Paediatrics

MBBS Recap: high-yield notes for revision

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Revision notes for the MD/MS entrance (MEC's PG paper): the facts an examiner asks in paediatrics, by topic, in the order of the practice questions.

Growth and development

Weight, length and head

- Birth weight about 3 kg, length 50 cm, head circumference 33-35 cm. A newborn may lose up to **10% of birth weight in week 1** and regains it by about 10-14 days.
- Weight **doubles at about 5 months (4-6), triples at 12 months, quadruples at 2 years**. Height doubles at about 4 years.
- Weight gain: about 30 g/day for 0-3 months, 400 g/month for 3-12 months, about 3 kg/year at 1-3 years, 2 kg/year at 3-7 years.
- Length gain: about 25 cm in year 1 (so about 75 cm at 1 year), 12 cm in year 2, 6-8 cm/year to 5 years, then about 5 cm/year.
- Head circumference gain: about 2 cm/month for 0-3 months, 1 cm/month for 3-6, 0.5 cm/month for 6-12 (so about 12 cm in year 1), 2 cm/year at 1-3 years, 1 cm/year at 3-5. It equals chest circumference at 9-12 months. Growth faster than about 1 cm per 2 weeks in the first 3 months suggests hydrocephalus.
- Worked example: head 35 cm at birth reaching 43 cm: 35 + 6 (first 3 months) = 41 cm at 3 months, then +1 cm/month, so 43 cm at about 5 months. Use the rate for each age band.
- **Microcephaly**: Nelson defines it as a head circumference **more than 3 SD below the mean** for age and sex (the textbook answer); WHO (2016) and most current guidelines use **below -2 SD**, with below -3 SD as severe. Macrocephaly is above +2 SD.
- Fontanelles: posterior closes at 6-8 weeks (by 2-3 months at the latest), **anterior at 12-18 months** (normal range 9-18 months; Nelson accepts up to 24 months).

Bedside formulae

Quantity	Formula
Weight, 3-12 months	(age in months + 9) / 2 kg, so 3 months gives 6 kg
Weight, 1-6 years	2 x age + 8 kg
Weight, 7-12 years	(7 x age - 5) / 2 kg
Height, up to 12 years	6 x age + 77 cm
Mid-parental height	boys (father + mother + 13) / 2; girls (father + mother - 13) / 2

- Upper:lower segment ratio is about 1.7 at birth, 1.3 at 3 years and **1.0 at 7-10 years**. Standing height is about 0.7 cm less than recumbent length.
- **Arm span** is an accurate substitute for height when a child cannot stand or be measured; it equals height at about 10 years and exceeds it in later childhood (the ratio matters in Marfan syndrome).
- **Fetal growth** is driven mainly by **insulin and the insulin-like growth factors (IGF-2, IGF-1)**, given an adequate supply of nutrients across the placenta. **Growth hormone** is present in the fetus but has little effect on fetal size (babies with GH deficiency have near-normal birth length); it becomes the main driver of growth after the first months. Thyroid hormone is essential for fetal brain development but not for fetal size; glucocorticoids mature organs (lung, gut) rather than promote growth.

Short stature

	Familial	Constitutional delay
Family history	short parents	a late-maturing parent possible
Growth velocity	normal	normal
Bone age	equals age	delayed
Puberty	on time	delayed
Final height	short	normal

Milestones

Domain	Milestone and age
Gross motor	head holding 3 months; sits without support 8 months; stands with support 9; **walks alone 12-15 months (normal range 9-18)**; runs 18 months; tricycle 3 years; hops 4 years
Fine motor	transfers 7 months; mature pincer 12 months; scribbles, copies a line 2 years; circle 3 years; cross and rectangle 4 years; triangle 5 years
Social and language	**social smile 2 months** (a reflex smile in the first weeks does not count); stranger anxiety 7 months; waves bye-bye 9 months; two-word phrases 2 years; three-word sentences 3 years

- A child of 15 months who is not yet walking but sits, crawls, pulls up, uses a pincer grasp, speaks and has a normal examination is normal: reassure and review. Creatine kinase and imaging are for regression, weakness or abnormal tone.
- **Corrected age** for a preterm infant = chronological age minus weeks born early. A baby born at 32 weeks (8 weeks early) who is 6 months old is assessed as 4 months. Correct until about 2 years.
- **Cube tower**: 2 cubes at 15 months, 3 at 18 months (with walking backwards and scribbling), **about 6 at 2 years**, 9 or a three-cube bridge at 3 years. Recognises the mother at about 3 months.

Primitive reflexes

- **Moro reflex**: appears at 28-32 weeks, fades by **4-6 months**. Persistence beyond 6 months suggests a neurological disorder such as cerebral palsy. **An asymmetrical response** points to a lesion on the weaker side: Erb palsy, clavicle fracture, hemiplegia or shoulder dislocation; a weak or absent response on both sides goes with prematurity, depression from drugs or asphyxia, or severe illness.
- The rooting and palmar grasp reflexes go by 3-4 months and the **asymmetric tonic neck reflex by 4-6 months**. Postural reactions appear as these fade: the **Landau reflex appears at 3-10 months** and goes by about 2 years; the **parachute reflex appears at 8-9 months and persists for life**. Plantar grasp lasts until 9-12 months.
- In cerebral palsy, **spastic CP is the commonest type overall** (about 70-80%). Preterm birth goes with **spastic diplegia** (periventricular leukomalacia); in term-born children spastic hemiplegia (perinatal stroke) and quadriplegia are commoner, and the **dyskinetic (choreoathetoid) type goes with term asphyxia affecting the basal ganglia and with kernicterus**.

Breath-holding spells

- Between 6 months and 5 years: a provoked cry (anger, frustration, pain), breathing stops in expiration, then cyanosis (or pallor in the pallid type, a vagal bradycardia or brief asystole), limpness and sometimes a few jerks, with full recovery in under a minute and a normal examination. Not epilepsy and harmless; **reassure, and look for and treat iron deficiency, which reduces the spells** even without anaemia (Nelson). No antiepileptic, EEG or cardiac work-up for the typical cyanotic spell; an ECG is reasonable for frequent pallid spells (long QT).

Intelligence

- Classical **IQ = mental age / chronological age x 100**. An 8-year-old with IQ 50 has a mental age of 4 years and manages tasks of a 4-year-old (copying a cross or rectangle). IQ 75 at 8 years gives a mental age of 6.

Teeth

	Primary (milk)	Permanent
Starts	about 6 months	about 6 years
First tooth	**lower central incisor**	**first molar** (behind the last milk molar; replaces no tooth)
Number	20	32
Formula per quadrant	2 incisors, 1 canine, 0 premolars, 2 molars	2, 1, 2, 3
Last	second molar	third molar

- Primary dentition is complete by 2-2.5 years. Delayed eruption is no tooth by 13 months. In the mixed dentition, permanent teeth present are about (age in years - 5) x 4.

Puberty

- Girls: thelarche first, then pubic hair, the growth spurt (peak height velocity comes early, before menarche), then menarche about 2 years after breast buds. Boys: testicular enlargement (4 mL or more) is first; the growth spurt comes late.
- **Precocious puberty**: secondary sex characters before **8 years in girls, 9 years in boys**. In girls it is usually idiopathic and central (early GnRH pulse generator activation), with advanced bone age and a growth spurt. Premature thelarche is isolated breast budding without growth acceleration or advanced bone age. In boys, organic causes (CNS lesion) are commoner. McCune-Albright syndrome: polyostotic fibrous dysplasia, café-au-lait spots and peripheral precocity.
- Delayed puberty: no changes by 13 years in girls, 14 in boys. Primary amenorrhoea: no menses by 13 without secondary characters, or by 15 with them; commonest cause Turner syndrome.

Enuresis

- Bedwetting is normal until about **5 years** (textbooks say 5-6): under 5, reassure; no drugs and no alarm. From 5 years (NICE CG111), behavioural measures and an **enuresis alarm** are first-line; **desmopressin** is the drug of choice when rapid or short-term control is needed or an alarm is unsuitable; imipramine is rarely used now (specialist care only) and oxybutynin is added for daytime bladder symptoms.

Neonatology

Neonatal jaundice

- **Physiological**: unconjugated, appears after 24 hours (day 2-3), peaks about day 3-4 in term babies and stays below about 12 mg/dL. Causes: more red-cell breakdown, immature UGT1A1 (about 1% of adult activity at term, reaching adult levels by about 6-14 weeks) and a sterile gut that favours enterohepatic recycling.
- **Jaundice in the first 24 hours is always pathological**; commonest cause **haemolytic disease of the newborn** (Rh or ABO incompatibility, G6PD deficiency). Jaundice beyond 2 weeks (3 in preterms) needs a split bilirubin.
- Conjugated hyperbilirubinaemia: the classic definition is direct bilirubin above 1 mg/dL when total is under 5, or above 20% of total; **current practice (NASPGHAN/ESPGHAN 2017): a direct or conjugated bilirubin above 1.0 mg/dL is abnormal whatever the total**. It is never physiological.

	Breastfeeding (failure) jaundice	Breast milk jaundice
Onset	first week	after the first week (from about day 5-7), peaks at 2-3 weeks, may last up to 12 weeks
Mech-anism	low intake, dehydration, slow stooling, more enterohepatic circulation	factors in milk (beta-glucuronidase, free fatty acids) deconjugating bilirubin in the gut
Baby	poor feeding, weight loss, few wet nappies	thriving, normal examination, normal stool and urine colour
Action	improve feeding	continue breastfeeding; benign

- **Kernicterus** (bilirubin encephalopathy): unconjugated bilirubin not bound to albumin enters the brain and stains the **basal ganglia (globus pallidus)**, subthalamic nucleus and brainstem nuclei. Survivors have athetoid cerebral palsy, sensorineural deafness, upward-gaze palsy and dental enamel hypoplasia. Risk rises with prematurity, acidosis, sepsis and drugs that displace bilirubin (sulphonamides, ceftriaxone).
- **Phototherapy**: blue light; bilirubin absorbs most strongly at about **460 nm**, and AAP 2022 specifies narrow-band light at **460-490 nm** (LED). It converts bilirubin to water-soluble isomers, chiefly **lumirubin (structural isomerisation)**, excreted without conjugation; photo-oxidation contributes least. Start and exchange thresholds depend on age in hours, gestation and neurotoxicity risk factors (AAP 2022 revised the nomograms and raised the thresholds for infants of 35 weeks

or more); exchange transfusion for failed phototherapy or signs of encephalopathy.

- Phenobarbitone induces UGT but is not used to treat neonatal jaundice in practice (its use is Crigler-Najjar type II). IV immunoglobulin is used in isoimmune haemolysis when bilirubin is rising despite phototherapy.
- Heme is broken to biliverdin and then unconjugated bilirubin (water-insoluble, albumin-bound), conjugated by UDP-glucuronosyl transferase in the liver; gut bacteria make urobilinogen, which is partly reabsorbed (enterohepatic circulation), excreted as urobilin in urine or as stercobilin in stool.
- Haemolysis gives unconjugated hyperbilirubinaemia with normal-coloured urine, because unconjugated bilirubin is protein-bound and not filtered; dark urine in haemolysis is haemoglobinuria. "Haemolysis raises conjugated bilirubin" is the false statement in "all except" questions. Obstructive jaundice has conjugated bilirubin, bilirubinuria, pale stools and pruritus; hepatic jaundice has both fractions raised.
- Hereditary: Gilbert (mild, unconjugated, after puberty, normal biopsy), Crigler-Najjar I (no UGT1A1, severe, liver transplant) and II (phenobarbitone-responsive); Dubin-Johnson (conjugated, black liver) and Rotor.
- **Biliary atresia**: progressive obliteration of the extrahepatic ducts; a well baby with jaundice, **pale stools and dark urine** in the first weeks, hepatomegaly. Ultrasound (absent or abnormal gallbladder, triangular cord sign) first; HIDA scan shows no excretion into the gut; operative cholangiogram is the gold standard. **Kasai portoenterostomy ideally before 8 weeks (60 days)**: the chance of restoring bile flow falls steeply after that, and after 3 months a transplant is more likely; most still need transplant. It is the commonest cause of paediatric liver failure and liver transplantation.

Neonatal seizures

- Commonest cause: **perinatal hypoxic-ischaemic encephalopathy**, then intracranial haemorrhage, infection and metabolic disturbance (hypoglycaemia, hypocalcaemia, hyponatraemia).
- Seizure type: **subtle seizures** (eye deviation, lip smacking, cycling, apnoea) are the commonest in the classical (Volpe) teaching; focal clonic seizures carry the best outlook and myoclonic the worst. ILAE 2023 classifies by EEG and calls many clinical-only events non-epileptic; the subtle-commonest answer is the exam one. Timing: HIE in the first 24 hours; early hypocalcaemia (preterm, asphyxia, infant of a diabetic mother) at 24-72 hours; **late hypocalcaemia (phosphate-rich cow milk or formula) at 5-10 days, with a good prognosis**; "fifth-day fits" are benign idiopathic neonatal seizures, not hypocalcaemia.
- Correct glucose and calcium first. **Phenobarbitone is first-line** (confirmed by the ILAE neonatal seizure guideline, 2023, whatever the cause); phenytoin, levetiracetam, midazolam or lidocaine are second-line. Valproate and carbamazepine are not used. EEG (ideally continuous or amplitude-integrated) is needed, since many neonatal seizures are electrographic only.

