. Infective diarrhea
D. Dysentery

Ans A

149.An eight-year-old boy had abdominal pain, fever with bloody diarrhea for 18 months. His height is 100 cm and weight is 14.5 kg. Stool culture was negative for known enteropathogens. The sigmoidoscopy was normal. During the same period child had an episode of renal colic and passed urinary gravel. The Mantoux test was 5 × 5 cm. The most probable diagnosis is:

A. Ulcerative colitis
B. **Crohn's disease**
C. Intestinal tuberculosis
D. Strongyloidiasis OTHER DISORDERS OF INTESTINE

Ans B • The boy in question is obviously malnourished (Expected weight 20-25 kg, height 125-130 cm)
• A normal sigmoidoscopy rules out ulcerative colitis as it extends in a continuous fashion proximally from the rectum
• A negative mantoux test goes against active tuberculosis and a negative stool culture also rules out strongyloidosis. Hence, the obvious choice left is option 'b', i.e. Crohn's disease.

150.A 12-year-old girl has history of recurrent bulky stools and abdominal pain since 3 years of age. She has moderate pallor and her weight and height are below the 3rd percentile. Which of the following is the most appropriate investigation to make a specific diagnosis?

A. **Small intestinal biopsy**
B. Barium studies
C. 24 hours fecal fat estimation
D. Urinary d-xylose test

Ans A /e p 1831-1838
• The given child has malabsorption syndrome associated with chronic diarrhea
• Intestinal biopsy is necessary to differentiate chronic diarrhea due to different pathophysiologic reasons
• Work-up of a child suspected to have chronic diarrhea and malabsorption includes the following investigations- Repeated stool examination (especially for giardia), fecal fat excretion studies, D-xylose test, intestinal biopsy, specific tests, e.g. sweat

151. Aphthous-like lesions may be associated with the following conditions EXCEPT

A. inflammatory bowel disease
B. Behcet disease
C. gluten-sensitive enteropathy
D. SWEET syndrome
E. **herpetic gingivostomatitis**

Ans E The aphthous ulcer (canker sore) is a distinct oral lesion, prone to recurrence. Aphthous ulcers are reported to develop in 20% of the population. Their etiology is unclear, but allergic or immunologic reactions, emotional stress, genetics, and injury to the soft tissues in the mouth have been implicated.

152.A neonate is presented with watery and voluminous stool which is mistaken for urine. Of the following, the MOST likely cause is

A. congenital glucose-galactose malabsorption
B. **microvillus inclusion disease**
C. congenital bile acid malabsorption
D. congenital enterokinase deficiency
E. congenital trypsinogen deficiency

Ans B In secretory diarrhea caused by disorders such as congenital chloride diarrhea and microvillus inclusion disease, the stool is watery and voluminous and can be mistaken for urine.

153.Acrodermatitis enteropathica requires long term treatment with elemental zinc

A. 0.5 mg/kg/day
B. **1 mg/kg/day**
C. 1.5mg/kg/day
D. 2 mg/kg/day
E. 2.5 mg/kg/day

Ans B Acrodermatitis enteropathica requires long term treatment with elemental zinc 1 mg/kg/day.

154.Increased fecal calprotectin concentration is seen in

A. fat malabsorption
B. carbohydrate malabsorption
C. **intestinal inflammation**
D. pancreatic function
E. reduced intestinal surface

Ans C Abnormalities in the digestive–absorptive function tests suggest small bowel involvement, whereas intestinal inflammation, as demonstrated by increased fecal calprotectin or lactoferrin, supports colitis.

155.Stippled calcifications of the patellas and greater trochanter are found in the following intrahepatic cholestasis

A. idiopathic neonatal hepatitis
B. Aagenaes syndrome
C. **Zellweger (cerebrohepatorenal) syndrome**
D. neonatal iron storage disease
E. Byler disease

Ans C Affected infants have severe, generalized hypotonia and markedly impaired neurologic function with psychomotor retardation. Patients have an abnormal head shape and unusual facies, hepatomegaly, renal cortical cysts, stippled calcifications of the patellas and greater trochanter, and ocular abnormalities.

156.One of the following rules out biliary atresia

A. history of prematurity
B. normal size of liver
C. normal consistency of liver
D. **consistently pigmented stools**
E. no familial incidence

Ans D rule against biliary atresia. The finding of bile- stained fluid on duodenal intubation also excludes biliary atresia.

157. The success rate for establishing good bile flow after the Kasai operation is much higher (90%) if performed before

A. 4 wk of life
B. 6 wk of life
C. **8 wk of life**
D. 10 wk of life
E. 12 wk of life

Ans C The success rate for establishing good bile flowafter the Kasai operation is much higher (90%) if performed before 8 wk of life. Therefore, early referral and prompt evaluation of infants with suspected biliary atresia is important.

Pancreatic Disorders

158.An infant presents with 15-20 watery stools since 9 days. Along with Zinc, what else should be given to the patient?

A. ORS alone
B. ORS and low lactose feed
C. ORS with antibiotics
D. ORS with probiotics

Ans A This child is suffering from acute gastroenteritis (as duration is less than 14 days); treatment of Acute Gastroenteritis in children: includes oral rehydration therapy (ORS), zinc supplementation, normal diet.

159.A child presents to the emergency with a history of ingestion of a button battery, 6 hrs ago. On X-ray it was found in the stomach or duodenum. What is the next step?

A. Endoscopic removal of battery
B. Wait and watch
C. Repeat X-ray after 5 days
D. Immediate laparotomy

Ans B • Ingestion of batteries → symptoms from leakage of alkali/Hg
• If symptoms such as vomiting/abdominal pain, if a large battery (> 20 mm) remains in stomach for > 48 hr, or if a lithium battery is ingested, the battery should be removed
• In children < 6 yr age, batteries > 15 mm, are not likely to pass spontaneously & should be removed endoscopically
• If patient develops peritoneal signs surgical removal
• Batteries beyond duodenum pass per rectum in 85% within 72 hr

160.Exocrine pancreatic insufficiency is seen in:

A. Shwachman-Diamond syndrome
B. Diamond-Blackfan syndrome
C. Rubinstein-Taybi syndrome
D. Seckel syndrome

Ans A Disorders associated with pancreatic insufficiency include cystic fibrosis, Shwachman-Diamond syndrome, Pearson syndrome, Johanson-Blizzard syndrome, isolated enzyme deficiencies, enterokinase deficiency, chronic pancreatitis & protein-calorie malnutrition. fo Shwachman-Diamond syndrome is an autosomal recessive syndrome with pancreatic insufficiency, neutropenia, neutrophil chemotaxis defects, metaphyseal dysostosis, failure to thrive & short stature.

161. Eruption cyst over the erupting tooth is characterized by the following EXCEPT

A. smooth
B. painful
C. blue or blue-black
D. no treatment is indicated
E. resolves after eruption of the tooth

Ans B Eruption cyst is a smooth, painless swelling over the erupting tooth.

162.Abetalipoproteinemia is a rare autosomal recessive disorder of lipoprotein metabolism (Bassen-Kornzweig syndrome). It is characterized by the following EXCEPT

A. presence of acanthocytes in the peripheral blood smear
B. extremely low plasma levels of cholesterol
C. very low triglycerides level
D. absent deep tendon reflexes
E. normal intellectual development

Ans E Intellectual development tends to be slow.

163.The MOST common congenital disorder associated with exocrine pancreatic insufficiency is

A. Shwachman- Diamond syndrome
B. Johanson-Blizzard syndrome
C. Pearson bone marrow syndrome
D. isolated pancreatic enzyme deficiency
E. cystic fibrosis

Ans E is the most common congenital disorder associated with exocrine pancreatic insufficiency.

164.The following are alarm symptoms of functional abdominal pain EXCEPT

A. persistent left lower quadrant pain
B. unexplained fever
C. genitourinary tract symptoms
D. dysphagia
E. nocturnal diarrhea

Ans A Pain that wakes up the child from sleep, persistent right upper or right lower quadrant pain.

165.The test of choice for acute pancreatitis is

A. serum amylase
B. serum lipase
C. hyperglycemia
D. hypocalcemia
E. elevated γ-glutamyl transpeptidase

Ans B is now considered the test of choice for acute pancreatitis as it is more specific than amylase for acute inflammatory pancreatic disease and should be determinedwhen pancreatitis is suspected.

166.The first choice for diagnosis of pancreatic pseudocysts is

A. transabdominal ultrasonography
B. CT scanning
C. magnetic resonance cholangiopancreatography
D. endoscopic retrograde cholangiopancreatography
E. endoscopic ultrasound

Ans A Because of its ease, availability, and reliability, ultrasonography is the first choice.

17

CHAPTER 17

Pediatric Hepatology



WHAT THIS CHAPTER COVERS

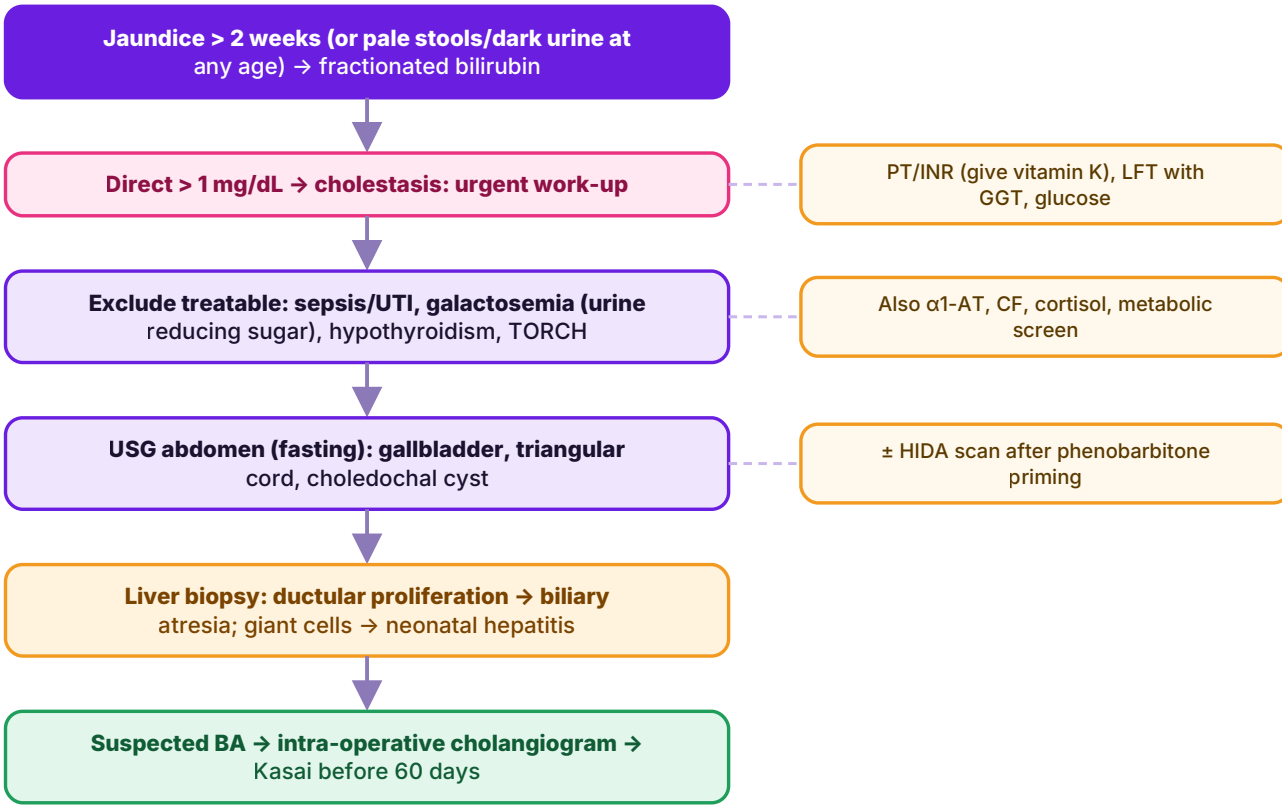
- Neonatal Cholestasis
- Biliary Atresia
- Neonatal Hepatitis
- Viral Hepatitis
- Autoimmune Hepatitis
- Metabolic Liver Disease
- Wilson Disease
- Liver Failure
- Portal Hypertension
- Liver Transplantation

25 MCQs **10** subtopics

17.1 Neonatal Cholestasis

- **Definition**: **conjugated (direct) bilirubin > 1 mg/dL (if total ≤ 5) or > 20% of total bilirubin** (NASPGHAN 2017: **direct/conjugated > 1 mg/dL** abnormal at any level); **any jaundice beyond 2 weeks (term) needs fractionated bilirubin**.
- **Red flags**: **pale/clay-coloured (acholic) stools**, **dark urine staining the nappy**, hepatomegaly, failure to thrive, bleeding (vitamin K deficiency – check PT).
- **Stool colour card** screening (Taiwan model) – used in India in some states.

Extrahepatic	Intrahepatic
Biliary atresia (MCC surgical/MC overall in many series)	**Idiopathic neonatal hepatitis (MC in some Indian series)**
Choledochal cyst	Infections – **sepsis (UTI – E. coli)**, TORCH (CMV), HSV, syphilis, HIV
Inspissated bile/spontaneous perforation of bile duct	Metabolic – **galactosemia**, tyrosinaemia, **α1-antitrypsin deficiency**, CF, Niemann–Pick C, bile-acid synthesis defects, **hypothyroidism, hypopituitarism**
	Genetic – **Alagille (paucity of bile ducts)**, **PFIC (low GGT – types 1, 2; high GGT – type 3)**, Down syndrome
	Parenteral nutrition-associated cholestasis, drugs



Work-up of neonatal cholestasis – rule out biliary atresia early

17.2 Biliary Atresia (BA)

- **Progressive fibro-obliterative cholangiopathy** of extrahepatic (± intrahepatic) bile ducts; **MC indication for liver transplantation in children**; girls slightly > boys.
- Types: **type III (atresia at porta hepatis – MC, 90%)**; **syndromic BA (BASM – polysplenia, situs inversus, preduodenal portal vein, interrupted IVC, malrotation – 10%)**.

- **Clinical**: **well-looking, thriving term baby** with **jaundice persisting beyond 2 weeks, pale stools, dark urine, firm hepatomegaly** → later splenomegaly, ascites, portal hypertension, **biliary cirrhosis by 3–6 months**.
- **Investigations**: ↑ direct bilirubin, **↑↑ GGT (> 300 IU/L)**; **USG – "triangular cord sign" (fibrotic remnant > 3–4 mm anterior to portal vein), absent/small/abnormal gallbladder (< 1.5 cm, irregular, "gallbladder ghost triad")**; **HIDA (hepatobiliary) scan – good uptake but NO excretion into bowel at 24 h** (after **phenobarbitone priming 5 mg/kg × 5 days**) – high sensitivity, poor specificity; **liver biopsy – ductular (bile duct) proliferation, bile plugs, portal fibrosis/oedema** (most reliable pre-op test); **intra-operative cholangiogram – GOLD STANDARD (confirms)**; **serum MMP-7 (new biomarker)**.
- **Treatment: Kasai portoenterostomy (hepatic portoenterostomy) – best results if performed before 60 days of age (≤ 45 days ideal)**; post-op – ursodeoxycholic acid, antibiotics (cholangitis – MC complication), fat-soluble vitamins (ADEK), MCT; ~1/3 clear jaundice long-term; **liver transplant** for failed Kasai/late presentation.

Feature	Biliary atresia	Neonatal hepatitis
Baby	**Term, normal weight, well-looking**	Often preterm/SGA, unwell
Stools	**Persistently acholic**	Intermittently pigmented
Liver	**Firm, enlarged**	Soft–firm
GGT	**Very high**	Moderate
HIDA	**No excretion**	Delayed uptake, excretion present
Biopsy	**Bile ductular proliferation, portal fibrosis, bile plugs**	**Giant-cell transformation, lobular disarray**, hepatocyte necrosis

- **Choledochal cyst** (Todani type I – fusiform dilatation of CBD, MC): **triad – jaundice, RUQ mass, abdominal pain** (classic triad in < 20%); **anomalous pancreaticobiliary junction**; cholangitis, pancreatitis, **cholangiocarcinoma risk**; USG/MRCP; Rx **excision + Roux-en-Y hepaticojejunostomy**. **Caroli disease (type V)** – intrahepatic cystic dilatation with congenital hepatic fibrosis (ARPKD).

17.3 Viral Hepatitis in Children

Virus	Key points
HAV	Faeco-oral; **MC cause of acute hepatitis & acute liver failure in Indian children**; **mostly anicteric in < 6 y**; **IgM anti-HAV**; no chronicity; complications – **relapsing/cholestatic hepatitis**, ALF; vaccine
HEV	Faeco-oral; epidemic (waterborne); **severe in pregnancy**; IgM anti-HEV
HBV	Parenteral/**vertical (MC route in endemic areas)**; **chronicity inversely related to age – 90% in perinatally infected, 25–50% at 1–5 y, < 5% adults**; immune-tolerant phase in children; **HBsAg (infection), anti-HBc IgM (acute; window period), HBeAg (replication), anti-HBs (immunity)**; HCC risk; Rx (selected) – **entecavir, tenofovir, peg-IFN**
HCV	Vertical (5%); mostly chronic, mild in children; Rx **DAAs (sofosbuvir-based) from 3 y**
HDV	Needs HBV; co-/super-infection → fulminant

- **Acute liver failure (paediatric – PALF)**: **INR ≥ 1.5 with encephalopathy or INR ≥ 2 without** (not corrected by vitamin K), no chronic liver disease; causes – **HAV (MC in India), HEV, drugs (paracetamol – MC in West; anti-TB drugs, valproate), Wilson disease (> 5 y), autoimmune, metabolic (galactosemia, tyrosinaemia – infants), HLH**, neonatal haemochromatosis (gestational alloimmune liver disease – IVIG in pregnancy). Rx supportive, **NAC (paracetamol)**, avoid sedatives, manage cerebral oedema, **liver transplant (King's College criteria)**.

17.4 Autoimmune Hepatitis & Metabolic Liver Disease

- **Autoimmune hepatitis**: girls; type 1 – **ANA, ASMA (anti-smooth muscle)**; **type 2 – anti-LKM-1 (liver-kidney microsomal) – younger, more severe, acute presentation**; **↑ IgG**; interface hepatitis, plasma cells; Rx **prednisolone + azathioprine**.

- **α1-antitrypsin deficiency** (PiZZ, SERPINA1): neonatal cholestasis, cirrhosis; **PAS-positive diastase-resistant globules** in hepatocytes; emphysema in adults.
- **Reye syndrome**: **aspirin during viral illness (influenza B, varicella)** → **acute non-inflammatory encephalopathy + microvesicular fatty liver**; vomiting, encephalopathy, **↑ ammonia, ↑ AST/ALT, normal bilirubin, hypoglycaemia**, raised ICP; mimics – FAO defects (MCAD), urea cycle defects; **avoid aspirin < 16 y** (except Kawasaki, JIA).
- **NAFLD/MASLD** – MC chronic liver disease in obese children/adolescents; ALT screen from 9–11 y in obese.

17.5 Wilson Disease

- **ATP7B (chr 13q14), AR** – defective **biliary copper excretion** & incorporation into ceruloplasmin → copper accumulates in **liver → brain (basal ganglia – lenticular), cornea, kidney, RBCs**.
- **Presentation**: **hepatic (MC in children, > 3–5 y)** – asymptomatic ↑ ALT, acute hepatitis, chronic hepatitis/cirrhosis, **acute liver failure with Coombs-negative haemolytic anaemia** (**low ALP:bilirubin ratio < 4, AST:ALT > 2.2**); **neurological (adolescents)** – **dysarthria, dystonia, tremor ("wing-beating"), drooling, parkinsonism, risus sardonicus**, deterioration in school performance; **psychiatric**; **Kayser–Fleischer ring** (copper in **Descemet's membrane** of cornea – **slit-lamp**; present in **> 95% neurological, ~50% hepatic**); **sunflower cataract**; renal – **Fanconi syndrome**, rickets; haemolysis.
- **Investigations**: **↓ serum ceruloplasmin (< 20 mg/dL)**, **↑ 24-h urinary copper (> 100 µg/day; > 40 µg suggestive in children)** – **penicillamine challenge** (> 1600 µg/24 h after 500 mg × 2); ↑ free serum copper; **hepatic copper > 250 µg/g dry weight – gold standard**; **ATP7B mutation analysis**; MRI brain – **"face of the giant panda" sign (midbrain)**, T2 hyperintense putamen; **Leipzig score ≥ 4**.
- **Treatment**: **chelation – D-penicillamine** (side effects – nephrotic syndrome, lupus, marrow suppression, **initial neurological worsening**; give **pyridoxine**) or **trientine** (better tolerated); **zinc acetate (↓ intestinal copper absorption via metallothionein – maintenance & presymptomatic; DOC in pregnancy/asymptomatic sibs)**; **ammonium tetrathiomolybdate** (neuro); low-copper diet (avoid shellfish, nuts, chocolate, liver, mushrooms); **liver transplant (curative) – fulminant Wilson (New Wilson Index ≥ 11). Screen all siblings.**
- **Indian childhood cirrhosis (ICC)**: copper toxicosis (brass vessels for milk), 1–3 y, **Mallory hyaline, orcein-positive copper** – now rare.

17.6 Portal Hypertension

Pre-hepatic	Intrahepatic	Post-hepatic
Extrahepatic portal vein obstruction (EHPVO) – MCC in Indian children (umbilical sepsis/catheterisation, dehydration; **cavernoma**)	**Cirrhosis** (biliary atresia, Wilson, AIH, metabolic), **congenital hepatic fibrosis (ARPKD)**, non-cirrhotic portal fibrosis, schistosomiasis	**Budd–Chiari** (hepatic vein/IVC – membranous obstruction), constrictive pericarditis, veno-occlusive disease (post-HSCT)

- **EHPVO**: **well child with massive haematemesis (variceal bleed) + splenomegaly + normal liver function & size**; hypersplenism, growth failure, portal biliopathy; **USG Doppler – portal cavernoma**. Rx: acute bleed – resuscitation, **octreotide**, **endoscopic variceal ligation (EVL)/sclerotherapy**; definitive – **meso-Rex bypass (restores physiological flow – preferred)**, proximal splenorenal shunt.
- **Non-selective β-blocker (propranolol)** for primary/secondary prophylaxis (less evidence in children).

17.7 Liver Transplantation – Indications

- **Biliary atresia (MC – > 50%)**, metabolic (Wilson, tyrosinaemia, urea cycle defects, Crigler–Najjar type I, PFIC, primary hyperoxaluria), **acute liver failure**, unresectable hepatoblastoma, Alagille, cirrhosis.
- **PELD score** (< 12 y; bilirubin, INR, albumin, growth failure, age < 1 y) – allocation; **MELD** ≥ 12 y.
- Living-related **left lateral segment** grafts common in India.

Practice MCQs · Pediatric Hepatology

25 questions · correct option highlighted in green · cover the answer strip to self-test

Neonatal Cholestasis

1. Which of the following viral markers signifies the ongoing viral replication in the case of Hepatitis-B infection?

A. Anti-HBs
B. Anti-HBc
C. HBe Ag
D. HBs Ag

Ans C HBeAg is a marker of active viral replication and high infectivity in hepatitis B infection.

2. In neonatal cholestasis direct bilirubin is:

A. >10%
B. >15%
C. >20%
D. >25%

Ans C Neonatal cholestasis is defined biochemically as prolonged elevation of the serum levels of conjugated bilirubin (> 2 mg/dL or > 20% of total bilirubin level) beyond the 1st 14 days of life.

3. In neonatal cholestasis, if the serum gamma glutamyl-transpeptidase (GGT) is more than 600 IU/L the most likely diagnosis is:

A. Neonatal hepatitis
B. Choledochal cyst
C. Sclerosing cholangitis
D. Biliary atresia

Ans D A serum GGT above 600 IU/L in neonatal cholestasis is characteristic of biliary atresia, in contrast to the low/normal GGT seen in some forms of progressive familial intrahepatic cholestasis.

Biliary Atresia

4. Which of the following if seen on Day 10 of life, is worrisome?

A. Conjugated hyperbilirubinemia
B. Dolls Eye Reflex
C. No weight gain
D. Unconjugated hyperbilirubinemia

Ans A Discussing about the options one by one, a. Presence of conjugated hyperbilirubinemia is abnormal at anytime of life including Neonatal period b. A Neonate may fixate on face or light in line of vision and "Doll's-eye" movement of eyes is seen c. Physiological weight loss is seen in neonates and birth weight is regained by day 10 of life in term neonates, after which weight gain begins d. Jaundice is seen in 60% of term infants and 80% of preterm infants; Physiological jaundice is always unconjugated.

5. In a child presenting with obstructive Jaundice all are elevated except:

A. Acholic stool
B. Unconjugated hyperbilirubinemia
C. Conjugated hyperbilirubinemia
D. Absence of radionuclide in duodenum in HIDA scan
E. Jaundice is presenting feature

Ans C Glutamate dehydrogenase
• Elevations in serum Alkaline phosphatase, 5′ nucleotidase, and γ-glutamyl transpeptidase levels are sensitive indicators of obstruction or inflammation of the biliary tract.

Neonatal Hepatitis

6. Most common cause of cholestatic jaundice in new born is:

A. Hypoplasia of biliary tract
B. Neonatal hepatitis
C. Choledochal cyst
D. Physiological

Ans B Neonatal (idiopathic) hepatitis is the most common cause of true cholestatic (conjugated) jaundice in the newborn, though biliary atresia must always be excluded.

7. A neonate presents with jaundice and clay colored stools. Liver biopsy shows giant cells diagnosis is:

A. Neonatal hepatitis with physiological jaundice
B. Neonatal hepatitis with extrahepatic biliary atresia
C. Physiological jaundice
D. Physiological jaundice with extrahepatic biliary atresia

Ans B • Jaundice with clay colored stools suggests cholestasis
• Both neonatal hepatitis and extrahepatic biliary atresia cause cholestasic jaundice
• "Giant cells" are characteristics of both neonatal hepatitis and extrahepatic biliary atresia
• Therefore the answer is neonatal hepatitis with extrahepatic biliary atresia. Idiopathic neonatal hepatitis (Also caused "giant cell hepatitis") is cholestatic jaundice of unknown cause that has a typical liver biopsy appearance.

Viral Hepatitis

8. False about kernicterus is:

A. No long-term effect
B. Occurs with bilirubin more than 25 mg%
C. Deposition in basal ganglion
D. Opisthotonus

Ans A Kernicterus is due to deposition of conjugated bilirubin in basal ganglia. Mnemonic for features of Chronic Bilirubin Encephalopathy (Kernicterius): "S-A-D M-U-M" (

9. Hepatitis B incubation period is:

A. < 7 days
B. 20-30 days
C. 60-90 days
D. 10-15 days

Ans C

10. A 30-year-old lady delivered a healthy baby at 37 week of gestation. She was known case of chronic hepatitis B infection and positive for HbsAg but negative for HbeAg. Which of the following is the most appropriate treatment for the baby?

A. Both active and passive immunization soon after birth
B. Passive immunization soon after birth and active immunization at 1 year of age
C. Only passive immunization soon after birth
D. Only active immunization soon after birth

Ans A

11. A 5-year-old boy is detected to be HBsAg positive on two separate occasions during a screening program for hepatitis B. He is otherwise asymptomatic. Child was given three doses of recombinant hepatitis B vaccine at the age of 1 year. His mother was treated for chronic hepatitis B infection around the same time. The next relevant step for further investigating the child would be to:

A. Obtain HbeAg and anti-Hbe levels (AIPGMEE 03)
B. Obtain anti-Hbs levels
C. Repeat HbsAg
D. Repeat another course of Hepatitis B vaccine

Ans B Obtain anti Hbs levels The child in question has received complete vaccination for Hepatitis B, is currently asymptomatic but has tested positive for HBsAg. In order to know whether the infection is acute or chronic, as well as for knowing the immune status of child, anti HBsAg levels should be done next.

12. A 45-day-old infant developed icterus and two days later symptoms and signs of acute liver failure appeared. Child was found to be positive for HbsAg. The mother was also HBsAg carrier. The mother's hepatitis B serological profile is likely to be:

A. HbsAg positive
B. HbsAg and HbeAg positivity
C. HbsAg and anti-Hbe antibody positivity
D. Mother infected with mutant HBV Chapter 17: Pediatric Hepatology 393 Answers

Ans B • Most important risk factor that determines perinatal transmission of Hepatitis B is HBeAg
• HBsAg carrier mothers who are HBe Ag positive almost invariably (> 90%) transfer infection to their offsprings. Embryological basis
• Lung develops from outpouching of foregut during 4th week of gestation
• Stages in lung development are: Embryonic stage ↓ Pseudoglandular or canalicular stage ↓ Saccular stage ↓ Alveolar stage Identify the congueensittailo lnung abnor- mality seen inQ this chest X 1-ray of a 2 year child

Autoimmune Hepatitis

13. Aspirin is associated with:

A. Reye's syndrome
B. Sjogren syndrome
C. Reiter synderome
D. None of above

Ans A

14. Chronic hepatitis is seen in infection due to which of the following viruses?

A. Hep A
B. Hep B
C. Hep E
D. Hep D

Ans B Hepatitis B & Hepatitis C can present as chronic hepatitis.

Metabolic Liver Disease

15. A 12-year-old boy presents with hematemesis, melaena and mild splenomegaly. There is no obvious jaundice or ascites. The most likely diagnosis is:

A. EHPVO
B. NCPF
C. Cirrhosis
D. Malaria with DIC

Ans A • The given history is suggestive of portal hypertension
• Extrahepatic portal vein obstruction is an important cause of portal hypertension in childhood.
• The obstruction can occur at any level of the portal vein.

16. A neonate born to infected hepatitis-B mother, should be treated with:

A. Isolation
B. Immunoglobulins
C. Hepatitis-B-vaccine
D. Immunoglobulins and hepatitis-B-vaccine

Ans D "Infants born to HbsAg positive mothers should receive both Hep B Ig and Hep B vaccine at separate sites within 12 hours of birth, followed by 2nd and 3rd dose of vaccine at 1 and 6 months.

Wilson Disease

17. True about Wilson's disease is:

A. Increased serum ceruloplasmin
B. Decreased liver copper
C. Increased urinary copper excretion
D. Decreased urine copper excretion

Ans C • In Wilson disease serum ceruloplasmin is decreased (<20 mg/dL), "free" copper level may be elevated in early ds (>1.6 µmol/L)
• Urinary copper excretion is increased to >100-1000 µg/day & hepatic copper content is > 250 µg/g dry weight in Wilson disease

18. Wilson disease true all except:

A. AR
B. KF ring
C. Raised copper level
D. Raised ceruloplasmin level

Ans D • Wilson disease is an autosomal recessive disorder caused by a mutation of ATP7B gene on chromosome 13.

Liver Failure

19. In a child with acute liver failure, the most important abnormal serum biochemical test that indicates poor prognosis:

A. Vaccine
B. Interferon
C. Gamma-globulin
D. Corticosteroids

Ans C Increasing prothrombin time • Hepatic synthetic function is reflected in serum albumin and protein levels and in the PT or INR
• Unresponsiveness of PT to vitamin K suggests severe hepatic disease
• Persistently low levels of factor VII are evidence of a poor prognosis in fulminant liver disease
• Serum aminotransferase activities do not correlate well with the severity of the illness.

20. Which one of the following hepatitis viruses have significant perinatal transmission:

A. Hepatitis E virus
B. Hepatitis C virus
C. Hepatitis B virus
D. Hepatitis A virus

Ans B

Portal Hypertension

21. Most common cause of portal hypertension in children is:

A. Extrahepatic portal vein obstruction
B. Budd-Chiari syndrome
C. Veno-occlusive disease
D. Postnecrotic

Ans A • Extrahepatic portal vein obstruction is an important cause of portal hypertension in childhood
• The obstruction can occur at any level of the portal vein
• Umbilical infection (omphalitis) with or without a history of catheterization of the umbilical vein may be causal in neonates. Three most common causes of portal hypertension in children in India: are Extrahepatic portal vein obstruction (most common), Cirrhosis, Non-cirrhotic portal fibrosis. fo 15. b.

22. Portal hypertension in children in India is commonly due to:

A. Indian childhood cirrhosis
B. Extrahepatic portal venous obstruction
C. Idiopathic portal hypertension
D. Hepatic outflow tract obstruction

Ans B

23. A 7-year-old girl from Bihar presented with three episodes of massive hematemesis and melena. There is no history of jaundice. On examination, she had a large spleen, non-palpable liver and mild ascites. Portal vein was not visualized on ultrasonography. Liver function tests were normal and endoscopy revealed esophageal varices. The most likely diagnosis is:

A. Kala azar with portal hypertension
B. Portal hypertension of unknown etiology
C. Chronic liver disease with portal hypertension
D. Portal hypertension due to extrahepatic obstruction

Ans D Splenomegaly and esophageal varices point to the diagnosis of portal hypertension while absence of jaundice and deranged liver function test indicate an extrahepatic etiology. 29. Ans. d. Biliary atresia 3 Enzymes reflect cholestasis: Alkaline phosphatase, 5'nucleotidase & Gamma glutamyl transpeptidase. About GGT in Cholestasis: (Normal value of GGT is 5-40 IU/L) "GGT levels are commonly elevated to values more than 10 times normal in biliary atresia while in hepatic causes it is raised to about three

24. Ramu, an 8-year-old boy presents with upper GI bleeding. On examination, he is found to have splenom egaly; there are no signs of ascites, or hepatom egaly; esophageal varices are found on UGIE. Most likely diagnosis is:

A. Budd Chiari syndrome
B. Non cirrhotic portal fibrosis
C. Cirrhosis
D. Veno-occlusive disease

Ans B Clues in this question are: Upper GI bleeding from esophageal varices, Splenomegaly, No ascites or hepatomegaly, No jaundice; All these indicate non-cirrhotic portal fibrosis. About other options:
• Budd-Chiari syndrome (Hepatic vein thrombosis) and venocclusive disease: Hepatomegaly, jaundice and ascites seen.
• Cirrhosis: jaundice and ascites will be seen.

Liver Transplantation

25. Regarding Gilberts syndrome all the following are true except:

A. More common in females
B. Autosomal dominant
C. Phenobarbitone inproves condition
D. Fasting increases the bilirubin condition

Ans A Gilbert Syndrome Feature Gilbert's Syndrome Inheritance (all autosomal) Promoter mutation: recessive; Missense mutations: 7 of 8 dominant; 1 reportedly recessive Total serum bilirubin < 4 mg/dL in absence of fasting or hemolysis (mild unconjugated hyperbilirubinemia); Unconjugated Hyperbilirubinemia increased in stress, fatigue, reduced caloric intake and intercurrent illness Routine liver tests Normal Hepatic histology Usually normal; increased lipofuscin pigment in some Prognosis No increased risk of cirrhosis

18

CHAPTER 18

Pediatric Respiratory Disorders



WHAT THIS CHAPTER COVERS

- Upper Respiratory Infections
- Tonsillitis & Pharyngitis
- Otitis Media
- Sinusitis
- Croup (Laryngotracheobronchitis)
- Epiglottitis
- Bronchiolitis
- Pneumonia
- Asthma
- Tuberculosis
- Pleural Effusion & Empyema
- Bronchiectasis
- Cystic Fibrosis (Respiratory)
- Foreign Body Aspiration
- Sleep-Disordered Breathing
- Acute Respiratory Failure
- Mechanical Ventilation

140 MCQs **17** subtopics

18.1 Respiratory Basics & IMNCI Pneumonia Classification

WHO fast-breathing cut-offs (count for full 60 s)

Age	Fast breathing
< 2 months	**≥ 60/min**
2–12 months	**≥ 50/min**
12 months–5 years	**≥ 40/min**

- **Chest indrawing** = lower chest wall drawing in on inspiration (in 2 m–5 y now classed as pneumonia, not severe, per WHO 2014 revision); **severe chest indrawing in < 2 months = PSBI**.
- **General danger signs (2 m–5 y)**: **not able to drink/breastfeed, vomits everything, convulsions, lethargic/unconscious**, (convulsing now).

IMNCI (2 m–5 y)	Signs	Action
Severe pneumonia or very severe disease	**Any danger sign, stridor in calm child, SpO_2 < 90%**, central cyanosis	**Refer urgently**; first dose **IM ampicillin + gentamicin** (or ceftriaxone); oxygen
Pneumonia	**Fast breathing and/or chest indrawing**	**Oral amoxicillin 40 mg/kg/dose BD × 5 days** (3 days fast breathing only, in low-HIV settings)
Cough or cold	None of the above	Home care, soothe throat, safe remedy; follow up

- **Stridor** (inspiratory) → upper airway (supraglottic/glottic/subglottic); **wheeze** (expiratory) → lower airways; **grunt** → alveolar disease (pneumonia, RDS); **biphasic stridor → tracheal/subglottic (fixed) lesion**.

18.2 Upper Respiratory Tract Infections

- **Common cold (nasopharyngitis)**: **rhinovirus (MC)**, coronavirus, RSV; 6–8 episodes/year in young children normal; symptomatic Rx (saline drops); **no antibiotics, no cough-cold OTC combinations < 4 y**.
- **Acute pharyngitis/tonsillitis**: **viral (MC – adenovirus, EBV, Coxsackie)**; bacterial – **Group A β-haemolytic Streptococcus (GAS – 15–30%, 5–15 y)**.
  - **Modified Centor/McIsaac score**: fever > 38 °C, **absence of cough**, tender anterior cervical nodes, tonsillar exudate/swelling, **age 3–14 y (+1)**, ≥ 45 y (−1) → ≥ 4 → treat/test.
  - **Rapid antigen detection test / throat culture (gold standard)**.
  - **Rx: penicillin V or amoxicillin × 10 days** (prevents rheumatic fever – if started within 9 days); single **benzathine penicillin IM**; macrolide/cephalosporin if allergic.
  - Viral clues – conjunctivitis (**pharyngoconjunctival fever – adenovirus**), coryza, cough, oral ulcers, diarrhoea.
- **Peritonsillar abscess (quinsy)** – adolescents; trismus, **uvula deviation to opposite side**, "hot potato" voice; Rx aspiration/I&D + antibiotics.
- **Retropharyngeal abscess** – **< 4–5 y** (retropharyngeal nodes regress later); fever, neck stiffness/extension refusal, drooling, stridor; **lateral neck X-ray – widened prevertebral soft tissue (> 7 mm at C2)**, CT; Rx IV antibiotics ± drainage.
- **Diphtheria** – see Infectious diseases.

18.3 Otitis Media & Sinusitis (see also ENT)

- **Acute otitis media (AOM)**: **S. pneumoniae (MC)**, non-typeable H. influenzae, Moraxella; peak 6–24 m (**short, horizontal Eustachian tube**); ear pain, fever, **bulging TM – most specific sign**, ↓ mobility on pneumatic otoscopy.
  - Rx: **amoxicillin 80–90 mg/kg/day** (high-dose) × 10 days (< 2 y) / 5–7 days (older); **watchful waiting 48–72 h** for non-severe unilateral AOM in ≥ 2 y (6–23 m if non-severe unilateral); amoxicillin-clavulanate if treatment failure.
  - Complications: **mastoiditis (MC – postauricular swelling, ear pushed forward & down)**, hearing loss, TM perforation, facial palsy, meningitis, lateral sinus thrombosis.
- **Otitis media with effusion (OME/glue ear)**: **MCC of conductive hearing loss in children**; **type B (flat) tympanogram**; watchful waiting 3 months → **grommets (tympanostomy tubes)** ± adenoidectomy.
- **Acute sinusitis**: **maxillary & ethmoid present at birth**; sphenoid by 5 y, **frontal by 7–8 y (last)**; persistent (> 10 days) or severe or "double-worsening" URTI; **periorbital (preseptal) cellulitis** – ethmoid sinusitis is MC cause of **orbital cellulitis** in children; Rx amoxicillin(-clavulanate).

18.4 Croup, Epiglottitis & Other Stridor

Feature	Croup (laryngotracheobronchitis)	Acute epiglottitis (supraglottitis)	Bacterial tracheitis
Agent	**Parainfluenza virus type 1 (MC)**, RSV, influenza, measles	**H. influenzae type b** (↓ after vaccine; now S. pyogenes, S. aureus, pneumococcus)	**S. aureus (MC)**, Moraxella, after viral croup
Age	**6 months–3 years**	**2–7 years**	3–8 y
Onset	**Gradual**, coryza 1–2 days, worse at **night**	**Sudden (hours)**, toxic, high fever	Croup that worsens; toxic
Cough	**Barking/brassy ("seal-like")**	**Absent/minimal**	Brassy
Voice	**Hoarse**	**Muffled ("hot potato")**	Hoarse
Drooling/ dysphagia	No	**Yes – drooling, dysphagia, tripod/sniffing position**	No
X-ray (AP neck)	**"Steeple / pencil-tip sign"** (subglottic narrowing)	**Lateral neck: "thumb sign"** (swollen epiglottis), "vallecula sign"	Irregular tracheal margins, pseudomembrane
Rx	**Dexamethasone 0.15–0.6 mg/kg oral/IM single dose (all severities)**; **nebulised (racemic/L-) epinephrine** for moderate–severe (stridor at rest) – observe 2–4 h (rebound); budesonide neb; humidified air not effective	**Airway first – do NOT examine throat/lie supine/upset child**; secure airway in OT (intubation by most experienced); **IV ceftriaxone/cefotaxime**; rifampicin prophylaxis for contacts (Hib)	IV antibiotics (vancomycin + 3rd-gen cephalosporin), bronchoscopy, often intubation

- **Westley croup score** (stridor, retractions, air entry, cyanosis, consciousness): ≤ 2 mild, 3–7 moderate, ≥ 8 severe.
- **Spasmodic croup** – sudden night-time attacks, no fever, recurrent, atopic.
- **Laryngomalacia** – **MCC of congenital stridor & MC congenital laryngeal anomaly**; **inspiratory stridor worse when supine/feeding/crying, better prone**, appears at 2–4 weeks, peaks 6–8 m, **resolves by 12–24 months**; **omega-shaped epiglottis, short aryepiglottic folds** on flexible laryngoscopy; supraglottoplasty if severe (FTT, apnoea).
- Others: **vocal cord palsy (2nd MC; Arnold–Chiari)**, **subglottic stenosis** (post-intubation – MC acquired), **subglottic haemangioma** (with **"beard" cutaneous haemangioma**, biphasic stridor; **propranolol**), laryngeal web, vascular ring (**double aortic arch**), **foreign body**.

Foreign body aspiration

- **1–3 years** (peanuts/seeds MC); **right main bronchus (MC – wider, more vertical)**; choking episode → cough, **unilateral wheeze, ↓ breath sounds**; may be missed → recurrent pneumonia at same site.
- **CXR**: **obstructive emphysema (check-valve) – hyperinflation of affected side, mediastinal shift to opposite side on expiration film/lateral decubitus**; atelectasis (complete obstruction); mostly radiolucent.
- **Rigid bronchoscopy – diagnostic & therapeutic (TOC)**.

18.5 Bronchiolitis

- **RSV (MC – 50–80%)**, rhinovirus, hMPV, parainfluenza, adenovirus (**bronchiolitis obliterans**), influenza; **< 2 years (peak 2–6 months)**; winter/monsoon.
- **Prodrome of coryza (2–3 days) → cough, tachypnoea, chest retractions, wheeze + fine crepitations, hyperinflation (liver pushed down)**, apnoea in young infants; low-grade fever.
- High-risk: **prematurity, < 3 months, CHD (haemodynamically significant), BPD/CLD, immunodeficiency, neuromuscular disease, Down**.
- CXR (not routine): **hyperinflation, patchy atelectasis, peribronchial thickening**.
- **Treatment – supportive**: **oxygen if SpO_2 < 90–92%**, **hydration/NG feeds**, nasal suction, **HFNC/CPAP** for respiratory distress.
  - **NOT recommended routinely**: bronchodilators (salbutamol), adrenaline, **corticosteroids**, antibiotics, chest physiotherapy, ribavirin (except severe immunocompromised); **nebulised 3% hypertonic saline** – may ↓ hospital stay (inpatients).
- **Prevention**: **palivizumab (monthly IM 15 mg/kg in RSV season)** for high-risk; **nirsevimab (single-dose long-acting monoclonal)**; **maternal RSV vaccine (RSVpreF, 32–36 wk)**.
- Long-term – recurrent wheeze/asthma in some.

18.6 Pneumonia

Age	Common organisms
Neonate	**GBS, E. coli, Klebsiella**, Listeria; CMV; **Chlamydia trachomatis (afebrile pneumonia of infancy, 2–12 weeks, staccato cough, conjunctivitis, eosinophilia)**
1–3 months	Viruses (RSV), Chlamydia, Bordetella pertussis, S. pneumoniae, S. aureus
3 months–5 years	**Viruses (RSV MC overall)**; bacterial – **S. pneumoniae (MC bacterial)**, Hib, S. aureus
> 5 years	**Mycoplasma pneumoniae (MC – "walking pneumonia")**, Chlamydophila pneumoniae, S. pneumoniae
HIV-infected infant	**Pneumocystis jirovecii (3–6 m)**, CMV, TB

- **S. aureus pneumonia**: infants, after measles/influenza, skin infection; **rapid progression, pneumatoceles, empyema, pyopneumothorax**; Rx **cloxacillin/vancomycin (MRSA)**.
- **Mycoplasma**: school-age, gradual onset, headache, sore throat, **dry cough, few signs ("X-ray worse than patient")**, **cold agglutinins (IgM anti-I)**, bullous myringitis, rash (SJS), haemolysis, encephalitis; **Rx macrolide (azithromycin)**.
- **Klebsiella** – bulging fissure, red-currant jelly sputum. **Round pneumonia** in children (pneumococcus) – mimics mass.
- **Antibiotics (IAP/WHO)**: non-severe – **oral amoxicillin**; severe – **IV ampicillin/penicillin + gentamicin** or **ceftriaxone**; add **cloxacillin/vancomycin** if staphylococcal; **macrolide** for atypical (> 5 y); duration 5–7 days (10–14 for staph).
- **Recurrent pneumonia** (≥ 2 in 1 y or ≥ 3 ever with radiological clearing): **same lobe → local cause (foreign body, bronchial stenosis, sequestration, bronchiectasis)**; **different lobes → systemic (aspiration – GERD/ neurological, immunodeficiency, CF, PCD, CHD, sickle cell)**.

18.7 Bronchial Asthma

- **Chronic airway inflammation** with **variable airflow obstruction** & bronchial hyperresponsiveness; **MC chronic disease of childhood**; atopy (eczema, allergic rhinitis), family history, viral triggers, allergens (house-dust mite – MC), smoke, exercise, cold air.
- **Wheezing phenotypes (< 5 y)**: transient early (viral, resolves by 3 y), persistent (atopic), late-onset; **Asthma Predictive Index (API)** – frequent wheeze + 1 major (parental asthma, eczema, aeroallergen sensitisation) or 2 minor (food sensitisation, wheeze apart from colds, eosinophils ≥ 4%).
- **Diagnosis**: clinical; **spirometry (> 5–6 y) – FEV_1/FVC < 0.8–0.9 with reversibility ≥ 12% (and ≥ 200 mL) after salbutamol**; **PEFR variability > 13% (children)**; FeNO > 35 ppb (children) supports; methacholine/exercise challenge; CXR to exclude others.

NOT EVERYTHING THAT WHEEZES IS ASTHMA

Infants: **bronchiolitis, foreign body, GERD/aspiration, cystic fibrosis, PCD, vascular ring, tracheomalacia, CHF, TB lymph node compression, BPD**. Clues: onset at birth, FTT, focal signs, no response to bronchodilator, clubbing (**clubbing is NOT a feature of asthma** – think CF, bronchiectasis).

GINA 2024 – Assessment & stepwise treatment (6–11 years)

Step	Preferred controller	Reliever
1	**Low-dose ICS whenever SABA taken** (or daily low-dose ICS)	SABA
2	**Daily low-dose ICS** (or LTRA)	SABA
3	**Low-dose ICS-LABA** or **medium-dose ICS** or **very-low-dose ICS-formoterol MART**	SABA (or ICS-formoterol in MART)
4	**Medium-dose ICS-LABA** or low-dose ICS-formoterol MART; refer	
5	Refer – phenotyping, **add-on anti-IgE (omalizumab), anti-IL-5 (mepolizumab), anti-IL4R (dupilumab)**, tiotropium	

- **Adolescents ≥ 12 y (Track 1 – preferred)**: **as-needed low-dose ICS-formoterol (budesonide-formoterol) as reliever for steps 1–2**, maintenance-and-reliever (MART) steps 3–5. **SABA-only treatment no longer recommended** (↑ severe attacks).
- **≤ 5 years**: step 1 – SABA as needed (+ intermittent ICS); **step 2 – daily low-dose ICS (preferred)** or LTRA (montelukast – neuropsychiatric warning); step 3 – double low-dose ICS; step 4 – refer.
- **Devices**: **< 4 y – MDI + spacer with face mask**; **4–6 y – MDI + spacer with mouthpiece**; **> 6 y – DPI or MDI + spacer**; nebulisers only for acute severe. **pMDI + spacer is as effective as nebuliser** in acute attacks.
- **Control assessment** (last 4 weeks): daytime symptoms > 2/week, night waking, reliever > 2/week, activity limitation – **well controlled (0), partly (1–2), uncontrolled (3–4)**.
- ICS side effects: oral thrush (rinse mouth), dysphonia, **↓ growth velocity (~0.5–1 cm in first year, not cumulative)**.

Acute severe asthma

Severity	Features
Mild–moderate	Talks in phrases, prefers sitting, not agitated, HR/RR ↑, SpO_2 90–95%, PEF > 50%
Severe	**Talks in words, sits hunched forward, agitated, accessory muscles, RR > 30 (> 5 y)/> 40 (≤ 5 y), HR > 120–125, SpO_2 < 90–92%, PEF ≤ 50%**
Life-threatening	**Drowsy/confused, silent chest, cyanosis, poor respiratory effort, hypotension, normal or ↑ $PaCO_2$ (ominous)**, PEF < 33%

MANAGEMENT OF ACUTE EXACERBATION

1. **Oxygen to SpO_2 94–98%**.
2. **Salbutamol (SABA) – MDI + spacer 4–10 puffs every 20 min for 1 h** (or nebulised 2.5–5 mg) → then as needed; **ipratropium bromide** added in severe (first hour).
3. **Systemic corticosteroids early – oral prednisolone 1–2 mg/kg/day (max 40 mg) × 3–5 days** (IV hydrocortisone/methylprednisolone if vomiting/severe).
4. **IV magnesium sulfate 25–50 mg/kg (max 2 g) over 20 min** – severe/not responding (also nebulised).
5. Refractory: **IV salbutamol/terbutaline infusion, aminophylline** (narrow therapeutic index), **HFNC/NIV, ketamine for intubation**, heliox. - **No sedatives, no antibiotics routinely, no chest physio, no mucolytics.**

18.8 Tuberculosis (respiratory aspects) – see Infectious Diseases

- **Primary pulmonary TB** – hilar/mediastinal lymphadenopathy (MC radiological finding), **collapse-consolidation (compression of bronchus – epituberculosis)**, pleural effusion (older children), **miliary TB (infants, malnourished – CNS involvement in 30%)**, progressive primary disease, adult-type cavitary disease in adolescents.
- **Endobronchial TB** – localised wheeze, segmental collapse.

18.9 Pleural Effusion & Empyema

- **Parapneumonic effusion/empyema**: **S. pneumoniae (MC)**, **S. aureus (MC in India/developing countries – after skin infection/measles)**, GAS, H. influenzae; TB (lymphocytic exudate, high ADA > 40 U/L), malignancy (lymphoma), nephrotic (transudate).
- **Light's criteria (exudate)**: pleural protein/serum protein > 0.5, pleural LDH/serum LDH > 0.6, pleural LDH > 2/3 ULN of serum.
- **Empyema stages**: exudative → **fibrinopurulent (pH < 7.2, glucose < 40, LDH > 1000, loculations)** → organising (peel).
- **USG chest – best to detect & characterise (septations, loculation)**; CT for complicated cases.
- Rx: **IV antibiotics (cloxacillin + ceftriaxone; vancomycin if MRSA)** 2–4 weeks; **ICD (intercostal drain)** for large/purulent; **intrapleural fibrinolytics (urokinase/streptokinase/alteplase)** for loculated; **VATS decortication** if failure. Children have excellent long-term lung function.
- **Pneumatocele** (staph) – thin-walled air cysts, usually resolve spontaneously.

18.10 Bronchiectasis, CF & Others

- **Bronchiectasis (non-CF)**: **post-infectious (measles, pertussis, TB, adenovirus) – MCC in India**, **foreign body**, **primary ciliary dyskinesia (PCD)**, **immunodeficiency (XLA, CVID)**, **ABPA**, aspiration; **chronic wet/productive cough > 8 weeks, clubbing, coarse crepitations**, haemoptysis; **HRCT – IOC (signet-ring sign, tram tracks, lack of tapering)**; Rx airway clearance, antibiotics for exacerbations, azithromycin, treat cause, lobectomy for localised.
- **Primary ciliary dyskinesia** (AR, DNAH5/DNAI1): **neonatal respiratory distress (term), chronic wet cough, chronic rhinosinusitis, OME, bronchiectasis (lower/middle lobes)**, **situs inversus in 50% (Kartagener syndrome – triad: situs inversus, sinusitis, bronchiectasis)**, male infertility; Dx **nasal nitric oxide (low)**, high-speed video microscopy, **EM – absent dynein arms**, genetics.
- **Young syndrome**: bronchiectasis + sinusitis + **obstructive azoospermia** (normal cilia).
- **ABPA** – asthma/CF + Aspergillus: central bronchiectasis, ↑ IgE > 1000, eosinophilia, Aspergillus-specific IgE/IgG; Rx steroids + itraconazole.
- **Cystic fibrosis** – see Genetics chapter.
- **Pulmonary haemosiderosis** – recurrent haemoptysis, iron-deficiency anaemia, **haemosiderin-laden macrophages (BAL/gastric aspirate)**; **Heiner syndrome** (cow's milk-associated).

- **Interstitial lung disease (chILD)** – NEHI (neuroendocrine cell hyperplasia of infancy), surfactant protein defects (SFTPB, SFTPC, ABCA3).
- **Pneumothorax** – spontaneous in tall thin adolescents (Marfan), CF, asthma, staph; tension → needle decompression (2nd ICS MCL / 4th–5th ICS anterior-axillary).
- **Obstructive sleep apnoea** – adenotonsillar hypertrophy (MC), obesity, craniofacial; **polysomnography (AHI > 1 abnormal in children)**; Rx **adenotonsillectomy**.
- **Congenital central hypoventilation (Ondine's curse – PHOX2B)** – hypoventilation in sleep; Hirschsprung (Haddad syndrome), neuroblastoma.

18.11 Approach Tables

Chronic cough (> 4 weeks)	Clue
Wet cough	**Protracted bacterial bronchitis (MC – responds to 2 weeks amoxicillin-clavulanate)**, bronchiectasis, CF, PCD, aspiration
Dry cough	Asthma (cough-variant), post-viral, upper airway cough syndrome, GERD, habit/tic cough (disappears in sleep), TB, foreign body, ILD
Paroxysmal with whoop	Pertussis
Barking/brassy	Croup, tracheomalacia, habit cough (**"honking"**)
Staccato	**Chlamydia trachomatis** in infants
Cough with feeding	Aspiration, **H-type TEF**, laryngeal cleft

- **Haemoptysis in children**: infection (pneumonia, TB – MC in India), bronchiectasis/CF, foreign body, pulmonary haemosiderosis, CHD (Eisenmenger, pulmonary venous hypertension), AVM.
- **Digital clubbing**: **CF, bronchiectasis, empyema/lung abscess, cyanotic CHD, IE, ILD, IBD (Crohn's), cirrhosis/biliary atresia, celiac, thyrotoxicosis (acropachy)**, familial; **not in uncomplicated asthma or pneumonia**.

Practice MCQs · Pediatric Respiratory Disorders

140 questions · correct option highlighted in green · cover the answer strip to self-test

Upper Respiratory Infections

1. Congenital lobar emphysema is commonly seen in which lobe?

A. Left lower
B. Right upper
C. **Left upper**
D. Right lower

Ans C

2. A child with 3 days history of upper respiratory tract infection presents with stridor, which decreases on lying down position. What is the most probable diagnosis?

A. Acute epiglottitis
B. **Laryngotracheobronchitis**
C. Foreign body aspiration
D. Retropharyngeal abscess

Ans B The given clinical features are suggestive of a diagnosis of Croup or Laryngotracheobronchitis.

3. A 4-year-old malnourished child is brought to subcentre with breathing rate of 55 per minute, excessive crying, irritability, fever and not taking feeds. The ANM assesses the child and categorizes under the IMNCI guidelines for management of ARI as?

A. No pneumonia
B. **Very severe disease**
C. Pneumonia
D. Upper respiratory tract infection

Ans B New ARI classification (2014) by WHO: Diagnosis Clinical features Pneumonia Fast breathing and/or chest indrawing Severe pneumonia or Very severe disease General danger signs present As the child is not able to feed, he has very severe disease;

4. A 4-year-child with bronchial asthma presents history of 3 or more episodes during daytime and 2 wheezing episodes during night in a week. How will you grade this asthma?

A. Mild persistent
B. **Moderate persistent**
C. Severe persistent
D. Mild intermittent

Ans B

5. Grunting is produced by expiration against a partially closed glottis and is an attempt to maintain positive airway pressure during expiration for as long as possible. It is MOST commonly prominent in

A. epiglottitis
B. **hyaline membrane disease**
C. asthma
D. croup
E. choanal atresia

Ans B Grunting is most beneficial in alveolar diseases that produce widespread loss of FRC, such as in pulmonary edema, hyaline membrane disease, and pneumonia. Grunting is also effective in small airway obstruction (bronchiolitis) to maintain a higher positive pressure in the airway during expiration, decreasing the airway collapse.

6. The following may be effective as a common cold treatment EXCEPT

A. **vitamin C**
B. first generation antihistamine
C. topical or oral adrenergic agents
D. ipratropium bromide
E. honey

Ans A guaifenesin, and inhalation of warm, humidified air are no more effective than placebo for the treatment of cold symptoms.

7. Pleurisy or inflammation of the pleura is often accompanied by an effusion. Of the following, the MOST common cause of pleural effusion in children is

A. tuberculosis
B. heart failure
C. **bacterial pneumonia**
D. rheumatoid arthritis
E. metastatic intrathoracic malignancy

Ans C The most common cause of pleural effusion in children is bacterial pneumonia.

Tonsillitis & Pharyngitis

8. The most common cause of stridor in infant and children is:

A. Congenital subglottic stenosis
B. **Laryngomalacia**
C. Vocal Cord Paralysis
D. Foreign body in airway

Ans B Ghai 8/e p 369 Laryngomalacia is the most common cause of stridor in infants and children.

9. A 9-month-old HIV positive child following URTI developed sudden onset of breathlessness. The chest X-ray shows hyperinflation. The O saturation was greater 2 than 90%; the treatment of choice is:

A. Cotrimoxazole
B. **Ribavarin**
C. IV ganciclovir
D. Nebulized acyclovir

Ans B This HIV positive child has clinical features of Acute Bronchiolitis In immunocompromised child with Bronchiolitis, Ribavirin, an antiviral agent, shortens the course of illness.

10. Which of the following is the leading cause of mortality in under 5 children in developing countries?

A. Malaria
B. Acute lower respiratory tract infections (LRTI)
C. Hepatitis
D. **Prematurity**

Ans D Causes of under 5 child deaths: The area to the right of the dotted line indicates neonatal conditions

11. Chest CT is of a particular importance in the evaluation of the following lesions

A. mediastinal lesions
B. cystic parenchymal lesions
C. pulmonary embolisms
D. **vascular rings**
E. bronchiectasis

Ans D MRI is an excellent procedure to delineate hilar and vascular anatomy associated with vascular rings or slings.

12. Lemierre syndrome is characterized by septic thrombophlebitis of the internal jugular veins with septic pulmonary emboli, producing hypoxia and pulmonary infiltrates, it is a complication of pharyngitis caused by

A. group A β-hemolytic streptococcus
B. Arcanobacterium haemolyticum
C. Mycoplasma pneumoniae
D. **Fusobacterium necrophorum**
E. Corynebacterium diphtheriae

Ans D Lemierre syndrome is a serious complication of F. necrophorum pharyngitis and is characterized by septic thrombophlebitis of the internal jugular veins with septic pulmonary emboli, producing hypoxia and pulmonary infiltrates.

13. The classic laboratory findings of idiopathic pulmonary hemosiderosis (IPH) are the following EXCEPT

A. microcytic hypochromic anemia
B. elevated reticulocyte count
C. **normal plasma bilirubin**
D. reduced serum iron
E. elevated iron-binding capacity

Ans C The anemia of IPH can mimic a hemolytic anemia. Elevations of plasma bilirubin are caused by absorption and breakdown of hemoglobin in the alveoli.

14. The MOST common physical finding of the pulmonary exam in bronchopulmonary dysplasia (BPD) is

A. **tachypnea**
B. mouth breathing
C. increased anteroposterior diameter of the chest
D. intercostal retractions
E. clear breath sounds

Ans A Physical findings of the pulmonary exam vary with the severity of disease. Tachypnea is a common finding.

Otitis Media

15. Kartagener syndrome includes all of the following except:

A. **Dysphagia**
B. Bronchiectasis
C. Sinusitis
D. Dextrocardia

Ans A Kartagener syndrome includes situs inversus, chronic sinusitis and otitis, and airway disease leading to bronchiectasis.

16. Kartagener syndrome comprises of:

A. **Chronic sinusitis, situs inversus and bronchiectasis**
B. Chronic sinusitis, situs inversus and bronchitis
C. Chronic sinusitis, situs inversus and chronic ottitis media
D. Chronic sinusitis, dextrocardia and bronchitis

Ans A

17. A 5-year-old unimmunised child presents with pseu- domembrane in throat, fever and sore throat, the organ- ism responsible is:

A. Oral calcium
B. **Wait and watch**
C. Nebulization
D. Antibiotic

Ans B Pleomorphic gram positive bacilli The child is most probably suffering from Diphtheria, caused by Corynebacterium diphtheria Corynebacteria are aerobic, nonencapsulated, non-spore-forming, mostly nonmotile, pleomorphic, Gram-positive bacilli.

18. A child with cyanosis and choking on feeding at birth was diagnosed as TEF and had underwent a corrective surgery. He comes to you with complaints of barking cough and expiratory wheeze. What is the most likely diagnosis?

A. Recurrent papillomatosis
B. Croup
C. **Epiglottitis**
D. Tonsillitis

Ans C Tracheomalacia Nelson's 20/e p 2032-2034
• Tracheomalacia is the rule following correction of tracheoesophageal fistula
• Bronchomalacia is common following lung transplantation.
Clinical features of Tracheomalacia:
• Stridor is inspiratory, low pitched and exacerbated by any exertion; crying, agitation, or feeding.
• Results from the collapse of supraglottic structures inwards during inspiration.
• Symptoms appear within the first 2 weeks of life and increase in severity for up to 6 months

19. An 18-month-old child comes to subcenter with history of fever and cough. On examination weight was 11.5 kg, respiratory rate was 46/min, and there was no chest indrawing. Which is the most appropriate management?

A. IV antibiotics at PHC and observe
B. Give first dose of IV antibiotics & refer to higher center
C. **Oral cotrimoxazole for 5 days and send home**
D. Home remedies

Ans C Child has fast breathing with no danger signs; So he is suffering from Pneumonia. So, oral antibiotics need to be started & child can stay at home

20. Declines of 50% or more in rates of sudden infant death syndrome (SIDS0 in the USA and around the world have occurred in the past decade. The reductions in risk appear to be related primarily to

A. **decreases in placing infants prone for sleep**
B. exclusive breast-feeding
C. adequate immunizations
D. avoidance of soft sleeping surface
E. avoidance of bed sharing with parent(s)

Ans A The reductions in risk appear to be related primarily to decreases in placing infants prone for sleep and increases in placing them supine. A number of other risk factors also have significant associations with SIDS; although many are nonmodifiable and most of the modifiable factors have not changed appreciably, self-reported maternal smoking prevalence during pregnancy has decreased by 25% in the past decade.

21. Pulmonary edema is traditionally separated into two categories according to cause (cardiogenic and non-cardiogenic). Non-cardiogenic pulmonary edema, in its most severe state, is also known as acute respiratory distress syndrome (ARDS). The following radiographic features is usually present in non-cardiogenic pulmonary edema

A. septal lines
B. peribronchial cuffing
C. pleural effusions
D. **air bronchograms**
E. even or central distribution of edema

Ans D is usually present in non cardiogenic edema while the other findings are usually found in cardiogenic pulmonary edema.

22. The following are causes of diffuse alveolar hemorrhage (DAH) syndromes with pulmonary capillaritis EXCEPT

A. Goodpasture syndrome
B. Henoch-Schonlein purpura
C. Wegener granulomatosis
D. **Heiner syndrome**
E. Behcet syndrome

Ans D is a cause of diffuse alveolar hemorrhage (DAH) syndromes without pulmonary capillaritis.

23. The MOST common cause of pneumomediastinum in older children and teenagers is

A. dental extractions
B. adenotonsillectomy
C. tuberculosis
D. mycoplasma pneumonia
E. **acute asthma**

Ans E is the most common cause of pneumomediastinum in older children and teenagers. Simultaneous pneumothorax is unusual in these patients.

24. Bronchopulmonary dysplasia (BPD) is a syndrome characterized by signs and symptoms of chronic lung disease that originates in the neonatal period. An accepted definition includes a postnatal oxygen requirement for

A. 7 days
B. 14 days
C. 21 days
D. **28 days**
E. 35 days

Ans D An accepted definition includes an oxygen requirement for 28 days postnatally, and the disorder is graded as mild, moderate or severe on the basis of supplemental oxygen requirement and gestational age.

25. Asphyxiating thoracic dystrophy (Jeune syndrome) is associated with the following skeletal abnormalities EXCEPT

A. narrowed thorax
B. horizontal ribs
C. short extremities
D. bell-shaped chest cage
E. **low clavicles**

Ans E Physical examination reveals a narrowed thorax that, at birth, is much smaller than the head circumference. The ribs are horizontal, and the child has short extremities. Chest radiographs demonstrate a bell-shaped chest cage with short, horizontal, flaring ribs and high clavicles.

Sinusitis

26. Respiratory system develops from:

A. **Ventral wall of foregut**
B. Dorsal wall of foregut
C. Ventral wall of midgut
D. Dorsal wall of midgut

Ans A Respiratory system develops from: ventral wall of foregut.Q

27. An child with recent onset of URTI after 2 days presents with acute onset of breathlessness, cough and fever. All of the following can be given except: M (AIIMS May 2001) 408 Section 3: Systemic Pediatrics

A. Antipyretics
B. **Morphine**
C. Antibiotics
D. O inhalation

Ans B Morphine or any other sedative is to be avoided in a child with respiratory distress.

28. Pneumothorax could be a complication of:

A. Staphylococcal pneumonia
B. Pneumococcal pneumonia
C. Klebsiella pneumonia
D. Viral pneumonia ASTHMA

Ans A Pneumothorax could be a complication of Staphylococcal pneumonia, due to rupture of pneumatoceles. ASTHMA

29. Rigid bronchoscopy is preferentially indicated for

A. persistent pneumonia
B. extracting foreign bodies
C. atelectasis
D. persistent wheeze
E. interstitial disease

Ans B Rigid bronchoscopy is preferentially indicated for extracting foreign bodies, for removing tissue masses, and in patients with massive hemoptysis. In other cases, the flexible scope offers the advantages that it can be passed through endotracheal or tracheostomy tubes can be introduced into bronchi that come off the airway at acute angles, and can be safely and effectively inserted with topical anesthesia and conscious sedation.

30. Habit cough ("psychogenic cough" or "cough tic") must be considered in any child with a cough that has lasted for weeks or months, that has been refractory to treatment and is

A. abrupt
B. loud
C. barking quality
D. all day, never during sleep
E. paroxysmal

Ans D Habit cough ("psychogenic cough" or "cough tic") must be considered in any child with a cough that has lasted for weeks or months, that has been refractory to treatment, and that disappears with sleep or with distraction.

31. The treatment of choice of idiopathic pulmonary hemosiderosis (IPH) is

A. transfusion of blood products
B. systemic corticosteroids
C. cyclophosphamide
D. chloroquine
E. lung transplantation

Ans B In IPH, early treatment with systemic corticosteroids is the treatment of choice.

32. The following are features of pectus excavatum (funnel chest) EXCEPT

A. occurs in 1 : 400 births
B. usually associated with a connective tissue disorder
C. 9 : 1 male preponderance
D. accounts for >90% of congenital chest wall anomalies
E. positive family history in one third of cases

Ans B It may be associated with a connective tissue disorder (Marfan or Ehlers-Danlos syndrome).

Croup (Laryngotracheobronchitis)

33. Most common type of foregut cysts are?

A. Bronchogenic
B. Esophageal
C. Enteric
D. Mixed type

Ans A • Foregut cysts are abnormal detachments of primitive foregut most often located in hilum or middle mediastinum.
• 3 types: Bronchogenic (most common), Esophageal or Enteric.

34. Steeple sign is seen in:

A. Acute epiglottitis
B. Laryngotracheobronchitis (croup)
C. In both cases
D. None of above

Ans B 'Steeple sign' due to subglottic narrowing is characteristically seen in Croup

35. A 2-year-old child was brought to the emergency department at 3 AM. The child presented with fever and cough. On examination, respiratory rate was 36/min, temperature was 39°°C and saturation on pulse oximetry was 96%. The child had barking cough and stridor only on crying. Otherwise, the child was hydrated, able to drink and consolable. The next step in the management would be:

A. Single dose dexamethasone
B. Nebulized racemic epinephrine solution
C. Complete blood count and culture
D. Nasal washings for influenza virus and RSV

Ans A Single dose dexamethasone Nelson's 20/e p 2032-2034

36. A 4-year-old child has seal barking like croupy cough. Management includes all except:

A. Respiratory rate > 40/min in a child between 2-12 months age
B. Respiratory rate > 50/min in a child between 2-12 months age
C. Respiratory rate > 30/min in a child between 1-5 years age
D. Respiratory rate > 50/min in a child between < 2 months age
E. Salbutamol PULMONARY INFECTIONS 40. Which of the following is abnormal?

Ans B Antibiotic, d. Morphine, e. Salbutamol This child probably has Croup; Antibiotics, Morphine and Salbutamol are not indicated in the management of croup.

37. A "silent chest" with infiltrates should arouse suspicion of all the following EXCEPT

A. alveolar proteinosis
B. bronchopulmonary dysplasia
C. Pneumocystis jiroveci infection
D. interstitial pneumonitis
E. tumors

Ans B A "silent chest" with infiltrates should arouse suspicion of alveolar proteinosis, Pneumocystis jiroveci infection, genetic disorders of surfactant synthesis and secretion causing interstitial pneumonitis, or tumors.

38. The barking cough typical of croup is rare in

A. laryngotracheobronchitis
B. **acute epiglottitis**
C. acute infectious laryngitis
D. spasmodic croup
E. measles croup

Ans B This dramatic, potentially lethal condition is characterized by an acute rapidly progressive and potentially fulminating course of high fever, sore throat, dyspnea, and rapidly progressing respiratory obstruction. The barking cough typical of croup is rare.

39. Children with croup should be hospitalized for any of the following EXCEPT

A. progressive stridor
B. severe stridor at rest
C. cyanosis
D. depressed mental status
E. **congenital heart disease**

Ans E Children with croup should be hospitalized for any of the following: progressive stridor, severe stridor at rest, respiratory distress, hypoxia, cyanosis, depressed mental status, poor oral intake, or the need for reliable observation.

Epiglottitis

40. Blood supply of bronchogenic sequestration is:

A. **Aorta**
B. Pulmonary artery
C. Pulmonary vein
D. Bronchogenic artery

Ans A • Pulmonary sequestration refers to a discrete area of lung tissue that lacks any connectionQ to the airway system
• It has abnormal blood supply arising from aorta.

41. What is the most frequent causative agent for acute epiglottitis in children under 5 years of age?

A. **Haemophilus influenzae type B (UPSC-I 10)**
B. Influenza virus type A
C. Para influenza virus type
D. Staphylococcus aureus

Ans A Haemophilus influenzae type B

42. Which of the following is the etiological agent most often associated with epiglottitis in children?

A. Steptococcus pneumoniae
B. **H. influenza type b**
C. Neisseria spp
D. Moraxella catarrhalis

Ans B H. influenza type β

43. The following infections can cause sudden death in infants EXCEPT

A. bronchiolitis (respiratory syncytial virus)
B. infant botulism
C. pertussis
D. **Staph. pneumonia**
E. sepsis

Ans D The following infections can cause sudden death in infants: sepsis, meningitis, encephalitis, brain abscess, hepatitis, pyelonephritis, bronchiolitis (respiratory syncytial virus), infant botulism, and pertussis.

44. Bacterial tracheitis is characterized by all the following EXCEPT

A. Staphylococcus aureus is the most commonly isolated pathogen
B. incidence and severity do not differ by sex
C. mean age is between 5 and 7 yr
D. **considered a primary bacterial illness**
E. more common than epiglottitis in vaccinated populations

Ans D Bacterial tracheitis often follows a viral respiratory infection (especially laryngotracheitis), so it may be considered a bacterial complication of a viral disease, rather than a primary bacterial illness.

45. A commonly encountered risk factor for deep venous thrombosis (DVT) and pulmonary embolism (PE) in the pediatric population is

A. **central venous catheter**
B. hematologic malignancies
C. sickle cell diease
D. nephrotic syndrome
E. antiphospholipid antibody syndrome

Ans A A commonly encountered risk factor for DVT and PE in the pediatric population is the presence of a central venous catheter. The presence of a catheter in a vessel lumen as well as instilled medications can induce endothelial damage and favor thrombus formation.

46. Pectus carinatum is characterized by all the following EXCEPT

A. accounting for 5-15% of congenital chest wall anomalies
B. **females are affected 4 times more often than males**
C. high familial occurrence
D. common association of mild to moderate scoliosis
E. association with mitral valve disease

Ans B Males are affected 4 times more often than females.

Bronchiolitis

47. A 2-month-old infant has had inspiratory stridor since the 1st month of life, but has been otherwise well. Physical examination is unremarkable except for moderate inspiratory stridor and retractions which are worse when the infant is supine or agitated and better when he is prone and quiet. The most likely cause of these findings is:

A. Reactive airway disease
B. **Laryngomalacia**
C. Viral croup
D. An aspirated foreign body INFECTION OF AIRWAYS

Ans B The given clinical picture of stridor in an otherwise well infant which improves in prone position and when the baby is quiet, suggests a diagnosis of laryngomalacia.

48. Most common cause of Bronchiolitis is:

A. **RSV**
B. Adenovirus
C. Hospitalize and treat
D. Mycoplasma

Ans A Acute bronchiolitis
• It is predominantly a viral disease and RSV is responsible for more than 50% of cases
• Other agents include parainfluenza, adenovirus, rhinovirus, Mycoplasma, human metapneumovirus and bocavirus.

49. Treatment of bronchiolitis includes all except:

A. **Macrolides**
B. Humid oxygen
C. Bronchodilator
D. All of above

Ans A Treatment of bronchiolitis:
• Symptomatic; Moist oxygen inhalation remains the mainstay of treatment; Antibiotics have no role fo • Beta-2-adrenergic drugs and ipratropium are not recommended for infants < 6 months
• Ribavirin, shortens the course of illness in infants with congenital heart disease, chronic lung disease & immunodeficiency
• RSV Ig and palivizumab, (monoclonal antibody to the RSV F protein) should be considered for infants < 2 years of age with chronic lung

50. The most common etiological agent for acute bronchiolitis in infancy is:

A. Influenza virus
B. Para influenza virus
C. Rhino virus
D. **Respiratory syncytial virus**

Ans D

51. In bronchiolitis, following is/are seen:

A. **Seen in children 5 months–3 years of age**
B. Caused by Strep. pneumoniae
C. CXR shows bilateral hyperinflation
D. Symptomatic treatment is given
E. Antibiotics should be started

Ans A c. CXR shows bilateral hyperinflation, d. Symptomatic treatment is given with
• Most common cause of acute bronchiolitis is Respiratory Syncitial Virus (RSV)
• Treatment of Bronchiolitis is symptomatic and antibiotics are not indicated.

52. Features of bronchiolitis are:

A. **Caused by RSV**
B. Wheeze present
C. Pulmonary edema common
D. Seen in immunocompromised hosts
E. Common age group is from 5–15 years age

Ans A Caused by RSV, b. Wheeze present

53. Which of the following is/are true about bronchiolitis in children:

A. **Caused by RSV**
B. Hyperinflation of the chest
C. Pleural effusion
D. May lead to bronchial asthma later in life
E. Lymphopenia is seen

Ans A b. Hyperinflation of the chest, d. May lead to bronchial asthma later in life 2047
• X-ray chest in Bronchiolitis shows hyperinflation and infiltrates
• Relationship of acute bronchiolitis to bronchial asthma observed in about 1/4th those with a family history of allergy.

54. Bronchiolitis obliterans (BO) is a rare chronic obstructive lung disease of the bronchioles and smaller airways. An insult to the lower respiratory tract occurs, resulting in fibrosis of the small airways. Of the following, the BEST mean of establishing its diagnosis is

A. chest radiographs
B. pulmonary function tests
C. ventilation-perfusion scans
D. chest CT
E. **open lung biopsy**

Ans E or transbronchial biopsy remains the best means of establishing the diagnosis of BO.

55. The recurrent aspiration of small quantities of gastric, nasal, or oral contents can lead to several clinical presentations, including recurrent bronchitis or bronchiolitis; recurrent pneumonia; atelectasis; wheezing; cough; apnea; and/or laryngospasm. The MOST common underlying problem associated with recurrent pneumonias in hospitalized children is

A. oropharyngeal incoordination
B. esophageal foreign body
C. nasoenteric tube
D. poor oral hygiene
E. bronchopulmonary dysplasia

Ans A is reportedly the most common underlying problem associated with recurrent pneumonias in hospitalized children. In one series of 238 children hospitalized with recurrent pneumonia, 48% were found to have dysphagia as the underlying problem.

Pneumonia

56. Which one of the following life-threatening congenital anomaly in newborn presents with polyhydramnios, aspiration pneumonia, excessive salivation and difficulty in passing a nasogastric tube?

A. Gastroschisis
B. Diaphragmatic hernia
C. Tracheo-esophageal fistula
D. Choanal atresia

Ans C The given clinical picture with polyhydramnios, aspiration pneumonia, excessive salivation and difficulty in passing a nasogastric tube point towards a diagnosis of trachea esophageal fistula.

57. Which of the following clinical presentation is correct for a child with pneumonia?

A. <50/min RR, with age of 2 months
B. >40/min RR, for 2-12 months
C. >40/min RR, for 12-60 months
D. <60/min RR, for 0-2 months

Ans C

58. Pneumatoceles in chest X-ray are characteristically seen in pneumonia due to:

A. Streptococcus pyogenes
B. Haemophilus influenzae
C. Streptococcus pneumoniae
D. Staphylococcus aureus

Ans D Pneumatoceles in chest X-ray are characteristically seen in pneumonia due to Staphylococcus aureus.

59. WHO criteria for hospital admission in pneumonia:

A. High fever
B. Nasal flaring
C. Difficulty in breathing
D. Difficulty in feeding
E. Chest indrawing

Ans C d. Difficulty in feeding Indications of hospitalization in a child with pneumonia
• Age <6 months/Immunocompromised state • Complicated pneumonia
• Sickle cell anemia with acute chest syndrome • Dehydration
• Multiple lobe involvement • Vomiting or inability to tolerate oral fluids
• Toxic appearance • No response to appropriate oral antibiotic therapy
• Moderate to severe respiratory distress • Social factors (e.g., inability of caregivers to administer medications

60. An infant develops cough and fever. Chest X-ray is suggestive of broncho-pneumonia. All of the following viruses can be the causative agents except:

A. Mycobacterium TB
B. Staph aureus
C. Mycobacterium avium intracellulare (MAC)
D. Pneumocystis jiroveci

Ans D Mumps virus Viral causes of pneumonia in children:
• Of the respiratory viruses, respiratory syncytial virus (RSV) and rhinoviruses are the most common;
• Others include influenza virus, parainfluenza, adenoviruses, enteroviruses and human metapneumovirus.

61. A 4-year-old child is admitted in the ward with pneumonia. He develops sudden respiratory distress. The following chest X-ray was recorded. What will you do next?

A. Increase mechanical ventilation
B. Decrease mechanical ventilation
C. Immediate needle thoracostomy
D. Intercostal chest drainage tube insertion

Ans B In the given chest X-ray, we can see hyperinflated left lung (overinflation of lungs can give rise to volutrauma). Hence, decreasing mechanical ventilator settings is the management of choice.

62. The MOST frequent pathogens of pneumonia in children 5 yr and older is

A. Streptococcus pneumoniae
B. Mycoplasma pneumoniae
C. group A streptococci
D. H. influenzae (type b, nontypable)
E. adenovirus

Ans B Streptococcus pneumoniae (pneumococcus) is the most common bacterial pathogen in children 3 wk to 4 yr of age, whereas Mycoplasma pneumoniae and Chlamydophila pneumoniae are the most frequent pathogens in children 5 yr and older.

63. The MOST frequent pathogen of pneumonia in children 4 mo-4 yr is

A. Streptococcus pneumoniae
B. Mycoplasma pneumoniae
C. group A streptococci
D. H. influenzae (type b, nontypable)
E. Respiratory syncytial virus

Ans E The most frequent pathogens of pneumonia in children 4 mo-4 yr (in order of freguency) are Respiratory syncytial virus, other respiratory viruses (parainfluenza viruses, influenza viruses, and adenovirus), S. pneumoniae, H. influenzae (type b, nontypable), Mycoplasma pneumoniae, and group A streptococcus.

64. Recurrent pneumonia is defined as

A. 2 or more episodes in a single year, with radiographic clearing between occurrences
B. 2 or more episodes in a single year, without radiographic clearing between occurrences
C. 3 or more episodes in a single year, with radiographic clearing between occurrences
D. 3 or more episodes in a single year, without radiographic clearing between occurrences
E. 4 or more episodes in a single year

Ans A Recurrent pneumonia is defined as 2 or more episodes in a single year or 3 or more episodes ever, with radiographic clearing between occurrences.

65. The MOST consistent clinical manifestation of pneumonia is

A. subcostal retractions
B. grunting
C. tachypnea
D. fever
E. cough

Ans C is the most consistent clinical manifestation of pneumonia. Increased work of breathing accompanied by intercostal, subcostal, and suprasternal retractions, nasal flaring, and use of accessory muscles is common.

66. The following are indications for admission to a hospital in children with pneumonia

A. age <6 mo
B. sickle cell anemia
C. multiple lobe involvement
D. moderate to severe respiratory distress
E. vomiting

Ans B with acute chest syndrome.

Asthma

67. A 10-month-old child presents with an abnormal inspiratory sound; mother complaints that the sound increases with cry, and decreases when the child sleeps in prone position. What would be the probable diagnosis?

A. Croup
B. Acute epiglottitis
C. Laryngitis
D. Laryngomalacia

Ans D

68. An 11-month-old child presents with complaints of respiratory distress. On examination there is bilateral crepitation and wheezing. Which of the following is the most likely cause?

A. Pneumonia
B. Adenovirus
C. RSV
D. Rhinovirus

Ans C The clinical features are suggestive of Acute Bronchiolitis, for which the most common causative agent is RSV.

69. A 6-week baby presents with cough and cold for the past 3 days. Respiratory rate is 48/min. On examination, patient is febrile with wheezing but no chest indrawing. Which of the following is not true?

A. Child has pneumonia
B. Antibiotics are not required
C. Wheezing should be treated
D. Fever should be treated

Ans A This child does not have fast breathing; So there is no pneumonia; Hence antibiotics are not required;

70. Humanized monoclonal antibody that binds IgE to prevent its binding to the high affinity IgE receptor and blocking IgE mediated allergic response and inflam- mation in Bronchial Asthma is:

A. Palivizumab
B. Natalizumab
C. Omalizumab
D. Etilizumab

Ans C with An anti-IgE preparation, Omalizumab (Xolair), is approved by the FDA for use as an add-on therapy in children ≥12 yr who have moderate to severe allergic asthma that is difficult to control.

71. Which one of the following values is not a feature of Acute Severe Asthma?

A. Heart rate more than 110/min
B. PEF of 60 to 70% of expected
C. Pulsus paradoxus
D. PaO of less than 8 kPa

Ans B Note: 1 kPa = 7.5 mm Hg; So 8 kPa = 60 mm Hg Grading of severity of Asthma exacerbation Characteristics Mild Moderate Severe Heart rate <100 100-120 >120 PEF (Peak expiratory flow) ≥70% or response <40% 40-69% <40% Pulsus paradoxus Absent May be present Often present PaO Normal ≥60 mm Hg (8 KPa) <60 mm Hg (8 KPa)

72. A child of less than one year should use the following device for asthma treatment:

A. MDI with Spacer
B. MDI with Mask
C. MDI with Spacer with Mask
D. MPI with Mask

Ans C

73. Which of the following is NOT recommended in immediate treatment of acute severe asthma?

A. Oxygen supplementation
B. High doses of inhaled b –adrenoreceptor agonists
C. Systemic corticosteroids
D. **Intravenous aminophylline**

Ans D Immediate treatment of acute severe asthma includes oxygen supplementation, high doses of inhaled b2 – adrenoreceptor agonists, systemic corticosteroids and intravenous aminophylline

74. In a child with exercise induced asthma, which is done:

A. Prophylaxis with steroids
B. Prophylaxis with beta agonist
C. **Prophylaxis with Montelukast**
D. Breathing exercise

Ans C Leukotriene receptor antagonists like Montelukast and Zafirlukast, have bronchodilator and targeted anti-inflammatory properties and reduce exercise-, aspirin-, and allergen-induced bronchoconstriction.

75. Most common mode of treatment of a 1 year child with asthma is:

A. **Inhaled short acting β agonist**
B. Oral short acting theophylline
C. Oral ketotifen
D. Leukotriene agonist

Ans A Inhaled short acting b agonist 2 Most commonly beta agonists (bronchodilators) are used in the treatment of Asthma in all age groups.

76. A 3-year-old boy is brought to the casualty by his mother Questions with progressive shortness of breath for 1 day. The child has history of bronchial asthma. On examination, the child is blue, gasping and unresponsive. What will you like to do first:

A. Intubate
B. Administer 100% Oxygen by mask
C. **Ventilate with bag and mask with oxygen**
D. Administer nebulized Salbutamol CYSTIC FIBROSIS

Ans C As the child is cyanotic, gasping and unresponsive, there is respiratory failure, only oxygen supplementation will not help; So he must be ventilated. So bag and mask ventilation with oxygen is to be used.

77. Risk factors for persistent wheezing include all the following EXCEPT

A. parental history of asthma
B. **paternal smoking**
C. persistent rhinitis (apart from acute upper respiratory tract infections)
D. eczema at <1 yr of age
E. frequent episodes of wheezing during infancy

Ans B Multiple studies have tried to predict which early wheezers will go on to have asthma in later life. Risk factors for persistent wheezing include parental history of asthma and allergies, maternal smoking, persistent rhinitis (apart from acute upper respiratory tract infections), eczema at <1 yr of age, and frequent episodes of wheezing during infancy.

78. Cardiovascular causes of wheezing include

A. right atrial enlargement
B. **left atrial enlargement**
C. right ventricular enlargement
D. left ventricular enlargement
E. coarctation of aorta

Ans B Cardiovascular causes of wheezing include dilated chambers of the heart including massive cardiomegaly, left atrial enlargement, and dilated pulmonary arteries.

79. The etiology of wheezing in bronchopulmonary dysplasia (BPD) may be due to all the following EXCEPT

A. lower airway inflammation
B. bronchial smooth muscle irritation
C. **intraluminal exudate**
D. bronchial smooth muscle hypertrophy
E. airway malacia

Ans C The etiology of wheezing in BPD may be lower airway inflammation, bronchial smooth muscle irritation, bronchial smooth muscle hypertrophy, and airway malacia.

Tuberculosis

80. All are common causes of stridor except:

A. Foreign body in larynx and trachea
B. Laryngomalacia
C. **Atrophic laryngitis**
D. Multiple papillomas of larynx

Ans C Atrophic Laryngitis usually manifests with hoarseness & irritable cough; Rest 3 conditions can manifest as stridor.

81. Fast breathing in a child less than 2 months is defined as respiratory rate more than:

A. 40 breaths per minute
B. 50 breaths per minute
C. **60 breaths per minute**
D. 70 breaths per minute

Ans C

82. Wheeze in children is caused by:

A. **Foreign body**
B. Gastroesophageal reflux disease
C. Bronchial asthma
D. Epiglottitis
E. Laryngomalacia

Ans A b. Gastroesophageal reflux disease, c. Bronchinal asthma and e. Laryngomalacia Wheeze is a musical and continuous sound that originates from oscillations in narrowed airways. Important causes of Wheezing in Infancy: Category Causes Infection Viral/Tuberculosis Asthma Very common cause Anatomic abnormalities • Malacia of larynx, trachea, and/or bronchi • Mediastinal lymphadenopathy/mass/tumor • Tracheoesophageal fistula • Lung malformations • Laryngeal cleft • Congenital heart diseases with L → R shunt

83. **Foreign bodies are often placed in the nose by small children and developmentally delayed children. Of the following, the MOST common presenting clinical symptom is**

A. **mucopurulent nasal discharge**
B. foul nasal odor
C. epistaxis
D. nasal obstruction
E. mouth breathing

Ans A Presenting clinical symptoms include history of insertion of foreign bodies (86%), mucopurulent nasal discharge (24%), foul nasal odor (9%), epistaxis (6%), nasal obstruction (3%), and mouth breathing (2%).

84. **The MOST common congenital laryngeal anomaly that produces stridor is**

A. vocal cord paralysis
B. **laryngomalacia**
C. congenital subglottic stenosis
D. congenital laryngeal web
E. congenital subglottic hemangioma

Ans B is the most common congenital laryngeal anomaly and the most common cause of stridor in infants and children. Sixty percent of congenital laryngeal anomalies in children with stridor are due to laryngomalacia.

85. **The MOST common form of pulmonary malignancy in children is**

A. bronchial carcinoid
B. adenoid cystic carcinoma
C. mucoepidermoid carcinoma
D. **metastatic lesions**
E. pulmonary blastoma

Ans D are the most common forms of pulmonary malignancy in children; primary processes include Wilms tumor, osteogenic sarcoma, and hepatoblastoma.

86. **Acute respiratory insufficiency is often the MOST prominent clinical manifestation of**

A. Duchenne muscular dystrophy
B. spinal muscular atrophy
C. congenital myotonic dystrophy
D. myasthenia gravis
E. **Guillain-Barre syndrome**

Ans E Acute respiratory insufficiency is often the most prominent clinical manifestation of several acute neuromuscular disorders, such as high-level spinal cord injury, poliomyelitis, Guillain-Barre syndrome, and botulism. Although much more insidious in its clinical course, respiratory dysfunction constitutes the leading cause of morbidity and mortality in progressive neuromuscular disorders (e.g., Duchenne muscular dystrophy, spinal muscular atrophy, congenital myotonic dystrophy, myasthenia gravis, and...

Pleural Effusion & Empyema

87. **A child with fever and barky cough presented to emergency at 3 am. His respiratory rate is 36/min, stridor only on coughing, fever 39 degrees celsius. No other abnormality, What is the next logical step?**

A. **Dexamethasone**
B. Racemized epinephrine
C. Do nothing, just reassure
D. Give surfactant

Ans A Nelson's 20/e p 2032-2034 This child is suffering from Croup (Laryngotracheobronchitis): mild variety. Oral Dexamethasone is beneficial, even in mild croup; Antibiotics are not indicated in croup

88. **A mother in a village has brought her 1 month baby with a complaint of fever to the PHC. You find that the baby's respiratory rate is 69/minute. The baby is able to feed, does not have chest indrawing or stridor. There is no history of convulsions. According to the RCH program, the baby's illness is classified as:**

A. Severe pneumonia
B. Very severe disease
C. No pneumonia: cough or cold
D. **Pneumonia**

Ans D Pneumonia 39 above

89. **A child is brought to the pediatric OPD with fever of 24 hours duration. History reveals 3 episodes of chest infection and passage of foul smelling stools. The most probable diagnosis is:**

A. **Cystic fibrosis**
B. Maple syrup urine disease
C. Bilirubin conjugation defect
D. Criggler Najar syndrome

Ans A Recurrent chest infections with foul-smelling, greasy stools (steatorrhea) in a child suggest cystic fibrosis with pancreatic insufficiency.

90. **A 16-year-male presents with rapidly developing bilateral pleural effusion. The following chest radiograph was taken in the emergency. All are correct interpretation about this radiograph except:**

A. Pneumothorax is present
B. Bilateral ICD tubes are present
C. **Mediastinum is shifted**
D. Left sided pleural effusion is present

Ans C Mediastinum does not appear shifted in the given chest X-ray

91. Regarding congenital lobar emphysema (CLE), the following are true EXCEPT

A. familial occurrence has been reported
B. usually no cause of CLE can be identified
C. usually become apparent in the neonatal period
D. many cases are diagnosed by antenatal ultrasonography
E. left upper lobe is the most affected site

Ans B In 50% of cases, a cause of CLE can be identified. Congenital deficiency of the bronchial cartilage, external compression by aberrant vessels, bronchial stenosis, redundant bronchial mucosal flaps, and kinking of the bronchus caused by herniation into the mediastinum have been described as leading to bronchial obstruction and subsequent CLE and commonly affects the left upper lobe.

92. The following conditions are associated with increased intrathoracic pressure causing pneumothorax in children EXCEPT

A. asthma
B. pneumatocele
C. bronchiolitis
D. cystic fibrosis
E. airway foreign body

Ans B lung abscess, and bronchopleural fistula are infectious causes of pneumothorax in children. 51(E). Hydrothorax is most often associated with cardiac, renal, or hepatic disease. It can also be a manifestation of severe nutritional edema and hypoalbuminemia. Rarely, it results from vascular obstruction by neoplasms, enlarged lymph nodes, pulmonary embolism, or adhesions. It may occur from a ventriculoperitoneal shunt or peritoneal dialysis and has been reported in congenital parvovirus B19 infection.

93. Physical examination and an upright, posteroanterior radiograph with subsequent measurement of the angle of curvature (Cobb technique) remain the gold standard for assessment of scoliosis. Scoliosis is defined when curves

A. ≥5 degrees
B. ≥10 degrees
C. ≥15 degrees
D. ≥20 degrees
E. ≥25 degrees

Ans B Curves >10 degrees define the presence of scoliosis.

Bronchiectasis

94. An 18 months child came with LRTI with Respiratory rate of 50/min. Child was not lethargic but irritable and has stridor. Examination reveals bilateral rhonchi. SpO 2 is 88%. What is next best management?

A. Give O by mask with child on the lap of the mother, 2 along with beta agonist and adrenaline nebulization
B. Examine throat, give O by mask along with beta agonist 2 nebulization with one dose of steroid
C. Immediate intubation and ventilate the child and establish IV/IO access
D. Urgent referral to tertiary care

Ans B Examine throat, give O by mask along with beta agonist nebulization with one dose of steroid p 376, 398; Nelson's 20/e p 2032-2034 This child has croup along with hypoxia (severe croup); So O2 supplementation along with nebulization with Adrenaline (beta agonist) and single dose Dexamethasone (steroid) are indicated.

95. When mother complains that baby is more comfortable and breathes better when held against shoulder, what important clinical problem do you suspect?

A. Nose block
B. Psychological anxiety or insecurity
C. Aerophagy with abdominal fullness
D. Orthopnea

Ans D Baby being comfortable in a vertical position indicates orthopnea.

96. Pediatric airway differs from adult in having:

A. Large tongue
B. Short epiglottis
C. Narrowest part at glottis
D. Larynx in lower position

Ans A Differences between pediatric and adult airway are:
• Infants and small children have a disproportionately large head and short neck.
• Their tongues are relatively large with respect to the mandible
• Their larynx is more anterior and cephalad in comparison to the adult.
• They are obligate nasal breathers & find it more difficult to switch to mouth breathing when necessary.
• The epiglottis is relatively long 'floppy' & U-shaped.
• The narrowest part of the airway is at the level of the cricoid cartilage

97. Common causes of childhood epistaxis include the following EXCEPT

A. epistaxis digitorum
B. foreign bodies
C. significant gastroesophageal reflux into the nose
D. dry air
E. allergic rhinitis

Ans C Common causes of nosebleeds from the anterior septum include digital trauma, foreign bodies, dry air, and inflammation, including upper respiratory tract infections, sinusitis, and allergic rhinitis. Young infants with significant gastroesophageal reflux into the nose rarely present with epistaxis secondary to mucosal inflammation.

98. The MOST common etiology of pulmonary infiltrates with eosinophilia (PIE) is

A. simple pulmonary eosinophilia
B. acute eosinophilic pneumonia
C. chronic eosinophilic pneumonia
D. allergic bronchopulmonary aspergillosis
E. parasitic Infections

Ans E The most common etiology of PIE syndromes includes parasite infections and drug reactions.

99. In the developed world, the MOST common cause of clinically significant bronchiectasis is

A. primary ciliary dyskinesia
B. foreign body aspiration
C. aspiration of gastric contents
D. immune deficiency syndromes
E. cystic fibrosis

Ans E In the developed world, cystic fibrosis is the most common cause of clinically significant bronchiectasis.

100. **The MOST common complaint in patients with bronchiectasis is**

A. cough and production of copious purulent sputum
B. hemoptysis
C. fever
D. anorexia
E. poor weight gain

Ans A The most common complaints in patients with bronchiectasis are cough and production of copious purulent sputum. Younger children may swallow the sputum. Hemoptysis is seen with some frequency. Fever can occur with infectious exacerbations. Anorexia and poor weight gain may occur as time passes.

Cystic Fibrosis (Respiratory)

101. **A 3 weeks male child born to a Primi mother, is presenting with noisy breathing sound. Child is afebrile, sleeping and feeding well and on clinical examination, he is normal. Management protocol include:**

A. Begin IV antibiotics
B. Reassure and give saline nasal drop
C. Give racemic epinephnne Questions
D. Order for a chest X-ray
E. Give some decongestant

Ans B In this case, the baby appears to have no evidence of respiratory distress like tachypnea or retractions. The baby does not have any pneumonia, but only some coryza. So the appropriate management here is Reassure and give saline nasal drops.

102. **Under IMNCI programme in outpatient health facility, all of the following are danger signs except:**

A. Convulsions
B. Difficult breathing
C. Vomiting
D. Lethargy

Ans B

103. **Which of the following is seen in cystic fibrosis?**

A. Low sweat chloride levels
B. Elevated sweat chloride levels
C. Low sweat sodium levels
D. Elevated sweat potassium levels

Ans B Because the function of sweat gland duct cells is to absorb rather than secrete chloride, salt is not retrieved from the isotonic primary sweat as it is transported to the skin surface; chloride and sodium levels are consequently elevated. So sweat chloride is usually ≥ 60 mEq/L in cystic fibrosis.

104. **Which of the following exocrine glandular ducts are not obstructed in cystic fibrosis?**

A. Pancreas
B. Lung
C. Sweat gland
D. All of above

Ans C Excessive loss of salt in the sweat occurs in Cystic fibrosis a. Cystic fibrosis High grade fever and chest infection along with passage of foul smelling stools suggests systemic disease. In cystic fibrosis because of pancreatic exocrine insufficiency, foul smelling stool is common.

105. **Most common organism associated with cystic fibrosis:**

A. Pseudomonas aeruginosa (non mucoid)
B. Burkholderia cepacia
C. Pleisomonas
D. Aeromonas

Ans A In cystic fibrosis, there is high prevalence of airway colonization with Staphylococcus aureus, Pseudomonas aeruginosa and Burkholderia cepacia complex. Commonest organism to cause pulmonary infection in cystic fibrosis is Peudomonas aeruginosa, which is initially non-mucoid, later mucoid colonies form.

106. **Which of the following statements about cystic fibrosis (CF) is not true:**

A. Autosomal recessive, disorder
B. Abnormality in CFTR which leads to defective calcium transport
C. Mutation in cystic fibrosis transport regulator
D. CFTR protein has 1,480 amino acids

Ans B The cyclic adenosine monophosphate (cAMP)–mediated apical membrane conductance of Cl associated with the CF transmembrane regulator (CFTR) does not function in CF epithelia. So, there is defective transport of Chloride and not calcium in cystic fibrosis.

107. **A child presents with respiratory distress and failure to thrive. His sweat chloride levels were estimated 35 mEq/L and 41 mEq/L. What is next best test to do cystic fibrosis for diagnosis:**

A. Nasal transmembrane potential difference
B. DNA analysis ΔF 508 mutation
C. CT chest
D. 72 hours fecal fat estimation

Ans A

108. **Cystic fibrosis is associated with:**

A. Meconium ileus
B. Short bowel syndrome
C. Hirschsprung disease
D. Congenital pyloric stenosis

Ans A Intestinal manifestation of CF include meconium ileusQ, distal intestinal obstruction syndrome (DIOS), rectal prolapse.

109. **Infant with cystic fibrosis (CF) are likely to develop:**

A. Meconium ileus
B. Loose motions
C. Vomiting
D. Constipation MISCELLANEOUS

Ans A Meconium ileus MISCELLANEOUS

110. **Cystic fibrosis (CF) can be presented with all the following EXCEPT**

A. salt depletion
B. pansinusitis
C. **rectal polyposis**
D. pancreatitis
E. cholelithiasis

Ans C CF is responsible for most cases of exocrine pancreatic insufficiency in early life and is the major cause of severe chronic lung disease in children. It is also responsible for many cases of salt depletion, nasal polyposis, pansinusitis, rectal prolapse, pancreatitis, cholelithiasis, and insulin-dependent hyperglycemia. CF may manifest as failure to thrive and, occasionally, as cirrhosis or other forms of hepatic dysfunction.

111. **The earliest pathologic lesion in the lung of a patient with cystic fibrosis is**

A. **bronchiolitis**
B. bronchitis
C. bronchiolectasis
D. bronchiectasis
E. interstitial lung disease

Ans A The earliest pathologic lesion in the lung is that of bronchiolitis (mucous plugging and an inflammatory response in the walls of the small airways); with time, mucus accumulation and inflammation extend to the larger airways (bronchitis). 35 (D).

112. **Arterial blood gas study in cystic fibrosis shows**

A. hypernatraemic hypochloraemic metabolic alkalosis
B. hyponatraemic hypochloraemic metabolic acidosis
C. hyponatraemic hyperchloraemic metabolic alkalosis
D. **hyponatraemic hypochloraemic metabolic alkalosis**
E. hypernatraemic hyperchloraemic metabolic acidosis

Ans D Chronic salt loss through sweat in cystic fibrosis produces a hyponatremic, hypochloremic metabolic alkalosis (a 'pseudo-Bartter' picture).

113. **The MOST common presenting feature of cystic fibrosis is**

A. failure to thrive
B. **persistent respiratory symptoms**
C. abnormal stools
D. meconium ileus
E. hepatobiliary disease

Ans B Failure to thrive 37.5%; persistent respiratory symptoms 45.6%; abnormal stools 28.8%; meconium ileus 19.9%; hepatobiliary disease 1.2%

114. **Regarding cystic fibrosis, all the following are true EXCEPT**

A. delayed sexual development
B. more than 95% of males are azoospermic
C. **sexual function is generally impaired**
D. high incidence of inguinal hernia
E. diminished female fertility rate

Ans C More than 95% of males are azoospermic because of failure of development of Wolffian duct structures, but sexual function is generally unimpaired.

115. **The finding of the following pathogen on culture of the lower airways (sputum) strongly suggests a diagnosis of cystic fibrosis**

A.
B. gram-negative rods
C. nontuberculous mycobacterial species
D. Mycoplasma
E. **S. aureus**

Ans E The finding of S. aureus or P. aeruginosa on culture of the lower airways (sputum) strongly suggests a diagnosis of CF. In particular, mucoid forms of P. aeruginosa are often recovered from CF lungs. B. cepacia recovery also suggests CF. A wide range of other organisms are frequently recovered, particularly in advanced lung disease; they include a variety of gram-negative rods, fungi, and nontuberculous mycobacterial species.

116. **All the following inhalation therapies in cystic fibrosis are true EXCEPT**

A. β-agonists may decrease PaO2 acutely
B. human recombinant DNase improves pulmonary function
C. N-acetylcysteine is toxic to ciliated epithelium
D. nebulized hypertonic saline improves mucociliary clearance
E. **aerosolized antibiotics are often used when the airways are colonized with S. aureus**

Ans E Aerosolized antibiotics are often used when the airways are colonized with Pseudomonas as part of daily therapy. Aerosolized tobramycin, TOBI, used as a suppressive therapy (on 1 month, off 1 month) may reduce symptoms, improve pulmonary function, and alleviate the need for hospitalization.

Foreign Body Aspiration

117. **A girl child with fever, cough, dyspnea with X-ray showing right lower lobe patchy consolidation, for which treatment was given. After 8 weeks symptom improved but X-ray showed more dense consolidation involving the whole of the right lower lobe. What is the next best line of investigation?**

A. **Bronchoscopy**
B. Culture from nasopharynx
C. Barium esophagogram
D. Allergic skin test

Ans A Persistence of symptoms in the form of x-ray showing more dense consolidation involving the whole of the right lower lobe after treatment indicates some localized pathology in that area like foreign body. This will need bronchoscopy for further management and evaluation.

118. **A 10-month-child weighing 5 kg and with 65 cm length, presents with cough and cold. He was found to have a respiratory rate of 48 per minute with no retractions, grunting, cyanosis. There is no history of convulsions. Which is true?**

A. **No pneumonia, only cough and cold**
B. Child has pneumonia
C. Severe pneumonia
D. Very severe disease

Ans A Cut-off for fast breathing: Age <2 months 2 – 12 months 12 – 60 months Respiratory Rate ≥ 60/min ≥ 50/min ≥ 40/min So this baby does not have fast breathing; So this child has No pneumonia, only cough and cold, even though the child is malnourished (wt = 5 kg at 10 months against expected of 9 kg).

119. Level of trachea bifurcation in pediatric patient is:

A. Transient, migratory pulmonary infiltrations
B. Fibrosis in the pulmonary apices
C. Fibrosis in the base of one or both lungs
D. Miliary mottling

Ans A T2

120. In a child with foreign body in bronchus, next step is:

A. Rigid bronchoscopy
B. Chest X-ray
C. Flexible endoscopy
D. Direct laryngoscopy Chapter 18: Pediatric Respiratory Disorders 411 Answers

Ans A Treatment of choice in a child with foreign body in bronchus is Rigid bronchoscopy. 18: Pediatric Respiratory Disorders 417 syndrome" and is most commonly caused Answers e.g., ARDS, pneumonia, pulmonary edema) are with if accompanied by cyanosis or bradycardia. M
• Minimum urine osmolality (urine diluting ability) is well developed, even in preterm babies & is same as adults (40-50 mOsm/kg) I de n t i fy th e u n d e rul ye i sn tg ik oidn n e y a b n or m a li t y i n t h e f Qe tu s s h o w n b 1 e l o w : a.

121. All the following conditions are associated with false-positive sweat test results

A. eczema
B. anorexia nervosa
C. hypothyroidism
D. Klinefelter syndrome
E. malnutrition

Ans E Dilution, malnutrition, edema, insufficient sweat quantity, hyponatremia, and cystic fibrosis transmembrane conductance regulator (CFTR) mutations with preserved sweat duct function are causes of false negative sweat test.

122. One of the following is a rare cause of hydrothorax in children

A. cardiac disease
B. renal disease
C. hepatic disease
D. severe nutritional edema
E. vascular obstruction by neoplasms

Ans E Cardiac, renal, and hepatic disease along with severe hypoproteinemia are common causes of hydrothorax in children; obstruction of lymphatics/vessels by neoplasm is comparatively rare.

Sleep-Disordered Breathing

123. Bronchitis in children is caused by:

A. H. influenzae
B. RSV
C. Mycoplasma
D. EBV
E. Influenza virus

Ans E • Nonspecific bronchial inflammation is termed bronchitis; It is usually viral in origin, with cough as a prominent feature;
• It is most commonly caused by Influenza virus.

124. Most dangerous sign in LRTI in children is:

A. Abdominal breathing
B. Chest retraction
C. Grunting
D. Tachypnea

Ans C Of the above signs, grunting is most dangerous, as it indicates widespread lung involvement. About Grunting:
• It is produced by expiration against a partially closed glottis
• It is attempt to maintain positive airway pressure during expiration for as long as possible. dna Interpreting the Clinical Signs of Respiratory Disease Extrathoracic airway Intrathoracic extrapulmonary Intrapulmonary airway Parenchymal Sign obstruction airway obstruction obstruction pathology Tachypnea + + ++ ++++

125. Apnea in infant is cessation of breathing for:

A. > 10 sec
B. > 20 sec
C. > 30 sec
D. > 40 sec

Ans B In infants, apnea is defined as cessation of breathing for >20 sec, or any duration

126. Of the following, the MOST common childhood cause of nasal polyposis is

A. chronic sinusitis
B. allergic rhinitis
C. Samter triad
D. Kartagener syndrome
E. cystic fibrosis

Ans E is the most common childhood cause of nasal polyposis and should be suspected in any child <12 yr old with nasal polyps, even in the absence of typical respiratory and digestive symptoms; as many as 30% of children with cystic fibrosis acquire nasal polyps.

127. The following are clinical manifestations of primary ciliary dyskinesia (PCD) EXCEPT

A. recurrent pneumonia
B. chronic otitis media
C. chronic pansinusitis
D. dextrocardia
E. retinitis pigmentosa

Ans D Left-right laterality defects are found in PCD; 50% of patients have situs inversus totalis with transposition of the thoracic and abdominal organs.

128. Of the following, the LEAST likely cause of hemothorax in children is

A. chest trauma
B. intrathoracic neoplasms
C. costal exostoses
D. blood dyscrasias
E. rupture of an aneurysm

Ans E is unlikely during childhood.

Acute Respiratory Failure

129.A neonate presents with respiratory distress with enlargement of left upper lobe of lung and mediastinal shift towards the right. What is the most likely diagnosis?

A. Cong lobar emphysema
B. Alpha 1 antitrypsin deficiency
C. Pneumonia
D. Cystic fibrosis

Ans A Congenital Lobar Emphysema (CLE) Definition Over-distension of 1 or more lobes of lung Characteristics Left upper lobe is the most common site; It is over-distended with mediastinal shift to opposite side;

130.Two months old child presented with fever, respiratory distress, recurrent apnea followed by bradycardia and cyanosis. What is the probable diagnosis?

A. Congenital heart block
B. Whooping cough
C. Tracheobronchiomalacia
D. Epiglottitis

Ans B Whooping cough in Infants:
• Infants < 3 months of age do not display the classic stages of Whooping cough; Catarrhal phase is usually unnoticed,
• In Paroxysmal stage: – A well-appearing young infant begins to choke, gasp, gag and flail the extremities, with face reddened – Cough may not be prominent, especially in the early phase and Whoop infrequently occurs – Apnea & cyanosis can follow a coughing paroxysm; Apnea may be the only symptom

131. A 3-month-old child has moderate fever and non-productive cough and mild dyspnea. After a course of antibiotic, the condition of the child improved transiently but he again develops high fever, productive cough and increased respiratory distress. CXR shows hyperlucency and PFT shows obstructive pattern. Most probable diagnosis is:

A. Alveolar microlithiasis
B. Postviral syndrome
C. Follicular bronchitis
D. Bronchiolitis obliterans

Ans D Bronchiolitis obliterans (BO) A chronic obstructive lung disease of bronchioles and smaller airways, that results from an insult to the lower What is it? respiratory tract leading to fibrosis of small airways
• After the initial insult, inflammation affecting terminal bronchioles, respiratory bronchioles and alveolar ducts can result in the obliteration of the airway lumen Pathogenesis
• Epithelial damage resulting in abnormal repair is characteristic of BO

132.When can one diagnose acute respiratory distress in a child?

A. Bronchiectasis
B. Differential pulse
C. Sinusitis
D. Situs inversus

Ans D All of the above Diagnostic criteria of ARDS in children: Berlin definition criteria Timing Within 1 wk of a known clinical insult or new or worsening respiratory symptoms Chest X-ray or Bilateral opacitires not fully explained by effusions, lobar/lumg collapse, or nodules. (Illustrative clinical cases and tomography scan chest X-ray have been provided) Origin of edema Respiratory failure not fully explained by cardiac failure or fluid overload. Need objective assessment (e.g.,

133.Most common cause of ARDS in children is:

A. Aspiration
B. Injury
C. Severe pneumonia with sepsis
D. DIC

Ans C Pneumonia, sepsis and aspiration are the most common cause of ARDS and ALI in children

134.In mechanical ventilation of a newborn with ARDS, the end tidal volume is kept at?

A. 5 mL/kg
B. 7 mL/kg
C. 10 mL/kg
D. 15 mL/kg

Ans A Diseases associated with decreased time constants (decreased static compliance, best treated with small tidal volume of 5-6 mL/kg.

Mechanical Ventilation

135.A 6-month-old baby is brought with H/O increasing difficulty in breathing of 2 days duration. On examination, the baby is afebrile but B/L wheeze is audible; CXR shows B/L hyperinflation of the lungs with normal WBC count, the diagnosis is:

A. Bronchiolitis
B. Asthma
C. Ch. Bronchitis
D. Pneumonia
E. Foreign body bronchus

Ans A The given clinical picture with presence of dyspnea, wheeze, hyperinflation of lungs on chest X-ray and absence of fever and leukocytosis, suggest a diagnosis of Bronchiolitis.

136.Lecithin–Sphingomyelin ratio is used for:

A. Lung maturity
B. Brain maturity
C. Kidney maturity

Ans A By 35 wk of gestation, the lecithin: sphingomyelin (L: S) ratio averages about 2:1, indicative of lung maturity.

137. The pulmonary vascular resistance and the pulmonary artery pressure to the systemic arterial resistance, from 3 mo after birth through childhood and adolescence is approximately

A. 15% of the systemic values
B. 25% of the systemic values
C. 35% of the systemic values
D. 45% of the systemic values
E. 55% of the systemic values

Ans A The PVR is ~50% of the systemic arterial resistance 3 days after birth. In the next several wk after birth as pulmonary arterial musculature in the tunica media involutes, there is a further decline in PVR and therefore in pulmonary artery pressure. Two to 3 mo after birth, the PVR and the pulmonary artery pressure are ~15% of the systemic values, a relationship that exists through childhood and adolescence.

138. **Persistent cough may need to be sought beyond the lungs, because cough receptors also reside in the following regions EXCEPT**

A. pharynx
B. paranasal sinuses
C. stomach
D. **nose**
E. external auditory canal

Ans D Because cough receptors also reside in the pharynx, paranasal sinuses, stomach, and external auditory canal, the source of a persistent cough may need to be sought beyond the lungs.

139. **Pathologic findings of pulmonary hemosiderosis in association with cow's milk hypersensitivity include all the following EXCEPT**

A. **elevations of IgA**
B. peripheral eosinophilia
C. alveolar deposits of IgG, IgA
D. alveolar deposits of C3
E. high titers to cow's milk protein

Ans A Pathologic findings have included elevations of IgE.

140. **All the following are true in chylothorax EXCEPT**

A. fluid may be clear
B. fluid triglyceride level is >110 mg/dL
C. pleural fluid:serum triglyceride ratio is >1.0
D. pleural fluid:serum cholesterol ratio is <1.0
E. **cells are primarily B lymphocytes**

Ans E The cells are primarily T lymphocytes.

19

CHAPTER 19

Disorders of Kidney and Urinary Tract



WHAT THIS CHAPTER COVERS

- UTI & Vesicoureteral Reflux
- Nephrotic Syndrome
- Acute Glomerulonephritis
- RPGN & Crescentic GN
- IgA Nephropathy
- Henoch-Schönlein Purpura Nephritis
- HUS & TTP
- Acute Kidney Injury
- Chronic Kidney Disease
- Renal Tubular Disorders
- RTA (Renal Tubular Acidosis)
- Nephrogenic DI & Diabetes Insipidus
- Wilms Tumor
- Posterior Urethral Valves
- Hypospadias & Epispadias
- Cryptorchidism
- Hydrocele & Hernia
- Enuresis
- Renal Transplantation
- Dialysis

259 MCQs **20** subtopics

19.1 Renal Physiology & Assessment in Children

- **Nephrogenesis completes at 34–36 weeks**; ~1 million nephrons per kidney; preterms have fewer nephrons (↑ later CKD/HTN risk).
- **GFR**: ~20 mL/min/1.73 m² at birth (term) → **adult values (~120) by 1–2 years**.
- **Normal urine output**: neonates/infants **1–3 mL/kg/h**, children 1–2 mL/kg/h; **oliguria < 1 mL/kg/h (infants) / < 0.5 mL/kg/h (children)**; **anuria < 0.5 mL/kg/h in neonates (or no urine 24 h)**.
- **Schwartz formula (bedside, 2009)**: **eGFR = 0.413 × height (cm) / serum creatinine (mg/dL)**. Old: k = 0.33 (preterm), 0.45 (term infant), 0.55 (children & adolescent girls), 0.7 (adolescent boys).
- **Proteinuria**: normal **< 4 mg/m²/h** or **spot urine protein/creatinine (UPCR) < 0.2 mg/mg** (< 0.5 in < 2 y); **nephrotic-range > 40 mg/m²/h or UPCR > 2 mg/mg** (or dipstick 3+/4+); significant 4–40 mg/m²/h.
- **Orthostatic proteinuria** – benign, common in adolescents (normal first-morning sample).
- **Haematuria**: **> 5 RBC/HPF** in centrifuged urine; **dysmorphic RBCs & RBC casts → glomerular**; **cola/smoky urine → glomerular**; bright red with clots → lower tract.
- Red urine without RBCs – **haemoglobinuria, myoglobinuria** (dipstick +, no RBC), beetroot, rifampicin, urates (pink in nappy – neonates), porphyria, Serratia.

19.2 Urinary Tract Infection (UTI) & Vesicoureteric Reflux (VUR)

- **E. coli (80–90%)** > Klebsiella, Proteus (**boys, phosphate stones – alkaline urine**), Enterococcus, Pseudomonas; **S. saprophyticus** (adolescent girls).
- **Male > female in first 3–6 months** (uncircumcised boys, posterior urethral valves); **female > male after 1 year**.
- Infants: **fever without focus**, poor feeding, irritability, vomiting, FTT, prolonged jaundice; older – dysuria, frequency, flank pain.
- **Diagnosis**: urine culture – **gold standard**:

Method	Significant growth
Suprapubic aspiration (most reliable)	**Any growth of gram-negative bacilli** (> 1000 CFU/mL gram-positive)
Catheter sample	**≥ 50,000 CFU/mL** (IAP: ≥ 10^4–5×10^4)
Clean-catch midstream	**≥ 10^5 CFU/mL** of a single organism
Bag samples	Useful only if negative (high contamination)

- Dipstick: **nitrite (specific, needs 4 h bladder incubation; not in infants who void often)**, **leucocyte esterase (sensitive)**; microscopy – pyuria ≥ 10 WBC/mm³ (uncentrifuged), bacteria.
- **Upper (pyelonephritis)**: fever > 38.5 °C, flank pain, toxicity, ↑ CRP; **DMSA scan – IOC for acute pyelonephritis & renal scarring** (scarring at 4–6 months after UTI).