Resuscitation and Apgar

- Primary aim: establish spontaneous breathing (effective ventilation). **Compressions only if heart rate stays below 60 after 30 seconds of effective ventilation**, at **3 compressions : 1 breath** (90 + 30 per minute), with oxygen increased to 100% (AHA/AAP Neonatal Resuscitation 2020, kept in the 2025 update; the 2025 update widens the ventilation rate to 30-60 breaths a minute). Delayed cord clamping (at least 30-60 seconds) for babies who do not need resuscitation at once.
- Start term babies on air (21%) and preterm babies under 35 weeks on 21-30%. Target pre-ductal (right hand) SpO2 about 60-65% at 1 minute, 80-85% at 5 minutes and 85-95% at 10 minutes. Never delay resuscitation to wait for the Apgar score.
- **Apgar** components: appearance, pulse, grimace, activity, respiration (effort), each 0-2 (total 10). **7-10 reassuring, 4-6 moderately abnormal, 0-3 low** (AAP/ACOG). Birth weight and respiratory rate are not components. The 1-minute score reflects intrauterine condition, the 5-minute score the response to resuscitation; neither predicts neurological outcome alone.
- Neonatal blood volume is about 85 mL/kg.
- **Prostaglandin E1** (alprostadil) keeps the duct open in duct-dependent lesions (pulmonary atresia, critical PS, critical coarctation, hypoplastic left heart, TGA); indomethacin or ibuprofen closes it.

Respiratory distress syndrome

- Surfactant deficiency: incidence rises as gestation falls. Other risks: male sex, caesarean without labour, perinatal asphyxia and **maternal diabetes**; maternal hypertension and prolonged rupture of membranes accelerate maturation.
- Surfactant: synthesised by type II pneumocytes from about 20 weeks, appears in amniotic fluid at 28-32 weeks; main component dipalmitoyl phosphatidylcholine (lecithin); SP-B is the most essential protein. **Lecithin : sphingomyelin ratio of 2 or more** = mature lungs; phosphatidylglycerol is more specific; shake (foam) test is a bedside test. Fetal hyperinsulinaemia opposes cortisol and delays surfactant synthesis, so infants of diabetic mothers get RDS.
- Picture: grunting, retractions, flaring, cyanosis within hours; chest X-ray **ground glass (reticulogranular), air bronchograms, low volumes**. Treatment: CPAP from birth, **intratracheal surfactant** (poractant, beractant), oxygen titrated to SpO2 90-94% (European Consensus 2022; older texts 90-95%). **INSURE** = intubate, surfactant, extubate to CPAP; modern practice also uses less invasive administration (LISA).
- **Antenatal corticosteroids** (24-34 weeks): betamethasone 12 mg IM, 2 doses 24 hours apart (or dexamethasone 6 mg 12-hourly, 4 doses). They reduce RDS, NEC, intraventricular haemorrhage and mortality, but not BPD; chorioamnionitis is a contraindication in older teaching.
- **Transient tachypnoea (TTN)**, **meconium aspiration (MAS)** and **hyaline membrane disease (HMD)** compared:

	TTN	MAS	HMD
Baby	term or near term, often elective caesarean (no labour, lung fluid not cleared)	post-term or stressed, meconium-stained liquor	preterm
Onset and course	within 1-2 hours; settles in 24-72 hours	at birth	within minutes to hours; worst at 48-72 hours in the untreated course
X-ray	streaky perihilar markings, **fluid in the fissure**, hyper-inflation	**coarse patchy opacities, air trapping**, flat diaphragms	**ground glass, air bronchograms, low volumes**

	TTN	MAS	HMD
Complication	usually none	pneumothorax, **persistent pulmonary hypertension** (the main threat)	bronchopulmonary dysplasia, air leak

- **Meconium-stained, non-vigorous baby**: the classic answer is tracheal suction under direct laryngoscopy before breathing; NRP 2015 and 2020 (kept in 2025) no longer advise routine intubation for suction: start the usual steps and ventilation within the first minute, suctioning the trachea only for obstruction. Vigorous babies are never suctioned.
- **Congenital diaphragmatic hernia** (Bochdalek, left posterolateral, commonest): scaphoid abdomen, bowel sounds in the chest, mediastinal shift; **no bag-and-mask ventilation** (it inflates the bowel in the chest and compresses the hypoplastic lungs): intubate at once and pass an orogastric tube; pulmonary hypoplasia and pulmonary hypertension decide survival; surgery after stabilisation. Morgagni hernias are anterior and present late.
- **Bilateral choanal atresia**: newborns breathe through the nose, so cyanosis with quiet breathing is **relieved by crying**; a catheter will not pass either nostril; CT confirms; an oral airway (McGovern nipple) then endoscopic repair; associated with CHARGE. Contrast tracheo-oesophageal fistula (choking and cyanosis with feeds) and laryngomalacia (stridor worse when crying).
- Apnoea of prematurity: treat with caffeine. **Bronchopulmonary dysplasia**: oxygen need at 28 days or 36 weeks corrected. Congenital pulmonary alveolar proteinosis is SP-B deficiency. Neonatal RDS affects type II pneumocytes; adult ARDS is diffuse alveolar damage.

Infant of a diabetic mother

- Macrosomia (maternal diabetes is the commonest cause; the insulin-sensitive trunk and shoulders grow, the brain does not, so **shoulder dystocia is the commonest birth complication**, with brachial plexus injury and clavicle fracture), **hypoglycaemia** (fetal hyperinsulinism continues after the glucose supply stops; babies have plenty of glycogen), **hypocalcaemia (and hypomagnesaemia) at 24-72 hours** from functional hypoparathyroidism, so a jittery or seizing baby with a normal glucose needs calcium and magnesium measured, and low magnesium corrected or the calcium will not stay up; polycythaemia, jaundice, RDS, **hypertrophic (septal) cardiomyopathy**, which regresses.
- Anomalies: commonest congenital heart lesion VSD; **most specific cardiac lesion TGA**; commonest neurological anomaly neural tube defect; **most specific of all: caudal regression (sacral agenesis)**; small left colon syndrome.
- Gestational diabetes: screen at 24-28 weeks. Classic two-step (US): 50 g challenge, a 1-hour value of 140 mg/dL or more (130 in some centres) leads to a 100 g 3-hour OGTT; two raised values diagnose. **WHO 2013 and IADPSG one-step**: 75 g 2-hour OGTT, any one of fasting 92 mg/dL or more, 1-hour 180 or more, 2-hour 153 or more.

Scalp swellings

	Caput succedaneum	Cephalohaematoma
Layer	subcutaneous oedema	**subperiosteal** blood
Crosses suture	yes	**no**, limited by periosteal attachment at sutures
Appears	present at birth	12-24 hours, may enlarge on day 2
Resolves	**2-3 days**	2 weeks to 3 months (may calcify)
Link	none	linear skull fracture, jaundice

Subgaleal haemorrhage crosses sutures and can cause shock.

Sepsis and meningitis

- Early-onset (under 72 hours): vertical, from the genital tract. **Group B streptococcus (GBS)** is the commonest cause of early sepsis and meningitis in high-income countries, with E. coli second and Listeria rarer. In low-resource settings including Nepal, Gram-negative rods (Klebsiella, E. coli) and coagulase-negative staphylococci account for much of hospital-onset disease; read the setting in the question.
- Early signs: poor feeding, lethargy, temperature instability, grunting, tachypnoea above 60/min. Best first step is **blood culture then IV ampicillin plus gentamicin** at once. Screening (any 2 of: total leucocytes under 5,000; ANC under 1,800; immature:total neutrophil ratio above 0.2; micro-ESR above 15 mm; CRP above 1 mg/dL) supports, never delays, treatment.
- GBS prevention: intrapartum penicillin for carriers (universal screening at 36-37 weeks in the US, ACOG 2020; risk-based in the UK). It prevents early-onset but not late-onset GBS disease. Late-onset sepsis (after 72 hours) is mostly hospital-acquired: coagulase-negative staphylococci in preterm infants with lines, S. aureus, Klebsiella and other Gram-negative rods.

Hypoxic-ischaemic encephalopathy and neonatal causes of death

- Preterm: **periventricular white matter** (periventricular leukomalacia, watershed). Term: parasagittal cortex and subcortical white matter; basal ganglia and thalami in acute profound insults. Sarnat staging. **Therapeutic hypothermia** (33.5 C for 72 hours, started within 6 hours) for moderate to severe HIE is the textbook answer and standard in high-income countries (it lowers death or disability). In low- and middle-income settings the **HELIX trial** (India, Sri Lanka, Bangladesh; Lancet Glob Health 2021) found that cooling did not reduce death or disability and **increased mortality**, so its benefit there is not established and its authors advise against routine use.
- Commonest causes of neonatal death worldwide: **prematurity**, then birth asphyxia, then sepsis, then congenital anomalies. After the neonatal period (1-59 months): pneumonia, diarrhoea, injuries, malaria.
- Eponyms: Bell (NEC), Sarnat (HIE), Ballard (gestational age), Brazelton (behaviour), Silverman-Andersen (respiratory distress in the preterm), Downes (respiratory distress, mainly term).

Prophylaxis at birth: vitamin K and hepatitis B

- **Vitamin K deficiency bleeding**: low stores, little vitamin K in breast milk and a sterile gut give low factors II, VII, IX and X (prolonged PT, then aPTT; normal platelets). **Early** (first 24 hours: maternal phenytoin, phenobarbitone, rifampicin, warfarin), **classic** (days 2-7: umbilical, gut, circumcision bleeding), **late** (2-12 weeks, up to 6 months: exclusively breastfed babies, cholestasis; often **intracranial**). Prevention: **1 mg intramuscularly once at birth** (AAP 2022: 1 mg within 6

hours of birth above 1,500 g; 0.3-0.5 mg/kg at 1,500 g or less); oral regimens need repeated doses and fail more often.

- **Hepatitis B, baby of an HBsAg-positive mother** (risk highest when HBeAg-positive or the viral load is high): **vaccine and hepatitis B immunoglobulin (0.5 mL) at separate sites, ideally within 12 hours of birth** (the vaccine always within 24 hours), then complete the series and test the baby for HBsAg and anti-HBs after it. The immunoglobulin neutralises virus at once, before the vaccine's active response exists; together they prevent about 90-95% of transmission. Current practice adds **tenofovir to the mother from the second or third trimester when her HBV DNA is 200,000 IU/mL or more or she is HBeAg-positive** (WHO 2024; WHO 2024 also allows it for every HBsAg-positive pregnant woman where those tests are unavailable). Breastfeeding is not stopped.

Congenital infections and drugs in pregnancy

- **CMV** is the commonest congenital infection and the commonest non-genetic cause of sensorineural hearing loss: microcephaly, **periventricular calcification**, petechiae, hepatosplenomegaly, chorioretinitis. Proof is virus in the baby **within the first 3 weeks** (later positives may be postnatal): **urine or saliva PCR**; infant IgG is maternal. **Valganciclovir for 6 months** in symptomatic babies improves hearing and development. **Toxoplasmosis**: hydrocephalus, chorioretinitis, **diffuse (scattered) calcification**; spiramycin to the mother before fetal infection, pyrimethamine-sulfadiazine once it is proved. Rubella (Gregg triad) is under the exanthems; varicella in early pregnancy gives limb scarring and hypoplasia; congenital syphilis snuffles, rash, osteochondritis, later Hutchinson teeth.
- Drugs: warfarin (nasal hypoplasia, stippled epiphyses), lithium (Ebstein anomaly), valproate (neural tube defect), phenytoin (fetal hydantoin syndrome), isotretinoin (ear, heart, brain), ACE inhibitors (renal failure, oligohydramnios), tetracycline (stained teeth); chloramphenicol given to the newborn causes the grey baby syndrome.