TREATMENT (IAP/ISPN 2023)

- **Infants < 3 months / toxic / vomiting** → **IV ceftriaxone/cefotaxime (or amikacin)** → switch to oral after 48–72 h when afebrile; total **10–14 days** (7–10 for older child febrile UTI).
- **Older, non-toxic febrile UTI** → **oral cefixime / amoxicillin-clavulanate** 7–10 days.
- **Cystitis (afebrile)** → oral antibiotic 3–5 days (nitrofurantoin, co-trimoxazole, cephalexin).
- Repeat culture not needed if clinical response.

Imaging after first UTI (ISPN 2023 / AAP)

- **USG kidney–urinary bladder (KUB) – all children with first febrile UTI** (esp. < 2 y).
- **MCU (micturating cystourethrogram) – IOC for VUR & posterior urethral valves** – indicated if **abnormal USG, recurrent febrile UTI, atypical UTI (non-E. coli, poor flow, raised creatinine, septicaemia, abdominal/bladder mass)**, or as per age-based protocols (ISPN: infants < 1 y with febrile UTI). Done after infection is treated.

- **DMSA scan – renal scars (4–6 months after UTI)**; MAG3/DTPA renogram – obstruction & differential function.

Vesicoureteric reflux

Grade (International)	Description
I	Reflux into **ureter only** (non-dilated)
II	Into ureter, pelvis & calyces, **no dilatation**
III	**Mild–moderate dilatation** of ureter & pelvicalyceal system; mild blunting of fornices
IV	Moderate dilatation & tortuosity; **obliteration of fornices** but papillary impressions maintained
V	**Gross dilatation & tortuosity; loss of papillary impressions**

- **Primary VUR** (short intravesical ureter – **MC, familial (siblings 30%)**); secondary – **PUV, neurogenic bladder**, bladder–bowel dysfunction.
- Lower grades (I–III) resolve spontaneously in most (80% grade I–II by 5 y); **reflux nephropathy → MCC of hypertension & a leading cause of CKD in children**.
- **Management**: treat **bladder–bowel dysfunction**, timely voiding, constipation; **continuous antibiotic prophylaxis (CAP) – grade III–V VUR (esp. < 1 y) / recurrent UTI: nitrofurantoin 1–2 mg/kg or co-trimoxazole (2 mg/kg TMP) at bedtime**; cephalexin < 3 m; circumcision in infant boys with high-grade VUR; **surgery – endoscopic injection (Deflux) or ureteric reimplantation (Cohen)** for breakthrough febrile UTI on prophylaxis, new scars, grade V.

19.3 Congenital Anomalies of Kidney & Urinary Tract (CAKUT)

Anomaly	Key points
Antenatal hydronephrosis (ANH)	**MC antenatally detected anomaly** (APD ≥ 4 mm 2nd tri / ≥ 7 mm 3rd tri); MC cause – **transient/ physiological**, then **PUJ obstruction**, VUR; postnatal USG **after 48–72 h** (dehydration underestimates)
Pelviureteric junction (PUJ) obstruction	**MCC of obstructive hydronephrosis/ANH**; **boys, left side**; flank mass, pain (Dietl crisis), haematuria, UTI; **DTPA/MAG3 diuretic renogram (obstructive curve, split function < 40%) – IOC**; Rx **Anderson–Hynes dismembered pyeloplasty**
Posterior urethral valves (PUV)	**MCC of bladder outlet obstruction & obstructive uropathy leading to CKD in boys**; antenatal – **bilateral hydroureteronephrosis, thick-walled bladder ("keyhole sign")**, oligohydramnios → pulmonary hypoplasia; postnatal – **poor/dribbling urinary stream, palpable bladder**, UTI, urinary ascites, AKI; **MCU – IOC (dilated posterior urethra, valve)**; Rx **bladder catheterisation → cystoscopic valve ablation**; lifelong follow-up (valve bladder, CKD)
Multicystic dysplastic kidney (MCDK)	**MC cystic renal disease of newborn / MC abdominal mass in newborn (with hydronephrosis)**; non-functioning kidney with non-communicating cysts ("bunch of grapes"); **contralateral VUR/ PUJO**; involutes spontaneously; conservative
ARPKD (PKHD1, fibrocystin)	**Bilateral enlarged echogenic kidneys ("bright kidneys")**, oligohydramnios, Potter sequence, **hypertension**, **congenital hepatic fibrosis → portal hypertension**, Caroli
ADPKD (PKD1/PKD2)	Usually adult; children – cysts, HTN, haematuria; **berry aneurysms**, liver cysts, MVP
Renal agenesis	Bilateral – **Potter sequence**, incompatible with life; unilateral – compensatory hypertrophy; MRKH (Müllerian) in girls
Horseshoe kidney	**Fusion of lower poles** – isthmus held under **inferior mesenteric artery**; **Turner syndrome, trisomy 18**; PUJ obstruction, stones, Wilms
Duplex kidney/ureter	**Weigert–Meyer rule – upper-pole ureter opens lower & medial (ectopic, obstructed – ureterocele); lower-pole ureter refluxes (VUR)**; ectopic ureter in girls → **continuous dribbling with normal voiding**
Ureterocele	Cystic dilatation of distal ureter ("cobra head" on IVU)

Anomaly	Key points
Prune belly (Eagle–Barrett) syndrome	**Triad: deficient abdominal wall musculature, bilateral cryptorchidism, urinary tract dilatation (megacystis, megaureter)**; boys; pulmonary hypoplasia
Bladder exstrophy–epispadias complex	Exposed bladder, **widened pubic symphysis**, epispadias
Urethral meatal stenosis, phimosis, hypospadias	See Surgery

19.4 Nephrotic Syndrome

- **Definition**: **nephrotic-range proteinuria (UPCR ≥ 2 mg/mg or ≥ 40 mg/m²/h or dipstick ≥ 3+) + hypoalbuminaemia (< 2.5–3 g/dL) + oedema** ± hyperlipidaemia (> 200 mg/dL cholesterol).
- **Idiopathic NS – 90% in children**; **minimal change disease (MCD) – MC (80% in < 6 y)**; **FSGS** (MC cause of steroid-resistant NS); MPGN, membranous (rare – HBV, SLE).
- Age **2–6 y**, boys 2:1; **periorbital puffiness (morning) → generalised oedema/anasarca, ascites**, pleural effusion, frothy urine; BP usually normal (MCD); **no gross haematuria** (microscopic in 20%).
- **Biochemistry**: **↓ albumin, ↑ cholesterol & TG, ↓ IgG, ↑ α2-globulin & fibrinogen, ↓ antithrombin III**, **normal complement (C3) in MCD**, pseudohyponatraemia, **↓ total calcium** (ionised normal).
- **MCD pathology**: **normal glomeruli on LM**, **IF negative**, **EM – effacement (fusion) of podocyte foot processes**; **loss of charge selectivity** (selective proteinuria – albumin).

TREATMENT OF FIRST EPISODE (ISPN 2021/KDIGO)

Prednisolone 2 mg/kg/day (60 mg/m²/day; max 60 mg) in single/divided doses × 6 weeks, then 1.5 mg/kg (40 mg/m²) on alternate days × 6 weeks (total 12 weeks), then stop (no taper needed per ISPN). **~90% of MCD respond (remission) within 4 weeks.** Supportive – salt restriction, **no fluid restriction unless severe oedema**, normal protein diet, albumin infusion + furosemide only for severe symptomatic oedema; treat infections; **vaccinate (PCV, PPSV23, varicella when in remission & off high-dose steroid)**.

Term (ISPN)	Definition
Remission	**UPCR < 0.2 or nil/trace dipstick for 3 consecutive days**
Relapse	**Dipstick ≥ 3+ (or UPCR ≥ 2) for 3 consecutive days** after remission
Frequent relapses (FRNS)	**≥ 2 relapses in first 6 months after initial response, or ≥ 3 (≥ 4 in some) in any 12 months**
Steroid dependence (SDNS)	**2 consecutive relapses during steroid therapy or within 14 days of stopping**
Steroid resistance (SRNS)	**No remission after 6 weeks of daily prednisolone 2 mg/kg** (ISPN: 4 wks + confirmation) → **renal biopsy**
Late resistance	Resistance after initial response

- **Relapse Rx**: prednisolone 2 mg/kg/day until remission (3 days) then 1.5 mg/kg alternate days × 4 weeks.
- **FRNS/SDNS** – steroid-sparing agents: **levamisole (2–2.5 mg/kg alternate days)**, **cyclophosphamide (oral 2 mg/kg/day × 12 weeks; gonadal toxicity – cumulative dose < 168 mg/kg)**, **mycophenolate mofetil (MMF)**, **calcineurin inhibitors (tacrolimus, ciclosporin – nephrotoxicity)**, **rituximab** (anti-CD20) for refractory SDNS.
- **SRNS**: **renal biopsy (FSGS MC)**, **genetic testing (NPHS1, NPHS2 – podocin, WT1)** – genetic forms don't respond to immunosuppression; Rx **calcineurin inhibitors (tacrolimus) – first line** + **ACEi/ARB (antiproteinuric)**; recurrence of FSGS after transplant (circulating factor).

Complications of nephrotic syndrome

- **Infections – MCC death**: **spontaneous bacterial peritonitis (S. pneumoniae – MC)**, cellulitis, sepsis, pneumonia (↓ IgG, ↓ factor B, steroids).

- **Thromboembolism (hypercoagulability)**: **renal vein thrombosis (flank pain, haematuria)**, cerebral venous sinus thrombosis, pulmonary embolism; due to ↓ ATIII, ↑ fibrinogen, ↑ platelets, haemoconcentration, diuretics.
- **Hypovolaemia/AKI** (aggressive diuretics), hypothyroidism (loss of TBG), **vitamin D deficiency/hypocalcaemia**, growth retardation, steroid toxicity (Cushingoid, cataract, hypertension, osteoporosis, behavioural).

Biopsy indications in NS
Age < 1 y (congenital/infantile) or > 12 y
Steroid resistance
Persistent hypertension, gross haematuria, low C3, persistent renal failure
Before calcineurin inhibitors (baseline) / suspicion of secondary cause (SLE)

- **Congenital nephrotic syndrome** (onset < 3 months): **Finnish type (NPHS1 – nephrin)** – **large placenta (> 25% of birth weight)**, ↑ maternal AFP, massive proteinuria from birth, steroid-resistant; Rx albumin infusions, ACEi, nephrectomy → dialysis → transplant. Other causes: congenital syphilis, CMV, toxoplasmosis, Denys–Drash (WT1).

19.5 Acute Glomerulonephritis (AGN) / Nephritic Syndrome

- **Post-streptococcal glomerulonephritis (PSGN) – MC cause of AGN in children** (5–12 y); follows **pharyngitis (latency 1–2 weeks)** or **pyoderma/impetigo (3–6 weeks)** with **nephritogenic GAS strains** (M types 12 – throat; 49 – skin).
- **Nephritic features**: **haematuria (cola/smoky urine), oedema (periorbital), hypertension, oliguria, azotaemia** ± proteinuria (sub-nephrotic).
- **Labs**: RBC casts (pathognomonic of glomerular bleeding), **↓ C3 (low for up to 6–8 weeks – returns to normal by 8 weeks)**, normal C4 (alternative pathway), **↑ ASO (throat), ↑ anti-DNase B (skin – best marker after pyoderma)**, streptozyme.
- **Pathology** (biopsy rarely needed): **LM – diffuse proliferative GN (hypercellular glomeruli, neutrophils)**; **IF – "starry-sky/lumpy-bumpy" granular IgG & C3**; **EM – subepithelial "humps"**.
- **Complications**: **hypertensive encephalopathy (seizures, headache, vision loss – PRES)**, **CHF/pulmonary oedema (volume overload)**, AKI.
- **Rx**: **fluid & salt restriction**, **diuretics (furosemide) for oedema/HTN** – first-line; antihypertensives (amlodipine, nifedipine); **penicillin to eradicate strep (doesn't alter course)**; dialysis if severe AKI/hyperkalaemia/fluid overload. **Prognosis excellent (> 95% recover)**; microscopic haematuria may persist 1–2 years.
- **Biopsy indications**: normal C3 at onset, **C3 persistently low > 8–12 weeks**, nephrotic-range proteinuria, **rapidly rising creatinine (RPGN)**, systemic features.

Low C3 GN	Normal C3 GN
PSGN (C3 ↓, C4 N), **MPGN/C3 glomerulopathy**, **lupus nephritis (C3 ↓, C4 ↓)**, shunt nephritis/IE, cryoglobulinaemia	**IgA nephropathy**, HSP nephritis, Alport, ANCA vasculitis, anti-GBM, thin basement membrane

19.6 IgA Nephropathy, HSP Nephritis & Hereditary Nephritis

- **IgA nephropathy (Berger)**: **MC primary GN worldwide**; **gross haematuria 1–2 days after URTI (synpharyngitic)**, recurrent; normal C3; **mesangial IgA deposits**; Rx ACEi for proteinuria, steroids if heavy proteinuria; Oxford MEST-C.
- **HSP (IgA vasculitis) nephritis**: haematuria ± proteinuria within weeks of rash; **renal involvement determines long-term prognosis**; nephrotic/nephritic or > 50% crescents → steroids ± immunosuppression.
- **Alport syndrome (hereditary nephritis)**: **type IV collagen – COL4A5 (X-linked, 80%)**, COL4A3/A4 (AR/AD); **persistent microscopic haematuria → proteinuria → ESRD (males by 20–30 y)**; **bilateral sensorineural hearing loss (high-frequency)**, **anterior lenticonus (pathognomonic)**, dot-and-fleck retinopathy; **EM –**

splitting/lamellation ("basket-weave") of GBM; Rx ACEi (delays progression), transplant (anti-GBM disease post-transplant risk).

- **Thin basement membrane nephropathy** (benign familial haematuria) – COL4A3/4 heterozygous; isolated microscopic haematuria, normal function.
- **RPGN** – crescents > 50%: anti-GBM (Goodpasture – linear IF), immune-complex (lupus, PSGN, IgA), **pauci-immune (ANCA – granulomatosis with polyangiitis/MPA)**; Rx **pulse methylprednisolone + cyclophosphamide/rituximab ± plasmapheresis**.
- **Lupus nephritis** – class IV (diffuse proliferative) MC & most severe; **wire-loop lesions**, full-house IF; Rx steroids + MMF/cyclophosphamide.

19.7 Haemolytic Uraemic Syndrome (HUS) & TTP

- **Triad: microangiopathic haemolytic anaemia (schistocytes, ↑ LDH, DAT negative) + thrombocytopenia + AKI**; **MC cause of AKI in young children (developed countries)**.
- **STEC-HUS (typical, 90%)**: **Shiga toxin-producing E. coli O157:H7** (also Shigella dysenteriae type 1 – India); **prodrome of bloody diarrhoea 5–10 days before**; **antibiotics & antimotility agents ↑ risk**; Rx **supportive – fluids, transfusion (avoid platelets unless bleeding), dialysis (~50%)**; good prognosis (mortality < 5%).
- **Pneumococcal HUS** – after pneumonia/empyema (**neuraminidase exposes T-antigen – DAT positive**); avoid plasma.
- **Atypical HUS (complement-mediated)**: **factor H/I/MCP mutations or anti-factor H antibodies (common in Indian children – DEAP-HUS)**, no diarrhoea, relapsing, poor prognosis; **low C3**; Rx **plasma exchange + steroids/MMF (anti-FH antibody)**, **eculizumab (anti-C5)**.
- **TTP**: **ADAMTS13 deficiency** (congenital Upshaw–Schulman or acquired autoantibody); **pentad – MAHA, thrombocytopenia, neurological symptoms, fever, renal involvement**; Rx **plasma exchange**, caplacizumab.

19.8 Acute Kidney Injury (AKI)

- **KDIGO/pRIFLE**: ↑ serum creatinine ≥ 0.3 mg/dL in 48 h or ≥ 1.5× baseline in 7 days, or urine output < 0.5 mL/kg/h for 6 h.
- Causes: **pre-renal (MC – dehydration from diarrhoea, shock, sepsis)**, renal (**ATN – MC intrinsic**, HUS, AGN, nephrotoxins – aminoglycosides, NSAIDs, contrast, amphotericin; rhabdomyolysis, tumour lysis), post-renal (PUV, bilateral obstruction, stones).

Index	Pre-renal	Intrinsic (ATN)
Urine osmolality	**> 500 mOsm/kg** (neonate > 400)	**< 350** (≈ 300)
Urine Na	**< 20 mEq/L** (neonate < 30)	**> 40** (neonate > 60)
FeNa	**< 1%** (neonate < 2.5%)	**> 2%** (neonate > 3%)
Urine/plasma creatinine	> 40	< 20
BUN/creatinine	> 20	< 10–15
Urine sediment	Bland, hyaline casts	**Muddy-brown granular casts**, epithelial casts
Response to fluid	Improves	No

- **Indications for dialysis ("AEIOU")**: **refractory Acidosis, Electrolyte (K > 6.5–7 with ECG changes), Intoxications, fluid Overload (pulmonary oedema, refractory HTN), Uraemia (encephalopathy, pericarditis, bleeding)**; **peritoneal dialysis – preferred in infants/young children**; CRRT in unstable patients.

19.9 Chronic Kidney Disease (CKD)

- **GFR < 60 mL/min/1.73 m² for > 3 months** (or kidney damage markers); stages 1–5 (ESKD < 15).

- **Causes in children: CAKUT (obstructive uropathy – PUV, reflux nephropathy, dysplasia/hypoplasia) – MCC in < 10 y**; **glomerulopathies (FSGS) – MCC in > 10 y**; hereditary (ARPKD, nephronophthisis, Alport, cystinosis).
- **Features & complications**: **growth failure (MC & characteristic)**, **anaemia (normocytic – EPO deficiency)**, **CKD–mineral bone disorder (renal osteodystrophy – secondary hyperparathyroidism, rickets)**, metabolic acidosis, hyperkalaemia, hypertension, **polyuria/enuresis (tubular disease)**, uraemic bleeding, pericarditis, encephalopathy, dyslipidaemia, developmental delay.
- **Management**: nutrition (calories), **sodium bicarbonate**, phosphate binders (calcium carbonate, sevelamer), **active vitamin D (calcitriol/alfacalcidol)**, **ESA (erythropoietin/darbepoetin) + iron (target Hb 11–12)**, **GH therapy** for short stature, ACEi/ARB (proteinuria, HTN), vaccines (Hep B – double dose), avoid nephrotoxins; **renal replacement therapy – pre-emptive kidney transplant (best)**, PD, HD.
- **Nephronophthisis** (NPHP genes, AR ciliopathy): **MC genetic cause of ESRD in first 3 decades**; **polyuria/polydipsia (concentrating defect), anaemia, growth failure, normal-sized kidneys with corticomedullary cysts, bland urine (no proteinuria/haematuria)**; **Senior–Løken** (+ retinitis pigmentosa), Joubert (molar tooth sign).

19.10 Renal Tubular Disorders

Renal tubular acidosis (normal anion-gap, hyperchloraemic metabolic acidosis)

Feature	Distal RTA (type 1)	Proximal RTA (type 2)	Type 4 (hyperkalaemic)
Defect	**↓ H^+ secretion in collecting duct** (H^+-ATPase, AE1)	**↓ HCO_3^- reabsorption in PCT** (↓ threshold)	**Aldosterone deficiency/resistance**
Urine pH	**> 5.5 always (can't acidify)**	**< 5.5 when acidotic** (threshold reached)	< 5.5
Serum K	**Low**	Low (worse with alkali)	**High**
Nephrocalcinosis/ stones	**Yes (hypercalciuria, hypocitraturia)**	No	No
Rickets/ osteomalacia	Yes	**Yes (with Fanconi)**	No
Associations	Primary (SLC4A1, ATP6V1B1 – **with SNHL**), Sjögren, SLE, **amphotericin**, obstructive uropathy	**Fanconi syndrome**, cystinosis, Wilson, Lowe, carbonic anhydrase inhibitors, ifosfamide	**CAH, hypoaldosteronism, pseudohypoaldosteronism, obstructive uropathy**, ACEi, K-sparing diuretics, heparin
Urine anion gap	**Positive**	Negative (variable)	Positive
$FeHCO_3$ (after loading)	< 5%	**> 15%**	5–10%
Alkali needed	**1–3 mEq/kg/day**	**Large (5–15 mEq/kg/day)**	Low; treat hyperkalaemia, fludrocortisone

Fanconi syndrome (generalised proximal tubular dysfunction)

- **Glycosuria (normal blood sugar), generalised aminoaciduria, phosphaturia (hypophosphataemic rickets), bicarbonaturia (proximal RTA), uricosuria, hypokalaemia, polyuria, low-molecular-weight proteinuria**.
- Causes: **cystinosis (MCC inherited in children – corneal crystals; Rx cysteamine)**, **Lowe syndrome (oculocerebrorenal – OCRL, X-linked: congenital cataract, glaucoma, hypotonia, ID)**, **Wilson, galactosemia, hereditary fructose intolerance, tyrosinaemia type 1**, GSD I, mitochondrial, Dent disease (CLCN5); acquired – **heavy metals (lead, cadmium), outdated tetracyclines, ifosfamide, cisplatin, tenofovir, valproate**, aminoglycosides.

Other tubulopathies

- **Bartter, Gitelman, Liddle, pseudohypoaldosteronism** – see Fluids & Electrolytes.

- **Nephrogenic diabetes insipidus**: **AVPR2 (X-linked, MC)**, AQP2 (AR); **polyuria, polydipsia, hypernatraemic dehydration, fever, FTT** in infants, hydronephrosis; **no response to desmopressin**; acquired – **lithium, hypercalcaemia, hypokalaemia, obstructive uropathy, amphotericin**. Rx **thiazides + low-sodium diet, indomethacin, amiloride**, free water.
- **Cystinuria, Hartnup** – see IEM.
- **Primary hyperoxaluria type 1** (AGXT): nephrocalcinosis, oxalate stones, ESRD, systemic oxalosis; Rx pyridoxine (some), **lumasiran (RNAi)**, liver-kidney transplant.

19.11 Urolithiasis, Enuresis & Others

- **Stones in children**: **calcium oxalate (MC)**; metabolic risk – **hypercalciuria (MC metabolic abnormality)**, hypocitraturia, hyperoxaluria, cystinuria, uric acid (Lesch–Nyhan, tumour lysis); **struvite (Proteus UTI, staghorn)**; **bladder stones in Indian children – endemic (ammonium urate, low-protein cereal diet)**; drugs – topiramate, furosemide (**nephrocalcinosis in preterm**), ceftriaxone (biliary/renal sludge). **Non-contrast CT/USG**; Rx hydration, potassium citrate, ESWL/ureteroscopy/PCNL.
- **Nocturnal enuresis**: **involuntary voiding at night in a child ≥ 5 years**, ≥ 2/week for 3 months (DSM-5: ≥ 2/week × 3 months or distress); **primary (never dry – 80%, familial – one parent 44%, both 77%)** vs **secondary (relapse after ≥ 6 months dry – UTI, diabetes mellitus/insipidus, constipation, stress, OSA, sexual abuse)**; monosymptomatic vs non-monosymptomatic (daytime symptoms).
  - Mechanisms: **nocturnal polyuria (↓ nocturnal ADH)**, ↓ bladder capacity/overactivity, **high arousal threshold**.
  - **Rx**: reassurance, **motivational therapy (star chart)**, bladder training, evening fluid restriction, treat constipation; **enuresis alarm – most effective long-term (TOC; ↓ relapse)**; **desmopressin (DDAVP) – first-line drug** (tablets/melts at bedtime; rapid effect, high relapse; **hyponatraemia – restrict fluids 1 h before & 8 h after**); anticholinergics (oxybutynin) for overactive bladder; **imipramine – 3rd line (cardiotoxic in overdose)**.
- **Neurogenic bladder** – spina bifida (MMC), sacral agenesis, tethered cord; **clean intermittent catheterisation (CIC) + anticholinergics (oxybutynin)**, botulinum toxin, augmentation.
- **Vincent's curtsy** – squatting with heel pressing perineum (detrusor overactivity in girls).
- **Hypertension** in children – MC secondary cause renal parenchymal (reflux nephropathy) – see Cardiology.

19.12 Renal Transplantation in Children

- **Best RRT for ESKD in children**; pre-emptive transplant ideal; living related donors common; **weight ≥ 8–10 kg**; recurrence risk – **FSGS (30–50%)**, atypical HUS, primary hyperoxaluria, MPGN.
- Complications: rejection, CMV/EBV (**PTLD**), **BK polyomavirus nephropathy (↑ with potent immunosuppression)**, non-adherence (adolescents – MCC late graft loss), growth & hypertension.

Practice MCQs · Disorders of Kidney and Urinary Tract

259 questions · correct option highlighted in green · cover the answer strip to self-test

UTI & Vesicoureteral Reflux

1. Most common anomaly of upper urogenital tract is:

A. Uretero pelvic junction stenosis
B. Ectopic urethral opening
C. Ureterocele
D. Ectopic ureter

Ans A Ureteropelvic junction (UPJ) obstruction is the most common obstructive lesion in childhood and usually is caused by intrinsic stenosis.

2. False regarding nephritic syndrome

A. Edema
B. Proteinuria <3.5g/day
C. Hypoalbuminemia
D. Hypertension

Ans C

3. A six years old male baby presents to a hospital with Questions recurrent gross hematuria for 2 years. There is no history of burning micturition or pyuria. Urine routine examination demonstrated no pus cells and urine culture was sterile. Serum C3 were normal. What is the most probable diagnosis?

A. Wilm's tumor
B. IgA nephropathy
C. Post streptococcal glomerulonephritis
D. Urinary tract infection

Ans B Recurrent painless gross hematuria with a normal C3 and sterile urine in a school-age child is the classic presentation of IgA nephropathy.

4. A 25-year-old primigravida with history of 20 weeks gestation comes for routine antenatal examination. The fetal ultrasonogram reveals unilateral hydronephrosis. The obstruction is likely to be at the level of:

A. Vesicourethral junction
B. Ureters 109.
C. Ureteropelvic junction
D. Urethra

Ans C

5. The treatment of choice for primary grade V VUR involving both kidneys in a 6 month old boy is:

A. Endoscopic injection of polyteflon at ureteric orifices
B. Ureteric reimplantation
C. Bilateral ureterostomy
D. Prophylactic antibiotics
E. Proteinuria occurs A child presented with intermittent episodes of left side flank pain. Ultrasonography reveals large hydro- nephrosis with dilated renal pelvis and cortical thinning with a normal ureter. Kidney differential function was observed to be 19%, which of the following is the best management:

Ans A Antibiotic prophylaxis , Latest VUR treatment guidelines by AUA & EUA, 2015; There is a lack of consensus regarding management of VUR in children, Latest treatment guidelines by the American Urological Association (AUA) and European Urological Association (EUA): Latest Updates I. Continuous Antibiotic prophylaxis (CAP): According to AUA, High-risk groups who would benefit from CAP included those 1 year of age, dilating VUR [grades 3-5] and/or a history of febrile UTI, those with bladder and bowel dysfunction;

6. A 4 months old female child presents with grade IV VUR, without dilation of urinary bladder. The treatment of choice is:

A. Septran + follow up
B. Re-implantation of ureter
C. Injection of collagen at ureteric orifices
D. Bilateral ureterostomy URINARY TRACT INFECTIONS

Ans A

7. A 4 months old infant with UTI was treated for 14 days with IV cefotaxime. What is the next step in management?

A. USG only
B. USG + MCU
C. USG + DMSA
D. USG + MCU + DMSA

Ans A EUA guidelines 2014 AAP recommends that in a typical first-episode of UTI, initial imaging should be ultrasonography of the kidneys, ureters, and bladder. VCUG is indicated if the ultrasound study is abnormal, the patient has atypical features, or after a recurrent febrile UTI. As per EUA (European Association of Urology), Renal and bladder USG is advised in all children with febrile UTI to exclude dilatation or anomalies of the upper & lower urinary tract, if no improvement is seen within 24 h

8. An 8 years old boy during a routine check up is found to have E. coli 1,00,000 cc/ml on a urine culture. The urine specimen was obtained by mid stream clean catch void. The child is asymptomatic. Which is the most appropriate next step in the management?

A. Treat as an acute episode of urinary tract infection
B. **No therapy**
C. Prophylactic antibiotics for 6 months
D. Administer long term alkalinizer

Ans B Revised Statement on Management of UTI. Indian Pediatrics, Vol 48, 2011 Criteria for diagnosis of UTI on midstream urine sample: Colony count > 105/ml Asymptomatic Bacteriuria:
• It is a form of chronic pyelonephritis characterized by destruction of renal parenchyma and the presence of granulomas, abscesses and collection of lipid laden foamy macrophages (foam cells)
• Rare in children; more common in adults; Focal form is more common in children;

9. True about UTI:

A. Common in female infants
B. If two episodes of UTI in female of 7 years occur, then cystometric evaluation needed.
C. If two episodes of UTI in male of 5 years occur, then cystometric evaluation needed.
D. **Urine should be obtained by suprapubic aspiration for culture in infants**
E. 70% of neonates with UTI have VUR

Ans D Urine for culture in infants should ideally be obtained by suprapubic aspiration (or catheterization), as bag specimens are prone to contamination.

10. All the following are risk factors for urinary tract infection EXCEPT

A. **male gender**
B. tight clothing (underwear)
C. pinworm infestation
D. poor toilet training
E. voiding dysfunction

Ans A Female gender and uncircumcised male.

11. According to the clinical guidelines, urine sample for diagnosis of urinary tract infection in children 2-24 mo should be from

A. an adhesive collection bag
B. mid-stream sample
C. urine bag
D. **catheterized sample**
E. all of the above

Ans D

12. Regarding vesicoureteral reflux VUR (the retrograde flow of urine from the bladder to the ureter and kidney). All the following are true EXCEPT

A. it is usually congenital
B. it may cause hypertension in children
C. **the mean age at VUR resolution is 10yr**
D. severity is graded according to voiding cystourethrogram
E. reimplantation of the ureters corrects the condition

Ans C The mean age at VUR resolution is 6yr.

13. Vesicoureteral reflux is usually discovered during evaluation for a UTI, which of the following is TRUE

A. **autosomal dominant inheritance**
B. males are mainly affected
C. antenatal hydronephrosis mainly seen in females
D. constipation is a rare association
E. average age at diagnosis is less than 1 year

Ans A Idiopathic VUR appears to be an autosomal dominant inherited trait with variable penetrance, 80% are females, primary VUR also may be discovered during evaluation for antenatal hydronephrosis, in this select population, 80% of affected children are male. Bladder and bowel dysfunction (constipation) may be present in 50% of children with reflux and the average age at diagnosis is 2-3 yr.

14. Regarding the voiding cystourethrogram (VCUG) for diagnosis of vesicoureteral reflux (VUR). Of the following, the MOST appropriate statement is

A. low-pressure VUR is occurring during voiding
B. high-pressure VUR is occurring during bladder filling
C. high-pressure VUR is significantly less likely to resolve spontaneously
D. contrast VCUG is significantly exposed to less radiation than radionuclide cystogram
E. **contrast VCUG provides more anatomic information**

Ans E VUR occurring during bladder filling is termed low-pressure VUR; VUR during voiding is termed high-pressure VUR.VUR in children with low-pressure VUR is significantly less likely to resolve spontaneously than in children who exhibit only high- pressure VUR. Radiation exposure during a radionuclide cystogram is significantly less than that from a contrast VCUG.

15. Struvite calculi (secondary to urinary tract infections) are MOST likely caused by

A. Escherichia coli
B. **Proteus**
C. Klebsiella
D. Pseudomonas
E. Staph. aurous

Ans B Caused by urea-splitting organisms (most often Proteus spp., and occasionally Klebsiella spp., Escherichia coli, Pseudomonas spp., and others).

Nephrotic Syndrome

16. Malformations of the following organ system of the fetus are found to be most commonly associated with single umbilical artery:

A. Central nervous system
B. Cardiovascular
C. **Genitourinary**
D. Skeletal

Ans C

17. Best method of estimation of amount of proteinuria in a 2 year child with nephrotic syndrome is

A. Dipstick testing
B. 24 hr urine protein
C. **Spot urine sample for protein/creatinine ratio**
D. Microalbuminuria

Ans C Though 24 hr urine for protein and creatinine, is more accurate, but due to difficulty in collecting 24 hour urinary sample in children, Spot urine sample for protein/creatinine ratio is preferred.

18. Which of the following is not a feature of Minimal change disease?

A. Nephrotic syndrome
B. **Hypertension**
C. Affects children
D. No abnormality detected on light microscopy

Ans B Important features of minimal change idiopathic nephrotic syndrome are the absence of hypertension and gross hematuria

19. In minimal change disease, which of the following is seen?

A. Immunodeposits in glomerulus
B. Immunodeposits in mesangium
C. Immunodeposits in blood vessels
D. **No immunodeposits**

Ans D In minimal change disease:
• Light microscopy: Glomeruli appear normal: No Ig/complement deposits, GBM: normal
• Immunofluorescence microscopy: No electron-dense deposits
• Electron microscopy: Diffuse effacement of foot processes of podocytes ("podocytopathy").

20. True statement about minimal change disease

A. Effacement of foot process on light microscopy
B. **Severe hypoalbuminemia**
C. Non selective proteinuria
D. Cyclosporine is the first line of treatment

Ans B

21. Nephrotic range proteinuria is defined as protein excretion of more than?

A. 10 mg/m2/hr
B. 30 mg/m2/hr
C. 20 mg/m2/hr
D. **40 mg/m2/hr**

Ans D • Microalbuminuria: Excretion of 30-300 mg/dayQ of albumin in urine or 30-300 mg albumin/gm of creatinine in urineQ
• Macroalbuminuria: Excretion of 300-3500 mg/day of albumin in urineQ
• Nephrotic range proteinuria: Excretion of > 3.5 gm/dayQ of albumin in urine or 40 mg/m2/hr

22. True about minimal change disease:

A. **Peak incidence is between 2 and 4 years of age**
B. Renal failure occur in adolescent
C. Steroid not very effect
D. Selective proteinuria

Ans A Peak incidence is between 2 and 4 years of age, d. Selective proteinuria

23. Most common cause of nephrotic syndrome in children

A. Membranous GN
B. **Minimal change disease**
C. PSGN
D. RPGN

Ans B

24. A child with nephrotic syndrome following an episode of diarrhea presented with acute kidney injury with a creatinine of 4.5 mg/dl. All of the following are possible reasons except?

A. Renal vein thrombosis
B. Diarrhoea induced water depletion
C. Frusemide
D. **Steroid induced diabetes**

Ans D Steroid-induced diabetes Diarrhoea induced water depletion (dehydration) is a common cause of Acute Renal Failure in children. Renal vein thrombosis can also predispose to AKI in a child with diarrhea and dehydration

25. Which is seen in nephrotic syndrome:

A. **Low serum calcium**
B. Raised AT-III
C. Low lipid
D. Platelet activation

Ans A In nephrotic syndrome there is hypocalcemia and secondary hyperparathyroidism due to loss of cholecalciferol binding protein; Secondary hyperparathyroidism causes hypophosphatemia by increasing renal phosphate excretion.

26. Most common cause of nephrotic syndrome in children:

A. **Minimal change disease**
B. IgA nephropathy
C. Mesangial GN
D. FSGN

Ans A

27. Patients with minimal change disease are at risk of?

A. **Spontaneous bacterial peritonitis**
B. Sepsis
C. DVT
D. Atherosclerosis
E. Renal Carcinoma

Ans A b. Sepsis, c. DVT, d. Atherosclerosis

28. A 7-year-old child with steroid dependent nephrotic syndrome has developed corticosteroid toxicity and posterior subcapsular cataracts. Which of the following is the best alternative for the treatment of the patient:

A. **Cause steroid resistant nephrotic syndrome**
B. Nephrin is a key component of the slit diaphragm
C. Coded by NPHS-1 gene
D. Symptom occur only after 1st month of age
E. Autosomal dominant pattern

Ans A Levamisole Levamisole has been successfully used in patients with Frequently relapsing Nephrotic syndrome (FRNS) & Steroid Dependent Nephrotic syndrome (SDNS); If relapses continue despite Levimasole therapy, Cyclophosphamide is recommended.

29. **Edema in nephrotic syndrome is mainly due to:**

A. Sodium and water retention
B. Increased venous pressure
C. **Hypoalbuminemia**
D. Hyperlipidemia

Ans C and Harrison 19th ed/pg 253 Edema is the most common presenting symptom of children with nephrotic syndrome. Causes of Edema: In Nephrotic Syndrome: (primarily hypoalbuminemia)
• Primary alteration in the nephrotic syndrome is a diminished colloid oncotic pressure due to losses of large
• quantities (3.5 g/day) of protein into the urine
• With severe hypoalbuminemia and reduced colloid osmotic pressure, the sodium and water that are retained cannot be restrained within

30. **A child comes with steroid resistant nephrotic synd rome secondary to FSGS, not responding to methyl prednisolone. What should be given next?**

A. Oral cyclophosphamide
B. **Oral cyclosporine**
C. Oral mycophenolate
D. IV Cyclophosphamide

Ans B FSGS:
• Renal biopsies show FSGS in about 30–35% children with non minimal nephrotic syndrome
• It is the most common cause of acquired kidney failureQ necessitating renal replacement therapies in children
• Most patients with FSGS have initial or late corticosteroid resistance; Cyclophosphamide is generally ineffective;
• The best results in FSGS have been obtained with the use of calcineurin inhibitors like cyclosporine

31. **Idiopathic nephrotic syndrome is associated with the following except:**

A. Focal segmental glomerulosclerosis
B. Minimal change disease
C. **Membranoproliferative glomerulonephritis**
D. Mesangioproliferative glomerulonephritis

Ans C is more often linked to secondary causes (e.g., autoimmune or infectious) rather than idiopathic nephrotic syndrome, unlike minimal change disease, FSGS, and mesangioproliferative GN.

32. **Spontaneous Bacterial Peritonitis in Nephrotic Syndrome is most commonly caused by:**

A. E. coli
B. Pseudomonas
C. Salmonella
D. **Streptococcus pneumoniae**

Ans D Infections in children with Nephrotic syndrome
• Especially susceptible to cellulitis, spontaneous bacterial peritonitis (SBP) and bacteremia
• Due to urinary losses of IgG and complement factors, impaired opsonization of microorganisms and increased risk of infection with encapsulated bacteria, especially pneumococcus
• SBP presents with fever, abdominal pain and peritoneal signs; Pneumococcus is the most frequent cause of SBP

33. **Hypercoagulation in nephrotic syndrome is caused by**

A. **Loss of antithrombin III**
B. Decreased fibrinogen
C. Decreased metabolism of Vitamin K
D. Increase in protein C

Ans A Hypercoagulability: Thrombosis (venous, arterial) of major vessels is seen in Nephrotic syndrome. Reason: urinary losses of antithrombotic factors such as antithrombin III (most important) and protein S due to vascular stasis, increased platelet number and aggregability, increase in hepatic production of fibrinogen.

34. **Edema in nephrotic syndrome is due to:**

A. Sodium and water retention
B. Increased venous pressure
C. **Hypoalbuminemia**
D. Hyperlipidemia

Ans C

35. **The most common gene defect in idiopathic steroid resistant nephrotic syndrome:**

A. ACE
B. **NPHS**
C. HOX II
D. PAX

Ans B

36. **Nephrotic syndrome in children is caused by:**

A. **Minimal change disease**
B. RPGN
C. MPGN
D. Membranous nephritis
E. Hemolytic uremic syndrome

Ans A b. RPGN c. MPGN; and d. Membranous nephritis

37. **Most common infection in a child with nephrotic syndrome:**

A. **Spontaneous bacterial peritonitis**
B. Pneumonia
C. UTI
D. Cellulitis

Ans A

38. **The most common gene defect in idiopathic steroid resistant nephrotic syndrome:**

A. ACE
B. **NPHS2**
C. HOX11
D. PAX

Ans B Pediatric Nephrology by Ellis D. Avner 6/e pg 645 Most common gene defect in idiopathic steroid resistant nephrotic syndrome is NPHS2.

39. The finnish type of congenital nephrotic syndrome occurs due to gene mutations affecting the following protein:

A. Podocin
B. Alpha – actinin
C. **Nephrin**
D. CD2 activated protein

Ans C Congenital Nephrotic syndrome more commonly occurs due to mutation of NPHS1gene that encodes nephrin

40. A 9 years old boy has steroid dependent nephrotic syndrome for the last 5 years. The patient is markedly cushingoid with blood pressure of 120/86 mmHg and small subcapsular cataracts. The most appropriate therapy of choice is:

A. Long term frusemide with enalapril
B. **Cyclophosphamide**
C. Intravenous immunoglobulin
D. Intravenous pulse corticosteroids

Ans B • This child with SDNS has evidence of steroid toxicity (cushingoid appearance, hypertension, cataracts);
• Cyclophosphamide prolongs the duration of remission in SDNS/FRNS patients
• Levamisole is usually the first line treatment for SDNS; In Levamisole failure, Cyclophosphamide is used.

41. Which of the following is included in definition of nephrotic syndrome:

A. Micro albuminuria
B. **Massive proteinuria**
C. Microscopic hematuria
D. Edema
E. Hyperlipidemia

Ans B d. Edema, e. Hyperlipidemia

42. True about nephrotic syndrome in a child:

A. **Minimal change disease is commonest cause**
B. Proteinuria of 4 gm/m2/hr is seen
C. Cyclosporine and azathioprine is mainstay of therapy
D. Pretreatment biopsy is done in all cases
E. Spontaneous bacterial peritonitis is associated with it

Ans A e. Spontaneous bacterial peritonitis is associated with it

43. A 5 years old child suffering from nephrotic syndrome is responding well to steroid therapy. What would be the most likely finding on light microscopy?

A. **No finding**
B. Basement membrane thickening
C. Hypercellular glomeruli
D. Fusion of foot process434 Section 3: Systemic Pediatrics
NEPHRITIC SYNDROME

Ans A This is a case of typical MCNS (Minimal Change Nephrotic Syndrome) with a good response to steroid therapy.

44. All the following are secondary causes of nephrotic syndrome EXCEPT

A. **measles**
B. malaria
C. syphilis
D. toxoplasmosis
E. schistosomiasis

Ans A Infections causes secondary nephrotic syndrome is : Endocarditis, Hepatitis B, C,HIV-1, infectious mononucleosis, malaria, syphilis (congenital and secondary), toxoplasmosis, schistosomiasis, filariasis.

45. Children with nephrotic syndrome are especially susceptible to infections such as cellulitis, spontaneous bacterial peritonitis, and bacteremia, all the following are contributory factors EXCEPT

A. urinary losses of immunoglobulin G
B. urinary loss of complement factors
C. impaired opsonization of microorganisms
D. immunosuppressive therapy
E. **urinary losses of immunoglobulin A**

Ans E

46. Nephrotic syndrome is a hypercoagulable state resulting from

A. hemodilution
B. decreased platelet number
C. **changes in coagulation factor levels**
D. decrease in hepatic production of fibrinogen
E. high level of antithrombin III

Ans C Vascular stasis from hemoconcentration,intravascular volume depletion , increased platelet number and aggregability, increase in hepatic production of fibrinogen,and low level of antithrombin III, cause hypercoagulable state.

47. When you investigate for first attack of minimal change nephrotic syndrome (MCNS), one of the following is TRUE

A. **microscopic hematuria is present in 20% of children**
B. spot urine protein : creatinine ratio should be <2.0
C. serum complement levels are low
D. renal biopsy is routinely performed
E. 2+ proteinuria

Ans A A spot urine protein : creatinine ratio should be >2.0, serum complement levels are normal, a renal biopsy is not routinely performed if the patient fits the standard clinical picture of MCNS, and the urinalysis reveals 3+ or 4+ proteinuria.

48. In treatment of minimal change nephrotic syndrome, all the following are true

A. tuberculosis must be ruled out prior to starting immunosuppressive therapy
B. steroid therapy may be initiated without a diagnostic renal biopsy
C. prednisone should be administered as a single daily dose of 60 mg/m2/day
D. remission consists of a urine<1+ protein on urine dipstick for3 consecutive days
E. pleural effusion, is a potential complication of parenteral albumin therapy

Ans E Symptomatic volume overload, hypertension, heart failure, and pulmonary edema, is a potential complication of parenteral albumin therapy, particularly when administered as rapid infusions.

49. Steroid resistance nephrotic syndrome is defined as failure to achieve remission after

A. 4wk of corticosteroid therapy
B. 6 wk. of corticosteroid therapy
C. 8 wk. of corticosteroid therapy
D. 10 wk. of corticosteroid therapy
E. 12 wk. of corticosteroid therapy

Ans C Steroid-resistant nephrotic syndrome is usually caused by focal segmental glomerulosclerosis (80%), minimal change nephrotic syndrome, or membranoproliferativeglomerulonephritis.

50. To reduce the risk of serious infections in children with nephrotic syndrome, all the following immunizations can be used EXCEPT

A. pneumococcal 13-valent conjugant vaccine
B. influenza vaccine annually to the child
C. pneumococcal 23-valent polysaccharide vaccine
D. varicella-zoster vaccine
E. influenza vaccination annually to their household contacts

Ans D Following close contact with varicella infection, give immune compromised children on immunosuppressive agents varicella-zoster immune globulin if available.

51. After achievement of remission in children with nephrotic syndrome, they should

A. restrict their activities
B. restrict sodium intake
C. be considered chronically ill
D. maintain an unrestricted diet
E. decrease water/fluid intake

Ans D To minimize the psychologic effects of the condition and its therapy, children with idiopathic nephrotic syndrome should not be considered chronically ill and should participate in all age-appropriate childhood activities.

52. Congenital nephrotic syndrome is defined as nephrotic syndrome manifesting at birth or within the 1st 3 mo of life, The etiologies include all the following EXCEPT

A. syphilis
B. herpes infection
C. mercury exposure
D. mutations in the NPHS1
E. infantile systemic lupus erythematosis

Ans B Congenital nephrotic syndrome may be classified as primary or as secondary to a number of etiologies such as in utero infections (cytomegalovirus, toxoplasmosis, syphilis, hepatitis B and C, HIV).

53. The definitive treatment of primary congenital nephrotic syndrome is

A. angiotensin-converting enzyme inhibitors
B. prostaglandin synthesis inhibitors
C. aggressive nutritional support
D. bilateral nephrectomies
E. renal transplantation

Ans E is the definitive treatment of congenital nephrotic syndrome, though recurrence of the nephrotic syndrome has been reported to occur after transplantation.

Acute Glomerulonephritis

54. Baby born at 30 weeks to 18 years old primigravida of weight 2 kg, died after 48 hours. Apgar scores were 5 and 8 at 1 and 5 minutes. On autopsy, bilateral enlarged kidney with multiple radially arranged cysts were seen. Which of the following finding is expected to be associ- ated with it?

A. Imperforate anus
B. Hepatic cyst and fibrosis
C. Absence of ureter
D. Holoprosencephaly

Ans B /e p 507-509 In the given question, neonatal death along with the presence of bilaterally enlarged kidneys with multiple cysts indicate ARPKD (autosomal recessive polycystic kidney disease). ARPKD is associated with hepatic fibrosis and portal hypertension which is the correct answer.

55. A 12 years old boy is referred for evaluation of nocturnal enuresis and short stature. The blood pressure is normal. The blood urea is 112 mg/dL, Creatinine 6 mg/dL, Sodium 119 meq/ L, Potassium 4 meq/L, Calcium 7 mg/dL, Phos- phate 6 mg/dL and alkaline phosphatase 400 U/L. Urinal ysis shows trace proteinuria with hyaline casts; no red and white cells are seen. Ultrasound shows bilateral small kidneys and the micturating cystourethrogram is normal. The most likely diagnosis is:

A. Reflux nephropathy
B. Nephronophthisis
C. Polycystic kidney disease
D. Medullary cystic kidney disease NEPHROTIC SYNDROME

Ans D Nephronophthisis /e p 509 In the given case scenario, a 12 years old boy presents with:
• Clinical features of chronic kidney disease (CKD) like short stature, nocturnal enuresis, and
• Biochemical evidence of CKD like high serum creatinine, low sodium, borderline calcium and raised ALP
• Ultrasound shows bilateral small kidneys – again an evidence of chronic kidney disease. About juvenile nephronophthisis (JN)–medullary cystic kidney disease complex (MCKD):

56. **A 10–years old child presents with edema, oliguria and frothy urine. He has no past history of similar complaints. On examination, his urine was positive for 3+ proteinuria, no RBCs/WBCs and no casts. His serum albumin was 2.5 g/dL and serum creatinine was 0.5 mg/ dL. The most likely diagnosis is:**

A. IgA nephropathy
B. **Minimal change disease**
C. Acute interstitial nephritis
D. Membranous nephropathy

Ans B In the present case, massive proteinuria (3+) along with hypoalbuminemia is suggestive of nephrotic syndrome. Out of the options, minimal change disease is the most common cause in children.

57. **A child presented with decreased albumin, creatinine 0.6 mg/dl, microscopic hematuria, proteinuria 3+, edema, hypercholesterolamia, C level normal. Most 3 likely diagnosis is**

A. **Minimal change disease**
B. Post streptococcal glomerulonephritis
C. Focal segmental glomerulonephritis
D. MPGN

Ans A

58. **A 10 year old boy presents with gross hematuria, loin pain, and diarrhea; urine examination is normal. Serum C3 levels are normal; Probable diagnosis is?**

A. **Berger's disease**
B. PSGN
C. Microangiopathic hemolytic anemia
D. None of the above

Ans A In Berger's disease (IgA Nephropathy), gross hematuria often occurs within 1-2 days of onset of an upper respiratory or gastrointestinal infection, in contrast to the longer latency period observed in acute post-infectious glomerulonephritis, and may be associated with loin pain.

59. **A 9-year-old boy is brought with history of decreased urine output, cola colored urine and swelling of the face and hands, of 2 days duration. He is hypertensive, has a puffy face and pitting edema of the lower limbs. He has history of skin lesions 4 weeks earlier. A diagnosis of post streptococcal glomerulonephritis is made. ASLO titers are likely to be?**

A. Elevated
B. **Not elevated**
C. Increased progressively over 2 weeks
D. Initially elevated, rapidly fall in 3-4 days

Ans B Lab Diagnosis of PSGN;
• Serum C3 level is reduced in > 90% of patients in acute phase of PSGN, and returns to normal in 6-8 wk
• Confirmation of the diagnosis requires evidence of a prior streptococcal infection eg positive throat culture or rising antibody titer to streptococcal antigen(s)
• Antistreptolysin O titer is commonly elevated after a pharyngeal infection but rarely increases after streptococcal skin infections.Q (as in this case)

60. **A 9-year-old boy is brought with history of decreased urine output, cola colored urine and swelling of the face and hands of 2 days duration. He is hypertensive, has a puffy face and pitting edema of the lower limbs. He has history of skin lesions 4 weeks earlier. A diagnosis of post streptococcal glomerulonephritis is made. C3 levels are likely to return to normal in?**

A. 1-2 years
B. 1-2 weeks
C. **6-8 weeks**
D. 3-4 months

Ans C

61. **A 9-year-old boy is brought with history of decreased urine output, cola colored urine and swelling of the face and hands of 2 days duration. He is hypertensive, has a puffy face and pitting edema of the lower limbs. He has history of skin lesions 4 weeks earlier. A diagnosis of post streptococcal glomerulonephritis is made. If an antibiotic is to be used to limit the spread of the nephritogenic organisms, the drug of choice is?**

A. Cefixime
B. **Penicillin**
C. Meropenem
D. Amoxycillin

Ans B Treatment of PSGN
• Management is directed at treating the acute effects of renal insufficiency and hypertension
• Although a 10 day course of systemic antibiotic therapy with penicillin is recommended to limit the spread of the nephritogenic organisms, antibiotic therapy does not affect the natural history of PSGN
• Treatment of hypertension: Calcium channel antagonists, vasodilators, or angiotensin-converting enzyme inhibitors.

62. **Which is seen in Electron microscopy in PSGN?**

A. **Epithelial humps**
B. Spike and dome appearance
C. Mesangial deposits
D. Subendothelial deposits

Ans A

63. **A 8 years old child has throat infection followed by hematuria; Diagnosis?**

A. E. coli
B. Shigella
C. Salmonella
D. **Pseudomonas**

Ans D Can be (a) or (b) As the time interval between sore throat and hematuria is not mentioned, it can either be IgA Nephropathy or PSGN.

64. A 5 years old child presents with perivascular IgA deposition and neutrophilic collection. There is erythema- tous rash on the lower limb and non blanching purpura. Probable diagnosis is:

A. Henoch-Schonlein purpura
B. Wegener's granulomatosis
C. Vasculitis
D. Kawasaki diseases

Ans A • The typical non blanching purpura with presence of IgA and neutrophils around vessels confirms the diagnosis of H.S.P
• Although palpable purpura may be present in Wegener's granulomatosis, biopsy characteristically shows necrotizing granulomatous vasculitis.

65. In case of PSGN complications commonly seen are all except:

A. LVF
B. Hypertensive encephalopathy
C. Hyperkalemia
D. Bleeding diathesis

Ans D

66. A child presented with cola coloured urine, proteinuria 2+ and h/o rash 2 weeks ago. Probable diagnosis is:

A. IgA nephropathy
B. HSP
C. HUS
D. Wegener's granulomatosis
E. Paroxysmal nocturnal hemoglobinuria

Ans B Ghai 8/e p 632
• Here, a child is presenting with acute nephritic syndrome with history of skin rash manifestations 2 weeks back
• Skin manifestations are typically absent in IgA nephropathy and Wegener's granulomatosis. About PNH: Hemoglobinuria is rarely seen in children as compared to adults with PNH. Chronic hemolysis, thrombocytopenia and leukopenia are often characteristic.

67. A 8 years old child with BP 180/100 mm Hg, Blood Urea 90 mg/dl, creatinine 5.3 mg/dl, urinalysis shows 15–20 pus cells, 1–2 RBC, protein 1+; Most likely diagnosis is:

A. Post Infective glomerulonephritis
B. Accelerated hypertension with ARF
C. Idiopathic RPGN
D. Chronic interstitial nephritis with VUR

Ans D In the given clinical scenario, child has hypertension, renal failure (increased urea and creatinine), pyuria, absence of hematuria (presence of 5 RBCs/HPF is considered significant) and mild proteinuria, Out of the options given, Chronic Interstitial Nephritis with VUR seems most appropriate diagnosis. Chronic Interstitial disease
• In children, it commonly occurs as a result of underlying obstructive uropathy or VUR

68. A child develops non-blanching macules and papules on lower extremities, mild abdominal pain and skin biopsy showed IgA deposition. Most appropriate diagnosis is:

A. Drug induced vasculitis
B. Henoch Schonlein purpura
C. Wegener's granulomatosis
D. Kawasaki disease

Ans B Classification criteria for H.S. purpura according to Pediatric Rheumatology European Society: Palpable purpura (in absence of coagulopathy or thrombocytopenia) and 1 or more of the following criteria:
• Diffuse abdominal pain • Biopsy of affected tissue demonstrating predominant IgA deposition
• Arthritis or arthralgia • Renal involvement (proteinuria, hematuria or red cell casts) fo Hence, it is a typical case presentation of Henoch–Schonlein purpura. weiveR 85. b. IgA nephropathy a.

69. A four-year old child presents with mild fever, malaise, purpura, arthritis, abdominal pain and microscopic hematuria. What would be the most likely diagnosis?

A. Thrombasthenia
B. Idiopathic thrombocytopenic purpura
C. Systemic lupus erythematosus
D. Henoch-Schonlein purpura

Ans D Henoch-Schonlein purpura /e p

70. Characteristic of acute Glomerulonephritis:

A. RBC cast
B. Hemoglobinuria 106.
C. Proteinuria
D. Broad cast
E. Hyaline cast

Ans A c. Proteinuria a. RBC cast Produced due to bleeding from within the glomerulus seen in acute glomerulonephritis (GN) b. Hemoglobinuria Hemoglobinuria without hematuria usually occurs in the presence of hemolysis. c. Proteinuria cola colored urine, proteinuria > 100 mg/dL, RBC casts and dysmorphic RBCs are all seen in acute GN d. Broad cast Broad casts are seen in chronic kidney disease. e. Hyaline cast Seen in Nephrotic syndrome and not nephritic

71. Child with B.P. 190/110, pedal edema, facial edema, absent ascites, gross hematuria. Diagnosis is:

A. Acute GN
B. Nephrotic syndrome

Ans A c. Renal thrombosis a. This is a classic case presentation of Acute glomerulonephritis with gross hematuria, hypertension, edema. b. Nephrotic syndrome: Massive proteinuria, hypoalbuminemia and edema seen but Gross hematuria is usually absent. c. Renal vein thrombosis: – It usually presents with hematuria and flank masses in presence of predisposing factors; Hypertension usually seen – Doppler flow studies of the inferior vena cava and renal vein confirm the diagnosis. d. Renal amyloidosis

72. A child was diagnosed as a case of pauci-immune crescentic glomerulonephritis. The treatment to be given in this child is:

A. Prednisolone + Cyclophosphamide
B. Methyl prednisolone
C. Cyclophosphamide
D. Immunoglobulins

Ans A Pauci-immune Crescentic GN Definition Crescentic GN with few or no immune deposits on immunofluorescence Causes
• Microscopic polyangitis • Churg Strauss syndrome
• Wegener's granulomatosis • Medications, e.g. Pencilllamine, hydralazine, propylthiouracil
• Idiopathic Crescentic GN Treatment Induction therapy with steroids and cyclophosphamide is used initially followed by maintenance with Azathioprine or MMF.

73. A 6-year-old girl presents with recurrent E. coli infection in urine. Ultrasound of abdomen shows hydroureter and hydronephrosis. Micturating cystourethrogam shows defect in urinary bladder. The likely diagnosis is:

A. Sacrococcygeal teratoma
B. Vesicoureteric reflux – grade II
C. Duplication of ureter
D. Ureterocele

Ans D In Ureterocele, IVP shows a round filling defect in the bladder;

74. A 6 years old girl presents with recurrent E.coli infection in urine. Ultrasound of abdomen shows hydroureter and hydronephrosis. Micturating cysto-urethrogram shows filling defect in urinary bladder. The likely diagnosis is:

A. Sacro coccygeal teratoma
B. Vesicouretric reflex – grade II
C. Duplication of ureter
D. Ureterocele ACUTE KIDNEY INJURY

Ans D Ureterocele

75. An 8 day old breast fed baby presents with vomiting, poor feeding and loose stools. On examination, the heart rate is 190/min, blood pressure 50/30 mm Hg, respiratory rate 72 breaths/min and CFT~4sec. Investigations show hemoglobin level of 15 gm/dL. Na+ 120 meq/L, K+ 6.8 meq/L, Cl– 81 meq/L, bicarbonate 15 meq/L, Urea 30 mg/ dL, and creatinine 0.6 mg/ dL, the most likely diagnosis is:

A. Congenital adrenal hyperplasia
B. Acute tubular necrosis
C. Congenital hypertrophic pyloric stenosis
D. Galactosemia 142.

Ans A Dehydration, salt wasting, hyponatremia and hyperkalemia suggest Congenital adrenal hyperplasia

76. An infant with severe dehydration secondary to diarrhea 143. suddenly presents with flank mass and blood in urine. The most probable diagnosis is?

A. Renal vein thrombosis
B. Pyelonephritis
C. Acute glomerulonephritis
D. Lower nephrosis 144.

Ans A Renal vein thrombosis (RVT) occurs in 2 distinct clinical settings:
• In newborns and infants: with asphyxia, dehydration, shock, sepsis, hypercoagulable states, and maternal diabetes; and
• In older children, RVT is seen in patients with nephrotic syndrome, cyanotic heart disease, inherited hypercoagulable states, sepsis, following kidney transplantation, and following exposure to angiographic contrast agents Diagnosis:
• Suggested by hematuria and flank masses in patients seen in the high-risk situations

77. In a child, non-functioning kidney is best diagnosed by:

A. Ultrasonography
B. IVU
C. DTPA renogram
D. Creatinine clearance 147. RENAL TUBULAR DISORDERS

Ans C In order to assess function of an apparent non-functioning kidney, the best modality would be DTPA renogram. DTPA renogram is a relatively non invasive radionuclide study which enables evaluation of function of individual kidney. 99Tc-DMSA scan is used for providing high quality images of renal morphology (especially useful in follow up of UTIs). About other options: a. Ultrasonography is the initial modality for imaging of the kidney and urinary tract. The procedure is operator dependent and

78. A 10-year-old girl present with polyuria and polydipsia with hypokalemia, hypercalciuria and metabolic alkalosis. What is the probable diagnosis?

A. Renal tubular acidosis
B. Diabetes insipidus
C. Bartter syndrome
D. Pseudo-hypoaldosteronism

Ans C Syndromes Bartter syndrome Gitelman syndrome Characterized by Hypokalemic metabolic alkalosis with hyper calciuria & Hypokalemic metabolic alkalosis, with hypocalciuriaQ salt wasting & hyponatremia and hypomagnesemia.Q Symptoms Polyuria, polydipsia, vomiting, consti pation, failure to Recurrent muscle cramps and spasms thrive. BP: Low/normal Molecular basis Mutations in genes that encode Na+/K+/2Cl– transporter Mutations in sodium chloride cotransporter NCCT,

79. A 3 years old boy is detected to have bilateral renal calculi. Metabolic evaluation confirms the presence of marked hypercalciuria with normal levels of calcium, magnesium, phosphate, uric acid and creatinine. A diagnosis of idiopathic hypercalciuria is made. The dietary management includes all, except:

A. Increased water intake
B. Low sodium diet
C. Reduced calcium intake
D. Avoid meat proteins

Ans C In Idiopathic hypercalciuria, dietary calcium is not restricted as low calcium diet are accompanied by higher urinary oxalate excretion, since intestinal oxalate is no longer bound to calcium.

80. **A 10 years old boy is having polyuria, polydipsia. Laboratory data showed (in meq/L): (AIIMS May 2002) Na+ – 154 K+ – 4.5 HCO – 22 3 S. Osmolality – 295 B. Urea – 50 Uric specific gravity – 1.005 What is the diagnosis?**

A. Diabetes insipidus
B. Renal tubular acidosis
C. Bartter's syndrome
D. Recurrent UTI

Ans A The given child in question has presented with clinical features: Polyuria, polydipsia. Lab data: Hypernatremia (S. Na+ – 154 meq/L), Raised S. Osmolality (S. Osmolality – 295), Raised B. Urea (B. urea – 50), Normal K+, HCO – (4.5, 22 respectively). The given clinical and laboratory features are compatible with a diagnosis of diabetes insipidus

81. **Which of the following is pathognomonic in diagnosis of X-linked type of hereditary nephritis?**

A. gross hematuria
B. discontinuous epidermal basement membrane staining
C. platelet abnormalities
D. hearing loss
E. macular flecks

Ans B Absence of epidermal basement membrane staining for the α5 chain of type IV collagen in male hemi zygotes and discontinuous epidermal basement membrane staining in female heterozygotes on skin biopsy is pathognomonic for X-linked AS and can preclude diagnostic renal biopsy.

82. **Renal biopsy in acute post streptococcal glomerulonephritis should be considered in all the following EXCEPT**

A. acute renal failure
B. nephrotic syndrome
C. absence of evidence of streptococcal infection
D. low C3 level in the first 2 months
E. initially normal complement levels

Ans D Renal biopsy is considered when hematuria and proteinuria, diminished renal function, and/or a low C3 level persist more than 2 mo. after onset.

83. **The diagnosis of Bartter syndrome typically include**

A. markedly low serum rennin
B. high urinary calcium levels
C. nephrocalcinosis
D. hypomagnesaemia
E. renal biopsy

Ans B Urinary calcium levels are typically elevated, as are urinary potassium and sodium levels, nephrocalcinosis resulting from Hypercalciuria may be seen on ultrasound examination. Serum renin, aldosterone, and prostaglandin E levels are often markedly elevated; hypomagnesaemia is seen in a minority of patients but is more common in Gitelman syndrome. Renal biopsy is rarely performed to diagnose this condition.

RPGN & Crescentic GN

84. **True about infantile polycystic kidney disease are all the following except:**

A. Autosomal dominant
B. Caroli disease associated
C. Renal cysts present at birth
D. Periportal fibrosis

Ans A /e p 507-509 Infantile polycystic kidney disease is autosomal recessive. with

85. **Type I RPGN is seen in**

A. SLE
B. IgA nephropathy
C. Henoch Schonlein purpura
D. Good pasture syndrome

Ans D Types of RPGN: Entity Type I (20%) Type II (25%) Type III (55%) Mechanism Anti-GBM Antibody Immune Complex Pauci-Immune Etiology Good pasture • Post-infectious: Post streptococcal GN ANCA-associated syndromeQ • Noninfectious: SLE, Henoch-Schönlein purpura, Mixed • Wegener granulomatosisQ with cryoglobulinemia • Microscopic polyangiitisQ
• Primary Renal Disease: IgA nephropathyQ • Hypersensitivity vasculitisQ
• Membranoproliferative glomerulonephritisQ

86. **A 5 years old child presents with a calculus of size 2 cm in the upper ureter. He also complains of hematuria. USG shows no further obstruction in the urinary tract. Treatment of choice for this patient would be:**

A. Ureterolithotomy
B. Endoscopic removal Questions
C. ESWL
D. Observation

Ans C 138 below; A calculus of size upto 2 cm in the upper ureter, the treatment of choice is ESWL (extra corporeal shock wave lithotripsy).

87. **Alport Syndrome AS, is a genetically heterogeneous disease caused by mutations in the genes coding for type IV collagen, it is characterized by**

A. asymptomatic microscopic hematuria
B. proteinuria<1 g/24 hr.
C. congenital sensor neural hearing loss
D. ocular abnormalities in 80% of X-linked type
E. leiomyomatosis of the esophagus is common

Ans A All patients with AS have asymptomatic microscopic hematuria, which may be intermittent in girls and younger boys. Progressive proteinuria, often exceeding 1 g/24 hr., is common by the 2nd decade of life and can be severe enough to cause nephrotic syndrome. Bilateral sensor neural hearing loss, which is never congenital, Ocular abnormalities, which occur in 30-40% of patients with X-linked AS, include anterior lentic onus (extrusion of the central portion of the lens into the anterior chamber), macular flecks, and...

88. **"Rapidly progressive" RPGN describes the clinical course of several forms of glomerulonephritis whose unifying feature is the histopathologic finding of crescents in the majority of glomeruli. Plasmapheresis has been reported to be of benefit in patients with**

A. post infectious glomerulonephritis
B. immunoglobulin A nephropathy
C. Henoch-Schönleinpurpura
D. **Good pasture disease**
E. endocarditis

Ans D Plasmapheresis may also benefit patients with ANCA-associated crescentic GN, in particular those with the most severe renal dysfunction at presentation. The possible benefits of plasmapheresis in other forms of RPGN are unclear.

89. **Medullary sponge kidney is a relatively rare sporadic disorder in children. It is characterized by**

A. cystic dilation of the proximal tubule
B. **nephrolithiasis**
C. proximal renal tubular acidosis
D. concentrated urine
E. cortical nephrocalcinosis

Ans B Medullary sponge kidney is characterized by cystic dilation of the terminal portions of the collecting ducts as they enter the renal pyramids, Ultrasonographically, patients often have medullary nephrocalcinosis, complications include nephrolithiasis, pyelonephritis, hyposthenuria (inability to concentrate urine), and distal RTA.

90. **Death in neonates with bilateral renal agenesis is due to**

A. renal failure
B. hypertension
C. **pulmonary insufficiency**
D. congenital heart disease
E. sepsis

Ans C Death is due to pulmonary insufficiency from pulmonary hypoplasia rather than renal failure.

91. **Ureter that drains outside the bladder is referred to as an ectopic ureter. Of the following, which statement is TRUE regarding ectopic ureter?**

A. male: female ratio is 2:
B. it mostly drains into the cervix in girls
C. it mostly drains into the vas deferens in boys
D. urinary tract infection (UTI) is uncommon
E. **nephrectomy is indicated if renal function is poor**

Ans E This anomaly is 3 times as common in girls as in boys.In girls, approximately 35% of these ureters enter the urethra at the bladder neck, 35% enter the urethra vaginal septum, 25% enter the vagina, and a few drain into the cervix, uterus, Gartner duct, or a urethral diverticulum.

92. **The most common cause of micropenis is failure of the hypothalamus to produce an adequate amount of gonadotropin-releasing hormone. Of the following, the syndrome associated with micropenis is**

A. **Kallmann syndrome**
B. fetal hydantoin syndrome
C. Apert syndrome
D. de Lange syndrome
E. Holt-Oram syndrome

Ans A Others syndromes associated with syndactyly.

93. **Dietary treatment of renal or ureteral calculi include**

A. increased dietary intake of sodium
B. reduced dietary intake in potassium
C. increased protein diet
D. **encourage lemon juice intake**
E. calcium restriction

Ans D An excellent source of citrate (inhibitor of calcium stones) is lemonade, reduction in dietary intake of sodium and increased potassium intake is indicated, low- protein diets reduce urinary calcium and oxalate excretion, and calcium restriction in children should be avoided.

IgA Nephropathy

94. **The most common congenital anomaly of the urinary tract:**

A. Epispadias
B. **Hypospadias**
C. Exstrophy of bladder
D. Posterior urethral valve

Ans B Hypospadias is a urethral opening on the ventral surface of the penile shaft affecting 1 in 250 male newborns. Exstrophy of urinary bladder:
• It occurs in 1 in 35,000-40,000 births; Male: female ratio is 2 : 1
• Severity ranges from simple epispadias (in boys) to cloacal exstrophy. Posterior urethral valve: The most common cause of severe obstructive uropathy in children is posterior urethral valves, affecting 1 in 8,000 boys.

95. **True regarding IgA Nephropathy**

A. Usually in children < 10 years
B. Microscopic hematuria is the most common presentation
C. **Recurrent gross hematuria following respiratory infection**
D. Decreased serum IgA

Ans C

96. **The renal biopsy of a 6 years old boy with recurrent gross hematuria shows IgA nephropathy. The urinary protein excretion is 130 mg/day. Which of the following is the most appropriate next step in the management:**

A. **Microangiopathic thrombotic anemia**
B. IgA nephropathy
C. PSGN
D. H.S. Purpura

Ans A Administer corticosteroids Treatment of IgA nephropathy: The primary t/t is proper BP control; Other drugs found useful are:
• Fish oil (omega-3 fatty acids): An anti-inflammatory agent and it prevents the rate of renal progression
• Immunosuppressive therapy: Done with corticosteroids with or without cytotoxic therapy
• ACE inhibitors and Angiotensin II receptor antagonists: role in reducing proteinuria

97. A child presents with abdominal colic and hematuria. On ultrasonography a stone 2.5 cm in diameter is seen in the renal pelvis. The next step in management of this case is:

A. Pyelolithotomy
B. Nephroureterostomy
C. Conservative
D. ESWL RENAL PHYSIOLOGY

Ans A Primary surgical treatment options for renal stones based on stone size and location. Stones Shock wave lithotripsy Ureteroscopy Percutaneous nephrolithotomy Renal <1 cm Most common Optional Optional 1–2 cm Most common Optional Optional >2 cm Optional Rare Most common Ureteral Proximal Most common Optional Occasional Distal Optional Most common Rare

98. IgA nephropathy is characterized by

A. hematuria occur after skin infection
B. proteinuria is often >1000 mg/24 hr
C. severe hypertension
D. normal serum levels of C3
E. serum IgA levels have a diagnostic value

Ans D Gross hematuria often occurs within 1-2 days of onset of an upper respiratory or gastrointestinal infection, in contrast to the longer latency period observed in acute post infectious glomerulonephritis, and may be associated with loin pain. Proteinuria is often <1000 mg/24 hr in patients with asymptomatic microscopic hematuria. Mild to moderate hypertension is most often seen in patients with nephritic or nephrotic syndrome, but is rarely severe enough to result in hypertensive emergencies.

99. The primary treatment of IgA nephropathy is appropriate blood pressure control and management of significant proteinuria. Regarding the treatment, which of the following is TRUE?

A. ACEI are ineffective in reducing proteinuria
B. corticosteroids not improve renal function
C. tonsillectomy is currently recommended
D. successful kidney transplantation
E. cyclophosphamide is effective in improving renal function

Ans D Angiotensin-converting enzyme inhibitors and angiotensin II receptor antagonists are effective in reducing proteinuria and retarding the rate of disease progression when used individually or in combination. Corticosteroids reduce proteinuria and improve renal function in those patients with a glomerular filtration rate>60 mL/min/m2.To date, additional immunosuppression with cyclophosphamide or azathioprine has not appeared to be effective, but further randomized clinical trials are in progress.

100. The juvenile nephronophthisis JN is a group of inherited genetically determined cystic renal diseases, it can be presented with all the following EXCEPT

A. oliguria
B. unexplained anemia
C. growth failure
D. chronic renal failure
E. retinal degeneration

Ans A Patients with JN typically present with polyuria, anemia that is seemingly disproportionate to the degree of renal insufficiency, JN often have characteristic small cysts in the corticomedullary region.

101. The MOST common cause of an abdominal mass in the newborn is

A. polycystic kidney disease
B. neuroblastoma
C. renal vein thrombosis
D. hydronephrosis
E. multicystic dysplastic kidney

Ans E

102. Ureteroceles is a cystic dilation of the terminal ureter. Of the following, the MOST appropriate statement is

A. it is more common in boys
B. it cannot be diagnosed prenatally
C. it is associated with ureteral duplication in girls
D. it commonly causes bladder outlet obstruction
E. the orthotopic type extends into the urethra

Ans C Ureteroceles are much more common in girls than in boys. Affected children usually are discovered by prenatal ultrasonography. Rarely, large ectopic ureteroceles can cause bladder outlet obstruction. Ureteroceles may be ectopic, in which case the cystic swelling extends through the bladder neck into the urethra, or orthotopic, in which case the ureteroceles is entirely within the bladder.

103. Priapism of high-flow type MOST commonly follows

A. sickle cell disease
B. perineal trauma
C. sildenafil ingestion
D. leukemia
E. all the above

Ans B Non ischemic (high-flow) priapism most commonly follows perineal trauma, such as a straddle injury, while others are causes of ischemic (venoocclusive, low-flow) priapism.

104. Which of the following drugs can precipitate and form renal stones?

A. acetazolamide
B. indanivir
C. probenecid
D. theophylline
E. vitamin C and Children

Ans B All the others distractors increase the risk of stone formation but do not themselves precipitate as stones.

Henoch-Schönlein Purpura Nephritis

105. Which of the following is the most common renal cystic disease in infants is?

A. Renal anomalies
B. Severe oligohydramnios
C. Flattened nose
D. All the above

Ans C Unilateral renal dysplasia Multicystic dysplastic kidney
• It is the most frequent form of renal cystic disease in childhood; It is usually unilateral,
• DTPA renography shows no function and USG or CT scanning shows replacement of kidney with cysts of varying sizes that do not communicate with each other Simple, renal solitary cysts: Mostly observed in adults, are uncommon in children.

106. **Henoch Schonlein purpura commonly involves which age group?**

A. 4–8 years
B. 10–15 years
C. 15–20 years
D. > 25 years

Ans A • Henoch-Schönlein purpura (HSP) is the most common small vessel vasculitis in childhood
• 90% of HSP cases occur in children, usually between 3-10 yr age; Presents with purpura, arthritis and abdominal pain.

107. **Regarding FeNa, which of the following is true?**

A. FeNa is lower in neonates when compared to children
B. FeNa is higher in intrinsic renal failure than prerenal failure
C. Measurement of FeNa is not affected by use of diuretic
D. FeNa is similar in both preterm and term neonate

Ans B Fractional excretion of sodium (FeNa)
• FeNa is the urine:plasma (U:P) ratio of sodium divided by U:P of creatinine x 100.
• Neonates have higher FeNa because of tubular immaturity,
• FeNa is higher in preterm neonates due to immaturity of sodium reabsorptive capacity.
• FeNa may be affected by diuretic use. But fractional excretion of urea (FEurea) is dependent on passive forces and is less influenced by diuretic therapy. Classic laboratory findings in AKI U with Type UOsm Na Fe BUN/ Na Cr

108. **Thin basement membrane disease TBMD is defined by the presence of persistent microscopic hematuria and isolated thinning of the GBM on electron microscopy, all the following are true EXCEPT**

A. significant proteinuria is rare
B. episodic gross hematuria can be present
C. may be transmitted as an autosomal dominant trait
D. heterozygous mutations in the COL4A4 genes
E. renal insufficiency is common

Ans E Rare cases of TBMD progress, and such patients develop significant proteinuria, hypertension, or renal insufficiency.
[heterozygous mutations in the COL4A3 and COL4Agenes, which encode the α3 and α4 chains of type IV collagen present in the GBM, result in TBMD while homozygous mutations in these same genes result in Autosomal recessive Alport syndrome].

109. **All the following causes hematuria associated with rapid development of microangiopathic hemolytic anemia or enlargement of the kidney(s) EXCEPT**

A. nephrotic syndrome
B. renal vein thrombosis
C. polycystic kidney disease
D. Wilms tumor
E. hemolytic-uremic syndrome

Ans A These include hemolytic uremic syndrome, hydronephrosis, polycystic kidney disease, Wilms tumor, and intrarenall abscess or hematoma. All patients should be evaluated for congenital and acquired hypercoagulable states.

110. **Hyperkalemia can lead to cardiac arrhythmia, cardiac arrest, and death. Serum potassium can be lowered by all the following EXCEPT**

A. regular insulin with glucose
B. calcium gluconate
C. sodium bicarbonate
D. Kayexalate
E. Dialysis

Ans B counteracts the potassium-induced increase in myocardial irritability but does not lower the serum potassium level.

111. **One of the complications of the horseshoe kidney is**

A. renal abscess
B. neuroblastoma
C. bladder outlet obstruction
D. familial renal dysplasia
E. multicystic dysplastic kidney

Ans E Wilms tumors are 4 times more common in children with horseshoe kidneys than in the general population, stone disease and hydronephrosis secondary to ureteropelvic junction obstruction are other potential late complications.

112. **Prune-belly syndrome, also called triad syndrome or Eagle-Barrett syndrome, is characterized by**

A. female predominance
B. polydramnios
C. small bladder
D. cardiac abnormalities (in 10% of cases)
E. all the above

Ans D 95% of affected children are male, oligohydramnios, very large bladder.

113. **The MOST common male urethral anomaly associated with prune-belly syndrome is**

A. congenital urethral fistula
B. urethral duplication
C. megalourethra
D. urethral hypoplasia
E. urethral atresia

Ans C

HUS & TTP

114. **Which one of the following is the most common cause of abdominal mass in neonates?**

A. Neuroblastoma
B. Wilm's tumor
C. Distended bladder
D. Multicystic dysplastic kidneys

Ans D a. Neuroblastoma is the MC extra cranial solid tumor in children & most commonly diagnosed malignancy in infants b. Wilm's tumor or Nephroblastoma is the most common primary malignant renal tumor of childhood and the second most common malignant abdominal tumor in childhood c. Distended bladder: examination after voiding rules out swelling due to distended bladder d. Multi cystic dysplastic kidney (MCDK) is the most common cause of an abdominal mass in the newborn, but the vast majority are non palpable at birth.

115. In Post-streptococcal glomerulonephritis:

A. C3 decreases, ASO increases
B. C3 increases, ASO increases
C. C3 decreases, ASO decreases
D. C3 increases, ASO decreases

Ans A

116. In shigella dysentery associated hemolytic uremic syndrome, the false statement is:

A. Leucocytosis
B. Neurological abnormalities
C. Hepatic failure
D. Thrombotic angiopathy

Ans C Hepatic failure does not occur in HUS. Laboratory findings of HUS include:
• Thrombocytopenia, Leukocytosis • Peripheral smear: Helmet cells, burr cells, fragmental RBCs (Schistocytes)
• ARF: Increased BUN and creatinine • Hemoglobinuria with hemosiderinuria
• Microscopic hematuria and Proteinuria • Normal PT and APTT
Option 'b' requires some explanation: Though neurological manifestations are not common in HUS, but they may occur.

117. A 2-year-old child with length 85 cm and weight of 11 kg was found to have serum urea–49 mg/dL, & serum creatinine 2 mg/dL. What is the estimated GFR of this child, as per Schwartz formula?

A. 90
B. 48
C. 18
D. 9

Ans C 18 Ref: Hari P, Biswas B, Pandey R, Kalaivani M, Kumar R, Bagga A. Clin Exp Nephrol 2012;16:697-705. Schwartz formula for creatinine clearance calculation = k x length/serum creatinine For Indian children, k = 0.42 eGFR = 0.42 × 85/2 = 17.8

118. The best single test to document cutaneous streptococcal infection is

A. low serum C3 level
B. elevated antistreptolysin O titer
C. positive streptozyme screen
D. antideoxyribonuclease B level
E. depressed serum CH50

Ans D Four differences in APSGN follow throat or skin infections: Post streptococcal GN commonly follows streptococcal pharyngitis during cold-weather months and streptococcal skin infections or pyoderma during warm-weather months. Epidemics of nephritis have been described in association with throat (serotypes M1, M4, M25, and some strains of M12) and skin (serotype M49) infections. The typical patient develops an acute nephritic syndrome 1-2 wk. after an antecedent streptococcal pharyngitis or 3-6 wk.

119. In hemolytic-uremic syndrome (HUS). Of the following, the MOSTcorrect statement is

A. majority of patients of diarrhea-associated enteropathictype develops HUS
B. stool culture is often positive in patients who have diarrhea-associated HUS
C. kidney biopsy is rarely indicated to diagnose HUS
D. partial thromboplastin and prothrombin times are usually low
E. Coombs test is usually negative in in pneumococci-induced HUS

Ans C Kidney biopsies are only rarely performed in HUS because the diagnosis is usually established by clinical criteria and the risks of biopsy are significant during the active phase of the disease. The organisms that cause HUS may be rapidly cleared, therefore, the stool culture is often negative in patients who have diarrhea-associated HUS, Partial thromboplastin and prothrombin times are usually normal, the Coombs test is negative, with the exception of pneumococci-induced HUS, where the Coombs test is usually...

120. Recurrent disease in the renal graft occur in all cases of

A. focal segmental glomerulosclerosis
B. primary oxalosis
C. membranoproliferative glomerulonephritis type II
D. IgA nephropathy
E. congenital nephrotic syndrome

Ans C

121. Pyelonephritis usually requires total or partial nephrectomy in

A. renal abscess
B. perinephric abscess
C. xanthogranulomatous pyelonephritis
D. pyelonephritic scarring
E. acute lobar nephronia

Ans C

122. Classic bladder exstrophy is characterized by

A. hypospadias
B. narrow based gait
C. normal upper urinary tracts
D. posteriorly displaced anus
E. upward displaced umbilicus is

Ans C The umbilicus is displaced downward, the anus is displaced anteriorly in both sexes, and the wide separation of the pubic rami causes a characteristic broad-based gait but no significant disability. In boys, there is complete epispadias with dorsal chordee; girls also have epispadias, with separation of the 2 halves of the clitoris and wide separation of the labia.

123. The MOST common cause of testicular pain in a 12- yr-old boy is

A. epididymitis
B. testicular tumor
C. testicular torsion
D. scrotal hematoma
E. incarcerated inguinal hernia

Ans C

Acute Kidney Injury

124.Which of following is not seen in nephritic syndrome:

A. Edema
B. Hematuria
C. Hypertension
D. **Hypocholesterolemia**

Ans D Features of nephritic syndrome are: Hematuria and RBC casts, Nephritic range Proteinuria (<3.5g/day), hence edema is present, Hypertension, Uremia and Oliguria.

125.In Henoch-Schönlein purpura renal involvement is not seen generally if no involvement till?

A. 1 month after onset
B. 2 months after onset
C. 3 months after onset
D. **4 months after onset**

Ans D Screening in a patient of HSP:
• Most patients who develop nephritis have urinary abnormalities by 1 month, nearly all by 3 months after onset of HSP
• Therefore, a urinalysis should be performed weekly in patients with HSP during the period of active clinical disease
• Thereafter, a urinalysis should be performed once a month for up to 6 months; after that nephritis is unlikely So among the given options 4 months is the best answer, as it is closest to 6 months.

126.Most common cause of urinary obstruction in a male infant is:

A. Anterior urethral valves
B. **Posterior urethral valves**
C. Stone
D. Stricture

Ans B Most common cause of severe obstructive uropathy in children is posterior urethral valves, affecting 1 in 8,000 boys.

127. Which of the following is true for a child with acute kidney injury?

A. **Urine Specific gravity is higher in pre-renal failure**
B. Schwartz formula uses only serum Cr to calculate GFR
C. Greater loss of sodium through urine in pre-renal failure
D. S. Creatinine value may remain normal 438 Section 3: Systemic Pediatrics

Ans A

128.Renal causes of acute renal failure in children include:

A. Minimal changes disease
B. Renal amyloidosis
C. Pre-eclampsia
D. **Malignant hypertension**
E. Hemolytic uremic syndrome

Ans D e. Hemolytic uremic syndrome with a. Minimal change disease usually does not cause renal failure b. Renal amyloidosis usually presents with nephrotic syndrome or CKD, but is exceedingly rare in children. c. Pre-eclampsia refers to high BP with proteinuria in pregnant females, so this option is not applicable to children. d. Malignant hypertension or as Hypertensive emergency is hypertension with acute impairment 1 or of more organ systems esp CNS, CVS or renal system.

129.Hemolytic-uremic syndrome is a common cause of community acquired acute kidney injury in young children; atypical type may be triggered by

A. toxin-producing E-coli
B. raw milk
C. Streptococcus pneumoniae
D. shiga toxin
E. **cyclosporine**

Ans E

130.Renal cortical necrosis is a rare cause of acute renal failure occurring secondary to extensive ischemic damage of the renal cortex. Of the following, the LEAST common cause is

A. perinatal asphyxia
B. severe congenital heart disease
C. severe hemolytic-uremic syndrome
D. amniotic fluid embolism
E. **infectious endocarditis**

Ans E Less-common causes of cortical necrosis include malaria, extensive burns, snakebites, infectious endocarditis, and medications (e.g., nonsteroidal anti- inflammatory agents). Acute renal cortical necrosis has also been reported to occur in systemic lupus erythematosis–associated antiphospholipid antibody syndrome.

131. Acute kidney injury AKI is a clinical syndrome in which a sudden deterioration in renal function results in the inability of the kidneys to maintain fluid and electrolyte homeostasis. Of the following, the MOST correct matching is

A. low urine sodium (UNa< 20 mEq/L) : intrinsic AKI
B. fractional excretion of sodium >2% : prerenal AKI
C. elevated urine osmolality (UOsm>500 mOsm/kg) : post renal AKI
D. **specific gravity of <1.010 : intrinsic AKI**
E. red blood cell casts :prerenal AKI

Ans D Patients whose urine shows an elevated specific gravity (>1.020), elevated urine osmolality (UOsm>500 mOsm/kg), low urine sodium (UNa< 20 mEq/L), and fractional excretion of sodium <1% (<2.5% in neonates) most likely have prerenal AKI. Those with a specific gravity of <1.010, low urine osmolality (UOsm< 350 mOsm/kg), high urine sodium (UNa> 40 mEq/L), and fractional excretion of sodium >2% (>10% in neonates), red blood cell casts most likely have intrinsic AKI and low urine osmolality (UOsm< 350 mOsm/kg) in Post...

132.Medical management of acute kidney injury include all the following EXCEPT

A. bladder catheter should be placed immediately
B. intravenous administration of boluses isotonic saline
C. furosemide may be administered as a single IV dose
D. fluid restriction is essential if there is no response to a diuretic challenge
E. **mannitol is not effective in prevention of pigment induced renal failure**

Ans E Mannitol may be effective in prevention of pigment (myoglobin, hemoglobin) induced renal failure.

133.Treatment of hypocalcemia in acute kidney injury include all the following EXCEPT

A. low-phosphorus diet
B. **intra venous phosphate binders**
C. calcium should not be given intravenously
D. sevelamer
E. avoid aluminum-based binders

Ans B Hypocalcemia is primarily treated by lowering the serum phosphorus level,phosphate binders should be orally administered to bind any ingested phosphate and increase GI phosphate excretion. Calcium should not be given i.v except in cases of tetany to avoid deposition of calcium salts into tissues.

134.Indications for dialysis in acute kidney injury include all the following EXCEPT

A. anuria/oliguria
B. **persistent hypercalcemia**
C. severe metabolic acidosis
D. volume overload
E. blood urea nitrogen >100-150 mg/dl

Ans B Persistent hyperkalemia.

135.Many patients with acute kidney injury require dialysis support for1-3 wk, which of the following is TRUE statement

A. Intermittent hemodialysis is useful in patients with unstable hemodynamic status
B. continuous renal replacement therapy is useful in patients with stable hemodynamic status
C. in peritoneal dialysis ,hypo-osmolar dialysate is infused into the peritoneal cavity
D. **anticoagulation is not necessary in peritoneal dialysis**
E. urea and creatinine clearance more with peritoneal dialysis than intermittent hemodialysis

Ans D Intermittent hemodialysis is useful in patients with relatively stable hemodynamic status, while continuous renal replacement therapy is useful in patients with unstable hemodynamic status, concomitant sepsis, or multiorgan failure in the intensive care setting, hyperosmolar dialysate is infused into the peritoneal cavity via a surgically or percutaneously placed peritoneal dialysis catheter, urea and creatinine clearance more with intermittent hemodialysis than peritoneal dialysis.

136.Pollakiuria is characterized by

A. dysuria
B. nocturia
C. occurrence at 7-10 yr of age
D. daytime incontinence
E. **voiding every 10-15 min during the day**

Ans E This condition is termed the daytime frequency syndrome of childhood or Pollakiuria; without dysuria, UTI, daytime incontinence, or nocturia. The most common age for these symptoms to occur is 4-6 yr.

Chronic Kidney Disease

137. A child with Malignancy is most likely to have which of the following:

A. **Minimal change disease**
B. Membranous nephropathy
C. Rapidly progressive glomerulonephritis
D. Focal segmental Glomerulosclerosis

Ans A Most likely kidney biopsy finding associated with malignancy in children is Minimal change disease dna Most likely kidney biopsy finding associated with malignancy in Adults is Membranous Glomerulonephropathy.

138.Not seen in HSP:

A. **Thrombocytopenia**
B. Arthritis
C. Abdominal pain
D. Renal involvement

Ans A

139.Most common cause of acute retention of urine in a child of 2 years age:

A. Posterior urethral valve
B. Duplication of renal pelvis
C. **Meatal ulceration with scabbing**
D. Urethral stones

Ans C Meatal ulceration with scabbing can cause acute retention of urine. Dont get confused by option 'a' – Though posterior urethral valve is the most common cause of obstructive uropathy in children, it does not cause acute retention. Posterior urethral valve usually presents with chronic obstructive symptoms like weak urinary stream, distended bladder, uremia or sepsis caused by infection in the obstructed urinary tract.

140.Following parameters are used in Schwartz formula for estimating glomerular filtration rate:

A. 24 hour fluid intake & Urine output
B. Serum creatinine & urine output
C. **Height & serum creatinine**
D. Height & muscle mass

Ans C Schwartz formula is the most widely used pediatric formula for estimating GFR, and is based on serum creatinine, patient height, and an empirical constant. According to Schwartz formula, GFR= k x height (cm)/serum Creatinine (mg/dl), where K is 0.33 in preterm infants, 0.45 in term infants & 0.55 for children

141. **A 10-year-old male (had a boil in the left thigh and mild fever one month ago) presented with headache, malaise, lethargy, lion pain, decreased urine output, and puffiness of the face; GUE: RBC cast ++, albumin ++; low C3 level and normal C4 level. Which of the following is TRUE about his disease?**

A. acute nephritic syndrome 40%
B. nephrotic syndrome 50%
C. acute renal failure 90%
D. ↑ ASO titers 70%
E. **hypertension 70%**

Ans E Acute nephritic syndrome 90%, nephrotic syndrome 10-20%, acute renal failure 50%, the antistreptolysin O titer is commonly elevated after a pharyngeal infection but rarely increases after streptococcal skin infections, hypertension 70%.

142. **Hypercalciuria can be seen in**

A. hypoparathyroidism
B. **corticosteroid therapy**
C. vitamin D deficiency
D. oral thiazide diuretics therapy
E. Addison`s disease

Ans B Hypercalciuria can accompany conditions resulting in hypercalcemia, such as hyperparathyroidism, vitamin D intoxication, immobilization, and sarcoidosis. Hypercalciuria may be associated with Cushing syndrome, corticosteroid therapy, tubular dysfunction secondary to Fanconi syndrome (Wilson disease, oculocerebrorenal syndrome), Williams syndrome, distal renal tubular acidosis, or Bartter syndrome.

143. **Chronic kidney disease in children <5-yr-old is MOST commonly a result of**

A. lupus nephritis
B. familial juvenile nephronophthisis
C. Alport syndrome
D. **focal segmental glomerulosclerosis**
E. autosomal dominant polycystic kidney disease

Ans D After 5 yr of age, acquired diseases (various forms of glomerulonephritis including lupus nephritis) and inherited disorders (familial juvenile nephronophthisis, Alport syndrome) predominate.

144. **Pathogenesis of chronic kidney disease (CKD) include all the following EXCEPT**

A. hyperfiltration injury
B. **hyperkalemia**
C. proteinuria
D. hypertension
E. hyperphosphatemia

Ans B is the result of CKD not a cause.

145. **End-stage renal disease (ESRD) represents the state in which a patient's renal dysfunction has progressed to the point at which homeostasis and survival can no longer be sustained with native kidney function and maximal medical management. Of the following, the TRUE statement is**

A. the ultimate goal for children is dialysis
B. renal replacement therapy be initiated at stage 5 CKD
C. **the time to actually initiate dialysis include impaired school performance**
D. peritoneal dialysis is performed 3 times weekly
E. peritoneal dialysis use cycler-driven therapy increases the risk of peritonitis

Ans C The ultimate goal for children with ESRD is successful kidney transplantation. It is recommended that plans for renal replacement therapy be initiated when a child reaches stage 4 CKD.Hemodialysisis performed 3 times weekly. Peritoneal dialysis use cycler-driven therapy reduction in the number of dialysis catheter connections and disconnections (which decreases the risk of peritonitis).

146. **The risk of renal stone formation increases in the presence of**

A. **low urine pH**
B. urinary citrate
C. urinary magnesium
D. urinary glycosaminoglycan
E. urinary osteopontin

Ans A That's why treatment of renal or ureteral calculus by urinary alkalization (the urine pH should be ≥6.5).Inorganic (e.g., citrate, magnesium) and organic (e.g., glycosaminoglycan's, osteopontin) substances are known to inhibit stone formation. Organic inhibitory compounds adsorb to the surface of the crystal, thereby inhibiting crystal growth and nucleation.

Renal Tubular Disorders

147. **Most common nephropathy in world is?**

A. **IgA nephropathy**
B. FSGS
C. Minimal Change ds
D. Adult PSGN

Ans A • IgA nephropathy is the most common type of glomerulonephritis worldwide
• IgA nephropathy is the most common cause of gross hematuria

148. **Post-streptococcal reactive arthritis is differentiated from Acute Rheumatic Fever by all except:**

A. Small joint involvement and often symmetric
B. **Caused by non-group A hemolytic streptococci**
C. Non-responsiveness to salicylate
D. Shorter incubation period

Ans B

149.A 5 years old male child presents with complaints of fever and abdominal distension. He is having vomiting for the last five days. On examination there are 6–8 pus cells/hpf in urine. WBC count shows 78% neutrophils. What is the best line of management:

A. Send urine for culture and sensitivity and wait for results
B. **Send urine for culture and sensitivity and start IV antibiotics immediately**
C. Send urine for culture, do an USG and start chloroquine
D. Radionuclide studies

Ans B Send urine for culture and sensitivity and start IV antibiotics immediately The given case scenario indicates an upper UTI (pyelonephritis) as the presence of fever in the presence of pus cells in urine indicates renal parenchymal involvement
• In acute febrile infections suggesting pyelonephritis, a 10–14 day course of broad spectrum antibiotics is preferable.
• Parenteral Ceftriaxone/cefotaxime, or ampicillin with an aminoglycoside is preferable.

150.Maximum urine osmolality in preterm neonate:

A. 200
B. 400
C. **500**
D. 800 440 Section 3: Systemic Pediatrics

Ans C Pediatric Urology By John G. Gearhart pg 15-20 Premature infants have an even more limited capacity to concentrate their urine than those born at term, with a maximum urine osmolality of about 500 mOsm/kg H2O. M
• Most commonly diagnosed adenoma during childhood is prolactinomaQ > corticotropinoma >somatotropinoma
• Most common tumor causing hypopituitarism is craniopharyngioma
• Trophic hormone failure with pitui- tary compression or destruc tion: GHQ > FSH > LH > TSH > ACTH

151. Acute post- infectious glomerulonephritis is one of the most common glomerular causes of gross hematuria in children. Of the following, the TRUE statement is

A. **can follow fungal infection**
B. early systemic antibiotic, eliminate the risk of glomerulonephritis
C. all family members of patient need antibiotic treatment
D. antibiotic affect the natural history of the disease
E. recurrences are common

Ans A Acute GN can occur after certain fungal, rickettsial, protozoan, parasitic, or viral diseases (influenza and parvovirus) thatis why the terms APSGN and acute post infectious GN are used synonymously. Early systemic antibiotic therapy for streptococcal throat and skin infections does not eliminate the risk of GN. Family members of patients with acute GN, especially young children, should be considered at risk and be cultured for group A β-hemolytic streptococci and treated if positive.

152.Clinical manifestations of sickle cell nephropathy SSN include all the following

A. hematuria
B. polyuria
C. **hemolytic-uremic syndrome**
D. renal tubular acidosis
E. nephrotic-range proteinuria

Ans C Polyuria caused by a urinary concentrating defect, renal tubular acidosis, and proteinuria associated with the glomerular lesions. Approximately 20-30% of patients with sickle cell disease develop proteinuria, Nephrotic-range proteinuria with or without clinically apparent nephrotic syndrome occurs in up to 30% of patients with SSN, and when present generally heralds progressive renal failure.

153.Treatment of cystinosis is directed at correcting the metabolic abnormalities associated with Fanconi syndrome or chronic renal failure, and the use of

A. cystine eye drops
B. oral cysteine
C. prostaglandin synthesis inhibitor
D. **growth hormone**
E. angiotensin-converting enzyme inhibitors

Ans D Cysteamine, which binds to cystine and converts it to cysteine. Oral cysteamine does not achieve adequate levels in ocular tissues, so additional therapy with cysteamine eye drops is required. Patients with growth failure that does not improve with cysteamine might benefit from treatment with growth hormone. Kidney transplantation is a viable option.

154.Urinalysis in patients with Fanconi syndrome may show all the following EXCEPT

A. **urine pH is alkaline**
B. glycosuria
C. uricosuria
D. phosphaturia
E. elevated urinary sodium

Ans A The urine pH is acidic (<5.5) because distal acidification mechanisms are intact in these patients.

155.Pathogenesis of hyperkalemic (Type IV) renal tubular acidosis is due to

A. hyperaldosteronism
B. **inhibiting ammonia genesis**
C. pseudohyperaldosteronism
D. massive bicarbonate wasting
E. all of the above

Ans B Type IV RTA occurs as the result of impaired aldosterone production (hypoaldosteronism) or impaired renal responsiveness to aldosterone (pseudohypoaldosteronism), proximal RTA pathogenesis is massive bicarbonate wasting. Acidosis results because aldosterone has a direct effect on the H+/ATPase responsible for hydrogen secretion. In addition, aldosterone is a potent stimulant for potassium secretion in the collecting tubule; consequently, lack of aldosterone results in hyperkalemia.

156. **Bartter syndrome is a group of disorders characterized by hypokalemic metabolic alkalosis with hypercalciuria and salt wasting. It can be presented with all of the following EXCEPT**

A. maternal polyhydramnios
B. severe episodes of recurrent dehydration
C. failure to thrive in classic Bartter syndrome
D. deafness is associated with antenatal Bartter syndrome
E. **hypertension due to hyperaldosteronism**

Ans E Blood pressure is usually normal. Patients with the antenatal form can have severe salt wasting, resulting in dehydration and hypotension.

157. **All the following are options in the treatment of Bartter syndrome EXCEPT**

A. potassium supplementation
B. aldosterone antagonist
C. high-sodium diet
D. Indomethacin
E. **kidney transplantation**

Ans E

158. **Gitelman syndrome is a rare cause of autosomal recessive hypokalemic metabolic alkalosis, the distinctive feature is**

A. Hypercalciuria
B. **hypermagnesuria**
C. hypernatremia
D. low aldosterone levels
E. elevated prostaglandin E secretion

Ans B The urinary calcium level is usually very low (in contrast to the elevated urinary calcium level often seen in Bartter syndrome), hypokalemia, aldosterone levels usually normal, prostaglandin E secretion is not elevated.

RTA (Renal Tubular Acidosis)

159. **A 10 year old child presents with edema and decreased urine output. On evaluation, serum albumin is 2.5 g/dL, S.Creatinine is 0.5 mg/dL, urine protein is 3+ with no RBC casts. Pathological change expected is?**

A. **Minimal change disease**
B. Interstitial nephritis
C. IgA nephropathy
D. FSGS.

Ans A

160. **Which of the following is not seen in Henoch-Schonlein purpura?**

A. Purpura
B. Abdominal pain
C. **Seizure**
D. Arthralgia

Ans C

161. **Least likely cause of renal papillary necrosis?**

A. Sickle cell disease
B. Analgesic nephropathy
C. **Posterior urethral valves**
D. Diabetes with UTI

Ans C Sickle cell disease, use of analgesics and Diabetes are important causes of Renal papillary necrosis.

162. **The part of the glomerulus which serves as a supporting and probably has a role in the regulation of glomerular blood flow, filtration, and removal of macromolecules is the**

A. glomerular basement membrane
B. **mesangium**
C. bowman's capsule
D. parietal epithelial cell
E. visceral epithelial

Ans B

163. **The MOST common etiology of secondary membranous nephropathy (MN) in children is**

A. neuroblastoma
B. measles
C. **systemic lupus erythematosis**
D. chronic hepatitis C
E. penicillin

Ans C MN also associated with chronic hepatitis B infection and congenital syphilis, other chronic infections, including malaria, which is likely the most common cause of nephrotic syndrome worldwide. Certainmedications, such as penicillamine and gold, or chronic factor replacement in patients with hemophilia. Rare causes such as neuroblastoma, or other idiopathic systemic diseases.

164. **Autosomal dominant polycystic kidney disease is the most common hereditary human kidney disease, all the following are true EXCEPT**

A. gross or microscopic hematuria
B. may be seen in neonates
C. abnormal renal sonography in the absence of symptoms
D. is a multiorgan disorder
E. **right sided valvular heart disease**

Ans E Mitral valve prolapse is seen in approximately 12% of children; aortic and coronary artery aneurysms and aortic valve insufficiency are noted in affected adults. It is a systemic disorder with possible cyst formation in multiple organs (liver, pancreas, spleen, brain) and the development of saccular cerebral aneurysms.

165. **Distal RTA can be presented with**

A. hypocalciuria
B. hypercitraturia
C. phosphaturia
D. conductive deafness
E. **growth failure**

Ans E Hypercalciuria, hypocitraturia, and sensorineural deafness are seen in distal RTA while phosphate and massive bicarbonate wasting are characteristic of proximal RTA.

166. **Urinary indices in patients with type IV RTA include all the following EXCEPT**

A. may be alkaline or acidic
B. elevated sodium level
C. inappropriately low potassium level
D. foul-smell
E. **aminoaciduria**

Ans E Urinary indices in patients with Fanconi syndrome demonstrate varying degrees of aminoaciduria, and elevated urinary sodium or potassium; while in type IV RTA elevated urinary sodium levels with inappropriately low urinary potassium levels reflect the absence of aldosterone effect.

167. **Polyomavirus nephropathy is an important cause of allograft dysfunction; the increased incidence is thought to be the result of**

A. reduction in immunosuppression doses
B. **more potent immunosuppressive regimens**
C. cidofovir use
D. leflunomide use
E. ciprofloxacin therapy

Ans B That is why reducing immunosuppression is the main form of therapy, and cidofovir and leflunomide are used as adjunctive therapies.

168. **Irritative symptoms of dysuria, urgency, and frequency usually indicate a calculus in the**

A. urethra
B. bladder
C. **distal ureter**
D. middle part of ureter
E. ureteropelvic junction

Ans C If the stone passes into the bladder, the child usually is asymptomatic. if the stone is in the urethra, dysuria and difficulty voiding can result, particularly in boys.

Nephrogenic DI & Diabetes Insipidus

169. **Mutation in NPHS1 gene causes:**

A. Alport syndrome
B. **Congenital Finnish type nephrotic syndrome**
C. Focal segmental glomerulosclerosis
D. Nail patella syndrome

Ans B

170. **Which of the following is/are not the features of Henoch-Schonlein purpura (HSP)?**

A. Abdominal pain
B. Splinter hemorrhage
C. **Thrombocytopenia**
D. Epistaxis
E. Arthritis

Ans C • HSP is characterized by a tetrad of palpable purpura, arthritis, glomerulonephritis and abdominal pain
• Less common skin lesions are: macules, vesicles, bullous lesions, splinter hemorrhage and ulcerations; Rarely, epistaxis may occur.

171. **Straining and dribbling of urine in a male infant with recu rrent urinary infection should lead to the suspicion of:**

A. Vesico-ureteric reflux
B. **Posterior urethral valve**
C. Pelvic ureteric junction obstruction
D. Phimosis

Ans B Straining and Dribbling suggest the diagnosis of obstructive uropathy, for which posterior urethral valve is the commonest cause.

172. **Collectively, the glomerular endothelial cell, basement membrane, and podocytes form the**

A. glomerulus
B. **filtration membrane**
C. juxtamedullary nephron
D. nephron
E. renal corpuscle

Ans B

173. **The well-known complication of membranous nephropathy is**

A. **renal vein thrombosis**
B. hypertension 2%
C. hematuria acute renal failure
D. gross hematuria
E. nephritic syndrome

Ans A A subset of patients with MN present with a major venous thrombosis, commonly renal vein thrombosis, this well-known complication. Approximately 20% of children have hypertension at presentation, most patients also have microscopic hematuria and only rarely present with gross hematuria. MN is most common in the 2nd decade of life, but it can occur at any age, including infancy.The disease usually manifests as nephrotic syndrome and accounts for 2-6% of all cases of childhood nephrotic syndrome.

174. **Hemorrhagic cystitis can occur in response to all the following EXCEPT**

A. cyclophosphamide
B. adenovirus infection
C. **cyclosporine**
D. polyoma BK virus infection
E. amyloidosis

Ans C The hemorrhagic cystitis can occur in response to chemical toxins (cyclophosphamide, penicillin's, busulfan, thiotepa, dyes, insecticides), viruses (adenovirus types 11 and 21 and influenza A), radiation, and amyloidosis. The polyoma BK virus, present latently in immunocompetent hosts, is associated with the development of drug-induced cystitis in immunosuppressed patients.

175. **Nephrogenic diabetes inspidus is a disorder of water metabolism characterized by an inability to concentrate urine, even in the presence of antidiuretic hormone. Of the following, the MOST correct statement is**

A. most common inheritance is autosomal dominant
B. **can be caused by hypokalemia**
C. mutations in the AQP2 gene in X-linked form
D. irritability and crying are uncommon features
E. secondary form present with hyponatremia

Ans B The most common pattern of inheritance in congenital NDI is an X-linked recessive disorder, mutations in the AQP2 gene have been identified in patients with the rarer autosomal dominant and recessive forms, Irritability and crying are common features, patients with the secondary form generally present later in life, primarily with hypernatremia and polyuria.

176. **All the following are options in the treatment of nephrogenic diabetes insipidus**

A. free access to water
B. **furosemide**
C. hydrochlorothiazide
D. amiloride
E. indomethacin

Ans B

177. **Neuropathic bladder dysfunction in children is usually congenital resulting from neural tube defects or other spinal abnormalities. All the following are options in the treatment EXCEPT**

A. botulinum toxin
B. **cholinergic drugs**
C. cutaneous vesicostomy
D. antimicrobial prophylaxis
E. clean intermittent catheterization

Ans B Treatment includes reduction of bladder pressure with anticholinergic drugs (e.g., oxybutynin, 0.2 mg/kg/24 hr in 2 or 3 divided doses).

178. **Uric acid stones occur in**

A. **inflammatory bowel disease**
B. corticosteroids therapy
C. vitamin D excess
D. distal renal tubular acidosis
E. hyperoxaluria

Ans A Others distracters are causes of calcium stones.

Wilms Tumor

179. **In which one of the primary Glomerulonephritides the glomeruli are normal by light microscopy but shows loss of foot processes of the visceral epithelial cells and no deposits by electron microscopy**

A. Poststreptococcal glomerulonephritis
B. Membranoproliferative glomerulonephritis type
C. IgA nephropathy
D. **Minimal change disease**

Ans D

180. **A 8 years old male had non blanching rashes over the skin and swelling of knee joint with hematuria +++ and protein + in urine. Microscopic analysis of his renal biopsy specimen is most likely to show:**

A. Tubular necrosis
B. Visceral podocyte fusion
C. **Mesangial deposits of IgA**
D. Basement membrane thickening

Ans C The given case scenario (palpable purpura, arthritis, hematuria and proteinuria) suggests a diagnosis of Henoch Schonlein purpura In H.S. purpura and IgA nephropathy, IgA deposits are present in mesangium (by immunofluorescence)

181. **A boy suffering from acute pyelonephritis, most specific Questions urinary finding will be:**

A. Leukocyte esterase test
B. WBC casts
C. Nitrite test
D. **Bacteria in gram stain**

Ans D • UTI may be suspected based on symptoms or findings on urinalysis, or both.
• A urine culture is necessary for confirmation and appropriate therapy. Hence, although WBC casts in urine indicate renal parenchymal involvement and both leukocyte esterase and Nitrite test indicate UTI, identification of bacteria on gram staining while doing urine culture is the most specific urinary finding.

182. **The ultra-filtrate which is filtered through the glomerular capillary walls contains all of the following EXCEPT**

A. electrolytes
B. low-molecular-weight proteins
C. creatinine
D. **albumin**
E. glucose

Ans D The ultra-filtrate, which is cell free, contains all of the substances in plasma (electrolytes, glucose, phosphate, urea, creatinine, peptides, and low-molecular-weight proteins except proteins having a molecular weight of ≥68 kDa *such as albumin and globulins]).

183. **Type I membranoproliferative glomerulonephritis is**

A. 100% of patients
B. 80% of patients
C. 60%of patients
D. 40% of patients
E. **20% of patients**

Ans E Type II MPGN (dense deposit disease) is less common than type I membranoproliferative glomerulonephritis, not to be mediated by immune complexes, demonstrate an associated systemic disease called partial lipodystrophy, serum C3 complement levels are low in the majority of cases(while in membranous nephropathy is normal), Patients present in equal proportions with nephrotic syndrome, acute nephritic syndrome, or persistent asymptomatic microscopic hematuria and proteinuria (while in membranous nephropathy usually...

184. **All the following are treatment options of hemorrhagic cystitis EXCEPT**

A. **antibiotic**
B. intensive intravenous hydration
C. forced dieresis
D. analgesia
E. spasmolytic drugs

Ans A

185. **Risk factor for renal graft thrombosis include**

A. **prior history of peritoneal dialysis**
B. arterial hypertension
C. donor age (>2 yr.)
D. recipient age (>5 yr.)
E. cold ischemia time <24 hr.

Ans A

186. **Symptoms of cystitis include**

A. fever
B. polyuria
C. nausea
D. **suprapubic pain**
E. urinary retention

Ans D Symptoms include dysuria, urgency, frequency, suprapubic pain, incontinence, and malodorous urine. Cystitis does not cause fever and does not result in renal injury; malodorous urine is not specific for a UTI.

187. **Complications of augmentation cystoplasty includes all the following EXCEPT**

A. bladder calculi
B. **metabolic alkalosis**
C. malignant neoplasm
D. urinary tract infection
E. spontaneous perforation

Ans B The enteric mucosal surface in contact with the urine absorbs ammonium, chloride, and hydrogen ions and loses potassium; hyperchloremic metabolic acidosis can result, possibly requiring medical treatment.

188. **The MOST accurate study in a child with suspected renal stone is**

A. renal ultra-sonogram
B. plain radiograph of the abdomen
C. **an unenhanced spiral CT scan**
D. radioisotope studies
E. magnetic resonance urography

Ans C This study takes only a few minutes to perform, has 96% sensitivity and specificity in delineating the number and location of calculi, and demonstrates whether the involved kidney is hydronephrotic or not.

Posterior Urethral Valves

189. **A 7-year-old girl is brought with complains of generalized swelling of the body. Urinary examination reveals grade 3 proteinuria and the presence of hyaline and fatty casts. She has no history of hematuria. Which of the following statements about her condition is true?**

A. **No IgG deposits or C3 deposition on renal biopsy**
B. Her C3 level will be low
C. IgA nephropathy is the likely diagnosis
D. Alport's syndrome is the likely diagnosis

Ans A This is a classic presentation of Minimal change nephrotic syndrome (MCNS); In MCNS, light microscopy is normal and glomerular deposits are not seen on immunofluorescence examination. About the other options. Disease Characteristic features IgA nephropathy Recurrent gross hematuria is the typical manifestation On light microscopy, focal segmental proliferation of mesangial matrix is seen Immunofluorescence shows mesangial deposits of IgA Alport syndrome X–linked recessive;

190. **Most common cause of renal artery stenosis in children in India is:**

A. **Takayasu aortoarteritis**
B. Fibromedial hypertrophy
C. Fibrointimal hyperplasia
D. Polyarteritis nodosa

Ans A Renovascular hypertension and Renal artery stenosis:
• The commonest cause for renovascular hypertension in children reported in western literature is fibromuscular dysplasia (FMD) and the midaortic syndrome.
• However, in India, Takayasu disease (idiopathic aortoarteritis) is the most important cause, accounting for upto 87% of children with renovascular hypertension.
• Renal artery stenosis may also occur in association with neurofibromatosis, Williams syndrome, Marfan syndrome, rubella syndrome,

191. **Which of the following is the most appropriate method for obtaining a urine specimen for culture in an 8 month old girl?**

A. **Suprapubic aspiration**
B. Indwelling catheter sample
C. Clean catch void
D. Urinary bag sample

Ans A O.P. Ghai: "Urine is obtained by suprapubic bladder aspiration or urethral catheterization in children below 2 years." Pediatric Nephrology protocols, AIIMS says: "In neonates and infants, the best technique for obtaining an uncontaminated urine specimen is supra pubic aspiration from the bladder."

192. **In contrast to the concentration of blood urea nitrogen, the serum creatinine level is primarily influenced by**

A. state of hydration
B. nitrogen balance
C. **muscle mass**
D. hemorrhage
E. vomiting

Ans C Creatinine is affected by sex, height, muscle mass, bilirubin, and red blood cell hemolysis, while the concentration of blood urea nitrogen is affected by state of hydration and nitrogen balance.

193. **WHO classification of lupus nephritis is based on a combination of features including light microscopy, immunofluorescence, and electron microscopy. Of the following, the WORST outcome is associated with**

A. class I nephritis
B. class II nephritis
C. class III nephritis
D. **class IV nephritis**
E. class V nephritis

Ans D Renal biopsy should be performed in all patients with SLE because histopathologic findings are used to determine the selection of specific immunosuppressive therapies.

194.In exercise hematuria; one of the following is TRUE

A. A blood clots rarely seen in urine
B. no dysuria
C. abnormal findings on cystoscopy
D. resolution after 7 days
E. less common in males

Ans A Findings on urine culture, intravenous pyelography, voiding cystourethrography, and cystoscopy are normal in most patients, hematuria generally resolves within 48 hr. after cessation of exercise, exercise hematuria is less common in females and can be associated with dysuria.

195.Antithymocyte globulin and thymoglobulin act against

A. interleukin-2 Receptor Antibodies
B. T-lymphocyte antigens
C. B cells
D. CD20 epitope
E. mammalian target of rapamycin

Ans B

196.In pyelonephritis, a high risk of renal scarring is associated with an elevated

A. erythrocyte sedimentation rate
B. C-reactive protein
C. procalcitonin
D. white blood cell count
E. serum ferritin

Ans C Others distracters are nonspecific markers of inflammation.

197. Posterior urethral valves, is the most common cause of severe obstructive uropathies in children. Of the following, the MOST appropriate statement

A. its incidence 1 in 40,000 boys
B. vesicoureteral reflux occurs in 5% of patients
C. the urinary stream is strong
D. Foley catheter should be avoided
E. it is adiverticulum in the penile urethra

Ans D Foley (balloon) catheter should not be used, because the balloon can cause severe bladder spasm, which can produce severe ureteral obstruction, affecting 1 in 8,000 boys. Vesicoureteral reflux occurs in 50% of patients and the urinary stream is weak.The urethral valves are tissue leaflets fanning distally from the prostatic urethra to the external urinary sphincter.

198.The normal values for 24-hr urine calcium is

A. <4mmol/1.73 m2
B. >4mmol/1.73 m2
C. <4 mg/kg
D. >4 mg/kg
E. = 4 mg/kg

Ans C Hypercalciuria if >4 mg/kg/24 hr.

Hypospadias & Epispadias

199.A 7 years old girl is brought with complaints of generalized Questions swelling of the body. Urinary examination reveals grade 3 proteinuria and presence of hyaline and fatty casts. She has no history of hematuria. Which of the following statements about her condition is true?

A. No IgG deposits or C3 deposition on renal biopsy
B. C3 level will be low
C. IgA nephropathy is the likely diagnosis
D. Alport's syndrome is the likely diagnosis.

Ans A

200.A 3 years old child presents with four days history of puffi ness of face, fever and tea colored urine. During the course of his disease, he can have any of the compli- cations except:

A. Hypokalemia
B. Hypertensive encephalopathy
C. Acute renal failure
D. Acidosis

Ans A It is a case of acute nephritic syndrome which can result in Hypertensive encephalopathy, Acute renal failure and metabolic acidosis.

201.Which one of the following statement is FALSE with regard to pyuria in children:

A. Presence of more than 5 WBC/hpf for girls and more than 3 WBC/hpf for boys
B. Infection can occur without pyuria
C. Pyuria maybe present without UTI
D. Isolated pyuria is neither confirmatory nor diagnostic for urinary tract infection

Ans A • Pyuria suggests infection, but infection can occur in the absence of pyuria; Conversely, pyuria can be present without UTI;
• Sterile pyuria (positive leukocytes, negative cultures) occurs in partially treated bacterial UTI, viral infections, renal tuberculosis, renal abscess, interstitial nephritis, inflammation near ureter or bladder In an uncentrifuged urine specimen > 5 WBCs/hpf is abnormal and is called pyuria.

202.Mesangial cell disease include all the following EXCEPT

A. IgA nephropathy
B. membranous nephropathy
C. mesangioproliferative glomerulonephritis
D. diabetic nephropathy
E. class II lupus nephritis

Ans B result from epithelial cell injury, epithelial cells, in conjunction with basement membrane, allow filtration of plasma solutes but retard passage of cells and plasma proteins. Disease related to these cells is typified by the presence of sub epithelial deposits and flattening of the foot processes that engage the basement membrane, resulting in disruption of the filtration barrier and proteinuria.

203.In treatment of lupus nephritis, plasmapheresis is effective in

A. all patients
B. classes III and IV
C. maintenance therapy
D. reducing proteinuria
E. **accompanying thrombotic thrombocytopenic purpura**

Ans E Plasmapheresis is ineffective in lupus nephritis unless there is accompanying thrombotic thrombocytopenic purpura or antineutrophilic cytoplasmic antibody associated disease.

204.False-ngative results in urine dipstick measurement of protein can occur in

A. very high urine pH (>7.0)
B. **large volume of urine output**
C. contamination of the urine with blood
D. highly concentrated urine specimen
E. contamination of the urine with antiseptic

Ans B Dilute urine or a large volume of urine output or in disease states in which the predominant urinary protein is not albumin cause false-negative test results.

205.The side-effect profile of cyclosporine in children include all the following EXCEPT

A. hypertrichosis
B. gingival hyperplasia
C. **neutropenia**
D. coarsening facial features
E. hyperlipidemia

Ans C Gastrointestinal and hematologic side effects occur in mycophenolate mofetil, but not in cyclosporine.

206.Acute cystitis should be treated promptly to prevent possible progression to pyelonephritis. Of the following, the LEAST effective drug is

A. trimethoprim-sulfamethoxazole
B. cephalexin
C. ciprofloxacin
D. **nitrofurantoin**
E. amoxicillin

Ans D should not be used routinely in children with a febrile UTI because it does not achieve significant renal tissue levels.

207.In children up to the age of 14 yr, the mean bladder capacity in ounces is equal to the age (in years) plus

A. **2**
B. 3
C. 4
D. 5
E. 6

Ans A

208.Secondary hyperoxaluria can occur in patients with

A. furosemide therapy
B. sarcoidosis
C. **pyridoxine deficiency**
D. primary hyperparathyroidism
E. immobility

Ans C Others distracters are causes of Hypercalciuria.

Cryptorchidism

209.A 2 year old child presents with hematuria. There is also history of cough and cold 2-3 days back. Which of the following is the most likely electron microscopy picture?

A. **Irregular focal thickening of the glomerular basement membranes**
B. Absence of IgG deposits and/or complement on immunofluorescence.
C. Subepithelial humps
D. Segmental sclerosis

Ans A The given history of hematuria 2-3 days after an upper respiratory infection suggests a diagnosis of IgA Nephropathy. Electron microscopy in IgA nephropathy reveals electron microscopy reveals diffuse thickening, thinning, splitting, and layering of the glomerular and tubular basement membranes fo 59. b. Conservative therapy weiveR The given clinical picture suggests a diagnosis of nephritic syndrome, most probably post-streptococcal glomerulonephritis (PSGN). Management of PSGN involves:

210.The most common leukocytoclastic vasculitis affecting children is:

A. Takayasu disease
B. Mucocutaneous lymph node syndrome (Kawasaki disease)
C. **Henoch-Schonlein purpura**
D. Polyarteritis nodosa

Ans C Henoch-Schonlein purpura (HSP) is the most common vasculitis of childhood and is characterized by leukocytoclastic vasculitis and IgA deposition in the small vessels in skin, joints, GIT and kidney.

211. Which one of the following statements is false with regard to xanthogranulomatous pyelonephritis in children:

A. **Often affects those younger than 8 years of age**
B. It affects the kidney focally more frequently than diffusely
C. Boys are affected more frequently
D. Clinical presentation in children is same as in adults

Ans A Xanthogranulomatous pyelonephritis
• It is a form of chronic pyelonephritis characterized by destruction of renal parenchyma and the presence of granulomas, abscesses and collection of lipid laden foamy macrophages (foam cells)
• Rare in children; more common in adults; Focal form is more common in children;
• Those who are affected diffusely, present with non-specific symptoms like Weight loss, Fever, Lethargy, Failure to thrive
• Proteus is the most common causative organism.

212.Hematuria is defined as the presence of at least 5 red blood cells per microliter of urine, false-positive results may be seen in urinary dipstick reading in all the following

A. an alkaline urine (pH > 8)
B. contamination with hydrogen peroxide
C. **presence of formalin**
D. hemoglobinuria
E. myoglobinuria

Ans C False negative results can occur in the presence of formalin (used as a urine preservative) or high urinary concentrations of ascorbic acid (i.e., in patients with vitamin C intake >2000 mg/day).

213.Henoch-Schönlein purpura is the most common small vessel vasculitis in childhood, aggressive therapy may be reasonable in those with

A. isolated microscopic hematuria
B. insignificant proteinuria
C. severe systemic manifestations
D. **>50% crescents on renal biopsy**
E. IgA deposits intensity

Ans D Spontaneous and complete resolution of the nephritis typically occurs in the majority of patients with mild initial manifestations (isolated hematuria with insignificant proteinuria), and the severity of the systemic manifestations does not correlate with the severity of the nephritis.

214.Which of the following methods offer more precise information regarding urine protein excretion?

A. frothy urine
B. dipstick testing
C. micro albuminuria
D. spot urine for protein/creatinine ratio
E. **24 hr. urine for protein and creatinine excretion**

Ans E

215.Corticosteroids remain an integral part of many immunosuppressive protocols despite their multifaceted toxicities. In children, the MOST noteworthy side effect is

A. obesity
B. hypertension
C. aseptic necrosis of bone
D. **retarded skeletal growth**
E. cushingoid faces

Ans D

216.All the following are indications for admission to hospital in patients with urosepsis

A. dehydration
B. unable to drink fluids
C. neonatal period
D. vomiting
E. **microscopic hematuria**

Ans E is common in acute cystitis, but micro hematuria alone does not suggest UTI.

217. The MOST common cause of daytime incontinence is

A. **an overactive bladder**
B. infrequent voiding
C. detrusor–sphincter dyssynergia
D. bladder outlet obstruction
E. behavioral

Ans A

218.The consequences of cryptorchidism include all the following EXCEPT

A. infertility
B. **hydrocele**
C. inguinal hernia
D. testicular malignancy
E. poor testicular growth

Ans B Retractile testes may be misdiagnosed as undescended testes.

Hydrocele & Hernia

219.A 10 years boy presented with Cola coloured urine, oliguria for 3 days, facial puffiness, edema and hypertension. Urine albumin is positive and C3 levels are reduced. BP is 130/80. He had skin infection two weeks back. What is the next appropriated management?

A. Renal biopsy
B. **Conservative therapy**
C. Cyclosporine
D. Steroids

Ans B This presentation (cola-colored urine, oliguria, edema, hypertension, low C3, preceding skin infection) is classic post-streptococcal glomerulonephritis, which is generally managed conservatively/supportively as it is usually self-limited.

220.Henoch-Schonlein purpura is characterized by the deposition of the following immunoglobulin around the vessels:

A. IgM
B. IgG
C. **IgA**
D. IgE

Ans C

221.A 4-year-old male child presents with fever, anemia and 139. azotemia after an episode of dysentery 9 days earlier. The commonest organism responsible for this condition is?

A. Meningococcus
B. **E.coli**
C. E. histolytica
D. Staphylococcus 140.

Ans B E. coli The given condition is most likely HUS for which the most common organism is E. coli, among the given options

222.Rhabdomyolysis is always clinically significant as it may lead to acute renal injury, it can occurs secondary to

A. **severe electrolyte abnormalities**
B. bacterial endocarditis
C. transfusion reactions
D. mycoplasma infection
E. autoimmune hemolytic anemia

Ans A Rhabdomyolysis can occurs secondary to viral myositis, crush injury, severe electrolyte abnormalities (hypernatremia, hypophosphatemia), hypotension, disseminated intravascular coagulation, toxins (drugs, venom), metabolic disorders of muscles, and prolonged seizures. While other distractors are causesof hemolysis and hemoglobinuria.

223.All the following diseases can cause a pulmonary-renal syndrome EXCEPT

B. systemic lupus erythematosis
C. Henoch-Schönlein purpura
D. microscopic polyangiitis
E. **granulomatosis with polyangiitis F. IgA nephropathy**

Ans E

224.Transient proteinuria can be seen in all the following EXCEPT

A. temperature >38.3°C
B. exercise
C. **over hydration**
D. cold exposure
E. seizures

Ans C Defined contributing factors include a temperature >38.3°C (101°F), exercise, dehydration, cold exposure, heart failure, seizures, or stress.

225.The following formulas are true EXCEPT

A. sodium requirement= 0.6 ×weight in kg ×(125− serum sodium in mEq/L)
B. blood anion gap=*Na+ − *Cl +HCO3+
C. blood anion gap=*Na − Cl -HCO3]
D. urine anion gap (*urine Na + urine K++ − urine Cl−)
E. **estimated GFR = 0.43 × height in m/serum creatinine in mg/dL**

Ans E Schwartz formula for estimated GFR = 0.43 × height in cm/ serum creatinine in mg/Dl.

226.The antenatal hydronephrosis is graded by the trimester and the antero-posterior diameter of the renal pelvis. Of the following, the MOST likely cause is

A. vesicoureteral reflux
B. ureteropelvic junction obstruction
C. **transient hydronephrosis**
D. posterior urethral valve
E. ureterovesical junction obstruction

Ans C

227.Staccato urinary stream is seen in

A. hypospadias
B. meatal stenosis
C. ureteral ectopia
D. posterior urethral valves
E. **non neurogenic bladder**

Ans E In posterior urethral valves the urinary stream is weak. Hypospadias is typically ventral deflection or severe splaying. Meatal stenosis; if the meatus is pinpoint, boys void with a forceful, fine stream that goes a great distance. Ureteral ectopia characterized by constant urinary dripping all day.

228.Cystinuria is characterized by

A. X-linked recessive inheritance
B. decreased cystine urinary excretion
C. renal tubular acidosis
D. alkaline urine
E. **faint radiopaque renal calculi**

Ans E Autosomal recessive disorder of the epithelial cells of the renal tubule that prevents absorption of the 4 dibasic amino acids (cystine, ornithine, arginine, lysine) and results in excessive urinary excretion of these products. The only known complication of this familial disease is the formation of calculi, because of the low solubility of cystine; the patients usually have acidic urine, which leads to a higher rate of precipitation.

Enuresis

229.Following all are syndromes associated with renal disease except

A. Lowe Syndrome
B. Denys-Drash Syndrome
C. Alport Syndrome
D. **Sinding-Larsen-Johansson Syndrome**

Ans D Discussing about the options one by one, Options Clinical manifestations a. Lowe Syndrome Oculocerebrorenal syndrome b. Denys-Drash Syndrome Wilms tumor in kidney c. Alport Syndrome Hematuria, eye defects and sensorineural hearing loss d. Sinding-Larsen – Johansson Syndrome Point tenderness at the inferior pole of the patella associated with focal swelling

230.95. A picture resembling microthrombi in renal vasculature occurs in:

A. **Pre eclampsia**
B. Henoch-schonlein purpura
C. Benign hypertension
D. Hemolytic uremic syndrome 105.
E. Malignant hypertension

Ans A Posterior urethral valve Voiding cystourethrogram in an infant with posterior urethral valves showing the dilation of the prostatic urethra and the transverse linear filling defect corresponding to the valves.

231.In children, renal failure in terms of urine output is defined as?

A. **Less than 0.3 ml/kg/hr**
B. Less than 0.5 ml/kg/hr
C. Less than 0.8 ml/kg/hr
D. Less than 1 ml/kg/hr 141.

Ans A Acute kidney injury (AKI) in children is defined as eCCl (estimated creatinine clearance) decrease by 75% or eCCl <35 mL/ min/1.73 m2 or Urine output <0.3 mL/kg/hr for 24 hr or anuric for 12 hr

232.Glomerular hematuria is associated with

A. **urinary microscopic findings of RBC casts**
B. minimal proteinuria on dipstick (<100 mg/dL)
C. gross hematuria that is bright red or pink
D. the presence of leukocytes cast
E. normal urinary RBC morphology

Ans A Hematuria from within the glomerulus is often associated with urinary microscopic findings of RBC casts, and deformed urinary RBCs (particularly acanthocytes), brown, cola- or tea-colored, or burgundy urine, and proteinuria >100 mg/dLvia dipstick. Hematuria originating within the tubular system may be associated with the presence of leukocytes or renal tubular casts.

233.In hemolytic-uremic syndrome, thrombotic microangiopathies are associated with all the following EXCEPT

A. genetically determined factor H deficiency
B. neuraminidase-producing Streptococcus pneumonia infection
C. systemic lupus erythematosis
D. malignancy
E. **hypotension**

Ans E Malignant hypertension.

234. **Orthostatic proteinuria is increased amounts of protein in the upright position. Of the following, the MOST appropriate answer is**

A. usually symptomatic
B. hematuria may be present
C. **renal dysfunction are absent**
D. the cause is well-known
E. edema usually positive

Ans C The absence of proteinuria (dipstick negative or trace for protein; and a normal ratio of urinary protein [mg/dL] to urinary creatinine [mg/dL]= [uPr/uCr] <0.2) on the first morning urine sample for 3 consecutive days confirms the diagnosis of orthostatic proteinuria, children with this condition are usually asymptomatic;

235. **Glomerular filtration rate in full term neonate is**

A. 15 mL/min/1.73 m2
B. **25 mL/min/1.73 m2**
C. 35mL/min/1.73 m2
D. 45 mL/min/1.73 m2
E. 55 mL/min/1.73 m2

Ans B

236. **Ureteropelvic junction obstruction is the most common obstructive lesion in childhood. Of the following, the MOST correct statement is**

A. it is usually caused by extrinsic stenosis
B. it mainly occurs on the right side
C. there is a female preponderance
D. **voiding cystourethrogram (VCUG) is necessary**
E. it is usually bilateral

Ans D VCUG is necessary because 10-15% of patients have ipsilateralvesicoureteral reflux.

237. **Family history in nocturnal enuresis is positive in**

A. 10%
B. 30%
C. **50%**
D. 70%
E. 90%

Ans C

238. **The pathogenesis of nocturnal enuresis is**

A. defective sleep arousal
B. nocturnal polyuria
C. genetic factors
D. overactive bladder
E. **multifactorial**

Ans E

239. **The MOST effective way of treatment in older children with nocturnal enuresis is**

A. motivational therapy
B. **conditioning therapy**
C. desmopressin acetate
D. oxybutynin chloride
E. imipramine

Ans B This form of therapy has a reported success of 30-60%, although the relapse rate is significant, while motivational therapy; there is no evidence that this approach is beneficial, Pharmacologic therapy is intended to treat the symptom of enuresis and thus is regarded as second line and is not curative.

Renal Transplantation

240. **A 5-year-old boy presents with hematuria 2 days after sore throat. Most probable cause is?**

A. Post-streptococcal glomerulonephritis
B. **Berger disease**
C. Paroxysmal nocturnal Hemoglobinuria
D. Microscopic polyangitis

Ans B IgA Nephropathy or Berger disease presents as Gross hematuria 1-2 daysQ after a respiratory, urinary tract or GI infection.

241. **A child has diarrhea since 8 days. He is dehydrated and urine output is reduced. Which of the following is not correct regarding the renal failure in this patient?**

A. **Urinary sodium > 40 mEq/L**
B. Urinary osmolality > 500 mosm/L
C. FENa < 1%
D. BUN/Creatinine >

Ans A The child in question has pre-renal azotemia, urinary sodium should be less than 20 meq/L. Classic laboratory findings in AKI U Type UOsm Na Fe BUN/ Na Cr Prerenal >500 <10 <1% >20 Intrinsic <350 >20 >2% <15 Postrenal <350 >40 >4% >15

242. **In hematuria, renal biopsy is indicated in all the following EXCEPT**

A. some children with persistent microscopic hematuria
B. decreased renal function
C. proteinuria
D. hypertension
E. **persistent high serum complement C3**

Ans E Renal biopsy is indicated for children with persistent low serum complement C3.

243. **In the treatment of diarrhea-associated hemolytic-uremic syndrome. One of the following options has no beneficial role**

A. dialysis
B. early intravenous volume expansion
C. control of hypertension
D. **eculizumab**
E. red cell transfusions

Ans D is an anti-C5 antibody that inhibits complement activation, a pathway that contributes to active disease in some forms of atypical familial HUS, Eculizumab is FDA approved for the treatment of atypical HUS, while initial reports suggested that eculizumab provided benefit in patients with diarrhea associated HUS, subsequent systematic analysis showed no benefit from either plasma exchange or eculizumab.

244.All the following matching are true EXCEPT

A. proximal RTA----trimethoprim
B. distal RTA----amphotericin B
C. hyperkalemic RTA----cyclosporine
D. distal RTA----lithium
E. proximal RTA----gentamicin

Ans A Hyperkalemia RTA---- Trimethoprim/sulfamethoxazole.

245.Kidney transplantation is the optimal therapy for children with end-stage renal disease, all the following are true EXCEPT

A. relative contraindication is preexisting metastatic malignancy
B. successful transplantation leads to improvement in their linear growth
C. preemptive transplantation mean transplantation without prior dialysis
D. optimal outcomes result when the child weighs >15 kg
E. urologic problems should be addressed before surgery

Ans D Optimal outcomes result when the child weighs ≥10 kg.

246.Indications for bilateral native nephrectomies before renal transplantation include

A. Denys-Drash syndrome
B. IgA nephropathy
C. chronic glomerulonephritis
D. Prune belly syndrome
E. medullary cystic disease

Ans A Owing to the high risk of developing Wilms tumor, patients with Denys-Drash syndrome should undergo bilateral nephrectomy before transplantation.

247.Before renal transplantation, hemoglobin levels should be maintain at the level between

A. 7 and 8 g/dL
B. 9and 10 g/dL
C. 11 and 12 g/dL
D. 13 and 14 g/dL
E. 15 and 16 g/dL

Ans C

248.Infections need to be identified and treated before renal transplantation. Infectious disease screening includes all the following EXCEPT

A. tuberculosis skin test
B. cytomegalovirus IgG
C. measles antibody
D. hepatitis A serology
E. varicella titer

Ans D Infections need to be identified and treated before renal transplantation are; Hepatitis B and Hepatitis C

249.Of the following, the BEST choice for maintenance immunosuppression in renal transplantation is

A. cyclosporin
B. basiliximab
C. daclizumab
D. rituximab
E. belatacept

Ans A Central to many current pediatric immunosuppressive regimens is a calcineurin inhibitor (cyclosporine or tacrolimus) in combination with steroids and an adjunctive anti proliferative agent (azathioprine, sirolimus, or MMF), other for induction therapies.

Dialysis

250.Post-streptococcal glomerulonephritis results in

A. Nephrotic syndrome
B. Nephritic syndrome
C. Interstitial nephritis
D. Acute tubular necrosis

Ans B

251.Most common cause of hydronephrosis in children

A. PUJ obstruction
B. Posterior urethral valve

Ans A Practical Algorithms in Pediatric Nephrology edited by Israel Zelikovic, Israel Eisenstein p 49 Most common cause of hydronephrosis in children is PUJ obstruction; However, a neonate with a history of hydronephrosis on prenatal ultrasound and a palpable bladder most likely has posterior urethral valves.

252.In Schwartz formula for calculation of creatinine clearance in a child, the constant depends on the following except:

A. Age
B. Method of estimation of creatinine
C. Mass 146.
D. Severity of renal failure

Ans D Schwartz formula (for creatine clearance in child) is Creatinine clearance = K × height/creatinine where K = constant that depends upon age, muscle mass, method of creatinine estimation.

253.A 5-year-old male presented with dark color urine 1 to 2 days after an upper respiratory tract infection, GUE: RBC +++, albumin +; blood urea: 25 mg/dl; serum creatine: 0.6 mg/dl; with normal serum complement. Poor prognostic indicators at follow-up include all of the following EXCEPT

A. persistent hypertension
B. diminished renal function
C. significant proteinuria
D. histologically diffuse mesangial proliferation
E. recurrent macroscopic hematuria

Ans E is a good prognostic indicator.

254.All the following matching are true EXCEPT

A. nephrotic syndrome---Penicillin
B. nephrogenic diabetes inspidus ---cisplatin
C. nephrolithiasis ---furosemide
D. renal tubular acidosis---lithium
E. interstitial nephritis---cimetidine

Ans A Nephrotic syndrome--- penicillamine while interstitial nephritis--- penicillin

255.Cystinosis is a systemic disease caused by a defect in the metabolism of cysteine that results in accumulation of cystine crystals in most of the major organs of the body. All the following are recognized features EXCEPT

A. renal insufficiency
B. rickets
C. hypothyroidism
D. glaucoma
E. fever

Ans D Ocular presentations include photophobia, retinopathy, and impaired visual acuity. Patients also can develop hepatosplenomegaly, and delayed sexual maturation.

256.Potter syndrome (bilateral renal agenesis) is characterized by all the following

A. widely separated eyes
B. polyhydramnios
C. low set ears
D. broad nose
E. limb anomalies

Ans B Oligohydramnios.

257.Prompt surgical repair in ureteropelvic junction obstruction is indicated in infants with all the following EXCEPT

A. solitary kidney
B. abdominal mass
C. bilateral severe hydronephrosis
D. hematuria after minimal trauma
E. diminished function in the involved kidney

Ans D

258.All the following are contraindications to circumcision in neonates EXCEPT

A. hypospadias
B. chordee without hypospadias
C. dorsal hood deformity
D. small penis
E. penile torsion

Ans E In most cases, penile development is normal, and the condition is unrecognized until circumcision is performed or the foreskin is retractable

259.Nephrocalcinosis refers to calcium deposition within the renal tissue. All of the following can cause nephrocalcinosis EXCEPT

A. furosemide
B. distal RTA
C. hypoparathyroidism
D. hyperoxaluria
E. Cushing syndrome

Ans C Hyperparathyroidism.

20

CHAPTER 20

Pediatric Endocrinology



WHAT THIS CHAPTER COVERS

- Growth Hormone Disorders
- Thyroid Disorders
- Diabetes Mellitus
- Diabetic Ketoacidosis
- Adrenal Disorders
- Congenital Adrenal Hyperplasia
- Cushing Syndrome
- Precocious Puberty
- Delayed Puberty
- Disorders of Sexual Development
- Hypopituitarism
- Diabetes Insipidus
- Calcium & Parathyroid Disorders
- Rickets & Vitamin D
- Obesity & Metabolic Syndrome
- Ambiguous Genitalia

217 MCQs **16** subtopics

20.1 Growth Hormone Disorders & Hypopituitarism

- **GH**: pulsatile, mainly during **slow-wave sleep**; acts via **IGF-1 (liver)**; GH is **not important for fetal growth** (insulin, IGF-2, placental factors are) → **GH-deficient babies have normal birth length**.
- **Congenital GH deficiency/hypopituitarism**: **neonatal hypoglycaemia, prolonged jaundice (cholestasis), micropenis (with gonadotropin deficiency)**, midline defects (**septo-optic dysplasia – optic nerve hypoplasia + absent septum pellucidum + pituitary deficiency; HESX1**), single central incisor, cleft lip/palate; **growth failure apparent after 6–12 months**.
- **Clinical GHD**: **short stature with ↓ growth velocity, "chubby/cherubic" appearance (truncal adiposity), immature (doll-like) face, frontal bossing, high-pitched voice, delayed dentition, delayed bone age (BA = height age)**; normal proportions; **normal intelligence**.
- Acquired: **craniopharyngioma (MC)**, cranial irradiation, germinoma, LCH, head trauma, infections, hypophysitis.
- **Diagnosis**: low **IGF-1 & IGFBP-3** (screening), **two GH stimulation tests – clonidine, glucagon, arginine, insulin-induced hypoglycaemia (gold standard but risky), L-dopa; peak GH < 10 ng/mL (some use < 7) = deficiency**; sex-steroid priming in peripubertal; **MRI brain (pituitary)**.
- **Rx**: **recombinant GH (somatropin) 0.025–0.05 mg/kg/day SC at bedtime** until growth velocity < 2 cm/y or epiphyseal fusion; long-acting weekly GH (somapacitan). Side effects – **pseudotumor cerebri, SCFE, scoliosis progression**, hyperglycaemia, gynaecomastia.
- **Laron syndrome (GH insensitivity – GH receptor defect)**: **high GH, very low IGF-1**, severe short stature, protruding forehead; Rx **recombinant IGF-1 (mecasermin)**.
- **GH excess**: **pituitary gigantism (before epiphyseal fusion)** – McCune–Albright, MEN-1, Carney complex, AIP mutation; acromegaly after fusion.

20.2 Thyroid Disorders

Congenital hypothyroidism (CH)

- **MC preventable cause of intellectual disability**; incidence **~1 : 1000–1 : 2500 in India** (higher than West).
- **Causes**: **thyroid dysgenesis (MC – 85%: ectopic thyroid (MC – lingual/sublingual), aplasia/agenesis, hypoplasia) – sporadic, no goitre**; **dyshormonogenesis (10–15% – AR, goitre present; thyroid peroxidase (TPO) defect MC; Pendred syndrome – SLC26A4, SNHL + goitre)**; central (TSH/TRH deficiency); transient (maternal antithyroid drugs, **iodine deficiency/excess (povidone-iodine)**, TSH-receptor blocking antibodies, prematurity).
- **Clinical** (most are **normal at birth** – maternal T4): **prolonged physiological jaundice (earliest)**, **large anterior & posterior fontanelle**, feeding difficulty, constipation, **lethargy/somnolence, hoarse cry, macroglossia, umbilical hernia**, dry mottled skin, **hypothermia, bradycardia**, hypotonia, coarse facies, **myxoedema**, delayed milestones; **post-term, large birth weight** often.
- **Newborn screening**: **heel-prick TSH on day 2–5 (ideally 48–72 h)**; cord blood TSH (in early discharge; higher cut-off); **TSH > 20 mU/L (screen) → confirm with serum TSH & FT4**. **Diagnosis: ↑ TSH + ↓ FT4** (primary).
- **Investigations**: **bone age – absent distal femoral epiphysis at birth (normally present at 36 wk)**, **epiphyseal dysgenesis (stippled)**; **thyroid scan (99mTc/123I) or USG** – aetiology (do not delay treatment).
- **Rx: L-thyroxine 10–15 µg/kg/day orally as early as possible (within first 2 weeks)** – crushed tablet with water/breast milk (not with soy/iron/calcium); goal – normalise FT4 within 2 weeks, TSH within 4 weeks; keep **FT4 in upper half of normal**; monitor every 1–2 months in first 6 months. **IQ normal if treated before 2–4 weeks**; trial off therapy at **3 years** if transient suspected (not if ectopic/agenesis).

Acquired hypothyroidism

- **Hashimoto (autoimmune) thyroiditis – MCC of acquired hypothyroidism & goitre in children (non-endemic areas)**; adolescent girls; **anti-TPO, anti-thyroglobulin antibodies**; associated – **Down, Turner, T1DM, celiac**; firm, pebbly goitre.
- **Iodine deficiency** – MC cause globally (endemic goitre).

- Features: **growth failure (↓ height velocity – often first sign), delayed bone age (most delayed of all causes)**, weight gain, school performance may be maintained, constipation, cold intolerance, **delayed puberty (or precocious – Van Wyk–Grumbach syndrome: hypothyroidism + precocious puberty + multicystic ovaries; TSH acts on FSH receptor)**, **pseudotumor**, **SCFE**, galactorrhoea, muscle pseudohypertrophy (**Kocher–Debré–Sémélaigne**).
- Rx L-thyroxine 2–5 µg/kg/day (older children).

Hyperthyroidism

- **Graves disease – MCC in children** (adolescent girls); **TSH-receptor stimulating antibodies (TRAb)**; goitre, tachycardia, weight loss, tremor, **accelerated growth & bone age**, behavioural change (ADHD-like), exophthalmos (milder).
- **Neonatal thyrotoxicosis** – transplacental TRAb from Graves mother; irritability, tachycardia, goitre, craniosynostosis; transient (2–3 months); Rx methimazole + propranolol.
- **McCune–Albright** – autonomous; **hashitoxicosis**, toxic adenoma.
- **Rx**: **methimazole (carbimazole) – DOC in children** (**PTU avoided – hepatotoxicity**, except 1st trimester pregnancy/thyroid storm), β-blocker (propranolol) for symptoms; **radioactive iodine (> 10 y)** or **total thyroidectomy** for relapse.
- **Thyroid nodules** – higher malignancy risk in children (**papillary carcinoma MC**; RET in **MEN-2 → medullary – prophylactic thyroidectomy**).

20.3 Diabetes Mellitus

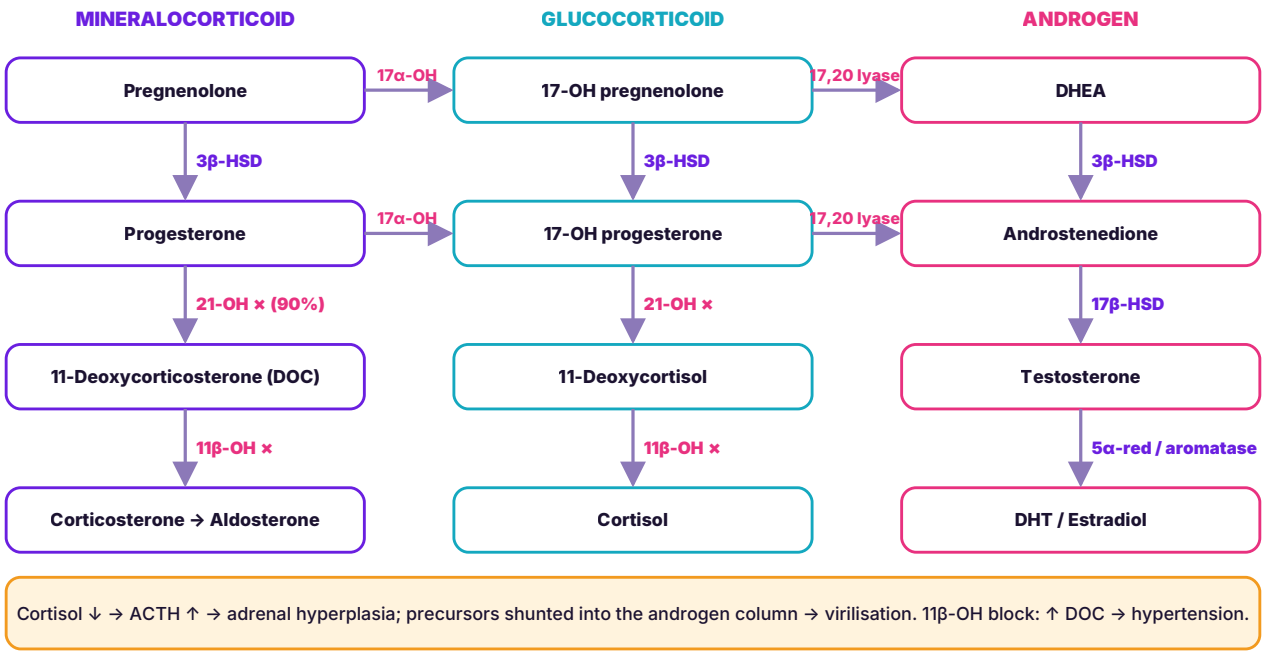
Feature	Type 1 DM	Type 2 DM	MODY
Frequency in children	**MC (> 90%)**	↑ (obesity), adolescents	1–4%
Mechanism	**Autoimmune β-cell destruction** (HLA-DR3/DR4, DQ8)	Insulin resistance	**Monogenic AD β-cell defect**
Age	Peaks **5–7 y & puberty**	Puberty	< 25 y, 3 generations
Presentation	**Polyuria, polydipsia, weight loss, DKA (30–40% at diagnosis)**, **secondary enuresis**	Obese, **acanthosis nigricans**, PCOS, mild symptoms	Mild hyperglycaemia, no ketosis
Antibodies	**GAD65, IA-2, insulin (IAA – earliest in young), ZnT8**	Negative	Negative
C-peptide	**Low**	Normal/high	Normal
Rx	**Insulin**	**Metformin** (+ insulin, GLP-1 RA – liraglutide)	**GCK-MODY (MODY 2): no Rx**; **HNF1A (MODY 3, MC)/HNF4A: sulfonylureas**

- **Diagnosis (ADA/ISPAD)**: **fasting glucose ≥ 126 mg/dL**, **2-h OGTT ≥ 200 mg/dL**, **random ≥ 200 with symptoms**, **HbA1c ≥ 6.5%**.
- **Neonatal diabetes** (< 6 months): **KCNJ11/ABCC8 (KATP channel) mutations → respond to oral sulfonylureas (glibenclamide)**; transient (6q24 imprinting) vs permanent; IPEX, Wolcott–Rallison.
- **Insulin regimens**: **basal-bolus (MDI) – long-acting (glargine/degludec/detemir) + rapid-acting (aspart/lispro/glulisine) with meals – preferred**; split-mixed (NPH + regular) twice daily (resource-limited); **insulin pumps (CSII), CGM, hybrid closed-loop**. Total daily dose ~**0.5–1 U/kg/day** (prepubertal ~0.7; puberty 1–1.2; honeymoon < 0.5).
- **Honeymoon (remission) phase**: weeks–months after starting insulin – ↓ requirement (< 0.5 U/kg/day).
- **Dawn phenomenon** (early-morning hyperglycaemia from GH/cortisol surge) vs **Somogyi effect** (nocturnal hypoglycaemia → rebound morning hyperglycaemia) – check 3 am glucose.
- **Targets (ISPAD 2022)**: **HbA1c < 7% (53 mmol/mol)**; time in range (70–180 mg/dL) > 70%; pre-meal 70–144 mg/dL.

- **Hypoglycaemia (< 70 mg/dL)** – MC acute complication: **15 g fast-acting glucose (0.3 g/kg)** orally → recheck in 15 min; severe/unconscious – **glucagon IM/SC (0.5 mg < 25 kg; 1 mg ≥ 25 kg) or intranasal 3 mg**, IV 10% dextrose 2 mL/kg.
- **Screening for complications (ISPAD)**: **from 11 y (or 2–5 y after diagnosis)** – **retinopathy, nephropathy (urine ACR annually)**, neuropathy; **thyroid (TSH) at diagnosis & every 2 y**, **celiac (tTG) at diagnosis & 2 & 5 y**, lipids, BP.
- **Mauriac syndrome** – poorly controlled T1DM: **growth failure, delayed puberty, hepatomegaly (glycogen), Cushingoid**.
- **Wolfram (DIDMOAD)** – **D**iabetes **I**nsipidus, **D**iabetes **M**ellitus, **O**ptic **A**trophy, **D**eafness (WFS1).
- **DKA** – see Fluids & Electrolytes.

20.4 Adrenal Disorders

Congenital adrenal hyperplasia (CAH)



Adrenal steroid pathway – the CAH enzyme blocks

Enzyme deficiency	Frequency	Cortisol	Aldosterone	Androgens	Features	Marker
21-hydroxylase (CYP21A2)	**> 90–95% (MC)**	↓	**↓ (salt-wasting – 75%)** / N (simple virilising)	**↑**	**Girls: ambiguous genitalia (virilised 46,XX – MCC of DSD in genetic females)**; **boys: normal genitalia → salt-wasting crisis at 1–3 weeks** (vomiting, dehydration, **hyponatraemia, hyperkalaemia, metabolic acidosis**, hypoglycaemia, shock); later – precocious pseudopuberty, tall child → short adult; non-classic – hirsutism, PCOS-like	**↑ 17-hydroxyprogesterone (17-OHP)** – screening/ diagnosis; ↑ renin, ↑ ACTH
11β-hydroxylase (CYP11B1)	5–8%	↓	↓ but **↑ DOC (11-deoxycorticosterone) – mineralocorticoid**	↑	**Virilisation + HYPERTENSION + hypokalaemia**	**↑ 11-deoxycortisol & DOC**
17α-hydroxylase (CYP17A1)	Rare	↓	↓ (but ↑ DOC, corticosterone)	**↓**	**Hypertension, hypokalaemia**; **46,XY – undervirilised (female phenotype)**; 46,XX – **primary amenorrhoea, no puberty**	↑ DOC, progesterone
3β-HSD type 2	Rare	↓	↓	↑ DHEA (weak)	**Salt wasting**; **ambiguous genitalia in BOTH sexes** (mild virilisation of XX, undervirilisation of XY)	↑ 17-OH pregnenolone, DHEA
StAR (congenital lipoid adrenal hyperplasia)	Rare	↓↓	↓↓	↓↓	Severe salt wasting; **all 46,XY appear female**; **lipid-laden enlarged adrenals**	All steroids low
P450 oxidoreductase	Rare				Antley–Bixler skeletal anomalies, ambiguity in both sexes	

IF THE DEFICIENT ENZYME STARTS WITH 1 → HYPERTENSION; ENDS WITH 1 → VIRILISATION

11β-OH: starts with 1 & ends with 1 → **hypertension + virilisation**. **17α-OH**: starts with 1 → **hypertension, no virilisation**. **21-OH**: ends with 1 → **virilisation, salt wasting (hypotension)**.

MANAGEMENT OF CAH

- **Salt-wasting crisis**: **IV NS bolus 20 mL/kg + 10% dextrose**, **IV hydrocortisone 50–100 mg/m² (or 25 mg infant) stat**, then 50–100 mg/m²/day divided; treat hyperkalaemia.
- Maintenance: **hydrocortisone 10–15 mg/m²/day in 3 divided doses** (preferred in children – least growth suppression); **fludrocortisone 0.05–0.2 mg/day + NaCl 1–2 g/day (infants)** for salt-wasters.
- **Stress dosing** (fever, surgery, illness): **double/triple hydrocortisone dose**; emergency card/injection.
- Monitor: **growth velocity, bone age, 17-OHP, androstenedione, renin**.
- **Feminising genitoplasty** (individualised; parents' informed choice); **female sex of rearing for 46,XX (fertile)**.
- **Prenatal dexamethasone** (to prevent virilisation – experimental/controversial).

Adrenal insufficiency & Cushing syndrome

- **Primary (Addison)**: autoimmune (APS-1/APECED, APS-2), **X-linked adrenoleukodystrophy (boys – check VLCFA)**, CAH, adrenal haemorrhage (**Waterhouse–Friderichsen – meningococcaemia**), TB, **adrenal hypoplasia congenita (DAX1/NR0B1, X-linked – with hypogonadotropic hypogonadism)**, familial glucocorticoid deficiency (ACTH resistance), **triple A (Allgrove)**; **hyperpigmentation**, salt craving, hyponatraemia, hyperkalaemia, hypoglycaemia; **↑ ACTH, ↓ cortisol; short Synacthen test (cortisol < 18 µg/dL at 30/60 min)**.
- **Secondary**: **abrupt withdrawal of long-term steroids (MCC of adrenal insufficiency)**, hypopituitarism – no hyperpigmentation, no hyperkalaemia.
- **Cushing syndrome**: **iatrogenic (exogenous steroids) – MCC**; endogenous – **adrenal tumour (MC in < 7 y – carcinoma/adenoma, often with virilisation)**, **Cushing disease (pituitary ACTH adenoma – MC endogenous cause > 7 y)**, McCune–Albright, PPNAD (Carney); **growth failure with weight gain (hallmark in children – "short & fat")**, moon face, buffalo hump, striae, HTN, osteoporosis. Screen – **24-h urinary free cortisol, late-night salivary cortisol, 1 mg overnight dexamethasone suppression**.
- **Pheochromocytoma** (children – bilateral/extra-adrenal more often; **VHL (MC), MEN-2, NF-1, SDH**): **sustained hypertension (vs paroxysmal in adults)**, sweating, headache, palpitations; **plasma free metanephrines (best)**; Rx **α-blockade (phenoxybenzamine) first, then β-blocker**, then surgery.

20.5 Disorders of Calcium & Bone Metabolism (endocrine)

- **Hypoparathyroidism**: DiGeorge (22q11), autoimmune (APECED), post-surgical, **hypomagnesaemia**, activating CaSR mutation; **↓ Ca, ↑ PO_4, ↓ PTH**.
- **Pseudohypoparathyroidism (PHP 1a – GNAS, maternal; Albright hereditary osteodystrophy)**: **short stature, round face, obesity, short 4th–5th metacarpals (knuckle-knuckle-dimple-dimple), subcutaneous calcifications, ID**; **↓ Ca, ↑ PO_4, ↑ PTH (resistance)**; multi-hormone resistance (TSH, GnRH); **pseudo-pseudohypoparathyroidism (paternal GNAS)** – AHO phenotype with normal biochemistry.
- **Hyperparathyroidism** – rare (MEN-1, MEN-2A); **neonatal severe hyperparathyroidism (homozygous CaSR loss)**.
- Rickets – see Nutrition chapter.

20.6 Puberty Disorders

Precocious puberty – girls < 8 y, boys < 9 y

	Central (gonadotropin-dependent, true)	Peripheral (gonadotropin-independent, pseudo)
Mechanism	**Premature activation of HPG axis**	Sex steroids from gonad/adrenal/exogenous
Sex	**Girls (MC – 90% idiopathic)**; **boys – CNS lesion likely (50–75%)**	Both
Causes	**Idiopathic (MC in girls)**; **hypothalamic hamartoma (MC CNS lesion – gelastic/laughing seizures)**, CNS tumours (glioma – NF-1, germinoma), hydrocephalus, cranial irradiation, trauma, MKRN3/KISS1 mutations, **secondary central PP after long-standing peripheral PP (e.g. treated CAH)**	**CAH (MC in boys)**, **McCune–Albright** (girls), **testotoxicosis (LH-receptor activating mutation – familial male-limited)**, **Leydig cell tumour**, **hCG-secreting tumours (hepatoblastoma, germinoma – boys)**, ovarian cysts/tumours (granulosa cell), adrenal tumours, exogenous steroids, **severe primary hypothyroidism (Van Wyk–Grumbach – TSH acts on FSH receptor; bone age delayed)**
Consonance	**Consonant (normal sequence)**	Often **non-consonant**
Testes (boys)	**Bilaterally enlarged (≥ 4 mL)**	**Small (prepubertal) in adrenal causes**; **unilateral enlargement – Leydig cell tumour**; bilateral – testotoxicosis/hCG
Basal/GnRH-stimulated LH	**↑ (LH > 0.3 IU/L basal; stimulated LH > 5 IU/L)**	**Suppressed**
Rx	**GnRH agonists (leuprolide depot, triptorelin) – DOC**; treat cause	Treat cause; **aromatase inhibitors (letrozole), anti-androgens (bicalutamide, spironolactone)**, ketoconazole

- **McCune–Albright syndrome**: **somatic GNAS activating mutation (mosaic)** – **triad: polyostotic fibrous dysplasia, café-au-lait spots with irregular "coast of Maine" borders (vs smooth "coast of California" in NF-1), peripheral precocious puberty (girls – autonomous ovarian cysts)**; also hyperthyroidism, GH excess, Cushing, hypophosphataemia (FGF-23).
- **Benign variants**: **premature thelarche** (isolated breast development 6 m–2 y, no growth acceleration, normal bone age, regresses) & **premature adrenarche** (isolated pubic/axillary hair, body odour, ↑ DHEAS; girls 6–8 y; obesity, SGA; later PCOS risk).

Delayed puberty – no breast development by 13 y (girls) / testes < 4 mL at 14 y (boys)

Type	LH/FSH	Causes
Constitutional delay (CDGP)	Low (prepubertal)	**MC cause (esp. boys)**; family history; delayed BA; spontaneous puberty
Hypogonadotropic hypogonadism	**Low**	**Kallmann syndrome (anosmia/hyposmia + HH; KAL1/ANOS1 X-linked – GnRH neuron migration defect; renal agenesis, synkinesia)**, isolated GnRH deficiency, **Prader–Willi, Laurence–Moon–Bardet–Biedl**, CNS tumours (**craniopharyngioma**), hypopituitarism, **chronic disease/malnutrition/anorexia nervosa/excessive exercise (functional)**, hyperprolactinaemia, hypothyroidism
Hypergonadotropic hypogonadism	**High**	**Turner (MC in girls)**, **Klinefelter (MC in boys)**, gonadal dysgenesis (Swyer 46,XY), chemo/radiation, galactosemia (ovarian failure), autoimmune oophoritis, anorchia, 17α-OH deficiency

- Rx: CDGP – reassurance or **low-dose testosterone (boys)/oestrogen (girls)** short course; hypogonadism – sex-steroid replacement; fertility – gonadotropins/pulsatile GnRH.

20.7 Disorders of Sex Development (DSD)

- **Ambiguous genitalia = medical & social emergency** – don't assign sex until evaluation; check for **salt-wasting CAH (electrolytes, glucose, 17-OHP)**.
- Features suggesting DSD: **bilateral non-palpable testes in apparent male**, **perineal hypospadias with bifid scrotum**, clitoromegaly, posterior labial fusion, **inguinal/labial mass in a girl (testis)**, discordance between genitalia & prenatal karyotype.
- **First investigations**: **karyotype (rapid FISH/QF-PCR for SRY)**, **pelvic USG (uterus?)**, **17-OHP, electrolytes**, testosterone, DHT, LH/FSH, AMH, hCG stimulation test, genitogram/cystoscopy.

Category	Examples
46,XX DSD (virilised female)	**CAH (21-OH – MCC of ambiguous genitalia overall)**, 11β-OH, maternal androgens (luteoma, progestins), placental aromatase deficiency, ovotesticular (SRY+)
46,XY DSD (undervirilised male)	**Androgen insensitivity syndrome (AIS – AR gene, X-linked)**: complete (CAIS – **female phenotype, blind vaginal pouch, no uterus, inguinal hernia with testes, primary amenorrhoea, normal breasts (aromatisation), scanty pubic hair**; high testosterone & LH); partial (PAIS); **5α-reductase type 2 deficiency (↑ T:DHT ratio > 20; ambiguous at birth → virilisation at puberty – "penis at 12" – Dominican guevedoces)**; testosterone biosynthesis defects (17β-HSD3, 3β-HSD, StAR, 17α-OH); **gonadal dysgenesis (Swyer – 46,XY complete: female phenotype with uterus, streak gonads – gonadoblastoma risk)**; Leydig cell hypoplasia (LH receptor); **persistent Müllerian duct syndrome (AMH/AMHR defect – normal male with uterus, hernia uteri inguinale)**; Denys–Drash/Frasier (WT1)
Sex chromosome DSD	**45,X/46,XY mixed gonadal dysgenesis** (streak + testis; asymmetric), Turner, Klinefelter, **ovotesticular DSD (46,XX/46,XY chimera; both ovarian & testicular tissue)**

- **Gonadoblastoma risk**: dysgenetic gonads with **Y-chromosome material** (Swyer, mixed gonadal dysgenesis) → **gonadectomy**; CAIS – gonadectomy after puberty (lower risk).
- **Micropenis**: stretched penile length **< 2.5 SD (< 2.5 cm in term newborn)** with normal structure – hypopituitarism, Kallmann, hypogonadism, PAIS; Rx short course of **testosterone (IM 25 mg monthly × 3)**.

20.8 Diabetes Insipidus & Other Pituitary-Hypothalamic Disorders

	Central DI	Nephrogenic DI	Primary polydipsia
Defect	↓ ADH secretion	Renal resistance to ADH	Excess water intake
Causes	**Idiopathic, craniopharyngioma, germinoma, LCH (MC infiltrative), trauma/surgery, septo-optic dysplasia, Wolfram**, meningitis	**X-linked AVPR2**, AQP2; lithium, hypercalcaemia, hypokalaemia, obstructive uropathy	Psychogenic
Serum Na / osm	**High–normal/high**	High	**Low–normal**
Water deprivation test	**Urine osmolality fails to rise (< 300)**	Fails to rise	**Rises (> 750)**
Desmopressin response	**Urine osm rises > 50%**	**No rise (< 10%)**	Already concentrated
Copeptin	Low	**High**	
Rx	**Desmopressin (intranasal/oral/SC)**	**Thiazides, low-salt diet, NSAIDs (indomethacin), amiloride (lithium)**	Fluid restriction

- **SIADH** – see Fluids chapter.
- **Hyperprolactinaemia** – prolactinoma (adolescents – galactorrhoea, amenorrhoea, delayed puberty); drugs (risperidone, metoclopramide); **cabergoline**.

- **Craniopharyngioma** – see Neurology/Tumours.

20.9 Childhood Obesity & Metabolic Syndrome (endocrine view)

- See Nutrition chapter; **endocrine obesity → short stature + delayed BA**; exogenous → tall + advanced BA.
- **Metabolic syndrome (IDF, ≥ 10 y)**: waist ≥ 90th centile + ≥ 2 of TG ≥ 150, HDL < 40, BP ≥ 130/85, FPG ≥ 100 mg/dL.
- **PCOS in adolescents** – see Adolescent chapter.

20.10 Multiple Endocrine Neoplasia & Autoimmune Polyendocrine Syndromes

Syndrome	Components
MEN-1 (MEN1 – menin)	**3 Ps – Parathyroid hyperplasia (MC), Pituitary adenoma, Pancreatic NET (gastrinoma, insulinoma)**
MEN-2A (RET)	**Medullary thyroid carcinoma (MTC), pheochromocytoma, parathyroid hyperplasia**
MEN-2B (RET M918T)	**MTC (earliest, most aggressive – thyroidectomy in 1st year), pheochromocytoma, mucosal neuromas, marfanoid habitus, intestinal ganglioneuromatosis**
APS-1 (APECED – AIRE, AR)	**Chronic mucocutaneous candidiasis (first), hypoparathyroidism, Addison disease** (childhood)
APS-2 (Schmidt)	**Addison + autoimmune thyroid disease ± T1DM** (adults)
IPEX (FOXP3)	Neonatal T1DM, enteropathy, eczema

Practice MCQs · Pediatric Endocrinology

217 questions · correct option highlighted in green · cover the answer strip to self-test

Growth Hormone Disorders

1. A 3-year male presents with recurrent episodes of hypoglycemia. He also has short stature & micropenis. He might be suffering from:

A. Hypothyroidism
B. **Hypopituitarism**
C. Adrenal insufficiency
D. Diabetes mellitus

Ans B

2. Acromegaly is due to:

A. Increased secretion of cortisol
B. Decreased secretion of cortisol
C. **Increased secretion of growth hormone**
D. Decreased secretion of growth hormone

Ans C In young persons with open epiphyses, overproduction of GH results in gigantism; in persons with closed epiphyses, the result is acromegaly.

3. Growth hormone deficiency is associated with all except:

A. Micropenis
B. Doll like facies
C. **IUGR**
D. Obesity THYROID DISORDERS

Ans C

4. In a 2–5 year-old child with DM, target HbA is: 1C

A. < 8%
B. **< 7.5%**
C. < 7%
D. < 6.5%

Ans B As per the latest ISPAD guidelines 2015, Target HbA1c <7.5% for children of all ages.

5. A 16-year-old woman athlete presents with complaints of primary amenorrhea. Her height is 153 cm, weight is 51 kg. She has well developed breasts. She has no pubic or axillary hair and no hirsutism. She has been given 117. 6 months of progesterone and estrogen but no benefit. Which of the following is the most probable diagnosis?

A. Bartter syndrome
B. Mutation of the calcium sensing receptor
C. Pseudo-pseudohypoparathyroidism
D. **Parathyroid adenoma**
E. McCune Albright syndrome The karyotype of a patient with androgen Insensitivity Syndrome is: (AI 05)

Ans D Complete androgen insensitivity syndrome In the given scenario, a phenotypic female with tall stature, primary amenorrhea has well-developed breasts and absent pubic hairs. All these point towards a diagnosis of Complete Androgen insensitivity syndrome.

6. Main causes of hypocalcemia in nephrotic syndrome in children are the following

A. **decreased activity of renal α-hydroxylase**
B. lowered serum albumin
C. decreased intestinal absorption of calcium
D. increased urinary losses of cholecalciferol-binding globulin
E. prednisone therapy

Ans A (which is involved in converting the less-active 25-hydroxyvitamin D into the more active 1,25-(OH)2 D) is a cause in renal insufficiency.

7. Physiologic factors play a role in stimulating and inhibiting growth hormone (GH). One of the following inhibit GH release

A. sleep
B. exercise
C. **hyperglycemia**
D. acute illness
E. fasting

Ans C Sleep, exercise, physical stress, trauma, acute illness, puberty, fasting, and hypoglycemia stimulate the release of GHwhereas hyperglycemia, hypothyroidism, and glucocorticoids inhibit GH release.

8. Patients with mineralocorticoid deficiency can develop the following EXCEPT

A. weight loss
B. hypotension
C. **metabolic alkalosis**
D. hyponatremia
E. hyperkalemia

Ans C Patients with mineralocorticoid excess can develop hypertension, hypokalemia, and metabolic alkalosis.

9. Significant gynecomastia is seen in

A. hyperthyroidism
B. **Klinefelter syndrome**
C. 17-ketosteroid reductase deficiency
D. 11β-hydroxylase deficiency
E. Peutz-Jeghers syndrome

Ans B Significant gynecomastia is seen in 50% of adolescents with Klinefelter syndrome.

10. Subjects with type 1 DM have evidence for celiac disease in

A. **5-10%**
B. 11-15%
C. 16-20%
D. 21-25%
E. 26-30%

Ans A Fifteen to 30% of subjects with T1DM have elevated thyroid-stimulating hormone (TSH) and antithyroid antibodies and close to 5-10% have evidence for celiac disease. These diseases share common genes and likely the same interplay between environmental and immunologic factors.

Thyroid Disorders

11. All are involved in MEN IIA except:

A. Pituitary
B. Parathyroid
C. Thyroid
D. Adrenal

Ans A In Multiple Endocrine Neoplasia (MEN) IIA, features seen are: "HAPPY", i.e. Hirschsprung disease, Amyloidosis, Pheochromocytoma, Parathyroid hyperplasia/Adenoma and thyroid Carcinoma (Medullary).

12. Neonatal hypothyroidism when is the best time for screening?

A. 2–4 hours after birth
B. 6–12 hours after birth
C. 1–2 days after birth
D. 2–4 days after birth

Ans D

13. Most common cause of congenital Hypothyroidism is?

A. Iodine deficiency
B. Thyroid agenesis
C. Thyroid dysgenesis
D. Thyroid dyshormonogenesis

Ans C Most cases of congenital hypothyroidism are not hereditary and result from thyroid dysgenesis.

14. A 2-year-old male child presents with congenital hypothyroidism with generalized muscular pseudo-hypertrophy. What is the diagnosis?

A. Lyonized Duchenne muscular dystrophy
B. Limb girdle dystrophy with congenital hypothyroidism
C. Kocher-Debre-Semelaigne syndrome
D. Isolated Pseudohypertrophy with congenital hypothyroidism

Ans C

15. The most common cause of Permanent Congenital Hypothyroidism is:

A. TSH Receptor Mutations
B. Thyroid Dysgenesis
C. Defective Iodide Transport
D. PIT – 1 Mutations

Ans B

16. All are true about Hypothyroidism except:

A. Delayed dentition
B. Widened fontanelle
C. Distended abdomen
D. All are true

Ans D If left untreated, a child with Hypothyroidism develops the following features:
• • Growth will be stunted, the extremities are short
• • Face: Depressed bridge of nose, swollen eyelids, thick, broad, protruding tongue
• • Delayed Dentition, hoarse voice; Broad hands with short fingers; Dry and scaly skin with little perspiration

17. Most common cause of congenital hypothyroidism:

A. Thyroid dysgenesis
B. Dyshormonogenesis
C. Antithyroid antibodies
D. Maternal Hypothyroidism

Ans A

18. A 1-year-old child is brought to the hospital with thyroid swelling. The child has abnormal weight gain and has poor activities. His TSH is found raised and T4 is very low. Which of the following is most likely cause of his disease?

A. Hypothalamic disease
B. Thyroid dysgenesis
C. Dyshormonogenesis
D. End organ receptor insensitivity

Ans C Most common cause of congenital hypothyroidism in a child with goiter is Thyroid dyshormonogenesis. a. Triangular facies with craniosyntosis •• Craniosynostosis refers to premature fusion of sutures and closure of fontanelle
• • But in congenital hypothyroidism, there is delayed closure of fontanelle.

19. The features of neonatal hypothyroidism include all except:

A. Growth retardation
B. Mental retardation within 2 years
C. Delayed puberty
D. Umbilical Hernia
E. Moist skin

Ans E Congenital/neonatal hypothyroidism causes dry, coarse skin (not moist skin, which is a feature of hyperthyroidism), along with growth and mental retardation if untreated, delayed puberty, and umbilical hernia.

20. Features of hypothyroidism in infancy include the following except:

A. Premature closure of posterior fontanelle
B. Coarse facies
C. Umbilical hernia
D. Constipation

Ans A

21. Clinical features of "hypothyroidism" in a newborn are all except:

A. Sluggishness +++
B. Large tongue
C. Large posterior fontanel
D. Mental retardation

Ans D Mental retardation (Intellectual disability) develops by 3-6 months of age (not in neonatal period).

22. Blood specimen for neonatal thyroid screening is obtained on:

A. Cord blood
B. 24 hours after birth
C. 48 hours after birth
D. **72 hours after birth**

Ans D This is a controversial question
• • Blood for neonatal thyroid screening is ideally obtained by heel prick between 2 and 5 days of life is placed on a filter paper card and sent to a central screening laboratory
• • Cord blood may also be used, especially in regions where the neonates may not be brought for follow-up after 48 hrs life; But the best answer, amongst the given options is 72 hours after birth.

23. Congenital hypothyroidism is characterized by all except:

A. Open posterior fontanel
B. Large tongue
C. Large abdomen
D. **Hyperthermia**

Ans D

24. Manifestations of endemic cretinism include:

A. Deafness and facial nerve involvement
B. Blindness and hypothyroidism
C. **Strabismus and spastic diplegias**
D. Multinodular goitre and mental retardation

Ans C The term endemic cretinism includes two different overlapping syndromes: Neurologic and Myxedematous. In Neurological Cretinism, there is goiter, but the child is euthyroid; Deaf mutism, squint and cerebral diplegia are usually present

25. Which of the following is true regarding cretinism?

A. Short limbs compared to trunk
B. Proportionate shortening
C. **Short limbs and short stature**
D. Short limbs and long stature

Ans C Hypothyroidism is a cause of disproportionate short stature with short limbs.

26. Which of the following is/are true in cretinism?

A. **Goiter present at birth**
B. Can be diagnosed by serum T4 levels
C. Prolonged physiological jaundice present
D. Common in iodine deficiency endemic areas
E. Delayed skeletal development ADRENAL DISORDERS

Ans A b. Can be diagnosed by serum T4 levels, c. Prolonged physiological jaundice present, d. Common in iodine-deficiency endemic areas, e. Delayed skeletal development

27. Which of the following is/are true about neonatal hypoparathyroidism?

A. **Familial isolated hypoparathyroidism is where a genetic mutation like SOX3 is the underlying cause**
B. Calcium sensing receptor (CaSR) mutation is another disorder of genetic origin
C. DiGeorge syndrome (22q11 deletion syndrome) is a condition which includes hypoparathyroidism at birth
D. Hypercalcemia is a salient feature
E. Seizures are not seen

Ans A b. Calcium sensing receptor (CaSR) mutation is another disorder of genetic origin, c. DiGeorge syndrome (22q11 deletion syndrome) is a condition which includes hypoparathyroidism at birth Discussing about the options one by one, a. True; Causes of Congenital hypoparathyroidism include:
• Familial isolated hypoparathyroidism: Autosomal recessive (GCMB, PTH), dominant (CaSR), X-linked (SOX3)
• DiGeorge syndrome (TBX1)
• Kenny-Caffey syndrome 1 (short stature, medullary stenosis) (TBCE)

28. Pseudohypoparathyroidism is characterized by:

A. Normal serum Ca++ and decreased serum PTH
B. Decreased serum Ca++ and decreased serum PTH
C. **Decreased serum Ca++ and Increased serum PTH**
D. Normal serum Ca++ and Increased serum PTH

Ans C Ghai 8/e Pseudohypoparathyroidism (PHP)
• • Parathyroid glands are normal or hyperplastic and they can synthesize and secrete parathyroid
• • Serum levels of immunoreactive PTH are elevated even when the patient is hypocalcemic normocalcemic
• • Neither endogenous nor administered PTH raises the serum levels of calcium or lowers the levels

29. Congenital goiter is seen in which percent of newborns with congenital hypothyroidism?

A. 10%
B. **20%**
C. 30%
D. 40%
E. 50%

Ans B Congenital goiter is seen in only 20% of newborns with congenital hypothyroidism.

30. During a routine physical examination, a solitary thyroid nodule is palpated in asymptomatic 10-year-old child. Of the following, the MOST likely cause is

A. **thyroid carcinoma**
B. thyroid adenoma
C. thyroid abscess
D. thyroid cyst
E. subacute thyroiditis

Ans A In children with a solitary nodule, about 30-40% has a carcinoma, 20-30% has an adenoma, and the remainder will have thyroid abscess, thyroid cyst, multinodular goiter, Hashimoto thyroiditis, subacute thyroiditis, or nonthyroidal neck mass.

31. The MOST accurate test of thyroid function is

A. T4
B. free T4
C. T3
D. thyroglobulin
E. TSH

Ans E Serum TSH levels are the most accurate test of thyroid function. Serum TSH levels are elevated in primary hypothyroidism and suppressed in hyperthyroidism.

32. The thyroid hormones are transported in plasma bound to thyroxine-binding globulin (TBG), a glycoprotein synthesized in the liver. TBG binds approximately 70% of T4 and 50% of T3. TBG level increase with administration of

A. estrogens
B. androgens
C. glucocorticoids
D. nicotinic acid
E. l-asparaginase

Ans A TBG levels increase in pregnancy, in the newborn period, with hepatitis, and with administration of estrogens (oral contraceptives), selective estrogen receptor modulators, heroin or methadone, mitotane, 5-fluorouracil, and perphenazine, and they decrease with androgens, anabolic steroids, glucocorticoids, nicotinic acid, and l- asparaginase.

33. The MOST common cause of permanent congenital hypothyroidism is

A. dyshormonogenesis
B. thyroid dysgenesis
C. Iodine deficiency
D. defect of iodide transport
E. Pendred syndrome

Ans B Some form of thyroid dysgenesis (aplasia, hypoplasia, or an ectopic gland) is the most common cause of permanent congenital hypothyroidism, accounting for 80-85% of cases.

34. Approximately 10% of infants with congenital hypothyroidism have associated congenital anomalies. Of the following, the MOST common are

A. nervous system anomalies
B. lungs anomalies
C. eye anomalies
D. cardiac anomalies
E. genitourinary anomalies

Ans D

35. The normal level of serum thyroid-stimulating hormone in first week of term Infants is up to

A. 4.4 mIU/L
B. 9.5 mIU/L
C. 13.6 mIU/L
D. 17.6 mIU/L
E. 27.0 mIU/L

Ans D Thyroid-stimulating hormone levels: premature Infants (28-36 wk); 1st wk of life 0.7-27.0 mIU/L; term Infants: birth to 4 days 1.0-17.6 mIU/L; 2-20 wk 0.6-5.6 mIU/L; 5 mo-20 yr 0.5-5.5 mIU/L.

36. Retardation of osseous development in congenitally hypothyroid infants can be shown radiographically at birth in approximately

A. 20%
B. 40%
C. 60%
D. 80%
E. 100%

Ans C Retardation of osseous development can be shown radiographically at birth in approximately 60% of congenitally hypothyroid infants and indicates some deprivation of thyroid hormone during intrauterine life. The distal femoral and proximal tibial epiphyses, normally present at birth, are often absent.

37. Levothyroxine (l-T4) given orally is the treatment of choice in congenital hypothyroidism. The recommended initial starting dose is

A. 2-5 µg/kg/day
B. 4-7 µg/kg/day
C. 5-9 µg/kg/day
D. 8-12 µg/kg/day
E. 10-15 µg/kg/day

Ans E The recommended initial starting dose is 10-15 µg/kg/day (totaling 37.5-50.0 µg/ day for most term infants). The starting dose can be tailored to the severity of hypothyroidism. Rapid normalization of thyroid function has been demonstrated to be important in achieving optimal neurodevelopmental outcome.

38. The MOST common cause of acquired hypothyroidism is

A. craniopharyngioma
B. Hashimoto thyroiditis
C. meningoencephalitis
D. drug-induced
E. irradiation

Ans B The most common cause of acquired hypothyroidism is chronic lymphocytic (Hashimoto) thyroiditis.

39. Antithyroid antibodies develop and subclinical or overt hypothyroidism occurs in the following conditions EXCEPT

A. Down syndrome
B. Turner syndrome
C. type 1 diabetes mellitus
D. Sj ̲gren syndrome
E. Williams syndrome

Ans E is associated with subclinical hypothyroidism; this does not appear to be autoimmune, as antithyroid antibodies are negative.

40. Risk for subclinical hypothyroidism is seen in children with

A. chronic hepatitis B infection
B. chronic hepatitis C infection
C. chronic hepatitis D infection
D. hepatitis A infection
E. hepatitis E infection

Ans B Children with chronic hepatitis C infectionare at risk for subclinical hypothyroidism; this does not appear to be autoimmune, because antithyroid antibodies are negative.

41. **The first clinical manifestation of acquired hypothyroidism is**

A. poor schoolwork
B. goiter
C. deceleration of growth
D. weight gain
E. constipation

Ans C is usually the first clinical manifestation, but this sign often goes unrecognized.

42. **One of the following is not routinely done in children with suspected hypothyroidism**

A. sonography
B. serum free T4
D. antithyroglobulin
E. antiperoxidase antibodies

Ans A is not indicated unless there is a suspicion of a thyroid nodule on neck palpation. In such cases, ultrasound examination is the most accurate study to confirm the presence of a nodule and determine if other, smaller nodules are present. In addition, an ultrasound examination can determine the nodule dimensions, texture (solid vs cystic nature), and presence or absence of other features that might influence a decision to undertake fine-needle aspiration, such as microcalcifications, blurred margins,...

43. **Chronic lymphocytic thyroiditis is the most common cause of acquired hypothyroidism, with or without goiter. All the following are true EXCEPT**

A. more common in girls
B. peak incidence during adolescence
C. most of the affected children are asymptomatic
D. clinical course is constant
E. familial clusters are common

Ans D The clinical course is variable. The goiter might become smaller or might disappear spontaneously, or it might persist unchanged for years while the patient remains euthyroid.

44. **Thyroid peroxidase antibodies are absent in the following thyroiditis syndrome**

A. Hashimoto thyroiditis
B. painless sporadic thyroiditis
C. painful subacute thyroiditis
D. acute suppurative thyroiditis
E. Riedel thyroiditis

Ans D

45. **Endemic cretinism is the most serious consequence of iodine deficiency; it includes two different but overlapping syndromes: a neurologic type and a myxedematous type. The neurologic syndrome is characterized by the following EXCEPT**

A. intellectual disability
B. deaf-mutism
C. disturbances in gait
D. patellar hyperreflexia
E. delayed pubertal development

Ans E Affected persons are goitrous but euthyroid, have normal pubertal development and adult stature, and have little or no impaired thyroid function.

46. **All the following conditions are associated with Graves' disease EXCEPT**

A. type 1 diabetes mellitus
B. vitiligo
C. iron deficiency anemia
D. alopecia areata
E. celiac disease

Ans C Pernicious anemia.

47. **A painless nodule in the thyroid or in the neck is the usual presentation of childhood thyroid cancer. Of the following, the MOST common site of distant metastasis is**

A. mediastinum
B. long bones
C. skull
D. brain
E. lungs

Ans E

48. **Aplasia or hypoplasia of the parathyroid glands is often associated with the DiGeorge/velocardiofacial syndrome. All the following regarding this syndrome are true**

A. occurs in 1 in 4,000 newborns
B. caused by a deletion of chromosome 22q11.2
C. hypocalcemia is transitory in the majority
D. hypocalcemia can have its onset later in life
E. conotruncal defects of the heart occur in 75% of cases

Ans E Associated abnormalities of the third and fourth pharyngeal pouches are common; these include conotruncal defects of the heart in 25%, velopharyngeal insufficiency in 32%, cleft palate in 9%, renal anomalies in 35%, and aplasia of the thymus with severe immunodeficiency in 1%.

49. **There is a spectrum of parathyroid deficiencies with clinical manifestations varying from no symptoms to those of complete and longstanding deficiency. Early manifestations include**

A. tingling of the hands and feet
B. muscular pain and cramps
C. carpopedal spasms
D. convulsions
E. delayed irregular teeth eruption

Ans B are early manifestations; they progress to numbness, stiffness, and tingling of the hands and feet.

50. **Laboratory findings in hypoparathyroidism include the following EXCEPT**

A. low serum calcium (5-7 mg/dL)
B. elevated serum phosphorus (7-12 mg/dL)
C. elevated serum alkaline phosphatase
D. level of 1,25(OH)2D3 is usually low
E. normal serum magnesium

Ans C The serum level of alkaline phosphatase is normal or low.

51. Type Ia accounts for the majority of patients with pseudohypoparathyroidism (PHP), they have the following skeletal abnormalities EXCEPT

A. brachydactyly
B. dimpling of the dorsum of the hand
C. **the three middle fingers are the same length**
D. bowing
E. thickening of the calvaria

Ans C The 2nd metacarpal is involved least often. As a result, the index finger occasionally is longer than the middle finger. Likewise, the 2nd metatarsal is only rarely affected.

52. Parathyroid crisis is manifested by the following EXCEPT

A. serum calcium levels >15 mg/dL
B. **progressive polyuria**
C. azotemia
D. stupor
E. coma

Ans B Progressive oliguria.

53. The MOST consistent and characteristic radiographic finding of primary hyperparathyroidism is

A. gross trabeculation of the skull
B. generalized rarefaction
C. cysts
D. **resorption of subperiosteal bone**
E. radiographic signs of rickets

Ans D The most consistent and characteristic radiographic finding is resorption of subperiosteal bone, best seen along the margins of the phalanges of the hands.

54. When diabetes and thyroid disease coexist, the possibility of autoimmune adrenal insufficiency should be considered. It may be heralded by the followings EXCEPT

A. **increasing insulin requirements**
B. increasing pigmentation of the skin
C. salt craving
D. asthenia
E. postural hypotension

Ans A Decreasing insulin requirements.

Diabetes Mellitus

55. A 9-year-old boy presents with growth retardation and propensity to hypoglycemia. Physical examination reveals short stature, micropenis, increased fat and high- pitched voice. The skeletal survey reveals bone age of 5 years. Which of the following is most appropriate diagnosis?

A. Malabsorption
B. **Growth hormone deficiency**
C. Adrenal tumor
D. Thyroxine deficiency

Ans B The given clinical features like growth retardation, short stature, hypoglycemia, micropenis, increased fat, high-pitched voice and bone age less than chronological age, all point towards a diagnosis of growth hormone deficiency.

56. Most common enzyme deficiency leading to childhood hypertension is:

A. 17-Alpha hydroxylase
B. 21-Beta Hydroxylase
C. **11-Beta Hydroxylase**
D. 3-Beta Hydroxy steroid dehydrogenase

Ans C All the above enzyme deficiencies lead to CAH. Out of these, hypertension is seen in 11-b hydroxylase deficiency deficiency. But 11-b hydroxylase deficiency is more common of the two.

57. A 3-month-old male child with normal genitalia presents to the emergency department with severe dehydration of hyperkalemia and hyponatremia. The measurement of blood levels of which of the following will be helpful?

A. **17-hydroxy progesterone**
B. Renin
C. Cortisol
D. Aldosterone

Ans A

58. All are features of type I Diabetes mellitus except:

A. Polyuria
B. Polydipsia
C. Polyphagia
D. **Diarrhea**

Ans D

59. Definition of Diabetes mellitus includes:

A. **Fasting blood glucose > 126 mg/dL**
B. Random blood glucose > 140 mg/dL
C. HbA1c < 7%
D. Post prandial blood glucose > 180 mg/dL

Ans A Fasting blood glucose >126 mg/dL Fasting blood glucose > 12.6 mg/dL or 2 hr post prandia or rangdom glucose 200 mg/dL or HbA1c >6.5% in presence of symptoms like polyuria and polydiapsia are diagnostic of diabetes mellitus.

60. Which of the following is a rapid acting insulin?

A. Glargine
B. Detemir
C. Lente
D. **Glulisine**

Ans D Ultrashort acting Insulins include: Insulin Lispro, Glulisine and Aspart.

61. Monogenic transmission of diabetes mellitus occurs in:

A. Insulin Dependent Diabetes Mellitus (IDDM)
B. Non-insulin Dependent Diabetes Mellitus (NIDDM)
C. Latent Autoimmune Diabetes in Adults (LADA)
D. **Maturity Onset Diabetes of the Young (MODY)**

Ans D MODY refers to forms of diabetes are associated with monogenic defects in pancreatic b-cell function.

62. Percentage of dose given as basal insulin in basal-bolus regimen in children is:

A. 0–25%
B. **25–50%**
C. 50–75%
D. None of these

Ans B Basal-bolus regimen of Insulin:
• • Basal insulin glargine should be 25–30% of the total dose in toddlers and 40-50% in older children
• • Remaining portion of the total daily dose is provided as bolus insulin that is directed by both the carbohydrate content of the meal as well as the preprandial glucose value
• • Glargine (basal insulin) may be given either as a single dose or every 12 hr in young children.

63. In children with type I Diabetes mellitus, when is ophthalmologic evaluation indicated:

A. At the time of diagnosis
B. After 1 year
C. **After 2 years**
D. After 5 years
E. After 10 years

Ans C and d. After 5 years •• In IDDM, initial retinal examination should be done 5 years after the onset of disease in prepubertal children and after 2 yrs in pubertal children; As the sight threatening retinopathy usually does not appear immediately after diagnosis
• • Thereafter 1–2 yearly examination is required.

64. In India the commonest cause of Juvenile Onset of Diabetes mellitus:

A. IDDM
B. **Fibrocalcific pancreaticopathy**
C. MODY
D. Gallstones DISORDERS OF PUBERTY 108.

Ans B "In tropical countries 50% of young onset diabetes is constituted by protein deficient diabetes mellitus; next in frequency is Fibrocalculus Pancreatic diabetes". DISORDERS OF PUBERTY Answers

65. Typical insulin dosage requirement in prepubertal children after the "honeymoon" period is

A. 0.4 U/kg/day
B. **0.5 U/kg/day**
C. 0.6 U/kg/day
D. 0.7 U/kg/day
E. 1 U/kg/day

Ans B Prepubertal children generally require about 0.5 U/kg/day. During the middle of puberty, dosages often exceed 1 U/kg/day, whereas postpubertal individuals require 0.75-1.0 U/kg/day. Athletes or those with a low caloric intake may require less insulin. For most children with type 1 diabetes, there are a variety of strategies and combinations of insulin that are currently used. There is a trend toward three or more doses of insulin each day. In addition, more children are on insulin pump therapy.

66. Falsely low HbA1c levels are noted in the following conditions EXCEPT

A. **aplastic anemia**
B. pure red cell aplasia
C. blood transfusions
D. cirrhosis
E. renal disease treated with erythropoietin

Ans A Falsely low HbA1c levels are noted in hemolytic anemias, pure red cell aplasia, blood transfusions, and anemias associated with hemorrhage, cirrhosis, myelodysplasias, or renal disease treated with erythropoietin.

67. Diabetic ketoacidosis (DKA) is the end result of the metabolic abnormalities resulting from a severe deficiency of insulin or insulin effectiveness. DKA is characterized by the following EXCEPT

A. ketonuria
B. **normal ion gap**
C. decreased serum bicarbonate
D. decreased pH
E. elevated effective serum osmolality

Ans B An increased ion gap.

68. To continue the insulin infusion in treaterment of DKA without causing hypoglycemia, glucose must be added to the infusion. We typically recommend that glucose be added as a 10% solution when the serum glucose has decreased

A. <100 mg/dL
B. <150 mg/dL
C. **<200 mg/dL**
D. <250 mg/dL
E. <300 mg/dL

Ans C To continue the insulin infusion without causing hypoglycemia, glucose must be added to the infusion. We typically recommend that glucose be added as a 5% solution when the serum glucose has decreased <300 mg/dL and as a 10% solution when the serum glucose has decreased <200 mg/dL. The insulin infusion can also be lowered from the initial maximal rate if, despite the above outlined interventions, the serum glucose falls further.

69. Any child with diabetic ketoacidosis (DKA) can be easily transitioned to oral intake and subcutaneous insulin when the following criteria are found EXCEPT

A. total CO2 >15 mEq/L
B. **pH >7.25**
C. sodium stable between 135 and 145 mEq/L
D. no emesis
E. no dehydration

Ans B Any child can be easily transitioned to oral intake and subcutaneous insulin when DKA has resolved (total CO2 >15 mEq/L; pH >7.30; sodium stable between 135 and 145 mEq/L; no emesis). The first dose of short acting subcutaneous insulin is given with a meal, and the insulin drip is discontinued approximately 30 min later.

70. **HbA1c values may be spuriously elevated in**

A. **thalassemia**
B. sickle cell disease
C. iron deficiency anemia
D. leukemia
E. aplastic anemia

Ans A Depending on the method used for determination, HbA1c values may be spuriously elevated in thalassemia (or other conditions with elevated hemoglobin F) and spuriously lower in sickle cell disease (or other conditions with high red blood cell turnover).

71. **Signs of insulin resistance or conditions associated with insulin resistance in T2 DM in children include the following EXCEPT**

A. acanthosis nigricans
B. hypertension
C. dyslipidemia
D. **increased waist : hip ratio**
E. polycystic ovary syndrome

Ans D

72. **Therapy with one of the following drugs usually results in significant insulin resistance leading to glucose intolerance and overt diabetes**

A. streptozotocin
B. rodenticide Vacor
C. **high-dose oral steroid**
D. cyclosporin
E. tacrolimus

Ans C High-dose oral or parenteral steroid therapy usually results in significant insulin resistance leading to glucose intolerance and overt diabetes. The immunosuppressive agents cyclosporin and tacrolimus are toxic to β cells, causing IDDM in a significant proportion of patients treated with these agents. Their toxicity to pancreatic β cells was 1 of the factors that limited their usefulness in arresting ongoing autoimmune destruction of β cells.

Diabetic Ketoacidosis

73. **Thyroxine secretion begins in fetus at how many weeks of gestation?**

A. 6 weeks
B. **12 weeks**
C. 20 weeks
D. 28 weeks

Ans B

74. **Pseudohermaphroditism in a female child is most commonly due to:**

A. **21-hydroxylase deficiency**
B. 17-hydroxylase deficiency
C. 11-hydroxylase deficiency
D. 3-hydroxylase deficiency

Ans A & Classic 21-hydroxylase deficiency is the most common cause of ambiguous genitalia in a female usually apparent at birth in females, i.e. Pseudohermaphroditism of a female child.

75. **All of the following statements about androgen insensitivity syndrome are true except:**

A. It is an X linked disorder
B. XY genotype is present
C. Affected individuals have female phenotype
D. **Abundant pubic hairs are present**

Ans D Sexual hairs are absent in Androgen insensitivity syndrome.

76. **The following is true about Nesidioblastosis except:**

A. Presents with hypoglycemic attacks
B. **More common in adults than in children**
C. Histopathology shows hyperplasia of islet cells
D. Diazoxide is used for treatment

Ans B Nesidioblastosis or congenital hyperinsulinism is the most common cause of hyperinsulinemic hypoglycemia in neonates and infancy; Drugs used for treatment include: IV Glucose; Glucagon, Diazoxide, Octreotide and Nifedipine.

77. **True statement about testicular feminising syndrome; are all except:**

A. Absent uterus
B. **Absent vagina**
C. Chromosome pattern 46 XY
D. Absent ovary

Ans B Blind vagina is present in testicular feminizing syndrome (androgen insensitivity syndrome) and genotype is 46, XY with presence of testis and absence of ovary & uterus.

78. **The following criteria are needed for the diagnosis of syndrome of inappropriate secretion of antidiuretic hormone (SIADH) EXCEPT**

A. excessive urinary sodium concentration
B. **hyponatremia with normal serum osmolality**
C. elevated urine osmolality
D. normal renal, adrenal, and thyroid function
E. absence of volume depletion

Ans B Hyponatremia with reduced serum osmolality.

79. **The following are main features of diabetic ketoacidosis (DKA) EXCEPT**

A. hyperglycemia (glucose usually >300 mg/dL)
B. ketonemia (serum ketones >3 mmol/L)
C. ketonuria
D. **venous pH <7.20**
E. serum HCO3<15 mEq/L

Ans D Venous pH <7.30.

80. **What percent of newly diagnosed diabetics present with symptoms of diabetic ketoacidosis (DKA)?**

A. 10%
B. 20%
C. **30%**
D. 40%
E. 50%

Ans C The percentage is higher in younger children (<5 years old).

81. All prepubertal gynecomastia cases, as well as pubertal cases with suspicious features, should be investigated. Of the following, the initial laboratory evaluation should include

A. karyotype
B. dehydroepiandrosterone sulfate
C. liver function tests
D. **thyroid function tests**
E. renal function tests

Ans D No laboratory evaluation is indicated in routine cases with no other associated abnormality but all prepubertal cases, as well as pubertal cases with suspicious features, should be investigated; initial laboratory evaluation should include thyroid function tests (to rule out hyperthyroidism), testosterone, estradiol, human chorionic gonadotropin, luteinizing hormone, and prolactin levels.

82. Diabetic ketoacidosis (DKA) may be arbitrarily classified as mild, moderate, or severe. Venous pH in severe DKA is

A. <7.05
B. <7.10
C. **<7.15**
D. <7.20
E. <7.25

Ans C

83. Cerebral edema complicating DKA remains the major cause of morbidity and mortality in children and adolescents with T1 DM. All the following are true EXCEPT

A. early bolus administration of insulin is a risk factor
B. high volumes of fluid is a risk factor
C. the incidence of cerebral edema in children with DKA has not changed over the past 15-20 yr
D. **radiographic imaging is frequently helpful in making the diagnosis of cerebral edema**
E. its etiology remains unknown

Ans D Radiographic imaging is frequently unhelpful in making the diagnosis of cerebral edema.

Adrenal Disorders

84. Clinical manifestations of hoarse cry, umbilical hernia, hypotonia, mottling of skin, lethargy, prolonged jaundice is seen in:

A. Gaucher's disease
B. Mucopolysaccharidosis
C. Growth hormone deficiency
D. **Congenital hypothyroidism**

Ans D The given clinical features, i.e. hoarse cry, umbilical hernia, hypotonia, mottling of skin, lethargy, prolonged jaundice suggest a diagnosis of congenital hypothyroidism

85. Adrenal cortex of fetus mainly releases which hormone during early part of pregnancy?

A. **Cortisol**
B. Corticosterone
C. Progesterone
D. DHEAS

Ans A Shortly afer the fetal adrenal gland forms (wk 8-10), it efficiently secretes cortisol, which is able to negatively feedback on fetal pituitary and hypothalamus to suppress ACTH secretion. After wk 12, 3b hydroxy steroid dehydrogenase activity in the fetal adrenal gland decreases and steroid sulfokinase activity increases. Thus, the major steroid products of the midgestation fetal adrenal gland are DHEA and DHEA sulfate (DHEAS).

86. Congenital adrenal hyperplasia, most commonly presents as:

A. Male pseudohermaphroditism
B. **Female pseudohermaphroditism**
C. True hermaphroditism
D. 46,XY intersex

Ans B Note: Male pseudohermaphrodite is now called 46,XY DSD, while female → 46 XX DSD; So female pseudohermaphrodite is genotypically female (46,XX), whose external genitalia looks like that of a male; In CAH due to 21-hydroxylase deficiency, females have ambiguous genitalia, while males have normal genitalia

87. MC enzyme defect seen in a patient of congenital adrenal hyperplasia is:

A. **21-hydroxylase**
B. 11-hydroxylase
C. 3-hydroxylase
D. 17-hydroxylase

Ans A More than 90% of CAH cases are caused by 21-hydroxylase deficiency.

88. Congenital adrenal hyperplasia with hypotension due to deficiency of enzymes:

A. Aromatase
B. 5 a reductase
C. 17 b hydroxylase
D. **21 hydroxylase**
E. 11 hydroxylase

Ans D 21 hydroxylase Answers

89. Congenital adrenal hyperplasia with hypertension is due to deficiency of:

A. 21-hydroxylase
B. 3-b HSD
C. **17-a hydroxylase**
D. All of the above

Ans C

90. In congenital adrenal hyperplasia, deficient enzyme is most commonly:

A. 11b hydroxylase deficiency
B. **21a hydroxylase deficiency**
C. 3 hydroxylase deficiency
D. 17 hydroxylase deficiency

Ans B

91. What percentage of pheochromocytomas are malignant?
A. 5
B. **10**
C. 20
D. 15

Ans B 10 M neMonic 'Rule of ten' for Pheochromocytoma: ("BG MEN"): 10% of Pheochromocytoma are:
• • Bilateral •• Extra-adrenal: paragangliomas
• • Malignant •• Normotensive (No hypertension) Note: 10% of sporadic Pheochromocytoma are bilateral; but 50% of familial tumors are bilateral.

92. In Pheochromocytoma, which of the following is increased in urine?
A. **VMA**
B. Aldosterone
C. Cortisol
D. 17 hydroxyprogesterone

Ans A Biochemical tests for Pheochromocytoma:
• • Pheochromocytomas synthesize and store catecholamine, so elevated plasma and urinary levels of catecholamines methylated metabolites (VMA or vanillyl mandelic acid), metanephrines, are the cornerstone for the diagnosis
• • Among these tests, the fractionated metanephrines and catecholamines are the most sensitive.Q
• • In contrast to adults with pheochromocytoma in whom both norepinephrine and epinephrine are elevated, children with

93. False about Addison's disease is:
A. Hyperpigmentation
B. Hypoglycemia
C. Hypotension
D. **Hypertension 468 Section 3: Systemic Pediatrics**

Ans D Hypertension Hypotension is seen in Addison's disease due to glucocorticoid deficiency and not hypertension.

94. Pheochromocytoma is the disease of:
A. Pituitary gland
B. **Adrenal medulla**
C. Pancreas
D. Adrenal cortex

Ans B Pheochromocytoma is a neoplasms of chromaffin cells in medulla that release catecholamines.

95. Which of the following effects is NOT SEEN in primary hyperaldosteronism (Conn's syndrome)?
A. Hypertension
B. Metabolic Alkalosis
C. **Hyperkalemia**
D. Expansion of Extracellular and Plasma volume

Ans C Primary aldosteronism encompasses disorders caused by excessive aldosterone secretion independent of the reninangiotensin system. These disorders are characterized by hypertension and hypokalemia.

96. All are features of Autoimmune Addison Disease except:
A. Hypoglycemia
B. Ketosis
C. Hyponatremia
D. **Hypokalemia**

Ans D Hyperkalemia is a feature of adrenal insufficiency due to mineralocorticoid insufficiency and not hypokalemia.

97. Addison's disease is characterized by all, except:
A. **Hyperglycemia**
B. Hypotension
C. Hyperkalemia
D. Hyponatremia

Ans A Hypoglycemia is a feature of glucocorticoid deficiency in Addison's disease.

98. Accepted screening test for Hyperaldosteronism is:
A. Measurement of sodium levels
B. Measurement of aldosterone levels
C. **Calculation of aldosterone renin ratio**
D. Measurement of potassium levels

Ans C The ratio of plasma aldosterone concentration to renin activity is always high, and this represents a cost-effective screening test for primary aldosteronism (hyperaldosteronism).

99. Congenital Adrenal Hyperplasia is due to deficiency of:
A. **21 beta hydroxylase**
B. 17 alpha hydroxylase
C. 5 alpha reductase
D. 17 beta reductase

Ans A Most common cause of Congenital Adrenal Hyperplasia is 21 beta hydroxylase deficiency. a. Adrenalectomy Adrenalectomy, in a case of pituitary adenoma, may lead to increased ACTH secretion by an unresected evidenced mainly by marked hyperpigmentation; this condition is termed Nelson syndrome.

100. A 2-month-old infant is presented with failure to thrive, recurrent emesis, hepatosplenomegaly, and adrenal insufficiency. Adrenal calcification is noted radiologically. The most likely diagnosis is:
A. Adrenal hemorrhage
B. **Wolman's disease**
C. Pheochromocytoma
D. Addison's disease

Ans B Wolman's disease What is it? A rare lipid disorder caused by a deficiency of lysosomal acid lipase Clinical features Presents in 1st few wks of life with failure to thrive, vomiting, abdominal distention, steatorrhea hepatosplenomegaly Important finding Extensive bilateral calcifications of adrenal glands; Hyperlipidemia

101. Most common type of congenital adrenal hyperplasia Questions (CAH):
A. **21-hydroxylase deficiency**
B. 11-beta hydroxylase deficiency
C. 3-hydroxylase deficiency
D. 17-alpha hydroxylase deficiency

Ans A

102. Adrenal hyperplasia due to 21 hydroxylase deficiency is treated with low dose:
A. Androgen
B. Estrogen
C. **Cortisone**
D. Anti-androgen

Ans C Oral hydrocortisone with or without mineralocorticoid supplementation is the treatment of choice for CAH due to 21 hydroxylase deficiency.

103. **A female has previous child with congenital adrenal hyperplasia. In the present pregnancy, steroid therapy should be started:**

A. After karyotyping and determination of sex of the baby
B. At the time of delivery
C. Before conception
D. **As soon as pregnancy is diagnosed**

Ans D

104. **The MOST common cause of primary adrenal insufficiency in infancy is**

A. **congenital adrenal hyperplasia**
B. isolated glucocorticoid deficiency
C. X-linked adrenal hypoplasia congenita
D. Lemli-opitz syndrome
E. adrenoleukodystrophy

Ans A The most common causes of adrenocortical insufficiency in infancy are the salt- losing forms of congenital adrenal hyperplasia. Approximately 75% of infants with 21- hydroxylase deficiency, almost all infants with lipoid adrenal hyperplasia, and most infants with a deficiency of 3β-hydroxysteroid dehydrogenase manifest saltlosing symptoms in the newborn period because they are unable to synthesize either cortisol or aldosterone.

105. **The clinical presentation of adrenal insufficiency depends on the age of the patient, the usual presentation of adrenal insufficiency in infancy is**

A. ketosis
B. hyperpigmentation
C. orthostatic hypotension
D. **hypoglycemia**
E. hypernatremia

Ans D Hyperkalemia, hyponatremia, and hypoglycemia are prominent presenting signs of adrenal insufficiency in infants.

106. **The MOST definitive test for adrenal insufficiency is measurement of**

A. blood sugar
B. **cortisol before and after administration of ACTH**
C. serum sodium
D. arterial blood gases
E. urinary excretion of sodium and chloride

Ans B The most definitive test for adrenal insufficiency is measurement of serum levels of cortisol before and after administration of ACTH; resting levels are low and do not increase normally after administration of ACTH.

107. **Patients with autoimmune Addison disease must be closely observed for the development of other autoimmune disorders. Of the following, the MOST commonly associated disorder in children is**

A. alopecia
B. vitiligo
C. chronic active hepatitis
D. type 1 diabetes mellitus
E. **hypoparathyroidism**

Ans E Patients with autoimmune Addison disease must be closely observed for the development of other autoimmune disorders. In children, hypoparathyroidism is the most commonly associated disorder, and it is suspected if hypocalcemia and elevated phosphate levels are present.

108. **The signs and symptoms of congenital adrenal hyperplasia (salt wasting form of the disease) caused by 21-hydroxylase deficiency typically first develop in affected infants at approximately**

A. 1 week of age
B. **2 weeks of age**
C. 3 weeks of age
D. 4 weeks of age
E. 6 weeks of age

Ans B Progressive weight loss, anorexia, vomiting, dehydration, weakness, hypotension, hypoglycemia, hyponatremia, and hyperkalemia. Typically first develop in affected infants at approximately 10-14 days of age. Without treatment, shock, cardiac arrhythmias, and death may occur within days or weeks.

109. **Significantly virilized females with congenital adrenal hyperplasia (CAH) usually undergo surgery between**

A. **2- 6 mo of age**
B. 7-12 mo of age
C. 13-24 mo of age
D. 5-9 year of age
E. 10-14 year of age

Ans A Significantly virilized females usually undergo surgery between 2-6 mo of age. If there is severe clitoromegaly, the clitoris is reduced in size, with partial excision of the corporal bodies and preservation of the neurovascular bundle; however, moderate clitoromegaly may become much less noticeable even without surgery as the patient grows. Vaginoplasty and correction of the urogenital sinus usually are performed at the time of clitoral surgery; revision in adolescence is often necessary.

110. **Cortisol deficiency in congenital adrenal hyperplasia (salt wasting form of the disease) is treated with hydrocortisone**

A. 5-10 mg/m2/24 hr
B. 10-15 mg/m2/24 hr
C. **15-20 mg/m2/24 hr**
D. 20-25 mg/m2/24 hr
E. 25-30 mg/m2/24 hr

Ans C Cortisol deficiency is treated with glucocorticoids. This often requires larger glucocorticoid doses than are needed in other forms of adrenal insufficiency, typically 15-20 mg/m2/24 hr of hydrocortisone daily administered orally in 3 divided doses. Affected infants usually require dosing at the high end of this range. Double or triple doses are indicated during periods of stress, such as infection or surgery.

111. **Clinical manifestations of pheochromocytomas include the following EXCEPT**

A. hypertension
B. convulsions
C. pulmonary edema
D. **obesity**
E. good appetite

Ans D Patients have a good appetite but because of the hypermetabolic state may not gain weight, and severe cachexia may develop.

112. Calcification within the adrenal glands may occur in a wide variety of situations, some serious and others of no obvious consequence. Adrenal calcifications are bilateral in

A. neuroblastomas
B. ganglioneuromas
C. **Wolman disease**
D. cortical carcinomas
E. pheochromocytomas

Ans C Infants with Wolman disease, a rare lipid disorder caused by a deficiency of lysosomal acid lipase, have extensive bilateral calcifications of the adrenal glands.

Congenital Adrenal Hyperplasia

113. Newborn with prolonged jaundice and umbilical hernia—the likely diagnosis is:

A. **Congenital hypothyroidism**
B. Down's syndrome
C. Turner's syndrome
D. Noonan syndrome

Ans A

114. Drug used in the treatment of CAH in a child is:

A. Dexamethasone
B. Betamethasone
C. Prednisolone
D. **Hydrocortisone**

Ans D Treatment of CAH (21 hydroxylase deficiency) involves use of Hydrocortisone 15-20 mg/m2/24 hr, administered orally in 3 divided doses. Double or triple doses are indicated during periods of stress, such as infection or surgery.

115. Which of the following statements about 21 alpha hydroxylase deficiency is false?

A. Most common cause of congenital adrenal hyperplasia in children
B. Affected females present with ambiguous genitalia
C. Affected males present with precocious puberty
D. **Hypokalemic alkalosis is seen**

Ans D In congenital adrenal hyperplasia due to 21 alpha hydroxylase deficiency, there is hyperkalemia (not hypokalemia).

116. Oral glucose tolerance test in children is done with:

A. 1.5 gm/kg glucose
B. **1.75 gm/kg glucose**
C. 2 gm/kg glucose
D. 2.5 gm/kg glucose
E. 75 gm as an adult

Ans B Dose of glucose used in Oral Glucose Tolerance Test (OGTT) in adults → 75 gm and in children → 1.75 gm/kg of ideal weight, up to a maximum of 75 gm.

117. A baby girl presents with bilateral inguinal masses, thought to be hernias but are found to be testes in the inguinal canals. Which karyotype would you expect to find in the child?

A. 46,XX
B. **46,XY**
C. 46,XXY
D. 47,XXY

Ans B This baby girl with bilateral inguinal masses (testis) is probably suffering from Androgen insensitivity syndrome, where karyotype is 46,XY.

118. The following features suggest constitutional delay as a cause of short stature

A. no signs or symptoms of systemic disease
B. **bone age delayed beyond the height age**
C. period of poorest growth often occurring between the ages of 18 and 30 months
D. parental or sibling history of delayed development
E. height predictions consistent with family characteristics

Ans B Bone age delayed up to 2-4 years but consistent with height age

119. Hyponatremia in cerebral salt wasting is accompanied by the following EXCEPT

A. elevated urinary sodium excretion
B. **low urine output**
C. hypovolemia
D. normal or high uric acid
E. suppressed vasopressin

Ans B Cerebral salt wasting appears to be the result of hypersecretion of atrial natriuretic peptide and is seen primarily with central nervous system disorders including brain tumors, head trauma, hydrocephalus, neurosurgery, cerebrovascular accidents, and brain death. Hyponatremia is accompanied by elevated urinary sodium excretion (often >150 mEq/L), excessive urine output, hypovolemia, normal or high uric acid, suppressed vasopressin, and elevated atrial natriuretic peptide concentrations (>20 pmol/L).

120. High-dose glucocorticoids (the equivalent of >10 times physiologic cortisol secretion) can be administered without requiring a subsequent tapering for

A. 5 day
B. **7 day**
C. 10 day
D. 14 day
E. 21 day

Ans B

121. Many patients with Turner syndrome are recognizable at birth because of

A. **loose skin folds at the nape of the neck**
B. cubitus valgus
C. webbing of the neck
D. widely spaced nipples
E. hyperconvex fingernails

Ans A Many patients with Turner syndrome are recognizable at birth because of a characteristic edema of the dorsa of the hands and feet and loose skinfolds at the nape of the neck. Low birthweight and decreased birth length are common.

122.Nonketotic hyperosmolar coma is characterized by the following EXCEPT

A. severe hyperglycemia
B. nonketotic acidosis
C. severe dehydration
D. **hypothermia**
E. positive babinski signs

Ans D This syndrome is characterized by severe hyperglycemia (blood glucose >800 mg/dL), absence of or only slight ketosis, nonketotic acidosis, severe dehydration, depressed sensorium or frank coma, and various neurologic signs that may include grand mal seizures, hyperthermia, hemiparesis, and positive Babinski signs.

Cushing Syndrome

123.True about pendred syndrome:

A. **Diffuse colloid goiter**
B. Nodular goiter
C. Mental retardation
D. B/L sensory neural deafness
E. Normal cochlea

Ans A d. B/L sensory neural deafness Pendred Syndrome is an autosomal recessive disorder caused by mutations in the PDS gene on chr 7, which codes for pendrin (SLC26A4), a transporter protein found in the cochlea, thyroid and kidney; Congenital b/l SNHL and goitre (after puberty) with hypothyroidism seen.

124.The most common cause of Cushing's syndrome is:

A. Pituitary adenoma
B. Adrenal adenoma
C. Ectopic ACTH
D. **Iatrogenic steroids**

Ans D The most common cause of Cushing's syndrome is prolonged exogenous administration of glucocorticoid hormones, especially at the high doses used to treat lymphoproliferative disorders.

125.About Cushing syndrome all are true except:

A. Purple striae
B. Plethora
C. **Hypoglycemia**
D. Obesity

Ans C Hyperglycemia is a feature of Cushing's syndrome and not hypoglycemia.

126.Which one of the following is the earliest manifestation of Cushing's syndrome:

A. **Thyroid adenomas**
B. Thyroid carcinomas
C. Parathyroid adenomas
D. Pituitary adenomas

Ans A Loss of diurnal variation In patients with Cushing's syndrome loss of circadian rhythm of corticosteroids is the earliest finding. d. Pituitary adenomas Hyperpituitarism causes tall stature (Giants), with weak muscles; Most common cause of Hyperpituitarism is Pituitary adenoma.

127. True of testicular feminization syndrome is:

A. **Testes are present**
B. Female habitus
C. XY genotype
D. Secondary amenorrhea
E. Uterus present OBESITY

Ans A b. Female habitus, c. XY genotype OBESITY

128.Corticotropin-releasing hormone (CRH) and ACTH release are inhibited by

A. arginine vasopressin
B. oxytocin
C. angiotensin II
D. **atrial natriuretic peptide**
E. cholecystokinin

Ans D Arginine vasopressin, oxytocin, angiotensin II, and cholecystokinin stimulate release of CRH and ACTH, whereas atrial natriuretic peptide and opioids inhibit release of CRH and ACTH.

129.The cord serum T4 is decreased in proportion to gestational age and birthweight. The serum T4 gradually increases and enters the T4 range seen in term infants by the age of

A. 2 wk
B. 4 wk
C. **6 wk**
D. 8 wk
E. 10 wk

Ans C The cord serum T4 is decreased in proportion to gestational age and birthweight. The postnatal TSH surge is reduced, and the more premature, verylow- birthweight infants with complications of prematurity, such as respiratory distress syndrome, actually experience a decrease in serum T4 in the 1st wk of life. As these complications resolve, the serum T4 gradually increases so that generally by 6 wk of life it enters the T4 range seen in term infants.

130.Classic 21-hydroxylase deficiency occurs in approximately 1 in 15,000- 20,000 births in most populations. Of affected infants, the salt-losing form constitute about

A. 40%
B. 50%
C. 60%
D. **70%**
E. 80%

Ans D Approximately 70% of affected infants have the salt-losing form, whereas 30% have the simple virilizing form of the disorder.

131. In older children with Cushing syndrome, in addition to obesity, a common early manifestation is

A. purplish striae on abdomen
B. **short stature**
C. hypertension
D. hyperglycemia
E. osteoporosis

Ans B In older children, in addition to obesity, short stature is a common presenting feature. Gradual onset of obesity and deceleration or cessation of growth may be the only early manifestations.

132.All the following tests are of diagnostic utility in the diagnosis of Cushing syndrome

A. midnight cortisol levels
B. nighttime salivary cortisol levels
C. **glucose tolerance test**
D. urinary excretion of free cortisol
E. dexamethasone suppression test

Ans C The glucose tolerance test is often abnormal but is of no diagnostic utility. Levels of serum electrolytes are usually normal, but potassium may be decreased, especially in patients with tumors that secrete ACTH ectopically.

Precocious Puberty

133.Pendred syndrome is:

A. **Consistently associated with deafness**
B. Hypothyroidism is seen
C. Mutation in connexin coding gene
D. Mutation in chromosone 21 causing receptor defect

Ans A Consistently associated with deafness, b. Hypothyroidism is seen

134.All of the following can cause precocious puberty in males except:

A. **17 alpha hydroxylase deficiency**
B. 21 alpha hydroxylase deficiency
C. 11 hydroxylase deficiency
D. None of the above

Ans A

135.A five-year-old boy has precocious puberty along with BP 130/80 mm Hg. Estimation of which of the following will help in diagnosis?

A. 17-Hydroxyprogesterone
B. **11-Deoxycortisol**
C. Aldosterone
D. DOCA

Ans B Hypertension and precocious puberty in males can be seen in 11 beta hydroxylase deficiency, which can be diagnosed by increased 11 deoxycortisol levels.

136.Injection of Glucagon is effective for management of persistent hypoglycemia in all except:

A. Large for date baby
B. **Galactosemia**
C. Infant of diabetic mother
D. Nesidioblastosis

Ans B Glucagon will not be able to release Glucose from an already damaged liver, as seen in Galactosemia.

137. Most common cause of central precocious puberty in girls:

A. Exogenous estrogen
B. **Idiopathic**
C. CNS tumor
D. Hypothyroidism 109.

Ans B Although approximately 90% of girls have an idiopathic form, a structural central nervous system (CNS) abnormality can be demonstrated in up to 75% of boys with central precocious puberty. with

138.Drug of choice for precocious puberty:

A. Cyproterone acetate
B. Danazol
D. **GnRH agonists**

Ans D Nelson latest ed says: "Long-acting formulations of GnRH agonists, which maintain fairly constant serum concentrations of the drug for weeks or months, constitute the preparations of choice for treatment of central precocious puberty."

139.Most common brain lesion causing central precocious puberty is:

A. Tuberculous meningitis
B. Tuberous sclerosis
D. **Hypothalamic hamartoma**

Ans D Hypothalamic hamartomas are the most common brain lesion causing central precocious puberty.

140.Central precocious puberty in a girl is defined as breast development before the age of?

A. 6 years
B. **8 years**
C. 12 years
D. 10 years

Ans B Central precocious puberty is defined by onset of breast development before the age of 8 yrs in girls and by onset of testicular development (vol 4 mL) before the age of 9 yrs in boys, as a result of early activation of hypothalamic-pituitary-gonadal axis.

141. Precocious puberty is seen in:

A. **Hypothyroidism**
B. CNS irradation
C. McCune-Albright syndrome
D. Turner's syndrome
E. Congenital adrenal hyperplasia

Ans A c. McCune-Albright syndrome and e. Congenital adrenal hyperplasia •• In children with hypothyroidism, sexual maturation may also be delayed or might not take place at all.
• • Adolescents with acquired hypothyroidism also typically have delayed puberty;
• • Younger children with hypothyroidism might present with galactorrhea or pseudoprecocious puberty.
• • Galactorrhea is a result of increased TRH stimulating prolactin secretion and precocious puberty (breast development and vaginal

142.Precocious puberty in girls is defined by the onset of secondary sexual characteristics before the age of

A. 7 yr
B. **8 yr**
C. 9 yr
D. 10 yr
E. 11 yr

Ans B Precocious puberty is defined by the onset of secondary sexual characteristics before the age of 8 yr in girls and 9 yr in boys.

143. **The MOST common brain lesion causing central precocious puberty is**

A. postencephalitic scar
B. tuberculous meningitis
C. **hypothalamic hamartoma**
D. tuberous sclerosis
E. hydrocephalus

Ans C s are the most common brain lesion causing central precocious puberty.

Delayed Puberty

144. **A 6-month-old infant is brought with a history of constipation and excessive sleepiness. On examination, he is lethargic, has periorbital puffiness, large tongue and umbilical hernia. The investigation which will help to diagnose this condition is:**

A. **T4, TSH assay**
B. Karyoptyping
C. Rectal mucosal biopsy
D. Knee X-ray

Ans A It is a typical presentation of hypothyroidism, so thyroid hormone assay is required.

145. **A 2-year-old child presented with diarrhea and failure to thrive, Blood examination shows: Na = 122, K = 6. He is most probably suffering from:**

A. Bartter syndrome
B. **21 Hydroxylase deficiency**
C. 11-b Hydroxylase deficiency
D. 17-Hydroxylase deficiency

Ans B Presence of failure to thrive along with salt wasting leading to hyponatremia and hyperkalemia are compatible fo of Congenital adrenal hyperplasia due to 21 hydroxylase deficiency.

146. **Male pseudohermaphroditism is seen in:**

A. **5a reductase deficiency**
B. 21 hydroxylase deficiency
C. 17 hydroxylase deficiency
D. Gonadal dysgenesis
E. 11 beta hydroxylase deficiency

Ans A c. 17 Hydroxylase deficiency and d. Gonadal dysgenesis True Hermaphroditism: Both ovarian and testicular tissues are present either in the same (ovotestis) or opposite gonads. Male Pseudohermaphroditism: Genotype is XY, but the external genitalia is female. Causes of Male Pseudohermaphroditism A. Defect in testicular differentiation B. Defect in testicular hormone synthesis
• • Deletion of short arm Y chromosome •• Leydig cell aplasia

147. **Which disease is the most likely diagnosis as depicted by these pictures of a child, his hands and retina?**

A. Carpenter syndrome
B. Alstrom syndrome
C. **Bardet-Biedl syndrome**
D. Prader-Willi syndrome

Ans C The given pictures show obesity, hypogonadism, post axial polydactyly and pigmentary retinal changes, which are all suggestive of Laurence-Moon-Bardet Biedl syndrome.

148. **Indications for GH treatment to promote growth include the following EXCEPT**

A. GH deficiency
B. chronic renal failure before transplantation
C. **celiac disease**
D. Prader-Willi syndrome
E. Noonan syndrome

Ans C The FDA has approved 8 indications for GH treatment to promote growth. They are GH deficiency, Turner syndrome, chronic renal failure before transplantation, idiopathic short stature, small-for-gestational age short stature, Prader-Willi syndrome, SHOX gene abnormality, and Noonan syndrome.

149. **Adverse reaction reported exclusively with propylthiouracil is**

A. **severe liver disease**
B. agranulocytosis
C. lupus-like polyarthritis syndrome
D. glomerulonephritis
E. antineutrophilic cytoplasmic antibody–positive vasculitis

Ans A including liver failure requiring transplantation, has been reported exclusively with propylthiouracil.

150. **In primary aldosteronism, one of the following is low**

A. serum PH
B. serum carbon dioxide
C. serum sodium
D. **serum chloride**
E. serum calcium

Ans D Hypokalemia occurs frequently. Serum pH and the carbon dioxide and sodium concentrations may be elevated and the serum chloride and magnesium levels decreased. Serum levels of calcium are normal, even in children who manifest tetany.

151. **The MOST common cardiac defect in Turner syndrome is**

A. aortic coarctation
B. **isolated nonstenotic bicuspid aortic valves**
C. aortic stenosis
D. mitral valve prolapse
E. anomalous pulmonary venous drainage

Ans B Regardless of the age, all patients with Turner syndrome at the time of diagnosis need comprehensive cardiovascular evaluation by a cardiologist specializing in congenital heart disease. Complete cardiologic evaluation, including echocardiography, reveals isolated nonstenotic bicuspid aortic valves in one third to one half of patients. In later life, bicuspid aortic valve disease can progress to dilation of the aortic root.

152. **Microvascular complications of DM include**

A. **nephropathy**
B. accelerated coronary artery disease
C. cerebrovascular disease
D. peripheral vascular disease
E. peripheral neuropathies

Ans A Complications of DM can be divided into 3 major categories: (1) microvascular complications, specifically, retinopathy and nephropathy; (2) macrovascular complications, particularly accelerated coronary artery disease, cerebrovascular disease, and peripheral vascular disease; and (3) neuropathies, both peripheral and autonomic, affecting a variety of organs and systems. In addition, cataracts may occur more frequently.

Disorders of Sexual Development

153.A 8-year-old-child presents with lethargy, multiple epiphyseal breaks, wormian bones with growth retardation and mental retardation. Diagnosis is:

A. Rickets
B. **Hypothyroidism**
C. Scurvy
D. Hypoparathyroidism

Ans B

154.A 3-week-female infant is brought for ambiguous genitalia and hyperpigmentation of skin. She has hyponatremia and hyperkalemia. Which one of the following is the most likely diagnosis?

A. **21 hydroxylase deficiency**
B. 17 alpha hydroxylase deficiency
C. 17, 20 lyase deficiency
D. 11 beta hydroxylase deficiency

Ans A The given clinical scenario of ambiguous genitalia, hyperpigmentation and electrolyte disturbances suggest a diagnosis of CAH due to 21 hydroxylase deficiency. Features Seen in Adrenal Enzyme Deficiency 17-a-hydroxylase 11-b hydroxylase 21 hydroxylase and 3b-HSD Salt retention and hypertension Salt retention and hypertension Salt losing Feminization of male Virilization of female Virilization of female Precocious puberty in male Precocious puberty in male

155.A 7-year-old child presents to the pediatric clinic with ambiguous genitalia increasing with age. On examination her height, weight and blood pressure were recorded within normal limits. Labia appeared bifid with 2 separate perineal opening, phallic length was 2.5 cm and no palpable gonads were noted in the inguinal region. USG shows presence of mullerian structure. The most probable diagnosis is:

A. Classic Salt-Wasting 21 Hydroxylase deficiency
B. **Simple virilizing congenital adrenal hyperplasia**
C. Complete Androgen Insensitivity Syndrome
D. 5-Alpha Reductase Deficiency

Ans B As this child with ambiguous genitalia (with presence of Mullerian structures) never had features of shock most probably suffering from 'simple virilizing' type of CAH due to 21 hydroxylase deficiency;

156.Which of the following is the least common cause of ambiguous genitalia in a female child?

A. **Fetal placental steroid sulfatase deficiency**
B. Fetal placental aromatase deficiency
C. Congenital adrenal hyperplasia
D. WNT-4 gene mutation

Ans A Discussing the options one by one, a. Fetal placental steroid sulfatase deficiency has X-linked inheritance Affected individuals with this disorder are mostly males; so, it is uncommon in females b. Fetal placental aromatase deficiency with
• • Aromatase catalyzes conversion of androgens into estrogens; So in aromatase deficiency, estrogen synthesis cannot occur;
• • So, DHEA-S produced by fetal adrenal glands cannot be converted to estrogen by the placenta, so is converted to testosterone

157. The most common cause of ambiguous genitalia in a newborn is:

A. **21 hydroxylase deficiency**
B. 11-b hydroxylase deficiency
C. 17-b hydroxyalse deficiency
D. 3-b hydroxysteroid dehydrogenase deficiency

Ans A

158.Reported side effects of GH treatment include the following EXCEPT

A. pseudotumor cerebri
B. slipped capital femoral epiphysis
C. gynecomastia
D. worsening of scoliosis
E. **6-fold increase in the risk for type 1 diabetes**

Ans E Recent studies indicate that GH treatment is associated with a 6-fold increase in the risk for type 2 diabetes and no significant increase in the risk for type 1 diabetes.

159.Hypocalcemia is common in neonates between 12 and 72 hr of life, especially in infants with

A. **birth asphyxia**
B. sepsis
C. exchange transfusion
D. hypomagnesemia
E. hyperbilirubinemia

Ans A Hypocalcemia is common in neonates between 12 and 72 hr of life, especially in premature infants, in infants with asphyxia, and in infants of diabetic mothers (early neonatal hypocalcemia).

160.The major causes of incidentalomas are

A. **benign adenomas**
B. pheochromocytomas
C. adrenocortical carcinoma
D. hemorrhagic cysts
E. myelolipomas

Ans A Benign, hormonally inactive adrenocortical adenomas make up the majority of incidentalomas.

161. The 47,XXX (trisomy) chromosomal constitution is the most frequent extra-X chromosome abnormality in females, occurring in almost 1 in 1,000 liveborn females. It is characterized by the following EXCEPT

A. maternal meiotic nondisjunction is the most common cause
B. normal female phenotype
C. **sexual development and menarche are delayed**
D. tall and gangly
E. behavior disorders

Ans C Sexual development and menarche are normal. Most pregnancies have resulted in normal infants.

162.Guidelines suggest that prepubertal T1DM patients should commence screening for retinopathy

A. at diagnosis
B. 1 yr after diagnosis
C. 3yr after diagnosis
D. **5 yr after diagnosis**
E. 10yr after diagnosis

Ans D Screening for retinopathy commenced after 5 yr duration in prepubertal children, after 2 yr in pubertal children.

Hypopituitarism

163. Which is false in congenital hypopituitarism?

A. Growth hormone levels 7 ng/mL
B. Hypoglycemia
C. Baby small at birth
D. Delayed puberty

Ans C The child with hypopituitarism usually has normal size & weight at birth, although those with MPHD (Multiple pituitary hormone deficiency) & genetic defects of the GH1 or GHR gene have birth lengths <1 SD below the mean. Clinical Features of Growth Hormone deficiency: Craniofacies Growth and development Other features Frontal bossing Birth weight and length: near-normal Hypoglycemia: in infants and children; Normal head circumference; Postnatal severe growth failure Motor milestones: delayed

164. A 4-week-old female child with normal genitalia presents to the emergency department with severe dehydration, hyperkalemia and hyponatremia. Measurement blood levels of which of the following will be helpful?

A. Increase 17-hydroxyprogesterone
B. Increase cortisol
C. Increase aldosterone
D. Increase 11 deoxycortisol

Ans D Aldosterone The serum electrolyte abnormalities (hyperkalemia and hyponatremia) along with normal genitalia deficiency or altered end-organ sensitivity to mineralocorticoids (Pseudohypoaldosteronism).
• • Both plasma renin and aldosterone are markedly elevated in Pseudohypoaldosteronism, but aldosterone hypoaldosteronism; so aldosterone level will help in diagnosing the illness
• • Levels of cortisol and ACTH are normal; If hypovolemia is severe, patients may develop prerenal azotemia.

165. A 7-year-old boy underwent neurosurgery for cranio-pharyngioma following which pituitary functions were lost. Which of the following hormone should be re- placed first?

A. Hydrocortisone
B. Thyroxine
C. Growth hormone
D. Prolactin

Ans A Hydrocortisone should be supplemented first if all pituitary function is lost.

166. Obesity in a child of age 2 years is defined as:

A. Weight for height more than double of expected
B. Weight for age more than double of expected
C. BMI >
D. BMI > 95th percentile

Ans D Obesity in children is defined as BMI > 95th percentile; Body mass index (BMI) is equal to weight/height2 (in kg/m2).

167. The pituitary gland is located in a saddle-shaped cavity of the

A. frontal bone
B. maxillary bone
C. sphenoid bone
D. ethemoid bone
E. temporal bone

Ans C The pituitary gland is located at the base of the skull in a saddle-shaped cavity of the sphenoid bone called the sella turcica.

168. The MOST common cause of acquired hypopituitarism is

A. craniopharyngioma
B. eosinophilic granuloma (histiocytosis)
C. tuberculosis
D. toxoplasmosis
E. meningitis

Ans A Any lesion that damages the hypothalamus, pituitary stalk, or anterior pituitary can cause pituitary hormone deficiency. Because such lesions are not selective, multiple hormonal deficiencies are usually observed. Diabetes insipidus is more frequent in acquired than in congenital hypopituitarism. The most common lesion is the craniopharyngioma.

169. Congenital hypogonadotropic hypogonadism include

A. gonadal dysgenesis
B. Klinefelter syndrome (47,XXY)
C. Noonan syndrome
D. cystic fibrosis
E. Laurence-Moon- Biedl syndrome

Ans E Follicle-stimulating hormone (FSH) and luteinizing hormone (LH) resistance Mutations in steroid synthetic pathways Gonadal dysgenesis Klinefelter syndrome (47,XXY) Noonan syndrome (PTPN-11 gene mutation in many cases) Cystic fibrosis (infertility) are congenital causes of hypergonadotropic hypogonadism (primary hypogonadism; testes).

170. The MOST common cardiac defect in Noonan syndrome is

A. aortic coarctation
B. pulmonary valvular stenosis
C. aortic stenosis
D. mitral valve prolapse
E. anomalous pulmonary venous drainage

Ans B The phenotype differs from Turner syndrome in several respects. Cognitive impairment is often present, the cardiac defect is most often pulmonary valvular stenosis or an atrial septal defect rather than an aortic defect, normal sexual maturation usually occurs but is delayed by 2 yr on average, and premature ovarian failure has been reported.

171. Guidelines suggest that diabetic patients should commence screening for celiac disease

A. at diagnosis
B. 1 yr after diagnosis
C. 3 yr after diagnosis
D. 5 yr after diagnosis
E. 10 yr after diagnosis

Ans A Screening for thyroid disease and celiac disease commenced at diagnosis.

Diabetes Insipidus

172. **Lesions of which of the hypothalamic nuclei cause diabetes insipidus:**

A. Dorsomedial nuclei
B. **Supraoptic and paraventricular nuclei**
C. Median preoptic nuclei
D. Ventromedial nuclei

Ans B Vasopressin, a 9-amino-acid peptide, has both antidiuretic and vascular pressor activity and is synthesized in the paraventricular and supraoptic nuclei of the hypothalamus. Deficiency of Vasopressin is responsible for Diabetes insipidus.

173. **Neonatal hypoglycemia is caused by:**

A. IV glucocorticoid administration
B. **Epinephrine deficiency**
C. Mesothelioma
D. Pancreatic atresia

Ans B Insulin and counter-regulatory hormones in Hypoglycemia:
• • Hypoglycemic effects of insulin are opposed by the actions of counter-regulatory hormones like glucagon, cortisol, and epinephrine, whose concentration in plasma increases as blood glucose falls; They act by: – Activating glycogenolytic enzymes (glucagon, epinephrine) – Inducing gluconeogenic enzymes (glucagon, cortisol) – Inhibiting glucose uptake by muscle (epinephrine, growth hormone, cortisol)

174. **Which protein secreted by adipocytes prevents obesity?**

A. Cathepsin
B. **Leptin**
C. Neuropeptide Y
D. Galanin DISORDERS OF CALCIUM METABOLISM

Ans B •• Leptin is a peptide produced by the ob gene; Its name derived from the Greek root leptos, meaning thin
• • Leptin is secreted by adipose cells and acts primarily through the hypothalamus
• • High leptin levels decrease food intake and increase energy expenditure, thereby preventing obesity

175. **The following stimulate vasopressin (VP) secretion EXCEPT**

A. hyperosmolality
B. hypovolemia
C. hypotension
D. **hyperglycemia**
E. nausea

Ans D Regulation of vasopressin (VP) secretion and serum osmolality. Hyperosmolality, hypovolemia, and hypotension are sensed by osmosensors, volume sensors, and barosensors, respectively. These stimulate both VP secretion and thirst. VP, acting on the kidney, causes increased reabsorption of water (antidiuresis). Thirst causes increased water ingestion. The results of these dual negative feedback loops cause a reduction in hyperosmolality or in hypotension or hypovolemia.