Growth restriction, NEC and others

- **Asymmetrical IUGR** (late placental insufficiency): blood is redirected to the brain, so **head circumference is spared** while weight and abdomen are small; ponderal index below 2. Symmetrical IUGR (early insults, infection, chromosomal): head, length and weight all small.
- **Necrotising enterocolitis**: prematurity, formula feeds, PDA, indomethacin. Bell stages: IIA **pneumatosis intestinalis** (pathognomonic; definite NEC), IIB portal venous gas, IIIA ascites, IIIB pneumoperitoneum (perforation).
- Neonatal vaginal bleeding (day 2-7) is a **withdrawal bleed after falling maternal oestrogen**: reassure.
- **Hypothermia** (WHO, axillary): cold stress 36.0-36.4 C, moderate 32.0-35.9 C, severe below 32 C; normal 36.5-37.5 C.
- Benign newborn findings: milia, miliaria, **erythema toxicum** (eosinophils on smear), transient neonatal pustular melanosis (neutrophils), Mongolian spots, salmon patch (stork bite), acrocyanosis, Epstein pearls, mastitis neonatorum, hymenal tags. Infantile (strawberry) haemangioma grows over the first months, then involutes slowly (most of the regression by about 4 years; classic teaching 50% by 5, 90% by 9); **oral propranolol** is the treatment of choice when treatment is needed (vision, airway, ulceration, disfigurement); port-wine stain is lifelong (Sturge-Weber). Meconium passes within 48 hours (meconium plug and Hirschsprung disease if delayed); urine within 24 hours.
- **Kangaroo mother care**: skin-to-skin contact for stable low-birth-weight babies, with exclusive breastfeeding and early discharge; it **shortens, not lengthens, hospital stay**, and lowers sepsis, hypothermia and mortality (WHO 2022 now recommends starting immediately after birth for small unstable babies as well).

Nutrition and malnutrition

Breastfeeding

- Start within 1 hour of birth, after caesarean too as soon as the mother is able (WHO/UNICEF Baby-friendly Initiative; older texts allowed up to 4 hours after caesarean); **exclusive for 6 months**, then complementary foods with continued breastfeeding to 2 years or beyond.
- **Hind milk**, late in a feed, is richer in fat (and energy) than foremilk, so a baby should finish one breast before the other. Colostrum has more protein, minerals and immunoglobulin and less fat and sugar than mature milk.
- The **predominant immunoglobulin in milk is secretory IgA**; it protects the gut and respiratory mucosa. IgG crosses the placenta and gives passive immunity at birth.
- Human versus cow's milk: human milk has **more lactose** (about 7 g/dL against 4.8), less protein and minerals (protein about 1.1 against 3.3 g/dL; calcium about 0.3 g/L against 1.2 g/L, so a lower renal solute load), casein : whey 40 : 60, better calcium : phosphate ratio (2 : 1), more taurine, vitamins A and C, and better iron bioavailability though total iron is low. Cow's milk has more casein, protein and minerals. Vitamin D (then K) is the vitamin most lacking in breast milk; it is a poor source of iron.

Classifying malnutrition

- Low weight-for-height = **wasting** (acute; best indicator of current state); low height-for-age = **stunting** (chronic); low weight-for-age = underweight (mixed).
- Z-scores: mild -1 to -1.99, moderate -2 to -2.99, severe below -3.
- **WHO severe acute malnutrition (6-59 months)**: weight-for-height below -3 SD, **MUAC below 11.5 cm**, or bilateral pitting oedema. Moderate acute malnutrition: MUAC 11.5 to under 12.5 cm. MUAC is independent of age.

Marasmus versus kwashiorkor

	Marasmus	Kwashiorkor
Deficit	energy	protein relative to energy
Age	infancy	after weaning, 1-3 years
Oedema	none	**bilateral pitting oedema**
Look	wasted, "old man" face, alert, hungry	moon face, apathy, irritability, **poor appetite**
Skin and hair	normal	flaky-paint dermatosis, sparse light hair, flag sign
Liver	normal	fatty enlarged liver
Prognosis	better	worse, more infections

- Oedema in kwashiorkor: the exam answer is **low plasma albumin (low oncotic pressure)**; current understanding adds that albumin alone does not explain it (oedema can clear before albumin rises), and **oxidative stress with glutathione depletion**, leaky capillaries, and the gut microbiota

contribute. Return of appetite is the first sign of recovery. Marasmic-kwashiorkor combines both.

- Unreliable signs in malnourished children (skin turgor, mental state, tongue); the most reliable is urine output. Use low-sodium **ReSoMal** (sodium 45, potassium 40 mmol/L, extra magnesium, zinc, copper) for rehydration, and give fluid slowly by mouth; IV fluid risks heart failure.

Management of severe acute malnutrition (WHO 2013, updated 2023)

- **Uncomplicated SAM** (alert, good appetite, passes the appetite test, no danger signs, no severe oedema) is treated as an outpatient with **ready-to-use therapeutic food** (about 150-200 kcal/kg/day), oral amoxicillin and follow-up (WHO 2013, kept in the 2023 wasting guideline).
- **Complicated SAM** (failed appetite test, +++ oedema, danger signs, infection, severe anaemia) goes to hospital: stabilise (hypoglycaemia, hypothermia, dehydration, infection, electrolytes) with F-75, then rehabilitate with F-100 or RUTF.
- **Rehydration in SAM** uses **ReSoMal** (sodium 45, potassium 40 mmol/L; table under Diarrhoea): the body holds excess sodium and has lost potassium and magnesium, and the heart is small and weak, so standard ORS or fast IV fluid risks overload. Give 5 mL/kg every 30 minutes for 2 hours, then 5-10 mL/kg an hour, orally or by nasogastric tube; IV fluid only for shock. **Hypoglycaemia in SAM is below 3 mmol/L (54 mg/dL)**: 50 mL of 10% glucose or F-75, then feeds every 2 hours day and night.
- **Iron is withheld during stabilisation**: transferrin is low, free iron fuels bacterial growth and oxidant injury; start in rehabilitation once appetite returns and infection is controlled.

Micronutrients

- **Vitamin A deficiency**: night blindness is the earliest symptom; conjunctival xerosis is the earliest sign; **Bitot spots** are the most specific sign; then corneal xerosis and keratomalacia (irreversible). Prophylaxis: 6-11 months 100,000 IU, 12-59 months 200,000 IU, every 4-6 months. Single doses up to 50,000 IU are safe under 6 months (bulging fontanelle is a side effect).
- Zinc: 20 mg daily for 10-14 days (10 mg below 6 months) in diarrhoea.
- **Zinc deficiency**: **acrodermatitis enteropathica** (autosomal recessive, SLC39A4) shows when breast milk, whose zinc is better absorbed, is stopped: perioral, perianal and acral scaly dermatitis, alopecia, diarrhoea, irritability; low serum zinc and alkaline phosphatase; **lifelong oral zinc** cures it. Acquired zinc deficiency gives the same picture. **Selenium**: Keshan disease (cardiomyopathy); Kashin-Beck disease (osteoarthropathy, selenium with iodine deficiency). **Niacin**: pellagra (dermatitis, diarrhoea, dementia, death; Casal necklace), in maize-eaters and Hartnup disease.
- **Scurvy** (vitamin C; infants of 6-24 months on boiled or processed milk): irritability, **pseudoparalysis** with frog-leg posture and tender legs from subperiosteal bleeding, swollen bleeding gums once teeth have erupted, scorbutic rosary (sharp, unlike the rounded rickety one). X-ray: ground-glass osteopenia, **Frankel white line** at the metaphysis, the lucent **Trummerfeld zone** beneath it, **Pelkan spurs**, and the **Wimberger ring** round the epiphysis. Rickets shows cupping and fraying instead; lead gives dense metaphyseal bands without the ring (Wimberger's corner sign, a metaphyseal erosion, is congenital syphilis).
- Albendazole (deworming, single oral dose): 12-23 months 200 mg, 24-59 months 400 mg.
- Rickets (nutritional): low vitamin D, secondary rise in PTH; low-normal calcium, low phosphate, high ALP; look for craniotabes (earliest sign, in infants), rickety rosary, widened wrists, bowed legs; X-ray: cupping, fraying and splaying of the metaphysis (wrist best seen). Treatment (Global Consensus 2016), for at least 12 weeks, with calcium: vitamin D 2,000 IU daily under 12 months (or a single 50,000 IU dose from 3 months), 3,000-6,000 IU daily at 1-12 years (or 150,000 IU once), 6,000 IU daily over 12 years (or 300,000 IU once); then maintenance 400-600 IU daily. Refractory types are in the metabolic section.

Diarrhoea and dehydration

Causes

- **Rotavirus** is the commonest cause of severe diarrhoea in children (secondary lactose intolerance can follow; EIA antigen test is the most specific); enterotoxigenic E. coli is the classic answer for the commonest bacterial cause in developing countries (the GEMS study, 2013, added Cryptosporidium and Shigella among the leading causes under 5); norovirus is the commonest cause in adults and, where rotavirus vaccine is routine, now in children too. Rotavirus vaccine is linked with a small risk of intussusception.
- Cholera toxin raises cyclic AMP and drives chloride secretion, but the **SGLT1 sodium-glucose co-transporter stays intact**, so glucose-containing ORS works even in secretory diarrhoea; absorbed sodium and glucose pull water along.

WHO low-osmolarity ORS

	Standard (old)	Reduced osmolarity	ReSoMal
Sodium (mmol/L)	90	**75**	45
Chloride	80	**65**	70
Potassium	20	20	40
Citrate	10	10	7
Glucose	111	75	125
Osmolarity (mOsm/L)	311	**245**	300

Low-osmolarity ORS lowers stool output, vomiting and need for IV fluid. Powder per litre: sodium chloride 2.6 g, glucose 13.5 g, potassium chloride 1.5 g, trisodium citrate 2.9 g.

Assessing dehydration (WHO/IMCI)

Grade	Signs	Plan
None	alert, normal eyes, drinks normally, skin pinch goes back at once	**Plan A**: more fluids at home; ORS after each loose stool (under 2 years 50-100 mL, 2-10 years 100-200 mL; about 10 mL/kg), zinc, continue feeding
Some	**restless or irritable, sunken eyes, drinks eagerly, skin pinch slow** (under 2 seconds)	**Plan B**: ORS **75 mL/kg over 4 hours**, then reassess

Grade	Signs	Plan
Severe	lethargic or unconscious, cannot drink, sunken eyes, skin pinch very slow (over 2 seconds)	**Plan C**: IV fluid

- **Plan C**: Ringer lactate (or normal saline). Under 12 months: **30 mL/kg in the first hour, then 70 mL/kg over 5 hours**. Age 12 months to 5 years: 30 mL/kg in 30 minutes then 70 mL/kg over 2.5 hours. Reassess often. Ringer lactate is the fluid of choice; dextrose alone is wrong because it supplies no sodium and risks hyponatraemia.
- Maintenance fluid (Holliday-Segar, 24 hours): 100 mL/kg for the first 10 kg, 50 mL/kg for the next 10, 20 mL/kg for the rest. The 4-2-1 rule gives it per hour (4, 2 and 1 mL/kg/hour for each band).
- **Zinc**: 20 mg/day for 10-14 days above 6 months, 10 mg/day below 6 months; it shortens the episode and lowers recurrence for 2-3 months. Antibiotics are not routine; they are for dysentery (blood in stool) and cholera.
- Continue feeding and breastfeeding throughout.

Hypernatraemic dehydration

- Brain cells make idiogenic osmoles to defend volume. Correcting sodium quickly pulls water into the brain: **cerebral oedema and seizures**. Lower sodium by no more than about 10-12 mmol/L per day.

Amoebic versus bacillary dysentery

	Amoebic (E. histolytica)	Bacillary (Shigella)
Onset	**gradual**	abrupt
Fever, toxicity	uncommon	common
Stools	6-8/day, copious, **offensive**, dark red, acidic, red cells in clumps, few pus cells, trophozoites	over 10/day, small, odourless, bright red, alkaline, many pus cells, tenesmus
Complications	liver abscess	convulsions, HUS, dehydration

The most sensitive test for invasive amoebiasis is antibody detection (ELISA). Shigella is treated with ciprofloxacin (WHO first-line), with azithromycin or ceftriaxone as alternatives where resistance is common; amoebiasis with metronidazole then a luminal agent (paromomycin, diloxanide).

Persistent diarrhoea and lactose intolerance

- **Persistent diarrhoea** (WHO): an episode that begins acutely and lasts **14 days or more** (acute under 14 days; chronic usually means a non-infectious cause over weeks to months). Manage with continued feeding and breastfeeding, zinc, micronutrients and nutritional rehabilitation, reducing lactose (yoghurt, a low-lactose diet) only when milk clearly worsens it.
- **Secondary lactose intolerance** is the commonest disaccharidase deficiency after gastroenteritis: unabsorbed lactose is fermented in the colon, giving frothy, acidic stool (pH below 5.5) with **reducing substances**; a short trial of a lactose-free or low-lactose feed settles it, while breastfeeding continues.