176. **Acquired nephrogenic diabetes insipidus (NDI) is associated with the following**

A. lithium
B. **vancomycin**
C. amphotericin
D. methicillin
E. rifampin

Ans B Acquired NDI can result from hypercalcemia or hypokalemia and is associated with lithium, demeclocycline, foscarnet, clozapine, amphotericin, methicillin, and rifampin.

177. **Neonates and young infants with central diabetes insipidus are often best treated with**

A. **fluid therapy**
B. vasopressin analogs
C. thiazide diuretics
D. indomethacin
E. amiloride

Ans A Neonates and young infants are often best treated solely with fluid therapy, given their requirement for large volumes (3 L/m2/24 hr) of nutritive fluid.

178. **Familial isolated gonadotropin deficiency associated with anosmia is**

A. Noonan syndrome
B. Bloom syndrome
C. Werner syndrome
D. **Kallmann syndrome**
E. Prader-Willi

Ans D

179. **Transient elevation of urinary albumin can occur in the following conditions EXCEPT**

A. **hypoglycemia**
B. strenuous exercise
C. urinary tract infections
D. heart failure
E. acute febrile illness

Ans A Short-term hyperglycemia, strenuous exercise, urinary tract infections, marked hypertension, heart failure, and acute febrile illness can cause transient elevation urinary albumin excretion. There is marked day-to-day variability in albumin excretion, so at least 2 of 3 collections done in a 3-6 mo period should show elevated levels before microalbuminuria is diagnosed and treatment is started.

Calcium & Parathyroid Disorders

180. **Commonest cause of female pseudohermaphrodism:**

A. **Congenital Adrenal hyperplasia**
B. Maternal ovarian tumor
C. Maternal drug intake
D. Chromosomal abnormalities

Ans A

181. **An 8-day-old male infant is brought to the emergency department with vomiting, lethargy, dehydration and features of shock. Clinical examination reveals hyperpigmentation of the genital skin and normal external genitalia. Blood investigations reveal Sodium 124 mEq/L, potassium of 7 mEq/L and hypoglycemia. Which of the following is the most likely diagnosis?**

A. Congenital Adrenal Hyperplasia (CAH)
B. Adrenal Hemorrhage
C. Acute Gastorenteritis with dehydration
D. Hyperaldosteronism

Ans A The given clinical scenario of dehydration, hyperpigmentation of genitalia and laboratory features of hypoglycemia, hyponatremia and hyperkalemia suggest a diagnosis of CAH due to 21 hydroxylase deficiency.

182. **Salt losing hydroxylase deficiency is characterized by:**

A. Hyponatremia
B. Hyperkalemia
C. Hypoglycemia
D. Hypocalcemia
E. Hypercalcemia

Ans A b. Hyperkalemia, c. Hypoglycemia

183. **Select the FALSE combination of chromosomal pattern and the syndrome:**

A. Mayer Rokitansky-46XY
B. Swyer's syndrome-46XY
C. Turner's syndrome-45XO
D. Klinefelter's syndrome-47XXY 113.

Ans A Mayer-Rokitansky-Küster-Hauser syndrome (also called' Mullerian Agenesis') is characterized by the failure of development of uterus and vagina, in women with normal ovarian function and normal external genitalia.

184. **Osteitis fibrosa cystica is a feature seen in:**

A. Paget's disease
B. Hyperparathyroidism
C. Multiple myeloma
D. Hypocalcemia

Ans B Brown tumor (also known as osteitis fibrosa cystica) is one of the manifestations of hyperparathyroidism Osteitis fibrosa may cause bone pain or tenderness. There may be fractures in the arms, legs, or spine.

185. **Broad forehead, hypertelorism, small chin, long philtrum, camptodactyly, and fetal finger pads are features of the following genetic overgrowth syndrome**

A. Perlman syndrome
B. Sotos syndrome
C. Weaver syndrome
D. Beals syndrome
E. Beckwith-Wiedemann syndrome (BWS)

Ans C

186. **Episodic symptomatic hypocalcemia occurs in the Kenny-Caffey syndrome, the latter is characterized by the following EXCEPT**

A. medullary stenosis of the long bones
B. short stature
C. delayed closure of the fontanel
D. advanced bone age
E. eye abnormalities

Ans D Delayed bone age.

187. **Acquired hypogonadotropic hypogonadism include**

A. chemotherapy
B. malnutrition
C. cystic fibrosis
D. infection (e.g., mumps)
E. infarction (testicular torsion)

Ans B Anorexia nervosa Drug use Malnutrition Chronic illness, especially Crohn disease Hyperprolactinemia Pituitary tumors Pituitary infarction Infiltrative disorders (e.g., histiocytosis, sarcoidosis) Hemosiderosis and hemochromatosis Radiation are acquired causes of hypogonadotropic hypogonadism (secondary hypogonadism; hypothalamic-pituitary).

188. **Children with type 2 DM often seek medical care because of**

A. weight loss
B. excessive weight gain
C. polyuria
D. polydipsia
E. anorexia

Ans B The presentation of T2DM is typically more insidious than that with T1DM. In contrast to patients with T1DM who are usually ill at the time of diagnosis and whose presentation rarely spans more than a few weeks, children with T2DM often seek medical care because of excessive weight gain and fatigue as a result of insulin resistance and/or the incidental finding of glycosuria during routine physical examination.

189. **The MOST commonly used and the only FDA-approved oral agent for the treatment of T2 DM in children and adolescents is**

A. thiazolidinediones
B. pramlintide
C. metformin
D. sulfonylureas
E. acarbose

Ans C The most commonly used and the only FDA-approved oral agent for the treatment of T2DM in children and adolescents is metformin. Renal function must be assessed before starting metformin as impaired renal function has been associated with potentially fatal lactic acidosis. Significant hepatic dysfunction is also a contraindication to metformin use, although mild elevations in liver enzymes may not be an absolute contraindication. The usual starting dose is 500 mg once daily.

Rickets & Vitamin D

190. **Aldosterone acts by promoting:**

A. Reabsorption of sodium and excretion of potassium
B. Reabsorption of potassium and excretion of sodium &
C. Excretion of both sodium and potassium
D. Reabsorption of both sodium and potassium

Ans A

191. **A 10-month-old baby previously normal, suddenly becomes distressed in his crib. The external appearance of genitalia was normal, except hyperpigmentation. Blood glucose showed a level of 30 mg%. What is the most probable diagnosis?**

A. 21 hydroxylase deficiency
B. Hyperinsulinism
C. Familial glucocorticoid deficiency
D. Cushing's syndrome

Ans C Hypoglycemia with hyperpigmentation suggests a diagnosis of glucocorticoid deficiency.

192. **A 10-day-old male pseudohermaphrodite child with 46 XY karyotype presents with BP of 110/80 mm Hg. Most likely enzyme deficiency is:**

A. 21 hydroxylase
B. 17 hydroxylase
C. 11 hydroxylase
D. 3-beta hydroxylase DIABETES MELLITUS

Ans B In 17-hydroxylase deficiency: All pregnonolone is converted to mineralocorticoid. So, there will be: fo •• Excess of mineralocorticoids → Salt retention and hypertension.
• • No androgens → Feminization of external male genitalia, while genitalia development will be normal in females Coming back to this question, this neonate has: Hypertension (normal BP in a neonate = 75/50) and male pseudohermaphroditism, both of which are features of 17-alpha hydroxylase deficiency.

193. **Androgen insensitivity syndrome – true is?**

A. 46XX
B. Vagina present
C. Androgen receptors working

Ans B Discussing about the options one by one, a. 46XX False Karyotype in Androgen insensitivity syndrome is 46, XY b. Vagina present True A blind vagina is present c. Androgen receptors working False Resistance or insensitivity of androgen receptors present d. Amenorrhea with normal secondary sexual characters False Pubic hair, which is a secondary sexual character is absent

194. **Familial hypocalciuric hypercalcemia is an autosomal dominant disorder characterized by mild elevation of calcium and PTH levels. It occurs secondary to what mutation?**

A. Missense mutation of mitochondrial calcium receptor
B. Missense mutation of ribosomal calcium surface protein
C. Missense mutation of Golgi complex calcium receptor
D. Mutations of the calcium-sensing receptor (CASR)

Ans D

195. **All the following are true in premature thelarche EXCEPT**

A. most often appears in the 1st 2 yr of life
B. growth and osseous maturation are usually normal
C. menarche occurs at the expected age
D. may be unilateral
E. often persists till puberty

Ans E Breast development may regress after 2 yr, often persists for 3-5 yr, and is rarely progressive.

196. **All the following are causes of infantile hypercalcemia EXCEPT**

A. subcutaneous fat necrosis
B. metaphyseal chondrodysplasia
C. lactase/disaccharidase deficiency
D. blue diaper syndrome
E. proximal renal tubular acidosis

Ans E Distal renal tubular acidosis.

197. **Klinefelter syndrome is the most common sex chromosomal aneuploidy in males, all the following features are true EXCEPT**

A. 80% of them have a 47,xxy chromosome complement
B. diagnosis is rarely made before puberty
C. psychiatric disorders may be apparent long before defects in sexual development
D. few complete high school
E. approximately 80% of adults have gynecomastia

Ans D Verbal cognitive defects and underachievement in reading, spelling, and mathematics are common. By late adolescence, many boys with Klinefelter syndrome have generalized learning disabilities, most of which are language based. Despite these difficulties, most complete high school.

198. **The current criteria for the diagnosis of type 1 and type 2 DM is a fasting blood glucose that exceeds**

A. 110 mg/dL
B. 115 mg/dL
C. 120 mg/dL
D. 125 mg/dL
E. 130 mg/dL

Ans D It should be noted that a fasting blood glucose thatexceeds 125 mg/dL (6.9 mmol/L) is the accepted criterion for the diagnosis of diabetes.

199. **Donohue syndrome is characterized by the following EXCEPT**

A. intrauterine growth restriction
B. fasting hypoglycemia
C. acanthosis nigricans
D. postprandial hyperglycemia
E. profound resistance to insulin

Ans C Extreme insulin resistance, acanthosis nigricans, abnormalities of the teeth and nails, and pineal hyperplasia are features of Rabson-Mendenhall syndrome.

Obesity & Metabolic Syndrome

200.A 1-week-old male newborn suddenly presents with lethargy, poor oral intake and shock. Investigations reveal hyperkalemia, hyponatremia and hypoglycemia. What is the most likely diagnosis?

A. Syndrome of inappropriate ADH secretion
B. Gram negative sepsis
C. Congenital adrenal hyperplasia
D. Phenylketonuria

Ans C The given laboratory findings along with dehydration in a male newborn with normal genitalia suggest a diagnosis of CAH, which is most commonly due to 21 hydroxylase deficiency.

201.Screening for nephropathy in prepubertal children with type 1 DM should be initiated after how many years of disease onset?

A. 2 years
B. 3 years
C. 4 years
D. 5 years

Ans D ADA recommendations 2017 Annual screening for albuminuria with a random spot urine sample for albumin-to-creatinine ratio should be considered once the child has had type 1 diabetes for 5 years.

202.In hypogonadotropic hypogonadism?

A. Both LH and FSH decreased
B. Both LH and FSH increased
C. LH increased and FSH decreased 116.
D. LH decreased and FSH increased

Ans A LH and FSH are increased in hypergonadotrophic hypogonadism.

203.A 3-year-old boy is detected to have bilateral renal calculi. Metabolic evaluation confirms the presence of marked hypercalciuria with normal blood levels of calcium, magnesium, phosphate, Uric acid and creatinine. A diagnosis of idiopathic hypercalciuria is made. The dietary management includes all, except:

A. Increased water intake
B. Low sodium diet
C. Reduced calcium intake
D. Avoid meat proteins

Ans C Dietary calcium restriction is not recommended in Idiopathic Hypercalciuria, because of obligate requirement

204.Premature adrenarche has traditionally applied to the appearance of sexual hair before the age of 8 yr in girls or 9 yr in boys without other evidence of maturation. It is characterized by the following EXCEPT

A. more frequent in girls
B. higher prevalence in african-american
C. axillary hair generally appears earlier
D. adult-type axillary odor is common
E. affected children are often slightly advanced in height

Ans C Hair appears on the mons and labia majora in girls and perineal and scrotal area in boys; axillary hair generally appears later.

205.Glucocorticoids play a major role in immune regulation. They increase circulating

A. polymorphonuclear cell
B. monocytes
C. eosinophils
D. lymphocytes
E. basophils

Ans A High doses of glucocorticoids deplete monocytes and lymphocytes, especially T cells. They do so at least in part by inducing cell-cycle arrest in the G1 phase and by activating apoptosis through glucocorticoid receptor–mediated effects. The effects on lymphocytes are primarily exerted on T-helper 1 cells and hence on cellular immunity, whereas the T-helper 2 cells are spared, leading to a predominantly humoral immune response.

206.True gynecomastia is characterized by the presence of a palpable fibroglandular mass (located concentrically beneath the nipple and areolar region) at least

A. 0.5 cm in diameter
B. 1 cm in diameter
C. 1.5 cm in diameter
D. 2 cm in diameter
E. 2.5 cm in diameter

Ans A

207.The clearest evidence of a role for viral infection in human type 1 DM is seen in

A. acquired rubella infection
B. live-virus rubella immunization
C. enteroviral infection
D. congenital rubella syndrome
E. mumps infection

Ans D The clearest evidence of a role for viral infection in human T1DM is seen in congenital rubella syndrome. Prenatal infection with rubella is associated with β-cell autoimmunity in up to 70%, with development of T1DM in up to 40% of infected children.

208.One of the following do not appear to be of concern in cystic fibrosis–related diabetes (CFRD)

A. ketoacidosis
B. macrovascular complications
C. microvascular complications
D. frequent infections
E. energy needs

Ans B do not appear to be of concern in CFRD, perhaps because of the shortened life span of these patients.

Ambiguous Genitalia

209.Nelson syndrome is seen in:

A. Adrenalectomy
B. Hypopitutarism
C. Deficiency of beta cells
D. Deficiency of growth hormone

Ans A Nelson syndrome refers to the development or enlargement of an ACTH-secreting pituitary adenoma following bilateral adrenalectomy for Cushing's disease.

210.A newborn presents with shock, hyperkalemia and hypoglycemia. What is the most likely diagnosis?

A. Septicemia
B. Inborn error of metabolism
C. Diabetes mellitus
D. **Congenital adrenal hyperplasia**

Ans D

211. A 5-year-old girl presents with hypertension and virilization. There is also finding of hypokalemia. What is the diagnosis?

A. 21-hydroxylase deficiency
B. 3-b hydroxy steroid deficiency
C. **11-b hydroxylase deficiency**
D. Conn's disease

Ans C Hypertension and Virilization in females is seen in 11-b hydroxylase deficiency

212.A 5-year-old girl presents with hypertension and virilization. There is also finding of hypokalemia. What is the diagnosis?

A. Hydrocortisone
B. Prednisolone
C. **Fludrocortisone**
D. Dexamethasone

Ans C 11-b hydroxylase deficiency Hypertension, hypokalemia and virilization suggest the diagnosis of 11-b hydroxylase deficiency.

213.A baby presents with tetany. First thing to be done is administration of:

A. Diazepam
B. Vitamin D
C. **Calcium gluconate**
D. Calcitonin Chapter 20: Pediatric Endo crinology 473 Answers

Ans C The baby presenting with tetany is hypocalcemic, so must be treated immediately with calcium gluconate Pediatric Endo crinology 481 mutation of Ca2+ sensing Answers during hypocalcemia. These (22q11 deletion) with p 521-524 hormone (PTH) of phosphorus. for details for growth. IV. What does the ufoelslotwioinng demon- strate in a fullQ term neona t1e? a. Normal tone b. Hypotonia c. Hypertonia d. Head control M What is the namuee sotf itohen congenital anomaly thiQs child is 2suffering from? a. Encephalocele b.

214.Peak serum concentrations of TSH at birth in full-term infants reach

A. 20 mU/L
B. 40 mU/L
C. **60 mU/L**
D. 80 mU/L
E. 100 mU/L

Ans C At birth, there is an acute release of TSH; peak serum concentrations reach 60 mU/L 30 min following delivery in full-term infants. A rapid decline occurs in the ensuing 24 hr and a more gradual decline over the next 5 days to <10 mIU/L. The acute increase in TSH produces a dramatic increase in levels of T4 to approximately 16 µg/dL and of T3 to approximately 300 ng/dL in about 4 hr. This T3 seems largely derived from increased peripheral conversion of T4 to T3.

215.Glucocorticoids readily penetrate the blood–brain barrier and have direct effects on brain metabolism. They have the following actions EXCEPT

A. decrease brain edema
B. insomnia
C. increased irritability
D. impaired memory
E. **improved ability to concentrate**

Ans E There is an increase in irritability and emotional lability, with an impairment of memory and ability to concentrate.

216.Weaker associations with gynecomastia are seen in

A. spironolactone
B. alkylating agents
C. ketoconazole
D. cimetidine
E. **opiates**

Ans E Weaker associations are seen with a large number of other medications and drugs of abuse, including opiates, alcohol, and marijuana, although the association with marijuana may not be as strong as previously thought.

217. The following factors are implicated in the pathogenesis of type 1 DM EXCEPT

A. prenatal influences
B. diet in infancy
C. viral infections
D. **excessive exposure to certain infections**
E. psychologic stress

Ans D A number of factors, including prenatal influences, diet in infancy, viral infections, lack of exposure to certain infections even psychologic stress, are implicated in the pathogenesis of T1DM, but their exact role and the mechanism by which they trigger or aggravate autoimmunity remains uncertain.

21

CHAPTER 21

Pediatric Neurology



WHAT THIS CHAPTER COVERS

- Seizures & Epilepsy
- Febrile Seizures
- Cerebral Palsy
- Meningitis
- Encephalitis
- Neurocysticercosis
- Tuberculous Meningitis
- Brain Abscess
- Hydrocephalus
- Neural Tube Defects
- Neurodegenerative Disorders
- Neuromuscular Disorders
- Neurocutaneous Syndromes
- Headache
- Neonatal Seizures
- Hypoxic Ischemic Encephalopathy
- Intraventricular Hemorrhage
- Periventricular Leukomalacia
- Developmental Delay
- Autism Spectrum Disorder
- ADHD

264 MCQs **21** subtopics

21.1 Seizures & Epilepsy – Basics

- **Seizure**: transient signs/symptoms due to abnormal excessive synchronous neuronal activity. **Epilepsy (ILAE 2014)**: **≥ 2 unprovoked seizures > 24 h apart**, or 1 unprovoked seizure with ≥ 60% recurrence risk, or an epilepsy syndrome.
- **ILAE 2017 classification**: **focal onset** (aware/impaired awareness; motor/non-motor; focal to bilateral tonic-clonic), **generalised onset** (motor – tonic-clonic, clonic, tonic, myoclonic, atonic, epileptic spasms; non-motor – **absence**), unknown onset.
- **MCC of seizures in children < 5 y: febrile seizures**; **MCC of focal seizures in Indian children: neurocysticercosis**; neonates – HIE.
- **Paroxysmal non-epileptic events (mimics)**: **breath-holding spells, syncope (reflex anoxic seizures)**, night terrors, **benign paroxysmal vertigo**, tics, **benign sleep myoclonus of infancy**, **self-gratification (infantile masturbation)**, Sandifer, shuddering attacks, psychogenic non-epileptic seizures.

Breath-holding spells

- **6 months–5 years** (peak 1–2 y); provoked by **crying/frustration (cyanotic type)** or minor injury/fright (**pallid type – vagal-mediated bradycardia/asystole**); child cries → holds breath in expiration → cyanosis → LOC ± brief jerks → recovery; **never during sleep**.
- **Associated with iron-deficiency anaemia** – **oral iron reduces frequency even without anaemia**; reassurance; resolve by 5–6 y; ECG to exclude long QT (pallid).

21.2 Febrile Seizures

- **Seizure with fever (≥ 38 °C) in a child aged 6 months–5 years (60 months)**, without CNS infection, metabolic cause or prior afebrile seizure; **2–5% of children**; peak **18 months**; **genetic predisposition** (family history), viral (**HHV-6 – roseola**, influenza); **seizure usually occurs on first day of fever (early in rise)**.

Simple febrile seizure (70–75%)	Complex febrile seizure
Generalised (tonic-clonic)	**Focal** onset or focal features
< 15 minutes	**≥ 15 minutes** (febrile status ≥ 30 min)
Single in 24 h (single in illness)	**Recurrent within 24 h**
No post-ictal deficit	Post-ictal **Todd's palsy**

- **Investigations**: none routinely for simple FS beyond finding fever focus; **LP** – **< 12 months if not fully immunised for Hib/pneumococcus or on antibiotics**, meningeal signs, prolonged altered sensorium; **EEG & neuroimaging – not indicated in simple FS**; consider in complex FS/focal deficits.
- **Recurrence risk ~30%** (overall); risk factors – **age < 18 months (or < 12 m) at first FS (strongest)**, **family history of FS**, **low peak temperature (< 40 °C) at seizure**, **short duration of fever (< 1 h) before seizure**; ≥ 3 risk factors → > 60%.
- **Risk of later epilepsy**: simple FS ~1–2% (slightly > general 1%); higher with **complex FS, neurodevelopmental abnormality, family history of epilepsy**, short fever duration.

MANAGEMENT

- Acute: **seizure > 5 min → benzodiazepine – buccal/intranasal midazolam 0.2–0.3 mg/kg or rectal diazepam 0.5 mg/kg or IV lorazepam 0.1 mg/kg**.
- **Antipyretics (paracetamol/ibuprofen) for comfort – do NOT prevent recurrence.**
- **No continuous prophylaxis for simple FS**; **intermittent oral clobazam (0.75–1 mg/kg/day × 2–3 days during febrile illness)** for frequent/prolonged FS or parental anxiety; continuous (valproate/levetiracetam) rarely.
- **Counsel parents** (benign; no brain damage; first aid – lateral position, nothing in mouth).

- **Febrile seizure plus (GEFS+)** – SCN1A; **Dravet syndrome** (SCN1A – prolonged febrile hemiclonic seizures in infancy → myoclonic, developmental decline; **avoid sodium-channel blockers – carbamazepine, phenytoin, lamotrigine**; Rx valproate, clobazam, stiripentol, fenfluramine, cannabidiol). **FIRES** – febrile infection-related epilepsy syndrome (super-refractory status).

21.3 Epilepsy Syndromes of Childhood

Syndrome	Age	Features	EEG	Treatment / prognosis
West syndrome (infantile spasms / IESS)	**3–12 months (peak 4–8 m)**	**Triad: epileptic spasms (flexor "salaam/jack-knife", in clusters on waking) + developmental arrest/ regression + hypsarrhythmia**	**Hypsarrhythmia – chaotic high-voltage slow waves with multifocal spikes**	**ACTH (DOC) or high-dose oral prednisolone**; **vigabatrin – DOC in tuberous sclerosis**; cause – **HIE (MC in India)**, TSC, CNS malformations, Down, IEM; poor prognosis (ID) – evolves to Lennox–Gastaut
Lennox–Gastaut syndrome	**1–8 y** (peak 3–5 y)	**Multiple seizure types – tonic (in sleep, characteristic), atonic (drop attacks), atypical absence**; ID	**Slow (< 2.5 Hz) spike-and-wave; paroxysmal fast activity (GPFA) in sleep**	Valproate, lamotrigine, rufinamide, clobazam, **cannabidiol**, felbamate; ketogenic diet, corpus callosotomy (drops); poor prognosis
Childhood absence epilepsy	**4–10 y**, girls	**Brief (5–15 s) staring spells, many/day, abrupt onset/ offset, no post-ictal state, eyelid flutter, automatisms (lip smacking)**; **provoked by hyperventilation (3–5 min) – bedside test**	**3 Hz generalised spike-and-wave**	**Ethosuximide – DOC (pure absence)**; valproate (if GTCS too), lamotrigine; **avoid carbamazepine/phenytoin (worsen)**; good prognosis – remits by adolescence
Juvenile myoclonic epilepsy (Janz)	**12–18 y**	**Morning myoclonic jerks (drops objects – toothbrush/cup) on awakening**, GTCS, absences; triggered by **sleep deprivation, alcohol, photic**	**4–6 Hz polyspike-and-wave**, photosensitivity	**Valproate (DOC in boys)**; **levetiracetam (DOC in girls of child-bearing age – avoid valproate)**, lamotrigine (may worsen myoclonus); **lifelong treatment (relapse on withdrawal)**, normal intelligence
Self-limited epilepsy with centrotemporal spikes (BECTS/ Rolandic)	**5–10 y (3–13)**	**Nocturnal focal seizures – hemifacial twitching, drooling, speech arrest, oropharyngeal sensations; awake; may generalise in sleep**	**Centrotemporal (Rolandic) spikes, activated by sleep**	**Often no treatment**; carbamazepine/levetiracetam if frequent; **excellent prognosis – remits by puberty**
Panayiotopoulos syndrome	3–6 y	**Autonomic seizures – vomiting, pallor, eye deviation**, prolonged	Occipital/multifocal spikes	Benign

Syndrome	Age	Features	EEG	Treatment / prognosis
Landau–Kleffner syndrome	3–7 y	**Acquired epileptic aphasia (verbal auditory agnosia)** after normal language; seizures	**ESES (electrical status epilepticus in sleep)**, temporal spikes	Steroids, valproate, benzodiazepines
Ohtahara / early infantile DEE	First weeks	Tonic spasms	**Burst-suppression**	Very poor; brain malformations, STXBP1, KCNQ2
Early myoclonic encephalopathy	Neonatal	Myoclonus	Burst-suppression	**NKH, IEM**
Rasmussen encephalitis	6–8 y	**Progressive hemiparesis + epilepsia partialis continua**, unilateral hemispheric atrophy		**Hemispherectomy**

DRUG OF CHOICE SUMMARY

Neonatal seizures – phenobarbitone · West – ACTH (vigabatrin in TSC) · Absence – ethosuximide (valproate if GTCS) · JME – valproate (levetiracetam in girls) · Focal seizures – carbamazepine/oxcarbazepine/levetiracetam · Generalised tonic-clonic – valproate (levetiracetam/lamotrigine in girls) · Lennox–Gastaut – valproate · Dravet – valproate + clobazam (+ stiripentol); avoid Na-channel blockers · Status epilepticus – IV lorazepam/midazolam → levetiracetam/phenytoin/valproate.

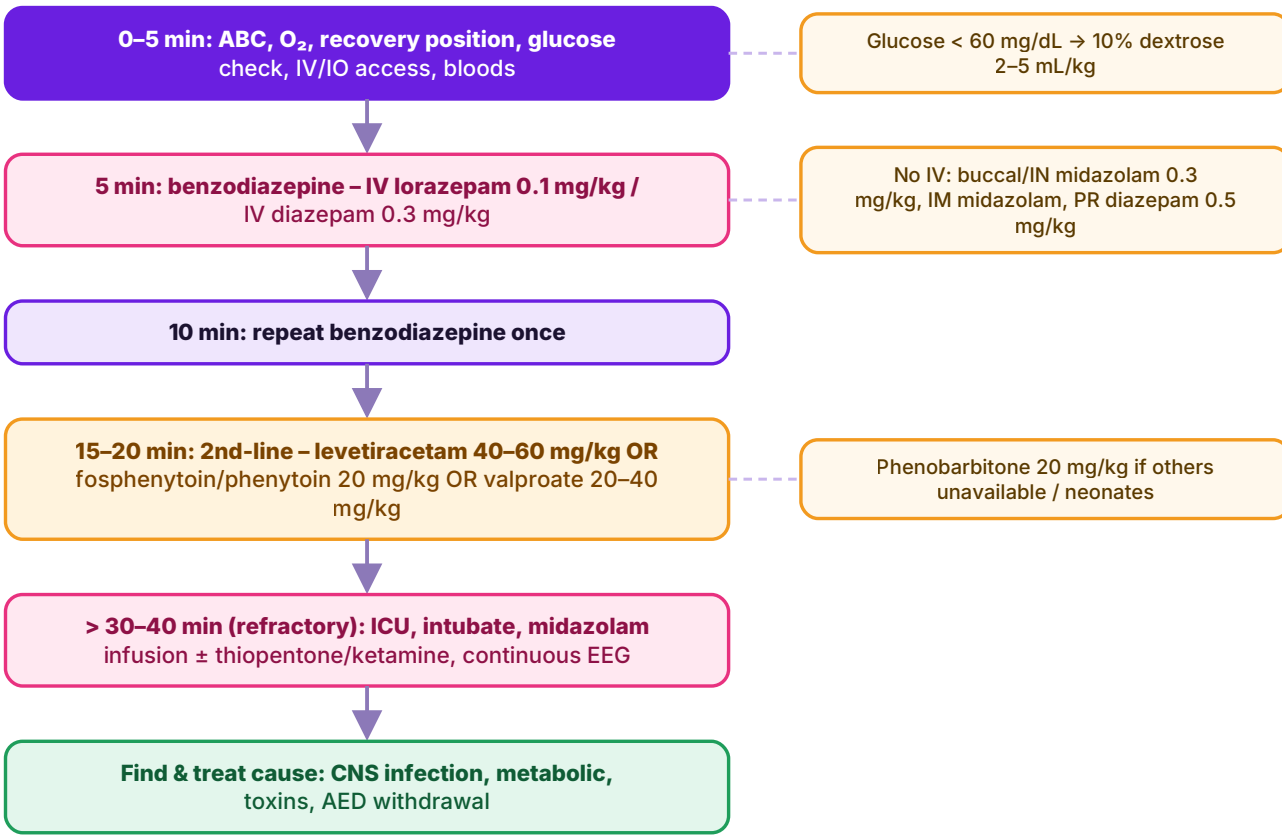
Antiseizure medication – side effects

Drug	Notable adverse effects
Phenytoin	**Gum hypertrophy, hirsutism, coarse facies**, nystagmus/ataxia, **zero-order kinetics**, megaloblastic anaemia, osteomalacia, **SJS (HLA-B*1502)**, **fetal hydantoin syndrome**, purple-glove (IV)
Valproate	**Hepatotoxicity (< 2 y, polytherapy, mitochondrial – POLG)**, pancreatitis, weight gain, **tremor, hair loss (curly regrowth)**, **thrombocytopenia**, hyperammonaemia, **PCOS**, **NTD (most teratogenic)**
Carbamazepine	**SJS/TEN (HLA-B*1502 in Asians)**, **hyponatraemia (SIADH)**, aplastic anaemia/agranulocytosis, diplopia, **enzyme inducer**, worsens absence/myoclonic
Ethosuximide	GI upset, hiccups, SLE-like, blood dyscrasia
Phenobarbitone	Sedation, **hyperactivity/behavioural problems in children**, cognitive dulling
Lamotrigine	**Rash/SJS (↑ with valproate – titrate slowly)**
Levetiracetam	**Behavioural (irritability, aggression, psychosis)**
Topiramate	**Weight loss, renal stones, metabolic acidosis, cognitive slowing ("dopamax"), oligohydrosis, glaucoma**
Vigabatrin	**Irreversible concentric visual field constriction**
ACTH/steroids	Hypertension, infection, irritability, hyperglycaemia, cardiomyopathy
Clobazam/benzodiazepines	Sedation, drooling, tolerance

- **Ketogenic diet** – **GLUT1 deficiency (DOC), PDH deficiency (DOC)**, refractory epilepsy (Dravet, LGS, infantile spasms).
- **Stopping AEDs**: after **2 seizure-free years** (except JME, structural lesions), taper over 2–3 months.
- **Epilepsy surgery**: mesial temporal sclerosis, focal cortical dysplasia, hemimegalencephaly; **VNS**, corpus callosotomy.

21.4 Status Epilepticus (SE)

- **Operational definition: continuous seizure ≥ 5 min, or ≥ 2 seizures without return to baseline** (t1 = 5 min – treat; t2 = 30 min – long-term damage).
- **Refractory SE** – failure of benzodiazepine + 1 second-line drug; **super-refractory** – persists ≥ 24 h after anaesthesia.



Algorithm for convulsive status epilepticus in children

Time	Action
0–5 min	ABC, oxygen, position, **check glucose (if < 60 mg/dL → 10% dextrose 2–5 mL/kg)**, IV access, labs (electrolytes, Ca, Mg, AED levels, toxicology, cultures)
5–10 min – 1st line	**IV lorazepam 0.1 mg/kg (max 4 mg)** or **IV diazepam 0.2–0.3 mg/kg**; no IV → **buccal/intranasal midazolam 0.2–0.3 mg/kg**, IM midazolam 0.2 mg/kg, rectal diazepam 0.5 mg/kg; **repeat once after 5–10 min**
10–30 min – 2nd line	**IV levetiracetam 40–60 mg/kg** or **fosphenytoin/phenytoin 20 mg/kg (rate ≤ 1 mg/kg/min, cardiac monitoring)** or **valproate 20–40 mg/kg** (avoid < 2 y/liver disease/mitochondrial) or **phenobarbitone 20 mg/kg** (ESETT trial – equal efficacy of LEV, fPHT, VPA)
> 30 min – refractory	**ICU; continuous midazolam infusion (0.1–0.2 mg/kg bolus, 1–24 µg/kg/min)**, thiopentone/ pentobarbitone, propofol (caution in children – propofol infusion syndrome), ketamine; **continuous EEG**

- Causes in children: **febrile status (MC)**, CNS infection (meningitis/encephalitis), **non-compliance/AED withdrawal**, metabolic (hypoglycaemia, hyponatraemia, hypocalcaemia), trauma, toxins, structural; pyridoxine (infants).

21.5 Cerebral Palsy (CP)

- **Group of permanent disorders of movement & posture, causing activity limitation, attributed to non-progressive disturbances in the developing fetal/infant brain**; often with sensation, cognition, communication, perception, behaviour, epilepsy & musculoskeletal problems.
- **MCC of chronic childhood motor disability**; prevalence 2–3/1000.
- **Risk factors**: **prematurity (most important – PVL, IVH)**, **LBW**, **birth asphyxia/HIE (term – ~10%)**, **kernicterus (dyskinetic)**, intrauterine infections (TORCH), multiple gestation, chorioamnionitis, neonatal stroke, meningitis, hypoglycaemia; **most causes are antenatal**.

Type	%	Lesion / cause	Features
Spastic (pyramidal) – MC (70–80%)		Corticospinal tracts (UMN)	**Hypertonia (clasp-knife), hyperreflexia, clonus, Babinski**, persistent primitive reflexes, scissoring, toe-walking, contractures
– **Spastic diplegia**	MC in **preterm**	**PVL**	**Legs >> arms**; scissoring gait; **normal intelligence usually**, strabismus
– **Spastic hemiplegia**	MC in term (some series)	**Perinatal arterial stroke (MCA)**, porencephaly	**One side (arm > leg)**, **early hand preference (< 12–18 m)**, **focal seizures**, growth arrest of affected limbs, cortical sensory loss
– **Spastic quadriplegia (tetraplegia)**	Most severe	**Severe HIE, multicystic encephalomalacia**, infections	All 4 limbs, **severe ID, epilepsy, microcephaly, pseudobulbar palsy (feeding problems, drooling), aspiration**
Dyskinetic (extrapyramidal) – 10–15%		**Basal ganglia – kernicterus (choreoathetoid), acute profound HIE (dystonic)**	**Involuntary movements (athetosis, chorea, dystonia)**, fluctuating tone, **dysarthria, drooling**; **intelligence often preserved**; **SNHL & upward gaze palsy (kernicterus)**
Ataxic – 5–10%		Cerebellum (genetic, malformations)	Hypotonia → wide-based gait, intention tremor, nystagmus
Mixed			Spastic + dyskinetic

- **Early signs**: **delayed motor milestones**, **persistence of primitive reflexes (Moro, ATNR beyond 6 m)**, early handedness, abnormal tone (**initial hypotonia → later spasticity**), poor head control, feeding difficulty, irritability, **fisting with adducted thumb ("cortical thumb")** beyond 4 months.
- Early diagnosis (< 5 months corrected): **General Movements Assessment (absent fidgety movements)**, **HINE (Hammersmith Infant Neurological Examination)**, **MRI (IOC for aetiology – abnormal in ~85%)**.
- **GMFCS (Gross Motor Function Classification System) levels I–V** – functional severity; MACS for hand.
- **Associated problems**: **ID (~50%), epilepsy (30–50% – most in quadriplegia/hemiplegia)**, visual (strabismus – MC, cortical visual impairment, ROP), hearing loss, speech, feeding/GERD, malnutrition, drooling, **hip subluxation/dislocation (spastic – surveillance X-ray)**, scoliosis, constipation, osteopenia, behavioural.

MANAGEMENT – MULTIDISCIPLINARY

Physiotherapy (mainstay), occupational & speech therapy, **orthoses (AFO)**, assistive devices; **spasticity – oral baclofen, diazepam, tizanidine; focal – botulinum toxin A injections (DOC for focal spasticity)**; **intrathecal baclofen**; **selective dorsal rhizotomy (spastic diplegia)**; orthopaedic surgery (tendon lengthening, hip/derotation osteotomy); **dystonia – trihexyphenidyl, levodopa, deep brain stimulation**; drooling – glycopyrrolate; nutrition (gastrostomy); AEDs; special education. **CP itself is non-progressive** (manifestations change with growth).

21.6 CNS Infections

CSF analysis

	Normal	Bacterial meningitis	Viral (aseptic)	Tuberculous meningitis	Fungal
Appearance	Clear	**Turbid**	Clear	Clear/opalescent, **cobweb (pellicle) on standing**	Clear
Cells (/mm³)	**< 5 (neonate < 20–30)**	**↑↑ (> 1000), neutrophils**	10–500, **lymphocytes** (early PMN)	**50–500, lymphocytes** (early neutrophils)	Lymphocytes
Protein (mg/dL)	15–40 (neonate up to 150)	**↑↑ (> 100)**	Normal/mild ↑	**↑↑ (100–500+)**	↑
Glucose	**> 60% of blood (> 40 mg/dL)**	**↓↓ (< 40% of blood)**	**Normal**	**↓**	↓
Others		Gram stain, culture, latex agglutination, PCR	**PCR (enterovirus, HSV)**	**ADA > 10 U/L, CBNAAT, AFB stain/ culture**	India ink, cryptococcal antigen

- **Partially treated bacterial meningitis** – mimics TBM/viral (lymphocytes ↑, culture negative).
- **LP contraindications**: **raised ICP signs (papilloedema, focal deficits, GCS ≤ 8, abnormal posturing, irregular breathing)**, cardiorespiratory instability, coagulopathy/platelets < 50,000, skin infection at site → **give antibiotics first**, CT later.

Encephalitis

- **HSV-1 (MC sporadic fatal encephalitis)** – **fever, altered sensorium, focal seizures, behavioural change; temporal lobe involvement (MRI – T2 hyperintensity, EEG – PLEDs/periodic lateralised discharges)**; **CSF HSV PCR – IOC**; **IV acyclovir 10 mg/kg 8-hourly (20 mg/kg in neonates) × 14–21 days – start empirically**.
- **Japanese encephalitis** (India – MC epidemic encephalitis): **thalamic & basal ganglia (bilateral) lesions**, parkinsonism, dystonia; Culex mosquito; no antiviral; vaccine.
- **Acute encephalitis syndrome (AES)** – India (Muzaffarpur – **litchi-associated hypoglycaemic encephalopathy (methylene cyclopropyl glycine)**, Gorakhpur – JE, scrub typhus); Nipah (Kerala).
- **Autoimmune encephalitis – anti-NMDA receptor** (MC autoimmune; children/young women; **psychiatric symptoms, dyskinesias (orofacial), seizures, autonomic instability, ↓ consciousness**; ovarian teratoma in older girls; Rx steroids, IVIG, PLEX, rituximab).
- **ADEM (acute disseminated encephalomyelitis)**: **post-infectious/post-vaccination (1–3 wk)**; multifocal demyelination – **encephalopathy + polyfocal deficits**; MRI – multiple large asymmetric white & grey-matter (thalami) lesions; **monophasic**; Rx **IV methylprednisolone**, IVIG; MOG-antibody associated. **Paediatric MS** – adolescents, relapsing.

Neurocysticercosis (NCC)

- **Taenia solium** larvae (**eggs ingested – faeco-oral from human carrier**; not by eating pork – that causes intestinal taeniasis); **MCC of acquired epilepsy in India**; **focal seizures (MC presentation)**, headache, raised ICP (racemose/ventricular – hydrocephalus).
- **Imaging**: **CT/MRI – single small enhancing lesion (SSECTL – ring or disc < 2 cm, eccentric scolex – "dot" sign/"hole-with-dot")**; stages – vesicular (cyst with scolex), colloidal (ring-enhancing with oedema), granular nodular, **calcified (nodular calcification)**; **MRI better for scolex/intraventricular; CT better for calcification**. **Starry-sky appearance** in heavy infestation (cysticercal encephalitis). EITB serology.
- **Rx**: **antiepileptics (carbamazepine/levetiracetam) for 6–12 months (≥ 6 months seizure-free after lesion resolution)**; **albendazole 15 mg/kg/day × 7–28 days (cysticidal) + steroids (prednisolone/dexamethasone –**

started before, to prevent inflammatory reaction) for viable/colloidal cysts; **no cysticidal therapy for calcified lesions or cysticercal encephalitis (raised ICP)**; **fundoscopy before albendazole (ocular cyst)**; praziquantel combination for multiple; surgery/shunt for hydrocephalus.

Tuberculous meningitis (TBM)

- Commonest in **< 5 y** (within 6–12 months of primary infection), malnourished, unvaccinated (BCG protects).
- **British MRC staging**: **Stage I – conscious, no focal deficits (non-specific: fever, irritability, vomiting, personality change, apathy)**; **Stage II – drowsy, meningeal signs, cranial nerve palsy (VI MC, III, VII)**, **focal deficits (hemiparesis – vasculitis of MCA/lenticulostriate arteries)**; **Stage III – coma, decerebrate/decorticate posture**, dense deficits.
- **Imaging (CECT/MRI)**: **basal exudates (enhancing basal meninges – most characteristic)**, **hydrocephalus (MC – communicating)**, **infarcts (basal ganglia/internal capsule)**, tuberculomas.
- CSF: as above; **CBNAAT**, culture (MGIT).
- Rx: **2HRZE + 10HRE + steroids (dexamethasone/prednisolone 2–4 mg/kg/day for 4 weeks, tapered over 4 weeks)** – ↓ mortality; **VP shunt/ETV** for obstructive hydrocephalus (communicating – acetazolamide/furosemide trial).
- Sequelae: hydrocephalus, ID, hemiparesis, blindness (optic atrophy/chiasmal arachnoiditis), deafness, endocrine (DI), seizures.

Brain abscess

- **Cyanotic CHD (TOF – > 2 y, MC predisposing in children)**, chronic otitis media/mastoiditis (**temporal lobe/cerebellum**), sinusitis (frontal), penetrating trauma, meningitis (neonates – Citrobacter); streptococci (S. milleri/anginosus), anaerobes, S. aureus.
- **Fever, headache, focal deficit, seizures, vomiting** (triad present in < 50%); **CECT/MRI – ring-enhancing lesion with surrounding oedema; DWI restriction (pus)**; **LP contraindicated**.
- Rx: **IV antibiotics (ceftriaxone + metronidazole ± vancomycin) 4–8 weeks**; aspiration/excision if > 2.5 cm, mass effect.

21.7 Hydrocephalus

- **Excess CSF with ventricular dilatation**; CSF produced by **choroid plexus (~500 mL/day, 0.35 mL/min)**, absorbed by arachnoid villi.
- **Obstructive (non-communicating)** – block within ventricular system: **aqueductal stenosis (MCC congenital – X-linked L1CAM with adducted thumbs)**, **Dandy–Walker**, **Chiari II**, tumours (posterior fossa), **post-haemorrhagic/post-infectious ventriculitis**, vein of Galen malformation, TBM exudates. **Communicating** – absorption defect: **post-meningitis (TBM, pyogenic), post-SAH/IVH**, choroid plexus papilloma (overproduction).
- **Infants**: **rapidly increasing head circumference (crossing centiles – earliest sign)**, **wide bulging anterior fontanelle, separated sutures (cracked-pot/MacEwen sign)**, **dilated scalp veins**, **"setting-sun" sign (upward gaze palsy – pressure on tectum)**, irritability, vomiting, poor feeding, spasticity of lower limbs, **Parinaud**; papilloedema rare (open sutures). Older: headache (morning), vomiting, papilloedema, **VI nerve palsy (false localising)**, **Cushing triad (bradycardia, hypertension, irregular respiration – late)**.
- Investigations: **cranial USG (open fontanelle)**, CT/MRI (IOC – cause).
- **Rx**: **ventriculoperitoneal (VP) shunt (MC)**; **endoscopic third ventriculostomy (ETV) – for obstructive (aqueductal stenosis) hydrocephalus in children > 6 months**; acetazolamide/furosemide (temporary); shunt complications – **obstruction (MC – proximal), infection (S. epidermidis – MC organism, first 6 months)**, overdrainage (slit ventricle), subdural haematoma, peritoneal pseudocyst, CSF ascites.

Malformation	Features
Arnold–Chiari II (Chiari II)	**Herniation of cerebellar vermis, 4th ventricle & medulla through foramen magnum**; **almost always with lumbosacral myelomeningocele**; hydrocephalus (90%), **tectal beaking**, syringomyelia, **stridor (vocal cord palsy), apnoea, dysphagia (brainstem)**; **"lemon" (frontal bone scalloping) & "banana" (cerebellum) signs on prenatal USG**

Malformation	Features
Chiari I	**Cerebellar tonsils > 5 mm below foramen magnum**; older children/adults – occipital headache (cough/Valsalva), **syringomyelia (cape-like dissociated sensory loss)**, scoliosis; no MMC
Dandy–Walker malformation	**Cystic dilatation of 4th ventricle + hypoplasia/agenesis of cerebellar vermis + enlarged posterior fossa with elevated tentorium (torcular–lambdoid inversion)**; hydrocephalus (90%), ataxia; associated agenesis of corpus callosum
Joubert syndrome	**"Molar tooth sign" (midbrain)**, vermian hypoplasia, **episodic hyperpnoea/apnoea**, abnormal eye movements (oculomotor apraxia), hypotonia, retinal dystrophy, renal cysts (nephronophthisis)
Vein of Galen malformation	**High-output cardiac failure in neonate**, cranial bruit, hydrocephalus; Rx endovascular embolisation
Holoprosencephaly	**Failure of forebrain (prosencephalon) cleavage**; alobar (single ventricle, **cyclopia/cebocephaly, single central incisor, proboscis**) → semilobar → lobar; **trisomy 13 (MC chromosomal cause)**, **SHH mutations**, maternal diabetes
Lissencephaly	Smooth brain (agyria/pachygyria) – **Miller–Dieker (17p13, LIS1)**, DCX (X-linked – double cortex in girls); infantile spasms, ID
Schizencephaly	Full-thickness clefts lined by grey matter
Hydranencephaly	Cerebral hemispheres replaced by CSF sac (**bilateral ICA occlusion**) – **transillumination positive**; brainstem intact (normal-appearing newborn)
Agenesis of corpus callosum	Aicardi syndrome (**girls – infantile spasms + chorioretinal lacunae**), "**racing car**/steer-horn" ventricles
Benign external hydrocephalus	Macrocephaly in infants with ↑ subarachnoid spaces, normal development; resolves

21.8 Neural Tube Defects (NTD)

- **Failure of neural tube closure at 3–4 weeks of gestation (day 24 – anterior neuropore; day 26–28 – posterior neuropore)**; **multifactorial**; risk factors – **folate deficiency**, previous NTD (recurrence 2–3%, 10% with two), **valproate/carbamazepine**, **maternal diabetes, obesity**, hyperthermia, MTHFR polymorphism.
- **Prevention**: **folic acid 0.4 mg/day from ≥ 1 month before conception till 12 weeks** (all women); **4 mg/day (high-risk – previous NTD, AED use)** – ↓ NTD by ~70%.
- **Prenatal diagnosis**: **↑ maternal serum AFP (15–20 wk)**, **↑ amniotic AFP & acetylcholinesterase**, **USG – lemon & banana signs**, anencephaly – **"frog-eye" appearance**.

Type	Description
Anencephaly	Absent cranial vault & cerebral hemispheres; **polyhydramnios** (impaired fetal swallowing); incompatible with life; **MC NTD in many series**
Craniorachischisis	Complete open neural tube – worst
Encephalocele	Herniation of brain & meninges through skull defect – **occipital (MC in West)**, frontal/nasal (Asia); **Meckel–Gruber syndrome** (occipital encephalocele + polycystic kidneys + polydactyly)
Myelomeningocele (spina bifida aperta/cystica)	**Sac with meninges + spinal cord/nerve roots**; **lumbosacral (MC – 75%)**; **flaccid paralysis below lesion, sensory loss, neurogenic bladder & bowel, clubfoot, hip dislocation**; **Chiari II + hydrocephalus (80–90%)**, **latex allergy**, tethered cord
Meningocele	**Sac with only meninges & CSF (no neural tissue)** – usually no neurological deficit; good prognosis
Spina bifida occulta	**Failure of vertebral arch fusion without herniation (L5–S1)**; **cutaneous markers – tuft of hair (hypertrichosis), dimple, lipoma, haemangioma, dermal sinus**; tethered cord
Iniencephaly	Retroflexion of head with occipital & cervical defect

MYELOMENINGOCELE – INITIAL CARE

Cover sac with sterile saline-soaked gauze & non-adherent dressing (keep moist), nurse prone, avoid latex, IV antibiotics; **surgical closure within 24–72 h** (↓ infection); **VP shunt** for hydrocephalus; **prenatal (fetal) surgery before 26 weeks – MOMS trial – ↓ shunt need, better motor function**; **clean intermittent catheterisation + oxybutynin**, bowel programme; orthopaedic care; lifelong follow-up (tethered cord, shunt malfunction, scoliosis).

- **Level of lesion & function**: **thoracic – no hip flexion (wheelchair)**; **L1–L2 – hip flexion**; **L3–L4 – knee extension (quadriceps) – walk with braces**; **L5–S1 – ankle function, community walkers**; **S2–S4 – bladder/bowel only**.

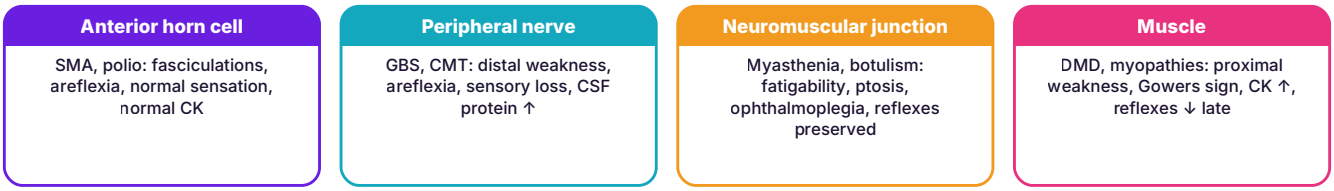
21.9 Neurodegenerative Disorders

- **Clue: regression (loss of previously acquired milestones)** after a period of normal development.

Grey-matter disease	White-matter disease (leukodystrophy)
Early seizures, dementia/cognitive decline, visual loss (retinal); late motor signs	**Early motor signs – spasticity, ataxia, pyramidal signs**, optic atrophy; seizures & dementia late
Examples: **Tay–Sachs, NCL (Batten), Rett, Alpers (POLG), Niemann–Pick, Gaucher type 2/3**, SSPE	Examples: **MLD, Krabbe, X-ALD, Canavan, Alexander, Pelizaeus–Merzbacher, vanishing white matter**

Disorder	Key points
Rett syndrome (MECP2, X-linked dominant – girls)	**Normal development 6–18 months → regression: loss of purposeful hand use → stereotypic hand-wringing/washing/clapping/mouthing**, loss of speech, **acquired microcephaly (deceleration of head growth)**, autistic features, **breathing abnormalities (hyperventilation, breath-holding while awake)**, gait apraxia, seizures, scoliosis, bruxism
Neuronal ceroid lipofuscinoses (Batten – CLN genes)	**Visual loss (retinal degeneration), seizures (myoclonic), dementia**; **fingerprint/curvilinear inclusions on EM**; CLN2 – **cerliponase alfa** (ERT)
Canavan (aspartoacylase, ASPA)	**Macrocephaly**, hypotonia → spasticity, blindness; **↑ urinary N-acetylaspartic acid (NAA)**, MRS – ↑ NAA peak; spongy degeneration
Alexander (GFAP, AD)	**Macrocephaly**, frontal-predominant leukodystrophy, **Rosenthal fibres**
Krabbe	Infancy – extreme irritability, hypertonia, **optic atrophy**, peripheral neuropathy, **↑ CSF protein**, **globoid cells**; HSCT pre-symptomatic
MLD (arylsulfatase A)	Late-infantile (MC) – gait disturbance, regression, neuropathy (↓ NCV); ↑ urinary sulfatides; gene therapy (atidarsagene)
X-ALD	Boys 4–10 y, ADHD-like → regression; **adrenal insufficiency**; ↑ VLCFA; parieto-occipital white matter
Pelizaeus–Merzbacher (PLP1, X-linked)	**Pendular nystagmus in early infancy**, head titubation, hypomyelination
Subacute sclerosing panencephalitis (SSPE)	Post-measles; **personality change → myoclonic jerks → dementia → vegetative**; EEG **periodic complexes (Radermecker – burst every 4–15 s)**; **↑ CSF measles antibody**
Wilson disease	Hepatic + neuropsychiatric (adolescent)
Huntington (juvenile, Westphal variant)	**Paternal transmission (CAG anticipation)**, rigidity > chorea, seizures
Friedreich ataxia (FXN, GAA repeat, AR)	**MC hereditary ataxia**; onset 5–15 y; **progressive ataxia, areflexia, extensor plantars, loss of position/vibration sense**, dysarthria, **pes cavus, scoliosis, HCM (MCC death), diabetes**
Ataxia telangiectasia	See Immunology
Hallervorden–Spatz (PKAN – PANK2)	Dystonia, rigidity, pigmentary retinopathy; **"eye-of-the-tiger" sign on MRI (globus pallidus)**
Leigh	Mitochondrial; **bilateral basal ganglia & brainstem lesions**; respiratory abnormalities

21.10 Neuromuscular Disorders



Localising weakness – anterior horn to muscle

Level	Disorder	Clues
Anterior horn cell	**SMA (SMN1)**, **poliomyelitis** (asymmetric, fever, CSF pleocytosis)	**Tongue fasciculations**, areflexia, normal sensation, normal CK
Peripheral nerve	**GBS**, CMT (hereditary motor-sensory neuropathy), diphtheritic, toxic	Distal weakness, areflexia, sensory signs
Neuromuscular junction	**Myasthenia gravis (juvenile, neonatal transient, congenital myasthenic syndromes)**, **infant botulism** (honey – constipation, ptosis, descending paralysis), tick paralysis, organophosphates	**Fatigable weakness, ptosis, ophthalmoplegia**; normal reflexes
Muscle	**DMD/BMD**, congenital myopathies, **myotonic dystrophy**, JDM, Pompe	**Proximal weakness, Gowers' sign**, ↑ CK (dystrophies)

Guillain–Barré syndrome (GBS)

- **MCC of acute flaccid paralysis (AFP) in children after polio eradication**; post-infectious (**Campylobacter jejuni – MC; AMAN axonal variant, anti-GM1**), CMV, EBV, Mycoplasma, Zika, vaccines (rare).
- **Ascending symmetric flaccid weakness**, **areflexia/hyporeflexia**, mild sensory symptoms (pain – back/legs common in children), **autonomic dysfunction (arrhythmias, BP fluctuation)**, **respiratory failure (~15–20%)**, facial/bulbar palsy; nadir by 2–4 weeks.
- **CSF – albuminocytological dissociation (↑ protein, normal cells < 10) – after 1st week**; **NCS – demyelinating (AIDP – MC in West) or axonal (AMAN – common in Asia/India)**.
- **Miller Fisher variant**: **ophthalmoplegia, ataxia, areflexia – anti-GQ1b**.
- **Rx: IVIG 2 g/kg over 2–5 days or plasmapheresis** (equally effective; **steroids NOT useful**); monitor **vital capacity (< 15–20 mL/kg → ventilate), single-breath count**; supportive.

Others

- **Juvenile myasthenia gravis** – anti-AChR antibodies; **ptosis, diplopia, fatigable bulbar/limb weakness**; **ice-pack test, neostigmine/edrophonium test**, repetitive nerve stimulation (decrement), single-fibre EMG (most sensitive); Rx **pyridostigmine**, steroids, thymectomy. **Transient neonatal MG** – baby of myasthenic mother (10–15%), weak suck & cry, hypotonia, resolves in weeks.
- **Infant botulism** – **honey/soil spores colonise gut** (< 12 m): **constipation (first sign), poor feeding, weak cry, ptosis, descending symmetric paralysis, dilated sluggish pupils**; Rx **BabyBIG (human botulism immune globulin)**; avoid aminoglycosides.
- **Charcot–Marie–Tooth (HMSN I – PMP22 duplication, AD)** – **distal wasting ("inverted champagne bottle"), pes cavus, hammer toes, areflexia, thickened nerves**, slow NCV.
- **Floppy infant (hypotonia)**: **central (MC – 60–80%)** – HIE, chromosomal (**Down, Prader–Willi**), brain malformations, IEM, hypothyroidism, sepsis → **hypotonia without significant weakness, normal/brisk reflexes, dysmorphism, seizures, ID**; **peripheral** – SMA, congenital myopathies, myotonic dystrophy (mother), myasthenia, botulism → **hypotonia with marked weakness, areflexia, frog-leg posture, poor antigravity movements, alert face**.
- **Transverse myelitis**, **acute flaccid myelitis (EV-D68 – grey matter of cord)**, spinal cord compression – urgent MRI.

- **Bell's palsy** – LMN facial palsy; steroids (within 72 h); good prognosis; exclude **hypertension (CoA), Lyme, Ramsay Hunt (VZV), leukaemia, otitis media**.

21.11 Headache, Stroke & Raised ICP

- **Migraine** – MC primary headache in children; **shorter (2–72 h, 1 h in children), often bilateral (frontal/temporal)**, nausea/vomiting, photophobia, family history; **migraine equivalents – cyclical vomiting, abdominal migraine, benign paroxysmal vertigo, benign paroxysmal torticollis**; Rx **ibuprofen/paracetamol, triptans (sumatriptan nasal ≥ 12 y; rizatriptan ≥ 6 y)**; prophylaxis – **flunarizine (India)**, propranolol, topiramate, amitriptyline, cyproheptadine.
- **Red flags**: **morning headache with vomiting, waking from sleep, progressive pattern, focal signs/papilloedema, change in personality, age < 3 y, occipital headache, worsened by cough/Valsalva** → neuroimaging.
- **Idiopathic intracranial hypertension (pseudotumor cerebri)**: headache, papilloedema, **VI nerve palsy**, normal imaging, **opening pressure > 28 cm H_2O (> 25 non-sedated)**; obesity, adolescent girls, **drugs – tetracyclines, vitamin A, steroid withdrawal, GH, OCP**; Rx **acetazolamide**, weight loss, LP; risk – visual loss.
- **Paediatric stroke**: arterial ischaemic – **sickle cell disease (MC)**, cyanotic CHD, **moyamoya ("puff of smoke" on angiography – progressive stenosis of terminal ICA; Down, NF-1, sickle cell, radiation)**, post-varicella arteriopathy (focal cerebral arteriopathy), dissection, prothrombotic states, vasculitis; **cerebral venous sinus thrombosis** – dehydration, nephrotic, otitis/mastoiditis, OCP; **haemorrhagic – AVM (MC)**.
- **Raised ICP management**: head end elevation 30°, neutral neck, avoid hypotonic fluids, **osmotherapy – 3% saline (2–5 mL/kg) or mannitol 0.25–1 g/kg**, brief hyperventilation ($PaCO_2$ 30–35) only for herniation, sedation, treat fever/seizures, neurosurgery; **Cushing triad = herniation**.

21.12 Movement Disorders & Others

- **Sydenham chorea** (rheumatic) – see Cardiology; **tics/Tourette** – see Behaviour.
- **Opsoclonus–myoclonus–ataxia** – **neuroblastoma** (paraneoplastic), post-viral.
- **Acute cerebellar ataxia** – **post-varicella (MC)**, other viruses; 1–4 y, **sudden truncal ataxia**, nystagmus, normal consciousness; self-limiting (weeks); exclude **posterior fossa tumour, drug ingestion (phenytoin, benzodiazepines, alcohol), ADEM, Miller Fisher, opsoclonus-myoclonus**.
- **Benign paroxysmal vertigo** – 2–4 y; sudden brief imbalance, nystagmus, no LOC; migraine precursor.
- **Spasmus nutans** – triad of **pendular nystagmus, head nodding, torticollis** (4–12 m); benign (exclude optic glioma).
- **Infantile tremor syndrome** – see Nutrition (B12).
- **Head injury in children** – GCS paediatric; **epidural haematoma (lucid interval, middle meningeal artery – biconvex/lens-shaped)**; **subdural (crescentic – shaken baby)**; **growing skull fracture (leptomeningeal cyst)**; **ping-pong (depressed) fracture of infants**.

Practice MCQs · Pediatric Neurology

264 questions · correct option highlighted in green · cover the answer strip to self-test

Seizures & Epilepsy

1. Which is the best marker for NTD?

A. **aFP**
B. hCG
C. Pseudocholinesterase
D. Inhibin-A

Ans A FP • Prenatal screening of maternal serum for AFP in the 16th-18th wk of gestation is an effective method for identifying pregnancies at risk for fetuses with neural tube defects in utero.

2. A 7-year-old boy presents with a right-sided hemangioma and left-sided focal seizures. The most likely diagnosis is:

A. Neurofibromatosis
B. Incontinentia pigmenti
C. Hypermelanosis of Ito
D. **Sturge-Weber syndrome**

Ans D Sturge-Weber Syndrome (SWS)
• Angiomas involving leptomeninges & skin of face, typically in ophthalmic & maxillary divisions distributions of trigeminal nerve.
• The hallmark of SWS is a facial cutaneous venous dilation, also referred to as a nevus flammeus or port-wine stain.
• Contralateral focal seizures, calcification of cerebral cortex and glaucoma on same side as skin lesions are seen.

3. A 10-year-old male child was presented to the pediatrician for evaluation of a seizure disorder. On examination a vascular plaque was found along the ophthalmic and maxillary divisions of the trigeminal nerve. The mother informed the pediatrician that the lesion was present since birth and there was no change in morphology. The most likely possibility is:

A. **Sturge-Weber syndrome**
B. Infantile hemangioma
C. Congenital hemangioma
D. Proteus syndrome

Ans A Sturge-Weber syndrome , Ghai p

4. A 2-year-old child is brought by parents with history of seizures and developmental delay. He has multiple hypopigmented macules over the back. What is the most probable diagnosis:

A. Neurofibromatosis type I
B. **Tuberous sclerosis**
C. Sturge Weber syndrome
D. Linear sebacous nevus syndrome

Ans B Ghai p 586 History of seizures and developmental delay along with multiple hypopigmented macules over the back suggests Tuberous sclerosis.

5. True about febrile seizures:

A. **Most common cause of seizure in childhood**
B. May persist beyond 5 years if associated with atypical feature
C. Family h/o febrile seizure is a risk factor
D. Febrile seizure is always associated with neurologically abnormal child
E. Anticonvulsants are the treatment of choice

Ans A b. May persist beyond 5 years if associated with atypical feature, c. Family h/o febrile seizure is a risk factor Febrile seizures are the most common cause of seizure in under 5 children. Long-term antiepileptic therapy is not required for simple febrile seizures.

6. Midazolam given after how much minutes after continuous febrile seizures?

A. 10 minutes of febrile seizures (midazolam nasal spray)
B. **5 minutes**
C. 3 minutes
D. 1 minute

Ans B If the seizure lasts for longer than 5 min, acute treatment with diazepam, lorazepam, or midazolam is needed.

7. What is correct about febrile seizures:

A. Causes developmental delay
B. Focal deficits
C. Repeated seizures in 24 hours
D. **Does not need long term antiepileptics**

Ans D Does not need long-term antiepileptics Long-term antiepileptic therapy is not required for simple febrile seizures.

8. Which of the following is not associated with increase in the risk of epilepsy in a child with febrile seizures:

A. Development delay
B. **Late age of onset**
C. Complex partial seizures
D. Family history positive for epilepsy

Ans B

9. Management of typical febrile seizures include all except:

A. Sponging
B. Paracetamol or ibuprofen
C. Intermittent diazepam
D. **Prophylactic phenobarbitone**

Ans D Measures to lower temperature (in a child with fever) like paracetamol and sponging and intermittent prophylaxis with Diazepam or Clobazam are used in the treatment of febrile seizures

10. An adolescent school girl complaints of dropping objects from hands, it gets precipitated during morning and during exams. There is no history of loss of consciousness and her cousin sister has been diagnosed with epilepsy. EEG was done and was suggestive of epileptic spikes. What is the diagnosis?

A. **Juvenile myoclonic epilepsy**
B. Atypical absence
C. Choreo–athetoid epilepsy
D. Centrotemporal spikes

Ans A Juvenile myoclonic epilepsy (Janz syndrome)
• Most common generalized epilepsy in young adults
• Starts in early adolescence with 1 or more of:
• – Myoclonic jerks in morning, causing patient to drop things;
• – Generalized tonic–clonic seizures upon awakening;
• – Juvenile absences.
• Sleep deprivation & photic stimulation can act as precipitants.
• EEG shows generalized 4-5 Hz polyspike& slow- wave discharges. Atypical absence seizures
• Less abrupt onset and offset of loss of awareness

11. Benign neonatal seizures is:

A. **Common in late preterms & terms**
B. Followed by febrile seizures
C. Followed by infantile spasms
D. Prolonged seizures
E. Also known as 'fifth day fits'

Ans A e. Also known as 'fifth day fits' fo Benign neonatal seizures This is a benign electroclinical syndrome that is seen in neonates. It is also referred to as fifth day fits or benign idiopathic neonatal convulsions. These seizures occur in term and later preterm infants born after an uneventful pregnancy, labor and delivery. Brief, but repetitive seizures lasting for 1–2 minutes seen.

12. Which of the following is not useful in the management of status epilepticus?

A. Lorazepam
B. Phenytoin
C. Phenobarbitone
D. **Carbamazepine**

Ans D

13. Which of the following is True regarding absence seizures?

A. Attack lasts for 60 to 90 seconds
B. **Hyperventilation often precipitates an attack**
C. Onset of disease in the first year of life
D. Attack followed by postictal confusion

Ans B Typical absence seizures
• • Usually start at 5–8 yr of age; They do not have an aura, usually last for only a few seconds and are accompanied by eyelid flutter or upward rolling of eyes and simple automatisms like lip-smacking or picking at clothing
• • Absence seizures do not have a postictal periodQ and there is immediate resumption of what patient was doing
• • Hyperventilation for 3–5 min can precipitate seizures & 3 Hz spike–and–slow-wave discharges

14. Absence seizures are seen in?

A. Grand mal epilepsy
B. Myoclonic epilepsy
C. **Petit mal epilepsy**
D. Hyperkinetic child

Ans C Absence seizures are sometimes referred to as petit mal seizures (from the French for "little illness").

15. Which of the following drugs is not used in Juvenile Myoclonic Epilepsy?

A. Topiramate
B. Zonisamide
C. **Carbamazepine**
D. Valproate

Ans C Drugs used for treatment of JME are Valproate (drug of choice), Levetiracetam; Lamotrigine, Topiramate, Zonisamide.

16. Treatment of rolandic epilepsy is:

A. Phenytoin
B. Lamotrigine
C. **Carbamazepine**
D. ACTH

Ans C Partial idiopathic epilepsy with rolandic spikes
• • Also called benign epilepsy with centrotemporal spikes
• • They are seizures with falling asleep or on awakening; focal sharp waves with centrotemporal location on EEG.
• • Carbamazepine is the most frequently used 1st line drug, but valproate, phenytoin, gabapentin and levetiracetam have been found effective as well.

17. Drug of choice for juvenile myoclonic epilepsy is:

A. Phenytoin
B. Lamotrigine
C. **Valproate**
D. Zonisamide

Ans C

18. Initial drug of choice in a child with status epilepticus:

A. **Lorazepam**
B. Phenobarbitone
C. Valproate
D. Phenytoin

Ans A Initial drug given in Status epilepticus in a child is a short acting Benzodiazepine like Lorazepam or Midazolam, to abort the seizures

19. A 7-year-old girl is easily distracted in class and exhibits poor scholastic performance. Seizures are precipitated by hyperventilation, what is the diagnosis?

A. Myoclonic seizures
B. **Absence seizures**
C. Atonic seizures
D. Myoclonia

Ans B This is a typical description of Absence seizures.

20. A school going boy was noted with vacant stare several times a day. There was no history of fever, seizures and neurological deterioration. What is the diagnosis?

A. Atonic seizures
B. Absence seizures
C. Myoclonic seizures
D. School phobia

Ans B Absence seizures with

21. Cause of status epilepticus in a child:

A. Hypernatremia
B. Hyponatremia
C. Hyperkalemia
D. Hypokalemia
E. Hypocalcemia

Ans B and e. Hypocalcemia Etiology of Status epilepticus in children:
• Metabolic disturbances: hypoglycemia; hypocalcemia; hyponatremia; hypomagnesemia;
• Drug intoxication (e.g., tricyclic antidepressants) in children and drug and alcohol abuse in adolescents;
• Drug withdrawal or overdose in patients on antiepileptic drugs;
• Acute head trauma; Infections: Encephalitis; meningitis;
• Autoimmune encephalitis (such as anti-NMDA receptor and anti–voltage-gated K+ channel antibody syndromes);

22. Not a cause of neonatal seizures:

A. Pyridoxine deficiency
B. Hypokalemia
C. Hypoxia
D. None

Ans B Pyridoxine deficiency and pyridoxal-5-phosphate deficiency, Hypoxia, Hypocalcemia, Hypoglycemia, Hypomagnesemia, Hypo- natremia and hypernatremia may precipitate neonatal seizure. Hypokalemia usually does not cause neonatal seizure.

23. True about Juvenile myoclonic epilepsy:

A. Drug of choice is sodium valproate
B. Mental retardation
C. Seizure can develop
D. Neurological examination abnormal
E. Life long treatment needed

Ans A c. Seizure can develop, d. Neurological examination abnormal, e. Life long treatment needed Juvenile myoclonic epilepsy (JANZ syndrome):
• • It is the most common generalized epilepsy in young adults, accounting for 5% of all epilepsies
• • It has been linked to mutations in many genes including CACNB4; CLNC2; EJM2, 3, 4, 5, 6, 7, 9; GABRA
• • Typically, it starts in early adolescence between ages of 12–16 years
• • Myoclonic jerks in the morning, often causing the patient to drop things

24. Which one of the following is the characteristic feature of juvenile myoclonic epilepsy?

A. Myoclonic seizures frequently occur in morning
B. Complete remission is common
C. Response to anticonvulsants is poor
D. Associated absence seizures are present in majority of patients

Ans A

25. True about juvenile myoclonic epilepsy:

A. Clonazepam
B. Phosphenytoin
C. Lamotrigine
D. Diazepam
E. Spike and waves in EEG498 Section 3: Systemic Pediatrics 82. A child in status epilepticus should not be given: (JIPMER 04)

Ans B Generalized seizure, c. Myoclonus, d. Response to sodium valproate, e. Spike and waves in EEG

26. All are feature of absence seizures except:

A. Usually seen in childhood
B. 3 Hz spike wave in EEG
C. Postictal confusion
D. Precipitation by hyperventilation CEREBRAL PALSY

Ans C Postictal confusion 56 above

27. Minor risk factors for recurrence of febrile seizure include the following EXCEPT

A. complex febrile seizure
B. age <1 yr
C. family history of febrile seizures
D. male gender
E. lower serum sodium

Ans B duration of fever <24 hr, and fever 38-390C are major risk factors for recurrence of febrile seizure.

28. There are several predictors of epilepsy after febrile seizures, the highest percent of risk factor for subsequent epilepsy after febrile seizure is

A. complex febrile seizure, any type
B. fever <1 hr before febrile seizure
C. recurrent febrile seizures
D. focal complex febrile seizure
E. family history of epilepsy

Ans D has 29% risk for subsequent epilepsy.

29. Electroencephalogram (EEG) in febrile seizure is characterized by the following

A. an EEG need not norrmally be performed in first simple febrile seizure
B. an abnormal EEG could predict the future recurrence of febrile seizures or epilepsy
C. spikes during drowsiness are often seen in children with febrile seizures
D. an EEG performed within 2 wk of a febrile seizure often have nonspecific slowing
E. an EEG should be used to delineate the type of epilepsy

Ans B An EEG would not predict the future recurrence of febrile seizures or epilepsy even if the result is abnormal.

30. Absence seizures are MOST often initially treated with

A. ethosuximide
B. valproate
C. lamotrigine
D. acetazolamide
E. clonazepam

Ans A Absence seizures are most often initially treated with ethosuximide, which is as effective as and less toxic than valproate and more effective than lamotrigine.

31. Epilepsy surgery is often used to treat refractory epilepsy of a number of etiologies including the following EXCEPT

A. cortical dysplasia
B. tuberous sclerosis
C. polymicrogyria
D. degenerative problems
E. Sturge-Weber syndrome

Ans D Epilepsy surgery is often used to treat refractory epilepsy of a number of etiologies including cortical dysplasia, tuberous sclerosis, polymicrogyria, hypothalamic hamartoma, and hemispheric syndromes, such as Sturge-Weber syndrome, hemimegalencephaly, Rasmussen encephalitis, and Landau-Kleffner syndrome. Patients with intractable epilepsy resulting from metabolic or degenerative problems are not candidates for resective epilepsy surgery.

32. The MOST common cause of neonatal seizures is

A. vascular events
B. intracranial infections
C. brain malformations
D. hypoxic-Ischemic encephalopathy
E. metabolic disturbances

Ans D is the most common cause of neonatal seizures, accounting for 50-60% of patients. Seizures secondary to this encephalopathy occur within 12 hr of birth.

33. All the following are true regarding diazepam in neonatal seizures EXCEPT

A. diazepam is highly lipophilic
B. it is cleared very quickly
C. recurrence of seizures is more than other anticonvulsants
D. it carries a risk of apnea and hypotension
E. it is currently recommended as a first-line agent

Ans E Because of the respiratory and blood pressure limitations and because the intravenous preparation contains sodium benzoate and benzoic acid, it is currently not recommended as a first-line agent.

34. The current antiepileptic drug (AEDs) of choice for primary generalized tonic-clonic seizures in children is

A. phenytoin
B. carbamazepine
C. phenobarbital
D. valproate
E. topiramate

Ans D Class I evidence demonstrates that phenytoin, carbamazepine, phenobarbital, primidone, valproate, topiramate, oxcarbazepine, and lamotrigine are effective for the treatment of primary generalized tonic-clonic seizures. On the basis of efficacy and tolerability, valproate and lamotrigine have emerged as the current drugs of choice for grand mal seizures.

35. The MOST common type of cerebral palsy (CP) associated with seizures is

A. spastic hemiplegia
B. spastic quadriplegia
C. spastic diplegia
D. hypotonic CP
E. ataxic CP

Ans A

36. The MOST common precipitant of status epilepticus in children is

A. CNS infection
B. fever
C. medication change
D. trauma
E. metabolic cause

Ans B /infection (36%) ,CNS infection (5%) , medication change (20%) , trauma (4%) , unknown (9%) , cerebrovascular (3%) , metabolic (8%) , ethanol/drug-related (2%) , congenital (7%) , tumor (1%) , and anoxia (5%).

37. Major risk factors for recurrence of febrile seizure include

A. fever 38-39°c
B. family history of febrile seizures
C. family history of epilepsy
D. complex febrile seizure
E. lower serum sodium at time of presentation

Ans A Risk factors for recurrence of febrile seizure include 🅾️ Age <1 yr 🅾️ Duration of fever <24 hr 🅾️ Fever 38-39°C (100.4-102.2°F) 🅾️ Family history of febrile seizures 🅾️ Family history of epilepsy 🅾️ Complex febrile seizure 🅾️ Daycare 🅾️ Male gender 🅾️ Lower serum sodium at time of presentation

38. In patients with febrile seizures, one of the following risk factors has the highest risk for subsequent epilepsy

A. recurrent febrile seizures
B. fever <1 hr before febrile seizure
C. neurodevelopmental abnormalities
D. family history of epilepsy
E. complex febrile seizures

Ans C Risk factor for subsequent epilepsy * simple febrile seizure 1% * recurrent febrile seizures 4% * complex febrile seizures (more than 15 min duration or recurrent within 24 hr) 6% * fever<1 hr before febrile seizure 11% * family history of epilepsy 18% * complex febrile seizures (focal) 29% * neurodevelopmental abnormalities 33%

39. The majority of patients who had prolonged febrile seizures and encephalopathy after vaccination and who had been presumed to have suffered from vaccine encephalopathy (seizures and psychomotor regression occurring after vaccination and presumed to be caused by it) turn out to have

A. generalized epilepsy with febrile seizures plus(GEFS+)
B. temporal lobe epilepsy secondary to mesial temporal sclerosis
C. myoclonic astatic seizures
D. Dravet syndrome
E. focal febrile seizures plus epilepsy variant

Ans D The majority of patients who had prolonged febrile seizures and encephalopathy after vaccination and who had been presumed to have suffered from vaccine encephalopathy(seizures and psychomotor regression occurring after vaccination and presumed to be caused by it) turn out to have Dravet syndrome mutations, indicating that their disease is caused by the mutation and not secondary to the vaccine. This has raised doubts about the very existence of the entity termed vaccine encephalopathy.

40. **Higher risk of recurrence of the febrile seizure is associated with lower serum**

A. **sodium**
B. potassium
C. chloride
D. calcium
E. magnesium

Ans A A low sodium level is associated with higher risk of recurrence of the febrile seizure within the following 24 hr.

41. **A history of personality change in a patient with seizure could suggest the following as a cause of seizure**

A. **intracranial tumor**
B. degenerative disease
C. metabolic disease
D. stimulants drugs
E. congenital brain dysfunction

Ans A A history of personality change or symptoms of increased intracranial pressure can suggest an intracranial tumor. Similarly, a history of cognitive regression can suggest a degenerative or metabolic disease. Certain medications such as stimulants or antihistamines, particularly sedating ones, can precipitate seizures. A history of prenatal or perinatal distress or of developmental delay can suggest etiologic congenital or perinatal brain dysfunction.

42. **Guidelines on the evaluation of a first unprovoked nonfebrile seizure, the following studies are recommended in specific clinical situations**

A. head CT
B. head MRI
C. **spinal tap**

Ans C is considered in patients with suspected meningitis or encephalitis, in children without brain swelling or papilledema, and in children in whom a history of intracranial bleeding is suspected without evidence of such on head CT.

43. **Drug therapy should be based on the type of seizure and the epilepsy syndrome as well as on other individual factors. Of the following, the drug of first choice for focal seizures and epilepsies is**

A. **carbamazepine**
B. ethosuximide
C. valproate
D. lamotrigine
E. clobazam

Ans A In general, the drugs of first choice for focal seizures and epilepsies are oxcarbazepine and carbamazepine; for absence seizures, ethosuximide; for juvenile myoclonic epilepsy, valproate and lamotrigine; for Lennox-Gastaut syndrome, clobazam, valproate, topiramate, lamotrigine, and, most recently, as add on, rufinamide; and for infantile spasms, adrenocorticotropic hormone (ACTH).

44. **Weight gain and alopecia are side effects of the following antiepileptic drugs AEDs**

A. primidone
B. **valproic acid**
C. oxcarbazepine
D. lamotrigine
E. acetazolamide

Ans B Side effects of valproic acid: weight gain, hyperammonemia, tremor, alopecia, and menstrual irregularities. Serious: hepatic and pancreatic toxicity.

45. **Discontinuation of antiepileptic drugs (AEDs) is usually indicated when children are free of seizures for at least**

A. 1 yr
B. 1.5 yr
C. **2 yr**
D. 2.5 yr
E. 3 yr

Ans C Discontinuation of AEDs is usually indicated when children are free of seizures for at least 2 yr.

46. **The following factors are associated with a higher risk of seizure relapse after antiepileptic drugs (AEDs) withdrawal EXCEPT**

A. **younger age of epilepsy onset**
B. longer duration of epilepsy
C. presence of multiple seizure types
D. need to use more than 1 AED
E. abnormal EEG before medication is discontinued

Ans A Older age of epilepsy onset.

47. **Sudden unexpected death in epilepsy (SUDEP) is the most common epilepsy related mortality in patients with chronic epilepsy. All the following are risk factors EXCEPT**

A. polypharmacology
B. **female gender**
C. age younger than 16 yr
D. long duration of epilepsy
E. frequent seizures

Ans B Male gender.

48. **There are 5 main neonatal seizure types: subtle, clonic, tonic, spasms, and myoclonic. One of the following seizures is frequently not associated with electrographic discharges**

A. spasms
B. focal clonic
C. **subtle**
D. focal tonic
E. generalized myoclonic

Ans C Spasms, focal clonic, focal tonic, and generalized myoclonic seizures are, as a rule, associated with electrographic discharges (epileptic seizures), whereas motor automatisms, the subtle, generalized tonic and multifocal myoclonic episodes are frequently not associated with discharges and thus are thought to often represent release phenomena with abnormal movements secondary to brain injury rather than true epileptic seizures.

49. **Subtle seizures include all the following EXCEPT**

A. blinking
B. mouthing
C. fluctuations in heart rate
D. **hypotension episodes**
E. apnea

Ans D Subtle seizures include transient eye deviations, nystagmus, blinking, mouthing, abnormal extremity movements (rowing, swimming, bicycling, pedaling, and stepping), and fluctuations in heart rate, hypertension episodes, and apnea. Subtle seizures occur more commonly in premature than in full-term infants.

50. **Many inborn errors of metabolism cause generalized convulsions in the newborn period. Prominent hiccups, persistent generalized seizures, and lethargy rapidly leading to coma are features of**

A. propionic academia
B. maple syrup urine disease
C. **nonketotic hyperglycinemia**
D. Leigh disease
E. neonatal adrenoleukodystrophy

Ans C an intractable condition characterized by markedly elevated plasma and CSF glycine levels, prominent hiccups, persistent generalized seizures, and lethargy rapidly leading to coma.

51. **The initial drug used to control acute neonatal seizures is usually**

A. diazepam
B. midazolam
C. phenobarbital
D. **lorazepam**
E. phenytoin

Ans D The initial drug used to control acute seizures is usually lorazepam. Lorazepam is distributed to the brain very quickly and exerts its anticonvulsant effect in <5 min. It is not very lipophilic and does not clear out from the brain very rapidly. Its action can last 6-24 hr. Usually, it does not cause hypotension or respiratory depression. The dose is 0.05 mg/kg (range: 0.02-0.10 mg/kg) every 4-8 hr.

52. **Intravenous phenytoin is not widely used to control acute neonatal seizures because of all the following EXCEPT**

A. reduced solubility
B. severe local cutaneous reactions
C. interaction with other drugs
D. possible cardiac toxicity
E. **not possible to mix with saline solutions**

Ans E It is not possible to mix phenytoin or fosphenytoin with dextrose solutions.

53. **Status epilepticus is a medical emergency that should be anticipated in any patient who presents with an acute seizure. It is defined as continuous seizure activity or recurrent seizure activity without regaining of consciousness lasting for more than**

A. **5 min**
B. 15 min
C. 30 min
D. 45 min
E. 60 min

Ans A

54. **The following studies are needed for all patients with status epilepticus EXCEPT**

A. glucose
B. magnesium
C. complete blood count
D. **lumbar puncture**
E. CT scan

Ans D comprehensive toxicologic screens, MRI, and other laboratory tests are performed depending on clinical suspicion and need.

55. **Currently, the level of the evidence for refractory treatment of status epilepticus is strongest for**

A. **midazolam**
B. propofol
C. levetiracetam
D. phenytoin
E. Phenobarbital

Ans A Currently, the level of the evidence for refractory treatment is strongest for midazolam and valproate, followed by propofol and pentobarbital/thiopental, followed by levetiracetam, phenytoin/fosphenytoin, lacosamide, topiramate, and phenobarbital.

56. **Bilateral cortical visual dysfunction, encephalopathy, and seizures are seen in**

A. global hypoxic–ischemic encephalopathy
B. herpes encephalitis
C. multiple sclerosis
D. **hypertensive encephalopathy**
E. hypoglycemia

Ans D The posterior reversible leukoencephalopathy syndrome is seen in children with hypertension, often in the context of an acute rise in blood pressure. Posterior regions are selectively involved, possibly resulting in symptoms of bilateral cortical visual dysfunction in addition to encephalopathy and seizures.

57. **Seizures are common during the course of bacterial meningitis. Immediate therapy for seizures includes intravenous diazepam (0.1- 0.2 mg/kg/dose) or lorazepam (0.05- 0.10 mg/kg/dose). After immediate management of seizures, patients should receive the following anticonvulsant to reduce the likelihood of recurrence**

A. valproic acid
B. **phenytoin**
C. phenobarbital
D. lorazepam
E. carbamazepine

Ans B After immediate management of seizures, patients should receive phenytoin (15-20 mg/ kg loading dose, 5 mg/kg/24 hr maintenance) to reduce the likelihood of recurrence. Phenytoin is preferred to phenobarbital because it produces less CNS depression and permits assessment of a patient's level of consciousness. Serum phenytoin levels should be monitored to maintain them in the therapeutic range (10-20 µg/mL).

58. The following viruses have clinical manifestations similar to that of the enteroviruses with the exception of more severe MRI lesions of the cerebral cortex and at times an absence of a CSF pleocytosis

A. arboviruses
B. **parechoviruses**
C. herpes simplex virus (HSV) type
D. rabies virus
E. Epstein-Barr virus

Ans B may be an important cause of aseptic meningitis or encephalitis in infants. The clinical manifestations are similar to that of the enteroviruses with the exception of more severe MRI lesions of the cerebral cortex and at times an absence of a CSF pleocytosis.

59. Congenital Guillain-Barré syndrome, manifest as generalized hypotonia, weakness, and areflexia in an affected neonate, fulfilling all electrophysiologic and CSF criteria and in the absence of maternal neuromuscular disease. Of the following, the MOST effective treatment is

A. steroids
B. **reassurance**
C. plasmapheresis
D. immunosuppressive drugs
E. intravenous immunoglobulin (IVIG)

Ans B Treatment might not be required, and there is gradual improvement over the 1st few months and no evidence of residual disease by 1 yr of age.

Febrile Seizures

60. Content of meningocele:

A. **Dura mater**
B. Spinal cord
C. Brain matter
D. Cauda equina

Ans A A meningocele consists of a CSF-filled meningeal sac only,

61. Rectal diazepam every 8 hourly for 2-3 days to reduce the risk of recurrence Select the correct answer using the code given below:

A. 1 and 2 only
B. 1 and 4 only
C. **3 and**
D. 1, 2 and

Ans C Lumbosacral

62. Continuous prophylactic anticonvulsant therapy is not needed in a child with febrile convulsions with?

A. 1–2%
B. 2–5%
C. **5–10%**
D. 10–15%

Ans C Typical simple febrile fits Continuous antiepileptic therapy is not required for simple febrile seizures, but intermittent prophylaxis may be given.

63. Treatment of simple febrile convulsion is:

A. Control of fever
B. **Rectal diazepam**
C. CSF finding
D. Blood reports

Ans B Rectal diazepam is prescribed for aborting the episode of febrile convulsions.

64. True regarding febrile convulsions is:

A. Paracetamol 6 hourly
B. **Paracetamol & diazepam**
C. Diazepam
D. Phenobarbitone
E. Continuous antiepileptic therapy is required59. A 4-year-old male had febrile seizures, best prophylaxis:

Ans B Patient with family h/o Febrile seizures have increased incidence of recurrence, d. Usually last for a short while Simple Febrile seizures last for < 15 minutes

65. A 9-month-old infant can do all the following EXCEPT

A. pulls to stand
B. pincer grasp
C. plays pat-a-cake
D. imitates sounds
E. **comes when called**

Ans E Walks with 1 hand held, releases an object on command, comes when called, and 1-2 meaningful words are milestones of 12-month-old child.

66. Infants with Tay-Sachs disease (TSD) are characterized by the following EXCEPT

A. convulsions
B. blindness
C. deafness
D. cherry-red spots
E. **microcephaly**

Ans E Macrocephaly becomes apparent by 1 yr of age and results from the 200-300 fold normal content of GM2 ganglioside deposited in the brain.

67. The MOST common presenting complaint of intramedullary spinal cord tumors is

A. gait disturbance
B. sensory deficits
C. scoliosis
D. **back pain**
E. urinary urgency

Ans D With the exception of the uncommon malignant glial tumors of the spinal cord, which tend to present precipitously, intramedullary spinal cord tumors present in a very insidious manner. Back pain related to the level of the tumor is a common presenting complaint. It is likely that this pain will awaken the child from sleep and improve as the day progresses.

68. The MOST definitive diagnostic test in Werdnig-Hoffmann disease is

B. muscle biopsy
C. serum creatine kinase
D. molecular genetic test
E. motor nerve conduction study

Ans D The serum creatine kinase level may be normal but more commonly is mildly elevated in the hundreds. Results of motor nerve conduction studies are normal, except for mild slowing in terminal stages of the disease. EMG shows fibrillation potentials and other signs of denervation of muscle. A secondary mitochondrial DNA depletion is sometimes demonstrated in the muscle biopsy of infants with SMA. The molecular genetic test of the SMN gene provides definite confirmation of the diagnosis.

69. Of the following, the LEAST recognized presentation of Charcot-Marie-Tooth disease is

A. foot drop
B. claw hand
C. pes cavus deformities
D. stork-like contour legs
E. tripping over their own feet

Ans B Atrophy of muscles of the forearms and hands is usually not as severe as that of the lower extremities, but in advanced cases contractures of the wrists and fingers produce a claw hand.

Cerebral Palsy

70. Tuft of hair over the lumbosacral region in a newborn is suggestive of:

A. Spina bifida occulta
B. Sinus tract
C. Tumor
D. Any of the above

Ans D 90% of patients with occult spinal dysraphism, have a cutaneous abnormality overlying the lower spine, including a small dimple, tuft of hair, dermal vascular malformation, or subcutaneous lipoma Occult spinal dysraphism includes spina bifida occulta, lipomeningocele, intradural lipoma, diastematomyelia, tight filum terminale, weiveR dermoid cyst-sinus, aberrant nerve roots, anterior sacral meningocele, and cauda equina tumor.

71. The most common cause of convulsions in a 9 month old infant with fever:

A. Septicemia
B. Febrile seizures
C. Brain tumor
D. Phenylketonuria EPILEPSY/SEIZURES

Ans B Febrile seizures EPILEPSY/SEIZURES

72. Diplegia means:

A. Spastic diplegia
B. Spastic quadriplegia
C. Mental retardation
D. Seizures

Ans B Weakness of lower limbs

73. The most common type of cerebral palsy is?

A. Spastic
B. Atonic
C. Extrapyramidal
D. Mixed

Ans A • Most common type of cerebral palsy is spastic diplegia in 35% cases, followed by Hemiplegia in 25% cases
• Spastic quadriplegia is the most severe form of CP because of marked motor impairment of all extremities and the high association with intellectual disability and seizures.

74. A child presented with mental retardation and spastic limbs with h/o perinatal distress. Diagnosis:

A. Cerebral palsy
B. HIE
C. Down's syndrome
D. Edward syndrome CNS INFECTIONS

Ans A The CNS consequences and Sequelae due to perinatal asphyxia is referred as cerebral plasy.

75. A child with head injury is admitted in ICU. Management of raised ICP includes all except:

A. Mannitol when ICP>20
B. Minimal use of sedation and analgesia
C. Controlled mechanical ventilation
D. Hypertonic saline may be used

Ans B ICP should be maintained <20 mm Hg; First-tier therapy for raised ICP includes elevation of head of bed, ensuring midline positioning of head, controlled mechanical ventilation, and sedation and analgesia (i.e., benzodiazepines and narcotics) & osmolar agents like mannitol and hypertonic saline.

76. The MOST severe form of CP is

A. spastic diplegia
B. spastic quadriplegia
C. spastic hemiplegia
D. athetoid CP
E. dyskinetic CP

Ans B is the most severe form of CP because of marked motor impairment of all extremities and the high association with intellectual disability and seizures.

77. All the following can be used for treatment of cerebral palsy (CP) EXCEPT

A. benzodiazepines
B. baclofen
C. dantrolene
D. botulinum toxin
E. hyperbaric oxygen

Ans E has not been shown to improve the condition of children with CP.

78. In Guillain-Barré syndrome, the paralysis usually follows a nonspecific gastrointestinal or respiratory infection by approximately 10 days. Of the following, the MOST likely respiratory infection that triggers the disease is

A. Chlamydia trachomitis
B. Staphylococcal aureus
C. Haemophilus influenzae
D. **Mycoplasma pneumoniae**
E. Streptococcal pneumonia

Ans D The original infection might have caused only gastrointestinal (especially Campylobacter jejuni, but also Helicobacter pylori) or respiratory tract (especially Mycoplasma pneumoniae) symptoms.

79. The earliest and MOST constant sign of myasthenia gravis is

A. **ptosis**
B. dysphagia
C. slurred speech
D. difficult chewing
E. poor head control

Ans A In juvenile autoimmune myasthenia gravis, unilateral or bilateral but usually asymmetrical ptosis and some degree of extraocular muscle weakness are the earliest and most constant signs.

Meningitis

80. Myelomeningocele most commonly involves which region of spine:

A. Cervico-dorsal
B. Dorsolumbar
C. **Lumbosacral**
D. Sacro-coccygeal

Ans C

81. Lennox-Gastaut Syndrome is characterized by:

A. Single seizure types
B. Good prognosis and adequate controlled epilepsy
C. Impaired cognitive function in all cases
D. **EEG showing slow (< 3Hz) spike and wave discharge**

Ans D EEG showing slow (< 3 Hz) spike and wave discharge
Lennox-Gastaut syndrome (LGS): High yield facts
• • It typically starts between the ages of 2 to 10 yr.
• • It consists of a triad of developmental delay, multipleQ seizure types and typical EEG changes.
• • Patients commonly have myoclonic, atypical absences atonic, astatic, and tonic seizures that are difficult to control, and most are left with long-term cognitive impairment and intractable seizures despite multiple therapies.

82. How long should a child be isolated after being diagnosed with bacterial meningitis to prevent further transmission?

A. **Till 24 hours after starting antibiotics**
B. Till cultures become negative
C. Till antibiotics course in complete
D. Till 12 hours after admission

Ans A Till 24 hours in after starting antibiotics The CSF becomes sterile within 24-48 hours of initiation of appropriate antibiotic therapy.

83. Most common cause of meningitis in post-neonatal period:

A. Klebsiella pneumoniae
B. **Streptococcus pneumoniae**
C. Staphylococcus aureus
D. Mycobacterium tuberculosis

Ans B

84. In a small child diagnosed with H. influenza meningitis. What investigation must to be done before discharging him from the hospital?

A. **BERA**
B. MRI
C. CT scan
D. X-ray skull

Ans A Sensorineural hearing loss is the most common sequelae of bacterial meningitis; So a hearing evaluation should be done before discharge of a child with meningitis, from hospital.

85. Residual auditory defect is a common complication in meningitis caused by:

A. Staphylococcus
B. Meningococcus
C. Escherichia coli
D. **Haemophilus influenzae**

Ans D Sensorineural hearing loss as a complication of bacterial meningitis, is a result of cochlear infection. It occurs in as many as 30% of patients with pneumococcal meningitis, 10% with meningococcal, and 5-20% of those with H. influenzae type b meningitis. So, the best answer among the given options is H. influenzae.

86. Meningitis in a 4-year-old child; microscopy of gram stained CSF sample will reveal:

A. **Gram pos cocci**
B. Gram pos bacilli
C. Gram neg diplococci
D. Gram neg bacilli

Ans A Meningitis in a 4 yr child is mostly caused by Streptococcus pneumoniae, which is a gram positive cocci.

87. The CSF findings in TB meningitis include:

A. Streptococcus pneumoniae
B. **H. influenzae**
C. Listeria
D. Neisseria meningitidis

Ans B Low sugar + high protein and lymphocytosis
Lymphocytosis is seen in Tuberculous meningitis, though in early stages, CSF neutrophilia may be seen

88. A 7-day-old newborn presented with meningitis. Common causes are:

A. **E. coli**
B. Strep. pneumoniae
C. N. meningitis
D. Strep. agalactiae
E. Staph aureus

Ans A d. Strep. agalactiae common causes of Acute bacterial meningitis in neonates are Gr. B Strepto, E. coli & Listeria

89. **Neonatal meningitis is caused by:**
A. Group A Streptococcus
B. **Group B streptococcus**
C. E. coli
D. H. influenzae
E. Klebsiella

Ans B Group B Streptococcus, c. E. coli

90. **The following bacteria are most often associated with acute neonatal meningitis except:**
A. E. coli
B. Step. agalactiae
C. **N. meningitides**
D. L. monocytogenes

Ans C

91. **A neonate develops signs of meningitis at 7 days of birth. The presence of which of the following infections agent in the maternal genital tract can be the causative agent of this disease:**
A. Neisseria gonorrhoeae
B. Chlamydia trachomatis
C. **Strep agalactiae**
D. H. ducreyi

Ans C

92. **Bacterial meningitis in children (2 months–12 years) is usually due to the following organisms except:**
A. Streptococcus pneumoniae
B. Neisseria meningitides
C. H. influenzae type B
D. **L. monocytogenes**

Ans D Listeria is an important cause of meningitis in the neonatal period, but not in older children.

93. **The most common agent associated with neonatal bacterial meningitis is:**
A. H. influenzae type b
B. N. meningitides
C. Strep. pneumoniae
D. **Strep. agalactiae**

Ans D

94. **The following is an alternative treatment for L. monocytogenes meningitis**
A. vancomycin
B. cefotaxime
C. chloramphenicol
D. **intravenous trimethoprimsulfamethoxazole**
E. meropenem

Ans D If L. monocytogenes infection is suspected, as in young infants or those with a T- lymphocyte deficiency, ampicillin (200 mg/kg/24 hr, given every 6 hr) should also be given because cephalosporins are inactive against L. monocytogenes. Intravenous trimethoprim sulfamethoxazole is an alternative treatment for L. monocytogenes.

95. **Reversible gallbladder pseudolithiasis, detectable by abdominal ultrasonography during treatment of bacterial meningitis is usually caused by**
A. ceftazidime
B. cefotaxime
C. **ceftriaxone**
D. meropenem
E. chloramphenicol

Ans C may cause reversible gallbladder pseudolithiasis, detectable by abdominal ultrasonography. This is usually asymptomatic but may be associated with emesis and upper right quadrant pain.

96. **Data support the use of intravenous dexamethasone, 0.15 mg/kg/dose given every 6 hr for 2 days, in the treatment of children older than 6 wk with acute bacterial meningitis caused by H. influenza type b. for the following reasons EXCEPT**
A. shorter duration of fever
B. lower CSF protein
C. **lower mortality**
D. lower CSF lactate levels
E. reduction in sensorineural hearing loss

Ans C Among children with meningitis caused by H. influenza type b, corticosteroid recipients have a shorter duration of fever, lower CSF protein and lactate levels, and a reduction in sensorineural hearing loss. Data in children regarding benefits, if any, of corticosteroids in the treatment of meningitis caused by other bacteria are inconclusive.

97. **The MOST common neurologic sequelae of bacterial meningitis is**
A. recurrent seizures
B. delay in acquisition of language
C. **hearing loss**
D. cognitive impairment
E. visual impairment

Ans C Sensorineural hearing loss is the most common sequela of bacterial meningitis and, usually, is already present at the time of initial presentation. It is a result of cochlear infection and occurs in as many as 30% of patients with pneumococcal meningitis, 10% with meningococcal, and 5-20% of those with H. influenzae type b meningitis. Hearing loss may also be caused by direct inflammation of the auditory nerve.

Encephalitis

98. **Porencephaly is due to:**
A. Dandy Walker syndrome
B. **Cerebral infarction**
C. Fetal alcohol syndrome
D. Trisomy 13 HYDROCEPHALUS

Ans B Porencephaly
• • Porencephaly is the presence of cysts or cavities within the brain that result from developmental defects or acquired lesions, including infarction of tissue. They represent developmental abnormalities of cell migration.
• • True porencephalic cysts are most commonly located in the region of the sylvian fissure and typically communicate with the subarachnoid space, the ventricular system, or both.
• • Often associated with microcephaly, abnormal patterns of adjacent gyri, and encephalocele.

99. A 15-year-old boy presented with day dreaming and decline in school performance. The likely possibility is?

A. Atonic seizure
B. Myoclonic seizure
C. **Typical absence seizure**
D. Atypical absence seizure

Ans C Typical absence seizures may present in adolescents with day dreaming and decline in school performance.

100. What is the most common lobe involved in Herpes Simplex virus encephalitis?

A. Frontal lobe
B. **Temporal lobe**
C. Occipital lobe
D. Parietal lobe

Ans B

101. Commonest cause of meningoencephalitis in children is:

A. HSV
B. Measles
C. Arbovirus
D. **Enterovirus**

Ans D Enteroviruses are the most common cause of viral meningoencephalitis in children.

102. A neonate develops encephalitis without any skin lesions. Most probable causative organism is:

A. HSV I
B. **HSV II**
C. Meningococci
D. Streptococci

Ans B with CNS infection by Herpes Simplex:
• In the neonatal period, HSV II (acquired from maternal genital lesions) is an important cause of encephalitis
• Beyond the neonatal period, HSV encephalitis is almost always caused by HSV I
• HSV encephalitis usually involves the (most common) & temporal frontal cortex and the limbic system
• HSV is a cause of aseptic meningitis and is the most common cause of recurrent aseptic meningitis (Mollaret meningitis).

103. Excessive irritability and crying, unexplained episodes of hyperpyrexia, vomiting, and difficulty feeding are the symptoms of the following neuro-degenerative disease

A. Tay-Sachs disease (TSD)
B. Sandhoff disease
C. juvenile GM2 gangliosidosis
D. **Krabbe disease (KD)**
E. metachromatic leukodystrophy

Ans D The symptoms of KD become evident in the 1st few mo of life and include excessive irritability and crying, unexplained episodes of hyperpyrexia, vomiting, and difficulty feeding. In the initial stage of KD, children are often treated for colic or milk allergy with frequent formula changes.

104. The MOST common cause of viral meningoencephalitis are

A. arboviruses
B. **enteroviruses**
C. herpes simplex virus (HSV) type
D. varicella-zoster virus
E. mumps

Ans B are the most common cause of viral meningoencephalitis. As of 2014, more than 70 serotypes of these small RNA viruses have been identified.

105. All the following vaccines may induce Guillain-Barré syndrome (GBS) EXCEPT

B. **Rota**
C. Rabies
D. Influenza
E. Conjugated meningococcal vaccine

Ans B

106. The Creatine kinase (CK) which is found in only 3 organs and may be separated into corresponding isozymes: MM for skeletal muscle, MB for cardiac muscle, and BB for brain. Of the following, the CK level is characteristically elevated in

A. Myasthenia gravis
B. Guillain-Barré syndrome
C. Werdnig-Hoffmann disease
D. **Duchenne muscular dystrophy**
E. Emery-Dreifuss Muscular Dystrophy

Ans D Serum CK determination is not a universal screening test for neuromuscular disease because many diseases of the motor unit are not associated with elevated enzymes. The CK level is characteristically elevated in certain diseases, such as Duchenne muscular dystrophy, and the magnitude of increase is characteristic for particular diseases.

Neurocysticercosis

107. A baby is born with fused eyes, single nasal chamber and undeveloped callosum. What is diagnosis?

A. **Holoprosencephaly**
B. Schizencephaly
C. Plagiocephaly
D. Brachycephaly

Ans A Holoprosencephaly
• An abnormality of brain development in which the brain does not properly divide into the right and left hemispheres.
• 3 main types (from most to least severe): Alobar, semi-lobar & lobar
• Severity of facial defects corresponds to severity of brain defect.
• Other signs and symptoms often include intellectual disability, seizures, pituitary dysfunction, short stature
• Associated with trisomy 13 (MC), trisomy 18, CHARGE syndrome, Smith-Lemli-Opitz syndrome, Rubinstein–Taybi syndrome

108. **In a premature baby with convulsion after 2 days of Questions birth, first investigation to be done is:**

A. Transcranial USG
B. MRI
C. Skull radiography
D. CT skull

Ans A Intracranial hemorrhage can be a common cause for seizures in a preterm neonate, which can be picked up easily on a transcranial USG through the open anterior fontanelle.

109. **Which of the following is the antihelminthic of choice for the treatment of neurocysticercosis ?**

A. Mebendazole
B. Albendazole
C. Thiabendazole
D. Ivermectin

Ans B Albendazole is the antithelminthic of choice for the treatment of neurocysticercosis.

110. **Pseudotumor cerebri is caused by all except:**

A. Medulloblastoma
B. Encephalocele
C. Vein of Galen malformation
D. Arachnoid cyst

Ans A Pseudotumor cerebri (idiopathic intracranial hypertension) is by definition a diagnosis of exclusion with no mass lesion on imaging; a true mass such as medulloblastoma causing obstructive hydrocephalus is excluded from the diagnosis.

111. **Regarding the CSF, all the following are true EXCEPT**

A. approximately 25% of CSF originates from extrachoroidal sources
B. in a normal child, about 20 mL/hr of CSF is produced
C. total volume of CSF approximates 150 mL in an infant
D. normally, CSF flows from the lateral ventricles through the foramina of Monro into the 3rd ventricle
E. CSF is absorbed primarily by the arachnoid villi

Ans C The total volume of CSF approximates 50 mL in an infant and 150 mL in an adult.

112. **A premature infant blinks in response to a bright light at**

A. 26 wk of corrected gestational age
B. 28 wk of corrected gestational age
C. 30 wk of corrected gestational age
D. 32 wk of corrected gestational age
E. 34 wk of corrected gestational age

Ans B At 28 wk of corrected gestational age, a premature infant blinks in response to a bright light, and at 32 wk, the infant maintains eye closure until the light source is removed. A normal 37 wk infant turns the head and eyes toward a soft light, and a term infant is able to fix on and follow a target, such as the examiner's face.

113. **Presenting symptoms in pediatric multiple sclerosis (MS) include the following**

A. hemiparesis
B. optic neuritis
C. ataxia
D. dysarthria
E. encephalopathy

Ans E is less common and suggests consideration of ADEM or possibly neuromyelitis optica (NMO).

114. **The MOST common involved segments in transverse myelitis (TM) are in the**

A. cervical region
B. thoracic region
C. lumbar region
D. lumbo-sacral region
E. sacral region

Ans B

115. **In Guillain-Barré syndrome, the onset is gradual and progresses over days or weeks. The maximal severity of weakness is usually reached by**

A. 2 wk
B. 4 wk
C. 6 wk
D. 8 wk
E. 10 wk

Ans B Weakness usually begins in the lower extremities and progressively involves the trunk, the upper limbs, and, finally, the bulbar muscles, a pattern known as Landry ascending paralysis. The onset is gradual and progresses over days or weeks; the process plateaus in 1-28 days. Weakness can progress to inability or refusal to walk and later to flaccid tetraplegia. Maximal severity of weakness is usually reached by 4 wk after onset.

116. **Dolichocephalic head is a feature of**

A. Central core myopathy
B. Myotubular myopathy
C. Nemaline rod myopathy
D. Becker muscular dystrophy
E. Emery-Dreifuss Muscular Dystrophy

Ans C Neonatal, infantile, and juvenile forms of the disease are known. The neonatal form is severe and usually fatal because of respiratory failure since birth. In the infantile form, generalized hypotonia and weakness, which can include bulbar-innervated and respiratory muscles and a very thin muscle mass, are characteristic. The head is dolichocephalic, and the palate high arched or even cleft. Muscles of the jaw may be too weak to hold it closed.

Tuberculous Meningitis

117. Identify the condition shown in the image below:

A. Dandy–Walker malformation
B. Vein of Galen malformation
C. Mega cistern magna
D. Arnold-Chiari malformation

Ans A Dandy-Walker malformation with Dandy-Walker malformation Consists of:
• Cystic dilation of 4th ventricle, Hypoplasia of the cerebellar vermis • Hydrocephalus and
• Enlarged posterior fossa Variable degrees of neurologic impairment seen. 17 a. Hypoplasia of cerebellar vermis Chiari malformation
• It is the most common malformation of the posterior fossa and hindbrain.
• It consists of herniation of the cerebellar tonsils though the foramen magnum.

118. Most common cause of convulsion on the first day of life in a newborn is:

A. Hypoxia
B. Head injury
C. Hypoglycemia
D. Hypocalcemia

Ans A Hypoxic–Ischemic Encephalopathy is the most common cause of neonatal seizures, accounting for 50-60% of patients. Seizures secondary to this encephalopathy usually occur within 12 hr of birth.

119. Which of the following is not seen in fetal alcohol syndrome?

A. Microcephaly
B. Macrocephaly
C. Holoprosencephaly
D. Thinning of corpus callosum

Ans B Fetal alcohol syndrome causes microcephaly, not macrocephaly, along with characteristic facial features and other CNS anomalies.

120. A 10-year-old boy, unconscious with 2 days h/o fever, comes to pediatric ICU with RR 46/min, BP 110/80 and E1 V1 M3 on Glasgow coma scale, next step of management includes:

A. Salicylic acid
B. Nalidixic acid
C. Tetracycline
D. Nitrofurantoin 502 Section 3: Systemic Pediatrics
E. Start antibiotic and order for CT scan Regarding polio which is true? Questions

Ans A Intubate and ventilate, b. Give 0.9% NaCl, e. Start antibiotic and order for CT scan GCS less than 8 (in this case 1 + 1 + 3 = 5), is an indication of intubation and mechanical ventilation IV fluid (Normal saline) should be started; Inotropes are not required in this case as blood pressure is maintained As fever is there, antibiotics should be started; A CT scan should be done to look for underlying intracranial pathology.

121. Trigoncephaly is a skull deformity that is a direct result of premature fusion of the following

A. sagittal suture
B. coronal suture
C. lambdoid suture
D. metopic suture
E. multiple sutures

Ans D Sagittal= Dolicocephaly or scaphocephaly (boat-shaped) Coronal= Unilateral: plagiocephaly Bilateral: brachycephaly, acrocephaly Lambdoid= Lambdoid/occipital plagiocephaly; right side affected in 70% of cases Multiple= Oxycephaly

122. The MOST common cause of anatomic megalencephaly is

A. Sotos syndrome
B. benign familial megalencephaly
C. Simpson-Golabi-Behmel syndrome
D. fragile X syndrome
E. Weaver syndrome

Ans B

123. The MOST common focal presentation of arterial ischemic stroke is

A. hemiparesis
B. acute visual deficit
C. speech deficit
D. sensory deficit
E. balance deficit

Ans A The most common focal presentation is hemiparesis, but acute visual, speech, sensory, or balance deficits also occur. Children with these presentations require urgent neuroimaging and consultation with a child neurologist, as emergency interventions may be indicated.

124. Gowers sign is fully expressed by the age of

A. 3 yr
B. 5 yr
C. 7 yr
D. 9 yr
E. 11 yr

Ans B An early Gower's sign is often evident by age 3 yr and is fully expressed by age 5 or 6 yr.

125. In Guillain-Barré syndrome, respiratory effort must be monitored to prevent respiratory failure and respiratory arrest. Of the following, the MOST recognized sign of impending respiratory failure is

A. dysphagia
B. tachypnea
C. tachycardia
D. altered sensorium
E. vasomotor instability

Ans A and facial weakness are often impending signs of respiratory failure.

Brain Abscess

126. **Arnold Chiari malformation is characterized by all except:**

A. **Hypoplasia of cerebellar vermis**
B. Herniation of cerebellum
C. Flattened base of skull
D. Adulthood syndrome not associated with hydrocephalus

Ans A In Chiari malformation the cerebellar vermis and tonsils are typically displaced/herniated downward through the foramen magnum rather than hypoplastic.

127. **Commonest cause of convulsions in a child with fever is:**

A. **Febrile convulsions**
B. Meningitis
C. Epilepsy
D. Hypothyroidism

Ans A Febrile seizures are the most common cause of convulsions in a child with fever.

128. **Age by which myelination of child completed is:**

A. By birth
B. 6 months
C. 6 years
D. **Adulthood**

Ans D There are several general rules regarding myelination: (1) Proximal pathways myelinate before distal pathways, (2) sensory pathways myelinate before motor pathways, and (3) projection pathways myelinate before association pathways. Myelination progresses from the central sulcus outward toward the occipital, frontal, and temporal poles. For example, pyramidal tract efferents (transmitting outwards) from the motor cortex are myelinated by the end of the first year.

129. **The skull tends to assume a square or boxlike shape in**

A. Kleinfelter syndrome
B. neurofibromatosis
C. storage disorder
D. **chronic subdural hemorrhages**
E. hydrocephalus

Ans D In chronic subdural hemorrhages, the skull tends to assume a square or boxlike shape, because the long-standing presence of fluid in the subdural space causes enlargement of the middle fossa.

130. **Crouzon syndrome is characterized by the following EXCEPT**

A. an autosomal dominant inheritance
B. brachycephaly
C. ocular proptosis
D. hypoplasia of the maxilla
E. **syndactyly**

Ans E Apert syndrome is characterized by syndactyly of the 2nd, 3rd, and 4th fingers, which may be joined to the thumb and the 5th finger.

131. **Craniosynostosis is defined as premature closure of the cranial sutures. Of the following, the MOST common form of craniosynostosis is**

A. frontal plagiocephaly
B. occipital plagiocephaly
C. **scaphocephaly**
D. trigonocephaly
E. turricephaly

Ans C Premature closure of the sagittal suture produces a long and narrow skull, or scaphocephaly, the most common form of craniosynostosis.

132. **The MOST common cause of childhood subarachnoid and intraparenchymal hemorrhagic stroke (HS) is**

A. moyamoya disease/syndrome
B. **arteriovenous malformations**
C. cerebral sinovenous thrombosis
D. hemolytic uremic syndrome
E. idiopathic thrombocytopenic purpura

Ans B are the most common cause of childhood subarachnoid and intraparenchymal HS and may occur anywhere.

133. **The predominant organisms causing brain abscesses in children are**

A. **aerobic and anaerobic streptococci**
B. Streptococcus pneumoniae
C. Enterococcus faecalis
D. bacteroides spp
E. Haemophilus aphrophilus

Ans A The predominant organisms causing brain abscesses in children are aerobic and anaerobic streptococci (60-70% of the cases) with Streptococcus milleri gp (Streptococcus anginosus, Streptococcus constellatus, and Streptococcus intermedius) being increasingly isolated from surgically drained brain abscesses.

134. **A brain abscess can be treated with antibiotics without surgery in the following conditions EXCEPT**

A. abscess is <2 cm in diameter
B. illness is of short duration (<2 wk)
C. **lesion is located in the posterior fossa**
D. no signs of increased intracranial pressure
E. child is neurologically intact

Ans C Surgery is indicated when the abscess is >2.5 cm in diameter, gas is present in the abscess, the lesion is multiloculated, the lesion is located in the posterior fossa, or a fungus is identified. Associated infectious processes, such as mastoiditis, sinusitis, or a periorbital abscess, may require surgical drainage. The duration of antibiotic therapy depends on the organism and response to treatment but is usually 4-6 wk.

Hydrocephalus

135.Which is the most common cause of ventriculomegaly in newborn?

A. Dandy-Walker syndrome
B. Arnold-Chiari malformation
C. **Aqueductal stenosis**
D. Arachnoid villi malfunction

Ans C Volpe's Neurology of the Newborn 6/e p62
Ventriculomegaly is seen in neonates with hydrocephalus; Obstructive or noncommunicating hydrocephalus develops most commonly in children because of an abnormality of the aqueduct of Sylvius or a lesion in the fourth ventricle. Nonobstructive or communicating hydrocephalus most commonly follows a subarachnoid hemorrhage, which is usually a result of intraventricular hemorrhage in a premature infant.

136.Commonly used shunt in hydrocephalus management:

A. Ventriculoatrial
B. **Ventriculoperitoneal**
C. Ventriculopericardial
D. Ventriculopleural

Ans B Therapy for hydrocephalus:
• • Medical management, including use of acetazolamide and furosemide, can provide temporary relief.
• • Most cases of hydrocephalus require extracranial shunts, particularly a ventriculoperitoneal shunt.
• • The major complications of shunting are occlusion (characterized by headache, papilledema, emesis, mental status changes) and bacterial infection (fever, headache, meningismus), usually due to Staph epidermidis.

137. Which of the following is not a cause of communicating hydrocephalus?

A. Cranial USG
B. **CT Scan**
C. MRI
D. X-ray skull

Ans B Vein of Galen malformation Vein of Galen is one of the causes of Obstructive or Non-communicating hydrocephalus.

138.Congenital hydrocephalus treatment:

A. **V–P shunt**
B. Acetazolamide
C. Glycerine
D. Furosemide

Ans A

139.Which of the following agents is likely to cause cerebral calcification and hydrocephalus in a newborn whose mother has history of taking spiramycin but was not compliant with therapy?

A. Rubella
B. **Toxoplasmosis**
C. CMV
D. Herpes

Ans B Cerebral calcification with hydrocephalus in seen in congenital toxoplasmosis.

140.Commonest cause of obstructive hydrocephalus in children:

A. **Aqueductal stenosis**
B. Aqueductal gliosis
C. Subarachnoid hemorrhage
D. Tubercular meningitis

Ans A is the most common cause of congenital/obstructive hydrocephalus in children.

141. Which type of shift in intracranial content is common in children with progressive hydrocephalus?

A. Transforaminal herniation (Karnataka PGMEE 06)
B. Upward cerebellar herniation
C. Unilateral transtentorial herniation
D. **Central transtentorial herniation**

Ans D Herniation of Brain:
• • It refers to displacement of brain tissue by an overlying or adjacent mass into a contiguous compartment that it normally does not occupy.
• • In the most common form of herniation, brain tissue is displaced from the supratentorial to the infratentorial compartment through the tentorial opening; this is referred to as transtentorial herniation.
• • Increases in ICP due to a progressive hydrocephalus can ultimately displace the brain downward into the foramen magnum—a

142.Which one of the following is a common cause of congenital hydrocephalus?

A. Craniosynostosis
B. Intrauterine meningitis
C. **Aqueductal stenosis**
D. Malformations of great vein of Galen

Ans C

143.Most common cause of hydrocephalus in children is:

A. **Post inflammatory obstruction (SGPGI 04)**
B. Budd-Chiari syndrome
C. Brain tumor
D. Perinatal injury

Ans A Postinflammatory obstruction

144.Acquired extracranial infection that causes Aqueductal stenosis is:

A. Bacterial endocarditis
B. **Mumps**
C. Measles
D. Staphylococcal septicemia NEUROCUTANEOUS SYNDROMES

Ans B Mumps virus is neurotropic and is thought to enter the CNS via the Choroid plexus and infect the choroidal epithelium and ependymal cells, both of which can be found in CSF along with mononuclear Leukocytes. Less-common CNS complications of mumps include transverse myelitis, aqueductal stenosis, and facial palsy.

145. **A 10-month-old child presents with 2 weeks history of fever, vomiting and alteration of sensorium. Cranial CT scan reveals basal exudates and hydrocephalus, the most likely etiological agent is:**

A. Mycobacterium tuberculosis
B. Cryptococcus neoformans
C. Listeria monocytogenes
D. Streptococcus pneumoniae

Ans A Fever for 2 weeks along with neurological features with CNS imaging showing basal exudates and hydrocephalus are compatible with a diagnosis of Tuberculous meningitis.

146. **Following feature differentiate hydranencephaly from hydrocephalus:**

A. Hydranencephaly is static while hydrocephalus continue to increase in size
B. Hydranencephaly does not cause increase in head size
C. Cerebral cortex is deficient/hypoplastic in hydranencephaly

Ans C Hydranencephaly
• • Cerebral hemispheres are absent or represented by membranous sacs with remnants of cortex dispersed over the membrane.
• • Midbrain and brainstem are relatively intact; Transillumination shows an absence of cerebral hemispheres.
• • Cause of hydranencephaly is thought to be bilateral occlusion of internal carotid arteries during early fetal development.
• • Affected infants can have a normal or enlarged head circumference at birth that grows at an excessive rate postnatally.

147. **Causes of communicating hydrocephalus include**

A. achondroplasia
B. aqueductal stenosis
C. Chiari malformation
D. Dandy-Walker malformation
E. Klippel-Feil syndrome

Ans A Aqueductal stenosis, Chiari malformation, Dandy-Walker malformation, and Klippel-Feil syndrome are causes of non-communicating hydrocephalus.

148. **Nonobstructive or communicating hydrocephalus may follow the following conditions EXCEPT**

A. a subarachnoid hemorrhage
B. pneumococcal meningitis
C. tuberculous meningitis
D. leukemic infiltrates
E. a vein of Galen malformation

Ans E can expand to become large and, because of its midline position, obstruct the flow of CSF.

Neural Tube Defects

149. **Marker of neural tube defects is:**

A. PAPPA
B. hCG
C. Estriol
D. Acetylcholinesterase

Ans D Diagnosis Tools for prenatal diagnosis of Neural tube Defects include detailed ultrasound examinations and measurements of maternal serum α-fetoprotein MSAFP, amniotic fluid a-fetoprotein, and amniotic fluid acetylcholinesterase (AChE).

150. **In a lady with history of birth of a previous child with neural tube defect, amount of folic acid to be given in microgram as secondary prophylaxis before next conception is:**

A. 40
B. 400
C. 4000
D. 500

Ans C • To prevent NTDs, it is recommended that all women of childbearing age, who are capable of becoming pregnant take
• 0.4 mg (400 g) of folic acid daily.
• If a pregnancy is planned in high risk women (previously affected child), supplementation should be started with 4 mg (4000 µmg) of folic acid daily, beginning 1 month before the time of the planned conception.

151. **Folic acid requirement in pregnancy for primary prevention of neural tube defects is:**

A. 200 mg
B. 400 mg
C. 600 mg
D. 800 mg

Ans B 400 g

152. **When should folic acid supplementation start, in order to be effective in preventing neural tube defects?**

A. As soon as pregnancy is diagnosed
B. Before the beginning of 2nd trimester
C. At least 1 month before conception
D. At least 1 week before conception

Ans C

153. **A case of meningomyelocele was posted for surgery. Till the patient is waiting for surgery, the covering of the sac will be protected by a gauze soaked in:**

A. Normal saline
B. Tincture iodine
C. Methylene blue
D. Mercurochrome

Ans A Meningomyelocele is covered with sterile saline-soaked gauze to prevent it from drying, trauma and infection

154. **Neural tube defects have which one of the following inheritance patterns?**

A. Autosomal dominant
B. Autosomal recessive
C. X-linked recessive
D. **Multifactorial**

Ans D Most Neural tube defects show multifactorial inheritance.

155. **Which of the following is a marker for neural tube defects:**

A. Phosphatidyl cholinesterase
B. Pseudo cholinesterase
C. **Acetyl cholinesterase**
D. Butyrylcholinesterase

Ans C

156. **Features of sacral meningomyelocele are all except:**

A. Hydrocephalus
B. Neuropathic bladder
C. **Spastic limbs**
D. Areflexia

Ans C Meningomyelocele in lower sacral region lead to: Bowel and bladder incontinence, Anesthesia in perineal area seen; but no impairment of motor function; So, spastic limbs are not seen.

157. **All of following are neural tube defects except:**

A. Myelomeningocele
B. Anencephaly
C. Encephalocele
D. **Holoprosencephaly**

Ans D Different types of neural tube defects are: Spina bifida occulta, meningocele, encephalocele, myelomeningocele, anencephaly, caudal regression syndrome, dermal sinus, tethered cord, syringomyelia, diastematomyelia and iniencephaly Holoprosencephaly is incomplete separation of the cerebral hemispheres across the midline.

Neurodegenerative Disorders

158. **Most common cause of fetal ventriculomegaly is:**

A. Arnold-Chiari Malformation – I
B. Arnold-Chiari malformation – II
C. **Aqueductal stenosis**
D. Dandy-Walker Malformation

Ans C Obstructive or non-communicating hydrocephalus develops most commonly in children because of an abnormality of the aqueduct of Sylvius or a lesion in the fourth ventricle.

159. **What is the commonest organism causing cerebrospinal fluid shunt related infections?**

A. Staph aureus
B. Gram Negative Bacilli
C. **Coagulase negative Staphylococcus**
D. Enterococci

Ans C CSF shunt infections increase the risk of meningitis caused by staphylococci (especially coagulase-negative species) and other low-virulence bacteria that typically colonize the skin.

160. **The clinical features of eye opening, best verbal response and best motor response in Glasgow coma scale of 7 will best fit as:**

A. **Mutation in sodium channel alpha 1 subunit**
B. Mutation in paired like homeobox 2B gene
C. Mutation in outer dynein arm
D. All of the above

Ans A Eye opening to pain, making incomprehensive sounds and has flexion as best motor response Glassgow coma scale (GCS): It is the most common scoring system used to describe the level of consciousness in a person esp following a traumatic brain injury. Modified GCS for infants and children: (Max score is 15 and minimum is 3) Child Infant Score Eye opening Spontaneous Spontaneous 4 To speech To speech 3 To pain only To pain only 2 No response No response 1 Best verbal response Oriented, appropriate Coos and babbles 5

161. **Smell can be tested reliably at**

A. **32nd wk of gestation**
B. birth
C. three months of age
D. six months of age
E. nine months of age

Ans A Although not a routine component of the examination, smell can be tested reliably as early as the 32nd wk of gestation by presenting a stimulus and observing for an alerting response, withdrawal, or both. Care should be taken to use appropriate stimuli, such as coffee or peppermint, as opposed to strongly aromatic substances (e.g., ammonia inhalants) that stimulate the trigeminal nerve. Each nostril should be tested individually by pinching shut the opposite side.

162. **Dravet syndrome is characterized by all the following EXCEPT**

A. the most severe of the phenotypic spectrum of febrile seizures
B. its onset is in the 1st yr of life
C. seizures subsequently start to occur without fever
D. developmental delay
E. **an autosomal dominant inheritance**

Ans E This syndrome is usually caused by a new mutation, although rarely it is inherited in an autosomal dominant manner. The mutated gene is located on 2q24-31 and encodes for SCN1A, the same gene mutated in GEFS+ spectrum. However, in Dravet syndrome the mutations lead to loss of function and thus to a more severe phenotype.

163. **The following are risk factors for deformational plagiocephaly EXCEPT**

A. **female sex**
B. firstborn child
C. congenital torticollis
D. developmental delay
E. exclusive bottle feeding

Ans A Male sex.

164. **The imaging studies are usually normal in the following stroke-like disorders of children**

A. alternating hemiplegia
B. inborn errors of metabolism
C. hypertensive encephalopathy
D. hypoglycemia
E. global hypoxic–ischemic encephalopathy

Ans A

165. **The characteristic features of Duchenne muscular dystrophy (DMD) are progressive weakness, intellectual impairment, hypertrophy of the calves, and proliferation of connective tissue in muscle. Of the following, the BEST initial test for diagnosis is**

B. muscle biopsy
C. electromyography
D. serum creatine kinase
E. nerve conduction study

Ans D The diagnosis should be confirmed by blood PCR or muscle biopsy in every case.

166. **Miller-Fisher syndrome (MFS) consists of acute external and occasionally internal ophthalmoplegia, ataxia, and areflexia. Of the following, the MOST likely cranial nerve involved is**

A. 3rd
B. 4th
C. 5th
D. 6th
E. 7th

Ans D

Neuromuscular Disorders

167. **Common deformity in Chiari II malformation is:**

A. Syringomyelia
B. Meningomyelocele
C. Hydrocephalus
D. All of above

Ans B c. Hydrocephalus Chiari malformation type I is usually associated with syringomyelia, while type II is usually associated with hydrocephalus & meningomyelocele

168. **Subdural empyema is a complication of all the following conditions except?**

A. Frontal Sinusitis
B. Skull vault osteomyelitis
C. Middle ear disease
D. Boil over face

Ans D Infections of the sinuses, ear and skull bones can lead to Subdural empyema. fo 94. a. Bacterial Meningitis weiveR Hypoglycorrhachia (Low CSF: serum glucose ratio) is commonly seen in Bacterial meningitis.

169. **disorder characterized by monophasic or polyphastic optic neuritis and transverse myelitis. The pathogenesis Includes demonstration of IgG antibodies against astrocyte foot processes within the periventricular region, brainstem, optic nerves and spinal cord. What is the name of these antibodies?**

A. Antineuronal antibody
B. Anticholinesterase antibody
C. Antiglial antibody
D. Anti Aquaporin-4 water channel antibody

Ans C Start oxygen by face mask, immobilize cervical spine and transfer to a tertiary centre accompanied by doctor The best management in this case is to supplement O as there is hypoxia, to prevent further CNS injury; Cervical spine should be immobilized and the patient should be transferred to a tertiary centre accompanied by doctor.

170. **Contraindications to performing a lumbar puncture include the following EXCEPT**

A. suspected mass lesion of the brain
B. suspected mass lesion of the spinal cord
C. symptoms and signs of impending cerebral herniation
D. skin infection at the site of the lumbar puncture
E. thrombocytopenia with a platelet count <50 × 109/L

Ans E Thrombocytopenia with a platelet count <20 × 109/L.

171. **Benign myoclonic epilepsies are often best treated with**

A. clonazepam
B. lamotrigine
C. topiramate
D. valproate
E. benzodiazepines

Ans D Benign myoclonic epilepsies are often best treated with valproate, particularly when patients have associated generalized tonic-clonic and absence seizures. Benzodiazepines, clonazepam, lamotrigine, and topiramate are alternatives for the treatment of benign myoclonic epilepsy.

172. **The ketogenic diet is absolutely contraindicated in**

A. primary carnitine deficiency
B. myoclonic– astatic epilepsy
C. tuberous sclerosis complex
D. Rett syndrome
E. infantile spasms

Ans A The ketogenic diet is absolutely contraindicated in carnitine deficiency (primary); carnitine palmitoyltransferase I or II deficiency; carnitine translocase deficiency; β-oxidation defects; medium-chain acyl dehydrogenase deficiency; long-chain acyl dehydrogenase deficiency; short-chain acyl dehydrogenase deficiency; long- chain 3-hydroxyacyl-coenzyme A deficiency; medium-chain 3-hydroxyacyl-coenzyme A deficiency; pyruvate carboxylase deficiency; and porphyrias.

173. **Small vessel childhood primary angiitis of the CNS (SVcPACNS) is usually presented with**

A. arterial ischemic stroke
B. **seizures**
C. cognitive dysfunction
D. personality changes
E. loss of social control

Ans B are a hallmark of SVcPACNS, as more than 80% of children with SVcPACNS present with seizures; all seizure types are seen.

174. **Duchenne muscular dystrophy (DMD) is the most common hereditary neuromuscular disease affecting all races and ethnic groups. All the following are features of DMD EXCEPT**

A. scoliosis
B. contracture
C. **fasciculation**
D. cardiomyopathy
E. intellectual impairment

Ans C s of muscle, which are often best seen in the tongue, are a sign of denervation is seen in spinal muscular atrophy.

175. **All the following may induce myopathy EXCEPT**

A. steroid use
B. hypothyroidism
C. hyperthyroidism
D. hyperaldosteronism
E. **hypoparathyroidism**

Ans E Most patients with primary hyperparathyroidism develop weakness, fatigability, fasciculation, and muscle wasting that is reversible after removal of the parathyroid adenoma.

176. **Guillain Barré syndrome is an autoimmune disorder often considered a postinfectious polyneuropathy involving mainly motor but also sensory and sometimes autonomic nerves. All the following are required in diagnosis EXCEPT**

A. CSF study
B. **muscle biopsy**
C. electromyography
D. sural nerve biopsy
E. motor nerve conduction study

Ans B is not usually required for diagnosis; specimens appear normal in early stages and show evidence of denervation atrophy in chronic stages.

177. **Charcot-Marie-Tooth disease, the most common genetically determined neuropathy. Of the following, the MOST severely affected nerve is**

A. radial
B. facial
C. femoral
D. **peroneal**
E. mandibular

Ans D The peroneal and tibial nerves are the earliest and most severely affected.

178. **Myasthenia gravis is a chronic autoimmune disease of neuromuscular blockade. Of the following, the MOST characteristic feature is**

A. **fatigue**
B. myalgia
C. fasciculation
D. sensory symptom
E. abnormal pupillary response

Ans A Fasciculations of muscle, myalgias, and sensory symptoms do not occur. Pupillary responses to light are preserved.

179. **Myasthenia gravis should be differentiated from other causes of neuromuscular blockade as organophosphate chemicals, botulism, and tick paralysis. Of the following, the MOST specific diagnostic test is**

A. muscle biopsy
B. **electromyography**
C. anti-AChR antibodies
D. serum creatine kinase
E. nerve conduction study

Ans B Myasthenia gravis is one of the few neuromuscular diseases in which electromyography (EMG) is more specifically diagnostic than a muscle biopsy. A decremental responseis seen to repetitive nerve stimulation; the muscle potentials diminish rapidly in amplitude until the muscle becomes refractory to further stimulation. Anti-AChR antibodies should be assayed in the plasma but are inconsistently demonstrated.

180. **Myotubular myopathy, maturational arrest of fetal muscle during the myotubular stage of development at 8-15 wk of gestation. All the following are a characteristic features EXCEPT**

A. high arch palate
B. **cardiomyopathy**
C. polyhydramnios
D. undescended testes
E. decrease fetal movement

Ans B At birth, affected infants have a thin muscle mass involving axial, limb girdle, and distal muscles; severe generalized hypotonia; and diffuse weakness. Respiratory efforts may be ineffective, requiring ventilatory support. The testes are often undescended. Facial muscles may be weak, but infants do not have the characteristic facies of myotonic dystrophy. Ptosis may be a prominent feature. Ophthalmoplegia is observed in a few cases. The palate may be high.

181. **Malignant hyperthermia is an acute hypermetabolic syndrome that is triggered by inhalational anesthetic agents and succinylcholine. Of the following, which myopathy is consistently associated with malignant hyperthermia?**

A. **Central core myopathy**
B. Myotubular myopathy
C. Nemaline rod myopathy
D. Becker muscular dystrophy
E. Emery-Dreifuss Muscular Dystrophy

Ans A Mutations in the RYR1 gene are the cause of both central core myopathy and malignant hyperthermia.

Neurocutaneous Syndromes

182. **About Dandy-Walker syndrome, true is:**

A. Mostly have hydrocephalus
B. Cystic expansion of 4th ventricle
C. Mid cerebellar hypoplasia
D. **All are true**

Ans D Dandy Walker malformation
• • Cystic expansion of 4th ventricle and midline cerebellar hypoplasia seen; 90% patients have hydrocephalus
• • Papilledema, pyramidal tract signs and cranial nerve palsies may occur along with symptoms of raised ICP.
• • It is managed by shunting the cystic cavity in the presence of hydrocephalus.

183. **Drug of choice for infantile spasms in a child with Tuberous sclerosis is:**

A. Diazepam
B. ACTH
C. Levetiracetam
D. **Vigabatrin**

Ans D Drug of choice for infantile spasms in a child with Tuberous sclerosis is Vigabatrin

184. **Which is not seen in tuberous sclerosis?**

A. Astrocytoma
B. **Ependymoma**
C. White matter lesions
D. Subependymal nodules

Ans B Ghai p 586 Diagnostic criteria of Tuberous sclerosis: Major features (Mnemonic: "PUSH CAR FAST") Minor features P: Pulmonary lymphangioleiomyomatosis F: Facial Angiofibroma Cerebral white matter migration lines U: Ungual or periungual fibroma A: Astrocytoma (giant cell) Multiple dental pits S: Shagreen patch, S: Subependymal nodule Gingival fibromas H: Hypomelanotic macules (>3) T: Tuber (Cortical) Bone cysts/ Multiple renal cysts C: Cardiac rhabdomyoma Retinal achromatic patch

185. **Which is not seen in tuberous sclerosis?**

A. Neurofibromatosis
B. **Sturge-Weber syndrome**
C. Tuberous Sclerosis
D. Incontinentia Pigmenti

Ans B Lisch nodules , Ghai p 586 Answers

186. **Which among the following is the most common tumor associated with neurofibromatosis in a child?**

A. **Juvenile myelomonocytic leukemia**
B. Acute lymphoblastic leukemia
C. Acute monocytic leukemia
D. Acute myeloid leukemia

Ans A Ghai p 586 with Patients with neurofibromatosis type 1 and Noonan syndrome have a predilection for JMML, since they have germline mutations involved in RAS signaling.

187. **Which one of the following is the most common tumor associated with type I neurofibromatosis?**

A. **Optic nerve glioma**
B. Meningioma
C. Acoustic Schwannoma
D. Low grade astrocytoma FEBRILE SEIZURES

Ans A Ghai p 586 Optic gliomas are present in 15% of patients with NF-1 and represent mostly low-grade astrocytomas. They are the main central nervous system tumor with a marked increased frequency in NF-1.

188. **The following are complications of neurofibromatosis (type 1) NF-1 EXCEPT**

A. learning disability
B. **cataracts**
C. seizures
D. precocious puberty
E. scoliosis

Ans B are found in 60-81% of patients with NF-2.

189. **Definite tuberous sclerosis complex (TSC) is diagnosed when at least 2 major or 1 major plus 2 minor features are present, all the following are minor features EXCEPT**

A. **shagreen patch**
B. cerebral white matter migration lines
C. multiple dental pits
D. gingival fibromas
E. bone cysts

Ans A Shagreen patch is a major feature.

190. **The MOST frequent lesion associated with neurofibromatosis type 2 NF-2 is**

A. **bilateral vestibular schwannomas**
B. intracranial
C. cataract
D. retinal hamartoma
E. skin plaque

Ans A Bilateral vestibular schwannomas 90-95%.

Headache

191. **Bulging anterior fontanelle is/are seen in:**

A. **Rickets**
B. CMV infection
C. Scurvy
D. Hypothyroidism
E. Tetracycline therapy

Ans A d. Hypothyroidism, e. Tetracycline therapy Rickets, tetracycline and Hypothyroidism are the causes of pseudotumor cerebri, which leads to manifestations of raised intracranial tension like bulging fontanelle;

192.A CSF/serum glucose ratio of < 0.4 is highly suggestive of:

A. Bacterial Meningitis
B. Viral Meningitis
C. Carcinomatous Meningitis
D. None of the above

Ans A A CSF-to-serum glucose ratio below 0.4 (hypoglycorrhachia) is highly suggestive of bacterial meningitis, reflecting increased glucose consumption by bacteria and leukocytes.

193.True about Migraine in children are all except:

A. Preceding aura
B. Recurrent headache lasting 2–72 hours per episode
C. Bilateral headache
D. Waking up with headache in the morning

Ans D Childhood Migraine weiveR • It is characterized by moderate to severe, episodic headache, focal in location, having throbbing.
• Quality & may be associated with nausea, vomiting, light/sound sensitivity.
• Compared to adults, pediatric migraine is shorter in duration & often has bilateral location.
• Migraine can also be associated with an aura that may be typical (visual, sensory, or dysphasic) or atypical (i.e., hemiplegic, "Alice in Wonderland" syndrome).

194.An elevated polymorphonuclear (PMN) cells count in cerebrospinal fluid (CSF) suggests

A. tuberculous meningitis
B. early phase of aseptic meningitis
C. fungal meningitis
D. demyelinating diseases
E. brain or spinal cord tumor

Ans B Normal CSF contains up to 5/mm3 white blood cells (WBCs), and a newborn can have as many as 15/mm3. Polymorphonuclear (PMN) cells are always abnormal in a child, but 1-2/mm3 may be present in a normal neonate. An elevated PMN count suggests bacterial meningitis or the early phase of aseptic meningitis. CSF lymphocytosis can be seen in aseptic, tuberculous, or fungal meningitis; demyelinating diseases; brain or spinal cord tumor; immunologic disorders, including collagen vascular diseases;

195.Gingival hyperplasia, coarsening of the facies, hirsutism, and cerebellovestibular symptoms (nystagmus and ataxia) are adverse effects of

A. carbamazepine
B. lamotrigine
C. phenytoin
D. valproic acid
E. succinimides

Ans C

196.Neuro-imaging is warranted in a child with headache in the following conditions

A. abnormal neurologic examination
B. afternoon headache
C. headache in children <6 yr old
D. brief cough headache
E. migrainous headache in the child with no family history of migraine

Ans B Neuroimaging is warranted when the neurologic examination is abnormal or unusual neurologic features occur during the migraine; when the child has headaches that awaken him or her from sleep or that are present on first awakening and remit with upright posture; when the child has brief headaches that only occur with cough or bending over; and when the child has migrainous headache with an absolutely negative family history of migraine or its equivalent (e.g., motion sickness, cyclic vomiting).

197. Tension-type headaches (TTH) are characterized by the following EXCEPT

A. diffuse in location
B. not affected by activity
C. throbbing quality
D. mild to moderate in severity
E. less frequently associated with nausea and photophobia

Ans C TTH are mild to moderate in severity, are diffuse in location, are not affected by activity (although the patient may not feel like being active), and are nonthrobbing (often described as a constant pressure).

198.Migraine is the most frequent type of recurrent headache that is brought to the attention of parents and primary care providers, but it remains under recognized and undertreated, particularly in children. Migraine is characterized by all the following EXCEPT

A. episodic attacks
B. moderate to severe in intensity
C. focal in location on the head
D. constant quality
E. may be associated with nausea and vomiting

Ans D Migraine is characterized by episodic attacks that may be moderate to severe in intensity, focal in location on the head, have a throbbing quality, and may be associated with nausea, vomiting, light sensitivity, and sound sensitivity. Compared to adults, pediatric migraine is shorter in duration and has a bilateral, often bifrontal, location. Migraine can also be associated with an aura that may be typical (visual, sensory, or dysphasic) or atypical (i.e., hemiplegic, "Alice in Wonderland" syndrome).

199.The aura associated with migraine is a neurologic warning that a migraine is going to occur. Of the following, the LEAST common type of typical auras is

A. sensory aura
B. dysphasic aura
C. visual aura
D. vertigo
E. distortion

Ans B s are the least-common type of typical aura and have been described as an inability or difficulty to respond verbally.

200.The following are indications for neuroimaging in a child with a headache EXCEPT

A. abnormal neurologic examination
B. headache worst on first awakening
C. brief cough headache
D. **frontal headache**
E. migrainous headache in the child with no family history of migraine

Ans D When the headache is mostly in the occipital area.

Neonatal Seizures

201.Macrocephaly is seen in which of the following syndromes?

A. Metachromatic leukodystrophy
B. Adrenoleukodystrophy
C. **Canavan disease**
D. Krabbe's disease

Ans C Canavan disease
• • Due to deficiency of the enzyme aspartoacylase, that leads to accumulation of N-acetylaspartic acid in brain
• • Usually normal at birth, but develop progressive macrocephaly, hypotonia, persistent head lag and delayed milestones
• • As the disease progresses, there is spasticity, joint stiffness, contractures, optic atrophy and seizures
• • Feeding difficulties, poor weight gain, and gastroesophageal reflux may occur in the 1st year

202.The appearance of cobweb formation in CSF indicates?

A. Pyogenic meningitis
B. Viral meningitis
C. **Tuberculous meningitis**
D. Fungal meningitis

Ans C Cob web coagulum formation in CSF is a feature of Tuberculous meningitis.

203.Xanthochromia of cerebrospinal fluid (CSF) suggests the following EXCEPT

A. **bloody tap**
B. subarachnoid hemorrhage
C. carotenemia
D. hyperbilirubinemia
E. markedly elevated CSF protein

Ans A Normal CSF contains no red blood cells (RBCs); thus, their presence indicates a traumatic tap or a subarachnoid hemorrhage. Progressive clearing of the blood between the first and last samples indicates a traumatic tap. Bloody CSF should be centrifuged immediately. A clear supernatant is consistent with a bloody tap, whereas xanthochromia (yellow color that results from the degradation of hemoglobin) suggests a subarachnoid hemorrhage.

204.Rickets is a potential side effect from all the following EXCEPT

A. phenytoin
B. **valproate**
C. phenobarbital
D. primidone
E. carbamazepine

Ans B Potential side effects are rickets from phenytoin, phenobarbital, primidone, and carbamazepine (enzyme inducers that reduce 25-hyrdroxy-vitamin D level by inducing its metabolism) and hyperammonemia from valproate.

205.The following are features of Aicardi syndrome EXCEPT

A. coloboma of the iris
B. retinal lacunae
C. agenesis of the corpus callosum
D. severe seizures
E. **aminoaciduria**

Ans E Brain malformations account for 5-10% of neonatal seizure cases. An example is Aicardi syndrome, which affects girls only and consists of retinal lacunae, agenesis of the corpus callosum, and severe seizures including subsequent infantile spasms with hypsarrhythmia that is sometimes initially unilateral on EEG.

206.The diagnosis of diffuse CNS infections depends on examination of cerebrospinal fluid (CSF) obtained by lumbar puncture (LP). The cerebrospinal fluid (CSF) protein may reach 3,000 (mg/dL) in

A. acute bacterial meningitis
B. **tuberculous meningitis**
C. fungal meningitis
D. amebic (naegleria) meningoencephalitis
E. subdural empyema

Ans B In tuberculous meningitis, CSF protein is 100-3,000; may be higher in presence of block, while in other options, it may reach 500.

207.In Duchenne muscular dystrophy (DMD), ambulation is important not only for postponing the psychologic depression but also for postponding

A. lordosis
B. **scoliosis**
C. kyphosis
D. ankles contracture
E. pseudohypertrophy

Ans B Ambulation is important not only for postponing the psychologic depression but also because scoliosis usually does not become a major complication as long as a patient remains ambulatory, even for as little as 1 hr per day; scoliosis often becomes rapidly progressive after confinement to a wheelchair.

208.The least significant finding in Miller-Fisher syndrome (MFS) is

A. ataxia
B. areflexia
C. **lower limbs weakness**
D. internal ophthalmoplegia
E. external ophthalmoplegia

Ans C Although areflexia is seen in MFS, patients do not have significant lower extremity weakness compared with Guillain Barré syndrome.

Hypoxic Ischemic Encephalopathy

209.In Arnold-Chiari malformation type-II there is:

A. Herniation of medulla oblongata
B. Herniation of medulla with cerebellum
C. **Meningomyelocele**
D. Aqueductal stenosis

Ans C dna 30. a. Aqueductal stenosis

210.India ink preparation of CSF is done to identify:

A. Streptococcus agalactiae
B. Naegleria fowleri
C. **Cryptococcus neoformans**
D. Coxsackie B

Ans C India ink preparation of CSF is a negative staining technique done to identify Cryptococcus neoformans.

211. Drooping eyelids in evening in boy can be seen in:

A. **Myasthenia gravis**
B. Oculomotor nerve palsy
C. Botulism toxin
D. All of above

Ans A Fatigable ptosis that worsens with sustained activity or as the day progresses is classic for myasthenia gravis.

212.Cranial CT is a valuable diagnostic tool in the evaluation of the following conditions

A. skull fractures
B. intracranial hemorrhages
C. **acute infarcts**
D. hydrocephalus
E. impending herniation

Ans C CT is less useful for diagnosing acute infarcts in children, because radiographic changes might not be apparent for up to 24 hr.

213.The facial port-wine stain of Sturge-Weber syndrome (SWS) is characterized by the following EXCEPT

A. present at birth
B. tends to be unilateral
C. always involves the upper face and eyelid
D. its distribution is consistent with the ophthalmic division of the trigeminal nerve
E. **most children with facial port-wine stain have SWS**

Ans E It is important to note that not all children with facial port-wine stain have SWS. In fact, the overall incidence of SWS has been reported to be 8-33% in those with a port-wine stain.

214.All the following are treatment options for breath-holding spells EXCEPT

A. education and reassurance of the parents
B. iron therapy
C. atropine sulfate
D. antiepileptic drug therapy
E. **providing more care and interest for the patient**

Ans E All parents should be taught not to provide secondary gain when the episodes occur, because this can reinforce the episodes.

215.The organism causing CNS infection is never seen on direct examination of CSF in

A. **tuberculous meningitis**
B. acute bacterial meningitis
C. partially treated bacterial meningitis
D. fungal meningitis
E. amebic (naegleria) meningoencephalitis

Ans A Acid-fast organisms almost never seen on smear. Organisms may be recovered in culture of large volumes of CSF. Mycobacterium tuberculosis may be detected by PCR of CSF.

216.Becker muscular dystrophy (BMD) is a disease that is fundamentally similar to Duchenne muscular dystrophy (DMD). Of the following, the MOST recognized feature of BMD is

A. cardiomyopathy
B. **longer ambulation time**
C. calf pseudohypertrophy
D. elevated creatine kinase
E. genetic defect at the Xp21.2 locus

Ans B In BMD, boys remain ambulatory until late adolescence or early adult life. The other distracters occur in both DMD and BMD.

217. All the following myopathies are characterized by proximal muscle wasting EXCEPT

A. Becker dystrophy
B. **myotonic dystrophy**
C. Duchenne dystrophy
D. central core myopathy
E. hypothyroid myopathy

Ans B The distal distribution of muscle wasting in myotonic dystrophy is an exception to the general rule of myopathies having proximal and neuropathies having distal distribution patterns.

218.Limb-girdle muscular dystrophies (LGMDs) are a heterogeneous group of progressive hereditary muscular dystrophies that mainly affect muscles of the hip and shoulder girdles. Of the following, the presenting complaint may be

A. **low back pain**
B. cardiomyopathy
C. difficulty in swallowing
D. characteristic facies
E. intellectual impairement

Ans A may be a presenting complaint because of the lordotic posture resulting from gluteal muscle weakness. The other distracters are less likely to be occurring.

219.The term congenital muscular dystrophy is misleading because all muscular dystrophies are genetically determined. A distinguishing feature of the congenital dystrophies from other muscular dystrophies, is a high association with malformations of

A. liver
B. lung
C. **brain**
D. bone
E. kidney

Ans C malformations, particularly disorders of cortical development such as lissencephaly/pachygyria and polymicrogyria, often complicated by severe epilepsy.

220.Bell palsy is an acute unilateral peripheral facial nerve palsy that is not associated with other cranial neuropathies or brainstem dysfunction. Of the following, the MOST traditional treatment is

A. acyclovir
B. **prednisone**
C. laser therapy
D. physiotherapy
E. surgical decompression

Ans B Oral prednisone (1 mg/kg/day for 1 wk, followed by a 1 wk taper) started within the 1st 3-5 days results in improved outcome and is a traditional treatment, its efficacy confirmed in a recent long-term prospective study in the United Kingdom.

221.All the following are features of chronic inflammatory demyelinating polyradiculoneuropathies (CIDPs) EXCEPT

A. motor deficits in 94%
B. sensory paresthesias in 64%
C. cranial nerve involvement in <30%
D. **CSF shows pleocytosis and high protein**
E. hyporeflexia or areflexia is almost universal

Ans D Patients are usually severely weak and can have a flaccid tetraplegia with or without bulbar and respiratory muscle involvement. Cerebrospinal fluid (CSF) shows no pleocytosis and protein is variably normal or mildly elevated.

Intraventricular Hemorrhage

222.Arnold chiari malformation is associated with:

A. **Meningomyelocele**
B. Leptomeningeal cyst
C. Agenesis of corpus callosum
D. Dandy-Walker anomaly

Ans A

223.A child presents with generalized petechiae. CSF shows gram negative diplsococci. Treatment:

A. **IV Ceftriaxone**
B. IV Penicillin G
C. IV Cefotaxime
D. IV Amoxicillin
E. IV Ofloxacin

Ans A b. IV Penicillin G, c. IV Cefotaxime The given clinical and laboratory findings are suggestive of Meningococcal infection Most strains of N. meningitidis are sensitive to penicillin and cephalosporins.

224.Cerebral perfusion pressure in children is:

A. 11-15 mm Hg
B. 16-20 mm Hg
C. 20-40 mm Hg
D. **40-60 mm Hg**

Ans D Cerebral perfusion pressure (CPP) is calculated as the difference between the systemic mean arterial pressure (MAP) and the intracranial pressure (ICP); So, CPP = MAP – ICP The normal range of CPP is thought to be approximately 50 to 150 mm Hg in healthy adults, and 40 to 60 mm Hg, in children.

225.An approach to imaging of the spine in patients with cutaneous lesions is indicated in the following conditions EXCEPT

A. hairy patch
B. subcutaneous mass or lipoma
C. dermal sinus
D. **coccygeal pits**
E. scarlike lesions

Ans D Imaging of the spine is not indicated in patients with simple dimples (<5 mm, <25 mm from anal verge) or coccygeal pits.

226.All the following are features of Von Hippel–Lindau (VHL) disease EXCEPT

A. its incidence is around 1: 36,000
B. **fifty percent have a de novo gene mutation**
C. hemangioblastoma of the spinal cord may be found
D. renal carcinoma is the most common cause of death
E. pheochromocytoma is a frequent association

Ans B Around 80% of individuals with VHL disease have an affected parent, and around 20% have a de novo gene mutation.

227.Café-au-lait macules are not specific for NF-1; they may be seen in the following

A. Noonan syndrome
B. ataxia telangiectasia
C. Fanconi anemia
D. Gaucher disease
E. **Hurler syndrome**

Ans E Hunter syndrome.

228.T-lymphocyte defects (congenital or acquired by chemotherapy, AIDS, or malignancy) are associated with an increased risk of infections of the CNS with

A. **Listeria monocytogenes**
B. Streptococcus pneumoniae
C. Neisseria meningitidis
D. Haemophilus influenzae type b
E. coagulase-negative Staphylococci

Ans A

229.Emery-Dreifuss Muscular Dystrophy (scapuloperoneal or scapulohumeral muscular dystrophy) is a rare X-linked recessive dystrophy. Of the following, the MOST characteristic feature is

A. myotonia
B. facial weakness
C. pseudohypertrophy
D. intellectual impairment
E. dilated cardiomyopathy

Ans E Also characterized by contractures of elbows and ankles that develop early, and muscle becomes wasted in a scapulohumeroperoneal distribution. The serum CK value is only mildly to moderately elevate.

230.The best diagnostic test in Guillain-Barré syndrome (GBS) is

A. CSF study
B. electromyography
C. sural nerve biopsy
D. serum creatine kinase
E. motor nerve conduction study

Ans A The dissociation between high CSF protein and a lack of cellular response in a patient with an acute or subacute polyneuropathy is diagnostic of Guillain-Barré syndrome.

Periventricular Leukomalacia

231.The following are recognized signs and symptoms of raised intracranial tension in a 9-month-old infant, except:

A. Bulging fontanel
B. Setting-sun sign
C. Papilledema
D. Increase in head size

Ans C Papilledema is usually not seen in infancy, as the fontanelle are open

232.After 5 days of birth, baby developed poor feeding, convulsions, fever with low protein low sugar and high chloride in CSF, is most likely due to:

A. L. monocytogenes
B. M. pneumoniae
C. TB
D. Leptospira MISCELLANEOUS Questions

Ans A Out of the given options, most common cause of neonatal meningitis is: Listeria monocytogenes.
MISCELLANEOUS

233.Regarding myelomeningocele, all the following are true EXCEPT

A. risk of recurrence after one affected child is 3-4%
B. maternal periconceptional use of folic acid supplementation reduces the incidence of neural tube defects (NTDs) in pregnancies at risk by at least 50%
C. anticonvulsant valproic acid causes NTDs in approximately 1-2% of pregnancies when administered during pregnancy
D. hydrocephalus in association with a type II Chiari malformation develops in at least 80% of patients with myelomeningocele
E. the lower the deformity is in the neuraxis (sacrum), the more likely is the risk of hydrocephalus

Ans E Generally, the lower the deformity is in the neuraxis (sacrum), the less likely is the risk of hydrocephalus.

234.PHACE syndrome denotes to the following EXCEPT

A. posterior fossa malformations
B. hemangiomas
C. anal anomalies
D. coarctation of the aorta
E. eye abnormalities

Ans C Arterial anomalies.

235.Slow, writhing, continuous, and involuntary movements are called

A. stereotypies
B. tics
C. tremor
D. athetosis
E. chorea

Ans D

236.The CSF leukocyte count in normal healthy neonates may reach

A. 10 leukocytes/mm3
B. 15 leukocytes/mm3
C. 20 leukocytes/mm3
D. 25 leukocytes/mm3
E. 30 leukocytes/mm3

Ans E Normal healthy neonates may have as many as 30 leukocytes/mm3 (usually <10), but older children without viral or bacterial meningitis have <5 leukocytes/mm3 in the CSF; in both age groups there is a predominance of lymphocytes or monocytes.

237.Myotonic muscular dystrophy (Steinert disease) is the second most common muscular dystrophy in North America, Europe, and Australia, inherited as an autosomal dominant trait. Of the following, the LEAST recognized feature is

A. cataract
B. myotonia
C. low serum IgG
D. cardiomyopathy
E. intellectual impairment

Ans D Cardiac involvement is usually manifested as heart block in the Purkinje conduction system and arrhythmias (and sudden death) rather than as cardiomyopathy, unlike most other muscular dystrophies.

238.Patients in early stages of Guillain-Barré syndrome should be admitted to the hospital for observation because the ascending paralysis can rapidly involve respiratory muscles during the next 24 hr. Of the following, the MOST effective treatment is

A. steroids
B. plasmapheresis
C. jejuni
D. immunosuppressive drugs
E. intravenous immunoglobulin (IVIG)

Ans E IVIG administered for 2, 3, or 5 days. A commonly recommended protocol is IVIG 0.4 g/kg/day for 5 consecutive days, but some studies suggest that larger doses are more effective (1g/kg/day for 2 consecutive days) and related to improved outcome.

Developmental Delay

239.MacEwen sign is seen in:

A. **Hydrocephalus**
B. Encephalitis
C. Meningitis
D. Microcephaly

Ans A In Hydrocephalus, percussion of the skull might produce a cracked pot sound or MacEwen sign, indicating separation of the sutures.

240.Most common cause of syncope in a child:

A. Breath holding spell
B. Tantrum spell
C. Hypoglycemia
D. **Neurocardiogenic**

Ans D Syncope is sudden transient loss of consciousness with inability to maintain postural tone. Most common cause of syncope in the normal pediatric population is neurocardiogenic syncope, also known as vasovagal syncope. Syncope is caused by a lack of adequate cerebral blood flow.

241.Puff of smoke appearance on cerebral angiography is seen in which of the following condition:

A. Takayasu's disease
B. Arteriovenous malformation
C. **Moyamoya disease**

Ans C Puff of smoke appearance on cerebral angiography is seen in Moyamoya disease.

242.Mobius syndrome is characterized by the following EXCEPT

A. bilateral facial weakness
B. paralysis of the abducens nerve
C. hypoplasia or agenesis of brainstem nuclei
D. feeding difficulties
E. **mental retardation**

Ans E The immobile, dull facies might give the incorrect impression of mental retardation; the prognosis for normal development is excellent in most cases.

243.The following are manifestations of increased intracranial pressure in an infant

A. increasing head circumference
B. bulging fontanel
C. failure to thrive
D. **persistent vomiting**
E. setting sun sign

Ans D The manifestations of increased intracranial pressure in an infant as compared with an older child are: Infant: Increasing head circumference, delayed closure of the fontanel, suture separation, bulging fontanel, failure to thrive, macrocephaly, setting sun sign, and shrill cry.

244.A slowly progressive ataxia that involves the lower extremities to a greater degree than the upper extremities. The Romberg test result is positive; the deep-tendon reflexes are absent (particularly at the ankle), and the plantar response is typically extensor (Babinski sign). Of the following, the MOST likely cause of this ataxia is

A. ataxia-telangiectasia
B. **Friedreich ataxia**
C. abetalipoproteinemia
D. Roussy-Levy disease
E. Ramsay Hunt syndrome

Ans B is inherited as an autosomal-recessive disorder involving the spinocerebellar tracts, dorsal columns in the spinal cord, the pyramidal tracts, and the cerebellum and medulla. The onset of ataxia is somewhat later than in ataxiatelangiectasia, but usually occurs before age 10 yr. The ataxia is slowly progressive and involves the lower extremities to a greater degree than the upper extremities. The Romberg test result is positive;

245.Detection of viral DNA or RNA by polymerase chain reaction is the test of choice in the diagnosis of CNS infection caused by

A. arboviruses
B. **parechoviruses**
C. West Nile virus (WNV)
D. rabies virus
E. Epstein-Barr virus

Ans B Detection of viral DNA or RNA by polymerase chain reaction is the test of choice in the diagnosis of CNS infection caused by HSV, parechovirus, and enteroviruses, respectively. CSF serology is the diagnostic test of choice for WNV.

246.In periodic paralysis, all the following are precipitating factors EXCEPT

A. **heavy fat meal**
B. licorice ingestion
C. emotional stress
D. hyperthyroidism
E. amphotericin B use

Ans A Heavy carbohydrate meal.

247.The last function to be recovered in Guillain-Barré syndrome (GBS) is

A. sensation
B. bulbar muscles
C. anal sphincters
D. **tendon reflexes**
E. respiratory muscles

Ans D The clinical course is usually benign, and spontaneous recovery begins within 2- 3 wk. The tendon reflexes are usually the last function to recover. Improvement usually follows a gradient opposite the direction of involvement: bulbar function recovering first, and lower extremity weakness resolving last.

Autism Spectrum Disorder

248.Lisch nodules are seen in which of the following conditions?

A. von Recklinghausen's disease
B. Louis-Bar syndrome
C. Tuberous sclerosis
D. von Hippel-Lindau syndrome

Ans A von Recklinghausen's disease Von Recklinghausen's disease is the other name for Neurofibromatosis type I.

249.You have been called to declare a brain dead 12-year- old child in PICU. All of the given are signs of brain death except:

A. Normal BP without pharmacological support
B. Positive spinal reflexes on stimulation
C. Sweating and tachycardia
D. Decorticate and decerebrate posturing

Ans D • Brain death is the irreversible cessation of all functions of the entire brain, including the brainstem.
• 3 key components of clinical brain death diagnosis are demonstrations of coexisting irreversible coma with a known cause, absence of brainstem reflexes, and apnea.
• To establish diagnosis of brain death, findings must remain consistent for 2 examinations separated by an observation period of 24 hr for neonates & 12 hr for infants & older children

250.Dandy-Walker malformation is characterized by the following EXCEPT

A. cystic dilatation of the fourth ventricle
B. herniation of the cerebellar tonsils though the foramen magnum
C. hypoplasia of the cerebellar vermis
D. hydrocephalus
E. an enlarged posterior fossa

Ans B Chiari malformation is the most common malformation of the posterior fossa and hindbrain. It consists of herniation of the cerebellar tonsils though the foramen magnum.

251.Characteristic features of pseudotumor cerebri include the following EXCEPT

A. headache
B. stiff neck
C. papilledema
D. fatigue
E. abnormal CSF profile

Ans E Normal CSF profile with the exception of an elevated opening pressure.

252.The MOST common acquired cause of chorea in childhood is

A. Huntington disease
B. paraneoplastic choreas
C. benign hereditary chorea
D. Sydenham chorea
E. chorea gravidarum

Ans D (St. Vitus dance) is the most common acquired chorea of childhood. It occurs in 10-20% of patients with acute rheumatic fever, typically weeks to months after a group A β-hemolytic streptococcal infection. Peak incidence is at age 8-9 yr, with a female predominance of 2:1.

253.Idiopathic intracranial hypertension, also known as pseudotumor cerebri, is a clinical syndrome that mimics brain tumors and is characterized by the following

A. increased intracranial pressure ≥280 mm Hg in nonobese, nonsedated children
B. normal cerebrospinal fluid (CSF) cell count and protein content
C. normal to slightly decreased ventricular size
D. normal ventricular anatomy and position documented by MRI
E. papilledema

Ans A Increased intracranial pressure ≥280 mm Hg in sedatedor obese children; ≥250 mm Hg in nonobese, nonsedated children.

254.In spinal muscular atrophy (SMA) type 1, all the following are spared EXCEPT

A. heart
B. diaphragm
C. intelligence
D. anal sphincter
E. extraocular muscles

Ans B In severe infantile form, also known as Werdnig-Hoffmann disease or SMA type 1, there is diaphragmatic involvement, even late.

255.Bell palsy usually develops abruptly about 2 wk after a systemic viral infection. Of the following, the MOST common viral cause is

A. Mumps virus
B. Cytomegalovirus
C. Epstein-Barr virus
D. Herpes simplex virus
E. Human herpes virus

Ans D Active or reactivation of herpes simplex or varicella-zoster virus may be the most common cause of Bell palsy.

ADHD

256.Lisch nodule is seen in?

A. Tuberous sclerosis
B. Sturge-Weber syndrome
C. Neurofibromatosis
D. Wilson disease

Ans C Important clinical features of Neurofibromatosis (NF1) are cafe-au-lait spots, neurofibromas, axillary freckling, Lisch nodules optic glioma and positive family history

257.A neonate with left to Right shunt & high cardiac output heart failure. CT Angiography of the neonate is shown. What is the diagnosis?

A. Sagittal venous thrombosis
B. Vein of Galen malformation
C. Intracranial hemorrhage
D. Arnold-Chiari malformation

Ans B CT angiography shows enhancing rounded lesion at tentorial apex, extending posteriorly & draining into dilated superior sagittal sinus, through dilated straight sinus, suggestive of vein of Galen malformation. The anomaly is actually due to a cerebral arteriovenous fistula of the median prosencephalic vein (a precursor of the vein of Galen) occurring at 6-11 weeks gestation. b. Macrocephaly Microcephaly, Growth retardation, ptosis, absent philtrum, congenital heart disease (ASD), feeding problems, neuroglial

258. **Wide open anterior fontanelle is found in following diseases except:**

A. Rickets
B. Cretinism 141.
C. Osteogenesis imperfecta
D. **Craniosynostosis**

Ans D In Craniosynostosis there is premature fusion of cranial sutures and so fontanelles close early and are small; While, Rickets, Cretinism (Hypothyroidism), Ostoeogenesis imperfecta & Achondroplasia are important causes of large anterior fontanelle. b. Acetazolamide

259. **Familial (autosomal recessive) microcephaly is characterized by the following**

A. slanted forehead
B. prominent nose and ears
C. **mild or borderline mental retardation**
D. prominent seizures
E. surface convolutional markings of the brain

Ans C Familial (autosomal recessive) microcephaly is characterized by severe mental retardation.

260. **How often are EEGs abnormal in healthy children?**

A. 5%
B. **10%**
C. 15%
D. 20%
E. 25%

Ans B Approximately 10% of "normal" children have mild, nonspecific abnormalities in background activity.

261. **Prematurity is a major cause of**

A. **spastic diplegia**
B. spastic quadriplegia
C. spastic hemiplegia
D. athetoid CP
E. dyskinetic CP

Ans A Prematurity, ischemia, infection, and endocrine/metabolic (e.g., thyroid) are major causes of spastic diplasia.

262. **The following are hematologic causes of childhood pseudotumor cerebri EXCEPT**

A. Wiskott-Aldrich syndrome
B. **megaloblastic anemia**
C. polycythemia
D. sickle cell disease
E. Fanconi anemia

Ans B Iron-deficiency anemia.

263. **Muscle denervation is any loss of nerve supply regardless of the cause. Of the following, the MOST specific clinical sign of denervation is**

A. myotonia
B. weakness
C. fatigability
D. **fasciculation**
E. abnormal sensation

Ans D s of muscle, which are often best seen in the tongue, are a sign of denervation. Sensory abnormalities indicate neuropathy. Fatigable weakness is characteristic of neuromuscular junctional disorders. Myotonia is specific for a few myopathies.

264. **All the following are preserved in Charcot-Marie-Tooth disease EXCEPT**

A. intelligence
B. **tibial nerves**
C. axial muscles
D. cranial nerves
E. anal sphincter

Ans B The peroneal and tibial nerves are the earliest and most severely affected. Children with the disorder are often described as being clumsy, falling easily, or tripping over their own feet. Muscles of the anterior compartment of the lower legs become wasted, and the legs have a characteristic stork-like contour. The muscular atrophy is accompanied by progressive weakness of dorsiflexion of the ankle and eventual footdrop. The process is bilateral but may be slightly asymmetric.

22

CHAPTER 22

Behavioral Disorders in Children



WHAT THIS CHAPTER COVERS

- Autism Spectrum Disorder
- ADHD
- Learning Disabilities
- Intellectual Disability
- Conduct Disorder & ODD
- Anxiety Disorders
- Depression & Mood Disorders
- Eating Disorders
- Elimination Disorders
- Tic Disorders & Tourette
- Schizophrenia & Psychosis
- Substance Abuse
- School Refusal
- Psychopharmacology

119 MCQs **14** subtopics

22.1 Autism Spectrum Disorder (ASD)

- **DSM-5 criteria**: **(A) persistent deficits in social communication & interaction** (social-emotional reciprocity, non-verbal communication – poor eye contact, lack of joint attention/pointing, developing relationships) **AND (B) restricted, repetitive patterns of behaviour, interests, activities** (≥ 2 of: stereotyped movements/speech – hand-flapping, echolalia; **insistence on sameness/routines**; highly restricted fixated interests; **hyper/hypo-reactivity to sensory input**); **onset in early developmental period**; causes impairment.
- **Prevalence ~1 in 36–100**; **boys 4:1**; genetic (high heritability), **fragile X (MC single-gene cause)**, tuberous sclerosis, Rett, Angelman, 15q dup, **advanced parental age**, prematurity, **maternal valproate**, congenital rubella. **Vaccines (MMR) do NOT cause autism.**
- **Earliest signs (12–18 m)**: **lack of joint attention (not pointing to show/share – "protodeclarative pointing" by 14 m)**, **no response to name**, poor eye contact, **no social smile/babbling**, **regression of language/social skills at 15–24 m (in 25–30%)**, lining up toys, unusual sensory interests.
- **Screening: M-CHAT-R/F at 18 & 24 months** (AAP); India – **INDT-ASD**, INCLEN tools; diagnosis – **ADOS-2 (gold standard observation)**, ADI-R, **CARS** (Childhood Autism Rating Scale), **ISAA (Indian Scale for Assessment of Autism)** for disability certification.
- **Associated**: **ID (~30–40%)**, epilepsy (25%; bimodal – infancy & adolescence), ADHD, anxiety, sleep problems, GI symptoms, selective eating (scurvy, rickets), self-injury, macrocephaly.
- **Work-up**: **hearing assessment (first)**, **chromosomal microarray (first-tier genetic test)**, fragile X DNA test (boys & girls), EEG if seizures/regression, MRI if focal signs/microcephaly, metabolic tests if suggestive.
- **Management**: **early intensive behavioural intervention – Applied Behaviour Analysis (ABA)**, Early Start Denver Model, **speech & language therapy**, occupational therapy (sensory integration), **TEACCH**, parent training, special education; **pharmacotherapy for associated symptoms only – risperidone & aripiprazole (FDA-approved for irritability/aggression)**, methylphenidate (hyperactivity), melatonin (sleep), SSRIs (anxiety).
- Good prognostic factors: **IQ > 70, functional language by 5 years**, early intervention.
- **Asperger syndrome (now within ASD)** – no language delay, normal/high IQ, social difficulty, circumscribed interests, motor clumsiness.

22.2 ADHD (Attention-Deficit/Hyperactivity Disorder)

- **MC neurobehavioural disorder of childhood** (5–7%); boys 3:1 (girls – inattentive type).
- **DSM-5**: **≥ 6 symptoms (≥ 5 if ≥ 17 y) of inattention and/or hyperactivity-impulsivity for ≥ 6 months**, **several symptoms present before 12 years**, **in ≥ 2 settings (home, school)**, impairing function, not better explained by another disorder.
- Types: **combined (MC)**, predominantly inattentive, predominantly hyperactive-impulsive.
- **Essential feature**: developmentally inappropriate **inattention, hyperactivity & impulsivity**.
- Rating scales: **Conners, Vanderbilt (parent & teacher)**; **diagnosis is clinical** (no lab test).
- Comorbidity: **learning disorders (MC), ODD/conduct disorder**, anxiety, depression, tics, ASD, sleep.
- Differentials: hearing/vision loss, absence epilepsy, OSA, hyperthyroidism, lead poisoning, **iron deficiency**, anxiety, family stress, drugs (phenobarbitone, antihistamines, β-agonists).

TREATMENT

- **< 6 years (preschool): parent training in behaviour management (PTBM) – first line**; methylphenidate if severe.
- **≥ 6 years: stimulants – methylphenidate (DOC)**, amphetamines (lisdexamfetamine) **+ behavioural therapy & school interventions**.
- **Non-stimulants**: **atomoxetine** (NE reuptake inhibitor – tic disorders, anxiety, substance abuse risk; **suicidal ideation, hepatotoxicity**), **α2-agonists (guanfacine ER, clonidine ER)** – tics, sleep, aggression; viloxazine.
- **Stimulant side effects**: **anorexia/weight loss (MC), insomnia**, headache, abdominal pain, ↑ HR/BP, **growth suppression (height – modest)**, **tics (unmasking)**, irritability, rebound; abuse potential; cardiac screening (history ± ECG) before starting. **Drug holidays** during school vacations for growth.

22.3 Learning Disabilities (Specific Learning Disorder) & Intellectual Disability

- **Specific learning disorder**: difficulty in **reading (dyslexia – MC, 80%), written expression (dysgraphia), mathematics (dyscalculia)** despite **normal intelligence**, adequate schooling & normal senses; **persistent ≥ 6 months despite targeted intervention**; boys > girls; often with ADHD.
- **Dyslexia** – phonological processing deficit; letter reversals beyond 7 y (b/d), slow reading; **dyspraxia (developmental coordination disorder)** – clumsy child.
- Assessment: **NIMHANS SLD battery**, educational psychologist; Rx **remedial education (special educator), accommodations (extra time, scribe, exemption from 2nd language – CBSE/RPWD Act 2016)**.
- **Intellectual disability (intellectual developmental disorder)** – deficits in intellectual (IQ ~< 70) **and adaptive functioning** with onset in developmental period; severity by **adaptive functioning (DSM-5)**; **mild (85%)** – educable; moderate – trainable; severe/profound – need supervision.
  - **MC genetic cause: Down syndrome**; **MC inherited: fragile X**; **MC preventable: iodine deficiency/ congenital hypothyroidism**; **fetal alcohol syndrome – MC preventable non-genetic cause (West)**.
  - Work-up: **CMA (first-tier)**, fragile X, thyroid, metabolic (if indicated), MRI (if microcephaly/focal signs), EEG (seizures), hearing/vision.
  - Indian tests: **Malin's Intelligence Scale for Indian Children (MISIC – Indian adaptation of WISC)**, **Binet–Kamat Test**, Seguin Form Board (non-verbal performance), VSMS (social quotient).

22.4 Disruptive Behaviour – ODD & Conduct Disorder

	Oppositional defiant disorder (ODD)	Conduct disorder (CD)
Features	**Angry/irritable mood, argumentative/ defiant behaviour, vindictiveness** towards authority ≥ 6 months	**Violation of others' rights & societal norms** – aggression to people/ animals (cruelty), destruction of property (fire-setting), deceitfulness/theft, **serious rule violations (truancy, running away)** ≥ 12 months
Age	Preschool–school	Childhood-onset (< 10 y – worse prognosis) / adolescent-onset
Outcome	May progress to CD	**Antisocial personality disorder in adulthood (if > 18 y)**, substance abuse
Rx	**Parent management training**	Multisystemic therapy, parent training, CBT; risperidone for aggression

22.5 Anxiety, Mood & Other Emotional Disorders

- **Separation anxiety disorder** – **MC anxiety disorder in children < 12 y**; excessive distress on separation from attachment figure ≥ 4 weeks; school refusal (somatic complaints on school days), nightmares.

- **School refusal vs truancy**: **school refusal – anxiety, child stays at home with parents' knowledge, good student**; **truancy – conduct problem, hides from parents, poor performance**.
- **Selective mutism** – consistent failure to speak in specific social situations (school) despite speaking elsewhere; anxiety-related.
- Generalised anxiety, specific phobias (**MC anxiety disorder overall**), social anxiety, panic, **OCD** (prepubertal boys; PANDAS); Rx **CBT – first line**, **SSRIs (fluoxetine, sertraline, fluvoxamine – OCD)**.
- **Depression in children**: **irritable mood (rather than sad)**, somatic complaints, school decline, social withdrawal, sleep/appetite changes; **adolescents – suicide risk**; Rx **CBT/IPT, fluoxetine (FDA-approved ≥ 8 y)/ escitalopram (≥ 12 y)** – **black-box warning: ↑ suicidal ideation** (monitor).
- **Disruptive mood dysregulation disorder (DMDD)** – severe recurrent temper outbursts (≥ 3/week) + persistent irritability; age 6–18, onset < 10 y.
- **Bipolar disorder** – rare before puberty; lithium (≥ 12 y), atypical antipsychotics.
- **Post-traumatic stress disorder**, adjustment disorders, **reactive attachment disorder** (neglect/institutional care – inhibited, withdrawn), **disinhibited social engagement disorder** (indiscriminate friendliness).
- **Suicide in adolescents** – leading cause of death in Indian adolescents (15–19 y); risk factors – depression, previous attempt (strongest), substance use, bullying, academic stress, family conflict, LGBTQ, access to means; **ask directly; safety planning; Tele-MANAS 14416**.

22.6 Eating & Feeding Disorders

Disorder	Key features
Anorexia nervosa	**Adolescent girls (peak 13–18 y)**; **restriction of energy intake → significantly low weight (BMI < 5th centile)**, **intense fear of weight gain, disturbed body image**; restricting vs binge-purge type; **bradycardia (MC CVS sign), hypotension, hypothermia, lanugo, amenorrhoea (no longer required in DSM-5), osteopenia, hypokalaemia (purging), prolonged QTc**, ↑ cholesterol, ↓ T3 (euthyroid sick), delayed gastric emptying; **highest mortality of psychiatric disorders (cardiac, suicide)**; Rx **family-based treatment (Maudsley) – first-line in adolescents**, nutritional rehabilitation (**watch refeeding syndrome – hypophosphataemia**), CBT-E; hospitalisation – HR < 50 (day)/< 45 (night), BP < 90/45, electrolyte disturbance, syncope, < 75% median BMI
Bulimia nervosa	Recurrent **binge eating + compensatory behaviour (vomiting, laxatives, exercise)** ≥ 1/week × 3 months; **normal/↑ weight**; **Russell sign (knuckle calluses), parotid enlargement, dental erosion**, hypokalaemic hypochloraemic alkalosis; Rx CBT, **fluoxetine**
Binge-eating disorder	Bingeing without compensation; obesity
ARFID (avoidant/ restrictive food intake disorder)	Restriction due to sensory aversion/fear of choking/lack of interest – **no body image disturbance**; younger children, ASD
Pica	**Persistent eating of non-nutritive substances (mud, chalk, paint, paper, ice) ≥ 1 month**, developmentally inappropriate (> 2 y); **iron deficiency (MC association), zinc deficiency**, ID, autism, pregnancy; complications – **lead poisoning, parasitic infection (toxocara), bezoars**; Rx treat deficiency, behaviour therapy
Rumination disorder	Repeated regurgitation & re-chewing of food; infants (neglect) & ID

22.7 Elimination Disorders

- **Enuresis** – see Renal chapter (age ≥ 5 y; **alarm = TOC; desmopressin = DOC**).
- **Encopresis** – faecal soiling ≥ 4 y; **retentive (MC – constipation with overflow)**; Rx disimpaction + laxatives + toilet training/behaviour therapy.

22.8 Tic Disorders & Tourette Syndrome

- **Tics**: **sudden, rapid, recurrent, non-rhythmic** motor movements or vocalisations; **suppressible temporarily (premonitory urge)**, ↑ with stress/fatigue, ↓ with concentration, **may persist in sleep (reduced)**; **simple (eye blinking – MC first tic, throat clearing)** / complex (coprolalia, echolalia, echopraxia).
- **Provisional tic disorder** < 1 year; **persistent (chronic) motor or vocal tic disorder** ≥ 1 year; **Tourette syndrome – multiple motor + ≥ 1 vocal tics for > 1 year, onset before 18 y (usually 5–7 y)**; peak severity 10–12 y, improves in adolescence.
- Comorbid: **ADHD (MC – 50–60%), OCD**, anxiety, learning problems.
- **Rx**: psychoeducation; **CBIT (comprehensive behavioural intervention for tics – habit reversal) – first line**; drugs – **α2-agonists (clonidine, guanfacine – first-line, esp. with ADHD)**, **aripiprazole**, risperidone, haloperidol/pimozide (FDA-approved), tetrabenazine, topiramate.

22.9 Habit Disorders, Sleep & Others

Behaviour	Key points
Thumb sucking	Normal till 3–4 y; persistent beyond **4–5 y → dental malocclusion (anterior open bite)**; Rx positive reinforcement, bitter nail paint, dental appliance (crib)
Nail biting	MC habit in school-age; anxiety
Bruxism	Teeth grinding in sleep (N2); anxiety, malocclusion
Temper tantrums	Peak 2–3 y; **ignore (planned ignoring)/time-out**, prevent triggers; persistent > 5 y → evaluate
Breath-holding spells	See Neurology; iron therapy
Head banging/body rocking	Sleep onset, < 4 y; usually benign; ASD/ID if persistent
Self-stimulation (infantile masturbation)	Toddlers; rhythmic thigh adduction, flushing, grunting – mimics seizure; reassurance
Stuttering (childhood-onset fluency disorder)	Repetitions, prolongations, blocks; onset 2–5 y; boys; **developmental dysfluency (normal) at 2–4 y**; speech therapy if persists > 6 m or > 5 y
Night terrors / nightmares / sleepwalking	See Development (NREM vs REM)
Narcolepsy	Excessive daytime sleepiness, **cataplexy**, hypnagogic hallucinations, sleep paralysis; ↓ **CSF hypocretin (orexin)**; MSLT

22.10 Schizophrenia, Psychosis & Substance Use

- **Childhood-onset schizophrenia (< 13 y)** – rare, insidious, poor prognosis; **early-onset (< 18 y)**; hallucinations (auditory), delusions, disorganisation, negative symptoms; **22q11 deletion** association; Rx **atypical antipsychotics (risperidone, aripiprazole, olanzapine; clozapine – refractory)**; monitor metabolic side effects.
- Exclude organic causes: **anti-NMDA receptor encephalitis**, SLE, Wilson, drugs (cannabis, stimulants), steroids, delirium.
- **Substance use in adolescents**: **tobacco, alcohol (MC substances), cannabis, inhalants (solvents – "whitener", glue; Indian street children – sudden sniffing death, neuropathy, leukoencephalopathy)**, opioids; **CRAFFT screening tool**; brief intervention, motivational interviewing; India – **NDPS Act**, COTPA (no tobacco sale < 18 y or within 100 yards of schools).

22.11 Child Rights, Abuse & Disability (behavioural context)

- **Child abuse forms**: physical, sexual, emotional, **neglect (MC form)**; **Munchausen syndrome by proxy (factitious disorder imposed on another)** – caregiver fabricates/induces illness (apnoea, bleeding, poisoning).
- **POCSO Act 2012** (sexual offences, < 18 y, mandatory reporting); **Juvenile Justice Act 2015**; **RPWD Act 2016 – 21 disabilities** (incl. autism, SLD, ID, CP, thalassaemia, haemophilia, sickle cell); **Right to Education Act 2009 (6–14 y)**; **Child Labour (Amendment) Act 2016** – < 14 y prohibited; childline **1098**.
- **Rashtriya Bal Swasthya Karyakram (RBSK, 2013)** – screening of children 0–18 y for **"4 Ds" – Defects at birth, Deficiencies, Diseases, Developmental delays including disabilities** (32 conditions); **District Early Intervention Centres (DEIC)**.

Practice MCQs · Behavioral Disorders in Children

119 questions · correct option highlighted in green · cover the answer strip to self-test

Autism Spectrum Disorder

1. True about Asperger's syndrome is:

A. Cognitively delayed
B. Normal social interaction
C. **Language skills relatively intact**
D. Normal occupational functioning

Ans C

2. True about Autistic disorder:

A. **Quality decrease in social interaction**
B. All affected children have subnormal intelligence
C. Treatment should be targeted toward speech development
D. Seen only after 3 yr of age
E. Stereotyped patterns of behavior

Ans A and e. Stereotyped patterns of behavior 182 ASD symptoms are typically recognized during the 2nd yr of life but can been seen earlier than 12 months, if it is severe. b. Repetitive activity pattern Discussing about the options one by one a. More common in girl Autistic spectrum disorder is more common in boys b. Repetitive activity pattern True c. Subnormal intelligence is consistent feature Intelligence may or may not be normal d.

3. True about autism:

A. Occurs in high economic strata
B. Normal communication
C. **Starts before 2-3 years of age**
D. More common in girls

Ans C Autism is more common in lower socioeconomic strata, more common in boys; Communication is impaired in Autism.

4. Which of the following is not true about the autistic spectrum disorder?

A. Impaired communication
B. Impaired imagination
C. Language developmental delay
D. **Vision problems**

Ans D

5. Autism is:

A. **Pervasive developmental disorder**
B. Attention deficit hyperactive disorder
C. Psychotic disorder
D. Neurotic disorder

Ans A

6. The following are characteristics of autism except:

A. **Onset after 6 years of age (AIPGMEE 06)**
B. Repetitive behavior
C. Delayed language development
D. Severe deficit in social interaction

Ans A Onset after 6 years of age

7. All the following are characteristic features of autistic spectrum disorder (ASD)

A. defective social communication
B. highly restricted fixated interests
C. scarceof gesture use
D. stereotyped motor movements
E. **absence of routines**

Ans E Autismis typically recognized during the 2nd yr of life but can been seen earlier than 12 mo if developmental delays are severe. Initial symptoms most frequently involve delayed language accompanied by lack of social interest or odd play patterns. During the 2ndyr, odd and repetitive behaviors and the absence of typical play become more apparent. It is typical for parents to report that there was no period of normal development or that there was a history of unusual behaviors.

8. Diagnosis of autistic spectrum disorder (ASD) depends partly but importantly on assessment of language. All the following may raise your concern regarding language development and may indicate ASD EXCEPT

A. **absent babbling by 6 months**
B. absent gestures by 12 month
C. absent single words by 16 month
D. absent 2-word purposeful phrases by 24 month
E. loss of language or social skills at any time

Ans A Absence of babbling by 12 months is a red flag for language development and should raise a suspicion of ASD (autistic spectrum disorder).

9. All the following are recognized in autistic spectrum disorder (ASD) EXCEPT

A. 3 grades of severity
B. superior intellectual functioning in select areas
C. motor deficits
D. epilepsy
E. **truancy**

Ans E All these mentioned are recognized associations with ASD, it has 3 grads of severity, early diagnosis usually improve outcome, but those in the severe form continue to adulthood with problems of independent living, employment, social relationships, and mental health. Small group (mild form with communication abilities) can grow up to live self-sufficient lives in the community with employment. Truancy is separate behavioral disorder.

10. The MOST consistent statement of structural MRI brain finding of autistic spectrum disorders (ASD) is

A. diffuse brain atrophy
B. **increase brain size**
C. focal fibrosis
D. white matter degenerative changes
E. gray matter degenerative changes

Ans B Increase in brain size and increased head circumference had been documented in autistic children. Other important finding in structural MRI is diffusion tensor imaging studies suggest aberrations in white matter tract development.

11. All the following should raise the suspicion of autistic spectrum disorders (ASD)

A. sibling with ASD
B. **playmate concern**
C. parental concern
D. caregiver concern
E. pediatrician concern

Ans B Playmate concern is not considered.

12. Ideally all children should be subjected to routine screening for autistic spectrum disorders (in USA) at age of

A. 06 and 12 mo
B. 12 and 18 mo
C. **18 and 24 mo**
D. 24 and 30 mo
E. 30 and 18 mo

Ans C

13. An 18-month-old male toddler was found to be symptomatic for autistic spectrum disorders (ASD) by routine screening testing. The recommended evaluation include all the following EXCEPT

A. physical examination for dysmorphic features
B. hearing tests
C. **brain CT**
D. wood's lamp
E. chromosomal microarray

Ans C The recommended evaluation should rule out dysmorphic syndromes; macrocephaly; hearing problems; tuberous sclerosis and chromosomal abnormalities and genetic causes like fragile X syndrome and Rett syndrome.

14. A 24-month-old child who is referred by health institute after parental concern of lonely play and delayed speech, he is pica eater. You have diagnosed him as autistic spectrum disorder (ASD). Of the following, the MOST important next action is

A. physical examination for dysmorphic features
B. hearing tests
C. **lead level**
D. Wood's lamp
E. chromosomal microarray

Ans C should be order in situation of that, the child is pica eater or he is living in area with high lead level in soil, air or water. Depending on clinical presentation, metabolic screen, thyroid stimulating hormone, EEG and specific genetic studies should be done.

15. You are following a 6-year-old boy with autism, he is under structured psychosocial behavioral training program, there is frequent complains of aggression and self- injurious behavior. Of the following, the BEST medication to control his behavior is

A. methylphenidate
B. **risperidone**
C. escitalopram
D. atomoxetine
E. amantadine

Ans B The drug of choice is risperidone, while methylphenidate is the drug of choice for ADHD in normal IQ children, atomoxetine is also a new medication for ADHD, and escitalopram is used for depression.

ADHD

16. Pervasive disorder is associated with all except:

A. Repetitive behavior
B. Impaired communication
C. Impaired language
D. **Impaired cognition**

Ans D Autistic spectrum disorders (ASD)
• • It includes childhood autism, atypical autism, Asperger disorder, childhood disintegrative disorder and pervasive developmental disorder not otherwise specified
• • Essential features are persistent impairment in reciprocal social communication and restricted, repetitive patterns of behavior or interests
• • Language impairment can range from nonverbal speech to 'phrase speech' to fluent speech

17. All are true about attention deficit hyperactivity disorder except:

A. In two third cases symptoms may continue in adulthood
B. **For making the diagnosis, the behavioral changes must begin before 5 years of age**
C. May be associated with mood disorder
D. Amphetamine may be used for treatment
E. More prevalent in boys

Ans B Attention deficit hyperactivity disorder (ADHD) It is Persistent in attention and/or Hyperactivity-Impulsivity, that interferes with functioning or development, present for at least 6 months, in 2 or more settings, beginning before 12 yr age. Must not be secondary to another disorder. 3 subtypes:
• • ADHD, predominantly inattentive type: includes cognitive impairment & is more common in females.
• • ADHD, predominantly hyperactive-impulsive type, and
• • ADHD, combined type, are more commonly diagnosed in males.

18. Drug not given in ADHD is:

A. Clonidine
B. Atomoxetine
C. Methylphenidate
D. **Barbiturate**

Ans D Drugs useful in the treatment of ADHD include methylphenidate, dexmethylphenidate, dextroamphetamine, atomoxetine, tricyclic antidepressants like imipramine and agonists like clonidine and guanfacine

19. Which is true concerning attention deficit hyperactivity disorder?

A. Impulsive behavior is a feature
B. Higher incidence in tic disorders
C. May respond to treatment with stimulants such as amphetamine
D. **All of the above**

Ans D Attention-deficit/hyperactivity disorder (ADHD) is characterized by inattention, including increased distractibility, difficulty sustaining attention; poor impulse control decreased self-inhibitory capacity; motor overactivity and restlessness. Diagnoses associated with ADHD behaviors are Fragile X syndrome, Fetal alcohol syndrome, Pervasive developmental disorders, Obsessive- compulsive disorder and Gilles de la Tourette syndrome (tic disorder).

20. All of the following are essential features of ADHD Questions except:

A. Lack of concentration
B. Impulsivity
C. Hyperactivity
D. **Mental retardation**

Ans D

21. Does not seem to listen when spoken to directly Which of the above diagnostic criteria are suggestive of inattention (attention deficit) in this child?

A. 1 and 2 only
B. 1 and 3 only
C. 2 and 4 only
D. **1,2, 3 and**

Ans D Motivational therapy

22. Drugs used in ADHD are:

A. **Atomoxetine**
B. Methylphenidate
C. Dextro-amphetamine
D. Quetiapine
E. Benzodiazepine

Ans A b. Methylphenidate, c. Dextro-amphetamine

23. All of the following are essential features of attention deficit hyperactive disease (ADHD) except:

A. Lack of concentration
B. Impulsivity
C. **Mental retardation**
D. Hyperactivity

Ans C with Affected children with ADHD commonly experience academic underachievement, problems with interpersonal relationships with family members and peers, and low self-esteem. But as such, mental retardation is not an essential feature of ADHD.

24. The behavioral changes in attention-deficit/hyperactivity disorder (ADHD) should met all the following criteria EXCEPT

A. developmentally inappropriately far comparing with other children of the same age
B. **must begin before age 6 yr**
C. must be present for at least for 6 mo
D. must be present in 2 or more settings
E. must not be secondary to another disorder

Ans B It is important to systematically gather and evaluate information from a variety of sources, including the child, parents, teachers, physicians, and, when appropriate, other caretakers, over the course of both diagnosis and subsequent management. These behaviors should be evident before age of 12 yr.

25. You are assessing an eight-year-old male child with attention-deficit/hyperactivity disorder (ADHD). Of the following, the LEAST useful test/investigation is

A. thyroid function test
B. lead level
D. **Blood film**
E. polysomnography

Ans D At first, hypertension, ataxia, and hyperthyroidism should be excluded. Other differential diagnosis should be considered as heavy metal poisoning (including lead), sensory deficits (hearing and vision), auditory and visual processing disorders, and neurodegenerative disorder, especially leukodystrophies. Fragile X syndrome, absence and sleeping disorders are also important differential diagnoses.

26. Of the following, the MOST common presenting behavior in girls with attention- deficit/hyperactivity disorder (ADHD) is

A. **inattentive**
B. hyperactive
C. impulsive
D. disruptive
E. combined

Ans A While the hyperactive-impulsive and combined types are common in boys.

Learning Disabilities

27. True about Asperger syndrome:

A. More common in girl
B. **Repetitive activity pattern**
C. Subnormal intelligence is consistent feature
D. Severe language impairment is characteristic

Ans B Asperger syndrome is characterized by repetitive/ restricted patterns of activity and interest, with preserved (not impaired) early language development and typically normal intelligence, and is more common in boys.

28. All are habit disorders except:

A. Pyridoxine
B. Zinc
C. **Iron**
D. Molybdenum

Ans C Temper tantrum •• Habits involve an action or pattern of behavior that is repeated often.
• • Habits are common in childhood and range from benign and transient (thumb sucking, nail biting) to more problematic (trichotillomania, bruxism).
• • Temper tantrums and breath-holding spells are common during the 1st years of life and are age-typical expressions of frustration or anger. Tics:
• • Sudden, rapid, recurrent, non-rhythmic motor movements or vocalizations.

29. Formula to calculate intelligence quotient (IQ):

A. Chronological age/Mental age ××100
B. Mental age–Chronological Age ×
C. **Mental age/Chronological age ×**
D. Mental age + Chronological ×

Ans C Mental age/chronological age×××100 Intelligence Quotient (IQ) = Mental age/Chronological age ×

30. A mother to a well 2-year-old girl with thumb sucking behavior, she is worried that the behavior may continue or may cause dental problem. Of the following, the BEST response is to

A. reassurance to mother
B. leave the behavior as the complications usually started after 5 years
C. **ignore thumb sucking and encouraging a substituted behavior**
D. use of bitter ointments will resolve the problem early
E. asses the social status of the family

Ans C Thumb sucking is common in infancy and in as many as 25% of children age 2 yr and 15% of children age 5 yr. Thumb sucking beyond 5 yr may be associated with sequelae (paronychia, anterior open bite). Like other rhythmic patterns of behavior, thumb sucking is self soothing. Basic behavioral management, including encouraging parents to ignore thumb sucking and instead focus on praising the child for substitute behaviors, is often effective treatment.

31. Approximately 90% of youths who complete suicide have a preexisting psychiatric illness. Of the following, the MOST commonly encountered illness is

A. **major depression**
B. schizophrenia spectrum disorders
C. conduct disorder
D. chronic anxiety
E. panic disorder

Ans A All mentioned distracters have a role in suicide but major depression is the most common one.

32. Which statement is FALSE regarding specific learning disorders (SLD)?

A. it is a type of neurodevelopmental dysfunctions
B. the overall estimates of the prevalence of SLD range from 3-10%
C. it focuses on academic skill development
D. **includes students did well in academic testing but not in intelligence testing**
E. terms as dysgraphia or dyscalculia had been revolutionized

Ans D Specific developmental disorders of scholastic Skills (specific learning disorders) includes reading disorder, spelling disorder, disorder of arithmetical skills, and mixed disorder of Scholastic Skills. Dyslexia (reading disorder) is included in ICD-10 in a separate category of symbolic dysfunction.

33. Witnessing violence, community violence, and media violence all are detrimental to children as it increases the impact of violence. All the following adverse effects can be increased by increasing the impact of violence

A. poor school performance
B. symptoms of anxiety and depression
C. lower self-esteem
D. **cyper bullying**
E. post traumatic stress disorder

Ans D All these (except D) represent the impact of violence on children, while cyber bullying is a newly emerging form of bullying that takes place using electronic technology.

34. In a child less than 3 years, the diagnosis of failure to thrive (FTT) is considered if

A. weight is below the 3rd percentile
B. **weight drops down more than 2 major percentile lines**
C. weight for height is less than the 1st percentile
D. BMI less than the 25th percentile
E. midarm circumference < 15 cm

Ans B Weight is below the 5th percentile, weight for height is less than the 5th percentile, BMI less than the 5th percentile, also defines FTT.

Intellectual Disability

35. A 3-year-old boy with normal developmental milestones with delayed speech, has difficulty in communication and concentration. He is not making friends. Most probable diagnosis is:

A. **Autism**
B. ADHD
C. Mental retardation
D. Specific learning disability

Ans A The given scenario where the child is having delayed speech and difficulty in communication and concentration and is not fo making friends, suggests a diagnosis of Autistic spectrum disorder.

36. Which of the following childhood disorder improves with increase in age?

A. Conduct disorder
B. Emotional problems
C. **Temper tantrum**
D. Sleep disorder

Ans C Temper tantrums and breath-holding spells are common during the first years of life and are age-typical expressions of frustration or anger; They improve as the child grows up.

37. Preventable cause of mental retardation is:

A. Downs
B. Phenylketonuria
C. **Cretinism**
D. Cerebral palsy

Ans C Hypothyroidism is the most important preventable or treatable cause of intellectual disability in children.

38. Conditions associated with mental retardation are:

A. **Trisomy**
B. Fragile X
C. Homocystinuria
D. Phenylketonuria
E. Tuberous sclerosis

Ans A 21, b. Fragile-X, c. Homocystinuria, d. Phenylketonuria, e. Tuberous sclerosis

39. A male child of 15 years with a mental age of 9 years has an IQ of:

A. 50
B. 60
C. 70
D. 80 Chapter 22: Behavioral Disorders in Children 519 Answers

Ans B 60 IQ = Mental age x 100/Chronological age ⇒ So, in this case, IQ = 9 × 100/15 = 60 23 Chapter Musculoskeletal DISORDERS INVOLVING MUSCLES A. MUSCULAR DYSTROPHIES Inherited disorders of skeletal muscle with progressive muscle damageQ 1. DUCHENNE MUSCULAR DYSTROPHY (DMD) M Epidemiology It is the most common hereditary neuromuscular disease affecting all races Genetics X-linked recessive: Xp21 (Dystrophin gene), one of the largest genes is affected

40. The following factors show psychosocial impact of war on children EXCEPT

A. loss of family members
B. separation from community
C. lack of education
D. acute stress reaction
E. displacement

Ans E It represents physical impact.

41. In patients with failure to thrive; chromosomal abnormality, intrauterine infection, and metabolic disorders need to be ruled out. All the following factors are suggestive of metabolic problems EXCEPT

A. insidious history
B. recurrent vomiting
C. neurologic symptoms
D. cardiomyopathy
E. renal symptoms

Ans A The history of metabolic disorder is usually acute, severe, and potentially life- threatening, recurrent vomiting, liver dysfunction, neurologic symptoms, cardiomyopathy and myopathy, impairment of special senses, renal symptoms, or distinct dysmorphic features and/or organomegaly all are my be a part of metabolic illness.

Conduct Disorder & ODD

42. A 14-year-old boy has difficulty in expressing himself in writing and makes frequent spelling mistakes, does not follow instruction and cannot wait for his turn while playing a game. He is likely to be suffering from:

A. Impaired vision
B. Impaired Neurobehavioral development
C. Impaired folate level
D. A socioeconomic hazard
E. Parenting

Ans C Specific learning disability Difficulty in expressing himself in writing and making frequent spelling mistakes, point towards the diagnosis of Dyslexia, a specific learning disability.

43. The following is not true regarding bruxism:

A. Associated with day time anxiety
B. If untreated, can result in dental malocclusion
C. Condition improves if proper deworming is done
D. Chronic bruxism leads to temporomandibular joint pain

Ans C Bruxism or teeth grinding can begin in the first 5 yr of life, and may be associated with daytime anxiety. Persistent bruxism can manifest as muscular or temporomandibular joint pain; Untreated bruxism can cause problems with dental occlusion.

44. A 5-year-old child is assessed to have developmental age of one year. His developmental quotient would be:

A. 100
B. 80 (DPG 10)
C. 60
D. 20

Ans D 20 DQ = Developmental age x 100/Actual age ⇒ So, in this case, DQ = 1 × 100/5 =

45. A 7-year-old child brought by his father to your clinic, the child often express fear of being injured by a car accident during transport to school. He expresses this fear to his teachers and parents. Of the following, the TRUE description of his reaction is

A. non pathological anxiety
B. school phobia
C. separation anxiety
D. generalized anxiety disorder
E. panic disorder

Ans A Generally anxiety classified to a pathological and non-pathological anxiety. In pathological form it is usually resulted in loss of function (i.e. become disabling, interfering with social interactions, development, and achievement of goals or quality of life, and can lead to low self-esteem, social withdrawal, and academic underachievement) also can be associated with somatic symptoms such as weight loss, pallor, tachycardia, tremors, muscle cramps, paresthesias, hyperhidrosis, flushing, hyperreflexia, and...

46. A 13-year-old adolescent female attained out-patient department (OPD), with a complaint of feeling fat especially over the stomach and thighs; she is also feeling cold, tired, weak, and lacking energy. Examination reveals, heart rate 46 beats/min, blood pressure 70/40 mm Hg, weight below 3rd centile, dry skin, and lanugo-type hair growth on face; lab investigations shows hypokalemia and hypophosphatemia. Of the following, the MOST appropriate next action is

A. admission to the hospital
B. referral to psychiatrist
C. referral to dietician
D. make an scheduled OP visit to you and to psychiatrist
E. offer partial hospital program

Ans A Heart rate <50 beats/min and blood pressure <80/50 mmHg with hypokalemia and hypophosphatemia are indications for admission in anorexia nervosa; other indications may include hypoglycemia, dehydration, temperature <36.1°C, <80% healthy body weight, hepatic, cardiac, or renal compromise, suicidal intent and plan.

47. The major chemical modulator of attention is

A. dopamine
B. serotonin
C. nor-epinephrine
D. troponin
E. epinephrine

Ans A Most brain processes are heavily dependent on functional arousal, alertness, and attention. Any malfunction within or across these systems will likely cause some degree of breakdown in other cognitive processes. Functional attention subsumes intact neuroanatomic and neurochemical brain systems. Structurally, brain regions involved include subcortical, cortical, and association areas throughout the brain.

48. The MOST reported type of child abuse in USA is

A. neglect
B. physical abuse
C. sexual abuse
D. psychological maltreatment
E. trafficking

Ans A Physical abuse includes beating, shaking, burning, and biting. Sexual abuse has been defined as the involvement of dependent, developmentally immature children and adolescents in sexual activities. Neglect refers to omissions in care, resulting in actual or potential harm which include inadequate healthcare, education, and supervision, protection from hazards in the environment, and unmet physical needs (e.g., clothing, food) and emotional support.

49. Approaching to a child with failure to thrive based on signs and symptoms. Of the following, the MOST common cause behind a child has spitting, vomiting, and food refusal is

A. gastroesophageal reflux
B. chronic tonsillitis
C. food allergies
D. eosinophilic esophagitis
E. inflammatory bowel disease

Ans A The first 4 causes are true causes but the commonest is (A). The choice (E) causes FTT with diarrhea and fatty stools.

Anxiety Disorders

50. A 6-year-old child has history of birth asphyxia, does not communicate well, has slow mental and physical growth, does not mix with people, has limited interests, and gets widely agitated if disturbed, diagnosis is:

A. Hyperkinetic child
B. Autistic disorder
C. Attention deficit disorder
D. Schizophrenia RETT SYNDROME

Ans B RETT SYNDROME 22. Ans c. Rett syndrome 23. Ans d. Macrocephaly Deceleration of head growth leading to acquired microcephaly is a feature of Rett syndrome.

51. Which of the following is not true regarding "thumb sucking"?

A. Is a sign of insecurity
B. Pleasurable sensation
C. Leads to dental problems
D. Seen only in child < 4 years of age

Ans D Thumb Sucking
• • Thumb sucking is a self-soothing behavior, common in infancy and is seen in 15% of children aged 5 yr
• • Thumb sucking beyond 5 yr may be associated with sequelae (paronychia, anterior open bite).
• • Management includes encouraging parents to ignore thumb sucking and focus on praising the child for substitute behaviors.

52. Mild MR features are:

A. Present in 5–10% population
B. Higher incidence in low socioeconomic group
C. Presents in 2 years
D. Genetic background present
E. Specific cause found in 75% cases

Ans B d. Genetic background present Mild intellectual disability (Note: New name for Mental retardation is Intellectual disability).
• • It is more likely in offspring of women who have not completed high school than those who have graduated.
• • This is presumably a consequence of both genetic and socioeconomic (poverty, malnutrition) factors.
• • Specific causes are identifiable in <50% of affected; Familial clustering is common.

53. All the following medical conditions can cause anxiety in a child EXCEPT

A. antihistamine medications
B. hypoparathyrodism
C. prolonged school absences
D. carbonated beverages
E. lead poisoning

Ans B Systemic disorders, hormonal disturbances, and drugs along with carbonated beverage can cause anxiety in children with it is symptoms and signs such as tachycardia e.g. hyperthyroidism, hyperparathyroidism, hypoglycemia, central nervous system disorders (brain tumors), migraine, asthma, lead poisoning, cardiac arrhythmias, and, rarely, pulmonary embolism, systemic lupus erythematosus, anaphylaxis, porphyria or pheochromocytoma.

54. All the following are characteristic features of separation anxiety disorder EXCEPT

A. not manifested below 3 years of age
B. common up to 5% of children
C. girls are more affected than boys
D. child thinking often include fear of harm affecting parents
E. not reported in children above 8 years of age

Ans E Separation anxiety disorder (SAD) is one of the most common childhood anxiety disorders with a prevalence of 3.5-5.4%. Separation anxiety behavior is developmentally normal when it begins about 10 mo of age and tapers off by 18 mo. By 3 yr of age, most children can accept the temporary absence of their mother or primary caregiver. SAD is more common in prepubertal children, with an average age of onset of 7.5 yr. Girls are more commonly affected than boys.

55. **In between specific learning disorders (SLD), the MOST refractory to treat is**

A. reading
B. **mathematics**
C. working memory
D. social cognition
E. writing

Ans B Delay in mathematical ability, known as mathematics disorder or dyscalculia, can be especially refractory to correction, partly because math involves the assimilation of both procedural knowledge (e.g., calculations) and higher-order cognitive processes (e.g., working memory).

56. **Regarding the physical abuse, the physical signs in abused children (as bite marks) may be found in up to**

A. 5%
B. 15%
C. **25%**
D. 35%
E. 45%

Ans C Sentinel injuries may be noted in approximately in 25% of abused infants and may precede the diagnosis by weeks or even months from the sentinel event.

57. **A 15-month-old male child with failure to thrive, diarrhea, and fatty stool. Of the following, the MOST common possible cause of his illness is**

A. **malabsorption**
B. intestinal parasites
C. milk protein intolerance
D. pancreatic insufficiency
E. immunodeficiency

Ans A

Depression & Mood Disorders

58. **A female child who initially had normal development followed by regression of developmental milestones, presented with slow, hand wringing movements; she also had microcephaly. What is the probable diagnosis?**

A. Autistic spectrum disorder
B. Lysosomal storage disorder
C. **Rett syndrome**
D. Congenital infection

Ans C Developmental regression with stereotypic hand-wringing movements and acquired microcephaly in a girl is characteristic of Rett syndrome.

59. **Waking up at night, screaming with fear, at 2.5 years age, is generally a manifestation of:**

A. Organic illness
B. Normal development pattern
C. Castration anxiety
D. **Separation anxiety**

Ans D •• Anxiety disorders are the most common psychiatric disorders of childhood
• • Separation anxiety disorder (SAD) is one of the most common childhood anxiety disorders
• • Separation anxiety is developmentally normal when it begins about 10 months of age and tapers off by 18 mo
• • By 3 yr of age, most children can accept the temporary absence of their mother or primary caregiver.

60. **The percentage of mental illnesses that is encountered at least once in any stage during childhood is about**

A. 10%
B. **20%**
C. 30%
D. 40%
E. 50%

Ans B

61. **You are asked to explain breath holding spells concept to a staff nurse during morning round. Of the following, the BEST statement; it is a/an**

A. manifestation of iron deficiency anemia
B. type of seizure
C. **age typical expressions of frustration or anger**
D. psychic problem manifest when the child becomes older
E. sort of tic disorder which resolve by age

Ans C Breath holding spells are common during the first years of life and are age- typical expressions of frustration or anger. Subtypes of breath holding spells include cyanotic, pallid, or mixed episodes. Cyanotic are the dominant type and may include a brief loss of consciousness and a very brief tonic-clonic seizure. Pallid spells may be similar to vasovagal related syncopal events in older children and initiated from similar stimuli.

62. **A parents came to your clinic complaining that their 5-year-old boy had attacks of frightening with imagination of a snake crawling over him and he is acting as trying to remove it. You assessed the child and find no acute physical illness and he is cooperative and quite intelligent. Of the following, the MOST appropriate explanation for the child behavior is**

A. **acute phobic hallucination**
B. early sign of schizophrenia
C. delusional infestation
D. night terror
E. bipolar disorder

Ans A In children with non-psychotic hallucinations, the symptoms of psychosis are absent; normal mental status examination is the key for it is diagnosis. Non-psychotic hallucinations commonly occur in the context of severe traumatic stress, developmental difficulties, social and emotional deprivation. Hallucinations related to fantasy, culture believes, grief, hypnogogic hallucinations, night terrors, fever, and acute phobic hallucinations all are considered normal.

63. **All the following are risk factors for specific learning disorders (SLD) EXCEPT**

A. extreme prematurity
B. low birth weight
C. history of sibs or parents with SLD
D. **history of admission to neonatal care unit**
E. chronic medical problem

Ans D is not a specific marker for serious problem. It could be for minor cause or for observation only.

64. The MOST common clue of physical abuse in children is

A. history of inflicted trauma
B. burn marks
C. **bruises**
D. fractures
E. retinal hemorrhages

Ans C Features suggestive of inflicted bruises include (a) bruising in a pre ambulatory infant (occurring in just 2% of infants), (b) bruising of padded and less-exposed areas (buttocks, cheeks, under the chin, genitalia), (c) patterned bruising or burns conforming to shape of an object or ligatures around the wrists, and (d) multiple bruises, especially if clearly of different ages. History is the least dependable because it is either hidden or skewed.

65. A 2-year-old child with failure to thrive, recurrent wheezing, and pulmonary infections. Of the following, the LEAST common cause of his illness is

A. asthma
B. aspiration
C. food allergy
D. cystic fibrosis
E. **immunodeficiency**

Ans E

Eating Disorders

66. A 2-year-old girl child is brought to the outpatient department with features of hand wringing stereotype movements, impaired language & communication, breath holding spells, poor social skills and deceleration of head growth after 6 months of age. The most likely diagnosis is:

A. Asperger's syndrome
B. **Rett syndrome**
C. Fragile X-syndrome
D. Calorad syndrome ADHD

Ans B Rett's syndrome The given clinical picture of a girl with hand wringing stereotype movements, impaired language and communication, breath holding spells, poor social skills and deceleration of head growth is consistent with the diagnosis of Rett syndrome. ADHD

67. Pica refers to:

A. IU sucking
B. Thumb sucking
C. **Foreign object being put in the mouth**
D. None of above

Ans C

68. Clinicians tools needed to recognize early symptoms of mental disorders are called "Mental Health Action Signs". All the following are true EXCEPT

A. involvement in many fights, wanting to badly hurt others
B. not eating, throwing up, or using laxatives to make yourself lose weight
C. **feeling very sad or withdrawn for more than 2 mo**
D. sudden overwhelming fear for no reason
E. worries or fears that get in the way of your daily activities

Ans C

69. Which one of the following statements is FALSE regarding rumination?

A. **seen only in infants and those with intellectual disability**
B. runs episodic course
C. results from neglect
D. in infancy can resolve spontaneously
E. behavioral treatment is the treatment of choice

Ans A Rumination disorder is the repeated regurgitation of food, where the regurgitated food may be rechewed, reswallowed, or spit out, for aperiod of at least 1 mo following a period of normal function. Rumination disorder has also been in healthy individuals across the life span in addition to infants and those with intellectual disability.

70. In pica disorder, the MOST appropriate statement is

A. it is only related to eating clay and earth
B. usually in children below 2 years
C. not present with other eating disorders
D. **it need a period of more than a month to be stated**
E. it resulted in high fatalities

Ans D Pica involves the persistent eating of non-nutritive, non-food substances (e.g., paper, soap, plaster, charcoal, clay, wool, ashes, paint, earth) over a period of at least 1 mo. The eating behavior is inappropriate to the developmental level (e.g., the normal mouthing and tasting of objects in infants and toddlers) and, therefore, a minimum age of 2 yr is suggested. Children with pica are at increased risk for lead poisoning; iron-deficiency anemia;

71. All the following are red flags in the history of children with specific learning disorders (SLD) EXCEPT

A. parents concern about academic performance
B. **inconsistency in marks report from grade to grade**
C. struggling with home work activities
D. positive vision or hearing test
E. positive standard screening test

Ans B Inconsistency in report from grade to grade may sometimes be caused by a difference in teaching styles or classroom demands.

72. In children with physical abuse, when the inflected trauma is burn injury. Of the following, the MOST suggestive one is

A. burn in napkins with involvement of flexures
B. presence of splash mark
C. unclear borders of burn
D. **glove and stock distribution**
E. folk remedies

Ans D Immersion burn injury patterns; napkin area that spares flexor creases, immersion stocking burn, immersion glove burn, immersion buttocks burn. It usually shows a clear delineation between the burned and healthy skin, and uniform depth. The splash marks are usually absent.

73. **In a child with failure to thrive, the indications for hospitalization include severe malnutrition or failure of outpatient management. The period after which the child needs hospitalization, if he has not been responded to outpatient management, is about**

A. 1-2 wk
B. 4 wk
C. **2-3 mo**
D. 4 mo
E. 5-6 mo

Ans C Inpatient care may include further diagnostic and laboratory evaluation, an assessment and implementation of adequate nutrition, and evaluation of the parent– child feeding interaction.

Elimination Disorders

74. **A 6-year old-child was brought with complaints of bedwetting at night and not during day time. His urine specific gravity was 1.020 and other tests were normal. What will you advice?**

A. Reassure
B. **Consult a child Psychologist**
C. USG abdomen
D. CT pelvis

Ans B In this case, the child has nocturnal enuresis, with normal findings on urine examination (normal urine specific gravity is 1.016-1.022) Nocturnal enuresis
• • It refers to the occurrence of involuntary voiding at night after 5 yr.
• • If no daytime symptoms and physical examination and urinalysis are normal, further evaluation for urinary tract pathology is not warranted.
• • An ultrasonogram is done in an older child (usually ≥ 7 yrs) with enuresis or in those who do not respond to therapy.

75. **Amongst various treatment modalities for nocturnal enuresis, the relapse rates have been observed to be lowest for:**

A. Desmopressin
B. Imipramine
C. **Bell alarm systems**
D. Oxybutinin

Ans C Bell alarm system has a reported success of 30-60%, although the relapse rate is significant. Direct comparisons of the bell alarm system with pharmacologic therapy favor the former because of lower relapse rates, although initial response rates are equivalent.

76. **Bedwetting is normal up to what age in girls and boys respectively?**

A. 3 years and 4 years
B. 4 years and 5 years
C. **5 years and 5 years**
D. 5 years and 6 years

Ans C Girls typically acquire bladder control before boys and bowel control is usually achieved before bladder control. Nocturnal enuresis refers to the occurrence of involuntary voiding at night after 5 yr, in both, boys and girls.

77. **A 5-year-old child presents with bedwetting. Rx of choice is:**

A. No treatment
B. Imipramine
C. Desmopressin
D. **Motivational therapy**

Ans D

78. **Nocturnal enuresis is abnormal after:**

A. 2 years
B. 3 years
C. 4 years
D. **5 years**

Ans D

79. **Nocturnal enuresis best t/t is:**

A. Positive reinforcement
B. Punishment
C. **Bed alarm**
D. Desmopressin

Ans C

80. **Lowest recurrence in nocturnal enuresis is seen with:**

A. **Bed alarms**
B. Desmopressin
C. Imipramine
D. Oxybutynin

Ans A Lowest recurrence rate is with combination therapy (bed alarm + pharmacotherapy), followed by bed alarm.

81. **If behavioral therapy fails in the management of enuresis, the pharmacological drug of choice for this case is:**

A. Phenytoin
B. Diazepam
C. **Imipramine**
D. Alprax

Ans C

82. **Which of the following nasal spray is very effective in control of enuresis:**

A. Pitressin
B. **Desmopressin**
C. Lypressin
D. None of the above AUTISTIC SPECTRUM DISORDER

Ans B •• Desmopressin ia a synthetic analog of antidiuretic hormone (ADH) that reduces urine production overnight
• • This medication is FDA-approved and is available as a tablet, with a dosage of 0.2-0.6 mg at bedtime
• • In the past a nasal spray was used, but some children experienced hyponatremia and convulsions with this formulation and the nasal spray is no longer recommended for nocturnal enuresis.

Tic Disorders & Tourette

83. All of the following are characteristics of Rett syndrome except:

A. Increased incidence of mental retardation
B. Seizures
C. Abnormal dendritic morphology in cortical pyramidal cells
D. **Macrocephaly**

Ans D Rett syndrome causes acquired (deceleration of) microcephaly, not macrocephaly, along with intellectual disability, seizures, and abnormal cortical dendritic morphology.

84. True regarding breath holding spells is all except:

A. **Antiepileptic treatment is necessary**
B. Atropine is sometimes used
C. Attacks of cyanosis can occur
D. Occurs between 6 months to 5 years

Ans A

85. Which is most common genetic cause of mental retardation?

A. Tuberous sclerosis
B. Cri-du-chat syndrome
C. **Fragile-X-syndrome**
D. Angel's syndrome

Ans C

86. A concerned parents of an 18-month-child which expressed a brief tonic movement after being upset and cried. All the following are true advices EXCEPT

A. investigate possibility of iron deficiency anemia
B. avoid over concern behavior
C. interfere early in the event by calming the child
D. try to behave calmly, observe the child without being seen
E. **arrange for home rectal diazepam**

Ans E All of the above in a sequence of BDCA represent an approach to deal with classical breath holding spells.

87. All the following are diagnostic criteria for major depressive episode EXCEPT

A. **hypomanic episode**
B. depressed mood
C. loss of interest or pleasure
D. significant weight loss
E. hypersomnia

Ans A Hypomanic or manic episodes are not features of major depressive episode. A period of at least 2 wks is required while in persistent depressive disorder require a period of 2 years for diagnosis.

88. All the following are established modalities of treatment in a child with specific learning disorders (SLD) EXCEPT

A. remediation of skills
B. demystification
C. bypass strategies
D. **dietary interventions**
E. strengthening of strength

Ans D

89. A 9-year-old male with good school performance and normal behavior. In the last 2 months he had changed his classroom. His teacher observed that he became introverted and his school performance reduced dramatically. He was always described by some of his new school mates as a "weak". Of the following, the MOST likely diagnosis is

A. school phobia
B. anxiety disorder
C. **bullying**
D. hypothyroidism
E. depression

Ans C Showing the power of some children on a weaker one can lead serious psychological and psychiatric effects. That child was a bullying-victim and should be protected.

90. Retinal hemorrhages are an important marker of abusive head trauma (AHT). Whenever AHT is being considered, a dilated indirect eye examination by a pediatric ophthalmologist should be performed. All the following are characteristic of retinal hemorrhage caused by AHT EXCEPT

A. multiple
B. involve more than one layer of the retina
C. **central**
D. traumatic retinoschisis
E. various sizes

Ans C It is often extending to the periphery (outside the posterior pole). D is highly characteristic.

Schizophrenia & Psychosis

91. A 10-year-old child is always restless, inattentive to study and always wants to play outside. Parents are extremely distressed. What would you advise?

A. It is a normal behavior
B. Behavior therapy
C. **It is a serious illness requires medical treatment**
D. Needs change in environment

Ans C The given history suggests ADHD; For ADHD, Stimulants have been more effective than behavioral treatments used alone.

92. Treatment for breath holding spells in a child is:

A. Give extra care and love to the child
B. Inflicting painful stimulus at the beginning of the attack
C. **Do not give attention to the child**
D. Fulfill all the wishes of the child to prevent the attack
E. Low dose barbiturates

Ans C For management of breath-holding spells, parents should be advised to intercede early in defiant behavior by calmly placing the child in timeout for a period of time approximating 1 min for each year of age.

93. All the following are true associations between psychiatric illness in childhood and their treatment EXCEPT

A. attention deficit/hyperactivity (ADHD) and atomoxetine
B. anxiety and antidepressant
C. aggression and atypical antipsychotic
D. **psychosis and typical antipsychotic**
E. depression and antidepressant

Ans D Atypical antipsychotic.

94. **You are evaluating a 5-year-old child with breath holding spells, the history given includes pallor with abnormal limb movement lasted for 5 minutes followed by sleep. All the following are true responses/advices EXCEPT**

A. reassurance, behavioral instruction to parents and follow up
B. order MRI brain
C. order an ECG
D. order an EEG
E. neurological consultation

Ans A In non-classical type of breath holding spells; medical conditions to consider should include seizures, Chiari crisis, dysautonomia, cardiac arrhythmias (as prolonged Q-T interval), and central nervous system lesions. So all the above optional investigations are justifiable.

95. **Childhood psychosis may include all the following EXCEPT**

A. delusions
B. loss of reality testing
C. disorganized speech
D. catatonic behavior
E. acute phobic hallucination

Ans E

96. **The MOST common specific learning disorder (SLD) of childhood is**

A. attention-deficit/hyperactivity disorder (ADHD)
B. reading disorder (dyslexia)
C. spelling disorder
D. arithmetical skills disorders
E. mixed disorder of Scholastic Skills

Ans B Reading disorder affect about 80% of children identified as learning disabled.

97. **Skeletal injuries with high specificity in child abuse may include all the following**

A. tibial metaphyseal corner lesions
B. posterior rib fracture
C. scapular fracture
D. first rib fracture
E. clavicle fracture

Ans E It is of low-specificity finding.

98. **You are treating a 10-month-old child with failure to thrive. Of the following, the minimal accepted catch-up weight gain is about**

A. 10 gm/kg/day
B. 20-30 gm/kg/day
C. 40-50 gm/kg/day
D. 60 gm/kg/day
E. 80 gm/kg/day

Ans B Minimal catch-up growth should generally be 2-3 times the average weight gain for corrected age.

Substance Abuse

99. **A 9-year-old child disturbs other people, is destructive, interferes when two people are talking, does not follow instructions and cannot wait for his turn while playing a game. He is likely to be suffering from:**

A. Emotional disorders
B. Behavioral problems
C. No disorder
D. Attention deficit hyperactive disorder

Ans D

100. **A 6-year-old boy presents with complaints of hypera cti-vity, excessive shoulder shrugging, throat clearing and eye blinking. During examination, he is not coo perative, uses obscene words and runs around in the consultation room. What is the most probable diagnosis?**

A. Attention deficit hyperactivity disorder
B. Tic disorder
C. Psychomotor seizures
D. Childhood psychosis

Ans B Tourette disorder (TD), persistent motor or vocal tic (PTD), and are characterized by involuntary, rapid, repetitive, single or multiple motor and/or vocal/phonic tics that wax and wane in frequency but have persisted for > 1 year since first tic onset.

101. **The following medications are truly matched to their major pharmacological groups**

A. methylphenidate ..... stimulant
B. atomoxetine ..... serotonin-norepinephrine reuptake inhibitor
C. escitalopram ..... selective serotonin reuptake inhibitors
D. fluoxetine..... tricyclic antidepressants
E. risperidone.....atypical antipsychotics

Ans D These are the common medications used in Pediatric psychiatric disorders which pediatrician should be familiar with. Floxtine is common antidepressant and it is another selective serotonin reuptake inhibitors

102. **A concerned young parents asking about frequent lying behavior experienced by their 3-year-old girl. The following advices are true EXCEPT**

A. it is a method of playing with the language
B. it is a part of their magical thinking
C. it indicates a potential for future lying behavior
D. it is an approach to avoid unwanted confrontation with adults
E. it is a way to describe things as they wish

Ans C Lying is a normal behavior between 2-4 years and represents all the above except the option C.

103. **The definition of intellectual function is the capacity to think in the abstract, reason, problem solve, and comprehend. The score at which significant weaknesses in adaptive skills are evident is equal to or below**

A. 55
B. 60
C. 65
D. **70**
E. 75

Ans D Neurodevelopmental dysfunctions reflect disruptions of neuroanatomic structure or psychophysiologic function and place a child at-risk for developmental, cognitive, emotional, behavioral, psychosocial, and adaptive challenges.

104. **Of the following, the TRUE statement of reading disorder is**

A. 25% of affected children have affected parents
B. **diagnosis is usually clinical**
C. IQ will remain same with time
D. functional MRI is diagnostic
E. boys are more affected than girls

Ans B The cardinal signs of dyslexia observed in school-age children and adults are a labored, effortful approach to reading involving decoding, word recognition, and text reading. 50% of affected children have affected parents. Boys and girls are equal.

105. **A 3-month-old baby girl admitted to pediatric intensive care unit with severe head injury (evident by CT scan), the history given by parents was trivial and not informative. You suspected child abuse. Of the following, the MOST helpful study to support your suspicion is**

A. infantogram (one shot for entire body)
B. lateral and AP view X ray of the spine
C. lateral and AP view X ray of skull
D. **radionuclide bone scan**
E. MRI brain

Ans D Of the given options, option D is the best because it detect acute skeletal injuries (as fractures of posterior rib fracture that indicate shaking injury) before it became evident on X ray. If there is suspension in the first scan, another scan can be performed after 2 days from the first one. Infantogram is not preferable as a one or 2 shot for entire body. Options B and C are only a part from skeletal survey, which is indicated at time of arrival and after 2 weeks.

School Refusal

106. **A 9-year-old child is restless. He is hyperactive and his teacher complaints that he does not listen to the teachings. Disturbs other students, he also shows less interest in playing. The likely diagnosis is:**

A. Cerebral palsy
B. **Attention deficit hyperactive child**
C. Delirium
D. Mania

Ans B

107. **Early strict toilet training can result in:**

A. Nocturnal enuresis
B. **Encopresis**
C. Night terror
D. Temper tantrums MENTAL RETARDATION

Ans B Encopresis is involuntary passage of feces into inappropriate places at least once a month for 3 consecutive months, after a chronologic or developmental age of 4 yr is reached. Early strict toilet training can cause it.

108. **A 7-year-old boy presented with recurrent eye blinking behavior and recurrent extension of extremities, mother describe the movement as sudden, rapid, and repetitive movements, it was present in the last 9 months. Of the following, the MOST appropriate diagnosis is**

A. Tourette's disorder
B. persistent motor tic disorder
C. **provisional tic disorder**
D. post-viral encephalitis
E. Sydenham chorea

Ans C A tic is a sudden, rapid, recurrent (in the same manner), non-rhythmic motor, movement or vocalization e.g. eye blinking, neck jerking, shoulder shrugging, extension of the extremities. Onset of tics is typically between ages 4 and 6 yr, in Tourette's disorder both multiple motor and one or more vocal tics have been present at some time during the illness, may be concurrently for > 1year.

109. **Regarding school refusal, which statement is FALSE?**

A. it is a complex disorder
B. selective mutism is overlapping
C. younger children usually have separation anxiety disorder
D. **older children usually suffer from obsessive disorders**
E. somatic symptoms are common

Ans D School refusal or phobia it is a common problem, it occurs in approximately 1- 2% of children, associated with anxiety in 40-50% of cases, depression in 50-60% ofcases, and oppositional behavior in 50% of cases. Younger anxious children who refuse to attend school are more likely to have separation anxiety disorder whereas older anxious children usually refuse to attend school because of social phobia.

110. **A school nurse called you to see a 6-year-old boy with school refusal, he is always crying after parental leaving, refuses to stay in the class room, the school nurse informed you that she tried her best during the last 4 weeks; and she met with the parents frequently. Of the following, the BEST action is to**

A. **refer the child to a pediatrics psychologist**
B. start selective serotonin reuptake inhibitor treatment
C. asses the home environment
D. arrange for parent management training
E. give the child special attention from the teacher

Ans A Management of school refusal typically requires parent management training and family therapy, with school personnel is always indicated; anxious children often require special attention from teachers, counselors, or school nurses. Parents who are coached to calmly sending child to school and to reward the child for each completed day of school are usually successful; in this scenario this option had been used. In cases of ongoing school refusal, referral to a child psychiatrist or psychologist is indicated.

111. **In USA, the newly arrived international adoptees are subjected to many lab tests. All the following tests need to be repeated after 3-6 months after arrival EXCEPT**

A. hepatitis B virus serology
B. hepatitis C virus serology
C. human immunodeficiency serology
D. tuberculin skin test
E. **treponemal test**

Ans E

112. **A 3-month-precious baby of a keen and highly educated family presented with history of URTI followed by deterioration of level of consciousness and seizure. CT brain shows intracranial bleeding with no evidence of skull fracture. Fundocscopy was normal. Mother gives history of difficult labour but with normal development, mild hypotonia, and macrocephaly which was reassured initially by general Pediatrician. Of the following, the MOST likely diagnosis is**

A. residual birth trauma
B. arteriovenous malformation
C. acute viral encephalitis
D. **glutaric aciduria type**
E. abusive head trauma (AHT)

Ans D Patients with birth trauma may have asymptomatic subdural hematoma which may resolve by the end of first month of life. Arteriovenous malformation usually has subarachnoid hemorrhage. Encephalitis is possibility but unusually present with intracranial bleed. AHT is a possibility but presence of hypotonia and macrocephaly and absence of retinal hemorrhage make the diagnosis of Glutaric aciduria type 1 is the most likely one.

Psychopharmacology

113. **About Fragile X Syndrome, true are all except:**

A. Large testis
B. **Large nose**
C. Large ears
D. High arched palate OTHER BEHAVIORAL DISORDERS

Ans B Elongated face with large ears, high arched palate and large testis (macro-orchidism) are seen in fragile X-syndrome.

114. **What is the cut off level of imbecile?**

A. 0-30
B. **30-49**
C. 50-69
D. >70

Ans B An Idiot has IQ < 30, an imbecile has IQ of 30-50, a moron has an IQ of 50-70

115. **All the following are recognized stereotypic movements EXCEPT**

A. hand shaking
B. **eye blinking**
C. body rocking
D. head banging
E. self-biting

Ans B considered as simple motor tic, others are sterotypic movements. Unlike tic disorders, the stereotypic movements typically begin within the first 3 yr of life. These movements resolve over time. Among individuals with intellectual disability, the stereotyped behaviors may persist for years. Stereotypic movements are a common manifestation of a variety of neurogenetic disorders, such as Lesch-Nyhan syndrome, Rett syndrome, fragile X syndrome, Cornelia de Lange syndrome, and Smith- Magenis syndrome.

116. **All the following are true about truancy EXCEPT**

A. **it is normal behavior in young children**
B. it represent disorganization within the home
C. it may reflect underlying child abuse
D. depression may be an association
E. adolescent may be at risk of substance abuse

Ans A Truancy (running from school) is a never developmentally appropriate. Truancy may represent disorganization within the home, care taking needs of younger siblings, developing conduct problems, or emotional problems including depression or anxiety. Whereas younger children may threaten to run away out of frustration or a desire to get back at parents, older children who run away are almost always expressing serious underlying problem within themselves or their family, including violence, abuse, and neglect.

117. **The neurodevelopmental function (cognition) is dependent mainly on the development of the following functions EXCEPT**

A. sensory and motor
B. language
C. visual–spatial
D. intellectual
E. **cerebeller**

Ans E Neurodevelopmental function is a basic brain process needed for learning and productivity. It needs all that process that mentioned above plus memory, social cognition and frontal lobe functions. The process of attention and executive functioning is heavily dependent on frontal lobe function.

118. **You are a newly employee Pediatrician to foster care children (children removed from their original families and put in suitable institutes for protection). Those children have many significant problems, but the MOST common one is**

A. chronic medical problems
B. abuse and neglect
C. mental health concerns
D. **family relationship problems**
E. developmental problems

Ans D All children in foster care have family relationship problems, while 40-60% for choice A, >70% for choice B, 80% for choice C, and around 60% for choice E.

119. **A 3-year-old female child with repeated admissions as bleeding tendency in a form of spontaneous ecchymosis and bleeding through the nose and urine. She had another daughter who died because of similar undiagnosed illness. The mother is a staff nurse and she is highly concerned about her child illness. Lab investigations usually show either PT and/or PTT prolongation with normal platelets, which usually normalized after empiric vitamin K administration. The father is a petrol engineer and never seen accompanying the family. Of the following, the MOST helpful investigation is**

A. factor VII assay
B. factor II assay
C. **drug level**
D. von Willebrand factor assay
E. platelets function test

Ans C The presentation of medical child abuse (MCA) formerly called Munchausen syndrome by proxy; may vary in nature and severity. Consideration of MCA should be triggered when the reported symptoms are repeatedly noted by only one parent, appropriate testing fails to confirm a diagnosis, and seemingly appropriate treatment is ineffective.

23

CHAPTER 23

Musculoskeletal System



WHAT THIS CHAPTER COVERS

- Rickets & Osteomalacia
- Scurvy
- Osteogenesis Imperfecta
- Achondroplasia & Skeletal Dysplasias
- Congenital Talipes Equinovarus
- Developmental Dysplasia of Hip
- Perthes Disease
- Slipped Capital Femoral Epiphysis
- Transient Synovitis
- Septic Arthritis
- Osteomyelitis
- Juvenile Idiopathic Arthritis
- Systemic Lupus Erythematosus
- Dermatomyositis
- Muscular Dystrophies
- Spinal Deformities
- Fractures & Trauma

125 MCQs **17** subtopics

23.1 Metabolic Bone Disease (orthopaedic view)

- **Rickets** – see Nutrition: genu varum (< 2 y) / valgum (> 3 y), **windswept deformity**, widened wrists, rachitic rosary; X-ray – **cupping, fraying, splaying**.
- **Physiological knee alignment**: **genu varum at birth–2 y (max at 6–12 m) → neutral at 18–24 m → genu valgum peaks at 3–4 y (up to 15°) → adult alignment by 7–8 y**. Pathological: unilateral, progressive, beyond expected age, short stature, > 2 SD.
- **Blount disease (tibia vara)**: **infantile (< 4 y) – obese early walkers, African; medial proximal tibial physis growth disturbance**; **metaphyseal–diaphyseal angle > 11° (Drennan)**, beak on X-ray (Langenskiöld stages); Rx bracing (< 3 y), osteotomy.
- **Scurvy** – see Nutrition (pseudoparalysis, subperiosteal haemorrhage, Frankel line, Wimberger ring).

23.2 Osteogenesis Imperfecta (OI)

- **Type I collagen defect – COL1A1/COL1A2, mostly AD** (some AR – CRTAP, LEPRE1); "brittle bone disease".
- **Triad: fragile bones (multiple fractures with minimal trauma), blue sclerae, hearing loss (conductive/otosclerosis – adolescence/adult)**; **dentinogenesis imperfecta (opalescent teeth)**, joint laxity, short stature, **Wormian bones (skull)**, bowing, scoliosis, basilar invagination.

Sillence type	Severity	Sclerae	Features
I (MC, mildest)	Mild, non-deforming	**Blue**	Fractures with walking age, ↓ after puberty, hearing loss (50%), normal height
II	**Perinatal lethal**	Dark blue	**Multiple intrauterine fractures, "crumpled" beaded ribs, concertina femurs**, pulmonary hypoplasia
III	**Progressively deforming (most severe surviving)**	Blue at birth → white	Severe short stature, triangular face, wheelchair-bound
IV	Moderate	**Normal/white**	Variable fractures, dentinogenesis imperfecta
V	Moderate	White	**Hyperplastic callus**, calcified interosseous membrane

- **Differential – non-accidental injury** (OI has blue sclerae, Wormian bones, osteopenia, family history, dentinogenesis).
- Rx: **bisphosphonates (IV pamidronate/zoledronate) – ↑ bone density, ↓ fractures & pain**; **intramedullary rodding (telescoping – Fassier–Duval/Sofield)**, physiotherapy, hearing aids; denosumab (type VI).

23.3 Achondroplasia & Skeletal Dysplasias

- **Achondroplasia** – **MC non-lethal skeletal dysplasia & MC cause of short-limbed dwarfism**; **FGFR3 gain-of-function (G380R), AD – 80% new mutations (advanced paternal age)**; defective **endochondral ossification** (membranous ossification normal).
- Features: **rhizomelic (proximal) limb shortening**, **macrocephaly with frontal bossing**, **midface hypoplasia, depressed nasal bridge**, **trident hand (gap between 3rd & 4th fingers)**, **exaggerated lumbar lordosis, thoracolumbar gibbus (infancy)**, genu varum, limited elbow extension, **normal trunk length & normal intelligence**; motor delay (hypotonia).
- **X-ray**: **"champagne-glass" pelvis (narrow, flat inlet), square iliac wings, horizontal acetabular roofs**, **narrowing of interpedicular distance in lumbar spine (caudally – spinal stenosis)**, short vertebral pedicles, bullet-shaped vertebrae (infancy), **"chevron" deformity of metaphyses**, small foramen magnum.
- **Complications**: **foramen magnum stenosis → cervicomedullary compression, central apnoea, sudden death in infancy**, hydrocephalus, OSA, **lumbar spinal stenosis (adults)**, otitis media, obesity.