Respiratory infections

Pneumonia and fast breathing (WHO IMCI)

- **Fast breathing**: under 2 months 60/min or more; **2-11 months 50/min or more**; 12-59 months 40/min or more. Count for a full minute in a calm child (crying inflates the rate).
- **Classic IMCI (the exam answer)**: fast breathing alone = pneumonia (oral antibiotic at home); **chest indrawing = severe pneumonia** (refer, injectable antibiotic); danger signs (unable to drink, persistent vomiting, convulsions, lethargy, stridor in a calm child, severe malnutrition) = very severe disease.
- **Current WHO classification (2014, in IMCI since)**: fast breathing **or chest indrawing** = pneumonia, treated with **oral amoxicillin** at home (40 mg/kg twice daily; 3 days for fast breathing, 5 days for indrawing); danger signs or SpO2 below 90% = severe pneumonia (admit, oxygen, ampicillin or penicillin plus gentamicin). Young infants under 2 months with fast breathing or indrawing are still referred.
- Hypoxaemia (SpO2 below 90%) is the strongest predictor of death; pulse oximetry and oxygen are now part of IMCI.
- Commonest killers after the neonatal period (1-59 months): pneumonia, then diarrhoea.

Pneumonia organisms by age

Age	Commonest organisms
Neonate	GBS, E. coli and other Gram-negative rods, Listeria
3 weeks to 3 months	viruses (RSV); **Chlamydia trachomatis** (afebrile, staccato cough, conjunctivitis, eosinophilia); pertussis
4 months to 5 years	**viruses (RSV commonest)**; bacterial: **Streptococcus pneumoniae** (commonest bacterial cause at all ages after the neonatal period), Hib in the unvaccinated
5 years and older	**Mycoplasma pneumoniae**, S. pneumoniae, Chlamydophila pneumoniae

- **Staphylococcus aureus** pneumonia: rapid progression, **pneumatoceles**, pyopneumothorax and empyema (commonest cause of empyema in children in developing countries; pneumococcus in the West). Mycoplasma: cold agglutinins, a macrolide.

Croup, epiglottitis, bronchiolitis

	Croup	Bronchiolitis	Epiglottitis
Cause	**parainfluenza** (type 1)	**RSV**	Hib (rare now; S. pyogenes, S. aureus, pneumococcus in the vaccinated)
Age	6 months-3 years	under 2 years (peak 2-6 months)	2-7 years
Picture	barking cough, hoarse voice, inspiratory stridor, **steeple sign** on frontal neck film	coryza, wheeze, crackles, feeding difficulty	toxic child, high fever, drooling, sits forward (tripod), muffled voice, **thumb sign** on lateral neck film

- Mild croup (stridor only on crying, alert, drinking): **single oral dexamethasone 0.15 mg/kg** (0.15-0.6). Stridor at rest: add **nebulised adrenaline** (effect lasts about 2 hours, so observe). Antibiotics are not indicated.
- Epiglottitis: do not examine the throat with a spatula, lie the child flat, take blood first or send the child to radiology: any of these can turn partial obstruction into complete. **Airway first**,

in theatre with an anaesthetist and ENT surgeon ready for a surgical airway; then blood culture and IV ceftriaxone.

- Bronchiolitis: supportive (oxygen if SpO2 below 90-92%, fluids or tube feeds, nasal suction); bronchodilators, steroids, hypertonic saline and routine antibiotics do not help (AAP 2014, NICE 2021). Prevention for high-risk infants: palivizumab (monthly), and since 2023 **nirsevimab**, a single-dose long-acting monoclonal antibody for all infants in their first RSV season, and a **maternal RSV prefusion F vaccine** at 32-36 weeks.

Stridor in infancy

- **Laryngomalacia** is the **commonest cause of congenital stridor**: inspiratory, worse when supine, feeding or crying, better prone; omega-shaped epiglottis on laryngoscopy; most resolve by 12-24 months; supraglottoplasty for severe cases. Others: vocal cord palsy (second commonest), subglottic stenosis, subglottic haemangioma (biphasic stridor, propranolol), vascular rings.
- Retropharyngeal abscess (under 5): fever, neck stiffness, drooling, widened prevertebral soft tissue on lateral film.

Whooping cough (pertussis)

- Bordetella pertussis: catarrhal (most infectious), paroxysmal (whoop, post-tussive vomiting) and convalescent stages, about 2 weeks each ("100-day cough"); infants may have apnoea instead of whoop. **Absolute lymphocytosis** (can exceed 20,000/uL) because **pertussis toxin blocks lymphocyte homing out of the blood**. Diagnosis: PCR of a nasopharyngeal swab (culture on Bordet-Gengou or Regan-Lowe medium).
- Treatment: macrolide; **azithromycin is preferred in young infants** because erythromycin is linked to infantile hypertrophic pyloric stenosis. It shortens contagiousness more than the illness, and alters the course only when given in the catarrhal stage. **Most infectious in the catarrhal stage**, when it looks like a cold; untreated, infectivity lasts until about 3 weeks after the cough begins, treated until 5 days of the antibiotic. Commonest complication: pneumonia (and cause of death); also subconjunctival haemorrhage, hernia, seizures.
- Maternal Tdap in pregnancy (from 16-32 weeks, UK; 27-36 weeks, US) protects the young infant.

Diphtheria

- Corynebacterium diphtheriae exotoxin (inhibits EF-2): grey-white **pseudomembrane** that bleeds when removed, **bull neck**, myocarditis (second week, commonest cause of death) and palatal palsy, then peripheral neuropathy. Treat with **diphtheria antitoxin** at once on clinical suspicion, plus penicillin or erythromycin; erythromycin for contacts. Albert stain shows metachromatic granules; Loeffler serum slope for culture.

Asthma in children (GINA 2024)

- Diagnosis under 5 is clinical (recurrent wheeze, response to a trial of treatment); spirometry with reversibility (FEV1 rise of more than 12%) from about 5-6 years.
- **Age 6-11 years**: reliever SABA, with an ICS taken whenever the SABA is taken or daily low-dose ICS from step 1-2; step 3 low-dose ICS-LABA or medium-dose ICS, or **low-dose ICS-formoterol as maintenance and reliever (MART)**; refer at step 4-5 (anti-IgE, anti-IL-5, anti-IL-4R biologics). **Age 12 and over**: as in adults, **as-needed low-dose ICS-formoterol is the preferred reliever** (Track 1); SABA alone is no longer recommended.
- **Under 5 years**: as-needed SABA; daily low-dose ICS (or a leukotriene receptor antagonist) if wheezing is frequent or severe; spacer with face mask under 3-4 years.
- Acute severe asthma: oxygen to SpO2 94-98%, repeated salbutamol by spacer or nebuliser, ipratropium, oral prednisolone (1-2 mg/kg) or IV hydrocortisone; IV magnesium sulphate for poor response. A silent chest, cyanosis, exhaustion or a normal or rising PaCO2 signals life-threatening asthma.

Tuberculosis in children

- Usually **primary TB**: Ghon focus with hilar nodes (primary complex); young children (under 2) are at highest risk of **miliary TB and TB meningitis**. Contact with an adult case is the key history.
- Diagnosis: sputum is often unobtainable; use **gastric aspirate**, induced sputum or nasopharyngeal aspirate, and since WHO 2022 **stool and Xpert MTB/RIF Ultra**; tuberculin skin test (induration of 10 mm or more; 5 mm in HIV or severe malnutrition) or IGRA shows infection, not disease.
- Treatment: classic **2HRZE/4HR** (6 months; 12 months for TB meningitis and osteoarticular TB, 2HRZE/10HR). **WHO 2022: a 4-month regimen (2HRZ(E)/2HR) for non-severe TB in children aged 3 months to 16 years** (SHINE trial). Child contacts under 5 without disease get preventive therapy (6H, or 3HR or 3HP).
- BCG protects mainly against **disseminated and meningeal TB in young children**, not pulmonary TB in adults.

Foreign body aspiration

- Sudden choking then cough and wheeze in a toddler (peak 1-3 years; peanuts and seeds). Commonest site: the **right main bronchus** (wider, shorter, more vertical). With a **ball-valve** obstruction, air enters on inspiration but cannot leave, so the affected lung is hyperinflated, and on **expiration the mediastinum shifts away** from it (expiratory or lateral decubitus film). Complete obstruction gives collapse with shift towards the lesion. Chest film may be normal; **rigid bronchoscopy** is the treatment (and the diagnosis when suspicion is high).

Bronchiectasis and cystic fibrosis lungs

- Commonest site of post-infectious bronchiectasis: left lower lobe; **bilateral upper lobes in cystic fibrosis**; **central (proximal) bronchiectasis in allergic bronchopulmonary aspergillosis**; unilateral upper lobe in tuberculosis; right middle lobe after primary TB is Brock syndrome (middle lobe syndrome, from node compression). HRCT is the investigation of choice. Primary ciliary dyskinesia (Kartagener: situs inversus, sinusitis, bronchiectasis) is a cause in children.

Immunisation

Nepal's national schedule (Nepal's 2025 report to WHO and UNICEF)

As Nepal's Ministry of Health and Population reported it on the WHO/UNICEF Joint Reporting Form for 2025 (WHO Immunization Data portal, read 2026-11-26), beside WHO's recommendation. The National Immunization Programme is run by the Family Welfare Division, Department of Health Services.

Vaccine	Nepal (2025)	WHO recommendation (position papers; summary table 2023-2024)
BCG	birth	single dose at birth
Hepatitis B	in the pentavalent at 6, 10, 14 weeks; **no birth dose listed in the report**	**birth dose within 24 hours**, then 2-3 doses
DTwP-HepB-Hib (pentavalent)	**6, 10, 14 weeks**; no booster listed	3 primary doses from 6 weeks, 4 weeks apart; **3 boosters**: 12-23 months, 4-7 years and 9-15 years
OPV (bivalent)	**6, 10, 14 weeks**	birth dose where polio risk is high, plus 3 doses
IPV, fractional (fIPV, intradermal)	**14 weeks and 9 months** (6 and 14 weeks until 2022)	at least 2 IPV doses (full or fractional), the first from 14 weeks
Pneumococcal conjugate	**6 weeks, 10 weeks, 9 months** (2 + 1)	2 + 1 or 3 + 0
Rotavirus	**6 and 10 weeks** (2 doses)	2 or 3 doses from 6 weeks, as early as possible
Measles-rubella	**9 and 15 months**	MCV1 at 9 months (high transmission), MCV2 at 15-18 months
Japanese encephalitis (live attenuated)	**12 months**	single dose in endemic areas
Typhoid conjugate	**15 months**	single dose from 6 months in endemic areas
HPV	**single dose to girls in grade 6, or out-of-school girls of 10** (routine since the 2025 national campaign)	**single dose to girls aged 9-14** (WHO 2022); 1 or 2 doses acceptable
Td, pregnant women	2 doses a month apart in the first pregnancy (from the first visit), 1 in each later pregnancy	TT/Td for mothers not fully immunised, 5 doses for lifelong protection
Vitamin A	every 6 months from 6 months to 5 years	(supplement, not a vaccine)

- What has changed in Nepal: **fIPV moved to 14 weeks and 9 months** (from late 2022, reported from 2023); **HPV was added after a national campaign in February 2025** that vaccinated about 1.46 million girls aged 10-14 (94% coverage, WHO Nepal 2025) and is now given each year to the routine cohort.
- A question on "the national schedule" in Nepal's exam means Nepal's, above; a question on WHO's recommendation means the right-hand column.

Route and type

- **BCG is intradermal**, 0.05 mL under 12 months (0.1 mL older, WHO 2018), over the insertion of the deltoid (left upper arm in most programmes), producing a papule, a pustule at about 6 weeks and then a scar. Subcutaneous injection risks a cold abscess. BCG revaccination is not recommended.
- Measles and MR vaccines are subcutaneous (WHO now allows intramuscular too); hepatitis B, DTP, Td and PCV are intramuscular (anterolateral thigh in infants); fractional IPV is intradermal (one-fifth of a full dose); OPV and rotavirus are oral.
- **Live attenuated vaccines**: BCG, OPV, measles, MMR, rotavirus, varicella, yellow fever, JE (SA 14-14-2), oral typhoid (Ty21a), intranasal influenza. Avoid in pregnancy and severe immunodeficiency. Non-live: **hepatitis B (recombinant)**, IPV (killed), tetanus and diphtheria toxoids, Hib and pneumococcal conjugate vaccines, typhoid Vi conjugate, HPV (virus-like particles).
- **Freeze-sensitive vaccines**: hepatitis B, DPT, Td/TT, pentavalent, PCV and HPV are damaged by freezing because their **aluminium adjuvant** clumps irreversibly (the **shake test** detects it); **IPV** is freeze-sensitive too (it loses potency on freezing). BCG, measles and OPV are freeze-dried or tolerate freezing and are damaged by heat and light instead (BCG and measles must be shielded from light and used within 6 hours of reconstitution). OPV is the most heat-sensitive vaccine.
- **Conjugate vaccines** (Hib, pneumococcal, meningococcal, typhoid) link a polysaccharide to a carrier protein, which makes the response T-cell dependent: memory, class switching and good response under 2 years. Plain polysaccharide vaccines are T-independent and fail in infants.
- Highest single-dose efficacy: **rubella** vaccine (about 95% or more), then measles (about 85% at 9 months, 95% at 12 months or more). The vaccine strains of bivalent OPV are types 1 and 3 (type 2 removed in the April 2016 switch, to lower the risk of vaccine-derived type 2 polio; novel OPV2 is used against type 2 outbreaks).
- Adverse events: thrombocytopenia after measles or MMR vaccine, toxic shock syndrome from a contaminated reconstituted measles vial reused, intussusception after rotavirus vaccine, BCG lymphadenitis and disseminated BCG in severe immunodeficiency. Severe egg allergy is a contraindication to yellow fever vaccine; **egg allergy is no longer a contraindication to influenza vaccine** (ACIP 2023) and never was to MMR.
- Contraindications are few: anaphylaxis to an earlier dose; for live vaccines, pregnancy and severe immunodeficiency; for pertussis-containing vaccines, encephalopathy within 7 days of an earlier dose. Mild illness, malnutrition, prematurity (vaccinate by chronological age) and antibiotics are not contraindications.

Congenital heart disease

Acyanotic (left-to-right shunts)

- Features: tachypnoea, sweating on feeds, poor weight gain, recurrent chest infections; cyanosis only if the shunt reverses (**Eisenmenger syndrome**).
- **Ventricular septal defect (VSD)** is the **commonest congenital heart disease overall** (about a quarter to a third; membranous type commonest); pansystolic murmur at the left lower sternal edge; **left atrium and left ventricle enlarge**; the CHD most often complicated by endocarditis. **Small (restrictive, especially muscular) defects often close by themselves** (30-50%, mostly in the first 2 years) and have the loudest murmurs; **large defects rarely close** and cause heart failure at about 6-8 weeks as pulmonary vascular resistance falls; repair before pulmonary hypertension becomes fixed (Eisenmenger in a minority). Device closure is used for suitable muscular defects.
- **Atrial septal defect (ASD)**: ostium secundum commonest; primum goes with endocardial cushion defects and is commoner in Down syndrome. **Wide, fixed splitting of S2**, soft ejection flow murmur, **right atrium and right ventricle**

enlarge (the left atrium is not enlarged, unlike VSD), "hilar dance" on fluoroscopy. Small defects need no closure. Paradoxical embolism possible; Lutembacher syndrome is ASD plus mitral stenosis.

- **Patent ductus arteriosus (PDA)**: **continuous machinery murmur** under the left clavicle, because aortic pressure exceeds pulmonary pressure in both systole and diastole; bounding pulses, wide pulse pressure; left atrium, left ventricle and aorta enlarge. It closes after birth as prostaglandins fall. In a preterm infant, close it with **indomethacin or ibuprofen** (paracetamol is also used). If pulmonary pressure rises (Eisenmenger) the murmur disappears and the shunt reverses, giving **differential cyanosis** (blue toes, pink fingers). Term PDA does not close with drugs: transcatheter device or surgical ligation.
- Coarctation: weak femoral pulses, upper-limb hypertension, rib notching, "figure of 3" sign; associated with Turner syndrome and bicuspid aortic valve.

Cyanotic

- Right-to-left shunts: tetralogy of Fallot (TOF), transposition of the great arteries (TGA), tricuspid atresia, TAPVC, truncus arteriosus, Ebstein anomaly. Hypoplastic left heart causes death in the first week (duct-dependent). Truncus is not duct-dependent.
- **TGA is the commonest cyanotic lesion presenting in the first week (first 24 hours)**: parallel circulations, egg-on-a-string film, plethoric lungs. Give prostaglandin E1, then **balloon atrial septostomy (Rashkind)**; definitive repair is the **arterial switch (Jatene)**, which moves the great arteries to their proper ventricles and transfers the coronaries, done **in the first 2-3 weeks with an intact septum**, before the left ventricle, pumping only to the low-pressure lungs, loses the muscle to be the systemic pump. The older **atrial switch (Mustard, Senning)** redirects blood at atrial level and leaves the right ventricle systemic (late right ventricular failure, arrhythmias).
- **Tetralogy of Fallot** is the **commonest cyanotic lesion beyond infancy**: pulmonary stenosis (infundibular is the key), VSD, overriding aorta, right ventricular hypertrophy; boot-shaped heart with oligaemic lungs; ejection systolic murmur and single S2. Pentalogy adds an ASD; "pink Fallot" has mild stenosis and little cyanosis; right aortic arch in a quarter.
- **Tet spells** (hypercyanotic episodes): knee-chest position, oxygen, then **IV morphine**, IV fluid, propranolol or esmolol, phenylephrine; sodium bicarbonate for acidosis. **Squatting raises systemic vascular resistance** (compresses femoral arteries), cutting the right-to-left shunt across the VSD and sending more blood to the lungs. Avoid digoxin, isoprenaline and diuretics (worsen infundibular spasm). Palliation: modified Blalock-Taussig shunt; definitive: total repair.
- **TAPVC**: equal oxygen saturation in all chambers; supracardiac type commonest, with the **snowman or figure-of-8** heart (vertical vein to innominate vein and SVC); the infracardiac type obstructs and resembles RDS.
- **Tricuspid atresia**: left axis deviation, right atrial enlargement, hypoplastic right ventricle; **Ebstein anomaly**: box- or balloon-shaped heart, maternal lithium.
- Chest X-ray signs: boot = TOF; egg on a string = TGA; box or balloon = Ebstein; square = tricuspid atresia; snowman or cottage loaf = TAPVC; figure of 3 and rib notching = coarctation.

Most common lesion in syndromes

Condition	Lesion
Down syndrome	**atrioventricular septal defect (endocardial cushion defect)**, then VSD
Infant of diabetic mother	VSD commonest; TGA most specific
Congenital rubella	PDA (or pulmonary artery stenosis)
Turner	bicuspid aortic valve; coarctation
Noonan	pulmonary stenosis
Williams	supravalvular aortic stenosis
Holt-Oram	ASD
Edwards and Patau	VSD
DiGeorge (22q11.2 deletion)	interrupted aortic arch type B, truncus arteriosus, TOF
Marfan	aortic root dilatation, mitral valve prolapse

Diagnosis

- **NADAS criteria** diagnose congenital heart disease: major (systolic murmur grade 3 or more, any diastolic murmur, cyanosis, heart failure) and minor criteria (softer systolic murmur, abnormal S2, abnormal ECG, X-ray or blood pressure); need 2 major or 1 major plus 2 minor.
- **Pulse oximetry screening** of newborns after 24 hours (AAP 2020 algorithm): pass at 95% or more in the right hand or a foot with a difference of 3% or less; fail at once below 90%; 90-94% or a difference above 3% is repeated once. It detects critical lesions; **echocardiography confirms** (investigation of choice).

Rheumatic fever, endocarditis and other cardiac conditions (curriculum)

- **Acute rheumatic fever**: 2-4 weeks after group A streptococcal pharyngitis (age 5-15); a type II hypersensitivity (molecular mimicry). **Revised Jones criteria (AHA 2015)**: evidence of preceding streptococcal infection (raised ASO or anti-DNase B, positive throat culture) plus 2 major or 1 major with 2 minor. Major: **carditis (including subclinical, shown on echocardiography)**, **migratory polyarthritis (large joints)**, **Sydenham chorea**, **erythema marginatum**, subcutaneous nodules; in moderate-to-high-risk populations monoarthritis and polyarthralgia also count as major. Minor: fever, raised ESR or CRP, prolonged PR interval, and (in low-risk groups) arthralgia. **Carditis, mainly mitral valvulitis, causes the lasting damage** (chronic rheumatic heart disease; mitral regurgitation acutely, stenosis years later); the arthritis heals without sequelae. Treat with penicillin to eradicate the organism, aspirin (or naproxen) for arthritis, prednisolone for moderate-to-severe carditis, and haloperidol or valproate for severe chorea. **Secondary prophylaxis: benzathine penicillin G 1.2 million units IM every 3-4 weeks** (oral penicillin V or sulfadiazine; erythromycin if allergic): duration is until age 21 or 5 years after the last attack whichever is longer for no carditis, 10 years or until 21 for carditis without residual valve disease, and 10 years or until 40 for persisting valve disease (AHA 2009).
- **Infective endocarditis**: the commonest organisms are **viridans streptococci** (Staph aureus in acute disease and in intravenous or catheter cases); fever, a new or changing murmur, splinter haemorrhages, Osler nodes, Janeway lesions, Roth spots; **modified Duke criteria** (updated as the

Duke-ISCVID criteria, 2023) with at least 2-3 blood cultures before antibiotics and an echocardiogram. Prophylaxis before dental procedures is advised only for the highest-risk group: prosthetic valves, previous endocarditis, unrepaired cyanotic congenital heart disease and recent repairs with prosthetic material (AHA 2007, reaffirmed 2021).

- **Congestive heart failure in infants**: tachypnoea, poor feeding and sweating, tachycardia, **hepatomegaly**, cardiomegaly; the commonest cause is a large left-to-right shunt (VSD, PDA) or an obstructive lesion; treat with **diuretics (furosemide), ACE inhibitors, then digoxin** (restrict its use) and treat the cause. **Pericardial effusion**: muffled sounds, pulsus paradoxus, Beck triad in tamponade, enlarged globular heart with small voltage on ECG; confirmed by echocardiography; **pericardiocentesis** for tamponade.
- **Hypertension in children**: blood pressure at or above the 95th centile for age, sex and height on three occasions (AAP 2017; adolescents from 13: 130/80). Secondary causes dominate in the young, mostly **renal parenchymal disease**, then renovascular disease and **coarctation of the aorta**; essential hypertension rises with age and obesity.
- **Systemic lupus erythematosus** (childhood): girls, adolescence; malar rash, arthritis, nephritis and cytopenias; **ANA** is the screen, **anti-dsDNA and anti-Smith** are specific, low C3 and C4 in active disease; hydroxychloroquine and steroids, with other immunosuppressants for nephritis. **Juvenile idiopathic arthritis** (arthritis lasting at least 6 weeks under age 16, other causes excluded): **oligoarticular is the commonest** (girls, **ANA positive, chronic anterior uveitis**, needing slit-lamp screening), systemic-onset (Still disease: quotidian spiking fever, evanescent salmon-pink rash, lymphadenopathy, high ferritin; macrophage activation syndrome is the dangerous complication), and RF-positive polyarticular; **methotrexate** is the first disease-modifying drug, with biologics (etanercept, tocilizumab) after.

Genetic syndromes

Syndrome	Cause	Key features
Down	**trisomy 21** (95% non-disjunction, 4% translocation, 1% mosaic)	hypotonia, flat facies, upslanting palpebral fissures, single palmar crease, **AVSD**, duodenal atresia, Hirschsprung, hypothyroidism, atlantoaxial instability, leukaemia (transient abnormal myelopoiesis in the newborn; AML-M7 under 4, ALL later), early Alzheimer disease; surveillance (AAP 2022): echocardiogram at birth, thyroid at birth, 6 and 12 months then yearly, hearing and eyes regularly, **polysomnography by 4 years**; **no routine neck X-rays** for atlantoaxial instability in a child without symptoms, and coeliac testing only for symptoms
Edwards	trisomy 18	clenched fists with overlapping fingers, rocker-bottom feet, prominent occiput, micrognathia, VSD
Patau	trisomy 13	holoprosencephaly, cleft lip and palate, polydactyly, cutis aplasia
Klinefelter	**47,XXY**	tall, small firm testes, gynaecomastia, azoospermia, hypergonadotropic hypogonadism, Barr body
Turner	45,X	short, webbed neck, shield chest, streak ovaries, coarctation, horseshoe kidney, commonest cause of primary amenorrhoea, absent Barr body
Noonan	normal karyotype, autosomal dominant	like Turner but either sex, pulmonary stenosis, fertile
47,XYY		tall, normal IQ, acne, behavioural problems
Cri du chat	5p deletion	high-pitched cry, downward slanting fissures
Williams	7q11 deletion	elfin face, friendly, stellate iris, supravalvular aortic stenosis, hypercalcaemia
Fragile X	**FMR1, CGG repeat expansion at Xq27**	macro-orchidism, long face; commonest inherited cause of intellectual disability (Down is the commonest genetic cause)
Marfan	FBN1 (chromosome 15)	arachnodactyly, superotemporal lens dislocation, aortic root dilatation, mitral valve prolapse
Fetal alcohol	alcohol	smooth philtrum, thin upper lip, small head
Pierre Robin		micrognathia, glossoptosis, cleft palate