- Rx: **vosoritide (C-type natriuretic peptide analogue) – ↑ growth velocity**; monitor (polysomnography, MRI craniocervical junction); surgical decompression; limb lengthening (controversial); **GH not effective**.
- **Hypochondroplasia** (FGFR3 – milder), **thanatophoric dysplasia (FGFR3 – lethal; "telephone-receiver" femurs, cloverleaf skull type II, small chest)**.
- **Pseudoachondroplasia (COMP)** – normal face/head at birth, short limbs appear at 2 y.
- **Spondyloepiphyseal dysplasia (COL2A1)** – short trunk, myopia/retinal detachment, cleft palate, odontoid hypoplasia.
- **Ellis–van Creveld (chondroectodermal dysplasia)** – **short limbs + postaxial polydactyly + natal teeth + CHD (common atrium) + nail dysplasia**; Amish.
- **Jeune (asphyxiating thoracic dystrophy)** – narrow chest, respiratory failure, renal disease.
- **Morquio** – short trunk; **Hurler** – dysostosis multiplex (see IEM).
- **Osteopetrosis (marble bone disease)** – defective osteoclasts; **dense brittle bones, "bone-within-bone", "Erlenmeyer flask"**, pancytopenia (marrow obliteration), **cranial nerve compression (blindness, deafness, facial palsy)**, hepatosplenomegaly (extramedullary haematopoiesis); infantile malignant (AR) – **HSCT**.
- **Cleidocranial dysplasia (RUNX2)** – absent clavicles, open fontanelles, supernumerary teeth.

23.4 Congenital Talipes Equinovarus (CTEV – Clubfoot)

- **MC congenital foot deformity**; 1–2/1000; **boys 2:1, bilateral 50%**; idiopathic (multifactorial) vs syndromic (**arthrogryposis, myelomeningocele, Edwards**).
- **Deformity components – "CAVE"**: **Cavus (midfoot), Adductus (forefoot), Varus (hindfoot – heel inversion), Equinus (ankle – plantar flexion)**; **talus is the primary bone involved (neck deviated medially & plantarward)**; small calf, short foot, deep medial & posterior creases.
- **Pirani score (0–6)** and Dimeglio for severity.
- Differentiate from **postural clubfoot** (fully passively correctable – positional in utero; physiotherapy).
- **Treatment – Ponseti method (gold standard; start within 1–2 weeks of birth)**: **serial manipulation & long-leg plaster casts weekly (5–6 casts) correcting in order CAVE – cavus first (by supinating forefoot/elevating 1st metatarsal), then adductus & varus together (abduct around talar head – fulcrum at head of talus), equinus last → percutaneous Achilles tenotomy (~80–90%) → foot abduction brace (Denis Browne/Steenbeek – 70° external rotation affected side) full-time 3 months then nights till 4–5 years** (non-compliance → relapse – MC cause of failure). **Never dorsiflex before correcting cavus/varus (rocker-bottom foot)**; never pronate.
- Relapse – repeat casting, **tibialis anterior tendon transfer (2.5–5 y)**; neglected – soft-tissue release (posteromedial), osteotomies, triple arthrodesis (> 10 y), Ilizarov.
- **Other foot deformities**: **metatarsus adductus (MC foot deformity in newborns)** – flexible, resolves; **congenital vertical talus (rocker-bottom – Edwards, MMC)**; **calcaneovalgus** (positional, benign); **flat feet (physiological < 6 y; flexible flatfoot – arch appears on tiptoe – reassure)**; tarsal coalition (rigid flatfoot, adolescents).

23.5 Developmental Dysplasia of the Hip (DDH)

- Spectrum: **dysplasia → subluxation → dislocation**; **left hip (MC – 60%; left occiput anterior position)**, bilateral 20%.
- **Risk factors – "4 Fs"**: **F**emale (6:1), **F**irst-born, **F**amily history, **F**rank **breech** (strongest risk factor) + oligohydramnios, **swaddling with hips extended**, torticollis, metatarsus adductus, CTEV (packaging disorders).
- **Examination (newborn – up to 3 months)**: **Ortolani test – reduction of dislocated hip (abduction + lifting greater trochanter → "clunk")**; **Barlow test – dislocation of a dislocatable (unstable) hip (adduction + posterior push)**. **"Ortolani – Out → In (reduces); Barlow – In → Out (dislocates)"**.
- **After 3 months**: **limited hip abduction (most sensitive sign > 3 m)**, **asymmetric thigh/gluteal folds**, **apparent limb shortening – Galeazzi (Allis) sign (knee height lower on affected side; unilateral)**, Klisic test; **walking age – painless limp, Trendelenburg gait, waddling gait (bilateral), lumbar lordosis, toe-walking**; **telescoping**.

- **Imaging**: **ultrasound (Graf method – α angle < 60° abnormal) – IOC < 4–6 months** (femoral head not ossified); **X-ray after 4–6 months** (ossific nucleus appears) – **Hilgenreiner's line, Perkin's line (head should be in lower inner quadrant), Shenton's line (broken), ↑ acetabular index (> 30°)**, delayed ossific nucleus; von Rosen view.

Age	Treatment
Birth – 6 months	**Pavlik harness** (flexion > 90° & abduction – dynamic; avoid forced abduction → **AVN**); triple nappies NOT effective
6–18 months	**Closed reduction under GA + adductor tenotomy + hip spica cast** (human position); arthrogram
18 months – 3–4 years	**Open reduction ± pelvic (Salter innominate) / femoral (derotation/shortening) osteotomy**
> 4–8 years	Open reduction + osteotomies (Pemberton, Dega); unilateral > 8 y, bilateral > 6 y – often left alone

- **Complication**: **avascular necrosis of femoral head (MC iatrogenic complication)**, redislocation, residual dysplasia → early OA.

23.6 Perthes Disease (Legg–Calvé–Perthes)

- **Idiopathic avascular necrosis of femoral capital epiphysis**; **boys 4–8 years (4:1)**, **bilateral 10–15% (not simultaneous)**, short stature, **delayed bone age**, ADHD, passive smoking, low birth weight.
- **Painless limp (MC) or limp with pain referred to the knee (obturator nerve)**, **↓ abduction & internal rotation**, thigh atrophy, Trendelenburg sign, mild shortening.
- **Waldenström stages**: **initial (sclerosis/dense, smaller epiphysis, widened medial joint space; crescent sign – subchondral fracture)** → **fragmentation** → **reossification** → healed (residual – coxa magna, coxa plana, short neck).
- **Earliest X-ray sign: widening of medial joint space (Waldenström sign)**; **MRI/bone scan** – earliest diagnosis (avascular).
- **Classification**: **Catterall (I–IV, % head involved)**, **Herring lateral pillar (A, B, B/C, C – best prognostic)**, Salter–Thompson; **"head-at-risk" signs (Catterall)** – Gage sign (V-shaped radiolucency lateral epiphysis), lateral calcification, lateral subluxation, horizontal growth plate, metaphyseal cysts.
- **Prognosis – best predictor: age at onset (< 6 y good)**, lateral pillar involvement, sphericity at maturity (Stulberg).
- **Rx – containment** of femoral head in acetabulum: **< 6 y or lateral pillar A – observation, NSAIDs, ROM exercises/physio**; **> 8 y or pillar B/C – surgical containment (femoral varus derotation osteotomy, Salter osteotomy, shelf)**; abduction braces (Petrie casts).

23.7 Slipped Capital Femoral Epiphysis (SCFE)

- **MC hip disorder of adolescents**; **obese (MC risk factor) boys 10–16 y (girls 10–14)**, during growth spurt; **left hip MC; bilateral 20–40%** (prophylactic pinning of contralateral in endocrine/young); endocrine – **hypothyroidism, GH therapy, hypogonadism, renal osteodystrophy, radiation**.
- **Epiphysis slips posteriorly & medially (inferiorly) relative to metaphysis** (metaphysis moves anteriorly & superiorly – external rotation).
- **Hip/groin pain or knee/thigh pain (referred) – often missed**, limp; **obligatory external rotation on hip flexion (Drehmann sign)**, ↓ internal rotation, abduction & flexion.
- **Stable (can bear weight) vs unstable (cannot walk even with crutches – high AVN risk 20–50%)** – Loder.
- **X-ray – AP & frog-leg lateral (lateral more sensitive)**: **Trethowan's sign – Klein's line (along superior femoral neck) fails to intersect the epiphysis**, **widening & irregularity of physis**, **Steel's blanch sign (metaphyseal blanch – double density)**, reduced epiphyseal height; Southwick angle (severity – mild < 30°, moderate 30–50°, severe > 50°).
- **Rx: urgent – stop weight-bearing; in-situ fixation with a single cannulated screw (TOC)** – **no forceful reduction (AVN)**; severe – Dunn osteotomy/modified Dunn.

- Complications: **AVN (unstable SCFE, forceful reduction)**, **chondrolysis**, femoroacetabular impingement, OA, slip of other side.

23.8 Transient Synovitis, Septic Arthritis & Osteomyelitis

The limping child – age-wise

Age	Common causes
1–3 y	**Toddler's fracture (spiral fracture of distal tibia)**, septic arthritis, DDH, NAI, infection, neuromuscular (CP), leukaemia
4–10 y	**Transient synovitis (MC cause of acute painful limp)**, **Perthes**, septic arthritis, JIA, leukaemia, osteomyelitis
11–16 y	**SCFE**, overuse (Osgood–Schlatter), tumour (osteosarcoma, Ewing), JIA (ERA), osteomyelitis, AVN

Feature	Transient synovitis (irritable hip)	Septic arthritis
Age	**3–8 y, boys**, after URTI	Any (< 2 y – hip MC)
General	**Well-looking**, afebrile/low-grade	**Toxic, high fever**
Weight bearing	Usually possible (limp)	**Refuses**
Hip position	Mild flexion; ↓ ROM at extremes	**Held in flexion, abduction & external rotation**; severe pain on any movement
Labs	Normal	**WBC > 12,000, ESR > 40, CRP > 2 mg/dL**
USG	Effusion	Effusion
Rx	**Rest, NSAIDs – resolves in 1–2 weeks**	**Emergency aspiration/arthrotomy & washout + IV antibiotics**

- **Kocher criteria (septic hip)**: **non-weight-bearing, fever > 38.5 °C, ESR > 40 mm/h, WBC > 12,000/mm³** (+ CRP > 2 mg/dL – Caird) – **4/4 → 99% probability**.
- **Septic arthritis**: **S. aureus (MC overall)**; **Kingella kingae (MC in 6 m–4 y in some regions)**, **neonates – S. aureus, GBS, gram-negatives**; **adolescents – N. gonorrhoeae**; **sickle cell – Salmonella**; Hib (unvaccinated). **Hip (MC in infants – AVN & destruction risk: surgical emergency)**, knee (MC overall in children).
  - **Joint aspirate – WBC > 50,000/mm³ (> 75% PMN), low glucose, Gram stain, culture** – diagnostic; blood culture (+ in 40%).
  - Rx: **urgent joint drainage (arthrotomy for hip)** + **IV cloxacillin/cefazolin (± vancomycin – MRSA)**, ceftriaxone (neonates/gonococcus); switch to oral after improvement (CRP); total 3–4 weeks.
  - **Tom Smith arthritis** – septic arthritis of infant hip with destruction of femoral head.
- **Acute haematogenous osteomyelitis**: **metaphysis of long bones (sluggish flow in metaphyseal sinusoids/ hairpin loops)** – **distal femur, proximal tibia (MC)**; **S. aureus (MC at all ages)**, **Salmonella (sickle cell)**, **Pseudomonas (puncture wound through sneaker – calcaneus)**, Kingella; **fever, severe localised bony tenderness (earliest sign), refusal to use limb (pseudoparalysis)**; **X-ray normal for first 10–14 days** (earliest – deep soft-tissue swelling at 3 days; periosteal reaction 10–14 d); **MRI – IOC (earliest)**; bone scan; blood culture; Rx **IV antibiotics → oral; total 4–6 weeks**; drainage of subperiosteal abscess; complications – **chronic osteomyelitis (sequestrum – dead bone; involucrum – new periosteal bone; cloaca)**, septic arthritis (**neonates, proximal femur/humerus where metaphysis is intra-articular**), growth disturbance, pathological fracture.
- **Brodie's abscess** – subacute osteomyelitis (localised abscess with sclerotic rim, tibia metaphysis).
- **Chronic recurrent multifocal osteomyelitis (CRMO)** – autoinflammatory, sterile.
- **TB of spine (Pott's)** – thoracolumbar, gibbus, cold abscess (psoas), paraplegia; **TB hip** – painful limp, flexion-adduction deformity later.

23.9 Juvenile Idiopathic Arthritis (JIA)

- **Arthritis of unknown cause, onset < 16 years, persisting ≥ 6 weeks** (ILAR), other causes excluded; **MC chronic rheumatic disease of childhood**.

Subtype (ILAR)	%	Features	Labs
Oligoarticular (≤ 4 joints in first 6 months)	**MC (40–50%)**	**Young girls (2–4 y)**, **large joints – knee (MC), ankle**, asymmetric; **chronic anterior uveitis (iridocyclitis – 20%, asymptomatic, ANA+) → regular slit-lamp screening (every 3 months if ANA+)**; leg-length discrepancy; persistent vs extended	**ANA + (70–80%)**, RF –
Polyarticular RF-negative (≥ 5 joints)	20–25%	Girls; symmetric, large & small joints	RF –, ANA ±
Polyarticular RF-positive	5%	**Adolescent girls; resembles adult RA** – symmetric small joints, **rheumatoid nodules**, erosive, worst joint prognosis	**RF +, anti-CCP +**
Systemic-onset (Still disease)	10%	**Quotidian (daily spiking) fever ≥ 2 weeks (once/twice daily, returns to baseline) + evanescent salmon-pink macular rash with fever + arthritis**; hepatosplenomegaly, lymphadenopathy, **serositis (pericarditis)**; any age, M = F; **macrophage activation syndrome (MAS – secondary HLH; ↓ ESR paradoxically, ↓ platelets, ↑↑ ferritin, ↓ fibrinogen) – life-threatening**	**↑↑ ESR, CRP, ferritin, leucocytosis, thrombocytosis, anaemia**; ANA & RF –
Enthesitis-related arthritis (ERA)	10%	**Boys > 6 y; lower-limb oligoarthritis, enthesitis (heel – Achilles/plantar fascia), sacroiliitis/back pain**; **acute symptomatic anterior uveitis (red painful eye)**; family history; may evolve to ankylosing spondylitis	**HLA-B27 + (~90%)**
Psoriatic	5–7%	Arthritis + psoriasis or **dactylitis, nail pitting**, family history	
Undifferentiated			

TREATMENT OF JIA (ACR)

NSAIDs (naproxen, ibuprofen) – symptomatic; **intra-articular triamcinolone hexacetonide (oligoarticular – first-line)**; **methotrexate (DMARD of choice – 10–15 mg/m²/week + folic acid)** for polyarticular/persistent; **biologics – anti-TNF (etanercept, adalimumab – also uveitis), tocilizumab (IL-6), abatacept**; **systemic JIA – IL-1 (anakinra, canakinumab) or IL-6 (tocilizumab) blockade, steroids**; MAS – high-dose methylprednisolone, ciclosporin, anakinra. **Uveitis – topical steroids, MTX, adalimumab.** Physiotherapy, calcium/vitamin D, ophthalmology screening.

- **Differentials of arthritis in children**: **reactive (post-enteric/post-streptococcal)**, septic, **ARF (migratory, aspirin-responsive)**, **leukaemia (night pain, bone pain disproportionate to arthritis, cytopenias, ↑ LDH)**, **HSP, Kawasaki**, SLE, serum sickness, haemophilia, Lyme, TB, scurvy, viral (parvovirus, rubella).

23.10 Other Paediatric Orthopaedic Conditions

Condition	Key points
Osgood–Schlatter disease	**Traction apophysitis of tibial tuberosity**; active adolescent boys (10–15 y – jumping/running); painful bump below knee; self-limiting (rest, ice, NSAIDs)
Sinding-Larsen–Johansson	Inferior pole of patella apophysitis
Sever disease	**Calcaneal apophysitis** (heel pain, 8–12 y)
Köhler disease	AVN of **navicular** (3–7 y boys)
Freiberg disease	AVN of **2nd metatarsal head** (adolescent girls)

Condition	Key points
Osteochondritis dissecans	**Lateral part of medial femoral condyle (MC)**; adolescents; locking
Kienböck / Panner	Lunate / capitellum osteochondrosis
Scheuermann disease	Adolescent thoracic kyphosis – **anterior wedging ≥ 5° of ≥ 3 adjacent vertebrae**, Schmorl nodes
Adolescent idiopathic scoliosis	**MC type of scoliosis (80%)**; **girls; right thoracic curve (MC)**; **Adams forward-bend test** (rib hump – scoliometer > 7°); **Cobb angle** on X-ray; Rx observe < 25°, **brace (Boston) 25–40° in growing child**, **surgery (spinal fusion) > 45–50°**; **left thoracic curve – suspect neural axis abnormality (syrinx, Chiari) → MRI**
Congenital scoliosis	Vertebral anomalies (hemivertebra) – associated renal & cardiac anomalies (VACTERL)
Torticollis	**Congenital muscular (SCM fibrosis – sternomastoid tumour, MC)** – stretching; others – Klippel–Feil, **Sandifer (GERD)**, posterior fossa tumour, atlantoaxial rotatory subluxation (**Grisel syndrome – after pharyngitis**), ocular (IV nerve palsy)
Klippel–Feil syndrome	**Congenital fusion of cervical vertebrae – triad: short neck, low posterior hairline, restricted neck movement**; **Sprengel shoulder (high scapula – omovertebral bar)**, renal anomalies, deafness
Sprengel deformity	**Undescended scapula** (MC congenital shoulder anomaly)
Pulled elbow (nursemaid's elbow)	**Radial head subluxation through annular ligament**; **1–4 y**, sudden pull on extended pronated arm; child holds arm pronated & refuses to use; **no X-ray needed – reduce by supination + flexion (or hyperpronation)**
Supracondylar humerus fracture	**MC elbow fracture in children (5–7 y), extension type (FOOSH)**; complications – **brachial artery injury, Volkmann's ischaemic contracture (compartment syndrome – pain on passive finger extension, earliest sign)**, **AIN (median) nerve injury (MC nerve)**, **cubitus varus ("gunstock" – malunion, MC late complication)**
Lateral condyle fracture	Intra-articular, unstable → **cubitus valgus → tardy ulnar nerve palsy**
Salter–Harris physeal fractures	**SALTR**: I – Straight across (slipped), **II – Above (through metaphysis – MC)**, III – Lower (below – epiphysis), IV – Through everything, V – Rammed (crush – worst prognosis, growth arrest, often missed)
Greenstick, torus (buckle), plastic deformation	Unique to children (soft, porous bone, thick periosteum); rapid healing, remodelling
Toddler's fracture	Spiral, non-displaced distal tibia fracture in 9 m–3 y
Arthrogryposis multiplex congenita	**Multiple congenital joint contractures**, ↓ fetal movement (amyoplasia – MC), normal intelligence, "wooden doll"; physiotherapy, splinting, surgery
Congenital pseudarthrosis of tibia	**NF-1 (50%)**, anterolateral bowing → fracture → non-union
Fibrous dysplasia	**Shepherd's crook deformity of femur**, ground-glass X-ray, McCune–Albright
Unicameral (simple) bone cyst	Proximal humerus metaphysis, **"fallen fragment" sign** after fracture
Aneurysmal bone cyst	Eccentric, expansile, **fluid-fluid levels on MRI**, "soap-bubble"
Osteoid osteoma	**Night pain relieved by aspirin/NSAIDs**, nidus < 2 cm with sclerosis (femur/tibia)
Osteochondroma (exostosis)	MC benign bone tumour; metaphysis pointing away from joint; multiple – HME (EXT1/2)
Muscle diseases (DMD, SMA)	See Genetics & Neurology

Practice MCQs · Musculoskeletal System

125 questions · correct option highlighted in green · cover the answer strip to self-test

Rickets & Osteomalacia

1. A 4-year-old child presented with painless genu varum. X-ray bilateral knee was done and is shown below: (AIIMS May 2018) Which of the following is most likely diagnosis?

A. **Rickets**
B. Scurvy
C. Congenital anomaly
D. Trauma to epiphysis

Ans A The given X-ray shows cupping, fraying and splaying of ends of long bones suggestive of Rickets.

2. Which of the following conditions typically has rickets with normal or low calcium, elevated phosphorus, elevated parathormone, and elevated alkaline phos- phatase?

A. Vitamin D deficiency
B. Fanconi syndrome
C. Dent's disease
D. **Chronic kidney disease**

Ans D All of the above conditions are causes of Rickets, so elevated alkaline phosphatase is seen in all the above. For all practical purposes, Chronic kidney disease is the only condition out of the above, where Phosphate is elevated.

3. Causes of Rickets include all EXCEPT:

A. Renal Tubular Acidosis
B. Chronic Kidney Disease
C. Benign Mesenchymal Tumors
D. **Severe Malnutrition**

Ans D Rickets is a disease of growing bones and is not seen in severe malnutrition.

4. Abnormality in RTA with rickets is:

A. Excessive ossification
B. Excessive mineralization
C. Defective resorption
D. **Defective mineralization**

Ans D All of above Rickets is mainly seen in Distal Renal tubular acidosis, where,
• • Acidosis is due to loss of Bicarbonate
• • There is also hypokalemia due to Potassium loss, along with hypocalcemia.

5. Which of the following is not a sign of active rickets?

A. Prominent fontanelle
B. Hot cross bun sign
C. **Saddle nose**
D. Caries teeth &

Ans C Saddle nose is not seen in rickets, but in Congenital Syphilis.

6. Rickets in infant present as all EXCEPT:

A. Craniotabes
B. Widened fontanel
C. Rachitic Rosary
D. **Bow legs**

Ans D Bowing of legs appears only after weight bearing; Hence, Bow legs is a feature of rickets in toddlers and not infants.

7. In Rickets which of the following is/are seen:

A. **Increased alkaline phosphatase**
B. Decreased alkaline phosphatase
C. Hypophosphatemia in blood
D. Hypocalcemia
E. Alkalosis

Ans A c. Hypophosphatemia in blood, d. Hypocalcemia

8. A 2-year-old boy has clinical features of rickets. His investi- gations revealed serum Calcium-9 mg/dL, Phosphate-2.4 mg/dL, alkaline phosphatase – 1041 IU, normal intact parathyroid hormone and bicarbonate 22 mEq/L. Which of the following is the most probable diagnosis:

A. Distal renal tubular acidosis
B. **Hypophosphatemic rickets**
C. Vitamin D dependent rickets
D. Hypoparathyroidism

Ans B Clues in this question are – Normal calcium, Low phosphate, Normal parathormone, & Elevated alkaline phosphatase All these suggest the diagnosis of hypophosphatemic rickets.

9. True about Vit D deficiency rickets:

A. Vit D given at a dose of 50–100 mg/day
B. **X-ray knee joint is diagnostic**
C. Rachitic rosary is tender
D. Increased chances of respiratory tract infections
E. Hyponatremia

Ans B d. Increased chances of respiratory tract infections Discussing about the options one by one, a. False Daily dose of Vit D for treatment of Rickets is 2,000–5,000 IU/day X 4-6 wks b. True X-ray wrist or knee are usually done to diagnose Rickets c. False Rachitic rosary is nontender (in contrast to scorbutic rosary which is tender & more angular or sharp) d. True Risk of pneumonia appears to be elevated in children with rickets e. False Not seen in Rickets; Hypocalcemia is usually seen

10. True about nutritional rickets:

A. **Craniotabes**
B. Multiple fractures
C. Wrist widening
D. Increased serum phosphate
E. Growth retardation

Ans A c. Wrist widening, e. Growth retardation

11. All of the following are seen in Rickets EXCEPT:

A. Bow legs
B. **Gum bleeding**
C. Pot belly
D. Craniotabes

Ans B Gum bleeding is a feature of Scurvy, and not Rickets.

Scurvy

12. Widening of epiphysis is seen in:

A. Scurvy
B. **Rickets**
C. Achondroplasia
D. Osteopetrosis

Ans B Wrist widening is an important clinical feature of Rickets.

13. All are radiologic signs in scurvy EXCEPT:

A. White line in metaphysis
B. **Cupping and fraying**
C. Ground glass appearance
D. Zone of rarefaction

Ans B Radiological Signs of Scurvy include Ground-glass appearance of long bones due to trabecular atrophy with Pencil thin cortex, White line of Fränkel, Wimberger sign (Sclerotic ring around epiphysis), Trumerfeld zone, Pelkan spur and Subperiosteal hemorrhages;

14. Primary metabolic bone disorder in scurvy is:

A. Decreased mineralization
B. **Decreased osteoid matrix formation**
C. Increased bone resorption
D. Decreased bone mass with normal mineralization and osteoid formation

Ans B Pathogenesis of scurvy:
• • There is defective formation of connective tissues & collagen in skin, cartilage, bone, & blood vessels•→•↑•fragility
• • In the long bones, osteoid is not deposited by osteoblasts, cortex is thin, trabeculae become brittle & fracture easily.

15. About scurvy, true are all EXCEPT:

A. Subperiosteal hematoma with tenderness
B. Zone of rarefaction
C. **Increased serum alkaline phosphatase**
D. Gingival bleeding
E. Pelkan spurs JUVENILE IDIOPATHIC ARTHRITIS

Ans C Note: Alkaline Phosphatase level is elevated in Rickets and not in Scurvy.

16. Pediatricians must understand normal limb development so as to recognize pathologic conditions during routine and targeted exams. Of the following, the TRUE statement is

A. normal tibiofemoral angle at birth is 10-15 degrees of physiologic valgus
B. physiologic varus up to 12 degrees is reached in between 3 and 4 yr of age
C. normal varus of 7 degrees is achieved by 5-8 yr of age
D. **persistence of varus beyond 2 yr of age may be pathologic**
E. more than 35% of developmental genu varum and genu valgum cases need intervention

Ans D The normal tibiofemoral angle at birth is 10-15 degrees of physiologic varus. The alignment changes to 0 degrees by 18 mo, and physiologic valgus up to 12 degrees is reached in between 3 and 4 yr of age. The normal valgus of 7 degrees is achieved by 5- 8 yr of age. Persistence of varus beyond 2 yr of age may be pathologic and is seen in conditions such as Blount disease. Overall, 95% of developmental physiologic genu varum and genu valgum cases resolve with growth.

17. Which medical condition is regarded a contraindication for sport participation?

A. atlantoaxial instability
B. splenomegaly
C. **carditis**
D. long-QT syndrome
E. advanced heart block

Ans C can result in sudden death with exertion. All the other conditions need individual assessment.

Osteogenesis Imperfecta

18. An infant presents with lethargy, frontal bossing and bowing of legs; X-ray shows widening of epiphysis. What is the diagnosis?

A. Scurvy
B. **Rickets**
C. Osteomalacia
D. Osteopetrosis

Ans B

19. Osteogenesis imperfecta with normal dentition and sclera:

A. **Type 1A**
B. Type
C. Type
D. Type 4B

Ans A Osteogenesis imperfecta (OI) is the most common genetic cause of osteoporosis; OI Type I is a mild disease. Both types I & IV are divided into A and B subtypes, depending on the absence (A) or presence (B) of dentinogenesis imperfecta.

20. **Juvenile localized scleroderma (JLS) is generally insidious with skin manifestations that vary according to disease subtype. Up to 25% of children with LS have extracutaneous manifestations, including arthritis and neurological symptoms. Of the following, the subtype that is MOST commonly associated with neurological manifestations is**

A. plaque morphea
B. eosinophilic fasciitis
C. generalized morphea
D. **en coup de sabre**
E. morphea profunda

Ans D Children with en coup de sabre may have symptoms unique to central nervous system involvement, such as seizures, hemifacial atrophy, ipsilateral uveitis, and learning/behavioral changes.

21. **You are evaluating a 1-year-old boy with an established diagnosis of Kawasaki disease (KD) since 4 days; the caring nurse asks you about cardiac involvement during this stage. All the following are possible cardiac complications at this stage EXCEPT**

A. myocarditis
B. pericarditis
C. **coronary artery aneurysm**
D. mitral regurgitation
E. cardiogenic shock

Ans C usually develops in the subacute phase in the 2nd or 3rd week of the illness.

22. **In evaluation of concerns relating to the limb, the examination should assess the exact torsional profile which include all the following EXCEPT**

A. foot progression angle
B. femoral anteversion
C. **knee – knee distance**
D. tibial version with thigh–foot angle
E. assessment of foot adduction and abduction

Ans C

23. **Adverse effect of anabolic–androgenic steroids include all the following EXCEPT**

A. infertility
B. gynecomastia
C. female virilization
D. **hypotension**
E. epiphyseal closure

Ans D Hypertension.

Achondroplasia & Skeletal Dysplasias

24. **Most potent form of vitamin D is:**

A. Ergocalciferol
B. 7 dehydrocholecalciferol
C. 25 hydroxycholecalciferol
D. **1,25 dihydroxycholecalciferol**

Ans D

25. **A 7-year-old boy developed small hypopigmented depressed scars after fingernail scratches; he has been treated with nonsteroidal antiinflammatory drugs (NSAIDs) for a rheumatic disease 6 weeks ago. Of the following, the NSAID that is MOST likely to cause such a unique skin reaction is**

A. celecoxib
B. meloxicam
C. indomethacin
D. **naproxen**
E. ibuprofen

Ans D is more likely than other NSAIDs to cause pseudoporphyria. Pseudoporphyria is more likely to occur in fair-skinned individuals and on sun-exposed areas. If pseudoporphyria develops; the inciting NSAID should be discontinued because scars can persist for years or be permanent.

26. **Juvenile systemic scleroderma (JSS) has a prolonged course with periods of remission and exacerbation. It is characterized by multisystem organ involvement including skin, locomotor system, CNS, and viscera. Of the following, the MOST common early cutaneous manifestation is**

A. **edema of the dorsum of the hands and fingers**
B. induration and fibrosis of the skin
C. flexion contractures at the elbows, hips, and knees
D. skin ulceration over pressure points
E. subcutaneous calcifications

Ans A

27. **A follow up two-dimensional echocardiography is performed to a 1.5-year-old boy with Kawasaki disease (KD) 3 weeks after the diagnosis; it shows a small solitary aneurysm of the left anterior descending coronary artery without thrombosis. Of the following, the MOST appropriate therapy for this child is**

A. aspirin for 6 months
B. **life-long aspirin**
C. IVIG and aspirin for 14 days
D. aspirin and clopidogrel antiplatelet for 8 weeks
E. aspirin and warfarin for 6 months

Ans B The patient with KD who has had a small solitary aneurysm should continue aspirin indefinitely. Patients with larger or numerous aneurysms may require the addition of other antiplatelet agents or anticoagulation.

28. **In-toeing gait most commonly results from excessive femoral anteversion. Of the following, the TRUE statements is**

A. it is more common in boys than girls (2 : 1)
B. patellas are pointing outward when the foot is straight
C. diagnosis is made by CT
D. **treatment is predominantly observation**
E. anteversion >25 degrees is an indication for operative intervention

Ans D It occurs more commonly in girls than boys (2: 1) in children 3-6 yr of age. The patellas are pointing inward when the foot is straight, and compensatory external rotation of the tibia is demonstrated. Diagnosis is made clinically on examination; CT can provide objective measurements but is rarely indicated. The torsion usually corrects with growth by 8-10 yr of age.

29. **Creatine is an amino acid mostly stored in skeletal muscle increasing muscle performance. Its use has been increased when other supplements have been withdrawn from the market. Of the following, the possible adverse effect include**

A. mouth ulcer
B. carditis
C. hepatitis
D. **nephritis**
E. genital sore

Ans D Concerns about nephritis in case reports have not been supported by controlled studies.

Congenital Talipes Equinovarus

30. **Alkaline phosphatase level are not elevated in:**

A. Renal rickets
B. Nutritional rickets
C. **Hypophosphatasia**
D. Hypophosphatemic rickets

Ans C

31. **Sulfasalazine is an effective drug in many rheumatic diseases; however, it is associated with severe systemic hypersensitivity reaction. It is approved in all the following rheumatic disease EXCEPT**

A. polyarticular juvenile idiopathic arthritis (JIA)
B. oligoarticular JIA
C. **systemic JIA**
D. peripheral arthritis associated with juvenile ankylosing spondylitis
E. enthesitis associated with juvenile ankylosing spondylitis

Ans C Sulfasalazine is generally considered contraindicated in children with active systemic JIA because of increased hypersensitivity reactions.

32. **You are discussing Raynaud phenomenon (RP) and Raynaud disease (RD) with medical students. You state that RP is usually associated with rheumatic diseases, while RD is independent of an underlying rheumatic disease. Of the following, the feature that is MOST likely consistent with RD, rather than RP is**

A. presence in early childhood
B. **absence of tissue necrosis and gangrene**
C. asymmetric occurrence
D. presence of periungual telangiectasia
E. associated pain and paresthesia

Ans B RD often begins in adolescence and is characterized by symmetric occurrence, the absence of tissue necrosis and gangrene, and the lack of manifestations of an underlying rheumatic disease. Children have normal nail-fold capillaries. Pain and paresthesia indicate ischemic changes, associated only with RP due to rheumatic diseases.

33. **Henoch-Schönlein purpura (HSP) is the most common vasculitis of childhood affecting small vessels in the skin, joints, gastrointestinal tract, and kidney. Gastrointestinal involvements occur in up to 80% of children with HSP. Of the following, the LEAST gastrointestinal manifestation that may occur in children with HSP is**

A. abdominal pain
B. vomiting and diarrhea
C. paralytic ileus
D. melena
E. **intussusception**

Ans E mesenteric ischemia, and intestinal perforation are uncommon in children with HSP.

34. **A school age child presented with a mass behind the knee. Physical examination reveals a firm mass in the popliteal fossa, medially located and distal to the popliteal crease. The mass is most prominent when the knee is extended. Knee radiographs are normal. Of the following, the MOST likely diagnosis is**

A. osteochondroma
B. osteochondritis dissecans
C. **popliteal cyst**
D. malignancy
E. juvenile idiopathic arthritis

Ans C s, or Baker cysts, are cystic masses filled with gelatinous material that develop in the popliteal fossa, the shallow depression located at the posterior part of the knee. They are considered rare in children. They most commonly occur in the region of the medial head of the gastrocnemius and semimembranosus. Transillumination of the cyst on physical examination is a simple diagnostic test.

35. **In competitive swimming, the MOST common injuries are of the**

A. neck
B. **shoulder**
C. spine
D. hip
E. knee

Ans B Injuries of the shoulder in competitive swimming are most common and generally a result of chronic, overuse. Swimmer's shoulder is a combination of subacromial bursitis and tendinosis of the rotator cuff and long-head of the biceps.

Developmental Dysplasia of Hip

36. **Windswept deformity is seen in:**

A. Scurvy
B. **Rickets**
C. Achondroplasia
D. Osteoporosis SCURVY

Ans B Windswept deformity Definition A valgus deformity of one in association with varus deformity of other knee. Causes •• Rickets •• Physeal osteochondromatosis
• • Rheumatoid Arthritis •• Hereditary dysplasia (epiphyseal dysplasia) of bone. SCURVY

37. **Oligoarthritis predominantly affects the joints of the lower extremities rather than upper extremity joints. Which of the following joints is never a presenting sign of oligoarthritis?**

A. **hip**
B. knee
C. ankle
D. metatarsal
E. interphalangeal

Ans A Isolated involvement of the hip is almost never a presenting sign and suggests extended oligoarticular juvenile idiopathic arthritis.

38. **Behçet disease (BD) is a primary variable vessel vasculitis, characterized by exacerbations and remissions. The hallmark of the disease is oral ulceration. All the following are characteristic features of the oral ulcers EXCEPT**

A. very painful
B. recurrent
C. involve any location in the oral cavity
D. **heal with scarring**
E. last 3-10 days

Ans D The oral ulcers heal without scarring.In contrast; the genital ulcers heal with scars.

39. **A 5-year-old boy has acute onset of abdominal pain and melena; he has been diagnosed with Henoch-Schönlein purpura (HSP) one week before. Examination reveals symmetrical palpable purpura overlying the lower extremities and buttock; other examinations are unremarkable. Of the following, the BEST treatment for this boy is**

A. supportive measures
B. **steroids**
C. azathioprine
D. cyclophosphamide
E. mycophenolate mofetil

Ans B Steroid is most often used to treat significant gastrointestinal involvement or other life-threatening manifestations.

40. **An adolescent girl presented with pain beneath the patella, walking up and down stairs aggravates the pain; squatting, running, and other vigorous physical activities also exacerbate the pain. There is no history of antecedent trauma and there is no swelling. Pain is often relieved through knee extension. Of the following, the MOST likely diagnosis is**

A. osteochondroma
B. osteochondritis dissecans
C. popliteal cyst
D. **patellofemoral pain syndrome**
E. juvenile idiopathic arthritis

Ans D (PFPS) is one of the most common causes of knee pain, particularly in adolescent girls. The precise etiology of the knee pain remains unknown and is likely multifactorial.

41. **There is marked geographic and racial variation in the incidence of developmental dysplasia of the hip (DDH). All the following are true EXCEPT**

A. 1.7 in 1,000 babies in Sweden
B. 75 in 1,000 in Yugoslavia
C. 188.5 in 1,000 in a district in Manitoba, Canada
D. **3.7 in 1,000 babies in Chinese newborn**
E. 0% in African newborn

Ans D The incidence of DDH in Chinese and African newborns is almost 0%, whereas it is 1% for hip dysplasia and 0.1% for hip dislocation in white newborns. These differences may be the result of environmental factors, such as child-rearing practices, rather than to genetic predisposition.

Perthes Disease

42. **Subperiosteal hemorrhage is due to deficiency of:**

A. Vit A
B. Vit K
C. **Vit C**
D. Vit D

Ans C

43. **Rheumatoid factor (RF)–positive polyarthritis is characterized by aggressive symmetric inflammation of joints of both upper and lower extremities. Of the following, the extra articular manifestation that is almost exclusively occur in RF- positive individuals is**

A. fever
B. evanescent rash
C. uveitis
D. **extensor surfaces nodules**
E. pericarditis

Ans D Rheumatoid nodules on the extensor surfaces of the elbows, spine, and over the Achilles tendons, although unusual, are associated with a more severe course and almost exclusively occur in RF-positive individuals.

44. **You are evaluating a 16-year-old female adolescent who has been diagnosed with Behçet disease since the age of 12 year; she recently develops oral genital ulcers; examination reveals multiple oral ulcers involving the hard palate, multiple genital ulcers involving the labia majora, and erythema nodosum. There is no major organ involvement and uveitis. Of the following, the BEST treatment for this patient is**

A. **colchicine**
B. azathioprine
C. cyclophosphamide
D. systemic steroids
E. interferon alpha

Ans A In patients without major organ involvement, colchicine significantly improves oral and genital ulcers, skin features, and disease activity.In pediatric patients with vascular involvement with venous thrombosis, steroids and azathioprine have been used, whereas those with pulmonary arterial or cardiac involvement are initially treated with cyclophosphamide. Interferon alpha can be used for CNS vasculitis.

45. **Renal disease is the major long-term complication, occurring in 1-2% of children with Henoch-Schönlein purpura (HSP). Chronic HSP renal disease is managed with a variety of immunosuppressants. Of the following, the LEAST effective drug for the treatment of renal disease in HSP is**

A. prednisolone
B. azathioprine
C. cyclophosphamide
D. cyclosporine
E. mycophenolate mofetil

Ans A

46. **The proximal femoral ossification center (in the center of the femoral head) appears between**

A. 1st and 3rd mo
B. 4th and 7th mo
C. 8th and 11th mo
D. 12th and 15th mo
E. 16th and 18th mo

Ans B Between the 4th and 7th mo of life, the proximal femoral ossification center (in the center of the femoral head) appears.

47. **Legg-Calvé-Perthes disease is a hip disorder of unknown etiology that results from temporary interruption of the blood supply to the proximal femoral epiphysis, leading to osteonecrosis and femoral head deformity. Of the following, the MOST common presenting symptom is**

A. limp of varying duration
B. pain
C. failure to ambulate
D. atrophy of the muscles of the thigh
E. an apparent leg-length inequality

Ans A The most common presenting symptom is a limp of varying duration.

48. **Differential diagnosis of legg-calvé-perthes disease include all the following EXCEPT**

A. osteochondromatosis
B. Schwartz-Jampel syndrome
C. Marfan syndrome
D. Maroteaux-Lamy syndrome
E. Martsolf syndrome

Ans C Differential diagnosis of legg-calvé-perthes disease include: Sickle cell disease, other hemoglobinopathies (e.g., thalassemia), chronic myelogenous leukemia, steroid medication, sequela of traumatic hip dislocation, treatment of developmental dysplasia of the hip, septic arthritis, mucopolysaccharidoses, and hypothyroidism.

Slipped Capital Femoral Epiphysis

49. **A 5-month-old baby (exclusively cow feed) presented to pediatric OPD with history of crying on touch. Cause may be:**

A. Scurvy
B. Rickets
C. JRA
D. Caffey's disease

Ans A In scurvy, there is painful pseudoparalysis of limbs, due to subperiosteal hemorrhage; so the child cries on touching.

50. **You are discussing with the medical students the role of systemic steroids for the management of rheumatologic diseases. Your discussion should include all the following statements EXCEPT**

A. they are recommended for management of severe systemic illness
B. they are used for bridge therapy during the wait for therapeutic response to a disease modified anti rheumatic drugs (DMARD)
C. they are effective for control of uveitis
D. they prevent joint destruction
E. they impose risks of severe toxicities

Ans D Systemic steroids do not prevent joint destruction.

51. **Primary Sjögren syndrome, although rare, may occur in children between 9-10 year. Of the following, the MOST common manifestation in children is**

A. recurrent parotitis
B. sicca symptoms
C. polyarthritis
D. vulvovaginitis
E. hepatitis

Ans A Recurrentparotid gland enlargement and parotitis are the most common manifestations in children (>70%), whereas sicca syndrome (dry mouth, painful mucosa, sensitivity to spicy foods, halitosis, widespread dental caries) predominate in adults.

52. **Childhood vasculitis encompasses a broad spectrum of diseases that share in common inflammation of the blood vessels. Vascular injury includes small, medium, and large vessels. Of the following, the disease that affects predominantly large blood vessels is**

A. Henoch-Schönlein purpura
B. granulomatosis with polyangiitis (wegener granulomatosis)
C. childhood polyarteritis nodosa
D. Kawasaki disease
E. Takayasu arteritis

Ans E

53. **Which torsional defotmity can follow a slipped capital femoral epiphysis and increase incidence of degenerative arthritis?**

A. external tibial torsion
B. external femoral torsion
C. internal tibial torsion
D. internal femoral torsion
E. metatarsus adductus

Ans B can follow a slipped capital femoral epiphysis. The disorder is associated with an out-toeing gait and increased incidence of degenerative arthritis. If slipped capital femoral epiphysis is detected, it is treated surgically.

54. **The MOST common ocular manifestation in Marfan syndrome is**

A. dislocation of the ocular lens
B. severe myopia
C. flat cornea
D. hypoplastic iris
E. retinal detachment

Ans A (ectopia lentis) occurs in approximately 60-70% of patients, although it is not unique to the disorder.

Transient Synovitis

55. Child with frog like position and resistance to move the limbs, is probably suffering from:

A. Scurvy
B. Rickets
C. Trauma
D. Congenital dislocation

Ans A It is seen in pseudoparalysis, caused by painful subperiosteal hemorrhage in Scurvy.

56. Spondyloarthritides are complex diseases in which susceptibility is largely genetically determined. Which of the following characteristics is never associated with such diseases?

A. enthesitis
B. peripheral arthritis
C. axial arthritis
D. HLA-B27 positivity
E. rheumatoid factor positivity

Ans E Positive rheumatoid factor is never associated with all subtypes of spondyloarthritides.

57. A 4-year-old boy has an acute severe left groin pain that refers to thigh. Examination reveals limitation of movement of the left hip joint. Lab investigations reveal normal blood count and indices, and normal ESR; radiologic imaging shows widening of the hip joint space with effusion. You suspect transient synovitis. Of the following, the BEST therapeutic approach for this boy is

A. systemic steroids
B. methotrexate
C. aspiration of joint fluid
D. intravenous antibiotics
E. observation

Ans C In transient synovitis (toxic synovitis), aspiration of joint fluid is often necessary to exclude septic arthritis and typically results in dramatic clinical improvement.

58. Normal full-term newborns can have up to 20-30 degree hip and knee flexion contractures, these contractures tend to resolve by

A. 1-3 mo
B. 4-6 mo
C. 7-9 mo
D. 10-12 mo
E. 13-15 mo

Ans B Normal full-term newborns can have up to 20-30 degree hip and knee flexion contractures. These contractures tend to resolve by 4-6 mo of age.

59. The MOST reliable sign of a dislocated hip after the 2nd month of life is

A. limited hip abduction
B. apparent shortening of the thigh
C. proximal location of the greater trochanter
D. asymmetry of the glutealor thigh folds
E. positive Ortolani test

Ans A As the baby enters the 2nd and 3rd mo of life, the soft tissues begin to tighten and the Ortolani and Barlow tests are no longer reliable. Limitation of abduction is the most reliable sign of a dislocated hip in this age group.

60. Transient synovitis (toxic synovitis) is a reactive arthritis and is one of the most common causes of hip pain in young children. All the following clinical manifestations are true EXCEPT

A. it is most prevalent between 3 and 8 yr of age
B. 70% of affected children have had a preceding nonspecific upper respiratory tract infection
C. symptoms often develop acutely
D. children are usually able to bear weight on the affected limb
E. usually associated with a high grade fever

Ans E They are often afebrile or have a low-grade fever <38°C (100.4°F). The hip is not held flexed, abducted, or laterally rotated unless a significant effusion is present.

Septic Arthritis

61. Wimberger ring sign is present in:

A. Rickets
B. Scurvy
C. Secondary syphilis
D. Tuberculosis

Ans B

62. You are discussing with medical students Juvenile Ankylosing Spondylitis (JAS) subtype of spondyloarthritides. One of the students asks you about the features that distinguish this disease from the adult-onset ankylosing spondylitis (AOAS). All the following are true regarding JAS in comparison with AOAS EXCEPT

A. it is present in patients <16 yr old
B. axial disease occurs more frequently early in the disease course
C. inflammatory back pain is less frequent at disease onset
D. enthesitis occurs more commonly
E. peripheral arthritis is more common

Ans B In comparison to adult-onset AS, axial disease and inflammatory back pain are less frequent at disease onset, while enthesitis and peripheral arthritis is more common.

63. Familial Mediterranean fever (FMF) is a recessively inherited autoinflammatory disease usually characterized by recurrent self-limited episodes of fever, serositis, arthritis, and skin rash. Of the following, the hallmark cutaneous finding of this disease is

A. erysipelas-like erythema overlying the dorsum of the foot
B. morbilliform rash
C. migratory rash overlying area of myalgia
D. cold-induced urticaria like lesions
E. generalized pustulosis

Ans A

64. The ossification centers that are typically present at birth include all the following

A. distal femur
B. proximal tibia
C. distal humerus
D. calcaneus
E. talus

Ans C The ossification centers that are typically present at birth are the distal femur, proximal tibia, calcaneus, and talus.

65. **Torticollis, literally meaning twisted neck, is not a diagnosis but rather a manifestation of a variety of underlying conditions. All the following conditions may cause torticollis EXCEPT**

A. positional deformation
B. Klippel-Feil syndrome
C. cervical lymphadenitis
D. **supratentorial brain tumor**
E. upper lobe pneumonia

Ans D Posterior fossa brain tumor.

66. **Septic arthritis in infants and children has the potential to damage the synovium, adjacent cartilage, and bone causing permanent disability. All the following statements regarding its epidemiology are true EXCEPT**

A. **septic arthritis is more common in young children**
B. half of all cases occur by 5 yr of age
C. adolescents and neonates are at risk of gonococcal septic arthritis
D. majority of infections in otherwise healthy children are of hematogenous origin
E. infection of joints following penetrating injuries are uncommon

Ans A Half of all cases occur by 2 yr of age and three-fourths of all cases occur by 5 yr of age.

Osteomyelitis

67. **Pseudoparalysis in an infant is suggestive of:**

A. Acute rheumatic fever
B. Vit B6 deficiency
C. Vit E deficiency
D. **Vit C deficiency**

Ans D Pseudoparalysis Definition A voluntary restriction of motion, mainly because of pain, but not due to actual muscular paralysis Causes Scurvy, Osteomyelitis, surgery, fractures, myositis, metabolic myopathy, Congenital syphilis, septic arthritis

68. **Reactive arthritis is defined as joint inflammation caused by a sterile inflammatory reaction following a recent infection. Of the following, the LEAST likely micro-organism that may cause reactive arthritis is**

A. **Clostridium difficile**
B. Salmonella paratyphi
C. Shigella flexneri
D. Yersinia enterocolitica
E. Campylobacter jejuni

Ans A Reactive arthritis typically follows enteric infection with Salmonella sp., Shigella flexneri, Yersinia enterocolitica, Campylobacter jejuni, orgenitourinary tract infection with Chlamydia trachomatis. Escherichiacoli and Clostridium difficile are also causative enteric agents, although less common.

69. **Amyloidosis is the most serious complication of familial Mediterranean fever (FMF), and in its absence FMF patients may live a normal life span. Of the following, the organ that is not affected by secondary amyloidosis of FMF is**

A. kidney
B. lung
C. **nerve**
D. heart
E. teste

Ans C Amyloidosis may develop most commonly in the kidneys, gastrointestinal tract, spleen, lungs, testes, thyroid, and adrenals. Rarely, cardiac amyloidosis may develop; macroglossia and amyloid neuropathy is generally not seen with the amyloidosis of

70. **The gait of a child become similar to that of an adult at**

A. 3 yr
B. 5 yr
C. **7 yr**
D. 9 yr
E. 11 yr

Ans C The gait characteristics of a 7 yr old child are similar to those of an adult.

71. **Klippel-Feil syndrome includes the classic triad of a low posterior hairline, short neck, and decreased cervical range of motion. Other associations include the following EXCEPT**

A. Sprengel's deformity
B. congenital scoliosis
C. genitourinary anomalies
D. **conductive hearing loss**
E. congenital heart disease

Ans D Other associations include Sprengel's deformity (congenital elevation of the scapula), congenital scoliosis, genitourinary anomalies (25-35%), sensorineural hearing loss (5%), and congenital heart disease (5-10%).

72. **The two MOST common causative organisms of osteomyelitis in children with sickle cell anemia are**

A. S. aureus and streptococci
B. **Salmonella spp. and S. aureus**
C. S. aureus and Pseudomonas aeruginosa
D. Enterobacteriaceae and S. aureus
E. S. aureus and Bartonella henselae

Ans B Streptococcus pneumoniae most commonly causes osteomyelitis in children younger than 24 mo of age and in children with sickle cell anemia, but its frequency has declined because of pneumococcal conjugate vaccines. Cases of Pseudomonas infection are related almost exclusively to puncture wounds of the foot, with direct inoculation of P. aeruginosa from the foam padding of the shoe into bone or cartilage, which develops as osteochondritis.

73. **The second MOST common cause of osteomyelitis in children younger than 5 yr of age in some parts of the world is**

A. S. aureus
B. **Kingella kingae**
C. Pseudomonas aeruginosa
D. Enterobacteriaceae
E. Bartonella henselae

Ans B may be the second most common cause of osteomyelitis in children younger than 5 yr of age in some parts of the world. The organism is increasingly recognized as a cause of osteomyelitis, spondylodiskitis, and septic arthritis, especially when polymerase chain reaction testing is employed.

74. **The MOST common site of osteomyelitis in children is**

A. **femur**
B. tibia
C. humerus
D. radius
E. ulna

Ans A 23-28%, tibia 20-26%, humerus 8-13%, radius and ulna 5-6%.

Juvenile Idiopathic Arthritis

75. **Characteristic of systemic juvenile idiopathic arthritis**

A. Begins after 16 years of age
B. Uveitis is a feature
C. **RA factor is negative**
D. NSAIDs are contraindicated

Ans C JIA begins at < 16 year age; Uveitis is usually seen in oligoarticulat JIA; NSAIDs are used for symptomatic relief

76. **In systemic form of JIA, all are true EXCEPT:**

A. **RA factor positive**
B. High fever with rash
C. Hepatosplenomegaly
D. Elevated ESR

Ans A Systemic juvenile idiopathic arthritis is characteristically rheumatoid factor NEGATIVE; RF positivity is atypical and more typical of polyarticular RF-positive JIA.

77. **Juvenile idiopathic arthritis has many subtypes. Arthritis must be present to make a diagnosis of any subtype; involved joints oftenly have the following signs EXCEPT**

A. swelling
B. warm
C. **erythema**
D. limitation of movement
E. pain on movement

Ans C Involved joints are often swollen, warm to touch, and painful on movement or palpation with reduced range of motion, but usually are not erythematous.

78. **Oligoarthritis, the most common subtype of juvenile idiopathic arthritis, is defined as involving ≤4 joints within the 1st 6 mo of disease onset. Of the following, the MOST commonly affected joint is**

A. hip
B. **knee**
C. elbow
D. wrist
E. metatarsal

Ans B In oligoarthritis, it predominantly affects the large joints of the lower extremities, such as the knees and ankles. Isolated involvement of upper extremity large joints is less common.

79. **A 4-year-old girl recently diagnosed with persistent oligoarticular juvenile idiopathic rheumatoid arthritis (JIA); she has 3 involved joints including the right knee, right ankle, and left elbow; antinuclear antigen (ANA) is significantly positive. Of the following, the MOST important step in the management of this girl is**

A. regular examination of locomotor system
B. **periodic slit-lamp examination**
C. periodic ANA monitoring
D. periodic erythrocyte sedimentation rate (ESR) monitoring
E. frequent C-reactive protein (CRP) monitoring

Ans B The presence of a positive ANA confers increased risk for asymptomatic anterior uveitis, requiring periodic slit-lamp examination.

80. **Antinuclear antigen (ANA) measurement test is useful in some rheumatologic diseases especially with persistent oligoarticular juvenile idiopathic rheumatoid arthritis (JIA). All the following are more likely to be correlated with ANA positivity EXCEPT**

A. anterior uveitis
B. younger age at disease onset
C. female sex
D. **symmetrical arthritis**
E. lower number of involved joints over time

Ans D ANA positivity is more likely to be correlated with asymmetric arthritis.

81. **A 5-year-old boy develops acute onset of high spiking fevers, lymphadenopathy, hepatosplenomegaly, and purpura; he has been diagnosed with systemic Juvenile idiopathic arthritis (JIA) since early childhood treated with antiinflammatory drugs. You suspect macrophage activating syndrome (MAS). Of the following, the BEST test that distinguishes MAS from a flare of the primary disease is**

A. leucopenia
B. falling ESR
C. hypofibrinogenemia
D. hypertriglyceridemia
E. **evidence of hemophagocytosis in the bone marrow**

Ans E The most important indicators of MAS include a falling platelet count, extreme hyperferritinemia, increased liver enzymes, falling leukocyte count, persistent, continuous fever ≥38∘C, falling ESR, hypofibrinogenemia, and hypertriglyceridemia. The diagnosis is confirmed by bone marrow biopsy demonstrating hemophagocytosis.

82. **Rheumatoid factor (RF)–positive polyarthritis usually accounts for < 10% of all juvenile idiopathic arthritis (JIA) cases. The articular manifestation pattern is characterized by involvement of ≥5 joints in both upper and lower extremities. Of the following, the LEAST effective drug to induce remission for this subtype of JIA is**

A. **NSAIDs**
B. methotrexate
C. TNF-α antagonists
D. IL-1 inhibitors
E. IL-6 inhibitors

Ans A alone rarely induces remission in children with polyarthritis or systemic JIA. Methotrexate is the oldest and least toxic effective drug. Biologic medications that inhibit proinflammatory cytokines, such as TNF-α, IL-1, and IL-6, demonstrated excellent disease control.

83. **A 5-year-old boy has systemic juvenile idiopathic arthritis (sJIA) with systemic manifestations including fever, hepatosplenomegaly, lymphadenopathy, and pericarditis. Of the following, the BEST initial treatment for this boy is**

A. NSAIDs
B. **systemic glucocorticoids**
C. TNF-α inhibitors
D. IL-1 antagonists
E. IL-6 antagonists

Ans B TNF inhibition is not as effective for the systemic symptoms found in sJIA. When systemic symptoms dominate systemic steroids are started followed by the initiation of IL-1 or IL-6 antagonist therapy, which often induces a dramatic and rapid response.

84. **Chronic uveitis is one of the extraarticular manifestations of various subtypes of juvenile idiopathic arthritis. All the following are risk factors for the development of uveitis EXCEPT**

A. oligoarthritis subtype
B. female gender
C. ANA-positivity
D. onset of arthritis earlier than 6 yr of age
E. **severity of arthritis**

Ans E There is no association between the activity or severity of arthritis and uveitis.

85. **The child with polyarticular JIA often has a more prolonged course of active joint inflammation and requires early and aggressive therapy. Of the following, the predictor that carries the WORST prognosis is**

A. old age at onset
B. rheumatoid factor (RF) seronegativity
C. absence of rheumatoid nodules
D. small numbers of affected joints
E. **hip joint involvement**

Ans E Disease involving the hip and hand and wrist is associated with a poorer prognosis and may lead to significant functional impairment. All other mentioned factors carry a good outcome.

86. **Spondyloarthritis may be overlapped clinically with other forms of juvenile idiopathic arthritis (JIA). Clinical manifestations that help to distinguish spondyloarthritis from other forms of juvenile arthritis include all the following EXCEPT**

A. arthritis of the sacroiliac joints
B. arthritis of the hips
C. enthesitis
D. **asymptomatic uveitis**
E. gastrointestinal inflammation

Ans D Symptomatic, rather than asymptomatic, eye inflammation (acute anterior uveitis) is a clinical feature suggestive of spondyloarthritis.

87. **Enthesitis-Related Arthritis (ERA) is a subtype of rheumatic disease that falls in the juvenile idiopathic arthritis (JIA) category. Of the following, the clinical feature that is highly suggestive of ERA is**

A. symmetrical arthritis early in the disease course
B. early involvement of more than 5 joints
C. involvement of upper limb joints
D. **Inflammation of the small joints of the foot**
E. asymmetrical enthesitis

Ans D In ERA, the arthritis is typically asymmetric and involves 4 or fewer joints during the 1st 6mo of the disease. The most frequently affected joints are the knees, ankles, and hips. Enthesitis is typically symmetric and most commonly affects the lower limbs. Inflammation of the small joints of the foot, or tarsitis, is highly suggestive of ERA.

88. **Amyloidosis is a disease caused by protein misfolding; the type referred as amyloid A (AA), amyloidosis usually develops in patients with chronic inflammatory states, especially juvenile idiopathic arthritis. In which of the following subtypes of JIA there is a highest prevalence of AA amyloidosis?**

A. **systemic JIA**
B. oligoarticular JIA
C. polyarticular JIA
D. psoriatic arthritis
E. enthesitis-related arthritis

Ans A JIA is a rheumatic disease that is associated with the development of AA amyloidosis with the highest prevalence in patients with systemic JIA followed by those with polyarticular disease.

Systemic Lupus Erythematosus

89. **Non-erosive arthritis is seen in:**

A. **Systemic lupus erythematosus**
B. Ankylosing spondylitis
C. Rheumatic fever
D. Juvenile idiopathic arthritis

Ans A Non-erosive arthritis includes those without significant radiographic changes, as in systemic lupus erythematosus (SLE); b. Juvenile idiopathic arthritis Criteria for Classification of Juvenile Rheumatoid Arthritis/juvenile Idiopathic arthritis.
• • Age at onset: <16 yr •• Duration of disease: at least ≥ 6 weeks
• • Arthritis in at least 1 joint •• Exclusion of other forms of juvenile arthritis

90. **Hydroxychloroquine sulfate is an antimalarial drug important in the treatment of SLE and dermatomyositis, particularly cutaneous manifestations of disease and to reduce lupus flares. Of the following, the MOST important procedure that should be done routinely during the course of administration is**

A. gastric endoscopy
B. bone marrow examination
C. muscle biopsy
D. glucose-6-phophate dehydrogenase enzyme level assay
E. **ophthalmological examination**

Ans E The most significant potential adverse effect is retinal toxicity, which occurs rarely but results in irreversible color blindness or loss of central vision. Complete ophthalmologic examinations, including assessment of peripheral vision and color fields, are conducted at baseline and every 6-12 mo to screen for retinal toxicity.

91. **You are meeting parents of a 14-year-old girl who has been treated with a monthly intravenous cyclophosphamide for SLE-associated renal failure for the last 6 months; the mother is asking about the long-term complications of this drug. All the following are long-term complications EXCEPT**

A. **bone marrow suppression**
B. bladder cancer
C. leukemia
D. lymphoma
E. infertility

Ans A Nausea, vomiting, anorexia, alopecia, mucositis, hemorrhagic cystitis, and bone marrow suppression are potential short-term adverse effects.

92. **Systemic lupus erythematosus (SLE) is a chronic autoimmune disease characterized by multisystem inflammation and the presence of circulating autoantibodies directed against self-antigens. It occurs in both children and adults. All the following are features of childhood SLE EXCEPT**

A. it is has a more severe course
B. there is more widespread organ involvement
C. **it usually presents before 5 year of age**
D. fever, fatigue, hematologic abnormalities, and arthritis are common clinical manifestations
E. although renal disease is asymptomatic, it is oftenly present as nephrotic syndrome in adolescent age group

Ans C Childhood SLE is rare before 5 yr of age.

93. **Skin is a commonly involved organ by SLE. There are different cutaneous manifestations. Of the following, the skin manifestation that is MOST suggestive of SLE in children is**

A. malar rash
B. **discoid rash**
C. photosensitive rash
D. cutaneous vasculitis
E. livedo reticularis

Ans B

94. **Drug-induced lupus refers to the presence of SLE manifestations triggered by exposure to specific medications, including antibiotics. Of the following, the drug that is MOST likely associated with drug-induced lupus is**

A. **isoniazid**
B. rifampin
C. nitrofurantoin
D. penicillin
E. tetracycline

Ans A

95. **Systemic lupus erythematosus (SLE) is often characterized by periods of flare and disease quiescence or may follow a more smoldering disease course. All the following lab tests correlate with active disease EXCEPT**

A. **positive anti-nuclear antibody titer**
B. positive anti–double-stranded DNA level
C. low serum complement level
D. high erythrocyte sedimentation rate
E. elevated C-reactive protein (CRP) value

Ans A ANA titers are not reflective of disease activity; therefore, repeating ANA titers is not helpful in disease management.

96. **A 12-year-old female adolescent has a recent diagnosis of SLE; she has a faint malar rash and mild arthritis including both elbow and knee joints as well as the joints of small fingers. Of the following, the MOST appropriate initial therapy for this patient is**

A. systemic steroids
B. methotrexate
C. leflunomide
D. cyclophosphamide
E. **hydroxychloroquine**

Ans E is recommended for all individuals with SLE if tolerated. In addition to treating mild SLE manifestations such as rash and mild arthritis, hydroxychloroquine prevents SLE flares, improves lipid profiles, and may have a beneficial impact on mortality and renal outcomes. Corticosteroids are a mainstay for treatment of significant manifestations of SLE.

97. **A 3-week-old male baby develops malar rash involving the face and periorbital area after exposure to sun light; you suspect neonatal lupus. Of the following, the NEXT step of management is**

A. measurement of anti-Ro and anti-La antibodies of the mother
B. measurement of anti-Ro and anti-La antibodies of the baby
C. cardiac evaluation of the mother
D. **cardiac evaluation of the baby**
E. hematological evaluation of the baby

Ans D The most serious complication due to neonatal lupus is congenital heart block.

98. **You are meeting a pregnant mother who has had a previous baby with congenital heart block due to neonatal lupus; she has a positive anti-SSA and anti-SSB antibodies. All the following are therapeutic modalities to prevent occurrence of congenital fetal cardiac complications EXCEPT**

A. fluorinated corticosteroids
B. intravenous immunoglobulin
C. plasmapheresis
D. hydroxychloroquine
E. **methotrexate**

Ans E

Dermatomyositis

99. **Phocomelia is a problem affecting:**

A. **Long bones**
B. Skull bones
C. Carpals
D. Metacarpals

Ans A Phocomelia is a birth defect in which the hands and feet are attached to abbreviated arms & legs. The term comes from phoco (meaning 'seal') & melia (meaning 'limb'), to indicate that a limb is like a seal's flipper; One of the important cause is exposure of fetus to thalidomide.

100. **The distinction between postinfectious arthritis and reactive arthritis is not always clear. All the following are common features of postinfectious arthritis, rather than reactive arthritis EXCEPT**

A. there is a transient joint swelling or pain
B. it lasts less than 6 wk
C. **chronic spondyloarthritis is a common sequelae**
D. it may be encountered after group A streptococcus infection
E. rubella infection is a common causative agent

Ans C Postinfectious arthritis does not necessarily share the typical spondyloarthritis pattern of joint involvement.

101. **Juvenile dermatomyositis (JDM) is the most common inflammatory myositis in children characterized by skin rash and proximal muscle weakness. All the following are common cutaneous manifestations of JDM EXCEPT**

A. heliotrope rash of the eyelids
B. photosensitivity to ultraviolet light
C. **facial erythema sparing the nasolabial folds**
D. Gottron papules
E. periungual telangiectasias

Ans C In JDM, facial erythema is crossing the nasolabial folds, in contrast to the malar rash without nasolabial involvement typical of systemic lupus erythematosus.

102. **Treatment of juvenile dermatomyositis (JDM) includes pharmacological and non- pharmacological therapy. The mainstay of pharmacological therapy is corticosteroids that should be integrated with non-pharmacological therapy. All the following are recommended as part of non-pharmacological treatment program in JDM EXCEPT**

A. physical therapy
B. occupational therapy
C. avoidance of sun exposure
D. **bed rest**
E. social and psychological support

Ans D is not indicated, because weight bearing improves bone density and prevents contractures.

103. **Most complications from Juvenile dermatomyositis (JDM) are related to prolonged and severe weakness; secondary complications from medical treatments are also common. Of the following, the LEAST likely recognized complications of JDM is**

A. aspiration pneumonia
B. gastrointestinal bleeding
C. **cardiac arrhythmias**
D. muscle atrophy
E. skin calcifications

Ans C Cardiac involvement by JDM is rare.

104. **Current pharmacologic approaches that are considered the standard of care in Marfan syndrome (MFS) are**

A. angiotensine converting enzyme inhibitors
B. **beta blockers**
C. calcium channel blockers
D. diuretics
E. angiotensine receptor II antagonists

Ans B β-Blockers have traditionally been considered the standard of care in MFS and multiple small observational studies suggest there is a protective effect on aortic root growth, with the dose typically titrated to achieve a resting heart rate <100 beats/min during submaximal exercise.

Muscular Dystrophies

105. **A baby was vigorously shaken by parents. What do you expect in the baby?**

A. Rib fractures
B. Pelvic fracture
C. Ruptured spleen
D. **Sub-dural hematoma** 534 Section 3: Systemic Pediatrics

Ans D Sub-dural hematoma Child abuse/Shaken baby syndrome/Inflicted childhood neurotrauma
• • It occurs secondary to violent, non-accidental, repetitive, unrestrained acceleration-deceleration head & neck movements, with or without blunt head trauma in children typically < 3 year of age.
• • Inflicted childhood neurotrauma accounts for 10% of all cases of child abuse and carries a mortality rate of up to 25%.
• • Retinal hemorrhage (multiple, involving > 1 layer) is the most common ophthalmic finding.

106. **You are meeting parents of 2-year-old girl who is going to receive rubella vaccine; the mother is concerned regarding arthritis that may develop after immunization. Of the following, the statement that should be included during the discussion is**

A. it is usually frequently encountered after immunization
B. it typically affects the large joints of the upper extremity
C. it is more common in girls
D. **it is uncommon in children**
E. arthralgia usually begins immediately after administration of immunization

Ans D Rubella-associated arthropathy may follow natural rubella infection and, infrequently, rubella immunization. It typically occurs in young women, with an increased frequency with advancing age, and is uncommon in preadolescent children and in males. Arthralgia of the knees and hands usually begins within 7 days of onset of the rash or 10-28 days after immunization.

107. **Although there is no established therapy to AA amyloidosis, colchicine may be an effective drug to prevent the development of amyloidosis. Of the following, the disease that is respond to colchicine in preventing AA amyloidosis is**

A. juvenile idiopathic arthritis
B. ankylosing spondylitis
C. **familial Mediterranean fever**
D. hyper IgD syndrome
E. cryopyrin-associated periodic syndrome

Ans C Unlike AA amyloidosis associated with FMF, AA amyloidosis associated with other autoinflammatory diseases (including TRAPS, cryopyrin-associated periodic syndrome, and HIDS) and chronic rheumatic diseases (JIA, RA, and ankylosing spondylitis) do not respond to colchicine.

108. **A rocker-bottom foot describes**

A. **congenital vertical talus**
B. flexible flatfoot
C. calcaneovalgus foot
D. congenital talipes equinovarus
E. metatarsus adductus

Ans A has also been described as a rocker-bottom foot or a Persian slipper foot. The plantar surface of the foot is convex, and the talar head is prominent along the medial border of the midfoot. The fore part of the foot is dorsiflexed and abducted relative to the hindfoot, and the hindfoot is in equinus and valgus.

109. **Polydactyly or duplication of a digit can occur either as a preaxial deformity (involving the thumb) or as a postaxial deformity (involving the small finger) All the following syndromes associated with polydactyly EXCEPT**

A. Carpenter syndrome
B. Ellis-van Creveld syndrome
C. **Trisomy**
D. Orofaciodigital syndrome
E. Rubinstein-Taybi syndrome

Ans C

Spinal Deformities

110. **Nonsteroidal anti-inflammatory drugs (NSAIDs) are prescribed to decrease acute and chronic inflammation associated with various rheumatic diseases; however, many adverse effects have been encountered with long term use. Of the following, the NSAID that has the highest toxicity is**

A. celecoxib
B. meloxicam
C. **indomethacin**
D. naproxen
E. ibuprofen

Ans C Certain agents (indomethacin) have a higher risk of toxicity than others (ibuprofen); naproxen has an intermediate risk. Selective COX-2 inhibitors (such as celecoxib and meloxicam) inhibit receptors responsible for promoting inflammation with potential for fewer gastrointestinal adverse effects.

111. **Postinfectious arthritis describes arthritis that occurs after infectious illnesses, mainly viruses. Of the following, the LEAST likely causative viral infection is**

A. rubella
B. varicella-zoster
C. cytomegalovirus
D. **Epstein-Barr virus**
E. herpes-simplex virus

Ans D

112. **Kawasaki disease (KD) is an acute febrile illness of childhood characterized by vasculitis with a predilection for the coronary arteries. Predictors of poor outcome across several studies include all the following EXCEPT**

A. **old age**
B. male gender
C. persistent fever
D. poor response to IVIG
E. elevated C-reactive protein levels

Ans A Young age is poor predictor. Other lab findings associated with poor outcome are neutrophilia, thrombocytopenia, transaminitis, hyponatremia, hypoalbuminemia, elevated levels of N-terminal-probrain natriuretic protein.

113. **Polydactyly is the most common congenital toe deformity, it may be preaxial (great toe) or postaxial (5th toe), and occasionally one of the central toes is duplicated. Which of the following statements is TRUE?**

A. it is seen in approximately 2 in 100,000 births
B. it is bilateral in 10% of cases
C. **one-third of patients will also have polydactyly of the hand**
D. it may be associated with Turner syndrome
E. surgical removal of the extra digit is generally performed after 2 years of age

Ans C It is seen in approximately 2 in 1,000 births and is bilateral in 50% of cases. Conditions that may be associated with polydactyly include Ellis-Van Creveld (chondroectodermal dysplasia), longitudinal deficiency of the tibia, and Down syndrome. Treatment is indicated for cosmoses and to allow for fitting with standard shoes. This involves surgical removal of the extra digit, and the procedure is generally performed between 9 and 12 mo of age.

114. **Scoliosis is a complex 3-dimensional spinal deformity that is defined in the coronal plane as a curve of at least 10 degrees, measured by the Cobb method, on a posteroanterior (PA) radiograph of the spine. Which statement is TRUE about this disease?**

A. adolescent idiopathic scoliosis (AIC) is 2-10 times more common in males than females
B. fathers with AIS transmit the disease to 50% of their children
C. high plasma melatonin levels have been noted in patients with progressive curvatures
D. girls with AIS have been noted to have a smaller foramen magnum
E. approximately one-third of girls with AIS have osteopenia

Ans E AIS is 2-10 times more common in females than males. Fathers with AIS transmit the disease to 80% of their children, but mothers with AIS transmit the disease to only 56% of their children. Lower plasma melatonin levels have been noted in patients with progressive curvatures. Girls with AIS have been noted to have a larger foramen magnum.

115. **The normal thoracic spine has 20-50 degrees of kyphosis as measured from T3 to T12. A thoracic kyphosis in excess of the normal range of values is termed hyperkyphosis. All the following conditions are associated with hyperkyphosis EXCEPT**

A. trauma causing spinal fractures
B. osteogenesis imperfacta
C. Marfan syndrome
D. glycogen storage disease
E. mucopolysaccharidoses

Ans D Conditions associated with hyperkyphosis are: Trauma causing spinal fractures, spinal infections resulting from bacterial, tuberculosis, and fungal diseases, metabolic diseases such as osteogenesis imperfecta or osteoporosis, iatrogenic (laminectomy, spinal irradiation), neuromuscular diseases, neoplasms, congenital/developmental, disorders of collagen such as Marfan syndrome, dysplasias such as neurofibromatosis, achondroplasia, and mucopolysaccharidoses.

Fractures & Trauma

116. **A 10-year-old boy has a fracture of femur. Biochemical evaluation revealed Hb 11.5 gm/dL and ESR 18 mm 1st hour. S. calcium 12.8 mg/dL, S. phosphorus 2.3 mg/ dL alkaline phosphate 28 KA units and blood urea 32 mg/dL. Which of the following is the most probable diagnosis in his case:**

A. Nutritional rickets
B. Renal rickets
C. Hyperparathyroidism
D. Skeletal dysplasia

Ans C The given case describes a 10-year-old boy with fracture femur. Blood investigations reveal normal ESR, Hypercalcemia, hypophosphatemia and raised ALP levels (normal = 3–13 kA units) with normal B. urea; Out of the options available, only hyperparathyroidism can give rise to this picture. About other options, with
• • In renal rickets, S phosphate is usually high and S. calcium low/normal with deranged kidney functions
• • In nutritional rickets, S phosphate/calcium can be normal/low but never high calcium

117. **Reactive arthritis has the potential for evolving to chronic arthritis, especially after bacterial enteric infection or genitourinary tract infection (UTI). Of the following, the microorganism that is MOST likely causing chronic arthritis after UTI is**

A. Escherichia coli
B. Chlamydia trachomatis
C. Proteus mirabilis
D. Pseudomonas aeruginosa
E. Staphylococcus saprophyticus

Ans B

118. **Kawasaki disease (KD) has unique clinical manifestations; however, less consistent clinical presentation may occur. All the following are common classic clinical features of KD EXCEPT**

A. bilateral non exudative bulbar conjunctivitis
B. cracked lips
C. edema and erythema of the hands and feet
D. vesicular rash
E. unilateral non suppurative cervical lymphadenopathy

Ans D Typical rash is maculopapular, erythema multiforme, or scarlatiniform. Bullous, pustular, or vesicular rashes are uncommon skin manifestations.

119. **Macrodactyly is seen in the following conditions EXCEPT**

A. Proteus syndrome
B. Neurofibromatosis
C. Tuberous sclerosis
D. Klippel-Trenaunay-Weber syndrome
E. Alport syndrome

Ans E

120. **Syndactyly is one of the common anomalies observed in the upper limb. All the following syndromes are associated with syndactyly EXCEPT**

A. Carpenter syndrome
B. Ellis-van Creveld syndrome
C. Laurence-Moon-Biedl syndrome
D. Fanconi pancytopenia
E. Trisomy

Ans B associated with polydactyly.

121. **Fractures are the second most common manifestation of child abuse after skin injury (bruises, burns/abrasions). All of the following fractures suggest nonaccidental injury EXCEPT**

A. femur fractures in nonambulatory children
B. distal femoral metaphyseal corner fractures
C. posterior rib fractures
D. scapular spinous process fractures
E. distal humeral fractures

Ans E Proximal humeral fractures.

122.Fractures of the wrist and forearm are very common fractures in children. All the following statements are true EXCEPT

A. the most common mechanism of injury is a fall on the outstretched hand
B. an eighty percent of forearm fractures involve the proximal radius and ulna
C. the majority of forearm fractures are torus or greenstick fractures
D. a significant malunion of a forearm diaphyseal fracture can lead to a permanent loss of pronation and supination
E. the AP and lateral radiographs of the forearm and wrist confirm the diagnosis

Ans B Eighty percent of forearm fractures involve the distal radius and ulna, 15% involve the middle third, and the rest are rare fractures of the proximal third of the radius or ulnar shaft.

123.All the following statements are true regarding clavicular fracture EXCEPT

A. most common site for fracture is the junction of the middle and medial 3rd of clavicle
B. biceps function is important to assess as it is a prognostic indicator for future function
C. posterior medial clavicular physeal injuries are particularly problematic
D. fractures heal rapidly usually in 3-6 wk
E. a palpable mass of callus is usually visible in thin children

Ans A The most common site for fracture is the junction of the middle and lateral 3rd of clavicle.

124.Toddler fractures occur in young ambulatory children. The age range for this fracture is typically around 1-4 yr. All the following statement are true EXCEPT

A. children in this age group are usually unable to describe the area of injury well
B. radiographs may show no fracture
C. classic symptom is refusal to bear weight
D. Inflammatory markers may be ordered to rule out infectious processes
E. fracture is treated with bed rest and analgesia for approximately 2 wk

Ans E The fracture is treated with an above-knee cast for approximately 3 wk.

125.All the following are indications for external fixation in pediatric fractures EXCEPT

A. grades II and III open fractures
B. fractures associated with severe burns
C. fractures with soft-tissue loss
D. pelvic fractures
E. fractures with associated head injuries and spasticity

Ans D Not all pelvic fractures only the unstable one.

24

CHAPTER 24

Pediatric Surgical Disorders



WHAT THIS CHAPTER COVERS

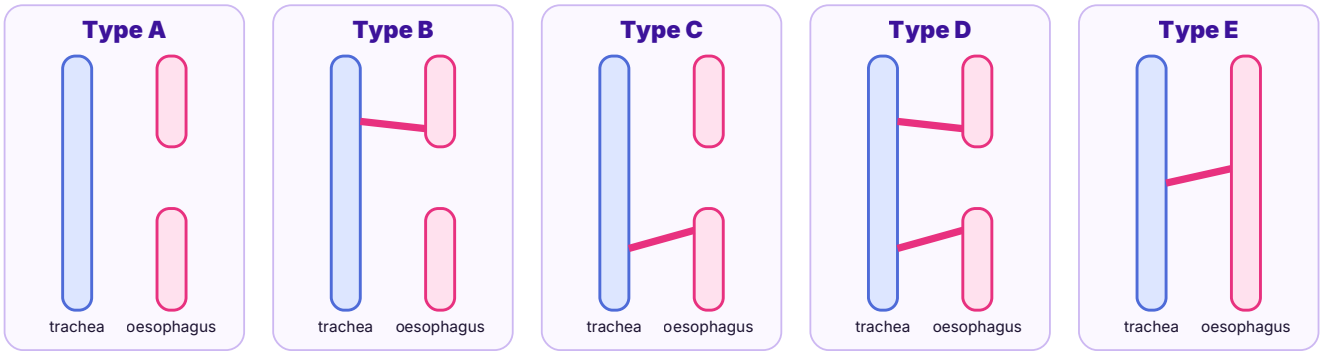
- Cleft Lip & Palate
- Esophageal Atresia & TEF
- Congenital Diaphragmatic Hernia
- Omphalocele & Gastroschisis
- Imperforate Anus
- Hirschsprung Disease
- Intussusception
- Pyloric Stenosis
- Appendicitis
- Meckel Diverticulum
- Hypospadias & Epispadias
- Undescended Testis
- Hydrocele & Hernia
- Torsion of Testis
- Wilms Tumor
- Neuroblastoma
- Trauma & Burns

47 MCQs **17** subtopics

24.1 Cleft Lip & Palate

- **MC congenital craniofacial anomaly**; **cleft lip ± palate (CL/P) – boys, left side (unilateral left MC)**; **isolated cleft palate (CP) – girls**, more syndromic (**Pierre Robin, Stickler, 22q11, Treacher Collins, Van der Woude – lip pits**).
- Embryology: **CL – failure of fusion of maxillary prominence with medial nasal prominence (4–7 wk)**; **CP – failure of fusion of palatal shelves (8–12 wk)**.
- Risk: multifactorial; **maternal smoking, phenytoin, valproate, retinoids, steroids, folate deficiency**, diabetes. Recurrence 4% with one affected child.
- **Problems**: **feeding difficulty (MC early problem – can't create negative pressure; use special bottles/ squeeze feeders, upright position; breastfeeding possible with CL alone)**, aspiration, **recurrent otitis media/ OME (tensor veli palatini dysfunction – conductive deafness; grommets)**, **speech (hypernasality, velopharyngeal insufficiency)**, dental anomalies, cosmetic/psychosocial.
- **Timing of surgery – "Rule of 10s" for lip repair: 10 weeks of age, 10 lb (4.5 kg) weight, Hb 10 g/dL** (+ TLC < 10,000):
  - **Cheiloplasty (lip): 3–6 months** (Millard rotation-advancement – MC technique; Tennison–Randall).
  - **Palatoplasty (palate): 9–18 months (before speech development; ideally ≤ 12 m)** (von Langenbeck, Bardach, Furlow double Z-plasty, Veau–Wardill–Kilner).
  - Alveolar bone grafting: **8–11 y (mixed dentition, before canine eruption)**; rhinoplasty – adolescence; **presurgical nasoalveolar moulding (NAM)** in infancy.
- Multidisciplinary cleft team; **Smile Train** (India).

24.2 Oesophageal Atresia (EA) & Tracheo-oesophageal Fistula (TEF)



Gross classification of oesophageal atresia / TEF

Gross type	Anatomy	Frequency
A	**Isolated EA, no fistula** (pure atresia) – gasless abdomen	~8%
B	EA with **proximal** TEF	~1%
C	**EA with DISTAL TEF (MC)**	**~85%**
D	EA with proximal & distal TEF	~1%
E (H-type)	**TEF without atresia**	~4%

- **Antenatal**: **polyhydramnios**, absent/small stomach bubble.
- **Newborn**: **excessive frothy salivation/drooling ("mucus baby")**, choking, coughing & cyanosis with first feed, respiratory distress (aspiration – **right upper lobe pneumonia**, reflux of gastric acid through distal fistula), abdominal distension (type C – air through fistula).

- **Diagnosis: inability to pass a stiff 10 Fr radio-opaque orogastric/nasogastric (red rubber) catheter beyond 10–12 cm – coils in upper pouch on X-ray (chest + abdomen)**; **air in stomach → distal fistula (type C)**; **gasless abdomen → type A (or B)**; small volume (0.5–1 mL) water-soluble contrast if needed (avoid barium); **H-type – prone pull-back tube oesophagogram / bronchoscopy** (recurrent pneumonia, cough with feeds – presents later).
- **Associations (50%)**: **VACTERL (MC)**, **CHD (MC – VSD, TOF; also right-sided aortic arch – check by echo before surgery)**, **CHARGE**, Down, Edwards, duodenal atresia, anorectal malformation.
- **Pre-op management**: **nurse head-up (30–45°) or prone**, **continuous upper-pouch suction (Replogle tube)**, NPO, IV fluids, antibiotics, avoid bag-mask ventilation (gastric distension through fistula), **echo (aortic arch side)**.
- **Surgery**: **right posterolateral extrapleural thoracotomy (left if right-sided arch), division of fistula & primary end-to-end oesophageal anastomosis** (thoracoscopic); **long-gap EA (type A) – gastrostomy + delayed anastomosis (Foker) or oesophageal replacement (gastric pull-up, colon interposition)**.
- **Spitz classification (prognosis)**: Group I – BW > 1500 g, no major CHD (~98% survival); II – < 1500 g or major CHD; III – both.
- **Complications**: **anastomotic leak (early), anastomotic stricture (MC – dilatation)**, **GERD (MC long-term)**, **tracheomalacia ("TEF cough" – barking, dying spells)**, recurrent fistula, dysmotility.

24.3 Congenital Diaphragmatic Hernia (CDH)

- **Bochdalek hernia – posterolateral defect, LEFT side (85%) – MC**; **Morgagni – anteromedial (retrosternal), right side, older child, often asymptomatic**; eventration – thin muscle.
- **Pathophysiology: pulmonary hypoplasia (both lungs, ipsilateral worse) + pulmonary hypertension (PPHN)** – the main determinants of mortality.
- Antenatal USG – **stomach/bowel in thorax, mediastinal shift**, polyhydramnios; prognosis – **lung-to-head ratio (LHR – observed/expected < 25% poor)**, liver herniation (poor); **FETO (fetoscopic endoluminal tracheal occlusion)** in severe.
- **Newborn**: **respiratory distress & cyanosis at birth, scaphoid (empty) abdomen, barrel chest, apex beat/heart sounds shifted to RIGHT, absent breath sounds & bowel sounds in left chest**.
- X-ray: **bowel loops in left hemithorax, mediastinal shift to right, NG tube tip in chest**.
- **Management**: **immediate intubation – avoid bag & mask ventilation (distends bowel in chest)**; **NG/OG decompression**; gentle ventilation (permissive hypercapnia, pre-ductal SpO_2 > 85%), **surfactant not routinely**, iNO/sildenafil for PPHN, **ECMO**; **surgery – delayed (after stabilisation of PPHN – 24–72 h); NOT an emergency**; reduction & primary repair/patch via abdominal approach.
- Associated: CHD, chromosomal (trisomy 18, 13), Fryns syndrome, Pallister–Killian (12p tetrasomy).

24.4 Abdominal Wall Defects

Feature	Omphalocele (exomphalos)	Gastroschisis
Defect	**Herniation through umbilical ring – failure of return of midgut (after 10–12 wk)**	**Full-thickness defect to the RIGHT of an intact umbilicus** (vascular accident – right omphalomesenteric artery/umbilical vein)
Sac	**Covered by a sac (amnion + peritoneum)**; umbilical cord inserted on sac	**No sac** – bowel exposed, **thickened, matted, oedematous (chemical peritonitis from amniotic fluid)**
Contents	Bowel ± **liver** (giant)	Bowel (no liver)
Maternal age	Older	**Young mothers (< 20 y)**, smoking, drugs
Associated anomalies	**Common (50–70%) – chromosomal (trisomy 13, 18, 21), CHD, Beckwith–Wiedemann, pentalogy of Cantrell, OEIS (cloacal exstrophy)**	**Rare (except intestinal atresia – 10%)**
Prognosis	Depends on associated anomalies	**Good (> 90% survival)**; intestinal dysmotility, short bowel

Feature	Omphalocele (exomphalos)	Gastroschisis
Rx	Small – primary closure; giant – **staged/"paint and wait" (silver sulfadiazine, epithelialisation) → delayed repair**	**Cover bowel with cling film/sterile bowel bag**, keep warm (heat & fluid loss), NG decompression, IV fluids, antibiotics; **primary closure or preformed silo with gradual reduction**
Raised AFP	Moderate	**Higher**

- **Umbilical hernia** – see Neonate chapter (most close by 4–5 y).
- **Prune belly syndrome** – see Renal.

24.5 Neonatal Intestinal Obstruction

Condition	Key features
Duodenal atresia	**Down syndrome (30%)**; **polyhydramnios**; **bilious vomiting from day 1 (if below ampulla – 85%)**, **no abdominal distension**; **X-ray: "double-bubble" sign (stomach + proximal duodenum) with no distal gas**; annular pancreas, malrotation; Rx **duodenoduodenostomy (Kimura diamond-shaped)**
Jejunoileal atresia	**Vascular accident in utero**; bilious vomiting, distension; **"triple bubble"/multiple dilated loops with air-fluid levels**; **apple-peel (type IIIb)** – SMA occlusion; CF (ileal)
Malrotation + volvulus	Bilious vomiting – emergency (see GI)
Meconium ileus	**CF (~ 90–100%)**; terminal ileum obstruction by inspissated meconium; **"soap-bubble"/ground-glass appearance (Neuhauser sign) in RLQ, few air-fluid levels**; **microcolon on contrast enema**; Rx **Gastrografin (hyperosmolar) enema** (therapeutic), surgery if complicated (meconium peritonitis – calcification)
Meconium plug syndrome / small left colon	IDM, maternal $MgSO_4$; contrast enema diagnostic & therapeutic; rule out Hirschsprung & CF
Hirschsprung	Delayed meconium > 48 h – see GI
Anorectal malformation	See GI
Hypertrophic pyloric stenosis	**Non-bilious** vomiting, 3–6 weeks

24.6 Intussusception, Pyloric Stenosis, Appendicitis & Meckel's

- See Gastroenterology chapter (ultrasound IOC; air/hydrostatic enema; Ramstedt pyloromyotomy after correcting alkalosis; rule of 2s; pertechnetate scan).

24.7 Hernia & Hydrocele

- **Inguinal hernia in children – indirect (99%)** due to **patent processus vaginalis**; **boys, right side (60%), preterm (up to 30%)**, CF, connective tissue disorders, VP shunt, peritoneal dialysis.
- **Risk of incarceration highest in first year (esp. < 6 months)** → **herniotomy (high ligation of sac) soon after diagnosis** (no herniorrhaphy needed); incarcerated – manual reduction (taxis) under sedation → surgery after 24–48 h (oedema subsides); strangulation/irreducible → emergency.
- **Hydrocele**: **communicating (patent processus – size varies, bluish, transilluminates, ↑ on crying/standing)** vs non-communicating (resolves by 1–2 y); **surgery if persisting > 1–2 years** (communicating → herniotomy approach via groin, not scrotal); **can get above the swelling** (vs hernia).
- **Encysted hydrocele of cord / hydrocele of canal of Nuck (girls)**.

- In a girl with bilateral inguinal hernia → think **complete androgen insensitivity (testes in sac)**.

24.8 Undescended Testis (Cryptorchidism)

- **MC congenital anomaly of male genitalia**; **3–5% term, ~30% preterm**; **most descend spontaneously by 3 months (corrected); after 6 months unlikely → 1% at 1 year**.
- **Retractile testis** (normal – brisk cremasteric reflex; can be brought to scrotum & stays) vs **undescended (cannot be brought down / springs back)** vs **ectopic (MC site – superficial inguinal pouch; perineum, femoral, prepenile)** vs **ascending testis** (descended earlier, later ascends).
- **Palpable (80%) vs non-palpable (20% – intra-abdominal, absent/vanishing testis)**; **bilateral non-palpable → rule out CAH/DSD (karyotype)**.
- **Complications**: **infertility (↓ spermatogenesis from 1–2 y)**, **malignancy (seminoma – 3–10× risk; risk not eliminated by orchidopexy but ↓ if done before puberty; allows self-examination)**, **torsion**, trauma, associated inguinal hernia (90%), psychological.
- **Investigations**: **clinical examination is key; imaging (USG/MRI) NOT recommended for localisation**; **diagnostic laparoscopy – IOC for non-palpable testis**; hCG stimulation test (bilateral non-palpable – anorchia vs intra-abdominal), AMH, inhibin B.
- **Treatment: orchidopexy at 6–12 months (by 18 months latest)** – inguinal (palpable) / laparoscopic (non-palpable; **Fowler–Stephens** staged for high intra-abdominal); orchidectomy for atrophic/post-pubertal intra-abdominal testis; **hormonal therapy (hCG/GnRH) not recommended routinely** (low success).

24.9 Hypospadias, Epispadias & Phimosis

- **Hypospadias**: **urethral meatus on ventral aspect** of penis (1 in 250 boys) – **distal/anterior (glanular, coronal, subcoronal – MC 70%)**, middle (penile), proximal (penoscrotal, scrotal, perineal); **hooded dorsal prepuce (deficient ventral foreskin)**, **chordee (ventral curvature)**; associated – **undescended testis, inguinal hernia**; **proximal hypospadias + undescended testis → evaluate for DSD (karyotype)**.
  - **DO NOT CIRCUMCISE (prepuce used for repair)**.
  - **Surgery at 6–18 months** – **TIP urethroplasty (Snodgrass – tubularised incised plate) – MC for distal**, MAGPI, Mathieu; proximal – staged (Bracka). Complications – **urethrocutaneous fistula (MC)**, meatal stenosis, stricture, diverticulum.
- **Epispadias**: **dorsal meatus**, almost always with **bladder exstrophy (exstrophy–epispadias complex)**; incontinence.
- **Physiological phimosis** – non-retractile foreskin is **normal till 3–5 y** (most retract by adolescence); **ballooning is normal**; **pathological phimosis – scarring (balanitis xerotica obliterans/lichen sclerosus)**, recurrent balanoposthitis, UTI; Rx **topical steroid (0.05% betamethasone) 4–8 weeks – first line**, circumcision.
- **Paraphimosis** – retracted foreskin trapped behind glans → emergency manual reduction (ice, compression, dorsal slit).
- **Indications for circumcision**: pathological phimosis/BXO, recurrent balanoposthitis, recurrent UTI with VUR (infants), religious; **contraindicated in hypospadias, chordee, buried penis, bleeding disorders**.

24.10 Acute Scrotum

Feature	Testicular torsion	Torsion of appendix testis (hydatid of Morgagni)	Epididymo-orchitis
Age	**Neonatal (extravaginal) & adolescents 12–18 y (intravaginal – bell-clapper deformity)**	**7–12 y (MC cause of acute scrotum in prepubertal boys)**	Adolescents (STI), infants (UTI/anomalies)

Feature	Testicular torsion	Torsion of appendix testis (hydatid of Morgagni)	Epididymo-orchitis
Onset	**Sudden severe pain**, nausea/vomiting	Gradual, localised to upper pole	Gradual, fever, dysuria
Examination	**High-riding, horizontal lie testis, absent cremasteric reflex**, diffusely tender	**"Blue-dot sign"**, tender nodule at upper pole; cremasteric reflex present	Tender epididymis, **Prehn's sign positive (relief on elevation)**, pyuria
Doppler USG	**↓/absent blood flow**, "whirlpool sign"	Normal/↑ flow	**↑ flow**
Rx	**EMERGENCY scrotal exploration – detorsion + bilateral orchidopexy within 6 h** (salvage ~100% < 6 h, ~20% > 12 h); orchidectomy if non-viable; **don't wait for imaging if strongly suspected**	Analgesia, rest (self-limiting)	Antibiotics

- **Neonatal torsion** – firm painless scrotal mass, discoloured; usually non-salvageable.
- Others: incarcerated hernia, **idiopathic scrotal oedema (HSP – scrotal involvement)**, trauma, tumour, **mumps orchitis (post-pubertal)**.

24.11 Other Paediatric Surgical Conditions

Condition	Key points
Sacrococcygeal teratoma	**MC neonatal tumour, girls**; Altman I–IV; **excise early with coccyx** (↑ malignancy with age), follow AFP
Cystic hygroma (lymphatic malformation)	**Posterior triangle of neck (MC)**, **transilluminant**, soft, compressible; **Turner, Noonan, Down**; airway compression; Rx **sclerotherapy (OK-432/bleomycin)**, sirolimus, excision
Thyroglossal cyst	**MC congenital neck mass**; **midline, moves with swallowing & tongue protrusion**; infection; **Sistrunk operation (excision with central part of hyoid)** – check normal thyroid (USG) first (may be only thyroid tissue)
Branchial cleft cyst/sinus	**2nd arch (MC)** – anterior border of SCM (lower 1/3 sinus opening); 1st – near ear/angle of jaw
Dermoid cyst	**External angular dermoid (MC)**, midline (nasal – rule out intracranial extension)
Ranula	Sublingual salivary gland mucocele (floor of mouth, bluish) – plunging ranula
Tongue-tie (ankyloglossia)	Frenotomy if feeding difficulty
Pectus excavatum	MC chest wall deformity; Marfan; **Nuss procedure**
Biliary atresia / choledochal cyst	See Hepatology
Intestinal duplication	Ileum (MC); mass/obstruction/bleeding (ectopic gastric mucosa)
Hirschsprung, ARM, Meckel's, intussusception	See GI
Wilms, neuroblastoma	See Tumours
Posterior urethral valves, PUJ obstruction	See Renal
Burns	See PALS (Parkland, Lund–Browder)
Foreign bodies	See GI/Respiratory
Bladder exstrophy	Exposed bladder plate, pubic diastasis, epispadias; staged reconstruction

Practice MCQs · Pediatric Surgical Disorders

47 questions · correct option highlighted in green · cover the answer strip to self-test

Cleft Lip & Palate

1. Failure to pass meconium within 48 hours of birth in a newborn with no obvious external abnormality should lead to the suspicion of:

A. Anal atresia
B. Congenital pouch colon
C. **Congenital aganglionosis**
D. Meconium ileus

Ans C Read what Nelson says about Hirschsprung's disease (Congenital aganglinosis):
• • "Ninety nine percent of full term infants normally pass meconium within 48 hrs of birth. Hirschsprung disease should be suspected in any full term infant with delayed passage of stool".

2. Diaphragmatic hernias can be diagnosed by:

A. Ultrasound of the chest
B. Barium meal follow through
C. None of the above
D. **Both the above**

Ans D Chest X-ray, Ultrasound of chest or barium meal followthrough can be used to diagnose Diaphragmatic hernia.

Esophageal Atresia & TEF

3. H type TEF presents with:

A. Dysphagia
B. **Aspiration**
C. Hematemesis
D. Hemoptysis
E. Recurrent pneumonia

Ans B e. Recurrent pneumonia The infant with an isolated TEF, in the absence of EA ("H-type" fistula) might come to medical attention later in life with chronic respiratory problems, including refractory bronchospasm and recurrent pneumonias.

4. Congenital eventration of diaphragm requires demonstration of:

A. **An intact diaphragm**
B. Diaphragmatic tear
C. A diaphragmatic defect
D. Normal diaphragmatic excursions

Ans A Eventration of the diaphragm is an abnormal elevation of the dome of diaphragm. There is no defect (hole) in diaphragm.

Congenital Diaphragmatic Hernia

5. Recurrent obstruction, mass per rectum and diarrhea are seen in a child diagnosis:

A. **Intussuception**
B. Rectal prolapse
C. Internal hernia
D. Hemorrhoids

Ans A Intussusception Discussing about the options one by one, Option a. Recurrent intussusception is noted in 5-8% Advancing intestine can prolapse through the anus in intussusception and is distinguished from rectal prolapse by separation between the protruding intestine and the rectal wall, which does not exist in rectal proplase Option b. Rectal prolapse usually occurs during defecation and is usually painless or produces mild discomfort Option c. There is no diarrhea in rectal prolapse, hernia and hemorrhoids

6. Diaphragmatic hernias can occur through the:

A. **Foramen of Morgagni**
B. Foramen of Magendie
C. Foramen of Winslow
D. Esophageal hiatus

Ans A In diaphragmatic hernia, the defect may be in the posterolateral (Bochdalek) portion of the diaphragm (most common), retrosternal (Morgagni) is the 2nd most common, at the esophageal hiatus (hiatal) or paraesophageal (adjacent to the hiatus).

7. Which of the following is associated with good prognosis in congenital diaphragmatic hernia?

A. Lung head ratio <
B. Detection of CDH in first trimester scan
C. **Symptoms develop after 48 hours**
D. Liver herniated into thorax

Ans C Predictors of a poor prognosis in CDH are:
• • Lung: Head ratio < 1 •• Severe pulmonary hypoplasia
• • An associated major anomaly •• Liver herniation into thorax
• • Symptoms before 24 hr of age •• Detection of CDH in 1st trimester scan
• • Herniation to the contralateral lung •• Need for ECMO

8. Congenital diaphragmatic hernia through foramen of Bochdalek is situated:

A. Anteriorly
B. **Posterolaterally**
C. Diaphragmatic hiatus
D. Antero laterally

Ans B

9. A neonate having congenital diaphragmatic hernia developed respiratory distress. Breath sounds were decreased on the left side. After bag and mask ventilation, ET tube was put and the maximal cardiac impulse shifted to the right side. What should be the next step in management:

A. Confirm the position of endotracheal tube by X-ray chest
B. **Remove tube and reattempt intubation**
C. Naso gastric tube insertion
D. Chest X-ray

Ans B Best answer would have been, confirm the position of endotracheal tube (but, not by X-ray!). Amongst the given options, best course of action is to remove the tube and reattempt intubation.

10. A newborn baby has been referred to the casualty as a case of congenital diaphragmatic hernia. The first clinical intervention is to:

A. Insert a central venous pressure line
B. Bag and mask ventilation
C. **Insert a nasogastric tube**
D. Ventilate with high frequency ventilator

Ans C If a child with congenital diaphragmatic hernia is otherwise stable & does not have severe respiratory distress, a nasogastric tube is inserted to decompress the stomach & bowel loops.

Omphalocele & Gastroschisis

11. A 6-month-old child woke up in night, crying with abdominal pain, which got relieved on passing red stools. What is the diagnosis?

A. Meckel's diverticulum
B. **Intususception**
C. Malrotation
D. Intestinal obstruction

Ans B Colicky abdominal pain relieved by passage of red 'currant jelly' stool in an infant is the classic presentation of intussusception.

12. Most important prognostic factor in congenital diaphragmatic hernia:

A. **Pulmonary hypertension**
B. Size of hernia
C. Timing of surgery
D. Gestational age

Ans A The two main factors that affect morbidity and mortality in CDH are pulmonary hypoplasia and pulmonary hypertension.

Imperforate Anus

13. X-ray detect congenital anorectal malformation at:

A. Immediately after birth
B. **24–48 hours**
C. 48–72 hours
D. After 72 hours

Ans B The invertogram or upside-down X-ray is of little value to detect congenital anorectal malformation, but a prone cross-table lateral plain X-ray at 24 hours of life (to allow time for bowel distention from swallowed air) with a radiopaque marker on the perineum is helpful in diagnosis.

14. A child presented with respiratory distress was brought to emergency with bag and mask ventilation. Now child is intubated. Chest X-ray shows right-sided deviation of mediastinum with scaphoid abdomen. His Pulse rate is increased. What is the next step?

A. Confirm the position of endotracheal tube
B. **Emergency surgery**
C. Naso gastric tube insertion
D. Chest X-ray to confirm position of the endotracheal tube

Ans B Put a nasogastric tube Mediastinal deviation, scaphoid abdomen with respiratory distress suggest a diagnosis of CDH. The resuscitation of CDH patient consists of: Stabilization by mechanical ventilation with 100% O & Nasogastric suction, In this case, the child is already intubated. Right sided deviation of mediastinum seen in this case could be due to herniated bowel loops, which would have been further distended by bag & mask ventilation. So, the best next step is to put a NG tube to decompress the bowel loops.

Hirschsprung Disease

15. FALSE statement regarding Hirschsprung disease is:

A. Aganglionosis always involves distal rectum
B. Non passage of meconium in 1st 24 hours is a cardinal feature
C. Diagnosis is established by Suction rectal biopsy
D. **No passage of stools after per rectal examination**

Ans D Clinical features of Hirschsprung disease:
• • It usually presents in the neonatal period with a distended abdomen, failure to pass meconium, delayed passage of stool and/or bilious emesis or aspirates with feeding intolerance. May present later with constipation
• • Rectal examination: Rectum is usually empty, but when finger is removed, there may be explosive discharge of foul smelling feces and gas.

16. In Hirschsprung disease, marker used for early diagnosis is:

A. **Acetyl cholinesterase**
B. Adrenaline
C. VIP
D. None of above

Ans A

17. Hirschsprung disease is confirmed by:

A. **Rectal biopsy**
B. Per Rectal examination
C. Rectal manometry
D. X-ray abdomen

Ans A

18. True about Hirschsprung's disease:

A. Aganglionic segment is contracted not dilated
B. Descending colon is most common site
C. Barium enema is diagnostic
D. Barium enema shows calcification
E. Rectal biopsy is the confirmatory investigation

Ans A e. Rectal biopsy is the confirmatory investigation Diagnosis of Hirschsprung's disease Name of investigation Features seen Rectal suction biopsy •• It shows Hypertrophied nerve bundles and absence of ganglion cells (gold standard) •• Acetylcholinesterase or Calretinin stains used Anorectal manometry •• Measures the pressure of internal anal sphincter while a balloon is distended in the rectum
• • In Hirschsprung's disease, there is paradoxical rise in pressure with rectal distention

19. True statement regarding Hirschsprung's disease:

A. Giant ganglia are present (AIIMS June 99)
B. Mucosa is involved and show foldings
C. Manometry excludes the disease
D. Rectal biopsy is contraindicated in infants

Ans C

Intussusception

20. Intussusception is caused by:

A. Submucous lipoma
B. Subfascial lipoma
C. Subserous lipoma
D. Intramural lipoma

Ans A Intussusception Predisposing Peyer's patches or prominent mounds of lymph tissue lead to mucosal prolapse of ileum into colon; conditions Other lead points: Meckel diverticulum, intestinal polyp, neurofibroma, intestinal duplication cysts, hamartomas, ectopic pancreatic tissue, hemangioma, lipoma & Clinical features Sudden onset of severe paroxysmal colicky pain in a previously well child, that recurs frequently 60% infants pass a stool containing red blood and mucus, the currant jelly stool

21. A 10-month-old infant presents with acute intestinal obstruction. Contrast enema X-ray shows the intussusception, likely cause is:

A. Peyer's patch hypertrophy
B. Meckel's diverticulum
C. Mucosal polyp
D. Duplication cyst

Ans A Peyer's patch lymphoid hyperplasia (often post-viral) is the most common lead point for idiopathic intussusception in infants, more so than Meckel's diverticulum, which is a more typical lead point in older children.

Pyloric Stenosis

22. A newborn has dribbling after feeds. He has respiratory distress and froths at the mouth. Diagnosis is:

A. Tracheoesophageal fistula
B. Tetralogy of Fallot
C. Respiratory distress syndrome
D. None of the above DISORDERS OF STOMACH

Ans A

23. The abdominal mass in congenital hypertrophic pyloric Stenosis is a new born child can be best palpated:

A. In the midepigastric area
B. In the right iliac quadrant
C. In the umbilical area
D. During feeding

Ans A •• Non-bilious vomiting is the initial symptom of pyloric stenosis, occurring immediately after a feeding.
• • Vomiting usually starts after 3 wk of age, but symptoms can develop as early as the 1st wk of life
• • As vomiting continues, a progressive loss of fluid, hydrogen ion, and chloride leads to hypochloremic metabolic alkalosis.
• • Diagnosis established by palpating the pyloric mass.

24. Investigation of choice for congenital hypertrophic pyloric stenosis is?

A. Barium meal
B. Ultrasound
C. CT scan
D. MRI

Ans B is the investigation of choice for congenital hypertrophic pyloric stenosis, demonstrating a thickened, elongated pylorus.

25. Melena Which of the above is/are the feature/features of infantile hypertrophic pyloric stenosis?

A. 1, 2 and
B. 1 and 3 only
C. 2 and
D. 4 only

Ans A Proximal end blind, distal end communicating with trachea

26. In Hypertrophic pyloric stenosis all the following electrolytes need to be replenished except:

A. Na+
B. K+
C. Cl–
D. HCO–

Ans D HCO – 3 Electrolyte abnormalities in congenital hypertrophic pyloric stenosis:
• • These infants can develop hypochloremic, hypokalemic, metabolic alkalosis & •• Hydrogen ion and Na+ re-absorption occurs with an accompanied potassium ion excretion
• • The resulting hypokalemia leads to the excretion of hydrogen ions in the face of alkalosis, a paradoxic aciduria
• • Treatment includes replacement of volume deficit with isotonic saline and potassium once adequate urine output is ensured

27. In a dehydrated child with hypertropic pyloric stenosis, the fluid of choice:

A. Normal saline
B. 5% dextrose
C. Ringer lactate
D. Darrows solution

Ans A

28. The metabolic derangement in congenital pyloric stenosis is:

A. Hypochloremic alkalosis
B. Hyperchloremic alkalosis
C. Hyperchloremic acidosis
D. Hypochloremic acidosis

Ans A Pyloric stenosis classically causes a hypochloremic, hypokalemic metabolic alkalosis from persistent vomiting of gastric (HCl-rich) contents.

29. Congenital hypertrophic pyloric stenosis usually presents:

A. Within 2 days after birth
B. Around 1 week after birth
C. Around 2 weeks after birth
D. Around 2 months after birth

Ans C Usual age of presentation of congenital pyloric stenosis is 2-6 weeks;

Appendicitis

30. A 3-month infant presents with palpable abdominal mass and non bilious vomiting. Most likely diagnosis is?

A. Hypertrophic biliary stenosis
B. Hypertrophic pyloric stenosis
C. Tracheoesophageal fistula
D. Duodenal atresia

Ans B fo 16. a. Hypochloremic alkalosis Congenital Hypertrophic Pyloric Stenosis
• • It is the commonest surgical disorder of the stomach during infancy
• • More common in boys and in infants of blood group 'B' and 'O'; Usually does not present at birth, but after 3 weeks age
• • Non-billious vomiting is the initial symptoms of pyloric stenosis; leads to Hypochloremic metabolic alkalosis

31. A previously healthy infant presents with recurrent episode of abdominal pain. The mother says that the child has been passing stool after episodes of pain, but gives no history of vomiting or bleeding per rectum. Which of the following is the most likely diagnosis?

A. Rectal polyps
B. Intussusception
C. Meckel's diverticulum
D. Necrotizing enterocolitis

Ans B In the given question, out of the given options, Intussusception is most appropriate one because
• • Usual presentation of rectal polyps and Meckel's diverticulum is painless rectal bleeding fo •• Necrotizing Enterocolitis is usually seen in premature infants with abdominal distension, bloody stools, lethargy and shock. weiveR 39. a. Peyer's patch hypertrophy

32. Best diagnostic investigation for acute appendicitis in children:

A. Tracheo-esophageal fistula
B. Tetralogy of Fallot
C. Congenital hypertrophic pyloric stenosis
D. ARDS
E. IV fluid & 43. A newborn with recurrent vomiting, cyanosis after each feed is likely to be suffering from: (APPG 05)

Ans A USG Gore & Levine Textbook of Gastrointestinal Radiology, 3/e, p 2282
• • Investigation of choice for acute appendicitis in a child is USG, as a child has thin abdominal wall
• • So, even though USG does not have much penetration power, appendix can be visualized
• • Best investigation would be CT abdomen, but that would expose the child to radiation, hence it is not the investigation of choice.

Meckel Diverticulum

33. Projectile vomiting is a feature of:

A. Esophageal atresia
B. Hypertrophic pyloric stenosis
C. Cholera
D. Intussusception DISORDERS OF INTESTINE

Ans B Discussing the options one by one a. Esophageal atresia Frothing & bubbling at mouth & nose & coughing, cyanosis, respiratory distress seen b. Hypertrophic pyloric Progressive non-bilious vomiting is the initial symptom of pyloric stenosis stenosis c. Cholera Profuse watery diarrhea, with profuse rice water stools with a fishy smell is seen d. Intussusception Sudden onset of severe paroxysmal colicky pain that is accompanied by straining efforts with legs and knees flexed.

34. Meckel's diverticulum is a derivative of?

A. Allantoic diverticulum
B. Vitellointestinal duct
C. Ventral mesogastrium
D. Ductus arteriosus

Ans B

35. Newborn with APGAR score of 2 at 1 min. and 6 at 5 min, has respiratory distress and mediastinal shift. What can be the diagnosis?

A. Congenital adenomatoid lung disease
B. Pneumothorax
C. Diaphragmatic hernia
D. Transient tachypnea of newborn
E. HMD Chapter 24: Pediatric Surgical Disorders 547 Answers

Ans A b. Pneumothorax & c. Diaphragmatic hernia Note

Hypospadias & Epispadias

36. Congenital mega colon is confirmed by:

A. Vitelloumbilical duct
B. Vitellointestinal duct
C. Vitelloportal duct
D. Vitellodiaphragmatic duct

Ans A Rectal biopsy •• Congenital Aganglionic Megacolon is the other name for Hirschsprung disease
• • Diagnosis of Hirschsprung disease is confirmed by rectal biopsy.

37. Infants with blood in stools and mass in abdomen, diagnosis is:

A. Intussusception
B. Volvulus
C. Idiopathic abdominal epilepsy
D. Hirschsprung's disease

Ans A About rest of the options, In volvulus: Features of obstruction predominate, i.e. Vomiting, abdominal distension but there is no abdominal mass palpable. In Hirschsprung's disease: history of constipation and abdominal distension may be present. In abdominal epilepsy: There is abdominal pain with/ without a seizure prodrome.

Undescended Testis

38. A child complains of fluid coming out of umbilicus on straining. What is the diagnosis?

A. Patent vitellointestinal duct
B. Urachal fistula
C. Umbilical hernia
D. Gastroschisis

Ans B •• A persistent urachus is due to failure of closure of allantoic duct and is associated with bladder outlet obstruction
• • Patency should be suspected if a clear, light yellow, urine-like fluid is being discharged from the umbilicus.

Hydrocele & Hernia

39. The absence of ganglion cells within the affected segment of bowel is a feature of:

A. Tropical sprue
B. Hirschsprung's disease
C. Celiac disease
D. Crohn's disease

Ans B

Torsion of Testis

40. Most common cause of Acute Intestinal Obstruction in neonates is:

A. Jejunal atresia
B. Malrotation
C. Duodenal atresia
D. Acute Intussusception

Ans C •• Most common cause of neonatal intestinal obstruction — Meconium plug syndrome > Duodenal atresia
• • Other important causes of intestinal obstruction in newborn are: Jejunoileal atresia, Malrotation of gut, Meconium ileus with
• • Intussusception is the most common cause of intestinal obstruction between 3 months and 6 years of age (not in neonate) Meconium plug syndrome—when meconium plug causes obstruction of colon.

41. A 6-month-old baby presents with history of bloody diarrhea of 2 days duration with abdominal distension and on examination the baby screams, diagnosis is:

A. Intussusception
B. HUS
C. Appendicitis
D. Ac. enterocolitis

Ans A Discussing the options one by one: a. Intussusception The typical age group with short history of bloody diarrhea and extreme tenderness on examination point towards intussusception b. HUS In HUS, the typical clinical picture is of sudden onset pallor, oliguria, edema, petechiae and lethargy; Usually there is history of severe gastroenteritis with blood stained stools but no severe pain abdomen; c. Appendicitis Typically presents with right lower quadrant pain with/ without gastroenteritis

Wilms Tumor

42. Commonest cause of intestinal obstruction in children is:

A. Intussusception
B. Volvulus
C. Hernia
D. Adhesions

Ans A Intussusception occurs when a portion of the alimentary tract is telescoped into an adjacent segment.

43. A 14-year-old girl with history of abdominal pain (peri-umbilical), postprandial, passing blood in stools, fever, weight loss since ten months. She also has episodes of passing blood in stools. What may be the likely diag- nosis?

A. Chronic appendicitis
B. Chronic pancreatitis
C. Crohn's disease
D. Bulimia DISORDERS OF ABDOMINAL WALL

Ans C Crohn's disease Blood in stools, abdominal pain and systemic features like fever and weight loss for months is suggestive of Inflammatory bowel disease. DISORDERS OF ABDOMINAL WALL Answers 48. Ans. b. Omphalocele Omphalocele
• • It is a midline defect of anterior abdominal wall, with herniation of abdominal viscera into base of umbilical cord. with
• • The gut is covered with peritoneum & amnion
• • Anomalies associated with omphalocele are: Chromosomal abnormalities (in ~43% cases) esp.

Neuroblastoma

44. A neonate presented with fever, lethargy, abdominal distension, vomiting and constipation. Clinically he was diagnosed as volvulus neonatorum with suspected perforation. Best investigation would be:

A. Plain X-ray
B. Barium enema
C. Upper GI endoscopy
D. Barium meal followthrough

Ans A Volvulus neonatorum What is it? Intestinal obstruction in a newborn due to twisting of bowel caused by malrotation or non-fixation of colon Investigations Barium meal followthrough is the investigation of choice as X-ray is not reliable in early stages of volvulus; However, if perforation is suspected, barium contrast should not be used because of risk of perotinitis

45. **Chromosomal anomalies more than 20% is associated with:**

A. Gastroschisis
B. **Omphalocele**
C. Spina bifida
D. Cleft palate

Ans B has a strong association with chromosomal anomalies (up to 30-40% in some series), unlike gastroschisis, which is rarely associated with karyotype abnormalities.

Trauma & Burns

46. **A male infant presented with distension of abdomen shortly after birth with passing of less meconium. Sub- se quently a full-thickness biopsy of the rectum was performed. The rectal biopsy is likely to show:**

A. Fibrosis of submucosa (AIIMS Nov 04)
B. **Lack of ganglion cells**
C. Thickened muscularis propria
D. Hyalinization of the muscular coat

Ans B "Whenever neonate presents with delayed passage of stool, Hirschsprung's disease should be suspected".

47. **A term newborn baby born by vaginal delivery had respiratory distress, grunt, with scaphoid abdomen. Most probable diagnosis is:**

A. Hyaline membrane disease
B. **Diaphragmatic hernia**
C. Pneumothorax
D. Meconium aspiration syndrome

Ans B

25

CHAPTER 25

General Pediatrics, Ethics & Laboratory Medicine



WHAT THIS CHAPTER COVERS

- Well Child Care
- Growth Monitoring
- Developmental Screening
- Immunization
- Nutrition Counseling
- Pediatric Ethics
- Informed Consent
- End of Life Care
- Research Ethics
- Laboratory Reference Ranges
- Common Lab Tests
- Blood Gas Interpretation
- CSF Analysis
- Urine Analysis

54 MCQs **14** subtopics

25.1 Well-Child Care & Child Health Programmes

- **Well-baby visits**: coincide with immunisation – **growth monitoring (plot on chart), development screening, nutrition counselling (IYCF), immunisation, anticipatory guidance (safety, sleep, oral health, screen time)**, vision/hearing screening.
- **Anticipatory guidance highlights**: **back to sleep (supine) – SIDS prevention** (firm mattress, no soft bedding, room-sharing without bed-sharing, avoid smoke exposure & overheating, pacifier at sleep, breastfeeding), car seats (rear-facing till ≥ 2 y), drowning prevention, poison storage, **no screen time < 18–24 months (except video calls); ≤ 1 h/day for 2–5 y** (IAP/AAP), first dental visit by 1 y, fluoride.
- **Sudden infant death syndrome (SIDS)**: unexplained death of infant < 1 y during sleep; **peak 2–4 months**; risk – **prone/side sleeping (most important modifiable)**, soft bedding, bed-sharing, maternal smoking, prematurity, overheating, winter, boys.
- **BRUE (brief resolved unexplained event)** – < 1 y, < 1 min, resolved: cyanosis/pallor, absent/irregular breathing, tone change, altered responsiveness; low-risk BRUE – reassurance.

Key national programmes (India)

Programme	Key points
ICDS (1975)	Anganwadi – supplementary nutrition, immunisation, health check-up, referral, pre-school education, nutrition & health education (6 services) for 0–6 y, pregnant/ lactating women
POSHAN Abhiyaan (2018) / Mission Poshan 2.0	Reduce stunting, undernutrition, anaemia, LBW
RMNCH+A (2013)	Continuum of care; life-cycle approach
IMNCI (2003)	Integrated management of neonatal & childhood illness (0–5 y) – assess, classify (colour-coded: pink – urgent referral, yellow – specific treatment, green – home care), treat, counsel
Facility-based newborn care	**SNCU (district), NBSU (FRU), NBCC (every delivery point)**
Home-Based Newborn Care (HBNC)	ASHA visits **6–7 times (day 1 (home deliveries), 3, 7, 14, 21, 28, 42)**
HBYC (Home-Based Young Child care)	ASHA visits at 3, 6, 9, 12, 15 months
RBSK (2013)	Screening 0–18 y for 4 Ds (32 conditions), DEICs
India Newborn Action Plan (INAP, 2014)	Target **single-digit NMR & SBR by 2030**
Anaemia Mukt Bharat (2018)	6×6×6 strategy, IFA, deworming
MAA (Mothers' Absolute Affection, 2016)	Breastfeeding promotion
National Deworming Day	**10 February & 10 August** – albendazole 1–19 y
Intensified Diarrhoea Control Fortnight (IDCF)/STOP Diarrhoea	ORS & zinc distribution
SAANS (2019)	Childhood pneumonia (social awareness & action)
LaQshya / MusQan	Labour room / child-friendly facility quality
JSY / JSSK	Institutional delivery incentive / **free care for sick infants up to 1 year**
Mission Indradhanush / IMI	Full immunisation coverage

- **Indicators (SRS 2021)**: **IMR 27, NMR 19, U5MR 31, MMR 97/1 lakh live births (2018–20)**; SDG 3.2 – **NMR ≤ 12, U5MR ≤ 25 by 2030**.

- **NFHS-5 (2019–21)**: stunting 35.5%, wasting 19.3%, underweight 32.1%, anaemia (6–59 m) 67.1%, exclusive breastfeeding < 6 m 63.7%, full immunisation 76.4%.

25.2 Growth Monitoring & Developmental Screening (summary)

- Growth: **WHO standards 0–5 y; IAP 2015 charts 5–18 y**; weigh every month till 1 y, 3-monthly 1–3 y (MCP card) – see Ch 1.
- Development: **TDSC, DDST-II, ASQ**, M-CHAT (autism) – see Ch 2.
- **Universal newborn hearing screening (OAE/AABR)**, red reflex, CCHD pulse oximetry, metabolic screen.

25.3 Paediatric Ethics & Law

Principle	Application
Autonomy	Respect for patient's choice – in children exercised by **parents (surrogate decision-makers)**; child's **assent** increasingly sought
Beneficence	Act in the **best interests of the child** (primary principle in paediatrics)
Non-maleficence	"First, do no harm"
Justice	Fair distribution of resources
Confidentiality	Adolescents – confidential care (sexual health, substance use) except risk of serious harm/abuse (POCSO mandates reporting)

- **Informed consent** – by **parent/legal guardian for < 18 years** (India – age of majority 18; **consent for medical examination ≥ 12 years for simple examination – IPC (now BNS) provision**; **assent from children ≥ 7 years**); emergency – treat without consent in best interest ("doctrine of necessity").
- **Mature minor / Gillick competence** (UK) – a child under 16 with sufficient understanding can consent; **Fraser guidelines** (contraceptive advice).
- **Parental refusal of life-saving treatment** (e.g. blood transfusion in Jehovah's Witness child) – doctor may seek court order / act in child's best interest in emergency.
- **Research ethics**: **ICMR National Ethical Guidelines for Biomedical Research involving Children (2017)**; **parental consent + child assent (≥ 7 y)**, minimal risk, prospect of direct benefit for higher risk, **Declaration of Helsinki**, Belmont Report; vulnerable population – Ethics Committee oversight; **audio-video recording of consent** in certain trials (India).
- **End-of-life care**: withholding/withdrawing life-sustaining treatment ethically equivalent; **palliative care** (pain control – WHO 2-step ladder for children: step 1 paracetamol/ibuprofen, step 2 morphine (strong opioid) – codeine & tramadol avoided in children); **Supreme Court (2018, simplified 2023) – passive euthanasia & advance directives legal in India**; DNR orders; **brain death** certification (Transplantation of Human Organs Act 1994 – panel of 4 doctors, 2 examinations 6 h apart).
- **Medical Termination of Pregnancy (Amendment) Act 2021**, **PCPNDT Act**, **POCSO**, **JJ Act**, **Right to Education**, **Child Marriage Prohibition (girls 18, boys 21)**.

25.4 Laboratory Reference Values (Paediatric)

Parameter	Neonate	Child	Notes
Hb (g/dL)	14–22 (cord 16.5)	11–14	Nadir 2–3 m (9.5–11)
HbF	60–80%	< 2% by 1–2 y	
WBC (/mm³)	9,000–30,000	5,000–15,000	**Lymphocyte predominance 4 days–4 years** (2 cross-overs: ~day 4–7 & ~4–7 y)

Parameter	Neonate	Child	Notes
Platelets	1.5–4.5 lakh	1.5–4.5 lakh	
Reticulocytes	3–7% (day 1)	0.5–2%	
MCV (fL)	100–120	70–86 (lower limit ≈ 70 + age in y)	Newborn macrocytosis normal
ESR	< 2–4 mm/h	< 10–20	
Blood glucose (mg/dL)	≥ 45–50	70–100 fasting	
Serum calcium	7–11	8.8–10.8	
Serum phosphate	4.5–9	**4–6.5 (higher than adults)**	
ALP (IU/L)	**↑↑ (up to 3–4× adult)**	**High during growth (esp. puberty)**	Bone isoenzyme
Creatinine (mg/dL)	0.3–1.0 (reflects maternal in first days)	0.2–0.7	↑ with age/muscle mass
BUN	3–12	5–18	
Serum albumin (g/dL)	2.5–3.5 (preterm)	3.5–5	
Total bilirubin	Physiological up to 12–15	< 1	
Ammonia (µmol/L)	**< 110 (neonate)**	< 50	Sample on ice, analyse in 30 min
Lactate (mmol/L)	0.5–2.0	0.5–2.0	
TSH (mU/L)	**Surge to 60–80 at 30 min after birth**, falls by 3–5 days	0.7–6	Screen after 48–72 h
IgG	= maternal (high)	Nadir 3–6 m	
CSF cells	< 20–30	< 5	
CSF protein (mg/dL)	40–150 (preterm up to 200)	15–40	

- **Neutrophil–lymphocyte crossover**: **neutrophils predominate at birth → lymphocytes predominate by 4–7 days → neutrophils again predominate by 4–7 years**.
- **Physiological "lymphocytosis" of childhood** – don't over-interpret.

25.5 Common Laboratory Tests – Interpretation Pearls

Test	Pearls
Sepsis screen	≥ 2 of: TLC < 5000, ANC low, I:T ≥ 0.2, CRP > 1 mg/dL, micro-ESR > 15 mm
CRP	Rises in 6–8 h, peaks 24–48 h, t½ ~19 h; **procalcitonin** rises in 2–4 h (bacterial specificity)
ESR	↑ in anaemia, hypergammaglobulinaemia (kala-azar, TB, SLE); **↓ in polycythaemia, sickle cell, spherocytosis, CHF, DIC, MAS (paradoxical), afibrinogenaemia**
Urine routine	Specific gravity 1.002–1.030; pH 4.5–8; glucose & ketones absent; **> 5 RBC/HPF haematuria; > 5–10 WBC/HPF pyuria**
Urinary reducing substances	Benedict (+) with glucose, galactose, fructose, lactose, pentose; **glucose oxidase dipstick – specific for glucose**
Sweat chloride	≥ 60 mmol/L = CF; 30–59 intermediate; < 30 normal
Tuberculin test	≥ 10 mm positive (≥ 5 mm immunocompromised/SAM/HIV/contact)

Test	Pearls
Widal	Single titre ≥ 1:160 (O & H) in endemic area, rising titre more significant
ASO	> 200 Todd units (children) – rises 1–3 weeks after pharyngitis; **anti-DNase B better for pyoderma**
Blood culture volume	Neonate ≥ 1 mL, child 1 mL per year of age (up to 10 mL)
G6PD assay	Falsely normal during haemolysis (repeat after 3 months)
HbA_2	> 3.5% = β-thal trait (masked by coexistent iron deficiency)
Ferritin	Acute-phase reactant – may be normal in IDA with infection

25.6 Blood Gas Interpretation (summary)

	pH	$PaCO_2$	HCO_3^-
Normal	7.35–7.45	35–45	22–26
Metabolic acidosis	↓	↓ (compensatory)	**↓ (primary)**
Metabolic alkalosis	↑	↑	**↑**
Respiratory acidosis	↓	**↑ (primary)**	↑
Respiratory alkalosis	↑	**↓**	↓

- **Cord blood gas**: **umbilical artery pH < 7.0 & base deficit ≥ 12 mmol/L** – significant asphyxia; normal UA pH ~7.25, venous higher.
- **Capillary/venous gas**: VBG pH ~0.03 lower, PCO_2 ~5–6 mmHg higher than arterial – adequate for pH/PCO_2 trends (DKA).
- **Oxygenation index (OI) = (MAP × FiO_2 × 100) / PaO_2; OI > 40 → ECMO** (neonates); A-a gradient.
- Classic paediatric ABG patterns: **pyloric stenosis – hypochloraemic hypokalaemic metabolic alkalosis**; **diarrhoea – normal-AG metabolic acidosis**; **DKA – high-AG metabolic acidosis**; **salicylate – respiratory alkalosis + high-AG acidosis**; **severe asthma – rising $PaCO_2$ (ominous)**; **CF in heat – hypochloraemic metabolic alkalosis**; **IEM (organic acidaemia) – high-AG acidosis + hyperammonaemia**; **urea cycle – respiratory alkalosis**.

25.7 Normal Vital Signs (quick reference)

Age	RR	HR	SBP
Newborn	40–60	120–160	60–90
Infant	30–50	100–160	80–100
Toddler (1–3 y)	24–40	90–150	90–105
Preschool (3–5 y)	22–34	80–140	95–110
School (6–12 y)	18–30	70–120	100–120
Adolescent	12–20	60–100	110–135

- **Temperature**: **fever = rectal ≥ 38 °C (100.4 °F)**; axillary ~0.5 °C lower; **fever without focus in < 3 months → full sepsis work-up**; hyperpyrexia > 41 °C.
- **Fever in < 1 month = medical emergency** (blood, urine, CSF cultures + IV antibiotics).

Practice MCQs · General Pediatrics, Ethics & Laboratory Medicine

54 questions · correct option highlighted in green · cover the answer strip to self-test

Well Child Care

1. The MOST common cause of under-5 mortality in developing world is

A. diarrheal disease
B. pneumonia
C. malaria
D. measles
E. neonatal disease

Ans E Causes of under-5 mortality differ markedly between developed and developing nations. In developing countries, 66% of all deaths resulted from infectious and parasitic diseases. Among the 42 countries having 90% of childhood deaths, diarrheal disease accounted for 22% of deaths, pneumonia 21%, malaria 9%, AIDS 3%, and measles 1%. Neonatal causes contributed to 33% of the total death but 24% of deaths are caused by severe infections and 7% by tetanus.

2. The well-child care (anticipatory guidance) intends to promote the physical and emotional well-being of children. The tasks of each well-child visit (which usually takes 18 min time) include all the following EXCEPT

A. disease detection
B. disease prevention
C. treatment plans
D. health promotion
E. accident prevention

Ans C This is a well child visit; treatment plans are not included here.

3. Children who grow up on farms have elevated rates of

A. mesothelioma
B. leukemia
C. skin tumor
D. lymphoma
E. carcinoma of lungs

Ans B Children who grow up on farms have elevated rates of leukemia; pesticides are suspected of playing an etiologic role.

4. Specific antivenoms (AV) are available for many venomous creatures of the world, particularly snakes, spiders, and scorpions. All the following about antivenoms are true EXCEPT

A. AV is capable of neutralizing only circulating venom
B. it is beneficial to give AV locally at the bite site
C. most AVs are given intravenously
D. skin tests are unreliable
E. intravenous AV should be started slowly

Ans B Skin tests, often recommended by AV manufacturers, are unreliable and should be omitted.

Growth Monitoring

5. The LEAST common cause of death < 1 yr in USA is

A. congenital malformations
B. short gestation and low birth weight
C. sudden infant death syndrome
D. unintentional injuries
E. bacterial sepsis of newborn

Ans E Bacterial sepsis constitutes only 2%. Other distracters are arranged from common to least common.

6. Osteopenia begins immediately after a spinal cord injury (SCI) occurs and plateaus 6- 12 mo later. Pathologic fractures occur as a consequence of loss of bone mineral density. Of the following, the MOST common site of fracture is

A. distal tibia
B. supracondylar region of the femur
C. lumbosacral spine
D. proximal humerus
E. distal radius

Ans B The most common sites of fracture include the supracondylar region of the femur and the proximal tibia.

7. Arsine gas is colorless, odorless, nonirritating, and highly toxic. Inhalation causes no immediate symptoms. After a latent period of 2-24 hr, exposed individuals experience

A. hematuria
B. sensorimotor peripheral neuropathy
C. respiratory failure
D. massive hemolysis
E. proteinuria

Ans D After a latent period of 2-24 hr, exposed individuals experience massive hemolysis, malaise, headache, weakness, dyspnea, nausea, vomiting, abdominal pain, hepatomegaly, pallor, jaundice, hemoglobinuria, and renal failure.

8. One of the following differentiate organophosphate poisoning and methamphetamine intoxication from bark scorpion envenomations

A. paresthesias
B. roving eye movements
C. cranial nerve dysfunction
D. seizures
E. hypertensive crisis

Ans B Two important ingestions that can be confused with a bark scorpion envenomation are organophosphate poisoning and methamphetamine intoxication. The opsoclonus-like roving eye movements seen in bark scorpion envenomations are not seen in the above ingestions and may help differentiate these conditions.

Developmental Screening

9. In mental retardation, all the following are true EXCEPT

A. constitute 1-3% of children
B. 80% are mild form
C. **in severe form socio-economic status is a considerable factor**
D. higher percentage in low birthweight babies
E. fragile X chromosome is a recognized cause

Ans C In severe form of mental retardation the socio-economic factor is not effective parameter while in mild forms it plays a role.

10. Abrupt withdrawal of baclofen may cause

A. depression
B. drowsiness
C. headache
D. euphoria
E. **seizure**

Ans E Abrupt withdrawal may cause seizures, hallucinations, rebound muscle spasms, and hyperpyrexia.

11. Acute ingestion of inorganic mercury salts (typically secondary to ingestion of a button battery) can manifest in a few hours as

A. tremor
B. neuropsychiatric disturbances
C. gingivostomatitis
D. **corrosive gastroenteritis**
E. necrotizing bronchiolitis

Ans D Acute ingestion of inorganic mercury salts (typically secondary to ingestion of a button battery) can manifest in a few hours as corrosive gastroenteritis, signified by metallic taste, oropharyngeal burns, nausea, hematemesis, severe abdominal pain, hematochezia, acute tubular necrosis, cardiovascular collapse, and death.

12. The closeness of a measured value to a standard or known value is termed as

A. **accuracy**
B. precision
C. sensitivity
D. specificity
E. none of the above

Ans A is a measure of the nearness of a test result to the actual value, whereas precision is a measure of the reproducibility of a result.

Immunization

13. An 8-month-old infant with confirmed diagnosis of spinal muscular atrophy type 1 referred back from tertiary care center with "DNAR" status. This means during acute illness or an emergency the child should

A. receive standard care and adrenaline injection
B. **receive supportive care**
C. receive active resuscitation but not mechanical ventilation
D. O2 is not part of treatment options
E. none of the above

Ans B DNAR status is a statement issued by medical ethical group (which is a group of consultants) regarding certain untreatable medical illnesses, it means do not attempt active resuscitation, so all supportive care, medications, and O2 are allowed, adrenaline is a medication used in active resuscitation.

14. Oral medications are often used as an early treatment for generalized spasticity. Which of the following works at the level of skeletal muscle to block calcium release from the sarcoplasmic reticulum?

A. **dantrolene sodium**
B. clonidine
C. tizanidine
D. baclofen
E. diazepam

Ans A works at the level of skeletal muscle to block calcium release from the sarcoplasmic reticulum. Despite its peripheral site of action, dantrolene may induce sedation, although to a lesser degree than other centrally acting agents.

15. Acrodynia, or pink disease, is a rare idiosyncratic hypersensitivity reaction to mercury that occurs predominantly in children exposed to mercurous powders. The symptom complex includes the following EXCEPT

A. generalized pain
B. paresthesias
C. an acral red-pink rash
D. photophobia
E. **generalized spasticity**

Ans E The symptom complex includes generalized pain, paresthesias, and an acral (hands, feet) rash that may spread to involve the face. The rash typically is red-pink, papular, pruritic, and painful; it may progress to desquamation and ulceration. Morbilliform, vesicular, and hemorrhagic variants have been described. Other important features include anorexia, apathy, photophobia, and hypotonia, especially of the pectoral and pelvic girdles.

16. You asked to obtain a measurement of serum sodium level in the hospital lab; you obtain a value of 125 mmol/L by repeating the test 5 times. The reference value of serum sodium at that lab is between 135-155 mmol/L. Your measurement is termed

A. accurate but not precise
B. **precise but not accurate**
C. neither accurate nor precise
D. both accurate and precise
E. sensitive and specific

Ans B Precision is the repeatability of the measurement. The measurement can be precise but not accurate, accurate but not precise, both accurate and precise, or not accurate nor precise.

Nutrition Counseling

17. A full term neonate with Down syndrome and esophageal atresia admitted in Pediatric intensive care unit (PICU). The parents want “no interference" approach. Of the following, the BEST response is to

A. act according to parents well
B. arrange the operation with the surgeon
C. **work for the patient best interest**
D. give supportive care only (DANR)
E. wait for a clear legislation

Ans C Pediatrician is a child advocate; he must take decision always to the "best interest" of the child that what is called beneficent paternalism in pediatrics which is the obligation to act in a younger child’s best interest that takes moral precedence over the wishes of the child’s parent(s), so in such a scenario arrangement with the surgeon to do the operation and calling the local hospital committee to protect the child is the best choice but this action is still need legislation in the third world.

18. Coadministration of botulinum toxin (BTX) and one of the following drugs should be performed with caution as the effect of the toxin may be potentiated

A. **aminoglycosides**
B. cephalosporines
C. penicillins
D. macrolids
E. sulphonamides

Ans A Coadministration of BTX and aminoglycosides or other agents interfering with neuromuscular transmission (curare-like nondepolarizing blockers, lincosamides, polymyxins, quinidine, magnesium sulfate, anticholinesterases, succinylcholine chloride) should be performed with caution as the effect of the toxin may be potentiated.

19. It is estimated that lead-poisoned children are identified by screening procedures rather than through clinical recognition of lead related symptoms in

A. **99% of cases**
B. 79% of cases
C. 59% of cases
D. 39% of cases
E. 19% of cases

Ans A It is estimated that 99% of lead-poisoned children are identified by screening procedures rather than through clinical recognition of lead related symptoms.

20. Assume you are going to estimate the prevalence of amoebic dysentery in a small country which harbors a total number of population of 530,000; you find that 57,000 of the population are infected by the disease. The prevalence of this disease is closest to

A. 5.33%
B. 7.45%
C. **10.75%**
D. 20.22%
E. 25.3%

Ans C Prevalence refers to the percentage of the infected people from the total number of population.

Pediatric Ethics

21. You are resident in Pediatrics, called at midnight to attend delivery of expected baby with multiple malformations and oligohydromnias as had been reported in antenatal scan, mother in stage I labor so far. Of the following, your BEST plan of action is to

A. counsel the parents that you will go for (DNAR) status
B. start your usual resuscitation waiting for your senior
C. explain that the result of antenatal scan is final
D. **call your consultant or senior resident**
E. apply palliative care for the coming neonate

Ans D In such situation when you have time (mother still in stage I labor), the best response is to call your direct senior resident or consultant then explain the result of antenatal scan trying to put the parents in the expected situation. Starting the usual resuscitation is the best choice when there is no time and the baby arrived. The decision of (DNAR) should be made by the ethical hospital committee or supervising consultant and should be powered by national legislations.

22. Risk factors for birth brachial plexus injury include the following EXCEPT

A. shoulder dystocia
B. birthweight greater than 4 kg
C. **primiparous mothers**
D. mothers with excessive weight gain
E. diabetic mothers

Ans C Risk factors for birth brachial plexus injury include prior infants with BBPP, shoulder dystocia, birthweight greater than 4 kg, multiparous mothers, mothers with excessive weight gain, and diabetic mothers. Delivering twins or triplets, as well as cesarean sections, have been described as protective from BBPP.

23. Mushrooms are an ideal food because they are

A. **low in calories**
B. high in calories
C. high in fat
D. low in fat
E. low in protein

Ans A Mushrooms are an ideal food. They are low in calories, fat free, and high in protein, making them a great source of nutrition.

24. Assume you have the results of PCR tests for Mycoplasma pneumoniae; the number of positive tests is 83 collected from a 100 truly-infected persons. The sensitivity of this test is

A. 17%
B. 55%
C. 60%
D. **83%**
E. 95%

Ans D Sensitivity means the probability that a patient who is disease positive will test positive. Sensitivity = number positive by test×100 /total number positive.

Informed Consent

25. A 7-year-old boy presented to the out-patient department with fever and cough, you have diagnosed him as a case of pneumonia and decided to admit and give parental therapy, the child refused. Of the following, the BEST response is to

A. explain to father about dangers of omitting treatment
B. try to explain the risk to the child
C. **give the chance for oral antibiotics and accept the child decision**
D. inform your consultant
E. inform hospital ethical committee

Ans C In older children and in noncritical situation the principle of the "child assent"which means the child voice must be heard should be applied. In this scenario, the situation is mild and the child non acceptance of injectable medication should be respected. It is still in critical situation or more acute illness the child best interest should be sought and it may be against his well.

26. The baby with birth brachial plexus palsy (BBPP) will start with occupational or physical therapy at approximately

A. 1 wk of age
B. **2 wk of age**
C. 3 wk of age
D. 4 wk of age
E. 5 wk of age

Ans B

27. One of the following is not recommended for treatment of Amanita poisoning

A. oral activated charcoal
B. **forced diuresis**
C. silibinin
D. intravenous penicillin G
E. acetylcysteine

Ans B should be avoided, as this increases renal exposure.

28. Assume you are evaluating the specificity of the indirect fluorescence antibody test for rapid diagnosis of Campylobacter enteritis. The number of normal persons tested is 150 and the test is positive in 30 persons. The specificity of this test is

A. 20%
B. 40%
C. 60%
D. 70%
E. **80%**

Ans E Specificity (also called the true negative rate) measures the proportion of negatives that are correctly identified as such (e.g., the percentage of healthy people who are correctly identified as not having the condition). Specificity = number negative by test×100 /total number without disease.

End of Life Care

29. You are treating acutely ill 6-year-old child with ascending paralysis, he eventually needs mechanical ventilation. Now you are in a situation of using the last bed in respiratory care unit (RCU). Of the following, the BEST approach is to

A. try to convince the parents that, there is no use of ventilation
B. keep the bed for more treatable condition
C. try bedside ventilation
D. **consider this case as potential treatable condition and use the last bed**
E. contact a nearby region RCU and transfer the child

Ans D It is clear in this scenario that the child suffers potentially treatable condition. In this situation use the best resources and the best care. The choices of (A&B) are definitely non ethical. Other choices are optional when there is no bed is available in RCU taking the best interest for the patient in consideration.

30. Prenatal screening is recommended for all pregnant women to detect neural tube defect. If a neural tube defect is present, one of the following is often elevated

A. human chorionic gonadotropin
B. **α-fetoprotein**
C. estriol
D. inhibin
E. prolactin

Ans B Prenatal screening is recommended for all pregnant women to detect neural tube defect. A simple blood test is done in the 2nd trimester to evaluate α-fetoprotein, human chorionic gonadotropin, estriol, and inhibin. If a neural tube defect is present, the α-fetoprotein is often elevated and further screening using high-resolution ultrasound is indicated.

31. Mushrooms of the genera Inocybe contain muscarine or muscarine-related compounds. These quaternary ammonium derivatives bind to postsynaptic receptors, 'producing an exaggerated cholinergic response. Of the following, the MOST serious complication is

A. diaphoresis
B. bradycardia
C. hypotension
D. vomiting
E. **bronchospasm**

Ans E Respiratory distress caused by bronchospasm and increased bronchopulmonary secretions is the most serious complication.

32. **You are assigned to evaluate the outcome of fecal occult blood test as a screening test for patients with bowel cancer. The total number of patients who underwent endoscopy is 2030; in those whom the disease is confirmed are 30. The results of fecal occult blood test are as follows: positive in 20 patients who has a confirmed bowel cancer (truly positive), and in 180 patients who don`t have the disease (falsely positive); and negative in 10 patients who truly has the disease (falsely negative), and in 1820 patients who are not affected by the disease (truly negative). All the following statements are true EXCEPT**

A. the prevalence of the disease for those population is 1.48%
B. the sensitivity of the test is 67%
C. the specificity of the test is 91%
D. **the positive predictive value is 20%**
E. the negative predictive value is 99.5%

Ans D PV of a positive test result = True-positive results× 100 / Total positive results. PV of a negative test result = True-negative results× 100 /Total negative results.

Research Ethics

33. **You are managing a 10-year-old child with spastic quadriplasia, who is globally retarded, had recurrent seizures, and severe recurrent chest infections which requires frequent admissions to respiratory care unit (RCU) and ventilation. You are trying to convince the mother about non-benefit of future RCU admissions. All of the following statements are true EXCEPT**

A. **use an obligations of no bed availability**
B. consider mother emotional status and religious believes
C. be empathetic
D. explain disease chronicity and non-response status
E. explain the suffering of child in each ventilation

Ans A In such difficult situation (chronic disabling illnesses) and the child in vegetative state; use the approach from B to E steps and allow the final decision to the parents especially if you are not covered with clear legislations.

34. **Hindbrain herniation or the Chiari type II malformation is seen in which percent of individuals with myelomeningocele**

A. 10-20%
B. 20-30%
C. 40-50%
D. 60-70%
E. **80-90%**

Ans E

35. **Pediatricians are likely to experience unique problems in managing childhood victims of biologic or chemical attack. The very rapid onset of neuromuscular symptoms after an exposure should lead the clinician to consider**

A. botulism
B. **nerve agent intoxication**
C. chlorine
D. phosgene
E. cyanide

Ans B The very rapid onset of neuromuscular symptoms after an exposure should lead the clinician to consider nerve agent intoxication. The nerve agents (tabun, sarin, soman, and VX) are organophosphate analogs of common pesticides that act as potent inhibitors of the enzyme acetylcholinesterase. They are hazardous via ingestion, inhalation, or cutaneous absorption.

36. **Rapid HIV antibody testing procedures using a finger stickor venipuncture to obtain whole blood, plasma, or serum, andtests using oral fluid were approved. All the following regarding this test are true EXCEPT**

A. they are simple and accurate as to render the likelihood of an erroneous result by the user negligible
B. a positive result does not need confirmation by Western blot analysis or immunofluorescence assay
C. they allow for implementation of antiretroviral therapy for HIV-infected women who have not been tested or are unaware of their HIV status
D. **they significantly reduce the risk of mother-to-child transmission**
E. The median turnaround time for obtaining results from blood collection to patient notification was only 66 minutes

Ans D A positive rapid HIV test result is then confirmed by Western blot analysis or immunofluorescenceassay. Rapid whole blood HIV testing is now the standard of care for women in labor with undocumented HIV status. Rapid HIV testing can also be used in developing countries. In resource-poor settings, because of the lack of properly equipped laboratories, skilled technologists, and basic resources, such as electricity and water.

Laboratory Reference Ranges

37. **After the first year of life, the MOST common cause of death in children is mainly**

A. none-accidental injuries
B. burn
C. drowning
D. **accidents**
E. war

Ans D The most common cause of death in children beyond the first few months of life is injuries which includes road traffic accidents and other none-intentional injuries, with up to 50-60% of total causes of death.

38. **In utero radiation exposure is associated with an excess risk of dying from leukemia before age 10 yr. Of the following, the MOST acceptable risk percent is**

A. 32%
B. 52%
C. 75%
D. **92%**
E. 180%

Ans D In utero radiation exposure is associated with a 92% excess risk of dying from leukemia before age 10 yr and a 180% excess risk of dying from other malignant diseases. Even 40 yr later there is a 228% increased relative risk of cancer associated with radiation in utero.

39. Pediatricians are likely to experience unique problems in managing childhood victims of biologic or chemical attack. A delayed onset of respiratory symptoms (days after exposure) is characteristic of

A. chlorine
B. phosgene
C. cyanide
D. **anthrax**
E. sarin

Ans D A delayed onset of respiratory symptoms (days after exposure) is characteristic of several infectious diseases and 1 toxin that might be adapted for sinister purposes by terrorists. Among the most threatening and problematic of these are anthrax, plague, tularemia, and ricin, the latter having garnered considerable media attention in recent years.

40. "Normal values" (reference intervals) are difficult to establish within the pediatric population. Many variables should be considered when developing reference intervals. All the following are variables that commonly considered to define reference intervals

A. genetic composition
B. physiologic development
C. environmental influences
D. subclinical disease
E. **ethnicity**

Ans E Other considerations for further defining reference intervals include partitioning based on sex and age.

41. The most commonly used reference range is generally given as the mean of the reference population ±2 standard deviations (SD). The term "normal distribution" refers to which of the following?

A. **gaussian distribution**
B. exponential distribution
C. skewed distribution
D. uniform distribution
E. unimodal distribution

Ans A All others are non-normal distribution.

Common Lab Tests

42. The MOST successful injury prevention strategy is

A. car seats
B. water safety
C. **changing in product design**
D. poison prevention
E. fall prevention

Ans C The most successful injury-prevention strategies generally are those involving changes in product design. Such as making more secure cars, more suitable car seats ...etc.

43. The following diseases are associated with sensitivity to radiation EXCEPT

A. ataxia-telangiectasia
B. **Edward syndrome**
C. Down syndrome
D. Fanconi anemia
E. Gardner syndrome

Ans B Diseases that are associated with sensitivity to radiation are 🅵🅾🅳 Ataxia-telangiectasia 🅵🅾🅳 Basal cell nevoid syndrome 🅵🅾🅳 Cockayne syndrome 🅵🅾🅳 Down syndrome 🅵🅾🅳 Fanconi anemia 🅵🅾🅳 Gardner syndrome 🅵🅾🅳 Nijmegen breakage syndrome 🅵🅾🅳 Usher syndrome

44. Pediatricians are likely to experience unique problems in managing childhood victims of biologic or chemical attack. Patients suffering from the sudden onset of severe neuromuscular symptoms may have nerve agent intoxication and should be given atropine (0.05 mg/kg) promptly for its antimuscarinic effects. Atropine has the following effects EXCEPT

A. relieves bronchospasm
B. relieves bradycardia
C. reduces bronchial secretions
D. ameliorates diarrhea
E. **improve skeletal muscle paralysis**

Ans E Pralidoxime (also known as 2-PAM) cleaves the organophosphate moiety from cholinesterase and regenerates intact enzyme if "aging" has not occurred. The effect is most prominent at the neuromuscular junction and leads to improved muscle strength. Its prompt use (at a dose of 25 mg/kg) as an adjunct to atropine is recommended in all serious cases.

45. Almost all of the diseases detected in neonatal screening programs have a very low prevalence; the strategy is to use the initial screening test to separate a highly suspect group of patients from normal infants (i.e., to increase the prevalence) and then to follow this suspect group aggressively. Of the following, the disease that has low prevalence rendering it not useful for neonatal screening testing is

A. phenylketonuria
B. hypothyroidism
C. sickle cell disease
D. cystic fibrosis
E. **neuroblastoma**

Ans E A simple spot test for VMA is not useful in general screening programs because of the low prevalence of neuroblastoma (3 cases/100,000) and the low sensitivity of the test (69%). Sickle cell disease, easily detected using liquid chromatography or isoelectric focusing, can betreated more effectively if it is diagnosed before clinical signs appear.In addition, the results of neonatal screening for cystic fibrosis show that there are clear benefits associated with preclinical diagnosis.

Blood Gas Interpretation

46. Around 44,000-98,000 patients die in U.S. hospitals each year because of preventable medical errors, these figures can be reduced if adapting an emerging new science of Quality Improvement (QI). The TRUE figure of children receive recommended health care in USA is about

A. 16%
B. 26%
C. 36%
D. **46%**
E. 80%

Ans D

47. The acute effects of radiation therapy (occurring less than 3 mo after therapy begins) are usually related to the area of the body being irradiated. Of the following, the MOST severe acute reactions is

A. **pneumonitis**
B. dermatitis
C. mucositis
D. esophagitis
E. cerebral edema

Ans A The acute effects of therapy (occurring less than 3 mo after therapy begins) are usually related to the area of the body being irradiated (except fatigue, which can begin during this time period). These acute effects include radiation-caused pneumonitis, dermatitis, mucositis and esophagitis, cerebral edema, and swelling of the organ irradiated. There may be changes in bowel movement patterns. Of these, one of the most severe acute reactions is pneumonitis.

48. In cases in which the delayed onset of respiratory symptoms may be the result of a terrorist attack, consideration should be given to the empirical administration of an antibiotic effective against anthrax, plague, and tularemia. A reasonable choice is

A. **ciprofloxacin**
B. rifampin
C. vancomycin
D. ampicillin
E. clarithromycin

Ans A (10-15 mg/kg IV q12h), levofloxacin (8 mg/kg IV q12h), or doxycycline (2.2 mg/kg IV q12h) is areasonable choice.

CSF Analysis

49. Patient culture can change the doctor treatment options and even the way of behavior with the patient. All the following culture values are expected if he is dealing with Muslim patients

A. fasting during Ramadan month
B. modesty and female body cover
C. forbiddance of touch and shake hands of opposite sex
D. **routine postmortem exam**
E. patriarchal way of decision making

Ans D All the above values are expected from Muslim culture, except submitting the dead body for routine postmortem exam. The value (E) means the way of decision making is usually left to older person in the family.

50. Primary malignancies with the highest cumulative incidence of a second neoplasm are

A. soft tissues sarcoma
B. cancers of bone
C. **Hodgkin disease**
D. leukemias
E. central nervous system (CNS) cancers

Ans C Primary malignancies with the highest cumulative incidence of a second neoplasm in the order of frequency are Hodgkin disease (7.6), soft tissuesarcoma (4.0), cancers of bone (3.3), leukemia (2.1), central nervous system (CNS) cancers (2.1), and non-Hodgkin disease lymphoma (1.9).

51. Infection is the most common complication of bite injuries, regardless of the species of biting animal. The MOST common microorganisms associated with cat bites are

A. Staphylococcus species
B. Streptococcus species
C. Eikenella species
D. **Pasteurella species**
E. Proteus species

Ans D P. multocida is the predominant species in at least 50% of cat bite wound infections.

Urine Analysis

52. Children with severe traumatic brain injury (TBI) may experience autonomic dysfunction characterized by all the following EXCEPT

A. elevated temperature
B. elevated heart rate
C. elevated respiratory rate
D. **lowered blood pressure**
E. diaphoresis

Ans D Children with severe TBI may experience autonomic dysfunction characterized by elevated temperature, heart rate, respiratory rate, and blood pressure, accompanied by diaphoresis and posturing. Autonomic dysfunction is a diagnosis of exclusion and is associated with poor outcome after acquired brain injury in children.

53. Children exposed to second-hand tobacco smoke have increased frequency of

A. **middle ear effusions**
B. bacterial respiratory illnesses
C. otitis externa
D. sinusitis
E. tonsilitis

Ans A Children exposed to second-hand tobacco smoke have increased frequency of lower respiratory illness, more middle-ear effusions, and more viral respiratory illnesses than unexposed children.

54. **The MOST important proposed field treatments for snake bites is**

A. tourniquets
B. **immobilization of injured body part at the level of the heart**
C. ice
D. incision
E. suction

Ans B Constrictive clothing, jewelry, and watches should be removed, and the injured body part should be immobilized in a position of function at the level of the heart. All proposed field treatments for snake bites, such as tourniquets, ice, electric shock, incision, and suction, have proven problematic, with most being ineffective and deleterious.

26

CHAPTER 26

Pediatric Pharmacology



WHAT THIS CHAPTER COVERS

- Pharmacokinetics in Children
- Drug Dosing
- Antibiotics in Pediatrics
- Analgesics & Antipyretics
- Antiepileptic Drugs
- Asthma Medications
- Corticosteroids
- Vaccines & Biologics
- Adverse Drug Reactions
- Poisoning & Overdose
- Drug Safety in Lactation
- Drug Safety in Pregnancy

89 MCQs **12** subtopics

26.1 Pharmacokinetics in Children

Process	Neonate/infant difference	Consequence
Absorption	**Higher gastric pH (achlorhydria at birth)**, delayed gastric emptying, thin stratum corneum (↑ skin absorption)	↑ absorption of **acid-labile drugs (penicillin G)**, ↓ of weak acids (phenobarbitone, phenytoin); **topical toxicity – hexachlorophene, boric acid, steroids, alcohol, iodine (hypothyroidism)**
Distribution	**↑ total body water & ECF**, ↓ body fat, ↓ plasma protein binding (↓ albumin, fetal albumin, bilirubin competes), **immature BBB**	**Larger Vd for water-soluble drugs (aminoglycosides – higher mg/kg dose)**; ↑ free drug; **sulfonamides/ceftriaxone displace bilirubin → kernicterus**
Metabolism	**↓ CYP450 & glucuronidation (UGT) at birth** – matures by 6 m–1 y; **sulfation well-developed**; **children 1–10 y – higher hepatic clearance than adults** (need higher mg/kg doses, e.g. theophylline, phenytoin, carbamazepine)	**Chloramphenicol → grey baby syndrome**; ↑ t½ of caffeine, theophylline, diazepam in neonates; **paracetamol – sulfation (less hepatotoxic in young children)**
Excretion	**GFR low at birth (adult by 1–2 y)**, tubular secretion matures by 6–12 m	**↑ dosing interval for renally excreted drugs (aminoglycosides, vancomycin, penicillins)** in neonates

- **Caffeine t½: ~100 h in neonates** (3–5 h adults) – once daily dosing.
- **Grey baby syndrome**: **chloramphenicol** in neonates – **↓ glucuronyl transferase + ↓ renal excretion** → vomiting, abdominal distension, **ashen-grey cyanosis, hypothermia, flaccidity, cardiovascular collapse**, metabolic acidosis.

26.2 Drug Dosing in Children

- **Most accurate: by body weight (mg/kg)** for most drugs; **body surface area (BSA)** for chemotherapy, some drugs (GH, steroids in nephrotic syndrome).
- **BSA (Mosteller) = √[height (cm) × weight (kg) / 3600]**; average adult BSA 1.73 m^2; **child dose = adult dose × BSA (m^2)/1.73** (≈ percentage method).
- Historical formulae (not recommended now): **Young's (age/(age + 12) × adult dose)**, **Clark's (weight in lb/150 × adult dose)**, Fried's (infants – age in months/150), Dilling's (age/20).
- **Never exceed adult maximum dose**; use **liquid formulations** (syrups, drops, dispersible tablets), measure with syringe (not kitchen spoons).
- Neonates: dose by **postmenstrual & postnatal age** (gentamicin – 4–5 mg/kg every 24–48 h by gestation).
- **Therapeutic drug monitoring**: aminoglycosides, vancomycin, phenytoin, phenobarbitone, digoxin, theophylline, ciclosporin/tacrolimus, methotrexate, busulfan.

Common doses (high-yield)

Drug	Dose
Paracetamol	**10–15 mg/kg/dose every 4–6 h** (max 60 mg/kg/day; up to 75 in hospital); IV 15 mg/kg (7.5 in neonates)
Ibuprofen	**5–10 mg/kg/dose every 6–8 h** (max 40 mg/kg/day); > 3 months
Amoxicillin	25–50 mg/kg/day (standard), **80–90 mg/kg/day (AOM, pneumonia – high dose)**
Ceftriaxone	50–75 mg/kg/day (100 mg/kg/day in meningitis)
Gentamicin / Amikacin	5–7.5 mg/kg/day / 15–20 mg/kg/day once daily
Vancomycin	15 mg/kg/dose 6–8-hourly

Drug	Dose
Azithromycin	10 mg/kg/day × 3–5 days
Cotrimoxazole	8–10 mg/kg/day TMP (treatment); 5 mg/kg TMP thrice weekly/daily (prophylaxis); **20 mg/kg/day TMP for PCP**
Salbutamol (neb)	0.15 mg/kg (min 2.5 mg)
Prednisolone	1–2 mg/kg/day (asthma), 2 mg/kg (nephrotic, max 60)
Dexamethasone (croup)	0.15–0.6 mg/kg single dose
Adrenaline (anaphylaxis)	**0.01 mg/kg IM (1:1000)**, max 0.5 mg
Adrenaline (arrest)	0.01 mg/kg IV/IO (1:10,000)
Phenobarbitone	20 mg/kg loading; 3–5 mg/kg/day maintenance
Midazolam (seizure)	0.2–0.3 mg/kg buccal/IN; 0.1–0.2 mg/kg IV
Ondansetron	0.15 mg/kg (max 8 mg)
ORS/Zinc	See Fluids
Iron (treatment)	3–6 mg/kg/day elemental
Vitamin K (newborn)	1 mg IM (0.5 mg < 1000 g)

26.3 Antibiotics – Paediatric Cautions

Drug	Paediatric concern
Tetracyclines (doxycycline, etc.)	**Tooth discolouration & enamel hypoplasia, ↓ bone growth** – avoid < 8 y (**short courses of doxycycline now accepted in all ages – rickettsial/scrub typhus**); pseudotumor cerebri
Fluoroquinolones	**Arthropathy/cartilage damage in juvenile animals** – restricted (allowed: MDR infections, CF, anthrax, dysentery, typhoid in some settings)
Chloramphenicol	**Grey baby syndrome**, aplastic anaemia (idiosyncratic), dose-related marrow suppression
Sulfonamides / ceftriaxone	**Kernicterus in neonates (displace bilirubin)**; **ceftriaxone + IV calcium → fatal precipitates in neonates (contraindicated in neonates receiving Ca-containing IV fluids)**; biliary pseudolithiasis
Erythromycin/ azithromycin	**Infantile hypertrophic pyloric stenosis (< 2 weeks)**
Aminoglycosides	Oto- & nephrotoxicity; **potentiate NM blockade (avoid in infant botulism)**
Nitrofurantoin	Haemolysis in G6PD deficiency & neonates (< 1 m)
Linezolid	Thrombocytopenia, lactic acidosis
Colistin	Nephro/neurotoxicity
Benzyl alcohol (preservative)	**"Gasping syndrome"** in neonates – metabolic acidosis, gasping, collapse
Propylene glycol	Hyperosmolality, seizures (neonates)

26.4 Analgesics & Antipyretics

- **Paracetamol** – first-line antipyretic/analgesic; toxicity > 150 mg/kg (> 200 in < 6 y) – **NAC**.
- **Ibuprofen** – avoid in dehydration (AKI), varicella (necrotising fasciitis/invasive GAS – debated), dengue, bleeding disorders, < 3 months.

- **Aspirin – avoid < 16 years (Reye syndrome)**; exceptions – **Kawasaki disease, ARF (anti-inflammatory), JIA, antiplatelet (post-cardiac surgery)**.
- **Nimesulide – banned in children < 12 y in India (hepatotoxicity)**; **metamizole (dipyrone) – agranulocytosis**.
- **Codeine & tramadol – contraindicated < 12 y and post-tonsillectomy (< 18 y)** – **CYP2D6 ultra-rapid metabolisers → respiratory depression/death**.
- **Morphine** – preferred opioid; neonates – ↑ sensitivity (respiratory depression).
- **Febrile seizures are not prevented by antipyretics**; **alternating paracetamol & ibuprofen not recommended** routinely; sponging with tepid water only adjunct.
- **Topical anaesthesia**: EMLA (prilocaine → **methaemoglobinaemia in neonates**), sucrose 24% (procedural analgesia in neonates).

26.5 Antiepileptics, Asthma Drugs & Corticosteroids (summary)

- AEDs – see Neurology (phenytoin gum hypertrophy; valproate hepatotoxicity < 2 y; carbamazepine SJS – HLA-B*1502; vigabatrin visual field loss; phenobarbitone hyperactivity).
- **Asthma**: ICS (budesonide, fluticasone) – growth velocity ↓ ~0.5 cm (first year); SABA (salbutamol – tremor, tachycardia, **hypokalaemia**); **montelukast – neuropsychiatric events (nightmares, agitation, suicidality – FDA box warning)**; theophylline – narrow therapeutic index (seizures, arrhythmias); **LABA never as monotherapy**.
- **Corticosteroids**: **growth suppression (alternate-day dosing minimises)**, Cushingoid, hypertension, hyperglycaemia, osteoporosis, cataract (posterior subcapsular), glaucoma, avascular necrosis, infections (varicella – severe), **adrenal suppression (taper if > 2 weeks)**, behavioural change; **live vaccines deferred** (≥ 2 mg/kg or ≥ 20 mg/day for ≥ 14 days).
- **Hydrocortisone stress dosing** for adrenal insufficiency/CAH during illness/surgery.

26.6 Vaccines & Biologics (pharmacology view)

- **Immunoglobulins**: IVIG (Kawasaki 2 g/kg, ITP 0.8–1 g/kg, GBS 2 g/kg, PID 0.4–0.6 g/kg every 3–4 wk); **HBIG, VZIG, RIG, TIG (250–500 IU), anti-D**; palivizumab/nirsevimab (RSV).
- **Monoclonals in paediatrics**: rituximab (SDNS, ITP), tocilizumab/anakinra (sJIA), infliximab/adalimumab (IBD, JIA, Kawasaki), **emicizumab (haemophilia A)**, eculizumab (aHUS), dinutuximab (neuroblastoma), omalizumab/mepolizumab/dupilumab (asthma, atopic dermatitis), nusinersen (ASO – SMA), burosumab (XLH), denosumab.
- **Gene therapies**: onasemnogene (SMA), betibeglogene (β-thal), exa-cel (SCD/β-thal), elivaldogene (CALD), voretigene (RPE65 retinal dystrophy), etranacogene (haemophilia B).

26.7 Adverse Drug Reactions – Paediatric "Named" Syndromes

Reaction	Drug
Grey baby syndrome	**Chloramphenicol**
Gasping syndrome	**Benzyl alcohol**
Kernicterus	**Sulfonamides, ceftriaxone, vitamin K analogues (menadione)** in neonates
Reye syndrome	**Aspirin**
Red man syndrome	**Vancomycin (rapid infusion – histamine release)**
Bronze baby	**Phototherapy** with conjugated hyperbilirubinaemia
Blue baby / methaemoglobinaemia	**Nitrates (well water), dapsone, prilocaine (EMLA), benzocaine, methylene blue (in G6PD)**
Purple glove syndrome	**IV phenytoin**
Fetal hydantoin syndrome	Phenytoin in pregnancy

Reaction	Drug
SJS/TEN	**Carbamazepine, lamotrigine, phenytoin, phenobarbitone, sulfonamides (co-trimoxazole), nevirapine, allopurinol, NSAIDs (oxicams)**
DRESS	Anticonvulsants (aromatic), allopurinol, vancomycin, minocycline
Serum sickness-like reaction	**Cefaclor (MC in children)**, amoxicillin, co-trimoxazole
Neuroleptic malignant syndrome	Haloperidol, antipsychotics, metoclopramide
Acute dystonia (oculogyric crisis)	**Metoclopramide, prochlorperazine, haloperidol** – Rx diphenhydramine/benztropine
Hepatotoxicity	**Valproate (< 2 y), INH/rifampicin/PZA**, paracetamol overdose, ketoconazole
Ototoxicity	Aminoglycosides, loop diuretics, cisplatin, quinine
Cardiotoxicity	Anthracyclines (doxorubicin), cisapride (QT), tricyclics
Pancreatitis	**L-asparaginase**, valproate, steroids, azathioprine
Haemorrhagic cystitis	Cyclophosphamide, ifosfamide (mesna)
Pulmonary fibrosis	Bleomycin, busulfan, nitrofurantoin, amiodarone
Gingival hyperplasia	**Phenytoin, ciclosporin, nifedipine**
Hirsutism	Phenytoin, ciclosporin, minoxidil, steroids
Tooth staining	**Tetracyclines** (also oral iron syrup – reversible black)
Pyloric stenosis	**Erythromycin/azithromycin in neonates; prostaglandin E_1 (antral hyperplasia)**
Cortical hyperostosis	**Prostaglandin E_1 (long-term)**, hypervitaminosis A (Caffey disease – infantile cortical hyperostosis)
Neonatal withdrawal	Maternal opioids, SSRIs (PPHN), benzodiazepines

26.8 Poisoning & Overdose (pharmacology summary)

- Antidotes – see PALS chapter: **NAC (paracetamol), desferrioxamine (iron), naloxone (opioids), flumazenil (benzodiazepines), atropine + pralidoxime (organophosphates), physostigmine (anticholinergics/datura), methylene blue (methaemoglobin), digoxin Fab, sodium bicarbonate (TCA, salicylates), fomepizole/ethanol (methanol, ethylene glycol), hydroxocobalamin (cyanide), BAL/EDTA/succimer (lead), vitamin K (warfarin), protamine (heparin), glucagon (β-blocker), octreotide (sulfonylurea), calcium (Ca-channel blocker), Intralipid (local anaesthetic toxicity), dantrolene (malignant hyperthermia)**.
- **Single pill can kill a toddler**: **TCAs, calcium-channel blockers, β-blockers, sulfonylureas, opioids (methadone, buprenorphine), clonidine, antimalarials (chloroquine, quinine), camphor, methyl salicylate (oil of wintergreen), lindane, benzocaine, diphenoxylate-atropine (Lomotil)**.

26.9 Drugs in Lactation

Safe (compatible)	Use with caution	Contraindicated
Paracetamol, ibuprofen, penicillins, cephalosporins, macrolides, **heparin, warfarin**, insulin, metformin, **levothyroxine**, **propylthiouracil/ methimazole (low dose)**, prednisolone, most AEDs (carbamazepine, valproate, levetiracetam), **SSRIs (sertraline preferred)**, labetalol, nifedipine, enalapril, **anti-TB drugs**, **ART**, vaccines (except smallpox, yellow fever – caution)	**Phenobarbitone, lamotrigine, benzodiazepines (sedation)**, **lithium (monitor infant levels)**, metronidazole (single 2 g dose – withhold 12–24 h), codeine/ tramadol (avoid), amiodarone, aspirin (high dose), fluoroquinolones, chloramphenicol, sulfonamides (neonatal jaundice/ G6PD)	**Antineoplastics (methotrexate, cyclophosphamide, doxorubicin)**, **radioactive isotopes (I-131 – stop BF)**, **ergotamine, bromocriptine & cabergoline (suppress lactation)**, **gold salts**, retinoids (isotretinoin), **drugs of abuse (cocaine, heroin, amphetamines, PCP)**, **combined OCPs (↓ milk – avoid < 6 weeks)**, tetracyclines (prolonged), statins, mycophenolate

26.10 Drugs in Pregnancy – Teratogens (Fetal Effects)

Drug	Fetal effect
Thalidomide	**Phocomelia** (day 20–36)
Isotretinoin / vitamin A excess	**Craniofacial (microtia/anotia), CNS (hydrocephalus), cardiac (conotruncal), thymic** – retinoic acid embryopathy
Valproate	**NTD (spina bifida – 1–2%)**, fetal valproate syndrome, ↓ IQ, autism
Carbamazepine	NTD (spina bifida), craniofacial, fingernail hypoplasia
Phenytoin	**Fetal hydantoin syndrome – nail & distal phalangeal hypoplasia, cleft lip/palate, microcephaly, ID**, neuroblastoma, **neonatal VKDB**
Warfarin	**Fetal warfarin syndrome (6–9 wk) – nasal hypoplasia, stippled epiphyses (chondrodysplasia punctata)**, CNS haemorrhage (later)
Lithium	**Ebstein anomaly**
ACE inhibitors/ARBs	**2nd/3rd trimester – renal tubular dysgenesis, oligohydramnios, skull hypoplasia**
Tetracyclines	**Teeth discolouration, bone growth inhibition**
Aminoglycosides (streptomycin)	**Ototoxicity (8th nerve deafness)**
Methotrexate / aminopterin	Craniofacial & limb defects, abortion
Misoprostol	**Möbius sequence** (facial & abducens palsy), limb defects
Diethylstilbestrol (DES)	**Clear-cell adenocarcinoma of vagina**, uterine anomalies
Danazol / androgens	**Virilisation of female fetus**
Carbimazole/ methimazole	**Aplasia cutis**, choanal/oesophageal atresia
Alcohol	**Fetal alcohol syndrome – smooth philtrum, thin upper lip, short palpebral fissures, microcephaly, IUGR, ID, CHD (VSD)** – MC preventable cause of ID
Cocaine	Vascular disruption – limb defects, **placental abruption**, IUGR, microcephaly, NEC
Smoking	**IUGR/LBW (MC)**, preterm, SIDS, cleft lip
Statins	VACTERL-like (contraindicated)
Mycophenolate	Microtia, cleft lip/palate
NSAIDs (3rd trimester)	**Premature closure of ductus arteriosus (PPHN), oligohydramnios**

Drug	Fetal effect
SSRIs (late)	**PPHN**, neonatal adaptation syndrome; **paroxetine – CHD (1st trimester)**
Quinine (high dose)	Deafness, thrombocytopenia
Iodides / radioactive iodine	**Congenital goitre/hypothyroidism**
Fluconazole (high dose)	Antley–Bixler-like (craniosynostosis)

- **Safe in pregnancy**: **penicillins, cephalosporins, azithromycin, paracetamol, insulin, metformin, heparin/LMWH, labetalol, nifedipine, methyldopa, levothyroxine, PTU (1st trimester), folic acid, iron**, most vaccines (inactivated – **Tdap, influenza, Td, COVID**); **live vaccines contraindicated (MMR, varicella)**.

Practice MCQs · Pediatric Pharmacology

89 questions · correct option highlighted in green · cover the answer strip to self-test

Pharmacokinetics in Children

1. Fentanyl anesthesia (used as analgesia to provide pain relief during operative procedures in neonates) reduces the incidence of all the following postoperative conditions EXCEPT

A. acidosis
B. **tachycardia**
C. hypotension
D. coagulation abnormalities
E. intraventricular hemorrhage

Ans B Reduce the incidence of other postoperative conditions as bradycardia, hypoglycemia, and death.

2. All the following substances are poorly adsorbed by activated charcoal EXCEPT

A. iron
B. caustics
C. alcohols
D. hydrocarbons
E. **benzodiazepines**

Ans E Charcoal is "activated" via heating to extreme temperatures, creating an extensive network of pores that provides a very large adsorptive surface area. Many, but not all, toxins are adsorbed onto its surface, thus preventing absorption from the GI tract.

3. The most important manifestation of hydrocarbon toxicity is aspiration pneumonitis via inactivation of the type II pneumocytes and resulting surfactant deficiency. Of the following, the propensity of a hydrocarbon to cause aspiration pneumonitis is

A. inversely proportional to its viscosity
B. inversely proportional to its viscosity and volatility
C. directly proportional to its viscosity and volatility
D. **inversely proportional to its viscosity, and directly proportional to its volatility**
E. directly proportional to its viscosity, and inversely proportional to its volatility

Ans D With low viscosity and high volatility, such as mineral spirits, naphtha, kerosene, gasoline, and lamp oil, spread rapidly across surfaces and cover large areas of the lungs when aspirated.

4. In digoxin toxicity, Fab fragments bind free digoxin in both the intravascular and the interstitial space to form a pharmacologically inactive complex that is subsequently renally eliminated. All the following are indications of Fab fragments EXCEPT

A. renal failure
B. **hypertension**
C. ingestion >4 mg
D. K+ value >5-5.5 mEq/L
E. life threatening dysrhythmias

Ans B Clinically significant hypotension or other cardiovascular instability.

Drug Dosing

5. Postoperative nausea and vomiting (PONV) may be related to the stress, trauma of surgery, and pain. Of the following, the MOST likely drug that cause PONV is

A. ketorolac
B. droperidol
C. acetaminophen
D. **opioid analgesics**
E. local anesthetic agents

Ans D The etiology may be also related to the stress and trauma of surgery combined with the emetic effects of anesthetic agents. Pain is an important cause of nausea and vomiting.

6. Alkalinization of urine is MOST useful in managing toxicity with

A. iron
B. lead
C. **methotrexate**
D. phenothiazine
E. cyclophosphoamide

Ans C Urinary alkalinization (UA) enhances the elimination of some drugs that are weak acids by forming charged particles that are "trapped" within the renal tubules and thus excreted. Urinary alkalinization is accomplished via a continuous infusion of sodium bicarbonate–containing intravenous fluids, with a goal urine pH of 7.5-8. Alkalinization of the urine is most useful in managing salicylate and methotrexate toxicity.

7. Hydrocarbons with low viscosity and high volatility (kerosene, gasoline) spread rapidly across surfaces and cover large areas of the lungs when aspirated. The quantity of aspirated hydrocarbon chemicals that can produce significant injury is less than

A. **1 mL**
B. 3 mL
C. 5 mL
D. 7 mL
E. 10 mL

Ans A Only small quantities (<1 mL) of such chemicals need be aspirated to produce significant injury.

8. Iron-containing products remain widely available, with the most potentially toxic being adult iron preparations. Of the following, the severity of iron toxicity is MOSTLY related to the

A. age of child
B. time of presentation
C. type of salt preparation
D. number of tablets ingested
E. **amount of elemental iron ingested**

Ans E The severity of an exposure is related to the amount of elemental iron ingested. Ferrous sulfate contains 20% elemental iron, ferrous gluconate 12%, and ferrous fumarate 33%. Multivitamin preparations and children's vitamins rarely contain enough elemental iron to cause significant toxicity.

Antibiotics in Pediatrics

9. Postoperative nausea and vomiting (PONV) occurs in the immediate postoperative period, within the 1st 1-2 hr, or several hours after surgery and anesthesia. All the following may decrease the incidence of PONV EXCEPT

A. hydration
B. use of ketorolac
C. **preoperative fasting**
D. glucose supplementation
E. prophylactic use of ondansetron

Ans C does not decrease the incidence of nausea and vomiting.

10. All the following drugs toxicity can be removed by hemodialysis EXCEPT

A. **iron**
B. methanol
C. salicylates
D. theophylline
E. valproic acid

Ans A Toxins that are amenable to dialysis have the following properties: low volume of distribution (<1 L/kg), low molecular weight, low degree of protein binding, and high degree of water solubility.

11. Certain hydrocarbons have unique toxicities and can cause symptoms after ingestion, inhalation, or dermal exposures. Of the following, the MOST likely cause of acute myelogenous leukemia is long-term exposure to

A. gasoline
B. **benzene**
C. kerosene
D. nitrobenzene
E. methyl alcohol

Ans B is known to cause cancer, most commonly acute myelogenous leukemia, after long-term exposure.

12. The amount of elemental iron ingested that should be referred to medical care for evaluation is more than

A. 10 mg/kg
B. 20 mg/kg
C. 30 mg/kg
D. **40 mg/kg**
E. 50 mg/kg

Ans D Pediatric patients who ingest >40 mg/kg of elemental iron should be referred to medical care for evaluation, although moderate to severe toxicity is typically seen with ingestions of >60 mg/kg.

Analgesics & Antipyretics

13. Malignant hyperthermia is an acute hypermetabolic syndrome that is triggered by inhalational anesthetic agents and succinylcholine. Of the following, the MOST recognized clue to the risk of malignant hyperthermia is

A. hypercarbia
B. rapid onset of fever
C. ventricular fibrillation
D. acute hemoglobinuria
E. **rigid clenching of the masseter muscles**

Ans E Acute myoglobinuria associated with a malignant hyperthermia triggering agent is another clue. 5 (A).

14. Intralipid emulsions therapy (IET) act by squestering fat-soluble drugs and decreasing their impact at target organs thus enhance the elimination in case of drug toxicity. All the following can be eliminated by IET EXCEPT

A. diltiazem
B. verapamil
C. **amiodrone**
D. imipramine
E. amitriptyline

Ans C Lipophilic drugs are potentially bound by Intralipid emulsions, including calcium channel blockers (verapamil and diltiazem) and tricyclic antidepressants.

15. Of the following, the SINGLE acute toxic dose of acetaminophen is

A. ≥100 mg/kg
B. ≥150 mg/kg
C. **≥200 mg/kg**
D. ≥250 mg/kg
E. ≥300 mg/kg

Ans C In acetaminophen overdose, glutathione stores are overwhelmed, and free N- acetyl-p-benzoquinone imine is able to combine with hepatic macromolecules to produce hepatocellular necrosis.

16. In toxic ingestion of acetaminophen, the serum level should be measured after

A. 2 hr
B. **4 hr**
C. 6 hr
D. 8hr
E. 10 hr

Ans B Acetaminophen levels obtained <4 hr after ingestion, unless "nondetectable," are difficult to interpret and cannot be used to estimate the potential for toxicity.

17. When considering the treatment of a patient with acetaminophen poisoning, and after assessment of the ABCs, it is helpful to place the patient into one of 4 stages. Of the following, stage 1 is characterize by

A. **nausea and vomiting**
B. elevated alanine aminotransferase
C. high international normalized ratio
D. right upper quadrant abdominal pain
E. elevated aspartate aminotransferase

Ans A Other findings present in stage 2.

18. The King's College criteria for consideration of liver transplantation in acetaminophen poisoning with acute liver failure (stage 3) include all the following

A. coagulopathy (INR >6)
B. acidemia (serum pH <7.3)
C. **serum lactic acid >3 mmol/L**
D. grade III or IV hepatic encephalopathy
E. renal dysfunction (creatinine >3.4 mg/dL)

Ans C A serum lactic acid >3 mmol/L (after IV fluids) adds to both the sensitivity and specificity of the criteria to predict death without liver transplant. The degree of transaminase elevation is not a factor in this decision making process.

19. **In acetaminophen poisoning, when the initiation of N-Acetylcysteine therapy is delayed, there is the greater the risk of acute liver failure. N-Acetylcysteine should be instituted no later than**

A. 2 hr after ingestion
B. 4 hr after ingestion
C. 6 hr after ingestion
D. **8 hr after ingestion**
E. 10 hr after ingestion

Ans D The importance of instituting therapy with either IV or oral N-Acetylcysteine (NAC) no later than 8 hr from the time of ingestion cannot be overemphasized. No patient, no matter the size of the ingestion, who receives NAC within 8 hr of overdose, should die from liver failure. The further out from the 8 hr mark the initiation of therapy is delayed, the greater the risk of acute liver failure.

20. **Ibuprofen is well tolerated even in overdose. The acute dose of ibuprofen that result in more serious effects (altered mental status and metabolic acidosis) is**

A. >100 mg/kg
B. >200 mg/kg
C. >300 mg/kg
D. **>400 mg/kg**
E. >500 mg/kg

Ans D In children, acute doses of <200 mg/kg rarely cause toxicity, but ingestions of >400 mg/kg can produce more serious effects, including altered mental status and metabolic acidosis.

21. **In acute toxic ingestion of ibuprofen, symptoms usually develop within 4-6 hr of ingestion and resolve within 24 hr. All the following can result from acute ingestion of variable dose of ibuprofen EXCEPT**

A. **gastric ulcers**
B. CNS depression
C. abdominal pain
D. renal insufficiency
E. nausea and vomiting

Ans A GI bleeding and ulcers have been described with chronic use; they are rare in the setting of acute ingestion.

Antiepileptic Drugs

22. **Postanesthetic apnea is most common within the 1st 12 hr after surgery and has been reported in premature infants up to 48 hr later. General anesthesia should be avoided (except for emergency surgery) in full-term infants younger than postconceptual age of**

A. **44 wk**
B. 46 wk
C. 48 wk
D. 50 wk
E. 52 wk

Ans A General anesthesia should be avoided electively in full-term infants younger than a postconceptual age of 44 weeks because of the risk of postanesthetic apnea; ex-preterm infants carry this risk until an even later postconceptual age.

23. **All the following methods can be used to enhance the elimination of toxins EXCEPT**

A. hemodialysis
B. urinary alkalinization
C. **whole bowel irrigation**
D. intralipid emulsions therapy
E. multiple-dose activated charcoal

Ans C Enhancing elimination is a potentially lifesaving intervention that results in improved clearance of a poison that has already been absorbed. Whole bowel irrigation used for gastrointestinal decontamination.

24. **Methemoglobinemia may result from exposure to**

A. gasoline
B. benzene
C. kerosene
D. **nitrobenzene**
E. carbon monoxide

Ans D aniline, and related compounds can produce methemoglobinemia.

25. **A 2.5 year-old-boy presented with tachycardia, pallor, and fatigue. The history reveals excessive iron ingestion with unknown duration. Of the following, the MOST likely time of ingestion is**

A. 30 min-6hr ago
B. **6hr-24 hr ago**
C. 12hr-36 hr ago
D. 36hr-48hr ago
E. 48hr-72hr ago

Ans B The 2nd stage, 6-24 hr after ingestion, is often referred to as the "quiescent phase," as GI symptoms typically have resolved. However, careful clinical exam can reveal subtle signs of hypoperfusion, including tachycardia, pallor, and fatigue.

Asthma Medications

26. **Upper respiratory illness (URI) can increase airway reactivity for up to 6 wk in both normal children and children with a history of reactive airway disease. All the following should be taken in concern when preparing a child for operation with history of URI EXCEPT**

A. fever
B. injected sclerae
C. **clear rhinorrhea**
D. productive cough
E. increased mucous secretions

Ans C Purulent green nasal discharge.

27. **The acute toxic dose of salicylates is >150 mg/kg, more significant toxicity is seen after ingestions of >300 mg/kg and severe, potentially fatal, toxicity is described after ingestions of**

A. ≥350 mg/kg
B. ≥400 mg/kg
C. ≥450 mg/kg
D. **≥500 mg/kg**
E. ≥550 mg/kg

Ans D

28. **Dysrhythmias may result from exposure to**

A. benzene
B. kerosene
C. nitrobenzene
D. carbon tetrachloride
E. **halogenated hydrocarbons**

Ans E (which contain a chlorine, bromine, or fluorine), can sensitize the myocardium to the effects of endogenous catecholamines. This can result in dysrhythmias and "sudden sniffing death".

29. **Cardiovascular (CV) and CNS symptoms dominate the clinical presentation of tricyclic antidepressant (TCA) toxicity, symptoms typically develop within 1-2 hr of ingestion, and serious toxicity usually manifests within 6 hr of ingestion. Of the following, the MOST common CV manifestation is**

A. hypotension
B. **sinus tachycardia**
C. widened QRS interval
D. ventricular arrhythmias
E. prominent R wave in lead aVR

Ans B is the most common cardiovascular manifestation of toxicity; however, patients can develop widening of the QRS complex, premature ventricular contractions, and ventricular arrhythmias. Refractory hypotension is a poor prognostic indicator and is the most common cause of death in TCA overdose.

Corticosteroids

30. **After Upper respiratory illness (URI), it is generally recommended to avoid general anesthesia in elective procedures for**

A. 1-2Wk
B. 2-4Wk
C. **4-6Wk**
D. 6-8Wk
E. 8-10 wk

Ans C URIs can increase the risk of laryngospasm and bronchospasm, reduce mucociliary clearance, and raise the risk of intraoperative atelectasis and hypoxemia. It is generally recommended to avoid general anesthesia for elective procedures for 4-6 wk after a URI.

31. **The child with asthma should be free of wheezing for at least several days before surgery, even if this necessitates an increase in β-agonist dosage and the addition of steroids. Of the following, the MOST appropriate preoperative steroids regime in children with asthma (who are receiving asthma therapy or who have received steroid within the last year) is**

A. **prednisone, 1 mg/kg given 24 and 12 hr before surgery**
B. prednisone, 2 mg/kg given 24 and 12 hr before surgery
C. dexamethason, 0.6 mg/kg given 24 and 12 hr before surgery
D. hydrocortisone IV, 5 mg/kg given 24 and 12 hr before surgery
E. hydrocortisone IV, 10 mg/kg given 24 and 12 hr before surgery

Ans A

32. **In ambient air, the average half-life of HbCO is 4-6 hr, while after administration of 100% oxygen the half-life is**

A. 0.5-1 hr
B. **1-1.5 hr**
C. 1.5-2 hr
D. 2-3 hr
E. 3-4 hr

Ans B In ambient air, the average half-life of HbCO is 4-6 hr. This is dramatically reduced to 60-90 min by providing 100% oxygen at normal atmospheric pressures via a non-rebreather facemask. Severely poisoned patients might benefit from hyperbaric oxygen (HBO), which decreases the half-life of HbCO to 20-30 minutes.

33. **Asymptomatic child with tricyclic antidepressant toxicity, should be observed with continuous cardiac monitoring and serial ECGs for at least**

A. 4hr
B. **6hr**
C. 8hr
D. 10hr
E. 12hr

Ans B Children who remain completely asymptomatic with normal serial ECGs may be candidates for discharge after 6 hr of close observation.

Vaccines & Biologics

34. **Children with anterior mediastinal masses are at serious risk for airway compromise, cardiac tamponade, and vascular obstruction. The indication to prohibit general anesthesia is CT scan showing compression of the airway at the carina**

A. ≥30%
B. ≥40%
C. **≥50%**
D. ≥60%
E. ≥70%

Ans C

35. **In early course of acute salicylism, the MOST dominate lab values are**

A. metabolic acidosis and hypoglycemia
B. metabolic acidosis and hyperglycemia
C. respiratory alkalosis and hypoglycemia
D. metabolic acidosis and normoglycemia
E. **respiratory alkalosis and hyperglycemia**

Ans E Classically, lab values from a patient poisoned with salicylates reveal a primary respiratory alkalosis and a primary, elevated anion gap, metabolic acidosis. Early in the course of acute salicylism, respiratory alkalosis dominates. As the respiratory stimulation diminishes, the patient will move toward the metabolic acidosis.

36. **Clinical manifestations of organophosphate toxicity relate to the accumulation of acetylcholine at peripheral nicotinic and muscarinic synapses and in the CNS. All the following are symptoms of cholinergic excess at muscarinic receptors EXCEPT**

A. miosis
B. emesis
C. salivation
D. bradycardia
E. **urine retention**

Ans E A commonly used mnemonic for the symptoms of cholinergic excess at muscarinic receptors is DUMBBELS, which stands for diarrhea/defecation, urination, miosis, bronchorrhea/bronchospasm, bradycardia, emesis, lacrimation, and salivation.

37. **Probiotics are oral supplement or a food product that contains a sufficient number of viable microorganisms (live microorganisms that are most often referred to as "friendly" or "good" bacteria). Probiotics are indicated in all the following EXCEPT**

A. constipation
B. irritable bowel syndrome
C. **protien losing enteropathy**
D. antibiotic-associated diarrhea
E. inflammatory bowel disorders

Ans C Probiotics also indicated in Clostridium difficile–associated diarrhea.

Adverse Drug Reactions

38. **During general anesthesia in children with Down syndrome, the anesthesiologists should be aware of all the following EXCEPT**

A. macroglossia
B. cardiac anomalies
C. atlantoaxial instability
D. upper airway obstruction
E. **temporo-mandibular sublaxation**

Ans E In children with Down syndrome, it is wise to exercise caution in stabilizing the cervical spine and also to avoid cervical flexion and extension.

39. **A 2.5 year-old- child brought to emergency room immediately upon finding the boy with open bottle of some drugs (used by his grandfather). The child has vomited once, fussy, and lethargic. Physical evaluation reveals a 13-kg child who has tachypnea and tachycardia. Laboratory results include a blood sugar=225mg/dl, pH=7.5, PCO2=20 mEq/L, and HCO3=20 mEq/L. Of the following, the MOST likely offending drug is**

A. iron
B. atropine
C. **salicylate**
D. methylxanthine
E. tricyclic antideprssnt

Ans C Early in the course of acute salicylism, respiratory alkalosis dominates. As the respiratory stimulation diminishes, the patient will move toward the metabolic acidosis. Hyperglycemia (early) and hypoglycemia (late) have been described.

40. **All the following are done in the initial assessment of digoxin toxicity EXCEPT**

B. **serum calcium**
C. serum potassium
D. renal function test
E. serum digoxin level

Ans B The serum digoxin level should be assessed at least 6 hr after ingestion and carefully interpreted in the setting of clinical symptoms because the digoxin level alone does not entirely reflect the severity of intoxication. In acute ingestions, serum potassium is an independent marker of morbidity and mortality, with levels >5.5 mEq/L predicting poor outcomes. In chronic toxicity, serum potassium is less useful as a prognostic marker and may be altered due to concomitant use of diuretics.

41. **Dietary supplements (as herbs) are commonly used and may be helpful adjunctive treatments for common childhood problems. All the following matching are true EXCEPT**

A. **Aloe vera ---------- lice**
B. Ginger ---------- Nausea
C. Chamomile ---------- dyspepsia
D. Tea tree oil ---------- acne remedies
E. Peppermint -- Irritable bowel syndrome

Ans A Tea tree oil --- Anti-bacterial (acne remedies), pediculicide (lice), and Aloe vera - -- Mild burns.

Poisoning & Overdose

42. **Of the following, the MOST acceptable hematocrit value for routine elective anesthesia is**

A. ≥28%
B. **≥30%**
C. ≥32%
D. ≥34%
E. ≥36%

Ans B If there are reasons to expect significant blood loss or prolonged convalescence, anemia should be corrected preoperatively. In the emergency setting, transfusion may be required.

43. **Nystagmus is a sign of poisoning with all the following EXCEPT**

A. **cocain**
B. alcohols
C. ketamine
D. anticonvulsant
E. dextromethorphan

Ans A Cocaine causes mydriasis.

44. **According to the National Poison Data Systems of the American Association of Poison Control Centers, approximately 50% of poisoning occurs in children**

A. ≤2 yr old
B. ≤4 yr old
C. **≤6 yr old**
D. ≤8 yr old
E. ≤10 yr old

Ans C

45. **In poisoning, obtaining an accurate problem-oriented history is of paramount importance. All the following are historical features that suggest a possible diagnosis of poisoning in patients without a witnessed exposure EXCEPT**

A. low levels of household stress
B. multiple system organ dysfunction
C. sudden alteration of mental status
D. age of the child (toddler or adolescent)
E. acute onset of symptoms without prodrome

Ans A High levels of household stress should suggest a possible diagnosis of poisoning.

46. **Acetone odor is a sign of poisoning with all the following EXCEPT**

A. cyanide
B. methanol
C. salicylates
D. paraldehyde
E. isopropyl alcohol

Ans A Bitter almonds odor is a sign of poisoning with cyanide.

47. **Lacrimation is a sign of poisoning with**

A. atropin
B. salicylate
C. imipramin
D. acetaminophine
E. organophosphates

Ans E

48. **Mydriasis is a sign of poisoning with**

A. opioids
B. clonidine
C. hypnotics
D. organophosphates
E. tricyclic antidepressant

Ans E The other distracters may cause miosis.

49. **Constipation can be seen in poisoning with**

A. iron
B. lead
C. colchicine
D. cholinergics
E. antimicrobials

Ans B Antimicrobials, iron, cholinergics, and colchicines cause diarrhea.

50. **Hematemesis is a recognized finding in the following poisoning EXCEPT**

A. lead
B. iron
C. NSAIDs
D. caustics
E. salicylates

Ans A Lead may cause constipation.

51. **Which of the following poisoning may cause hyperglycemia?**

A. ethanol
B. quinine
C. caffeine
D. Beta blockers
E. salicylates (late)

Ans C Hypoglycemia is a sign of other distracters.

52. **All the following poisonings may cause peripheral neuropathy EXCEPT**

A. lead
B. arsenic
C. lithium
D. mercury
E. organophosphates

Ans C may cause delirium/psychosis and ataxia.

53. **All the following poisonings may have a radiopaque shadow on KUB EXCEPT**

A. lead
B. arsenic
C. salicylates
D. calcium carbonate
E. enteric-coated pills

Ans C

54. **Prolonged PR interval is a sign of poisoning with**

A. lithium
B. fluconazole
C. erythromycin
D. carbamazepine
E. diphenhydramine

Ans A Erythromycin and fluconazole may cause QTc prolongation while diphenhydramine and carbamazepine may cause QRS prolongation.

55. **Prolonged QTc is a sign of poisoning with**

A. amiodarone
B. carbamazepine
C. diphenhydramine
D. cardiac glycosides
E. tricyclic antidepressants

Ans A The other distracters may cause wide QRS.

56. **Antidotes are available for relatively few toxins, but early and appropriate use of an antidote is a key element in managing the poisoned patient. The following matching are true EXCEPT**

A. β Blockers ---------- glucagon
B. opioids ---------- naloxone
C. iron ---------- deferoxamine
D. lead ---------- BAL (dimercaprol)
E. isoniazid ---------- thiamine

Ans E Pyridoxine is antidote for isoniazid.

57. The following matching are true regarding poisoning and antidotes EXCEPT

A. acetaminophen ------ N-Acetylcysteine
B. anticholinergics --------- physostigmine
C. **benzodiazepines ---------- nalorphene**
D. carbon monoxide---------- 100% oxygen
E. organophosphates --------- pralidoxime

Ans C Flumazenil is antidote for benzodiazepines.

58. The majority of poisonings in children are from ingestion, although exposures can also occur via inhalational, dermal, and ocular route. All the following are true about decontamination EXCEPT

A. the goal is to minimize absorption of the toxic substance
B. **decontamination should be routinely employed for every poisoned patient**
C. water should not be used for decontamination after exposure to phosphorus
D. the efficacy of the intervention decreases with increasing time since exposure
E. the specific method, depends on the properties of the toxin and the route of exposure

Ans B Decontamination should not be routinely employed for every poisoned patient. Instead, careful decisions regarding the utility of decontamination should be made for each patient and should include consideration of the toxicity and pharmacologic properties of the exposure, the route of the exposure, the time since the exposure, and the risks vs the benefits of the decontamination method.

59. All the following poisonings may cause QTc prolongation EXCEPT

A. **lithium**
B. cisapride
C. amiodarone
D. antipsychotics
E. clarithromycin

Ans A may cause PR interval prolongation.

60. Dermal and ocular decontamination begin with removal of any contaminated clothing and particulate matter, followed by flushing of the affected area with tepid water or normal saline. Of the following, water should be used for dermal decontamination after exposure to

A. phosphorus
B. calcium oxide
C. **organophosphate**
D. elemental sodium
E. titanium tetrachloride

Ans C Dermal decontamination, especially after exposure to adherent or lipophilic (e.g., organophosphate) agents, should include thorough cleansing with soap and water. Water should not be used for decontamination after exposure to highly reactive agents, such as elemental sodium, phosphorus, calcium oxide, and titanium tetrachloride.

61. Gastrointestinal (GI) decontamination is a controversial topic among medical toxicologists and is most likely to be effective in the 1st hour after an acute ingestion. GI decontamination can be done even 1 hr after ingestion of the following EXCEPT

A. **antipsychotics**
B. anticholinergics
C. massive pill ingestions
D. enteric-coated salicylates
E. sustained-release preparations

Ans A Gastrointestinal decontamination strategies are most likely to be effective in the 1st hour after an acute ingestion. GI absorption may be delayed after ingestion of agents that slow GI motility (anticholinergic medications, opioids), massive pill ingestions, sustained-release preparations, and ingestions of agents that can form pharmacologic bezoars (e.g., enteric-coated salicylates).

62. The majority of poisonings in children are from ingestion, although exposures can also occur via inhalational, dermal, and ocular routes. All the following are described methods of GI decontamination EXCEPT

A. gastric lavage
B. whole-bowel irrigation
C. induced emesis with ipecac
D. single-dose activated charcoal
E. **multiple-dose activated charcoal**

Ans E Whereas single-dose activated charcoal is used as a method of decontamination, multiple doses of activated charcoal (MDACs) can help to enhance the elimination of some toxins.

63. After a review of the evidence and assessment of the risks and benefits of ipecac use, the American Academy of Clinical Toxicology have published statements that advise to

A. use ipecac with caution
B. use ipecac in all poisoning
C. **abandon the use of ipecac**
D. use ipecac in some poisoning
E. not use ipecac in persistently vomiting patient

Ans C

64. Although gastric lavage was used routinely for many years, in most clinical scenarios of poisoning, the use of gastric lavage is no longer recommended due to all the following EXCEPT

A. painful
B. time-consuming
C. **can induce tachycardia**
D. only removes a fraction of gastric contents
E. can delay administration of activated charcoal

Ans C Gastric lavage can induce bradycardia via a vagal response to tube placement.

65. All the following poisonings may cause QRS prolongation EXCEPT

A. **sotalol**
B. quinidine
C. chloroquine
D. propranolol
E. procainamide

Ans A may cause QTc prolongation.

66. **Hypocalcemia is a sign of poisoning with**

A. ethanol
B. **fluoride**
C. Beta Blockers
D. salicylates (late)
E. calcium channel blockers

Ans B

67. **All the following poisonings may cause ataxia EXCEPT**

A. alcohols
B. **imipramin**
C. anticonvulsant
D. carbon monoxide
E. dextromethorphan

Ans B CNS toxicity of antidepressant can include lethargy, coma, myoclonic jerks, and seizures.

68. **Wintergreen odor is a sign of poisoning with**

A. arsenic
B. thallium
C. selenium
D. **methyl salicylate**
E. organophosphates

Ans D Garlic odor is a sign of poisoning with other distracters.

69. **Salicylate ingestions are classified as acute or chronic, and acute toxicity is far more common in pediatric patients. Which of the following is not a recognized feature of salicylate poisoning?**

A. tinnitus
B. tachypnea
C. diaphoresis
D. **bradycardia**
E. hyperthermia

Ans D The tachycardia results in large part from marked insensible losses from vomiting, tachypnea, diaphoresis, and uncoupling of oxidative phosphorylation.

70. **Bradycardia is a sign of poisoning with**

A. antipsychotics
B. anticholinergics
C. antidepressants
D. methylxanthines
E. **calcium channel blockers**

Ans E Tachycardia is a sign of poisoning with other distracters.

71. **In salicylate poisoning, hyperglycemia (early) and hypoglycemia (late) have been described. Of the following, the MOST appropriate clue to administer IV glucose is**

A. vomiting
B. convulsion
C. palpitation
D. diaphoresis
E. **altered mental status**

Ans E Parenteral glucose should be provided in altered mental status as they may have CNS hypoglycemia not noted in a peripheral serum glucose test.

72. **Salivation is a sign of poisoning with all the following EXCEPT**

A. **mercury**
B. ketamine
C. salicylates
D. corrosives
E. organophosphates

Ans A lead, arsenic, and bismuth cause gum lines.

73. **All the following are clinical manifestations of hydrocarbons poisoning EXCEPT**

A. transient mild CNS depression
B. cough is the first clinical finding
C. chest radiographs may be initially normal
D. pneumatoceles can appear 2-3 wk after exposure
E. **fever and leukocytosis indicate bacterial superinfection**

Ans E Fever and leukocytosis are common accompanying signs in patients with pneumonitis and don't necessarily imply bacterial superinfection.

74. **Carbon monoxide (CO) is a colorless, odorless gas produced during the combustion of any carbon-containing fuel (the less efficient the combustion, the greater the amount of CO produced). All the following are true about CO poisoning EXCEPT**

A. CO displaces nitric oxide (NO) from proteins
B. NO is responsible for headache, syncope, and hypotension
C. **HbCO levels are well correlated with clinical signs of toxicity**
D. CO bind to cytochrome oxidase, disrupt cellular respiration
E. CO binds to hemoglobin with an affinity >200 times that of oxygen

Ans C CO displaces oxygen and creates a conformational change in hemoglobin that impairs the delivery of oxygen to the tissues, leading to tissue hypoxia. HbCO levels are not well correlated with clinical signs of toxicity, likely because CO interacts with multiple proteins in addition to hemoglobin.

75. **Early symptoms of carbon monoxide (CO) poisoning are nonspecific and include headache, malaise, nausea, vomiting, and the skin may appear**

A. blue
B. pale
C. normal
D. grayish
E. **cherry-red**

Ans E At higher exposure levels, patients can develop mental status changes, confusion, ataxia, syncope, tachycardia, and tachypnea. Severe poisoning is manifested by coma, seizures, myocardial ischemia, acidosis, cardiovascular collapse, and potentially death. On exam, patients might have cherry-red skin.

76. **All the following poisoning may cause QTc prolongation EXCEPT**

A. sotalol
B. **propranolol**
C. ketoconazole
D. erythromycin
E. phenothiazines

Ans B may cause QRS prolongation.

77. **Caustics include acids and alkalis as well as a few common oxidizing agents, strong acids and alkalis can produce severe injury even in small-volume ingestions. All the following are true about a caustic poisoning EXCEPT**

A. pH of <5 produce significant injury
B. pH of >12 produce significant injury
C. acids produce a coagulative necrosis
D. alkalis produce a liquefaction necrosis
E. prolong contact produce significant injury

Ans A The severity of the corrosive injury depends on the pH and concentration of the product as well as the length of contact time with the product. Agents with a pH of <2 or >12 are most likely to produce significant injury.

78. **Nicotinic signs and symptoms of organophosphates poisoning include all the following EXCEPT**

A. tachycardia
B. hypotension
C. fasciculation
D. hypoventilation
E. muscle weakness

Ans B Hypertension.

79. **All the following are true about treatment of organophosphate poisoning EXCEPT**

A. activated charcoal is unlikely to be of benefit
B. without treatment, symptoms can persist for weeks
C. atropine is primarily targeted to correct bradycardia
D. washing exposed skin and removing exposed clothing
E. pralidoxime breaks the bond between the enzyme and the organophosphate

Ans C Atropine dosing is primarily targeted to drying the respiratory secretions.

80. **All the following poisoning may cause hypoglycemia EXCEPT**

A. quinine
B. ethanol
C. sulfonylureas
D. beta Blockers
E. calcium channel blockers

Ans E Calcium channel blockers may cause hyperglycemia.

81. **Cyanosis (unresponsive to oxygen) is a sign of poisoning with**

A. atropine
B. amiodarone
C. vancomycin
D. carbon monoxide
E. elemental mercury

Ans B The other distracters may cause erythema.

82. **Iron is directly corrosive to the GI mucosa, leading to hematemesis, melena, ulceration, infarction, and potential perforation. Of the following, the MOST appropriate method of gastrointestinal decontamination in iron toxicity is**

A. induced emesis with ipecac
B. gastric lavage
C. activated charcoal
D. whole-bowel irrigation
E. all of the above

Ans D

83. **The MOST common cause of death in tricyclic antidepressant (TCA) overdose is**

A. seizures
B. renal injury
C. respiratory failure
D. refractory hypotension
E. ventricular arrhythmias

Ans D is a poor prognostic indicator and is the most common cause of death in TCA overdose.

Drug Safety in Lactation

84. **Preoperative psychologic preparation programs decrease the incidence of postoperative behavioral changes which last for up to 1 mo. Of the following, the MOST appropriate advice is to use oral**

A. diazepam
B. midazolam
C. imipramine
D. pentobarbital
E. chloral hydrate

Ans B Oral midazolam (0.5 mg/ kg) may decrease negative behavioral changes after surgery. Midazolam has the benefit of providing not only rapid-onset anxiolysis in 10-20 min but also very effective and rapid (10 min) amnesia.

85. **Chronic salicylism have an insidious presentation, and patients can show marked toxicity at significantly lower salicylate levels than in acute toxicity. Of the following, the MOST appropriate primary mode of therapy in chronic salicylism is**

A. IV glucose
B. hemodialysis
C. urinary alkalinization
D. volume resuscitation
E. gastric decontamination

Ans C enhances the elimination of salicylates by converting salicylate to its ionized form, "trapping" it in the renal tubules, and thus enhancing elimination.

86. **All the following matching are true EXCEPT**

A. salicylates ---------- sodium bicarbonate
B. organophosphates ---------- atropine
C. methemoglobinemia --------- methylene blue
D. calcium channel blockers ---------- insulin
E. tricyclic antidepressants ---------- flumazenil

Ans E Sodium bicarbonate is antidote for tricyclic antidepressants.

Drug Safety in Pregnancy

87. Aspiration of gastric contents is a perioperative disaster and, if superimposed on lung disease, may be rapidly fatal. All the following are true about the guidelines for preoperative fasting EXCEPT

A. clear, sweet liquids --- 2hr
B. breast milk ---------- 4hr
C. infant formula --------- 6hr
D. solid food ---------- 8hr
E. **gelatin ----------10hr**

Ans E Guidelines for preoperative fasting ("2-4-6-8 Rule"), fruit juices, gelatin ----- 6hr.

88. Of the following, the MOST important manifestation of hydrocarbons toxicity is

A. dysrhythmias
B. hepatic toxicity
C. intestinal necrosis
D. bacterial pneumonitis
E. **aspiration pneumonitis**

Ans E The most important manifestation of hydrocarbon toxicity is aspiration pneumonitis via inactivation of the type II pneumocytes and resulting surfactant deficiency.

89. Digoxin has a very narrow therapeutic index, therapeutic plasma digoxin concentrations are 0.5-2.0 ng/mL; a level >2 ng/mL is considered toxic and a level >6 ng/mL is considered potentially fatal. All the following medications increase serum digoxin concentration EXCEPT

A. **tetracyclin**
B. amiodarone
C. itraconazole
D. clarithromycin
E. spironolactone

Ans A Numerous drug interactions affect plasma digoxin concentrations. Medications known to increase serum digoxin concentrations include the macrolides, erythromycin and clarithromycin, spironolactone, verapamil, amiodarone, and itraconazole.

27

CHAPTER 27

Adolescent Medicine & Gynecology



WHAT THIS CHAPTER COVERS

- Adolescent Growth & Development
- Adolescent Nutrition
- Menstrual Disorders
- Polycystic Ovary Syndrome
- Sexually Transmitted Infections
- Contraception
- Teenage Pregnancy
- Substance Abuse in Adolescents
- Mental Health in Adolescents
- Eating Disorders in Adolescents
- Acne
- Gyenecological Examination

48 MCQs **12** subtopics

27.1 Adolescent Growth & Development

- **Adolescence (WHO): 10–19 years** – early (10–13), middle (14–16), late (17–19). India – **~253 million adolescents (21% of population)**; **Rashtriya Kishor Swasthya Karyakram (RKSK, 2014)** – 6 priorities: **nutrition, sexual & reproductive health, mental health, injuries & violence, substance misuse, non-communicable diseases**; Adolescent Friendly Health Clinics (AFHCs – Ujala clinics), peer educators (Saathiya), **Weekly Iron Folic Acid Supplementation (WIFS)**, menstrual hygiene scheme.
- **Psychosocial development**: early – concern with body changes, same-sex peers; middle – **peak peer influence, risk-taking, conflicts with parents, sexual identity**; late – **abstract thinking, identity, future orientation** (Erikson – identity vs role confusion; Piaget – formal operational stage).
- **HEADSSS assessment**: **H**ome, **E**ducation/employment, **A**ctivities, **D**rugs, **S**exuality, **S**uicide/depression, **S**afety – interview adolescent alone (confidentiality with limits).
- **Physical**: **growth spurt – girls earlier (PHV at SMR 2–3, ~11.5 y) than boys (SMR 3–4, ~13.5 y)**; **adolescent growth spurt accounts for ~20% of adult height & 50% of adult weight**; **feet & hands grow first ("distal → proximal")**, then limbs, then trunk (awkward phase); bone mass accrual – **peak bone mass by late teens/early 20s**.
- **Sequence**: girls – **thelarche (first) → pubarche → PHV → menarche (~2–2.5 y after thelarche, SMR 4)**; boys – **testicular enlargement (first) → pubarche → penile growth → PHV → voice change, axillary/facial hair → spermarche (~13.5 y, SMR 3)**.
- **Physiological gynaecomastia** (boys SMR 2–4, up to 65%) – resolves in 1–2 y; pathological if pre-pubertal, > 5 cm, rapidly growing, with small testes (Klinefelter), drugs (spironolactone, ketoconazole, cannabis, anabolic steroids), tumours (Leydig, hCG), liver disease, hyperthyroidism.
- **Adolescent BMI/obesity** – IAP charts; **adolescent hypertension (≥ 130/80 at ≥ 13 y)**.

27.2 Adolescent Nutrition

- **Increased requirements**: energy (boys > girls), **protein, calcium (1200–1300 mg/day), iron (↑ needs from menstrual losses & expanding red-cell mass)**, zinc, folate, vitamin D.
- **Iron-deficiency anaemia** – **MC nutritional problem in adolescent girls** (NFHS-5: ~59% of 15–19 y girls anaemic); **WIFS – 60 mg iron + 500 µg folic acid weekly (blue tablet) + albendazole biannually** (Anaemia Mukt Bharat).
- Unhealthy patterns: skipped meals (breakfast), fast food, sugary drinks, **dieting**, vegetarian B12 deficiency, **eating disorders** (see below), athletes (female athlete triad/RED-S).
- **Pregnant adolescent** – high nutritional needs (competition between mother & fetus).

27.3 Menstrual Disorders

- **Menarche**: mean **12.5–13 y** (India 12–14); **normal cycle 21–45 days in first 1–2 years** (anovulatory cycles common), bleeding ≤ 7 days, 20–80 mL; **"menses as a vital sign"**.

Disorder	Definition / causes	Management
Primary amenorrhoea	**No menarche by 15 y with secondary sexual characters, or by 13 y without breast development**, or no menses 3 y after thelarche	**With breast development, absent uterus → MRKH (Müllerian agenesis – 46,XX, normal ovaries, vaginal agenesis) or CAIS (46,XY, testes, scanty pubic hair); imperforate hymen/ transverse vaginal septum (cryptomenorrhoea – cyclical pain, haematocolpos, bulging bluish membrane); no breast development → ↑ FSH: Turner (MC), gonadal dysgenesis; ↓ FSH: CDGP, Kallmann, hypopituitarism, chronic illness, anorexia**

Disorder	Definition / causes	Management
Secondary amenorrhoea	Absence of menses ≥ 3 months (regular) or ≥ 6 months (irregular) after menarche	**Pregnancy (always exclude first – β-hCG)**, **PCOS (MC pathological)**, **functional hypothalamic (stress, weight loss, exercise, anorexia)**, hyperprolactinaemia, thyroid, POI, Asherman
Primary dysmenorrhoea	**MC gynaecological complaint in adolescents**; starts with ovulatory cycles (1–2 y after menarche); **prostaglandin-mediated**, lower abdominal cramps on day 1–2	**NSAIDs (mefenamic acid, ibuprofen, naproxen) – first-line, start 1–2 days before/at onset**; OCPs if refractory
Secondary dysmenorrhoea	Pelvic pathology – **endometriosis (MC cause of chronic pelvic pain unresponsive to NSAIDs/ OCPs)**, obstructive Müllerian anomalies, PID	Laparoscopy
Abnormal uterine bleeding (AUB)/ heavy menstrual bleeding	**Anovulatory (immature HPO axis) – MC**; **bleeding disorders (vWD – MC inherited; ITP) in up to 20%** – screen if heavy from menarche; PCOS, thyroid, pregnancy complications	Haemodynamic assessment; **combined OCPs/high-dose progestins**, tranexamic acid, iron; acute severe – IV conjugated oestrogen/high-dose OCP
Premenstrual syndrome / PMDD	Luteal-phase symptoms, resolve with menses	Lifestyle, SSRIs, OCPs
Mittelschmerz	Mid-cycle ovulatory pain	Reassure

27.4 Polycystic Ovary Syndrome (PCOS) in Adolescents

- **Diagnosis in adolescents (International guideline 2023): BOTH (1) irregular menstrual cycles (> 1 year post-menarche: < 21 or > 45 days; > 3 years: < 21 or > 35 days, or < 8 cycles/year; or no menses by 15 y / > 3 y after thelarche) AND (2) clinical/biochemical hyperandrogenism** (hirsutism – modified Ferriman–Gallwey score; severe acne; ↑ free testosterone); **pelvic USG/AMH NOT used for diagnosis < 8 years post-menarche** (multifollicular ovaries are normal in adolescence).
- **Exclude**: thyroid disease, hyperprolactinaemia, **non-classic CAH (17-OHP)**, Cushing, androgen-secreting tumours (rapid virilisation, testosterone > 200 ng/dL).
- Associations: **obesity, insulin resistance (acanthosis nigricans), T2DM, dyslipidaemia, NAFLD, OSA, depression/anxiety**, later infertility, endometrial hyperplasia.
- **Rx**: **lifestyle (weight loss 5–10%) – first line**; **combined OCPs** (menstrual regulation, hirsutism, acne); **metformin** (insulin resistance, obesity); anti-androgens (spironolactone – with contraception); cosmetic (laser) for hirsutism; screen for diabetes, depression.

27.5 Sexually Transmitted Infections & Sexual Health

- Adolescents – **high-risk** (biological – cervical ectropion; behavioural); counsel confidentially, **POCSO considerations (< 18 y – sexual activity is reportable in India)**.

Syndrome / STI	Organism	Key points (NACO syndromic kits)
Urethral discharge / cervicitis	**Chlamydia trachomatis (MC bacterial STI)**, **N. gonorrhoeae**	**Kit 1 (grey)**: **azithromycin 1 g + cefixime 400 mg** single dose
Vaginal discharge	Trichomonas, **bacterial vaginosis (clue cells, whiff test)**, candida	**Kit 2 (green)**: secnidazole + fluconazole

Syndrome / STI	Organism	Key points (NACO syndromic kits)
Genital ulcer – non-herpetic	**Syphilis (painless chancre)**, chancroid (painful – H. ducreyi), LGV, donovanosis	**Kit 3 (white)**: benzathine penicillin + azithromycin; Kit 4 (blue) – doxycycline + azithromycin (penicillin allergy)
Genital ulcer – herpetic	**HSV-2** (painful grouped vesicles)	**Kit 5 (red)**: acyclovir
Lower abdominal pain (PID)	Gonorrhoea, Chlamydia, anaerobes	**Kit 6 (yellow)**: cefixime + metronidazole + doxycycline
Inguinal bubo	LGV, chancroid	**Kit 7 (black)**: doxycycline + azithromycin
Genital warts	**HPV 6, 11**	Podophyllin, cryotherapy; **HPV vaccine (9–14 y – 2 doses)**

- **HIV in adolescents** – perinatally acquired survivors & new infections; adherence challenges; **PrEP** in high-risk.
- **PID complications**: tubal infertility, ectopic, chronic pain, **Fitz-Hugh–Curtis (perihepatitis)**, tubo-ovarian abscess.

27.6 Contraception for Adolescents

- **WHO MEC** – age alone is not a contraindication to any method (IUD/implants – MEC 2 for < 20 y).
- **LARC (long-acting reversible contraception – implants, IUDs) – most effective, first-line for adolescents (AAP/ACOG)**; **etonogestrel implant (Nexplanon – most effective reversible method, 3 y)**, LNG-IUS (5 y), Cu-T 380A (10 y).
- **Dual protection – condoms (only method preventing STIs/HIV) + another method**.
- **Combined OCPs** – also useful for dysmenorrhoea, acne, PCOS, AUB; c/i – migraine with aura, thrombophilia, uncontrolled HTN, < 6 weeks postpartum (breastfeeding).
- **DMPA injection (Antara – India; every 3 months)** – ↓ bone mineral density (reversible).
- **Emergency contraception**: **levonorgestrel 1.5 mg single dose within 72 h (up to 120 h, ↓ efficacy)**; **ulipristal acetate 30 mg up to 120 h**; **Cu-IUD within 5 days (most effective EC)**.
- India: **Mission Parivar Vikas**, Chhaya (centchroman – weekly, non-steroidal SERM).

27.7 Teenage Pregnancy

- **Adolescent pregnancy (< 20 y)** – **NFHS-5: 6.8% of women 15–19 y had begun childbearing**; **child marriage (girls < 18 y)** – PCMA 2006.
- **Maternal risks**: **anaemia, pre-eclampsia/eclampsia, preterm labour, obstructed labour (cephalopelvic disproportion – immature pelvis), obstetric fistula**, unsafe abortion, STIs, depression, school dropout; **maternal death – leading cause of death in 15–19 y girls globally**.
- **Neonatal risks**: **LBW (MC), prematurity, stillbirth, neonatal death**, child malnutrition.
- **Prevention**: delay marriage, girls' education, sexuality education, contraception access, MTP services.

27.8 Substance Use, Mental Health & Eating Disorders in Adolescents

- **Substance use**: **tobacco & alcohol (MC)**, cannabis, inhalants (whitener, glue – India street children), opioids, prescription drugs; **CRAFFT screen (Car, Relax, Alone, Forget, Friends, Trouble)**; **brief intervention, motivational interviewing, family therapy**; e-cigarettes banned in India (PECA 2019).
- **Mental health**: **depression & anxiety (MC)**, **suicide (leading cause of death 15–29 y in India)**, self-harm, ADHD, conduct disorder, psychosis onset, internet/gaming disorder, cyberbullying; screening – **PHQ-A**; Tele-MANAS **14416**.

- **Eating disorders** – **anorexia nervosa** (restriction, low weight, fear of fatness, bradycardia, amenorrhoea, osteoporosis; **refeeding syndrome – hypophosphataemia**), **bulimia nervosa** (normal weight, binge-purge, **Russell sign**, parotid swelling, dental erosion, hypokalaemic alkalosis); **family-based therapy** – see Behaviour chapter.
- **Female athlete triad / RED-S**: low energy availability, menstrual dysfunction, low bone density.

27.9 Acne & Skin in Adolescents

- **Acne vulgaris** – pilosebaceous unit; **4 factors – androgen-induced ↑ sebum, follicular hyperkeratinisation (comedone – earliest lesion), Cutibacterium acnes, inflammation**; **comedonal (open – blackhead, closed – whitehead) → papulopustular → nodulocystic**; mid-face & T-zone; **neonatal acne (maternal androgens; Malassezia – neonatal cephalic pustulosis)**, infantile acne (3–6 m – evaluate for hyperandrogenism if severe/ persistent).
- **Treatment (stepwise)**:

Severity	Treatment
Mild (comedonal)	**Topical retinoid (adapalene, tretinoin) ± benzoyl peroxide**
Mild–moderate papulopustular	**Topical retinoid + BPO ± topical antibiotic (clindamycin – always with BPO to prevent resistance)**, azelaic acid
Moderate	**Oral doxycycline/minocycline (≥ 8 y; 3 months) + topical retinoid + BPO**; **combined OCPs/ spironolactone in girls**
Severe nodulocystic / scarring / refractory	**Oral isotretinoin (0.5–1 mg/kg/day, cumulative 120–150 mg/kg)** – **teratogenic (iPLEDGE – 2 forms of contraception, monthly pregnancy test)**, cheilitis (MC), dry skin, ↑ lipids & LFTs, **depression/suicidality**, pseudotumor cerebri (**never with tetracyclines**), night blindness

- **Acne fulminans** – sudden severe ulcerating acne with fever, arthralgia (boys) – oral steroids + isotretinoin.
- **Pityriasis rosea, tinea versicolor, hyperhidrosis, striae, hirsutism** common adolescent concerns.

27.10 Paediatric & Adolescent Gynaecology

- **Examination**: in young girls – **frog-leg/knee-chest position**, external inspection; **no speculum in pre-pubertal girls** (EUA/vaginoscopy if needed); chaperone, consent/assent.
- **Vulvovaginitis in pre-pubertal girls – MC gynaecological problem**: **non-specific (poor hygiene, irritants – bubble baths, tight clothes; thin unoestrogenised mucosa, close anus)**; specific – **pinworm (Enterobius)**, GAS (beefy-red), Shigella (bloody), candida (rare pre-puberty unless diabetic/antibiotics/nappies), **foreign body (foul bloody discharge – toilet paper MC)**, **sexual abuse (gonorrhoea, Chlamydia, Trichomonas, HPV warts beyond infancy – evaluate)**; Rx hygiene, sitz baths, front-to-back wiping, cotton underwear.
- **Labial adhesions (synechiae)** – 3 m–6 y, hypoestrogenism, irritation; **topical oestrogen cream** if symptomatic; most resolve at puberty.
- **Lichen sclerosus** – white atrophic "figure-of-8" plaques; **ultrapotent topical steroid (clobetasol)**.
- **Imperforate hymen** – newborn mucocolpos or pubertal **haematocolpos (cyclical pain, primary amenorrhoea, bulging bluish membrane, urinary retention)**; Rx hymenotomy/hymenectomy.
- **Ovarian masses** – **functional cysts (MC)**, **mature cystic teratoma (MC ovarian neoplasm)**; **ovarian torsion** (sudden severe pain, vomiting – emergency detorsion; ovarian conservation); germ-cell malignancies (dysgerminoma – LDH, yolk-sac – AFP).
- **Sarcoma botryoides (embryonal rhabdomyosarcoma)** – grape-like vaginal mass in girls < 5 y.
- **Precocious & delayed puberty, PCOS, DSD** – see Endocrinology.
- **Menstrual hygiene**: sanitary pads (MHS scheme – ₹6 pack), menstrual cups; school WASH facilities.

Practice MCQs · Adolescent Medicine & Gynecology

48 questions · correct option highlighted in green · cover the answer strip to self-test

Adolescent Growth & Development

1. Adrenal production of androgen with development of underarm odor and faint genital hair (adrenarche) may occur as early as

A. 4 yr
B. 5 yr
C. **6 yr**
D. 7 yr
E. 8 yr

Ans C Adrenal production of androgen (chiefly dehydroepiandrosterone sulfate [DHEAS]) may occur as early as 6 yr of age, with development of underarm odor and faint genital hair (adrenarche). 2.(B), 3.(C), 4.(D), 5.(C). In males, the first visible sign of puberty and the hallmark of SMR 2 is testicular enlargement, beginning as early as 9.5 yr, followed by the development of pubic hair. This is followed by penile growth during SMR 3. Peak growth occurs when testis volumes reach approximately 9-10 cm3 during SMR 4.

2. In males, the first visible sign of puberty is testicular enlargement, beginning as early as

A. 8.5 yr
B. **9.5 yr**
C. 10.5 yr
D. 11.5 yr
E. 12.5 yr

Ans B Testicular enlargement, the first visible sign of male puberty (SMR/Tanner stage 2), can begin as early as 9.5 years of age.

3. The first visible sign of puberty in females is the appearance of breast buds (thelarche) between

A. 5 and 9 yr
B. 6and 10 yr
C. 7 and 11 yr
D. **8 and 12 yr**
E. 9 and 13 yr

Ans D Thelarche, the first visible sign of female puberty, typically begins between about 8 and 12 years of age.

4. Menses typically begins 2.5 yr after the onset of puberty at an average age of

A. 9.5 yr
B. 10.5 yr
C. 11.5 yr
D. **12.5 yr**
E. 13.5 yr

Ans D Menarche typically occurs about 2 to 2.5 years after the onset of puberty, at an average age of around 12.5 years.

5. The gold standard diagnostic investigation for precocious puberty is

A. Tanner maturating rate
B. left wrist x-ray
C. serum luteinizing hormone levels
D. **gonadotropins measurement**
E. brain MRI

Ans D Measurement of gonadotropins after gonadotropin-releasing hormone or gonadotropin-releasing hormone–agonist stimulation.

Adolescent Nutrition

6. Penile growth occur during SMR

A. 1
B. 2
C. **3**
D. 4
E. 5

Ans C Penile growth begins during SMR (Tanner) stage 3, after testicular enlargement has already started in stage 2.

7. A 15-year-old female presented with delusions, paranoia, tachycardia, hypertension, hyperpyrexia, diaphoresis, piloerection, mydriasis, and hyperreflexia, later on she developed seizure, hypotension, and dysrhythmia. Of the following, the MOST likely cause is

A. antidepressant agents
B. **amphetamine**
C. barbiturates
D. benzodiazepines
E. edrophonium

Ans B Patient in the question presented with sympathomimetic syndrome and this syndrome can be caused by cocaine, amphetamine, methamphetamine, and OTC decongestants (phenylpropanolamine, ephedrine, and pseudoephedrine). In caffeine and theophylline overdoses, similar findings, except for the organic psychiatric signs, result from catecholamine release.

8. The MOST common cause of breast pain in adolescents is

A. benign fibroadenoma
B. **exercise**
C. mastitis
D. lipoma
E. Intraductal papilloma

Ans B Physiologic swelling and tenderness occur on a cyclic basis, most commonly during the premenstrual phase, and are secondary to hormonal stimulation and resulting proliferative changes. Hormonal imbalance can cause exaggerated responses in the breast tissue, especially in the upper and outer quadrants. Nodularity, poorly localized tenderness, and a soreness radiating to the axilla and arm are usual accompanying findings.

9. The tumor marker (α-fetoprotein) is used for the diagnosis of all the following ovarian cancers EXCEPT

A. Immature teratoma
B. embryonal carcinoma
C. mixed germ cell
D. **dysgerminoma**
E. endodermal sinus tumor

Ans D In dysgerminoma use E2, estradiol, hCG (human chorionicgonadotropin), and LDH (lactate dehydrogenase) as a tumor markers.

Menstrual Disorders

10. Sperm may be found in the urine and nocturnal emissions may be noted at SMR

A. 1
B. 2
C. **3**
D. 4
E. 5

Ans C Spermarche (first appearance of sperm, detectable in urine) and nocturnal emissions typically occur around SMR (Tanner) stage 3.

11. Evaluation for pubertal delay in female should be done if she lacks any pubertal signs by the age of

A. 12 yr
B. **13 yr**
C. 14 yr
D. 15 yr
E. 16 yr

Ans B 20.(D), 21.(D). Age 15 yr is commonly considered the age at which an evaluation for primary amenorrhea should be undertaken. Evaluation should begin sooner if 4 yr have elapsed since the onset of puberty (breast development in most females).

12. The age at which an evaluation for primary amenorrhea should be undertaken is

A. 12 yr
B. 13 yr
C. 14 yr
D. 15 yr
E. **16 yr**

Ans E Classic teaching defines evaluation for primary amenorrhea as warranted by age 16 if no menarche has occurred, even with otherwise normal pubertal development (more recent guidance suggests earlier evaluation at 15).

13. Primary amenorrhea generally requires evaluation, if menstruation does not occur within

A. 1 yr from the onset of puberty
B. 2 yr from the onset of puberty
C. **3 yr from the onset of puberty**
D. 4 yr from the onset of puberty
E. 5 yr from the onset of puberty

Ans C Primary amenorrhea evaluation is generally undertaken if menstruation has not begun within about 3 years of the onset of puberty (thelarche).

Polycystic Ovary Syndrome

14. The belief that major structural brain development is completed in childhood is outdated. It is now clear that neuromaturation continues into the

A. 2nd decade
B. **3rd decade**
C. 4th decade
D. 5th decade
E. 6th decade

Ans B This maturation is characterized by decreases in gray matter, increases in white matter, and an apparent increase in the efficiency of communication and connectivity between different brain regions.

15. The least likely cause of Infectious vulvovaginitis is

A. Escherichia coli
B. Staphylococcus aurous
C. Haemophilus influenza
D. Enterobiusvermicularis
E. **Candida spp.**

Ans E

16. Of the following, the MOST common solid mass seen in adolescent girls is

A. lipoma
B. **fibroadenoma**
C. lymphangioma
D. hematoma
E. hamartoma

Ans B

17. Pathogenesis of leiomyosarcoma is thought to be correlated with

A. **Epstein-Barr virus**
B. haemophilusinfluenzae
C. enterococcus
D. nocardia
E. Yersinia

Ans A

Sexually Transmitted Infections

18. Peak growth occurs during SMR 4 when testis volumes reach approximately

A. 3-4 cm3
B. 5-6 cm3
C. 7-8 cm3
D. **9-10 cm3**
E. 11-12 cm3

Ans D Peak height velocity in boys occurs during SMR stage 4, when testicular volume reaches approximately 9-10 cm3.

19. Urethritis is a sexually transmitted infections syndrome characterized by inflammation of the urethra, usually caused by an infectious etiology. All of the following are true EXCEPT

A. urethritis may present with meatal pruritus
B. approximately 30-50% of males are asymptomatic
C. **on examination, the classic finding is erythema of the urethral meatus**
D. N. gonorrhoeae is one of the most commonly identified pathogen
E. noninfectious causes of urethritis include urethral trauma or foreign body

Ans C On examination, the classic finding is mucoid or purulent discharge from the urethral meatus. If no discharge is evident on exam, providers may attempt to express discharge by applying gentle pressure to the urethra from the base distally to the meatus 3-4 times.

20. Which of the following is TRUE regarding pubertal gynecomastia?

A. occurring in up to 20% of normal adolescent males
B. onset typically is between 7 and 9 yr
C. usually regresses within 6 mo
D. **surgery may be indicated in severe or persistent cases**
E. medical therapies have been approved for use in adolescents

Ans D Occurring in up to 60% of normal adolescent males, onset typically is between 10 and 13 yr, usually regresses within 18-24 mo, no medical therapies for gynecomastia have been approved for use in adolescents.

21. Depo-provera: an injectable progestin, medroxyprogesterone acetate available as a deep intramuscular injection (150 mg) or as a subcutaneous injection (104 mg) with typicaluse, the failure rates is 6%. Potential adverse effect include

A. heavy menstrual bleeding
B. dysmenorrhea
C. acne
D. weight gain
E. **osteoporosis**

Ans E After 1 yr of use, 50% of users develop amenorrhea, which may be an added advantage for teens with heavy menstrual bleeding, dysmenorrhea, anemias, or blood dyscrasias. Although concern has been directed toward the potential for loss in bone mineral density in adolescents, thereby potentially increasing their risk for osteoporosis later in life, subsequent studies have found that bone density is recovered after discontinuation of the method and is considered safe for use in this population.

22. Amastia (complete absence of the breast) associated with all the following EXCEPT

A. Poland syndrome
B. ectodermal dysplasia
C. Crohn disease
D. **congenital adrenal hypoplasia**
E. hypogonadotropic hypogonadism

Ans D Congenital adrenal hyperplasia

Contraception

23. In assessing the seriousness of adolescent drug abuse score, which of the following take +2 in the score?

A. female sex
B. age >15 years
C. positive family history of drug abuse
D. **use before driving**
E. use of hallucinogens

Ans D The score consist of 0, +1 and +2. +2 include : use of drugs alone not in group, sad before drug use, recently poor school performance, use drugs before driving, history of accident, use drugs before and during school day and the use of Whiskey, opiates, cocaine, barbiturates.

24. The MOST commonly used method for contraception is

A. **condom**
B. withdrawal
C. oral contraceptive pill
D. spermicides
E. intrauterine device

Ans A The most commonly used method is the condom, followed by withdrawal and then the pill.

25. The contraceptive method with the highest failure rate even with perfect use is

A. progestin releasing intra uterine device
B. progestin-only injection
C. combined oral contraceptives
D. male condom
E. **spermicides**

Ans E Failure rate are as follow: Progestin releasing intra uterine device 0.2%, progestin-only injection 0.2%, combined oral contraceptives 0.3%, male condom 2%, spermicides 18%.

26. Approved treatment for condyloma acuminate of the vulva in children include all the following EXCEPT

A. topical trichloroacetic acid
B. local cryotherapy
C. electro cautery
D. laser ablation
E. **sinecatechins ointment**

Ans E Some products used to treat skin lesions in adults have not been approved for children, including provider application of podophyllin resin and home application of imiquimod, podofilox, and sinecatechins ointment.

Teenage Pregnancy

27. Which of the following scores regarded as serious according to the assessment of seriousness of adolescent drug abuse score?

A. 0-3
B. **3-8**
C. 8-13
D. 13-18
E. 18-23

Ans B 0-3 less worrisome, 3-8 serious and 8-18 very serious.

28. **Amoxicillin-clavulanate is the first-line treatment in vulvovaginal infections caused by**

A. Streptococcus pyogenes
B. **Staphylococcus aurous**
C. Haemophilus influenza
D. Shigella
E. Chlamydia trachomatis

Ans B In cases of treatment failure in H influenza or non-encapsulated H. influenzae, amoxicillin–clavulanate is recommended.

29. **The MOST common tumor metastasized to the breast is**

A. **rhabdomyosarcoma**
B. lymphoblastic leukemia
C. neuroblastoma
D. lymphoma
E. cystosarcomaphyllodes

Ans A Breast tumors also may be the first manifestation of relapse (extra medullary) in acute lymphoblastic leukemia.

30. **Hemihematometra is described as**

A. accumulation of mucus or nonsanguineous fluid in the vagina
B. **atretic segment of vagina with menstrual fluid accumulation**
C. accumulation of serous fluid in the fallopian tube
D. one cervix associated with 2 uterine horns
E. the result of failure of 1 müllerian duct to descend

Ans B

Substance Abuse in Adolescents

31. **A 13-years-old male presented with delirium and mumbling speech, tachycardia, dry flushed skin, dilated pupils, myoclonus, slightly elevated temperature, urinary retention, and decreased bowel sounds, later on he developed seizure and dysrhythmia. Of the following, the MOST likely cause is**

A. **antidepressant agents**
B. amphetamine
C. barbiturates
D. benzodiazepines
E. edrophonium

Ans A Patient in the question presented with anticholinergic syndrome and this syndrome can be caused by antihistamines, antiparkinsonian medication, atropine, scopolamine, amantadine, antipsychotic agents, antidepressant agents, antispasmodic agents, mydriatic agents, skeletal muscle relaxants, and many plants (notably jimson weed and Amanita muscaria).

32. **Ultra potent topical corticosteroids is the first-line therapy in**

A. labial agglutination
B. **lichen sclerosus**
C. psoriasis
D. atopic dermatitis
E. seborrheic dermatitis

Ans B

33. **The imaging modality of choice for breast abnormalities in the pediatric population is**

A. mammography
B. CT scan
C. **ultrasonography**
E. PET scan

Ans C Because the dense breast tissue of the adolescent obstructs the visualization of a palpable mass, mammography is not advised for this age group.

34. **The MOST sensitive and specific imaging technique used for evaluating müllerian anomalies is**

A. ultrasound
B. hysterosalpingogram
C. sonohysterography (saline-infusion sonography)
D. **MRI**
E. CT scan

Ans D Because it can image nearly all reproductive structures, blood flow, external contours, junctional zone resolution on T2-weighted images, and associated renal and other anomalies. MRI also has a high correlation with surgical findings because of its multiplanar capabilities and high spatial resolution.

Mental Health in Adolescents

35. **Alcohol acts primarily as a central nervous system depressant. It produces all the following EXCEPT**

A. euphoria
B. impaired short-term memory
C. increased pain threshold
D. **hyperthermia**
E. respiratory depression

Ans D Hypothermia.

36. **All the following are first-line treatment of diaper dermatitis EXCEPT**

A. frequent diaper changes
B. increasing diaper free period
C. frequent bathing
D. **antifungal treatment**
E. zinc oxide

Ans D If diaper dermatitis persists after these conservative measures, or if the classic satellite lesions of candida are present, treatment with an antifungal can decrease the inflammation.

37. **All the following may cause hirsutism EXCEPT**

A. hydrochlorothiazide
B. acetazolamide
C. **cyclophosphamide**
D. anabolic steroids
E. penicillamine

Ans C Cause alopecia.

38. **The MOST common structural uterine anomaly is**

A. **uterine septum**
B. bicornuate uterus
C. unicornuate uterus
D. uterine didelphys
E. arcuate uterus

Ans A

Eating Disorders in Adolescents

39. Smoking during pregnancy is associated with an average decrease in fetal weight by

A. 100 g
B. **200 g**
C. 300 g
D. 400 g
E. 500 g

Ans B Smoking during pregnancy is associated with an average decrease in fetal weight of 200 g; this decrease, added to the already smaller size of infants born to teenagers, increases perinatal morbidity and mortality.