- **Why Down syndrome risk rises with maternal age**: oocytes are held in meiosis I from fetal life, and cohesion between chromosomes deteriorates with age, so **non-disjunction in maternal meiosis** becomes commoner. Translocation Down syndrome is not age-related. Screening: first-trimester combined test; cell-free fetal DNA is the most accurate screening test (invasive diagnosis by amniocentesis or CVS).
- **Fragile X anticipation**: a premutation (55-200 CGG repeats) in a carrier mother expands to a full mutation (above 200) in oogenesis, so later generations have larger repeats and worse disease. Triplet-repeat anticipation also occurs in Huntington (CAG; expands in paternal transmission) and myotonic dystrophy.
- **Duchenne muscular dystrophy (DMD)**: X-linked recessive, dystrophin gene at **Xp21** (the longest human gene; high new-mutation rate); a frameshift mutation abolishes dystrophin, whereas Becker has an in-frame mutation and partial dystrophin. Boys of 3-5 years: delayed walking, proximal weakness, **Gower sign**, calf pseudohypertrophy. **CK is raised more than ten-fold** (first test; CK-MM); genetic testing (MLPA/sequencing) confirms, and muscle biopsy shows dystrophin loss. Treatment: **corticosteroids (prednisolone or deflazacort; vamorolone, US FDA 2023)** slow progression and prolong walking, with physiotherapy and cardiac and respiratory care; newer: exon-skipping antisense drugs for particular mutations (eteplirsen, US FDA 2016) and micro-dystrophin gene therapy (delandistrogene moxeparvovec, US FDA 2023; after deaths from acute liver failure in non-ambulatory boys, FDA in November 2025 added a boxed warning and limited it to walking boys aged 4 or more). Wheelchair by about 12, death from cardiomyopathy or respiratory failure, now often in the third or fourth decade. Note that the dystrophin gene is on the X chromosome, not chromosome 21.
- **Cystic fibrosis**: autosomal recessive, **CFTR on chromosome 7q31**, commonest mutation **Delta F508** (class II, trafficking defect). Recurrent chest infections (Pseudomonas, Staphylococcus aureus, Burkholderia cepacia complex), steatorrhoea and failure to thrive, meconium ileus, distal intestinal obstruction, rectal prolapse, biliary cirrhosis, male infertility (absent vas), nasal polyps. **Sweat chloride above 60**

mmol/L on two occasions diagnoses; newborn screen immunoreactive trypsinogen. Treatment: physiotherapy, pancreatic enzymes, and CFTR modulators (elexacaftor-tezacaftor-ivacaftor) in current practice.

- **Menkes disease**: X-linked recessive, **ATP7A copper-transport defect** (copper deficiency): kinky, steely hair (pili torti), seizures, neurodegeneration, low lysyl oxidase activity. **Wilson disease** is the reverse: autosomal recessive **ATP7B** (chromosome 13), copper overload; low ceruloplasmin, low total serum copper but raised free copper, raised urinary copper (above 100 ug/day) and liver copper; hepatic, neuropsychiatric (tremor, dystonia, psychosis) and haemolytic features; Kayser-Fleischer ring (Descemet membrane); treat with zinc (maintenance), chelators (D-penicillamine, trientine).
- **Prader-Willi**: loss of the paternal 15q11-q13 (paternal deletion in about 70%, maternal uniparental disomy in about 25%, an imprinting defect in a few): neonatal hypotonia and poor suck, then hyperphagia and obesity. **Angelman**: loss of the maternal copy (UBE3A): seizures, ataxia, happy demeanour.
- **Mitochondrial inheritance** is maternal: all children of an affected mother, **none of an affected father's** (MELAS, MERRF, Leber neuropathy; severity varies with heteroplasmy). **X-linked dominant**: an affected father has all daughters and no sons affected (X-linked hypophosphataemic rickets, X-linked Alport; Rett is X-linked dominant but almost always a new mutation, lethal in boys).
- **Potter sequence** (bilateral renal agenesis): oligohydramnios, **pulmonary hypoplasia** (cause of death), flattened facies, limb deformities.
- **Spinal muscular atrophy**: autosomal recessive, **homozygous SMN1 deletion on 5q13** (SMN2 copy number sets severity), loss of anterior horn cells; an alert baby, profoundly hypotonic, with tongue fasciculation, areflexia, a bell-shaped chest and paradoxical breathing, normal sensation and creatine kinase; type 1 (Werdnig-Hoffmann) begins before 6 months. Current treatment: nusinersen (intrathecal, US FDA 2016), onasemnogene abeparvovec gene therapy (2019) and oral risdiplam (2020), best given before symptoms, so newborn screening is spreading.
- **Kallmann syndrome**: hypogonadotropic hypogonadism with anosmia (failed migration of GnRH neurons); Klinefelter has raised gonadotropins.

Paediatric infections and emergencies

Exanthems

Disease	Clues
Measles (paramyxovirus)	cough, coryza, conjunctivitis; **Koplik spots** (pathognomonic, on the buccal mucosa opposite the molars) in the prodrome; maculopapular rash from behind the ears, no vesicles; incubation 10-14 days
Rubella	rash with postauricular lymphadenopathy; Forchheimer spots on the palate
Roseola (HHV-6)	high fever then rash as fever falls; Nagayama spots
Scarlet fever	sandpaper rash, strawberry tongue (white first, then red)
Erythema infectiosum (parvovirus B19)	slapped cheeks, then a lacy reticular rash on the limbs

- Measles pathology: **Warthin-Finkeldey multinucleate giant cells** in lymphoid tissue and **Hecht giant-cell pneumonia**. Commonest complication: otitis media; commonest cause of death: pneumonia; late complication: SSPE. Humans are the only reservoir with no carrier state. Infectious from 4 days before to 4 days after the rash. **Give vitamin A** (WHO: 2 doses 24 hours apart: 50,000 IU under 6 months, 100,000 IU at 6-11 months, 200,000 IU from 12 months) to every child with measles; it lowers mortality.
- **Congenital rubella syndrome (Gregg triad)**: cataract, sensorineural deafness and congenital heart disease (PDA or pulmonary artery stenosis). Congenital toxoplasmosis: chorioretinitis, hydrocephalus, intracranial calcification. Rubella infects the fetus across the placenta, mostly in the first trimester, so it is not a perinatal infection; HIV, hepatitis B and CMV are transmitted around birth or in milk.
- **Parvovirus B19** enters erythroid progenitors through the **P antigen** and halts red cell production for about a week: trivial in a healthy child, but a **transient aplastic crisis** (haemoglobin falls, **reticulocytes almost absent**) in sickle cell disease, spherocytosis and other short-survival anaemias; chronic anaemia in the immunocompromised; **hydrops fetalis** in pregnancy. Splenic sequestration differs: a big spleen and a high reticulocyte count. Roseola (HHV-6, also HHV-7): 3 days of high fever, then the rash as the fever falls; a common cause of febrile seizures.
- **Infectious mononucleosis** (EBV): fever, exudative sore throat, generalised lymphadenopathy, splenomegaly, atypical lymphocytes (Downey cells); a rash with amoxicillin or ampicillin is typical. Confirm by **heterophile antibody (Monospot)**, often negative under 4 years, or **EBV VCA IgM**, the test of choice in young children. No contact sport for 3-4 weeks (splenic rupture).

Meningitis and encephalitis

- 3 months to 5 years, now that Hib vaccine is widespread: **Streptococcus pneumoniae** and Neisseria meningitidis dominate. In neonates: GBS, E. coli, Listeria. Enterovirus is the commonest cause of viral meningitis; HSV-1 the commonest sporadic encephalitis; HSV-2 mainly neonatal; arboviruses (Japanese encephalitis in Nepal's Terai) the commonest epidemic cause.
- **Herpes simplex encephalitis**: fever, focal seizures, behaviour change, temporal lobe changes on MRI, red cells and a raised protein in the CSF; **CSF HSV PCR** confirms. Start **IV aciclovir** empirically, without waiting for the PCR, for 14-21 days (21 in neonates). Ganciclovir is for CMV.
- **Duration of IV antibiotic** for uncomplicated bacterial meningitis (Nelson): **pneumococcal 10-14 days**, meningococcal 5-7 days (7 in most texts), Hib 7-10 days, GBS 14-21 days, Gram-negative neonatal meningitis 21 days; longer for abscess or empyema; tuberculous meningitis 12 months.

Reye syndrome

- Encephalopathy with fatty liver after influenza or varicella in a child given **aspirin**; avoid aspirin in viral illness (exception: Kawasaki disease, supervised).

Febrile seizures

- Generalised seizure with fever above 38 C, age **6 months to 5 (or 6) years**, no CNS infection; usually in the rising phase of fever.
- **Simple**: generalised, under 15 minutes, one in 24 hours (the commonest). **Complex**: **focal**, over 15 minutes, or recurring within 24 hours.
- Risk of later epilepsy rises with complex features, family history of epilepsy, developmental abnormality and fever of under an hour before the seizure.
- Management: **reassure** (benign, outgrown by about 6 years); treat discomfort with paracetamol, but antipyretics do not prevent recurrence; **no long-term anticonvulsants, no EEG, no antibiotics** for a simple seizure; rescue benzodiazepine (diazepam or lorazepam) if a seizure continues. About a third recur. Lumbar puncture is considered under 12 months and with meningism.
- Epilepsy drugs of choice: infantile spasms (West syndrome: spasms, developmental delay, hypsarrhythmia) **ACTH or prednisolone, with vigabatrin first for tuberous sclerosis**; absence **ethosuximide**; focal oxcarbazepine or carbamazepine; juvenile myoclonic **valproate** (the textbook answer), with **levetiracetam or lamotrigine preferred in girls and women who may become pregnant** (valproate is teratogenic: MHRA 2018 and EMA restrictions); carbamazepine and phenytoin worsen myoclonic and absence seizures; self-limited epilepsy with centrotemporal spikes (benign rolandic) often needs no drug, otherwise carbamazepine or levetiracetam. **Cannabidiol** is approved (US FDA 2018 for Lennox-Gastaut and Dravet syndromes, 2020 for tuberous sclerosis complex), not for Rett syndrome. Fenfluramine (2020) and stiripentol are used in Dravet syndrome, where sodium-channel blockers worsen seizures.

Kawasaki disease

- Fever for **5 days or more plus 4 of 5**: bilateral non-purulent conjunctivitis, polymorphous rash, oedema and erythema of hands and feet, cervical lymphadenopathy, red cracked lips and strawberry tongue. Commonest medium-vessel vasculitis; age under 5, peak 18-24 months. **Coronary artery aneurysm** is the main complication; thrombocytosis in the second week.
- Treatment: **IV immunoglobulin 2 g/kg plus aspirin**, ideally within 10 days of onset; echocardiography at diagnosis and follow-up; a second IVIG dose, steroids or infliximab for IVIG-resistant disease (fever persisting or returning 36 hours or more after the IVIG ends). Incomplete Kawasaki (fewer criteria, typical in infants under 6 months) needs echocardiography and laboratory criteria. Aspirin dose: the classic answer is high-dose (80-100 mg/kg/day) until the fever settles, then low-dose (3-5 mg/kg/day) for 6-8 weeks; AHA 2017 accepts a moderate dose (30-50 mg/kg/day) as well, and many centres use it; the AHA scientific statement of 2024 defines high risk as age 6 months or less or a coronary (LAD or RCA) Z-score of 2.5 or more, and advises intensified first treatment for these children (IVIG with corticosteroids or infliximab).

Further infections and emergencies (curriculum)

- **Enteric fever**: Salmonella Typhi or Paratyphi; step-ladder fever, relative bradycardia, rose spots, hepatosplenomegaly, leucopenia; **blood culture** (best in the first week; bone marrow culture is the most sensitive); Widal is unreliable. Treatment: **azithromycin or ceftriaxone** (fluoroquinolone and multidrug resistance is common in South Asia); complications are intestinal perforation and haemorrhage in the third week. Vaccine: typhoid conjugate.
- **Tetanus**: Clostridium tetani toxin; trismus, risus sardonicus, spasms. **Neonatal tetanus**: the baby sucked and cried normally for the first 2 days, then could not suck and became stiff or had spasms, usually on days 3-14 (WHO case definition: days 3-28), from an unclean cord; prevented by maternal Td immunisation and clean delivery. Treat with **human tetanus immunoglobulin, metronidazole, benzodiazepines (diazepam, midazolam)** for spasms, quiet environment and airway care.
- **Poliomyelitis**: asymmetric flaccid paralysis with fever (see Neurology); there is no cure, and prevention is by vaccine. **Chickenpox**: vesicles in crops in different stages, aciclovir for the immunocompromised and neonates; the complication in healthy children is secondary skin infection. **Herpangina**: coxsackie A, fever with vesicles and ulcers on the soft palate and tonsillar pillars; hand, foot and mouth disease adds the hands and feet.
- **Malaria**: Plasmodium falciparum causes severe disease (cerebral malaria, severe anaemia, hypoglycaemia, acidosis); diagnose by thick and thin film or rapid antigen test; **severe malaria: IV artesunate** (WHO), then an artemisinin combination therapy; uncomplicated falciparum: artemether-lumefantrine; vivax: chloroquine with primaquine (check G6PD) for relapse. **Kala-azar (visceral leishmaniasis)** from Leishmania donovani, sandfly vector, in the Indian subcontinent: prolonged fever, massive splenomegaly, pancytopenia, hypergammaglobulinaemia; rK39 rapid test; amastigotes (LD bodies) on aspirate, **splenic aspirate the most sensitive (over 90%)**, bone marrow safer but less sensitive. Treatment: the older textbook answers are sodium stibogluconate (abandoned in Bihar for resistance) and then oral miltefosine; **current first line in South Asia is single-dose liposomal amphotericin B 10 mg/kg** (WHO Expert Committee 2010, used by Nepal's and India's elimination programmes), with miltefosine-paromomycin as a combination alternative. **Post-kala-azar dermal leishmaniasis** (macules or nodules months to years after cure) keeps the parasite in the community. **Giardiasis**: foul fatty diarrhoea, cysts or trophozoites in stool; metronidazole, tinidazole.
- **Leprosy**: Mycobacterium leprae; hypopigmented anaesthetic patch, thickened nerve; paucibacillary is 1-5 skin lesions with no nerve involvement and negative smears, multibacillary 6 or more lesions, nerve involvement or a positive smear. Multidrug therapy: the classic answer (WHO 1982, Park) is rifampicin and dapsone for 6 months in paucibacillary and rifampicin, dapsone and clofazimine for 12 months in multibacillary disease; **current (WHO 2018): the same three drugs for all**, 6 months for paucibacillary and 12 months for multibacillary. WHO 2018 also advises **single-dose rifampicin as post-exposure prophylaxis** for contacts aged 2 or more, and rifampicin resistance is treated with second-line drugs (ofloxacin or another quinolone, minocycline, clarithromycin). **Candidiasis**: oral thrush in infants (nystatin or miconazole gel), napkin dermatitis with satellite lesions; fluconazole for invasive disease.
- **HIV in children**: mother-to-child transmission is the main route; infants under 18 months are diagnosed by **HIV DNA PCR** (antibody tests reflect maternal IgG); **all infected**

children are treated, whatever the CD4 count (WHO 2016), with a **dolutegravir-based regimen** now preferred (WHO 2021); **cotrimoxazole prophylaxis** against Pneumocystis from 4-6 weeks in exposed infants; infant nevirapine prophylaxis; exclusive breastfeeding is advised where the mother is on treatment.

- **Skin**: impetigo (Staph aureus or group A streptococcus; honey-coloured crusts; topical mupirocin, oral cloxacillin or cephalexin for extensive disease; the streptococcal form may cause glomerulonephritis); **ringworm (tinea)**: topical clotrimazole or terbinafine, oral griseofulvin for tinea capitis; **scabies**: nocturnal itch, burrows, family contacts; **5% permethrin cream** to the whole body from neck down (the head too in infants), repeated after 7 days, with oral ivermectin as an alternative (not under 15 kg); furuncles and **cellulitis** (flucloxacillin or cloxacillin, drainage of an abscess); **urticaria** (antihistamines; adrenaline if anaphylaxis); **Stevens-Johnson syndrome** (drug-induced, e.g. sulfonamides, carbamazepine, lamotrigine, phenytoin: target lesions, mucosal erosions, skin detachment under 10% of body surface; stop the drug, supportive care).
- **Poisoning and drowning**: **kerosene**: do not induce vomiting or lavage (aspiration pneumonitis is the danger), treat as chemical pneumonitis; **organophosphate**: atropine until secretions dry, with pralidoxime; **paracetamol**: N-acetylcysteine; **iron**: desferrioxamine; **opioid**: naloxone; **carbon monoxide**: 100% oxygen. **Activated charcoal** for most oral drugs within an hour but not for hydrocarbons, corrosives, metals or alcohols. **Drowning**: airway and ventilation first (hypoxia is the injury, rescue breaths before compressions), and warm the child. **Shock**: hypovolaemic shock needs 10-20 mL/kg isotonic crystalloid boluses with reassessment (a cautious 10 mL/kg when cardiac failure is suspected; in severe acute malnutrition with shock WHO gives 15 mL/kg over 1 hour, Ringer lactate with 5% dextrose or half-strength Darrow, then reassesses); **anaphylaxis** needs **intramuscular adrenaline 0.01 mg/kg (1 in 1000, 0.01 mL/kg)** into the anterolateral thigh at once (maximum 0.5 mg a dose, repeated after 5 minutes if needed).

Renal, urinary and vasculitis

Glomerular disease

	Nephrotic	Nephritic
Features	proteinuria (40 mg/m2/hour or more, urine protein:creatinine ratio 2 mg/mg or more, or dipstick 3+), hypoalbuminaemia (under 2.5-3 g/dL), oedema, hyperlipidaemia	hypertension, haematuria (cola urine), oliguria, oedema
Children	**minimal change disease** (then FSGS)	post-streptococcal, IgA, MPGN

- **Alport syndrome**: a defect of **type IV collagen** (glomerular basement membrane, cochlea, lens capsule); X-linked COL4A5 in about 80%, autosomal forms COL4A3/4. Haematuria, then proteinuria, **sensorineural deafness**, **anterior lenticonus**; electron microscopy shows a split, laminated ("basket-weave") basement membrane. Boys progress to kidney failure in adolescence or early adult life; ACE inhibitors slow it. Thin basement membrane disease is the benign relative.

Nephrotic syndrome in children

- **Minimal change disease**: about 80% of childhood nephrotic syndrome (age 2-6); normal light microscopy, **foot-process effacement** on EM, selective albuminuria, steroid-responsive. **Gross haematuria, hypertension that persists, low C3 and renal failure are not features** (transient microscopic haematuria occurs in up to a fifth); any of them is a reason to biopsy.
- **Initial treatment**: prednisolone **60 mg/m2/day (2 mg/kg, maximum 60 mg) for 4-6 weeks, then 40 mg/m2 (1.5 mg/kg) on alternate days for 4-6 weeks** (8-12 weeks in all; IPNA 2023, KDIGO 2021). No biopsy before steroids in a typical child aged 1-12.
- Definitions (IPNA 2023): remission = urine protein nil or trace (UPCR under 0.2) for 3 days; **relapse = 3+ for 3 days**; frequent relapses = 2 or more in the first 6 months or 4 or more in any 12 months; steroid-dependent = relapse during the taper or within 14 days of stopping. **Steroid resistance**: the classic answer (ISKDC) is no remission after 8 weeks of prednisolone; **current definition (IPNA 2020/2023, KDIGO 2021): no remission after 4 weeks of daily prednisolone at full dose** (with a 2-week confirmation period). Steroid-resistant disease is biopsied and genetically tested (podocyte genes such as NPHS1, NPHS2); calcineurin inhibitors (tacrolimus, cyclosporine) are first-line.
- Steroid-sparing drugs for frequent relapses or dependence: levamisole, mycophenolate, cyclophosphamide, calcineurin inhibitors, and **rituximab** (now widely used).
- Complications: **infection** (the commonest; **spontaneous bacterial peritonitis from Streptococcus pneumoniae**, cellulitis), because immunoglobulin G and complement factors (such as properdin) are lost in the urine, so capsulated bacteria are poorly **opsonised** (pneumococcal vaccination is advised), **thrombosis** (loss of antithrombin III; renal vein thrombosis, more in membranous nephropathy in adults), hypovolaemia, hyperlipidaemia, and steroid side effects. Congenital nephrotic syndrome (first 3 months): Finnish type, NPHS1 (nephrin).

Nephritis

- **Post-streptococcal GN** (PSGN): nephritis **1-2 weeks after a sore throat** (about 10 days) or **3-6 weeks after a skin infection**, from nephritogenic group A streptococci; **low C3 that returns to normal within 6-8 weeks** (a C3 still low after 8 weeks suggests MPGN or lupus and needs biopsy); raised ASO titre after throat infection, anti-DNase B after skin infection; subepithelial humps. Treatment supportive (salt and fluid restriction, diuretics for hypertension); penicillin for the residual infection; it does not alter the nephritis. Excellent prognosis in children.
- **IgA nephropathy** causes visible haematuria **1-2 days after** an infection (synpharyngitic), recurrent, with **normal C3**; mesangial IgA deposits. It is the commonest glomerulonephritis worldwide and the commonest glomerular cause of recurrent visible haematuria in children (the commonest cause of visible haematuria overall in children is UTI).
- Low C3: PSGN, MPGN (C3 glomerulopathy), lupus, shunt nephritis, endocarditis; normal C3: IgA nephropathy, IgA vasculitis, anti-GBM disease, ANCA vasculitis.
- Microscopic haematuria: **5 or more RBCs per high-power field in children** (Nelson), 3 or more in adults (AUA); confirm

on 2 of 3 samples. Alport syndrome (type IV collagen, mostly X-linked COL4A5): haematuria, sensorineural deafness, anterior lenticonus.

IgA vasculitis (Henoch-Schonlein purpura)

- The **commonest small-vessel vasculitis in childhood** (age 3-10, peak 4-6, often after an upper respiratory infection). **EULAR/PRINTO/PRES 2010 criteria**: palpable purpura (mandatory) mainly on the legs and buttocks, plus at least one of abdominal pain, IgA-dominant deposits on biopsy, arthritis or arthralgia, renal involvement (haematuria or proteinuria).
- **Platelets and clotting are normal**: the purpura is vasculitic, not from thrombocytopenia. IgA immune complexes; complement normal; ANCA-negative. GI complications: **ileo-ileal intussusception** (unlike the usual ileocolic), bleeding. Kidney: nephritis in about a third, usually within 4-6 weeks, so **urine and blood pressure are checked for 6-12 months**; long-term outcome depends on the kidney.
- Treatment supportive (NSAIDs for joints if the kidneys are normal); steroids for severe abdominal pain or significant nephritis; early steroids do not prevent nephritis.

Haemolytic uraemic syndrome

- Triad: **microangiopathic haemolytic anaemia (schistocytes), thrombocytopenia and acute kidney injury**; the **commonest cause of acute kidney injury in young children** in many countries. Typical HUS follows bloody diarrhoea from **Shiga toxin-producing E. coli (O157:H7)**, or Shigella dysenteriae type 1 in South Asia. **Antibiotics and antimotility drugs are avoided** in STEC diarrhoea (they raise the risk); treatment is supportive with dialysis when needed. **Atypical HUS** (complement dysregulation, no diarrhoea) is treated with the anti-C5 antibody **eculizumab**. Coombs test negative (except pneumococcal HUS, from neuraminidase exposing the T antigen).

Urinary tract infection

- E. coli is the commonest organism. Diagnosis needs a clean sample: suprapubic aspirate (any growth), catheter (50,000 CFU/mL or more, AAP; 10,000 or more in some guidelines) or clean catch (100,000 or more); bag specimens are only for ruling out. Febrile UTI in an infant is pyelonephritis until shown otherwise; oral antibiotics (7-10 days) are as good as IV in a child who is not toxic.
- **First UTI imaging**: the exam answer (Ghai, after the Indian Society of Pediatric Nephrology, 2011): **under 1 year ultrasound, DMSA and MCUG**; 1-5 years ultrasound and DMSA, with MCUG if either is abnormal; over 5 years ultrasound, with further tests only if abnormal. Current practice is more selective: **AAP 2011 (reaffirmed 2016)** gives renal and bladder ultrasound after the first febrile UTI at 2-24 months and **MCUG only if the ultrasound is abnormal or the UTI recurs**; **NICE CG54** (2007, imaging advice unchanged in its 2022 update): under 6 months ultrasound for all, with DMSA and MCUG for atypical or recurrent UTI; 6 months-3 years DMSA for atypical or recurrent UTI; older children ultrasound if atypical.
- **Vesicoureteric reflux**: graded I-V on MCUG (International Reflux Study: I ureter only; II to the pelvis without dilatation; III mild dilatation; IV moderate dilatation with blunted calyces; V gross dilatation and tortuosity). **MCUG is the investigation of choice for VUR**; **DMSA is the investigation of choice for renal scarring** (done 4-6 months after the infection). Low grades often resolve; antibiotic prophylaxis or endoscopic injection or reimplantation for high grades with breakthrough infection. VUR with pyelonephritis is the commonest cause of renal scarring (reflux nephropathy).
- **PUJ obstruction** is the commonest cause of significant hydronephrosis in children (transient hydronephrosis is the commonest antenatal finding); diuretic renogram (MAG3 or DTPA) shows the obstruction; pyeloplasty. **Posterior urethral valves** are the commonest cause of lower urinary tract obstruction in boys and the most severe: antenatal bilateral hydronephrosis with a thick-walled, keyhole bladder and oligohydramnios; poor stream; **MCUG is diagnostic** (dilated posterior urethra); cystoscopic valve ablation. Many still progress to chronic kidney disease.
- **Revised bedside Schwartz equation**: eGFR = 0.413 x height (cm) / serum creatinine (mg/dL). The older formula used k x height / creatinine with k 0.33 for preterm infants, 0.45 term infants, 0.55 children and adolescent girls, 0.7 adolescent boys.