40. Vulvar dermatologic condition in children which necessitate evaluation of thyroid function is

A. lichen sclerosus
B. psoriasis
C. **vitiligo**
D. seborrheic dermatitis
E. atopic dermatitis

Ans C Although diagnosis is clinical, there is an association with other autoimmune or endocrine disorders (hypothyroidism, Graves' disease, Addison disease, pernicious anemia, and insulin-dependent diabetes mellitus) and workup should include evaluation for at least thyroid dysfunction.

41. Chocolate cysts are seen in

A. functional cysts
B. **endometriomas**
C. cyst adenomas
D. teratomas
E. ovarian carcinoma

Ans B

42. An imperforate hymen is characterized by all the following EXCEPT

A. the incidence is approximately 1 in 1,000
B. normal secondary sex characters
C. more often it is diagnosed at the time of menarche
D. **cannot be diagnosed In the newborn period**
E. primary amenorrhea

Ans D In the newborn period and early infancy, it may be diagnosed by a bulging membrane caused by a mucocolpos from maternal estrogen stimulation of the vaginal mucosa.

Acne

43. Exposure to smokeless tobacco increases the users risk for

A. lipoid pneumonia
B. chronic cough
C. **cancers of the esophagus**
D. irritability
E. decreased concentration

Ans C The 2 forms of smokeless tobacco (SLT) are “chew,” a leafy tobacco product sold in pouches, and “snuff,” a finely ground tobacco product sold in tins or packets. Exposure to SLT increases the users risk for oral cancers of the mouth, pharynx, larynx, and esophagus, as well as gum disease and nicotine addiction.

44. Lack of development of the breast is considered delayed and warrants endocrinology evaluation by the age of

A. 10 yr
B. 11 yr
C. 12 yr
D. **13 yr**
E. 14 yr

Ans D

45. 5-yr survival is 100% in the following germ cell tumors

A. **gonadoblastoma**
B. embryonal carcinoma
C. choriocarcinoma
D. dysgerminoma
E. endodermal sinus tumor

Ans A

Gyenecological Examination

46. Lysergic acid diethylamide (LSD) is a very potent hallucinogen that is made from lysergic acid found in ergot, a fungus that grows on rye and other grains. All the following are common somatic symptoms EXCEPT

A. dizziness
B. **miosis**
C. nausea
D. flushing
E. hyperthermia

Ans B Mydriasis.

47. Of the following, the MOST likely cause of bloody nipple discharge in infants is

A. chronic nipple irritation
B. ducts of Montgomery
C. hypothalamic tumors
D. Intraductal cysts
E. **mammary duct ectasia**

Ans E

48. The MOST common ovarian malignancy is

A. **gonadoblastoma**
B. embryonal carcinoma
C. choriocarcinoma
D. dysgerminoma
E. endodermal sinus tumor

Ans A

28

CHAPTER 28

Disorders of the Eye, Ear & Skin



WHAT THIS CHAPTER COVERS

- Conjunctivitis
- Ophthalmia Neonatorum
- Cataract
- Retinopathy of Prematurity
- Retinoblastoma
- Refractive Errors
- Otitis Externa
- Otitis Media
- Hearing Loss
- Eczema & Atopic Dermatitis
- Seborrheic Dermatitis
- Diaper Dermatitis
- Bacterial Skin Infections
- Viral Skin Infections
- Fungal Skin Infections
- Parasitic Skin Infections
- Birth Marks & Hemangiomas
- Acne
- Vitiligo & Pigmentary Disorders
- Urticaria & Angioedema
- Psoriasis
- Ichthyosis

181 MCQs **23** subtopics

28.1 Paediatric Ophthalmology – Essentials

- **Red reflex test (Brückner)** at birth & every visit – **leukocoria** (retinoblastoma, cataract, ROP, Coats, PHPV, toxocara); asymmetric reflex – strabismus, anisometropia.
- **Visual milestones** – see Development (6/6 by 5–6 y). **Amblyopia** – reduced vision from abnormal visual experience in critical period (< 7–8 y): **strabismic (MC), anisometropic, deprivation (cataract, ptosis – worst)**; Rx **correct cause + occlusion (patching) of the better eye/atropine penalisation – most effective before 7 y**.
- **Strabismus**: **pseudo-strabismus (broad epicanthus – MC cause of apparent squint)** – corneal light reflex symmetric (**Hirschberg test**); **cover test** – true squint; **any squint after 3–6 months needs evaluation**; **infantile esotropia** (surgery 6–24 m); **accommodative esotropia (2–3 y, hypermetropia – glasses)**; always rule out retinoblastoma/cataract (sensory strabismus).

28.2 Conjunctivitis & Ophthalmia Neonatorum

Ophthalmia neonatorum (conjunctivitis in first 28 days)

Cause	Onset	Features	Treatment
Chemical (silver nitrate)	**Within 24 h**	Mild, transient	None (self-limiting)
N. gonorrhoeae	**2–5 days**	**Hyperacute, profuse purulent discharge, chemosis, lid oedema → corneal ulceration/ perforation (emergency)**	**Single-dose IV/IM ceftriaxone 25–50 mg/kg (max 125 mg)** + frequent saline irrigation; treat parents
Chlamydia trachomatis (MC infective cause in developed countries)	**5–14 days**	Mild–moderate mucopurulent, **pseudomembrane**, **no follicles (neonates lack lymphoid tissue)**; **pneumonia (afebrile, staccato cough at 2–12 wk)**	**Oral erythromycin 50 mg/kg/day × 14 days** (or azithromycin 20 mg/ kg × 3 d) – systemic therapy (topical insufficient)
Other bacteria (S. aureus, Streptococcus, Haemophilus, Pseudomonas)	5–14 days	Purulent	Topical antibiotics
HSV	**6–14 days**	Watery, **vesicles on lids, dendritic keratitis**	**IV acyclovir + topical antiviral** (systemic disease risk)

- **Prophylaxis (Credé's)**: historically **1% silver nitrate**; now **0.5% erythromycin or 1% tetracycline ointment**, povidone-iodine 2.5% – within 1 h of birth.
- **Childhood conjunctivitis**: **bacterial (H. influenzae – with otitis media "conjunctivitis-otitis syndrome", S. pneumoniae)** – purulent, glued eyelids – topical antibiotics; **viral (adenovirus – pharyngoconjunctival fever, epidemic keratoconjunctivitis; preauricular nodes)**; **allergic – vernal keratoconjunctivitis (VKC – boys, hot climates, intense itching, ropy discharge, "cobblestone" giant papillae on upper tarsus, Horner–Trantas dots, shield ulcer)** – Rx mast-cell stabilisers/antihistamines (olopatadine), steroids, ciclosporin/tacrolimus.
- **Trachoma** (C. trachomatis A–C): **follicles & papillae (upper tarsus), Herbert's pits, Arlt's line, trichiasis, corneal opacity**; **SAFE strategy**; India declared trachoma-free as public health problem (2024 WHO validation) – azithromycin.

28.3 Paediatric Cataract & Glaucoma

- **Congenital/infantile cataract** – **MC treatable cause of childhood blindness**; **causes**: idiopathic (MC – ~50%), hereditary (AD – MC familial), **intrauterine infections (rubella – "pearly" nuclear cataract; others)**, **metabolic – galactosemia ("oil-drop"), hypocalcaemia, hypoglycaemia**, chromosomal (Down, trisomy 13, 18), **Lowe**

syndrome, Alport (anterior lenticonus), **steroids (posterior subcapsular)**, trauma (child abuse), radiation, prematurity (transient), Wilson (sunflower), myotonic dystrophy (Christmas-tree).

- **Leukocoria, absent/abnormal red reflex, nystagmus (late – after 2–3 months indicates poor prognosis), strabismus, photophobia**.
- **Rx: urgent surgery – dense unilateral by 4–6 weeks, bilateral by 6–10 weeks (before 8 weeks – critical period)**; **lensectomy + anterior vitrectomy (± primary posterior capsulotomy)**; **IOL after 1–2 years (aphakic contact lenses/glasses in infancy)**; amblyopia therapy; **complication – glaucoma (aphakic, MC long-term), posterior capsule opacification (visual axis opacification)**.
- **Congenital glaucoma (buphthalmos)**: **trabeculodysgenesis**, CYP1B1 (AR); **triad – epiphora (tearing), photophobia, blepharospasm** + **corneal enlargement (> 12 mm), corneal haze/Haab striae (Descemet tears)**, ↑ IOP, cupping; associations – **Sturge–Weber, NF-1, Lowe, aniridia, rubella, Axenfeld–Rieger**; **Rx surgical – goniotomy/trabeculotomy (first-line; medicines only temporising)**. Differentiate from **nasolacrimal duct obstruction** (tearing without photophobia/corneal enlargement).

28.4 Retinopathy of Prematurity (ROP)

- Vasoproliferative retinopathy of premature infants (retinal vascularisation completes at ~40 weeks – temporal periphery last).
- **Risk factors**: **low gestational age & birth weight (most important)**, **unmonitored/high oxygen therapy**, fluctuating oxygen, sepsis, blood transfusion, IVH, poor weight gain.
- **Screening (India – RBSK/NNF 2018)**: **all babies < 34 weeks GA and/or < 2000 g BW**, and 34–36 wk with risk factors (sick, ventilated, O_2); **first exam at 2–4 weeks postnatal age (by day 20–30; < 28 wk/< 1200 g – at 2–3 wk)**; indirect ophthalmoscopy with scleral depression.
- **ICROP3 (2021) classification**: **zone I (posterior pole – radius twice disc-to-fovea distance)**, zone II, zone III (temporal crescent); **stages 1 – demarcation line, 2 – ridge, 3 – extraretinal fibrovascular proliferation, 4 – partial retinal detachment (4A extrafoveal, 4B foveal), 5 – total detachment (leukocoria)**; **plus disease (dilated tortuous posterior pole vessels – ≥ 2 quadrants)**, **A-ROP (aggressive ROP – formerly AP-ROP)**.
- **Treatment indications (type 1 ROP – ETROP)**: **zone I – any stage with plus; zone I – stage 3 without plus; zone II – stage 2 or 3 with plus**; **A-ROP**. Treat within **48–72 h**.
- **Rx**: **laser photocoagulation of avascular retina (gold standard)**, **intravitreal anti-VEGF (bevacizumab, ranibizumab) – zone I/A-ROP** (late recurrences – longer follow-up); vitrectomy/scleral buckle for stage 4–5.
- **Prevention**: **targeted SpO_2 90–95% in preterm on oxygen**, avoid fluctuations, breast milk, infection control.
- Long-term: myopia (MC), strabismus, amblyopia, retinal detachment, glaucoma.

28.5 Retinoblastoma (eye aspects)

- See Tumours: **leukocoria (MC), strabismus (2nd)**; RB1 13q14; calcification on USG/CT; **MRI for optic nerve**; **no biopsy**; chemoreduction, focal therapy, intra-arterial chemo, enucleation.

28.6 Refractive Errors & Other Eye Conditions

Condition	Key points
Refractive errors	**MC cause of visual impairment in school children**; **myopia – MC refractive error in school-age (rising – near work, ↓ outdoor time)**; **newborns are hypermetropic (+2 to +3 D)** → emmetropisation; school vision screening (RBSK/NPCB&VI); **low-dose atropine (0.01–0.05%)** & outdoor activity (≥ 2 h/day) slow myopia progression
Nasolacrimal duct obstruction	Membrane at **valve of Hasner**; epiphora, mucoid discharge; **Crigler massage – 90% resolve by 1 y**; probing at 9–12 months; dacryocystocele
Preseptal vs orbital cellulitis	**Orbital – proptosis, painful/restricted eye movements, ↓ vision, chemosis, RAPD**; ethmoid sinusitis MCC; **CT orbit**; IV antibiotics ± drainage

Condition	Key points
Ptosis (congenital)	Levator dystrophy; surgery early if occluding pupil (amblyopia)
Coloboma	Failure of closure of **embryonic (choroidal) fissure – inferonasal**; **CHARGE**, Patau, cat-eye syndrome
Aniridia	PAX6 (11p13); **WAGR – Wilms tumour risk (sporadic)**; glaucoma, cataract, foveal hypoplasia, nystagmus
Ectopia lentis	**Marfan (up & out), homocystinuria (down & in)**, Weill–Marchesani (microspherophakia), sulfite oxidase deficiency
Optic neuritis	Post-viral, ADEM, MOG, MS; bilateral in children; steroids
Papilloedema	Raised ICP (tumour, IIH); no visual loss early (vs papillitis)
Cortical visual impairment	**MC cause of visual impairment in developed countries** (HIE, PVL); normal eye exam, poor visual behaviour
Vitamin A deficiency	**MC cause of preventable childhood blindness (developing countries)**; Bitot's spots, keratomalacia
Kayser–Fleischer ring	Wilson disease (Descemet)
Lisch nodules / phakomas	NF-1 / tuberous sclerosis
Cherry-red spot	Tay–Sachs, Niemann–Pick, sialidosis, CRAO
Uveitis	**JIA oligoarticular (ANA+, asymptomatic chronic anterior – screen)**, ERA (acute), sarcoid, Kawasaki (bilateral anterior), toxoplasma (posterior – MC infectious posterior uveitis)
Retinal haemorrhages	Abusive head trauma (multilayered, to ora serrata), birth (resolve by 4–6 weeks), leukaemia, sepsis

- **Causes of childhood blindness in India**: **corneal scarring (vitamin A, measles, ophthalmia neonatorum)** historically → now **ROP, congenital cataract, refractive errors, retinal dystrophies, CVI**; **"VISION 2020"** priorities.

28.7 Ear – Otitis, Hearing Loss

- **Acute otitis media, OME, mastoiditis** – see Respiratory chapter (S. pneumoniae MC; **amoxicillin 80–90 mg/kg/day**; **bulging TM most specific**; OME – MCC conductive hearing loss in children; grommets).
- **Otitis externa** – **Pseudomonas aeruginosa** (MC), S. aureus; swimmers; **pain on tragal pressure/pinna traction**; topical ciprofloxacin ± steroid drops; **malignant otitis externa** (diabetics/immunocompromised – Pseudomonas; rare in children). **Otomycosis** – Aspergillus niger (black spores), Candida.
- **Chronic suppurative otitis media (CSOM)**: **mucosal (safe – central perforation, mucoid discharge)** vs **squamosal (unsafe – attic/marginal perforation, cholesteatoma, foul scanty discharge – bone erosion, intracranial complications: brain abscess (temporal lobe/cerebellum), meningitis, lateral sinus thrombosis)**; Rx aural toilet + topical quinolone drops; mastoidectomy for cholesteatoma.
- **Foreign body/insect in ear** – kill insect with oil, remove; **button battery – emergency**.

Hearing loss in children

Conductive	Sensorineural (SNHL)
OME (MCC overall), wax, CSOM, foreign body, congenital aural atresia (Treacher Collins, Goldenhar), otosclerosis (rare), **OI (adolescent)**	**Genetic (50–60%) – non-syndromic AR (GJB2/connexin-26 – MC genetic cause)**; syndromic – **Pendred (goitre), Usher (retinitis pigmentosa – MC deaf-blind), Waardenburg, Alport (+ nephritis), Jervell–Lange-Nielsen (+ long QT), Treacher Collins (conductive), CHARGE, Stickler, BOR (branchio-oto-renal)**; **acquired – congenital CMV (MC non-genetic cause), rubella, toxoplasma, syphilis; meningitis (MC acquired postnatal cause – esp. pneumococcal/Hib)**, **kernicterus (auditory neuropathy)**, **ototoxic drugs (aminoglycosides – mitochondrial A1555G susceptibility, cisplatin, loop diuretics)**, prematurity/NICU stay, hypoxia, mumps (unilateral), measles, trauma, noise

- **High-risk registry (JCIH)**: family history, **NICU > 5 days**, ventilation, ECMO, ototoxic drugs, **hyperbilirubinaemia requiring exchange**, TORCH, craniofacial anomalies, syndromes, bacterial meningitis, head trauma, chemotherapy.
- **Universal newborn hearing screening**: **OAE (otoacoustic emissions – outer hair cells; quick, cheap; misses auditory neuropathy)** and **AABR/BERA (automated auditory brainstem response – detects auditory neuropathy; preferred in NICU graduates)**; **1-3-6 goals (screen by 1 month, diagnose by 3 months, intervene by 6 months)**; India – **RBSK, NPPCD**.
- **Behavioural audiometry**: **behavioural observation (0–6 m)**, **visual reinforcement audiometry (6 m–2.5 y)**, **play audiometry (2.5–5 y)**, pure-tone audiometry (> 5 y); **tympanometry** (middle ear – type B flat in OME).
- **Management**: **hearing aids (from 3–6 months)**, **cochlear implant – severe-to-profound bilateral SNHL, implant at ≥ 9–12 months (ideally before 2 y; best language outcomes)** – free under **ADIP scheme (India)**, speech therapy, sign language, auditory verbal therapy; **pneumococcal (PCV + PPSV23) & Hib vaccination before cochlear implant (meningitis risk)**.
- **Speech delay → always test hearing first.**

28.8 Atopic Dermatitis (Eczema)

- **MC chronic inflammatory skin disease of childhood**; onset **< 5 y (60% in 1st year, often 2–6 m)**; **filaggrin (FLG) loss-of-function** → barrier dysfunction + Th2 inflammation; atopic march.
- **Hanifin & Rajka / UK Working Party criteria**: **itch (essential)** + ≥ 3 of – flexural involvement (cheeks/extensors in infants), history of asthma/hay fever, dry skin, onset < 2 y, visible flexural eczema.
- **Distribution**: **infants – face (cheeks), scalp, extensor surfaces; nappy area spared**; **children – flexures (antecubital, popliteal), neck, wrists, ankles**; adolescents – hands, eyelids, flexures; **lichenification** in chronic.
- Associated: **Dennie–Morgan folds (infraorbital), allergic shiners**, keratosis pilaris, ichthyosis vulgaris, palmar hyperlinearity, **pityriasis alba (hypopigmented scaly patches on face)**, white dermographism.
- **Complications**: **secondary S. aureus infection (impetiginisation – MC)**, **eczema herpeticum (Kaposi varicelliform eruption – HSV; punched-out erosions, fever – IV acyclovir, emergency)**, molluscum, growth/sleep problems.
- **Severity – SCORAD, EASI**.

STEPWISE MANAGEMENT

Daily emollients (cornerstone – liberal, after bath "soak and seal"), avoid soaps/irritants/wool, short lukewarm baths; **topical corticosteroids for flares (low potency – face/folds: hydrocortisone 1%; moderate – body: mometasone/fluticasone)**, fingertip-unit dosing, **proactive twice-weekly maintenance**; **topical calcineurin inhibitors (tacrolimus 0.03% ≥ 2 y, pimecrolimus) – face/eyelids, steroid-sparing**; crisaborole; **wet wraps**; **oral sedating antihistamines for sleep only (no anti-itch benefit)**; antibiotics/bleach baths for infection; severe – **phototherapy (NB-UVB), ciclosporin, methotrexate, dupilumab (≥ 6 months), JAK inhibitors (adolescents)**. Food allergy testing only if suggestive history.

28.9 Other Dermatitis & Common Paediatric Skin Disorders

Condition	Key points
Infantile seborrhoeic dermatitis	**Onset 2–10 weeks**, **cradle cap (greasy yellow scales on scalp)**, face, flexures, **nappy area involved**, **non-itchy**, baby well; **Malassezia**; Rx emollients/oil, mild shampoo, ketoconazole 2%, mild steroid; resolves by 6–12 m. Severe generalised + FTT + diarrhoea → **Leiner's disease (C5 dysfunction)**, LCH (purpuric seborrhoeic-like rash), immunodeficiency (Omenn)
Napkin (diaper) dermatitis	**Irritant contact (MC) – convex surfaces involved, folds SPARED**; **candidal – fold involvement + satellite papules/pustules** (Rx clotrimazole/nystatin); seborrhoeic; **Jacquet's erosive**; psoriasis (napkin psoriasis – well-demarcated); **acrodermatitis enteropathica (zinc)**, LCH; Rx frequent changes, barrier creams (zinc oxide), air exposure
Impetigo	**MC bacterial skin infection in children**; **non-bullous (70% – honey-coloured crusts; S. aureus > GAS)**; **bullous (S. aureus phage II, exfoliative toxin)**; Rx **topical mupirocin/fusidic acid (localised)**, oral cephalexin/cloxacillin (extensive); **post-streptococcal GN (not ARF)**
Staphylococcal scalded skin syndrome (SSSS)	**< 5 y**; exfoliative toxins (desmoglein-1) → **superficial (subgranular) split**; fever, **tender erythema → flaccid bullae, Nikolsky +, perioral crusting, mucosa spared**; Rx IV antistaphylococcal antibiotics, fluids; heals without scarring
SJS/TEN	Drugs (anticonvulsants, sulfonamides, NSAIDs), Mycoplasma (**MIRM – mucositis-predominant**); **full-thickness epidermal necrosis (subepidermal), mucosal involvement (≥ 2 sites), Nikolsky +**; SJS < 10% BSA, TEN > 30%; SCORTEN; Rx stop drug, supportive (burns unit), IVIG/ciclosporin/ steroids (debated)
Scabies	**Sarcoptes scabiei**; **nocturnal itching, burrows (finger webs, wrists, genitalia, axillae); infants – palms, soles, face, scalp (vesicopustules)**; family members affected; **Rx permethrin 5% (> 2 months) – whole body neck down (head too in infants), repeat after 1 week; treat all contacts**; ivermectin (> 15 kg); **crusted (Norwegian) scabies** – immunocompromised
Pediculosis capitis	Head lice – **permethrin 1%**, combing; nits on hair shafts
Tinea capitis	**Trichophyton tonsurans (MC)**, Microsporum; scaly patches with alopecia, **black dots, kerion (boggy inflammatory mass)**; **Wood's lamp – green fluorescence (Microsporum)**; **Rx oral griseofulvin (DOC, 6–8 weeks) or terbinafine** – **topical alone ineffective**
Tinea corporis/cruris	**Annular plaques with raised scaly active edge, central clearing**; KOH mount; topical azoles/ terbinafine; **avoid topical steroid combinations (India – steroid-modified tinea epidemic)**
Pityriasis versicolor	**Malassezia**; hypo/hyperpigmented scaly macules (trunk); **"spaghetti & meatballs"** on KOH; Wood's lamp yellow-green; ketoconazole/selenium sulfide
Molluscum contagiosum	Poxvirus; **pearly umbilicated papules**; self-limiting (6–18 months); curettage, cantharidin
Warts	HPV; salicylic acid, cryotherapy
Pityriasis rosea	HHV-6/7; **herald patch → "Christmas-tree" pattern** on trunk (along Langer lines), collarette scale; self-limiting 6–8 weeks
Psoriasis	**Guttate psoriasis after streptococcal pharyngitis (MC type in children)**; Auspitz sign, Koebner; nail pitting
Urticaria	Acute – viral (MC), foods, drugs; non-sedating antihistamines
Papular urticaria	Insect bite hypersensitivity – grouped itchy papules on exposed areas
Erythema multiforme	**HSV (MC)**, Mycoplasma; **target lesions (3 zones) – acral**, minimal mucosa
Acne	See Adolescent chapter
Vitiligo	Depigmented macules; topical steroids/tacrolimus, NB-UVB
Alopecia areata	Patchy non-scarring alopecia, **exclamation-mark hairs**; nail pitting; topical/intralesional steroids, JAK inhibitors
Ichthyosis	**Ichthyosis vulgaris (filaggrin, AD – MC)**; **X-linked (steroid sulfatase – boys, dark adherent scales, corneal opacities, cryptorchidism, prolonged labour)**; **collodion baby**; **harlequin ichthyosis (ABCA12)** – severe

Condition	Key points
Epidermolysis bullosa	Simplex (keratin 5/14), junctional (laminin-332), dystrophic (collagen VII – scarring, SCC); blistering with minimal trauma
Mastocytosis (urticaria pigmentosa)	Brown macules; **Darier sign (urtication on rubbing)**
Haemangioma vs vascular malformations	**Infantile haemangioma – appears after birth, proliferates, involutes, GLUT-1 +; propranolol**; **port-wine stain (capillary malformation) – present at birth, grows proportionately, never involutes; Sturge–Weber (V1); pulsed-dye laser**; **salmon patch (naevus simplex) – fades**
Café-au-lait macules	≥ 6 → NF-1; irregular "coast of Maine" → McCune–Albright; also Fanconi, Bloom, Legius
Congenital melanocytic naevus	Giant (> 20 cm adult size) – **melanoma risk**, neurocutaneous melanosis
Kawasaki / scarlet fever / measles rashes	See Infections

28.10 Skin Signs of Systemic Disease (quick table)

Skin sign	Think of
Ash-leaf macules, shagreen patch, adenoma sebaceum	Tuberous sclerosis
Café-au-lait + axillary freckling	NF-1
Port-wine stain (V1)	Sturge–Weber
Acanthosis nigricans	**Insulin resistance (obesity, T2DM, PCOS)**
Butterfly (malar) rash sparing nasolabial folds	SLE
Heliotrope rash, Gottron papules	Juvenile dermatomyositis
Palpable purpura on buttocks/legs	HSP (IgA vasculitis)
Erythema marginatum	Rheumatic fever
Salmon-pink evanescent rash with fever	Systemic JIA
Erythema nodosum	**TB (MC in India), streptococcal infection (MC in West)**, sarcoidosis, IBD, drugs
Periorificial & acral dermatitis + alopecia + diarrhoea	Acrodermatitis enteropathica (zinc)
Flaky-paint dermatosis, flag sign	Kwashiorkor
Casal's necklace	Pellagra (niacin)
Perifollicular haemorrhage, corkscrew hair	Scurvy
Phrynoderma	Vitamin A deficiency (also EFA, B-complex)
Blueberry muffin rash	Congenital rubella, CMV, neuroblastoma, leukaemia (extramedullary haematopoiesis/infiltration)
Angiokeratomas	Fabry
Silvery hair + partial albinism	Chediak–Higashi, Griscelli
Kinky/steely hair	Menkes
Seborrhoeic-like purpuric rash + lytic bone lesions	LCH
Hypopigmented whorls along Blaschko lines	Incontinentia pigmenti (later stage), hypomelanosis of Ito

Skin sign	Think of
Collodion membrane	Lamellar ichthyosis
Aplasia cutis congenita (scalp)	Trisomy 13, methimazole
Xanthomas	Familial hypercholesterolaemia, GSD I, cholestasis (Alagille)
Telangiectasia of bulbar conjunctiva	Ataxia telangiectasia
Mongolian spots (extensive)	GM1, Hurler (sometimes); usually normal

Practice MCQs · Disorders of the Eye, Ear & Skin

181 questions · correct option highlighted in green · cover the answer strip to self-test

Conjunctivitis

1. Persistent deviation of an eye in an infant requires evaluation at the age of

A. 1 mo
B. 3 mo
C. **6 mo**
D. 9 mo
E. 12 mo

Ans C

2. Optic neuritis is inflammation or demyelinization of the optic nerve with attendant impairment of function, all the following may cause optic neuritis EXCEPT

A. lead poisoning
B. chloramphenicol
C. vincristine
D. **methylprednisolone**
E. hydroxychloroquine

Ans D Treatment of optic neuritis include high-dose intravenous methylprednisolone may help to speed the visual recovery in young adults, and it may prevent the development of multiple sclerosis in those at risk.

3. One of the following may support a diagnosis of drug eruption

A. neutrophilia
B. basophilia
C. **eosinophilia**
D. lymphocytosis
E. monocytosis

Ans C Medication eruptions begin on the trunk 7-10 days after exposure; they spread peripherally and are associated with pruritus and, less commonly, with fever, arthralgia, and lymphadenopathy. Eosinophilia may support a diagnosis of drug eruption but may be absent in the setting of bone marrow suppression. Penicillins, sulfa drugs, cephalosporins, nonsteroidal anti-inflammatory drugs, anticonvulsants, and aminoglycosides are common offenders.

4. Pemphigus vulgaris (PV) is a rare autoimmune blistering disorder, best treated initially with

B. methotrexate
C. azathioprine
D. cyclophosphamide
E. **systemic methylprednisolone**

Ans E The disease is best treated initially with systemic methylprednisolone 1-2 mg/kg/day. Azathioprine, cyclophosphamide, and methotrexate therapy all have been useful in maintenance regimens.

5. A rare but potentially life threatening complication of subcutaneous fat necrosis is

A. hyperkalemia
B. **hypercalcemia**
C. acidosis
D. septicemia
E. seizures

Ans B A rare but potentially life threatening complication is hypercalcemia. It manifests at 1-6 mo of age as lethargy, poor feeding, vomiting, failure to thrive, irritability, seizures, shortening of the QT interval on electrocardiography, or renal failure.

6. Aphthous stomatitis consists of solitary or multiple painful ulcerations occur on the labial, buccal, lingual, sublingual, palatal, or gingival mucosa; in severe, debilitating cases, systemic therapy with all the following may be helpful EXCEPT

A. corticosteroids
B. colchicine
C. **antibiotics**
D. dapsone
E. thalidomide

Ans C Treatment of aphthous stomatitis is palliative. The majority of mild cases do not require therapy. Relief of pain, particularlybefore eating, may be achieved with the use of a topical anestheticsuch as viscous lidocaine or an oral rinse with a combined solution of elixir of diphenhydramine, viscous lidocaine, and an oral antacid. Caution must be taken to avoid hot food and drink after topical anesthetic use.

Ophthalmia Neonatorum

7. The MOST widely used visual acuity test for preschool children is

A. **Tumbling E test**
B. Snellen letters
C. Lea symbols
D. Allen figures
E. HOTV test

Ans A The tumbling E test, in which the child indicates which direction the E is facing, is the most widely used visual acuity test for preschool children.

8. The symptoms of infantile glaucoma include the classic triad of

A. photophobia ,conjunctival injection, and blepharospasm
B. **tearing, photophobia , and blepharospasm**
C. tearing, photophobia , and ocular enlargement
D. corneal enlargement, photophobia , and blepharospasm
E. tearing, corneal edema , and blepharospasm

Ans B The symptoms of infantile glaucoma include the classic triad of epiphora (tearing), photophobia (sensitivity to light), and blepharospasm (eyelid squeezing). Each can be attributed to corneal irritation. Signs of glaucoma include corneal edema, corneal and ocular enlargement, and conjunctival injection.

9. In the use of topical medication, consideration of vehicle is as important as the specific therapeutic agent. Acute weeping lesions respond best to

A. ointments
B. gels
C. solutions
D. **wet compresses**
E. pastes

Ans D Acute weeping lesions respond best to wet compresses, followed by lotions or creams.

10. Dermatitis herpetiformis eruption is characterized by the following EXCEPT

A. symmetric
B. intensely pruritic
C. mostly on knees and elbows
D. **usual mucous membranes involvement**
E. occasional hemorrhagic lesions on the palms and soles

Ans D Mucous membranes are usually spared.

11. Histopathologic changes in sclerema neonatorum consist of

A. **increased size of fat cells**
B. fat necrosis
C. inflammation
D. giant cells
E. calcium crystals

Ans A Histopathologic changes in sclerema neonatorum consist of increases in the size of fat cells and in the width of the fibrous connective tissue septa. In contrast to SCFN, with which this disorder is mostly to be confused, fat necrosis, inflammation, giant cells, and calcium crystals are generally absent.

12. A 7-year-old child presented with painless, fluctuant, tense, 7mm, bluish papule on the floor of the mouth. Of the following, the MOST likely diagnosis is

A. **mucocele**
B. Epstein pearls
C. Fordyce spots
D. noma
E. canker sores

Ans A Lesions on the floor of the mouth are known as ranulas when the sublingual or submandibular salivary gland ducts are involved. Fluctuations in size are typical, and the lesions may disappear temporarily after traumatic rupture. Recurrenceis prevented by surgical excision of the mucus deposit and associated salivary gland(s).

Cataract

13. Children may have problems with going to sleep in a dark room (which may be mistaken for a behavioral problem) Of the following, the MOST likely cause is

A. dyslexia
B. diplopia
C. **nyctalopia**
D. amaurosis
E. amblyopia

Ans C or night blindness, is vision that is defective in reduced illumination.

14. Of the following, the MOST common benign tumor of the orbit is

A. **hemangioma**
B. rhabdomyosarcoma
C. lymphosarcoma
D. metastatic neuroblastoma
E. teratoma

Ans A Among malignant neoplasms, rhabdomyosarcoma, lymphosarcoma, and metastatic neuroblastoma are the most frequent.

15. Topical antibiotics have been used for many years to treat local cutaneous infections. Of the following, the MOST effective topical agents currently available is

A. polysporin
B. bacitracin
C. **fusidic acid**
D. gentamycin
E. tetracycline

Ans C Mupirocin, fusidic acid, and retapamulin are the most effective topical agents currently available and are as effectiveas oral erythromycin in treatment of mild to moderate impetigo.

16. Most cases of drug-induced linear IgA dermatosis are related to

A. **vancomycin**
B. anticonvulsants
C. ampicillin
D. cyclosporine
E. captopril

Ans A

17. The following drugs may cause hyperhidrosis EXCEPT

A. antipyretics
B. **antiemetics**
C. insulin
D. opiates
E. ciprofloxacin

Ans B Emetics.

18. Furunculosis is more common in individuals with low

A. serum zinc level
B. **serum iron level**
C. serum sodium level
D. serum calcium level
E. serum phosphorus level

Ans B Furunculosis is also more common in individuals with low serum iron levels, diabetes, malnutrition, HIV infection, or other immunodeficiency states.

Retinopathy of Prematurity

19. The MOST common cause of a dilated unreactive pupil is

A. internal ophthalmoplegia
B. tonic pupil
C. ocular trauma
D. **pharmacologic blockade**
E. Hutchinson pupil

Ans D The most common cause of a dilated unreactive pupil is purposeful or accidental instillation of a cycloplegic agent, particularly atropine and related substances.

20. **Hypertropia is seen in**

A. 3rd nerve palsy
B. **4th nerve palsy**
C. 5th nerve palsy
D. 6th nerve palsy
E. 7th nerve palsy

Ans B

21. **All the following agents are used as prophylaxis to prevent neonatorum ophthalmia**

A. 0.5% erythromycin drops
B. 1% silver nitrate drops
C. povidone iodine (2% solution)
D. single dose of ceftriaxone
E. **saline irrigation**

Ans E The eye should be irrigated initially with saline every 10-30 min, gradually increasing to 2-hr intervals until the purulent discharge has cleared,this is as part of treatment but not as prevention. An infant born to a woman who has untreated gonococcal infection should receive a single dose of ceftriaxone, 50 mg/kg (maximum 125 mg) IV or IM, in addition to topical prophylaxis.

22. **The risk factors associated with retinopathy of prematurity (ROP) are not fully known, but prematurity and the associated retinal immaturity at birth represent the major factors. Contributory factors include all the following EXCEPT**

A. oxygenation
B. apnea
C. heart disease
D. hypercarbia
E. **polycythemia**

Ans E Anemia and the need for transfusion are thought by some to be contributory factors.

23. **Retinopathy of prematurity (ROP) are classified into 5 stages. The stage which is characterized by the presence of a ridge and development of extraretinal fibrovascular tissue is**

A. stage
B. stage
C. **stage**
D. stage
E. stage

Ans C

24. **The MOST effective prophylaxis for recurrent otitis externa is**

A. **instillation of dilute alcohol immediately after bathing**
B. avoidance of swimming during the episode
C. ear protection
D. topical neomycin
E. use of hair dryer after swimming

Ans A The most effective prophylaxis is instillation of dilute alcohol or acetic acid (2%) immediately after swimming or bathing.

25. **A 3-day-old neonate presented with unvarying red purple hue restricted to left lower limb with atrophic underlying subcutaneous tissue. The lesions become more pronounced during changes in environmental temperature, physical activity, or crying. Of the following, the MOST likely diagnosis is**

A. reticulate capillary malformation
B. **cutis marmorata telangiectatica congenital**
C. harlequin color change
D. cutis marmorata
E. cutis verticis gyrate

Ans B

26. **A 10-year-old child with anterior uveitis developed a firm, smooth, erythematous papules that gradually enlarge to formannular plaques with a papular border and a normal, slightly atrophic central area several centimeters in size on the dorsum of the hands and feet. Of the following, the MOST likely diagnosis is**

A. tinea corporis
B. rheumatoid nodules
C. **granuloma annulare**
D. necrobiosis lipoidica
E. xanthomas

Ans C

27. **Infiltrative or destructive disorders that may produce atrophy of sweat glands by pressure or scarring include the following EXCEPT**

A. scleroderma
B. acrodermatitis chronica atrophicans
C. burns
D. **ichthyoses**
E. Sj ̲gren syndrome

Ans D Obstruction of sweat glands may occur in miliaria and in a number of inflammatory and hyperkeratotic disorders, such as the ichthyoses, psoriasis, lichen planus, pemphigus, porokeratosis, atopic dermatitis, and seborrheic dermatitis.

28. **Ecthyma gangrenosa is a necrotic ulcer covered with a gray-black eschar. It usually occurs in immunosuppressed patients with neutropenia. Of the following, the usual causative agent is**

A. S. aureus
B. **P. aeruginosa**
C. S. pneumoniae
D. H. influenzae type b
E. Clostridium perfringens

Ans B Ecthyma gangrenosum occurs in up to 6% of patients with systemic P. aeruginosa infection but can also occur as a primary cutaneous infection by inoculation.

29. **All the following are features of acrodermatitis enteropathica EXCEPT**

A. a rare autosomal recessive disorder
B. caused by an inability to absorb sufficient zinc from the diet
C. **Initial signs and symptoms usually occur in the 2nd year of life**
D. the cutaneous eruption consists of vesiculobullous, eczematous, dry, scaly, skin lesions
E. alopecia

Ans C Initial signs and symptoms usually occur in the 1st few mo of life, often after weaning from breast milk to cow's milk.

30. **Ocular manifestations of acrodermatitis enteropathica include all the following**

A. photophobia
B. **cataract**
C. conjunctivitis
D. blepharitis
E. corneal dystrophy

Ans B

31. **The replacement of elemental zinc for individuals with inherited acrodermatitis enteropathica is equal to**

A. 1 mg/kg/24 hr
B. **3 mg/kg/24 hr**
C. 5 mg/kg/24 hr
D. 7 mg/kg/24 hr
E. 10 mg/kg/24 hr

Ans B Oral therapy with zinc compounds is the treatment of choice. Replacement for individuals with inherited acrodermatitis enteropathica is with 3 mg/kg/24 hr of elemental zinc found in zinc sulfate, gluconate, or acetate (i.e., 220 mg of zinc sulfate contains 50 mg of elemental zinc). Zinc gluconate carries less risk of gastrointestinal distress.

Retinoblastoma

32. **Children with aniridia should be screened using renal ultrasonography every 3-6 mo until approximately 5 yr of age if there is an 11p13 region deletion because the child is at risk for**

A. rhabdomyosarcoma
B. neuroblastoma
C. **Wilms tumor**
D. gonadoblastoma
E. lymphoblastic leukemia

Ans C

33. **Retinoblastoma is the most common primary malignant intraocular tumor of childhood. It is unusual for a child to present with a retinoblastoma after the age of**

A. 1 yr
B. 2 yr
C. **3 yr**
D. 4 yr
E. 5 yr

Ans C

34. **The initial sign in the majority of patients with retinoblastoma is**

A. strabismus
B. **leukocoria**
C. pseudohypopyon
D. hyphema
E. vitreous hemorrhage

Ans B

35. **During the last decade there has been a dramatic shift in the treatment of retinoblastomas, by markedly reduced use of**

A. **external beam radiation**
B. systemic chemotherapy
C. laser therapy
D. cryotherapy
E. brachytherapy

Ans A Because those children who are irradiated during their 1st yr of life are 2-8 times more likely to develop second cancers as those irradiated after 1 yr of age. Patients treated with radiation tend to develop brain tumors and sarcomas of the head and neck. Secondary cataracts can also develop from radiation.

36. **The following disorders are associated with anhidrosis by unknown mechanisms**

A. dehydration
B. uremia
C. cirrhosis
D. Addison disease
E. **hypothyroidism**

Ans E Diverse disorders that are associated with anhidrosis by unknown mechanisms include dehydration; toxic overdose with lead, arsenic, thallium, fluorine, or morphine; uremia; cirrhosis; endocrine disorders such as Addison disease, diabetes mellitus, diabetes insipidus, and hyperthyroidism.

37. **Vaccination with BCG characteristically produces a papule approximately**

A. 1 wk after vaccination
B. **2 wk after vaccination**
C. 3 wk after vaccination
D. 4 wk after vaccination
E. 5 wk after vaccination

Ans B Vaccination with BCG characteristically produces a papule approximately 2 wk after vaccination. The papule expands in size, typically ulcerates within 2-4 mo, and heals slowly with scarring.

Refractive Errors

38. **Cat's-eye reflex is seen in all the following EXCEPT**

A. **juvenile xanthogranuloma**
B. persistent hyperplastic primary vitreous
C. cicatricial retinopathy of prematurity
D. retinoschisis
E. larval granulomatosis

Ans A

39. **Treatment of hyphema include all the following EXCEPT**

A. bed rest
B. **nonsteroidal anti-inflammatory drugs**
C. cycloplegic agent
D. topical steroids
E. systemicsteroids

Ans B All nonsteroidal anti-inflammatories and aspirin must be avoided.

40. **Topical corticosteroids are potent anti-inflammatory agents and effective antipruritic agents. Medium-potency topical corticosteroids include**

A. **betamethasone**
B. fluocinonide
C. halcinonide
D. clobetasol
E. hydrocortisone butyrate

Ans A Corticosteroids can be divided into 7 different categories on the basis of strength, but for practical purposes, 4 categories can be used: low, moderate, high, and super. Low-potency preparations include hydrocortisone, desonide, and hydrocortisone butyrate. Medium-potency compounds include amcinonide, betamethasone, flurandrenolide, fluocinolone, mometasonefuroate, and triamcinolone. High-potency topical steroids include fluocinonide and halcinonide.

41. **One of the MOST common causes of allergic contact dermatitis is**

A. **neomycin**
B. topical antihistamines
C. topical anesthetics
D. topical corticosteroids
E. ethylenediamine

Ans A sulfate is present in many nonprescription topical antibiotic preparations, and thus children are frequently exposed at an early age. It is one of the most common causes of allergic contact dermatitis, and use of combination products of neomycin with other antibiotics, antifungals, or corticosteroids may induce co- reactivity with these chemically-unrelated substances.

42. **Apocrine bromhidrosis develops after puberty as a result of the formation of short- chain fatty acids and ammonia by the action of the following bacteria on axillary apocrine sweat**

A. **anaerobic diphtheroids**
B. group A streptococcal infection
C. S. aureus
D. S. epidermidis
E. Pseudomonas aerogenosa

Ans A

43. **All the following matching are true EXCEPT**

A. Tinea capitis: dermatophyte infection of the scalp
B. Tinea corporis: infection of the glabrous skin
C. **Tinea cruris: infection of the axilla**
D. Tinea pedis: infection of the toe webs
E. Tinea unguium: dermatophyte infection of the nail plate

Ans C Tinea cruris, or infection of the groin, occurs most often in adolescent males and is usually caused by the anthropophilic species Epidermophyton floccosum or T. rubrum, but occasionally by the zoophilic species T. mentagrophytes.

Otitis Externa

44. **Bilateral facial palsy is a distinctive feature of**

A. **Möbius syndrome**
B. Brown syndrome
C. Parinaud syndrome
D. Duane syndrome
E. Bardet-Biedl syndrome

Ans A

45. **Purulent otorrhea is a sign of all the following conditions EXCEPT**

A. otitis externa
B. otitis media with perforation of the tympanic membrane
C. drainage from the middle ear through a patent tympanostomy tube
D. **skull base fracture**
E. drainage from first branchial cleft sinus

Ans D

46. **Of the following, the major trigger for otitis externa is**

A. eczema
B. trauma
C. **excessive wetness**
D. foreign body
E. previous infection

Ans C (swimming, bathing, increased environmental humidity) and dryness (dry canal skin and lack of cerumen) are the major triggers.

47. **The MOST common cause of otitis externa is**

A. **Pseudomonas aeruginosa**
B. Enterobacter aerogenes
C. Proteus mirabilis
D. Streptococci
E. Diphtheroids

Ans A External otitis (swimmer's ear) is caused most commonly by P. aeruginosa.

48. **The predominant symptom of otitis externa is**

A. tenderness by pressure on the tragus
B. **otalgia**
C. itching
D. hearing loss
E. otorrhea

Ans B

49. **The prominent sign of acute otitis externa is**

A. white cerumen
B. swollen and tender canal
C. opaque tympanic membrane
D. **otorrhea**
E. periauricular lymphadenopathy

Ans D Edema of the ear canal, erythema, and thick clumpy otorrhea are prominent signs of the acute disease.

50. **Necrotizing malignant otitis externa is probable with the finding of**

A. white cerumen
B. swollen and tender canal
C. **facial paralysis**
D. otorrhea
E. periauricular lymphadenopathy

Ans C Rarely, facial paralysis, other cranial nerve abnormalities, vertigo, and/or sensorineural hearing loss are present. If these occur, necrotizing (malignant) otitis externa is probable.

51. **The MOST common causative organism of necrotizing otitis externa is**

A. **Pseudomonas aeruginosa**
B. Enterobacter aerogenes
C. Proteus mirabilis
D. Streptococci
E. Diphtheroids

Ans A

Otitis Media

52. **Horner syndrome is an important cause of**

A. dyscoria
B. corectopia
C. microcoria
D. aniridia
E. **anisocoria**

Ans E meansinequality of the pupils. Dyscoria is abnormal shape of the pupil, and corectopia is abnormal pupillary position. Microcoria (congenital miosis) appear as a small pupil that does not react to light or accommodation and that dilates poorly, if at all, with medication. The term aniridiais a misnomer because iris tissue is usually present, although it is hypoplastic.

53. **Waardenburg syndrome is characterized by all the following EXCEPT**

A. an autosomal dominant inheritance
B. lateral displacement of the inner canthi
C. median white forelock
D. defective hearing
E. **hyperpigmentation of the skin**

Ans E Waardenburg syndrome is characterized principally by pigmentary disturbances (usually a median white forelock and patches of hypopigmentation of the skin).

54. **A major physical examination point to differentiate externa otitis from mastoiditis and otitis media is**

A. visualization of tympanic membrane
B. pus discharge from ear canal
C. **pain on manipulation of the auricle**
D. periauricular lymphadenopathy
E. hearing loss

Ans C

55. **The peak incidence of otitis media (OM) is in the age of**

A. **1-2 yr of life**
B. 4-5 yr of life
C. 6-7 yr of life
D. 9-11 yr of life
E. 13-15 yr of life

Ans A The peak incidence and prevalence of OM is during the 1st 2 yr of life. More than 80% of children will have experienced at least 1 episode of OM by the age of 3 yr.

56. **The mainstay of pain management for acute otitis media is**

A. **acetaminophen**
B. topical lidocaine
C. tympanostomy
D. myringotomy
E. narcotic analgesics

Ans A and Ibuprofen are the mainstay of pain management in acute otitis media.

57. **An important predictor for the development of recurrent and chronic otitis media is**

A. gender
B. race
C. **age**
D. genetic background
E. bottle feeding

Ans C The age of onset of OM is an important predictor of the development of recurrent and chronic OM, with earlier age of onset having an increased risk for exhibiting these difficulties later in life.

58. **The MOST likely reason for the higher rate of otitis media in infants is**

A. early exposure to unusual organisms
B. bottle feeding
C. oral appreciation of infancy
D. **less developed immunologic defenses**
E. high incidence of respiratory tract infections

Ans D and less-favorable eustachian tubal factors involving both the structure and function of the tube.

59. **Using standard culture techniques, the pathogens typically found in acute otitis media with effusion are recoverable in**

A. 10%
B. **30%**
C. 50%
D. 70%
E. 90%

Ans B

60. The reason behind progressive decline in the occurrence of otitis media as children grow older is

A. frequent exposure to the causative organisms
B. improved immune response
C. progressive reduction in eustachian tube wall compliance
D. the more-use of over-the counter medications
E. reduced incidence of respiratory tract infections

Ans C Progressive reduction in tubal wall compliance with increasing age may explain the progressive decline in the occurrence of OM as children grow older.

61. Otitis media is a universal finding in

A. Down syndrome
B. cleft lip
C. Patau syndrome
D. Ig A deficiency
E. cleft palate

Ans E In children with cleft palate, where OM is a universal finding, a main factor underlying the chronic middle-ear inflammation appears to be impairment of the opening mechanism of the eustachian tube.

62. Symptoms of acute otitis media include the following EXCEPT

A. irritability
B. change in sleep habit
C. upper respiratory tract symptoms
D. fever
E. facial pain

Ans E

63. Of the following, the LEAST sensitive and specific symptom of acute otitis media is

A. pulling at the ear
B. pain
C. pus discharge
D. hearing loss
E. fullness in the ear

Ans A

64. The MOST specific finding of the tympanic membrane in acute otitis media is

A. bulging
B. impairment of mobility
C. perforation
D. pale color
E. reduced translucency

Ans A

65. The following features specify non-typeable H. influenza otitis media EXCEPT

A. purulent conjunctivitis
B. often occurs in multiple family members
C. often affects infants
D. ineffective topical ocular antibiotics
E. bulging of tympanic membrane

Ans E This is a common finding in acute otitis media regardless the causative organism.

66. ''Watchful waiting'' and analgesics use in otitis media treatment is the right choice in

A. a four-month-old infant having presumed diagnosis of acute otitis media
B. a three-year-old girl having unilateral acute otitis media without otorrhea
C. a one-year-old infant having bilateral acute otitis media with otorrhea
D. a twelve-year-old girl having unilateral acute otitis media and fever of 390 c
E. an eighteen-month-old boy having bilateral acute otitis media without otorrhea

Ans B Younger children, children with otorrhea, and children with bilateral acute otitis media have a significantly enhanced benefit from antimicrobial therapy in comparison to older children, children without otorrhea, or children with unilateral acute otitis media.

67. The MOST crucial aspect in the treatment of acute otitis media is

A. choosing the right antibiotic
B. skillful examination of the ear
C. culture of middle ear aspirate
D. accurate diagnosis
E. analgesics use

Ans D

68. Of the following, the recommended first line treatment of acute otitis media is

A. amoxicillin
B. cefdinir
C. cefuroxime
D. clindamycin
E. cefpodoxime

Ans A 80-90 mg/kg/day in 2 divided doses for 10 days.

69. All the following are at greatest risk of harboring resistant bacteria in otitis media

A. those younger than 2 years of age
B. those in regular contact with large group of children
C. those who recently have received antimicrobial treatment
D. those who have bilateral otitis media with toxicity
E. those who are immune compromised

Ans D

70. Second line treatment for acute otitis media includes all the following agents

A. trimethoprim
B. cefdinir
C. amixicillin-clavulanate
D. cefuroxime
E. ceftriaxone

Ans A Drugs chosen for second-line treatment should be effective against β- lactamase–producing strains of non typeable H. influenzae and M. catarrhalis and against susceptible and most non- susceptible strains of S. pneumoniae. Only 4 antimicrobial agents meet these requirements: amoxicillin-clavulanate, cefdinir, cefuroxime axetil, and intramuscular ceftriaxone.

71. **Indications for myringotomy in children with acute otitis media include all the following EXCEPT**

A. severe refractory pain
B. hyperpyrexia
C. facial paralysis
D. immune compromised
E. **failure of first course of antibiotics**

Ans E Myringotomy should be considered as third-line therapy in patients that have failed 2 courses of antibiotics for an episode of AOM.

72. **The MOST common etiologic organism behind chronic suppurative otitis media is**

A. Klebseilla
B. Proteus
C. **Pseudomonas**
D.
E. Candida

Ans C The most common etiologic organisms are P. aeruginosa and S. aureus; however, the typical acute otitis media bacterial pathogens may also be the cause, especially in younger children or in the winter months.

73. **Gradenigo syndrome is the triad of**

A. suppurative otitis media, paralysis of the internal rectus muscle, and pain in the contralateral orbit
B. **suppurative otitis media, paralysis of the external rectus muscle, and pain in the ipsilateral orbit**
C. acute otitis media, paralysis of the external rectus muscle, and pain in the ipsilateral orbit
D. acute otitis media, paralysis of the external rectus muscle, and pain in the contralateral orbit
E. suppurative otitis media, paralysis of the internal rectus muscle, and pain in the ipsilateral orbit

Ans B

74. **Otitic hydrocephalus (a complication of chronic otitis media) is commonly associated with**

A. meningitis
B. **lateral sinus thrombosis**
C. subdural abscess
D. brain abscess
E. facial paralysis

Ans B

75. **Redundant skin over the posterior part of the neck is common in the following syndromes EXCEPT**

A. **Ehlers-Danlos**
B. Turner
C. Noonan
D. Down
E. Klippel-Feil

Ans A Loose folds of skin must be differentiated from a congenital defect of elastic tissue or collagen such as cutis laxa, Ehlers-Danlos syndrome, or pseudoxanthoma elasticum. Redundant skin over the posterior part of the neck is common in the Turner, Noonan, Down, and Klippel-Feil syndromes and monosomy 1p36; more generalized folds of skin occur in infants with trisomy 18 and short-limbed dwarfism.

76. **All the following treatments can be used in Kasabach-Merritt phenomenon EXCEPT**

A. corticosteroids
B. aminocaproic acid
C. vincristine
D. interferon-α
E. **propranolol**

Ans E

77. **An 8-year-old female child presented with a solitary, asymptomatic, smooth, dome shaped, blue-gray papule 8 mm in diameter on the dorsal aspect of her hand. Of the following, the MOST likely diagnosis is**

A. nevus spilus
B. nevus of ota
C. Spitz nevus
D. **blue nevus**
E. epidermal nevus

Ans D

78. **The Peutz-Jeghers syndrome is characterized by melanotic macules on the lips and mucous membranes and by gastrointestinal (GI) polyposis. Of the following, The MOST constant feature is**

A. diffuse hyperpigmentation of the nails
B. **buccal mucosal macules**
C. jejunal polyposis
D. melena
E. episodic abdominal pain

Ans B are the most constant feature of the disorder; in some families, occasional members may be affected only with the pigmentary changes.

79. **The following syndrome has a strong association with Café-au-lait spots**

A. Turner syndrome
B. **McCune-Albright syndrome**
C. Russell-Silver syndrome
D. Noonan syndrome
E. Rubinstein-Taybi syndrome

Ans B All other distracters have weak association.

80. **Stevens-Johnson syndrome is defined as affected body surface area**

A. <5%
B. **<10%**
C. <15%
D. <20%
E. <30%

Ans B SJS is defined as affected body surface area <10%, SJS-TEN overlap syndrome is affected body surface area between 10% and 30%, and TEN is affected body surface area >30%.

81. **All the following are advised in the treatment of Stevens-Johnson syndrome EXCEPT**

A. topical ocular steroid
B. topical oral anesthetics
C. systemic antibiotics for documented urinary or cutaneous infections
D. **prophylactic systemic antibiotics**
E. IV immunoglobulin

Ans D Systemic antibiotics are indicated for documentedurinary or cutaneous infections and for suspected bacteremia (Staphylococcus aureus or Pseudomonas aeruginosa) because infection is the leading cause of death. Prophylactic systemic antibiotics are not necessary.

82. **Classic Ehlers-Danlos syndrome is characterized by the following EXCEPT**

A. **post-term birth**
B. skin hyperelasticity
C. easy bruising
D. severe joint hypermobility
E. subcutaneous spheroids

Ans A Premature birth caused by rupture of membranes.

83. **Yellow nail syndrome manifests as thickened, excessively curved, slow-growing yellow nails without lunulae. Associated systemic diseases include the following EXCEPT**

A. bronchiectasis
B. recurrent bronchitis
C. chylothorax
D. **chronic hepatitis**
E. focal edema of the limbs and face

Ans D Deficient lymphatic drainage, caused by hypoplastic lymphatic vessels, is believed to lead to the manifestations of this syndrome.

84. **Staphylococcal scalded skin syndrome is caused predominantly by phage group 2 staphylococci, particularly strains 71 and 55, which are present at localized sites of infection. Of the following, the MOST common focus of infection is**

A. **nasopharynx**
B. umbilicus
C. urinary tract
D. superficial abrasion
E. conjunctivae

Ans A Foci of infection include the nasopharynx and, less commonly, the umbilicus, urinary tract, a superficial abrasion, conjunctivae, and blood.

85. **A 4-year-old child presented with a 2 pearly, skin-colored, smooth, dome-shaped, 4 mm papules with a central umbilication on the face. Of the following, the MOST likely diagnosis is**

A. **molluscum contagiosum**
B. ectopic sebaceous glands
C. keratoacanthoma
D. warty dyskeratoma
E. cryptococcosis

Ans A They typically have a central umbilication from which a plug of cheesy material can be expressed. The papules may occur anywhere on the body, but the face, eyelids, neck, axillae, and thighs are sites of predilection.

86. **Comedonal acne, particularly of the central face, is frequently the first sign of pubertal maturation. It occurs in**

A. 20% of adolescents
B. 40% of adolescents
C. 60% of adolescents
D. **80% of adolescents**
E. 100% of adolescents

Ans D

87. **The hair is sparse, thin, and depigmented, sometimes displaying a "flag sign "which is a characteristic finding in**

A. pellagra
B. scurvy
C. vitamin a deficiency
D. **kwashiorkor**
E. acrodermatitis enteropathica

Ans D Nails are thin and soft, and hair is sparse, thin, and depigmented, sometimes displaying a "flag sign" consisting of alternating light and dark bands that reflect alternating periods of adequate and inadequate nutrition.

Hearing Loss

88. **Spasmus nutans is a special type of acquired nystagmus in childhood. In its complete form, it is characterized by the triad of head nodding, torticollis and**

A. **pendular nystagmus**
B. latent nystagmus
C. Seesaw nystagmus
D. downbeat nystagmus
E. gaze-paretic nystagmus

Ans A The nystagmus is characteristically very fine, very rapid, horizontal, and pendular; it is often asymmetric, sometimes unilateral. Signs usually develop within the 1st yr or 2 of life. Components of the triad may develop at various times. In many cases, the condition is benign and self-limited, usually lasting a few months, sometimes years.

89. **Bloody otorrhea is associated with all the following EXCEPT**

A. neoplasm
B. trauma
C. foreign body
D. **branchial cleft sinus**
E. blood dyscrasia

Ans D

90. **The MOST common infectious cause of congenital sensorineural hearing loss is**

A. Measles virus
B. **Cytomegalovirus**
C. Toxoplasma gondii
D. Rubella virus
E. Treponema pallidum

Ans B

91. **The MOST common cause of bacterial meningitis that results in sensorineural hearing loss after the neonatal period is**

A. H. influenza
B.
C. **Streptococcus pneumoniae**
D. Listeria monocytogenes
E. Staphylococcus aureus

Ans C

92. **Of the following, the congenital infectious pathogen implicated in sensorineural hearing loss in children is**

A. Measles virus
B. Mumps virus
C. Streptococcus pneumoniae
D. **Lymphocytic choriomeningitis virus**
E. H. influenza

Ans D

93. **All the following are causes of sudden sensorineural hearing loss EXCEPT**

A. autoimmune disease
B. Epstein Barr virus infection
C. thromboembolic event
D. trauma
E. **Rubella virus infection**

Ans E

Eczema & Atopic Dermatitis

94. **All the following are causes of blepharitis EXCEPT**

A. Molluscum virus
B. Phthirus pubis
C. Staphylococcus epidermidis
D. Staphylococcus aureus
E. **Streptococci**

Ans E

95. **The MOST common cause of dizziness in young children is**

A. cholesteatoma in the mastoid or middle ear
B. **eustachian tube middle ear disease**
C. labyrinthitis
D. vestibular neuronitis
E. benign paroxysmal vertigo

Ans B

96. **Systemic adverse effects of high-potency and superpotent topical steroids occur with long-term use and include**

A. telangiectasia
B. acneiform eruptions
C. purpura
D. **cataracts**
E. increased hair growth

Ans D Poor growth, cataracts, and suppression of adrenal function.

97. **Pityriasis alba occurs mainly in children and causes lesions that are**

A. hyperpigmented
B. well-defined
C. severely erythematous
D. **finely scaly**
E. intensely pruritic

Ans D Pityriasis alba occurs mainly in children and causes lesions that are hypopigmented, ill-defined, round or oval patches. They may be mildly erythematous and finely scaly. Itching is minimal or absent.

98. **Nail pitting or grooves are usually seen in the following hair disorders**

A. telogen effluvium
B. trichotillomania
C. tinea capitis
D. **alopecia areata**
E. traction alopecia

Ans D

99. **A 3-month-old infant presented with an intensely erythematous, confluent plaque with a scalloped border, a sharply demarcated edge, and satellite pustules in the perianal skin, perineum, and inguinal folds. Of the following, the MOST likely diagnosis is**

A. seborrheic dermatitis
B. atopic dermatitis
C. primary irritant contact dermatitis
D. **candidal diaper dermatitis**
E. bacterial dermatitis

Ans D

Seborrheic Dermatitis

100. **All the following are options in the treatment of congenital nasolacrimal duct obstruction EXCEPT**

A. nasolacrimal massage
B. **cleansing of the lids with cold water**
C. topical antibiotic
D. systemic antibiotics
E. probing with topical anesthesia

Ans B Cleansing of the lids with warm water.

101. **All the following are included in the normal flora of the external ear canal EXCEPT**

A. Coagulase negative staphylococcus
B. Micrococcus
C. Diphtheroids
D. Pseudomonas aeruginosa
E. **coli**

Ans E The normal flora of the external canal consists mainly of aerobic bacteria and includes Coagulase-negative staphylococci, Corynebacterium (diphtheroids), Micrococcus, and, occasionally, Staphylococcus aureus, Viridans streptococci, and Pseudomonas aeruginosa.

102. **A 3-day-old neonate presented with numerous firm, yellow-white, 1-2 mm papules and pustules with a surrounding erythematous flare in several sites of the body surface but the palms and soles were spared. Intralesional contents demonstrated eosinophils in Wright-stained smears and culture was sterile. Of the following, the MOST likely diagnosis is**

A. pyoderma
B. **erythema toxicum**
C. candidiasis
D. herpes simplex
E. transient neonatal pustular melanosis

Ans B The cause of erythema toxicum is unknown. The lesions can mimic pyoderma, candidiasis, herpes simplex, transient neonatal pustular melanosis, and miliaria but can be differentiated by the characteristic infiltrate of eosinophils and the absence of organisms on a stained smear.

103. **Seborrheic dermatitis is a chronic inflammatory disease most common in infancy and adolescence, it is characterized by the following EXCEPT**

A. manifest in 1st mo of life
B. self-resolves by 1 yr
C. cradle cap
D. **pruritis**
E. post inflammatory pigmentary changes

Ans D A greasy, scaly, erythematous papular dermatitis, which is usually nonpruritic in infants, may involve the face, neck, retroauricular areas, axillae, umbilicus, and diaper area.

104. **Initial management for infantile seborrheic dermatitis is**

A. **conservative**
B. low-potency topical corticosteroids
C. topical antifungal
D. topical calcineurin inhibitors
E. oral antifungal agents

Ans A Initial management for infantile seborrheic dermatitis is generally conservative given the self-limited nature of this condition. Emollients, baby oil, gentle shampooing with non-medicated baby shampoo and gentle use of a soft brush to remove scales are usually effective measures.

105. **The nodules MOST commonly seen in children are**

A. fibrofolliculomas
B. **epidermoid cysts**
C. pilar cyst
D. pilomatricoma
E. trichoepithelioma

Ans B are the nodules most commonly seen in children. Such a cyst is a sharply circumscribed, dome-shaped, firm, freely movable, skin-colored nodule often with a central dimple or punctum that is a plugged, dilated pore of a pilosebaceous follicle.

Diaper Dermatitis

106. **Cicatricial adhesion between the conjunctiva of the lid and the globe is called**

A. dermoid cyst
B. pterygium
C. pinguecula
D. **symblepharon**
E. parinaudoculoglandular syndrome

Ans D

107. **Of the following, the treatment of choice for otomycosis is topical**

A. polymyxin
B. ciprofloxacin
C. **clotrimazole**
D. hydrocortisone
E. neomycin

Ans C A fungal infection of the external auditory canal, or otomycosis, is characterized by fluffy white debris, sometimes with black spores seen; treatment includes cleaning and application of antifungal solutions such as clotrimazole or nystatin; other antifungal agents include m-cresyl acetate, gentian violet, and thimerosal.

108. **Hypohidrotic ectodermal dysplasia is characterized by the following EXCEPT**

A. dry skin
B. sparse scalp hair
C. normal sexual hair growth
D. anodontia
E. **delayed sexual development**

Ans E Sexual development is usually normal.

109. **Diaper dermatitis can be treated with following measures EXCEPT**

A. frequent changing of the diapers
B. **overwashing**
C. application of a protective barrier agent
D. application of topical sucralfate
E. application of 2.5% topical hydrocortisone

Ans B should be avoided because it leads to chapping and a worsening of the dermatitis.

110. **Anagen effluvium is an acute, severe, diffuse inhibition of growth of anagen follicles, resulting in loss of >80-90% of scalp hair. All the following are causes of anagen effluvium EXCEPT**

A. radiation
B. alkylating agents
C. thallium
D. thiouracil
E. **hypervitaminosis D**

Ans E Hypervitaminosis A.

111. **A 7-year-old child presented with a smooth, pearly, pink, telangiectatic ulcerated papule that enlarge slowly on his face. Of the following, the MOST likely diagnosis is**

A. pyogenic granuloma
B. nevocellular nevus
C. epidermal inclusion cyst
D. dermatofibroma
E. **basal cell carcinoma**

Ans E is very rare in children in the absence of a predisposing condition, such as nevoid basal cell carcinoma syndrome, xeroderma pigmentosum, and nevus sebaceus of Jadassohn, arsenic intake, or exposure to irradiation. The lesions are smooth, pearly, pink, telangiectatic papules that enlarge slowly and may bleed or ulcerate. Sites of predilection are the face, scalp, and upper back.

Bacterial Skin Infections

112. **Keratoconus is characterized by progressive thinning and bulging of the central cornea (cone shaped), which of the following is a sign of keratoconus?**

A. **Munson sign**
B. Dalrymple sign
C. Stellwag sign
D. von Graefe sign
E. Pseudohypopyon sign

Ans A Signs of keratoconus include Munson sign (bulging of the lower eyelid on looking downward) and the presence of a Fleischer ring (a deposit of iron in the epithelium at the base of the cone). Myogenic retraction of the upper lid occurs in thyrotoxicosis, in which it is associated with 3 classic signs: a staring appearance (Dalrymple sign), infrequent blinking (Stellwag sign), and lag of the upper lid on downward gaze (von Graefe sign).

113. **The leading reason for physician visits and for use of antibiotics among children is**

A. urinary tract infection
B. lower respiratory tract infection
C. **otitis media**
D. infectious gastroenteritis
E. lymphadenitis

Ans C is a leading reason for physician visits and for use of antibiotics and figures importantly in the differential diagnosis of fever. It often serves as the sole or the main basis for undertaking the most frequently performed operations in infants and young children; myringotomy with insertion of tympanostomy tubes and adenoidectomy. OM is also the most common cause of hearing loss in children.

114. **Capillary malformations (CMs) are present at birth. The most effective treatment for CM is with the pulsed-dye laser. Therapy can begin**

A. **in infancy**
B. after 5 yr
C. after 10 yr
D. during puberty
E. after puberty

Ans A Therapy can begin in infancy, when the surface area of involvement is smaller.

115. **Phototoxic drug eruptions can be caused by**

A. **nalidixic acid**
B. tetracyclines
C. chlorothiazides
D. sulfonamides
E. barbiturates

Ans A Other distracters cause photoallergic drug eruptions.

116. **Nail anomalies are common in certain congenital disorders; large nails are seen in**

A. **Rubinstein-Taybi syndrome**
B. Ellis– van Creveld syndrome
C. ectodermal dysplasias
D. trisomy 18 syndrome
E. Turner syndrome

Ans A Large nails are seen in Pachyonychia congenita, Rubinstein-Taybi syndrome, hemihypertrophy, while smallness or absence of nails are seen in Ectodermal dysplasias, nail-patella, dyskeratosis congenita, focal dermal hypoplasia, cartilage-hair hypoplasia, Ellis–van Creveld, Larsen, epidermolysis bullosa, incontinentia pigmenti, Rothmund-Thomson, Turner, popliteal web, trisomy 13, trisomy18, Apert, Gorlin- Pindborg, long arm 21 deletion, otopalatodigital, fetal alcohol, fetal hydantoin, elfin facies, anonychia, and...

117. **The MOST common lesions that precede nonbullous impetigo are**

A. scabies
B. burns
C. **insect bites**
D. chickenpox
E. pediculosis

Ans C

118. **Ecthyma resembles nonbullous impetigo in onset and appearance but gradually evolves into a deeper, more chronic infection. Of the following, the usual causative agent is**

A. S. aureus
B. S. pneumoniae
C. H. influenzae type b
D. Clostridium perfringens
E. **group A-β hemolytic streptococcus (GABHS)**

Ans E The causative agent is usually GABHS; S. aureus is also cultured from most lesions but is probably a secondary pathogen.

Viral Skin Infections

119. **Epibulbar dermoids are choristomas found in 75% of**

A. Marfan syndrome
B. craniosynostosis
C. Alport syndrome
D. **Goldenhar syndrome**
E. Cogan syndrome

Ans D A dermoid usually appears as a well-circumscribed rounded or oval, gray or pinkish-yellow mass with a dry surface from which short hairs may protrude. It may affect only the superficial layers of the cornea, although full-thickness involvement is common.

120. **Middle ear aspirates in children with bronchiolitis regularly contain**

A. Respiratory syncytial virus
B. Parainfluenza virus
C. **Streptococcus pneumonia**
D. Pseudomonas aeruginosa
E. Staphylococcus aureus

Ans C Acute otitis media is a known complication of bronchiolitis; middle-ear aspirates in children with bronchiolitis regularly contain bacterial pathogens, suggesting that respiratory syncytial virus is rarely, if ever, the sole cause of their acute otitis media.

121. **A 25-day-old neonate presented with bright red, protuberant, compressible, and sharply demarcated lesion on right cheek. Of the following, the MOST likely diagnosis is**

A. strawberry hemangioma
B. cavernous hemangioma
C. **superficial infantile hemangioma**
D. deep infantile hemangioma
E. tufted angioma

Ans C Superficial IHs are bright red, protuberant, compressible, sharply demarcated lesions that may occur on any area of the body. Although sometimes present at birth, they more often appear in the 1st 2 mo of life and are heralded by an erythematous or blue mark or an area of pallor, which subsequently develops a fine telangiectatic pattern before the growth phase.

122. **The MOST common photosensitive reaction seen in children is acute sunburn, effective treatment of the desquamative phase is with**

A. topical corticosteroids
B. **bland emollient**
C. cool compresses
D. aloe vera products
E. calamine lotion

Ans B A bland emollient is effective in the desquamative phase.

123. **Terry nails are characterized by a white ground glass appearance of the entire or the proximal end of the nail and a normal pink distal 1-2 mm of the nail; this finding is associated with**

A. leprosy
B. tuberculosis
C. Hodgkin disease
D. arsenic poisoning
E. **hypoalbuminemia**

Ans E

124. **Facial eruption that frequently follows a butterfly distribution and a dermatitis encircling the neck "Casal's necklace." is usually seen in**

A. **pellagra**
B. scurvy
C. vitamin A deficiency
D. kwashiorkor
E. acrodermatitis enteropathica

Ans A

Fungal Skin Infections

125. **All the following are options in the treatment of dendritic keratitis EXCEPT**

A. trifluridine
B. topical ganciclovir
C. systemic acyclovir
D. cycloplegic agent
E. **topical corticosteroids**

Ans E Topical use of corticosteroids causes exacerbation of superficial herpetic disease of the eye and may lead to corneal perforation; eye drops combining steroids and antibiotics are therefore to be avoided in treatment of red eye unless there are clear- cut indications for their use and close supervision during therapy.

126. **The MOST sensitive finding of tympanic membrane in determining middle ear effusion is**

A. contour
B. translucency
C. color
D. **mobility**
E. structural changes

Ans D

127. **Ataxia-telangiectasia is transmitted as an autosomal recessive trait; the characteristic telangiectasias develop at approximately 3 yr of age, first on**

A. **bulbar conjunctivae**
B. nasal bridge
C. malar areas
D. external ears
E. upper anterior chest

Ans A The characteristic telangiectasias develop at approximately 3 yr of age, first on the bulbar conjunctivae and later on the nasal bridge, malar areas, external ears, hard palate, upper anterior chest, and antecubital and popliteal fossae. Additional cutaneous stigmata include café-au-lait spots, premature graying of the hair, and sclerodermatous changes. Progressive cerebellar ataxia, neurologic deterioration, sinopulmonary infections, and malignancies are also seen.

128. **The generalized eruption of pityriasis rosea resembles a number of other diseases. Of the following, the MOST important is**

A. **secondary syphilis**
B. drug eruptions
C. viral exanthems
D. guttate psoriasis
E. nummular dermatitis

Ans A

129. **Onycholysis indicates separation of the nail plate from the distal nail bed; drugs which may cause onycholysis include**

A. meronem
B. vancomycin
C. adriamycin
D. **indomethacin**
E. cyclophosphamide

Ans D Common causes are trauma, long-term exposure to moisture, hyperhidrosis, cosmetics, psoriasis, fungal infection (distal onycholysis), atopic or contact dermatitis, porphyria, drugs (bleomycin, vincristine, retinoid agents, indomethacin, chlorpromazine [Thorazine]), and drug-induced phototoxicity from tetracyclines or chloramphenicol.

130. **The MOST common risk factor for scurvy in children is**

A. prolonged antibiotics use
B. **psychiatric disease**
C. exclusive breast feeding
D. chronic diarrhea
E. cow milk protein allergy

Ans B In children, the most common risk factors are behavioralor psychiatric disease that results in poor nutrition.

Parasitic Skin Infections

131. **The MOST serious organism which can rapidly destroy stromal tissue and lead to corneal perforation is**

A. Neisseria gonorrhoea
B. **Pseudomonas aeruginosa**
C. Staphylococcus
D. Streptococcus
E. Yersinia

Ans B

132. **The MOST common finding of the tympanic membrane in middle ear effusion is**

A. bulging
B. **impairment of mobility**
C. opacification
D. white color
E. reduced translucency

Ans B

133. **All the following are risk factors for development of melanoma EXCEPT**

A. xeroderma pigmentosum
B. **dark complexion**
C. excessive sun exposure
D. giant congenital nevus
E. immunosuppression

Ans B Fair complexion.

134. **A keloid is a sharply demarcated, benign, dense growth of connective tissue that forms in the dermis after trauma. Of the following, the LEAST characteristic feature of keloid is**

A. firm
B. raised
C. pink
D. rubbery
E. **pruritic**

Ans E They may be tender or pruritic.

135. **Beau lines are transverse grooves in the nail plate. They are usually indicative of periodic trauma or episodic shutdown of the nail matrix secondary to the following systemic diseases EXCEPT**

A. mumps
B. measles
C. pneumonia
D. **celiac disease**
E. hand-foot-and-mouth disease

Ans D

136. **Scabies is transmitted only rarely by fomites because the isolated mite dies within**

A. 2-3 hours
B. 12-24 hours
C. **2-3 days**
D. 5-7 days
E. 10-20 days

Ans C

137. **In an immunocompetent host, scabies is frequently heralded by intense pruritus, particularly at night. Of the following, the classic lesions of scabies are**

A. bullae
B. pustules
C. wheals
D. red papules
E. **threadlike burrows**

Ans E are the classic lesion of scabies but may not be seen in infants.

138. **The treatment of choice for scabies is**

A. oral ivermectin
B. lindane 1% cream
C. sulfur ointment 5-10%
D. **permethrin 5% cream**
E. crotamiton 10% cream

Ans D The treatment of choice for scabies is permethrin 5% cream (Elimite) applied to the entire body from the neck down, with particular attention to intensely involved areas, which is also standard therapy.

139.The treatment of choice for head lice is

A. spinosad
B. pyrethroids
C. **malathion**
D. lindane shampoo
E. benzyl alcohol lotion

Ans C Because of resistance of head lice to pyrethroids, malathion 0.5% in isopropanol is the treatment of choice for head lice and should be applied to dry hair until hair and scalp are wet, and left on for 12 hr. A second application 7-9 days after initial treatment may be necessary.

Birth Marks & Hemangiomas

140.Several metabolic diseases produce distinctive corneal changes in childhood. Fine opacities radiating in a whorl or fan-like pattern is seen in

A. cystinosis
B. mucopolysaccharidosis
C. gangliosidosis
D. **Fabry disease**
E. Wilson disease

Ans D

141. Vascular tortuosity, arterial and venous occlusions, "salmon patches," refractile deposits, pigmented lesions, arteriolarvenous anastomoses, and neovascularization (with "sea-fan" formations); is retinopathy of

A. Iron deficiency anemia
B. polycythemia vera
C. leukemia
D. **sickling disorders**
E. β-thalassemia

Ans D

142.In the usual patient with infantile hemangiomas (IH) who has no serious complications or extensive growth resulting in tissue destruction and severe disfigurement, treatment consists of

A. **expectant observation**
B. pulsed-dye laser therapy
C. topical timolol solution
D. oral propranolol
E. oral corticosteroids

Ans A because almost all lesions regress spontaneously, therapy is rarely indicated. Parents require repeated reassurance and support.

143.In a disfiguring, life or vision threatening, or ulcerated infantile hemangiomas IH; the first-line treatment is

A. oral corticosteroids
B. **oral propranolol**
C. intralesional corticosteroid injection
D. vincristine
E. interferon-α

Ans B In a disfiguring, life- or vision-threatening, or ulcerated IH that is not responding to other treatment, oral propranolol is the first-line treatment in most cases. IHs typically responds with growth arrest and often early signs of involution within a couple weeks of treatment initiation.

144.Evaluate for PHACES in any patient with facial hemangioma involving significant area of face, one of the following is not a feature of PHACES

A. posterior fossa abnormalities
B. arterial cerebrovascular abnormalities
C. coarctation of the aorta
D. **ear abnormalities**
E. sternal abnormalities

Ans D PHACES stands for posterior fossa brain defects such as Dandy-Walker malformation or cerebellar hypoplasia, large segmental facial infantile hemangioma, arterial cerebrovascular abnormalities such as aneurysms and stroke, coarctation of the aorta, eye abnormalities. Sternal raphe defects such as pits, scars, or supraumbilical raphe are infrequently observed.

145.The association of spindle cell hemangiomas with nodular enchondromas in the metaphyseal or diaphyseal cartilaginous portion of long bones is known as

A. Angiokeratoma Corporis Diffusum
B. Osler-Weber-Rendu Disease
C. Angiokeratoma of Mibelli
D. Kaposiform Hemangioendothelioma
E. **Maffucci syndrome**

Ans E

Acne

146.The ectopia lentis (displacement of the lens) is often downward and forward, and the lens tends to be small and round in

A. Marfan syndrome
B. Homocystinuria
C. **Weill-Marchesani syndrome**
D. Sulfite oxidase deficiency
E. Ehlers-Danlos syndrome

Ans C In most cases of Marfan syndrome, the lens is displaced superiorly and temporarely, it is amost always bilateral and relatively symmetric. In homocystinuria, the lens is usually displaced inferiorly and sometimes nasally.

147.Purulent otorrhea of a recent onset is indicative of

A. acute otitis externa
B. chronic otitis externa
C. **acute otitis media**
D. chronic otitis media
E. otitis media with effusion

Ans C

148.The hallmark of Waardenburg type 1 is the

A. **white forelock**
B. depigmented skin
C. deafness
D. heterochromia irides
E. unibrow (synophrys)

Ans A The hallmark of Waardenburg type 1 is the white forelock, which is seen in 20- 60% of patients. Only 15% of patients have areas of depigmented skin. Deafness occurs in 9-37%, heterochromia irides in 20%, and unibrow (synophrys) in 17-69% of those affected. Dystopia canthorum (i.e., telecanthus) is seen in all patients with Waardenburg type 1.

149. **A 12-year-old child presented with brawny edema of the face and neck that spreads rapidly to involve the thorax and arms. The face acquires a waxy, mask-like appearance. The involved areas feel indurated, woody, nonpitting, and are not sharply demarcated from normal skin. Of the following, the MOST likely diagnosis is**

A. scleredema of buschke
B. scleroderma
C. morphea
D. dermatomyositis
E. subcutaneous fat necrosis

Ans A

150. **Nail changes may be particularly associated with various other diseases. The following matchings are true EXCEPT**

A. psoriasis : pitting, yellow-brown discoloration, and thickening
B. lichen planus : violaceous papules in the proximal nail bed
C. Darier disease: red or white streaks that extend longitudinally
D. alopecia areata: Transverse rows of fine pits
E. acrodermatitis enteropathica: subungual parakeratotic scaling and thickening

Ans E Patients with acrodermatitis enteropathica may have transverse grooves (Beau lines) and nail dystrophy as a result of periungual dermatitis.

151. **All the following drugs can induce acneiform lesions in susceptible individuals**

A. gold
B. isoniazid
C. phenytoin
D. vitamin B1
E. phenobarbital

Ans D Diagnosis of acne is rarely difficult, although flat warts, folliculitis, and other types of acne (drug induced: glucocorticoidagents, anabolic steroids, gold, dactinomycin, isoniazid, lithium, phenytoin, progestins) may be confused with acne vulgaris. The differential diagnosis includes sarcoidosis, angiofibromas, keratosis pilaris, chloracne, rosacea, and fibrofolliculomas.

152. **Little evidence shows that ingestion of the following foods can trigger acne flares**

A. high carbohydrate foods
B. high spicy foods
C. high sweet foods
D. high fatty foods
E. no certain foods

Ans E Little evidence shows that ingestion of particular foods can trigger acne flares. When a patient is convinced that certain dietary items exacerbate acne, it is prudent for the patient to omit those foods.

153. **All topical preparations for acne vulgaris must be used for 6-8 wk before their effectiveness can be assessed. Of the following, the primary topical preparation should be**

A. azelaic acid
B. salicylic acid
C. topical retinoid
D. Benzoyl peroxide
E. topical clindamycin

Ans C A topical retinoid should be the primary treatment for acne vulgaris.

154. **Antibiotics are indicated for treatment of patients whose acne has not responded to topical medications. Of the following, the antibiotic that may cause bluish discoloration of the skin and mucous membranes is**

A. tetracycline
B. doxycycline
C. minocycline
D. erythromycin
E. trimethoprim-sulfamethoxazole

Ans C Rarely, minocycline causes dizziness, intracranial hypertension, bluish discoloration of the skin and mucous membranes, hepatitis, a lupus-like syndrome, and drug reaction with eosinophilia and systemic symptoms.

155. **Approximately 20% of normal neonates demonstrate acne in the 1st mo of life.Treatment is usually unnecessary. If desired, the lesions can be treated effectively with**

A. azelaic acid
B. salicylic acid
C. topical retinoid
D. benzoyl peroxide
E. topical clindamycin

Ans D Topical antifungals and/or benzoyl peroxide.

Vitiligo & Pigmentary Disorders

156. **Anterior uveitis is caused by**

A. toxoplasmosis
B. toxocariasis
C. cytomegalovirus
D. rubella
E. brucellosis

Ans E The causes of posterior uveitis are numerous; the more common are toxoplasmosis, histoplasmosis, cytomegalic inclusion disease, sarcoidosis, syphilis, tuberculosis, and toxocariasis.

157. **Bezold abscess refers to**

A. neck abscess that is originated from mastoiditis involving the temporal bone
B. mastoid bone abscess that is originated from mastoiditis
C. orbital cavity abscess that is originated from mastoiditis involving the temporal bone
D. brain abscess that is originated from mastoiditis involving the temporal bone
E. paranasal abscess that is originated from mastoiditis

Ans A Neck abscess that is originated from mastoid bone infection involving the temporal bone.

158. **The following is a feature of generalized (nonsegmental) vitiligo**

A. often occurs in the face
B. usually not accompanied by other autoimmune diseases
C. **progressive, with flare-ups**
D. involves hair compartment soon after onset
E. usually responsive to autologous grafting

Ans C All other distracters are features of segmental type.

159. **Beaded papules on the eyelids is the classic sign of**

A. pseudoxanthoma elasticum
B. Ehlers-danlos syndrome
C. cutis laxa
D. **lipoid proteinosis**
E. anetoderma

Ans D may be noted initially in early infancy as hoarseness. Skin lesions appear during childhood and consist of yellowish papules and nodules that may coalesce to form plaques. The classic sign is beaded papules on the eyelids. Lesions also occur on the face, forearms, neck, genitals, dorsum of the fingers, and scalp, where they result in patchy alopecia.

160. **Periungual fibromas that appear in late childhood should suggest a diagnosis of**

A. mucous cysts
B. junctional nevi
C. Lichen planus
D. **tuberous sclerosis**
E. subungual exostoses

Ans D

Urticaria & Angioedema

161. **Cherry-red spots (a bright to dull red spot at the center of the macula surrounded and accentuated by a grayish white or yellowish halo) are seen in all the following**

A. **Tuberous sclerosis**
B. Tay-Sachs disease
C. Sandhoff disease
D. Sulfatide lipidosis
E. Niemann-Pick disease

Ans A

162. **All the following are common organisms in all variants of acute mastoiditis EXCEPT**

A. S. Pneumoniae
B. non-typable H. Influenza
C. Pseudomonas aeruginosa
D. **Klebsiella spp.**
E. group A Streptococcus

Ans D

163. **The MOST common agent implicated in the etiology of erythema multiforme (EM) is**

A. Mycoplasma pneumonia
B. **Herpes simplex virus (HSV) labialis**
C. Herpes simplex virus HSV genitalis
D. Streptococcus pneumonia
E. Staphylococcus epidermidis

Ans B HSV labialis and, less commonly, HSV genitalis are implicated in 60-70% of episodes of EM and are believed to trigger nearly all episodes of recurrent EM, frequently in association with sun exposure.

164. **Cutis laxa is characterized by all the following EXCEPT**

A. widespread folds of lax skin
B. characteristic facial features
C. **hypermobility of the joints**
D. normal tensile strength of the skin
E. hoarse cry

Ans C Hyperelasticity and hypermobility of the joints are not present as they are in the Ehlers-Danlos syndrome.

165. **Urticaria pigmentosa is the most common form of mastocytosis. It can be confused with the following EXCEPT**

A. drug eruptions
B. **herpes simplex**
C. pigmented nevi
D. insect bites
E. bullous impetigo

Ans B The differential diagnosis of solitary mastocytomas includes recurrent bullous impetigo, herpes simplex, congenital melanocytic nevi, and juvenile xanthogranuloma. Urticaria pigmentosa can be confused with drug eruptions, postinflammatory pigmentary change, juvenile xanthogranuloma, pigmented nevi, ephelides, xanthomas, chronic urticaria, insect bites, and bullous impetigo. Diffuse cutaneous mastocytoma may be confused with epidermolytic hyperkeratosis.

166. **All the following can be used in the treatment of papular urticaria EXCEPT**

A. oral antihistamines
B. cool compresses
C. potent topical corticosteroids
D. **topical antihistamines**
E. systemic steroids

Ans D are potent immunologic sensitizers and have no role in the treatment of insect bite reactions.

Psoriasis

167. **Phakomas are seen in all the following EXCEPT**

A. Sturge-Weber syndrome
B. von Hippel-Lindau disease
C. von Recklinghausen disease
D. **Niemann-Pick disease**
E. Tuberous sclerosis

Ans D

168. **The apocrine glands are located in the following areas EXCEPT**

A. axillae
B. **upper chest**
C. areolae
D. perianal
E. genital

Ans B

169. **Erythema multiforme (EM) is characterized by an abrupt, symmetric cutaneous eruption, MOST commonly on the**

A. face
B. trunk
C. legs
D. palms and soles
E. **extensor upper extremities**

Ans E Lesions are relatively sparse on the face, trunk, and legs. Lesions can be seen on the palms and soles.

170. **Guttate psoriasis can be confused with the following EXCEPT**

A. viral exanthems
B. secondary syphilis
C. **nummular dermatitis**
D. pityriasis rosea
E. pityriasis lichenoides chronica (PLC)

Ans C The differential diagnosis of plaque type psoriasis includes nummular dermatitis, tinea corporis, seborrheic dermatitis, postinfectious arthritis syndromes, and pityriasis rubra pilaris.

171. **The treatment of psoriasis should be viewed as a 4-tier process. The second tier of therapy is**

A. **phototherapy**
B. high-potency corticosteroids
C. tar preparations
D. methotrexate
E. infliximab

Ans A

Ichthyosis

172. **The first sign in hypertensive retinopathy is**

A. retinal edema
B. flame-shaped hemorrhages
C. cotton-wool spots
D. papilledema
E. **irregular narrowing of the arteriole**

Ans E

173. **Nail growth is relatively slow; complete fingernail regrowth takes**

A. 2 mo
B. 4 mo
C. **6 mo**
D. 8 mo
E. 10 mo

Ans C Nail growth is relatively slow; complete fingernail regrowth takes 6 mo, while complete toe nail regrowth requires 12-18 mo.

174. **Toxic epidermal necrolysis is characterized by the following EXCEPT**

A. widespread blister formation
B. skin tenderness
C. **target lesions**
D. sudden onset
E. confluent erythema

Ans C TEN is defined by (1) widespread blister formation and morbilliform or confluent erythema, associated with skin tenderness; (2) absence of target lesions; (3) sudden onset and generalization within 24-48 hr; (4) histologic findings of full-thickness epidermal necrosis and a minimal-to-absent dermal infiltrate. These criteria categorize TEN as a separate entityfrom EM.

175. **Ichthyosis vulgaris is the most common of the disorders of keratinization; with an incidence of 1/250 live births. Onset generally occurs in the 1st yr of life. Scaling is MOST prominent on the**

A. **extensor aspects the legs**
B. abdomen
C. neck
D. face
E. scalp

Ans A Scaling is most prominent on the extensor aspects of the extremities, particularly the legs. Flexural surfaces are spared, and the abdomen, neck, and face are relatively uninvolved.

176. **In X-Linked ichthyosis, scaling is MOST pronounced on the**

A. **sides of the neck**
B. elbow flexures
C. knee flexures
D. palms
E. soles

Ans A Scaling is most pronounced on the sides of the neck, lower face, preauricular areas, anterior trunk, and the limbs, particularly the legs.

Sun Protection

177. **Retinopathy in subacute bacterial endocarditis is present in approximately**

A. 5% of cases
B. 20% of cases
C. **40% of cases**
D. 60% of cases
E. 80% of cases

Ans C The lesions include hemorrhages, hemorrhages with white centers (Rothspots), papilledema, and, rarely, embolic occlusion of the central retinal artery.

178. **The presenting symptom of Behçet disease is**

A. erythema nodosum
B. **recurrent aphthous stomatitis**
C. genital ulcerations
D. perianal ulceration
E. purpuric lesions

Ans B is present in almost all patients and is commonly the presenting symptom.

179. **Severe mucosal blistering; GI involvement; laryngeal involvement (airway obstruction); and urologic involvement are the extracutaneous clinical features of**

A. epidermolysis bullosa simplex– generalized (AD)
B. epidermolysis bullosa simplex–Dowling- Meara (AD)
C. **junctional epidermolysis bullosa–herlitz (AR)**
D. dominant dystrophic epidermolysis bullosa (AD)
E. recessive dystrophic epidermolysis bullosa–hallopeau-siemens (AR)

Ans C

180. **The etiology is unknown in 30-50% of pediatric cases of erythema nodosum; one of the MOST common known etiologies in children is**

A. **group A streptococcal infection**
B. cat-scratch disease
C. leprosy
D. mycoplasma
E. brucellosis

Ans A Most common etiologies in children include: group A streptococcal infection, Yersinia enterocolitica gastroenteritis, medications (cephalosporins, penicillins, macrolides), and inflammatory disorders (inflammatory bowel disease); sarcoidosis should be considered in young adults.

181. **Angular cheilitis is characterized by inflammation and fissuring at the corners of the mouth, often with associated erosion, maceration, and crusting. Of the following, the LEAST likely cause is**

A. chapping
B. chronic lip lickers
C. excessive salivation
D. **nutritional deficiencies**
E. contact dermatitis to toothpaste

Ans D are a less-frequent etiology.

"Most Common" One-Liners

Growth, development & nutrition

- MC cause of short stature: **familial short stature / constitutional delay** (normal variants); pathological in India – **undernutrition**
- MC cause of delayed dentition: **idiopathic**; first permanent tooth: **first molar (6 y)**
- First primary tooth: **lower central incisor (6 m)**
- MC cause of large AF: **rickets / hypothyroidism**; MC craniosynostosis: **sagittal**
- First sign of puberty – girls **thelarche**, boys **testicular enlargement**
- Earliest sign of rickets: **craniotabes**; earliest X-ray sign: **loss of zone of provisional calcification**
- First sign of healing rickets: **line of provisional calcification**
- Earliest symptom of vitamin A deficiency: **night blindness**; earliest sign: **conjunctival xerosis**
- MC nutritional deficiency: **iron**; MC cause of anaemia in children: **IDA**
- MCC of death in SAM: **infection**; first step in SAM: **treat hypoglycaemia**
- Last fat to disappear in marasmus: **buccal pad of fat**
- MC preventable cause of ID: **iodine deficiency / congenital hypothyroidism**
- MC genetic cause of ID: **Down syndrome**; inherited: **fragile X**
- MC first social milestone: **social smile (6 wk)**
- Reflex persisting for life: **parachute**

Neonatology

- MCC neonatal mortality (India): **prematurity / LBW**
- MC cause of neonatal seizures: **HIE**; MC type: **subtle**; DOC: **phenobarbitone**
- MC cause of EOS (India): **Klebsiella / E. coli**; (West): **GBS**
- MC cause of LOS (India): **Klebsiella**
- Earliest sign of neonatal sepsis: **poor feeding / lethargy**
- MC cause of HDN: **ABO incompatibility**
- MC site of kernicterus: **globus pallidus (basal ganglia)**
- MC birth fracture: **clavicle**; MC brachial plexus injury: **Erb (C5–C6)**
- MC GI emergency in preterm: **NEC**; MC site: **terminal ileum**
- MC metabolic problem in IUGR & IDM: **hypoglycaemia**
- MC anomaly in IDM: **CHD (VSD)**; most specific: **caudal regression**
- MC cause of respiratory distress in preterm: **RDS**; in term (elective LSCS): **TTN**
- MC congenital infection: **CMV**; MC non-genetic SNHL: **CMV**
- MC neonatal tumour: **sacrococcygeal teratoma**
- MC cause of neonatal cholestasis needing surgery: **biliary atresia**
- MC abdominal mass in newborn: **hydronephrosis / MCDK**
- MC rash in newborn: **erythema toxicum**

Genetics & metabolism

- MC chromosomal disorder (liveborn): **trisomy 21**; in abortuses (single): **45,X**; trisomy: **16**
- MC cause of Down syndrome: **maternal meiosis I non-disjunction**
- MC heart lesion in Down: **AVSD** (characteristic) / VSD
- MC leukaemia in Down < 4 y: **AMKL (M7)**; overall: **ALL**
- MC cause of male hypogonadism: **Klinefelter**
- MC cardiac lesion in Turner: **bicuspid aortic valve**; MCC death: **aortic dissection/CoA**
- MC mutation in CF: **ΔF508**; earliest manifestation: **meconium ileus**
- MC β-thal mutation in India: **IVS 1-5 (G→C)**
- MC lysosomal storage disease: **Gaucher**
- MC urea cycle defect: **OTC deficiency (X-linked)**
- MC FAO defect: **MCAD**
- MC hepatic GSD: **type I (von Gierke)**
- MC inborn error screened in India: **congenital hypothyroidism**
- MC inherited bleeding disorder: **vWD**; MC inherited thrombophilia: **factor V Leiden**
- MC RBC enzyme defect: **G6PD deficiency**
- MC inherited marrow failure: **Fanconi anaemia**

Immunology & infections

- MC primary immunodeficiency: **selective IgA deficiency**
- MC SCID: **X-linked (IL2RG)**; MC complement deficiency: **C2**
- MC phagocyte defect: **MPO deficiency**; CGD MCC death: **Aspergillus**
- MC vasculitis of childhood: **HSP (IgA vasculitis)**
- MC acquired heart disease (developed countries): **Kawasaki**; India: **RHD**
- MC cause of AFP now: **GBS**
- MC cause of bronchiolitis: **RSV**; croup: **parainfluenza 1**
- MC cause of common cold: **rhinovirus**; pneumonia (bacterial): **S. pneumoniae**; > 5 y: **Mycoplasma**
- MC cause of epiglottitis (historic): **Hib**
- MC cause of AOM: **S. pneumoniae**
- MC complication of measles: **otitis media**; MCC death: **pneumonia**
- MC complication of varicella: **secondary bacterial skin infection**; neuro: **cerebellar ataxia**
- MC complication of mumps (children): **aseptic meningitis**; acquired unilateral SNHL: **mumps**
- MCC of meningitis > 3 m: **S. pneumoniae**; sequela: **SNHL**
- MCC diarrhoea < 5 y: **rotavirus**; dysentery: **Shigella**
- MC EPTB in children: **lymph node TB**

- MC cause of focal seizures in India: **neurocysticercosis**
- MC helminth: **Ascaris**
- MC opportunistic infection in HIV infants: **PCP**
- MC mode of HIV transmission in children: **vertical (intrapartum)**

Systemic paediatrics

- MC CHD: **VSD** (perimembranous); cyanotic: **TOF**; cyanotic in newborn: **TGA**
- MC cause of CHF in first week: **HLHS**; at 6–8 weeks: **large VSD**
- MC organism in IE: **viridans streptococci**
- MC valve in RHD: **mitral**; MC major Jones criterion: **arthritis**
- MC tachyarrhythmia: **SVT**
- MC childhood cancer: **ALL**; solid: **brain tumours**; extracranial solid: **neuroblastoma**
- MC brain tumour: **pilocytic astrocytoma**; malignant: **medulloblastoma**; supratentorial: **craniopharyngioma**
- MC renal tumour: **Wilms**; liver: **hepatoblastoma**; bone: **osteosarcoma**; soft-tissue: **rhabdomyosarcoma**; eye: **retinoblastoma**
- MC site of neuroblastoma: **adrenal**; of osteosarcoma: **distal femur**
- MC bone involved in LCH: **skull**
- MC cause of intestinal obstruction 3 m–3 y: **intussusception**; type: **ileocolic**; lead point: **Meckel's**
- MC surgical emergency in children: **appendicitis**
- MC cause of painless rectal bleeding 2–5 y: **juvenile polyp**; < 2 y massive: **Meckel's**
- MC cause of bright-red blood per rectum in infants: **anal fissure**
- MC cause of portal hypertension in Indian children: **EHPVO**
- MC cause of acute liver failure (India): **hepatitis A**
- MC indication for liver transplant: **biliary atresia**
- MC chronic disease of childhood: **asthma**; MC congenital stridor: **laryngomalacia**
- MC site of FB aspiration: **right main bronchus**
- MC organism of empyema (India): **S. aureus**
- MC cause of nephrotic syndrome: **MCD**; SRNS: **FSGS**; MCC death: **infection**
- MC cause of AGN: **PSGN**; MC primary GN worldwide: **IgA nephropathy**
- MC organism in UTI: **E. coli**; MCC obstructive uropathy (boys): **PUV**; hydronephrosis: **PUJ obstruction**
- MCC CKD < 10 y: **CAKUT**; MCC HTN in children: **renal parenchymal (reflux nephropathy)**
- MC form of CAH: **21-hydroxylase deficiency**; MCC ambiguous genitalia: **CAH**
- MCC congenital hypothyroidism: **thyroid dysgenesis (ectopic)**; acquired: **Hashimoto**
- MCC central precocious puberty in girls: **idiopathic**; CNS lesion: **hypothalamic hamartoma**
- MCC delayed puberty: **CDGP**
- MC type of CP: **spastic**; in preterm: **spastic diplegia (PVL)**
- MCC seizures < 5 y: **febrile seizure**
- MC epilepsy syndrome with 3 Hz spike-wave: **childhood absence**
- MC neurobehavioural disorder: **ADHD**; MC learning disability: **dyslexia**
- MC hip disorder in adolescents: **SCFE**; MCC acute painful limp 4–10 y: **transient synovitis**
- MC organism in osteomyelitis/septic arthritis: **S. aureus**; sickle cell: **Salmonella**
- MC JIA subtype: **oligoarticular**; MC congenital foot deformity: **CTEV**
- MC chest wall deformity: **pectus excavatum**; MC congenital neck mass: **thyroglossal cyst**
- MC TEF: **type C (EA with distal TEF)**; MC CDH: **Bochdalek (left)**
- MC cause of acute scrotum (prepubertal): **torsion of appendix testis**
- MC paediatric skin infection: **impetigo**; MC tinea capitis agent: **T. tonsurans**
- MC cause of visual impairment in school children: **refractive error (myopia)**
- MCC conductive deafness: **OME**; genetic SNHL: **GJB2 (connexin-26)**

Signs, Radiology & Eponyms

Radiological signs

Sign	Condition
Double-bubble	**Duodenal atresia** (Down), annular pancreas
Triple-bubble	Jejunal atresia
Soap-bubble (Neuhauser)	**Meconium ileus** (CF)
Football sign, Rigler sign	**Pneumoperitoneum (NEC perforation)**
Pneumatosis intestinalis	**NEC (stage II)**
Target / doughnut, pseudokidney (USG)	**Intussusception**
Claw / coiled-spring (enema)	Intussusception
String, double-track, shoulder, mushroom signs	**Hypertrophic pyloric stenosis**
Corkscrew duodenum, whirlpool sign	**Malrotation with volvulus**
Transition zone, saw-tooth	**Hirschsprung**
Coffee-bean sign	Sigmoid volvulus
Steeple / pencil sign	**Croup**
Thumb sign	**Epiglottitis**
Boot-shaped heart	**TOF**
Egg on a string	**TGA**
Snowman / figure of 8	**Supracardiac TAPVC**
Box-shaped heart	**Ebstein**
Rib notching, "3" / reverse-3	**Coarctation**
Scimitar	PAPVC (right)
Water-bottle heart	Pericardial effusion
Ground-glass, air bronchogram	**RDS**
Fluid in fissure, perihilar streaks	**TTN**
Coarse patchy infiltrates, hyperinflation	**MAS**
White-out hemithorax	Effusion / collapse / agenesis
Air crescent (Monod)	Aspergilloma
Signet-ring, tram tracks	**Bronchiectasis**
Cupping, fraying, splaying	**Rickets**
Frankel line, Wimberger ring, Pelkan spur, Trümmerfeld zone	**Scurvy**
Wimberger sign (medial tibia), periostitis, "celery stalk"	**Congenital syphilis** (celery stalk – also rubella)
Hair-on-end skull	**Thalassaemia**, sickle cell, IDA (severe)
Punched-out lytic skull lesion, vertebra plana, floating teeth	**LCH**
Sunburst, Codman triangle	**Osteosarcoma**

Sign	Condition
Onion-peel periosteal reaction	**Ewing sarcoma**
Bone-within-bone, Erlenmeyer flask	**Osteopetrosis** (flask – also Gaucher)
Champagne-glass pelvis, trident hand	**Achondroplasia**
Telephone-receiver femur	**Thanatophoric dysplasia**
Oar-shaped ribs, J-shaped sella, beaked vertebrae	**MPS (dysostosis multiplex)**
Crumpled bones, beaded ribs	**OI type II**
Stippled epiphyses	**Congenital hypothyroidism**, warfarin, Zellweger, chondrodysplasia punctata
Metaphyseal corner/bucket-handle fractures	**Child abuse**
Klein line fails to intersect epiphysis (Trethowan)	**SCFE**
Crescent sign, Gage sign	**Perthes**
Shenton line broken, ↑ acetabular index	**DDH**
Tram-track gyral calcification	**Sturge–Weber**
Periventricular calcification	**CMV**
Diffuse calcification + hydrocephalus	**Toxoplasma**
Candle-dripping subependymal nodules	**Tuberous sclerosis**
Molar tooth sign	**Joubert syndrome**
Eye-of-the-tiger	**PKAN (Hallervorden–Spatz)**
Face of giant panda	**Wilson disease**
Puff of smoke (angiography)	**Moyamoya**
Lemon & banana signs (USG)	**Chiari II / spina bifida**
Hot cross bun skull	Rickets (frontal-parietal bossing); hot cross bun pons – MSA (adult)
Starry sky (CT)	Neurocysticercosis (multiple); starry sky histology – Burkitt
Basal exudates + hydrocephalus + infarcts	**TB meningitis**
Ring-enhancing lesion with scolex	**Neurocysticercosis**
Triangular cord sign	**Biliary atresia**
Claw sign (kidney)	**Wilms tumour**
Drooping-lily sign	Duplex kidney (upper-pole obstruction) / neuroblastoma displacing kidney
Keyhole sign (fetal bladder)	**Posterior urethral valves**
Cobra-head	Ureterocele
Bright echogenic large kidneys	**ARPKD**

Clinical signs & eponyms

Sign	Condition
Koplik spots	**Measles**
Forchheimer spots	Rubella
Nagayama spots	Roseola
Pastia lines, circumoral pallor, strawberry tongue	**Scarlet fever**

Sign	Condition
Slapped cheek	**Parvovirus B19**
Dewdrop on rose petal	Varicella
Herald patch, Christmas-tree	Pityriasis rosea
Bitot spots	Vitamin A deficiency
Flag sign, flaky-paint dermatosis	**Kwashiorkor**
Casal necklace	Pellagra
Frog-leg posture with pseudoparalysis	**Scurvy**
Rachitic rosary (painless) vs scorbutic rosary (painful)	Rickets vs scurvy
Harrison sulcus, Marfan sign (double malleoli)	Rickets
Gowers sign	**DMD** (proximal weakness)
Trousseau, Chvostek	Hypocalcaemia
Kernig, Brudzinski	Meningitis
Setting-sun sign, MacEwen (cracked-pot)	Hydrocephalus
Ortolani (reduces), Barlow (dislocates), Galeazzi	**DDH**
Drehmann sign	**SCFE**
Trendelenburg sign	Hip abductor weakness (DDH, Perthes)
Steinberg (thumb), Walker–Murdoch (wrist)	**Marfan**
Dance sign, red-currant jelly stool	**Intussusception**
Olive, visible peristalsis	**Pyloric stenosis**
Squirt (blast) sign	**Hirschsprung**
Blue-dot sign	Torsion of appendix testis
Prehn sign	Epididymitis (relief)
Cremasteric reflex absent	Testicular torsion
Dennie–Morgan folds, allergic shiners	Atopy
Darier sign	Mastocytosis
Nikolsky sign	SSSS, TEN, pemphigus
Auspitz sign, Koebner	Psoriasis
Russell sign	**Bulimia nervosa**
Kayser–Fleischer ring	**Wilson**
Lisch nodules, Crowe sign	**NF-1**
Ash-leaf, shagreen, Koenen tumours	**Tuberous sclerosis**
Brushfield spots, sandal gap, simian crease	**Down syndrome**
Rocker-bottom feet, clenched overlapping fingers	**Edwards**
Hutchinson triad, saddle nose, saber shin, Clutton joints	**Late congenital syphilis**
Parrot pseudoparalysis	Early congenital syphilis
Gregg triad	**Congenital rubella**
Beck triad, pulsus paradoxus	Cardiac tamponade
Machinery murmur (Gibson)	**PDA**
Fixed wide split S2	**ASD**

Sign	Condition
Single S2 + ESM	**TOF**
Carey Coombs murmur	Acute rheumatic carditis
Milkmaid grip, pronator sign, spooning	**Sydenham chorea**
Erythema marginatum, SC nodules	ARF
BCG scar reactivation	**Kawasaki**
Heliotrope rash, Gottron papules	JDM
Salmon-pink evanescent rash	sJIA
Raccoon eyes, opsoclonus–myoclonus	**Neuroblastoma**
Leukocoria (cat's eye reflex)	**Retinoblastoma**
Cherry-red spot	**Tay–Sachs**
Cushing triad	Raised ICP (herniation)
Hypsarrhythmia	West syndrome
3 Hz spike-and-wave	Absence epilepsy
Radermecker complexes	SSPE
Burst-suppression	Ohtahara, severe HIE, NKH
Albuminocytological dissociation	**GBS**
Vincent curtsy	Detrusor overactivity
Chik sign (nose pigmentation)	Chikungunya
Eschar	Scrub typhus
Pseudomembrane bleeds on removal, bull neck	**Diphtheria**
Whoop, post-tussive vomiting	**Pertussis**
Risus sardonicus, trismus, opisthotonus	**Tetanus**

Named classifications & scores

Classification / score	Used for
Apgar	Newborn assessment at 1 & 5 min
New Ballard	Gestational age
Silverman–Andersen / Downes	Neonatal respiratory distress
Sarnat & Sarnat	HIE staging
Modified Bell	NEC staging
Papile	IVH grading
Kramer	Clinical jaundice zones
Bhutani nomogram	Hour-specific bilirubin (≥ 35 wk)
Finnegan	Neonatal abstinence
ICROP3	ROP
Pirani / Dimeglio	Clubfoot severity
Graf	DDH ultrasound
Catterall / Herring (lateral pillar) / Waldenström	Perthes

Classification / score	Used for
Southwick / Loder	SCFE
Salter–Harris	Physeal fractures
Gross	Oesophageal atresia/TEF
Spitz / Waterston	TEF prognosis
Sillence	Osteogenesis imperfecta
Tanner (SMR)	Puberty
Prader orchidometer	Testicular volume
Westley	Croup severity
Modified Centor (McIsaac)	Strep pharyngitis
Jones (revised 2015)	Rheumatic fever
Modified Duke	Infective endocarditis
Ross	Heart failure in infants
Wellcome / Gomez / IAP / Waterlow	Malnutrition
Marsh–Oberhuber	Celiac histology
Pediatric Appendicitis Score / Alvarado	Appendicitis
Kocher criteria	Septic hip
ILAR	JIA
GMFCS	Cerebral palsy function
British MRC	TB meningitis stages
Blantyre coma scale	Cerebral malaria (children)
PRISM / PIM	PICU mortality
PELD / MELD	Liver transplant allocation
Leipzig score	Wilson disease
Glasgow / paediatric GCS	Consciousness
SCORAD / EASI	Atopic dermatitis
ETROP type 1	ROP treatment threshold
Altman	Sacrococcygeal teratoma
Todani	Choledochal cyst
PRETEXT	Hepatoblastoma
INRG / Shimada	Neuroblastoma
Ann Arbor / Lugano	Hodgkin lymphoma
Lund–Browder	Burns (paediatric TBSA)
Parkland	Burns fluid

Drugs of Choice, Doses & Key Numbers

Drugs / treatment of choice

Condition	DOC / TOC
Neonatal seizures	**Phenobarbitone**
Status epilepticus (1st line)	**IV lorazepam / midazolam**
Infantile spasms	**ACTH** (TSC – **vigabatrin**)
Absence seizures	**Ethosuximide** (valproate if GTCS)
JME	**Valproate** (levetiracetam in girls)
Lennox–Gastaut	Valproate
Febrile seizure prophylaxis (intermittent)	**Clobazam**
Apnoea of prematurity	**Caffeine citrate**
PDA closure (preterm)	**Ibuprofen / indomethacin / paracetamol**
Duct-dependent CHD	**PGE_1**
Tet spell	Knee-chest → O_2 → **morphine** → propranolol
SVT (stable)	Vagal → **adenosine**
Kawasaki	**IVIG 2 g/kg + aspirin**
ARF secondary prophylaxis	**Benzathine penicillin every 3 weeks**
Strep pharyngitis	**Penicillin / amoxicillin × 10 d**
PPHN	**Inhaled nitric oxide**
RDS	**Surfactant + CPAP**
Croup	**Dexamethasone** (+ nebulised adrenaline)
Epiglottitis	Secure airway + **ceftriaxone**
Bronchiolitis	**Supportive (O_2, hydration)**
Pertussis	**Azithromycin**
Diphtheria	**ADS + penicillin/erythromycin**
Tetanus	**TIG + metronidazole + diazepam**
Typhoid (OPD)	**Cefixime / azithromycin**; IPD – **ceftriaxone**
Cholera (child)	**Azithromycin** (doxycycline > 8 y)
Shigella dysentery	**Ceftriaxone / azithromycin** (cipro – WHO)
Amoebic liver abscess	Metronidazole + luminal agent
Giardiasis	Metronidazole / tinidazole / nitazoxanide
Neurocysticercosis (viable)	**Albendazole + steroids + AED**
Scrub typhus	**Doxycycline** (azithromycin)
Severe malaria	**IV artesunate**
Vivax malaria	**Chloroquine + primaquine 14 d**
Kala-azar (India)	**Single-dose liposomal amphotericin B 10 mg/kg**
Congenital syphilis	**Aqueous crystalline penicillin G × 10 d**

Condition	DOC / TOC
Neonatal HSV	**IV acyclovir (14–21 d)**
Chlamydial conjunctivitis/pneumonia	**Oral erythromycin × 14 d**
Gonococcal ophthalmia	**Ceftriaxone single dose**
PCP	**High-dose co-trimoxazole + steroids**
Toxoplasmosis (congenital)	Pyrimethamine + sulfadiazine + folinic acid
Congenital CMV (symptomatic)	**Valganciclovir × 6 months**
Varicella (high-risk)	**Acyclovir**
TB (DS)	**2HRZE + 4HRE**; TBM – 2HRZE + 10HRE + steroids
Hypoglycaemia (neonate)	**10% dextrose 2 mL/kg**
Hypocalcaemia	**10% calcium gluconate 1–2 mL/kg**
Hyperkalaemia (cardioprotection)	**Calcium gluconate**
DKA	**NS → insulin 0.05–0.1 U/kg/h** (no bicarbonate)
Anaphylaxis	**IM adrenaline 0.01 mg/kg**
Hypothyroidism (congenital)	**L-thyroxine 10–15 µg/kg/day**
Graves disease	**Methimazole**
CAH	**Hydrocortisone + fludrocortisone**
Central precocious puberty	**GnRH agonist**
Central DI	**Desmopressin**
Nephrogenic DI	**Thiazides**
Nocturnal enuresis	**Alarm (TOC); desmopressin (DOC)**
Nephrotic syndrome	**Prednisolone**
SRNS	**Tacrolimus/ciclosporin + ACEi**
PSGN	**Diuretics + fluid/salt restriction**
Atypical HUS	**Eculizumab / plasma exchange**
Wilson disease	**Penicillamine/trientine**; maintenance – **zinc**
Biliary atresia	**Kasai before 60 days**
ITP (severe bleed)	**IVIG**
Haemophilia A	**Factor VIII** (mild – desmopressin); inhibitors – rFVIIa/emicizumab
vWD type 1	**Desmopressin**
Thalassaemia major	**Transfusion + chelation (deferasirox)**; cure – HSCT
Sickle cell (prevention)	**Hydroxyurea + penicillin prophylaxis**
Aplastic anaemia	**HSCT** (else ATG + ciclosporin)
Diamond–Blackfan	**Steroids**
Infantile haemangioma	**Propranolol**
JIA	**Methotrexate**; sJIA – IL-1/IL-6 blockers
Clubfoot	**Ponseti method**
DDH < 6 m	**Pavlik harness**
SCFE	**In-situ screw fixation**
Perthes	**Containment**

Condition	DOC / TOC
Septic arthritis	**Drainage + IV antibiotics**
Atopic dermatitis	**Emollients + topical steroids**
Scabies	**Permethrin 5%**
Tinea capitis	**Oral griseofulvin**
Acne (severe)	**Isotretinoin**
ADHD	**Methylphenidate** (< 6 y – parent training)
Autism irritability	**Risperidone / aripiprazole**
Tourette	**α2-agonists / aripiprazole + CBIT**
Anorexia nervosa (adolescent)	**Family-based therapy**
Organophosphate	**Atropine + pralidoxime**
Paracetamol overdose	**N-acetylcysteine**
Iron poisoning	**Desferrioxamine**
Lead (moderate/severe)	**Succimer / BAL + EDTA**
Kerosene	**Supportive – no emesis/lavage**
Scorpion sting	**Prazosin**
Snake bite	**Polyvalent ASV**
Methaemoglobinaemia	**Methylene blue**
Retinopathy of prematurity	**Laser** (anti-VEGF for zone I/A-ROP)
Congenital glaucoma	**Goniotomy / trabeculotomy**
Congenital cataract	**Surgery by 6–8 weeks**

Key numbers to remember

Item	Number
Birth weight doubles / triples / quadruples	**5 m / 1 y / 2 y**
Length at birth / 1 y / 4 y	**50 / 75 / 100 cm**
HC at birth / 1 y / 2 y	**34–35 / 45–46 / 48 cm**
AF closes	**12–18 months**; PF – 6–8 weeks
Primary teeth	**20**, start 6 m, complete 2½ y
Permanent teeth	**32**, start 6 y
Mid-parental height	(F + M)/2 **± 6.5 cm**
MUAC – SAM	**< 11.5 cm**
SAM WFH	**< −3 SD**
Vitamin A prophylaxis	**1 lakh at 9 m; 2 lakh 6-monthly to 5 y (9 doses)**
ORS osmolarity	**245 mOsm/L** (Na 75, glucose 75)
ReSoMal	**Na 45, K 40**
Zinc in diarrhoea	**10 mg (< 6 m) / 20 mg × 14 days**
Plan B ORS	**75 mL/kg over 4 h**
Plan C	**100 mL/kg RL – 30 in 1 h/30 min, 70 in 5 h/2½ h**

Item	Number
Holliday–Segar	**100/50/20 mL/kg/day**
Neonatal hypoglycaemia	**< 45 mg/dL**
SAM hypoglycaemia	**< 54 mg/dL**
Neonatal hypothermia – severe	**< 32 °C**
Phototherapy wavelength	**460–490 nm**
Exchange volume	**2 × 80–85 mL/kg**
Anti-D dose	**300 µg within 72 h**
NRP – PPV rate	**40–60/min**; compression ratio **3:1**
Neonatal adrenaline	**0.01–0.03 mg/kg IV**
CPR rate (child)	**100–120/min; 30:2 (1), 15:2 (2 rescuers)**
Defibrillation	**2 → 4 J/kg** (max 10)
Cardioversion	**0.5–1 → 2 J/kg**
ETT size	**age/4 + 4 (uncuffed)**
Min SBP (1–10 y)	**70 + 2 × age**
Fast breathing	**≥ 60 (< 2 m), ≥ 50 (2–12 m), ≥ 40 (1–5 y)**
Mantoux positive	**≥ 10 mm** (≥ 5 mm immunocompromised)
Sweat chloride – CF	**≥ 60 mmol/L**
Nephrotic proteinuria	**> 40 mg/m²/h; UPCR > 2**
Prednisolone (NS)	**2 mg/kg/day × 6 wk → 1.5 mg/kg alt days × 6 wk**
IVIG – Kawasaki	**2 g/kg**
Benzathine penicillin	**1.2 MU every 3 weeks**
PGE_1	**0.05–0.1 µg/kg/min**
Paracetamol	**15 mg/kg/dose** (toxic > 150 mg/kg)
Iron toxicity	**> 60 mg/kg elemental**
Enuresis age	**≥ 5 years**
Precocious puberty	**girls < 8, boys < 9 y**
Delayed puberty	**girls 13, boys 14 y**
Orchidopexy	**6–12 (≤ 18) months**
Cleft lip / palate repair	**3–6 m / 9–18 m** ("rule of 10s")
Kasai	**< 60 days**
Cochlear implant	**≥ 9–12 months**
ROP screening	**< 34 wk / < 2000 g at 2–4 weeks**
Congenital cataract surgery	**< 6–8 weeks**
SRS 2021	**IMR 27, NMR 19, U5MR 31**



Keep climbing.

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