Renal tubular acidosis

	Distal (type 1)	Proximal (type 2)	Type 4
Defect	H+ secretion in the collecting duct	HCO3- reabsorption in the proximal tubule	aldosterone deficiency or resistance
Urine pH in acidosis	**above 5.5**	below 5.5 once bicarbonate falls	below 5.5
Potassium	low	low	**high**
Stones, nephrocal-cinosis	**yes**	no	no
Clues	rickets, failure to thrive	Fanconi syndrome (glycosuria, aminoaciduria, phosphaturia); cystinosis the commonest inherited cause in children	obstructive uropathy

All are normal anion gap (hyperchloraemic) metabolic acidoses.

Wilms tumour (nephroblastoma)

- **Commonest primary renal malignancy in children**, peak 3-4 years (most under 5), bilateral in 5-10%. Usually an **asymptomatic, smooth flank mass that does not cross the midline**, found by a parent; also abdominal pain, **hypertension** (renin, about a quarter) and **haematuria** (in about 15-25%, from extension into the collecting system). Triphasic histology (blastema, stroma, epithelium).
- Genes and syndromes: **WT1 at 11p13** (**WAGR**: Wilms, aniridia, genitourinary anomalies, retardation; Denys-Drash: nephropathy and male pseudohermaphroditism), **WT2 at 11p15** (**Beckwith-Wiedemann**: macroglossia, omphalocele, hemihypertrophy, neonatal hypoglycaemia; ultrasound every 3 months to age 7).
- **Prognosis is decided by stage and histology**, not by the presenting symptom. **Anaplasia is the unfavourable histology** (diffuse anaplasia worst; COG); in the SIOP system, after preoperative chemotherapy, **blastemal-type** and diffuse anaplasia are high-risk histology. Molecular markers used for risk: **loss of heterozygosity at 1p and 16q, and 1q gain** (worse outcome; COG). Older texts call haematuria a sign of pelvic invasion and of a worse outlook; it is **not a prognostic factor in current staging or risk groups** (COG, SIOP-RTSG). Lungs are the commonest site of metastasis.
- Treatment: two approaches, both with survival above 90% overall: **COG (North America): upfront nephrectomy**, then

chemotherapy by stage and histology; **SIOP (Europe and most of the world): preoperative chemotherapy (vincristine and actinomycin D)**, then nephrectomy. Avoid percutaneous biopsy before nephrectomy in the COG system (it upstages the tumour); SIOP accepts a core biopsy for uncertain cases. Wilms is radiosensitive (flank radiotherapy for stage III).

	Wilms tumour	Neuroblastoma
Age	3-4 years	under 2 years
Mass	smooth, rarely crosses the midline	irregular, often crosses the midline
Calcification on CT	uncommon (about 10%)	**common (80-90%)**
Vessels	displaced; tumour thrombus in renal vein and IVC	**encased**
Markers	none	**urinary VMA and HVA**; MYCN amplification (poor prognosis)
Other	hypertension, haematuria	raccoon eyes (orbital metastasis), opsoclonus-myoclonus, blueberry-muffin skin nodules, Horner syndrome, stage MS (4S) regresses in infants

- Childhood tumours: commonest malignancy **ALL**; commonest solid tumour **brain tumours**; commonest extracranial solid tumour **neuroblastoma**; commonest renal tumour in early infancy (first 3 months) **congenital mesoblastic nephroma**; commonest neonatal tumour **sacrococcygeal teratoma**.

Genital problems

- **Undescended testis**: orchidopexy at **6-18 months** (EAU and AUA: by 18 months, ideally after 6 months, since few descend after then); hCG has no role. Risks: infertility and germ-cell tumour (seminoma), torsion, hernia. Retractile testis is normal.
- **Hypospadias**: meatus on the ventral side; **no circumcision** (the foreskin is used in the repair, done at 6-18 months). Phimosis is physiological up to about 3 years or later.

Metabolic and gastrointestinal

Meckel diverticulum

- Remnant of the vitello-intestinal (vitelline) duct; true diverticulum on the antimesenteric border with its own blood supply (vitelline artery). **Rule of 2s**: 2% of the population, **2 feet** from the ileocaecal valve, **2 inches** long, twice as common in males, symptomatic mostly by age 2, **two types of ectopic tissue** (gastric and pancreatic); only about 2% (4% lifetime) ever cause symptoms. Ectopic tissue is found in about half of resected diverticula, and in most that bleed.
- Commonest ectopic tissue: gastric. **Painless rectal bleeding** (from an ulcer in the adjacent ileum) is the commonest presentation in children; obstruction or intussusception (and diverticulitis) in adults. Best test for bleeding: **Tc-99m pertechnetate scan** (taken up by gastric mucosa; pentagastrin, H2 blockers raise its yield); treatment diverticulectomy (or segmental resection if there is ectopic tissue at the base or a broad base).

Pyloric stenosis, intussusception, atresias, Hirschsprung

- **Infantile hypertrophic pyloric stenosis**: first-born boys, **2-8 weeks**; **non-bilious projectile vomiting**, hungry after vomiting, visible peristalsis, an olive-shaped mass in the right upper quadrant; **hypochloraemic hypokalaemic metabolic alkalosis with paradoxical aciduria**; **ultrasound** is the investigation of choice (muscle thickness 3-4 mm or more, length 15-17 mm or more); correct fluids and electrolytes first (it is not a surgical emergency), then **Ramstedt pyloromyotomy** (open or laparoscopic). Erythromycin and azithromycin in early infancy raise the risk.
- **Intussusception**: commonest cause of intestinal obstruction from **3 months to 6 years** (peak 6-12 months); **ileocolic** commonest; lead point usually hypertrophied Peyer patches after a viral infection (lead points such as Meckel, polyp, lymphoma in older children); colicky pain with drawing up of the legs, vomiting, **red currant jelly stool** (late), sausage-shaped mass, empty right iliac fossa (Dance sign). **Ultrasound: target or doughnut sign** (investigation of choice); **pneumatic or hydrostatic enema reduction** under imaging is first-line (success about 80-90%), surgery for peritonitis, perforation or failure.
- **Duodenal atresia**: bilious vomiting from birth (the obstruction is usually below the ampulla), **double bubble** on X-ray, polyhydramnios; **a third have Down syndrome**. Oesophageal atresia with distal tracheo-oesophageal fistula (type C, about 85%) is the commonest type: frothy saliva, choking at the first feed, a nasogastric tube coiling in the pouch on X-ray; VACTERL associations.
- **Malrotation with volvulus**: **bilious vomiting in a newborn is a surgical emergency** until proved otherwise; upper GI contrast study (duodenojejunal flexure not left of the spine; corkscrew) is the investigation of choice; Ladd procedure.
- **Hirschsprung disease**: absent ganglion cells (failed neural-crest migration) in a segment beginning at the anus, rectosigmoid in about 75%; boys 4 : 1; **Down syndrome** and RET mutations. **Failure to pass meconium within 48 hours** (about 90% of affected babies), distension, bilious vomiting, explosive stool on rectal examination, enterocolitis (the main cause of death). **Rectal suction biopsy is diagnostic** (no ganglion cells, hypertrophied nerve trunks, raised acetylcholinesterase staining, absent calretinin); contrast enema shows the transition zone; anorectal manometry shows absent rectoanal inhibitory reflex. Pull-through operation (Swenson, Duhamel, Soave), often in one stage and transanally now.
- Delayed passage of meconium: think of **Hirschsprung disease** first (it must be excluded by biopsy), **meconium plug syndrome** (small left colon, infants of diabetic mothers; relieved by the contrast enema) and **meconium ileus**, which is almost always **cystic fibrosis** (soap-bubble appearance, no air-fluid levels; Gastrografin enema).

Other gastrointestinal facts

- Commonest cause of exocrine pancreatic insufficiency in children: cystic fibrosis, then Shwachman-Diamond syndrome (with neutropenia; not the same as Diamond-Blackfan anaemia, a pure red cell aplasia).
- **Coeliac disease**: gluten-sensitive enteropathy with HLA-DQ2 or DQ8; diarrhoea, failure to thrive, anaemia; screen with **IgA anti-tissue transglutaminase** (with total IgA); villous atrophy and intraepithelial lymphocytes on duodenal biopsy. **ESPGHAN 2020: no biopsy needed** when anti-tTG is 10 times the upper limit or more and anti-endomysial antibody is positive in a second sample. Lifelong gluten-free diet.
- Gastro-oesophageal reflux is normal in infants (peak at 4 months, settles by 12-18 months); treat only with

complications; proton-pump inhibitors are not for the happy spitter.

- Cholestasis and the hereditary hyperbilirubinaemias are under Neonatology.

Rickets

	Calcium	Phosphate	ALP	PTH	1,25(OH)2D
Nutritional	normal or low	low	high	high	low or normal
X-linked hypophosphataemic (PHEX; phosphate wasting)	normal	low	high	normal	low or inappropriately normal
VDDR type I (1-alpha-hydroxylase defect)	low	low	high	high	**low**
VDDR type II (vitamin D receptor defect)	low	low	high	high	**high**
Hypophosphatasia (ALPL)	high	high	**low**	normal	
Renal osteodystrophy	low	**high**	high	high	low

- A child with rickets, high PTH, **raised calcitriol** and no response to ordinary doses of vitamin D has **VDDR type II** (often with **alopecia**; treat with high-dose calcitriol and calcium, IV calcium for the most resistant; response may be partial). Type I responds to physiological doses of calcitriol.
- **X-linked hypophosphataemic rickets**: high FGF23 causes phosphate wasting; the classic treatment is oral phosphate with calcitriol; **burosumab**, an anti-FGF23 antibody (approved 2018), is now used for children. Hypophosphatasia: enzyme replacement with **asfotase alfa** (2015).
- Refractory rickets: VDDR I and II and hypophosphataemic rickets. Shortcut: high phosphate suggests CKD; low phosphate with acidosis suggests renal tubular acidosis (Fanconi); without acidosis, high PTH suggests vitamin D-dependent rickets and normal PTH hypophosphataemic rickets.
- Normal adult values: calcium 8.5-10.5 mg/dL; phosphate 2.5-4.5 mg/dL; PTH 10-65 pg/mL. **Phosphate and ALP are higher in growing children** (phosphate about 4-6.5 mg/dL in infancy; ALP up to several times the adult range), so read them against age.

Inborn errors of metabolism

Disorder	Defect	Key features
Phenylketonuria	phenylalanine hydroxylase (or its cofactor BH4)	normal at birth, then intellectual disability, seizures, **musty (mousy) odour**, fair skin and hair, eczema; newborn screening; low-phenylalanine diet for life (and before and during pregnancy: maternal PKU); sapropterin for BH4-responsive types
Galactosaemia	galactose-1-phosphate uridyltransferase (GALT)	vomiting, jaundice, hepatomegaly after milk feeds, **E. coli sepsis**, **oil-drop cataract**, reducing substance in urine; remove lactose at once (soy formula); breastfeeding is contraindicated
Maple syrup urine disease	branched-chain alpha-keto acid dehydrogenase	leucine, isoleucine, valine; encephalopathy in the first week, sweet-smelling urine
Homocystinuria	cystathionine beta-synthase	marfanoid habitus with **downward (inferonasal) lens dislocation**, intellectual disability, **thrombosis**; pyridoxine-responsive in about half
Urea cycle defects	**ornithine transcarbamylase (X-linked, commonest)**	hyperammonaemia with **respiratory alkalosis** and no ketoacidosis; orotic aciduria in OTC deficiency
Organic acidaemias (propionic, methylmalonic)		ketoacidosis with a high anion gap, hyperammonaemia, neutropenia
Von Gierke (GSD I)	glucose-6-phosphatase	fasting hypoglycaemia, lactic acidosis, hyperuricaemia, big liver and kidneys, doll face
Pompe (GSD II)	lysosomal acid maltase (acid alpha-glucosidase)	infantile **cardiomegaly**, hypotonia, macroglossia; enzyme replacement (alglucosidase alfa)
McArdle (GSD V)	muscle phosphorylase	exercise cramps, myoglobinuria, no lactate rise on exercise
Gaucher	glucocerebrosidase	commonest lysosomal storage disease; splenomegaly, bone crises, Erlenmeyer flask femur, "crumpled tissue paper" cells; enzyme replacement
Tay-Sachs	hexosaminidase A	**cherry-red spot without hepatosplenomegaly**, exaggerated startle
Niemann-Pick	sphingomyelinase	**cherry-red spot with hepatosplenomegaly**, foam cells
Hurler (MPS I) and Hunter (MPS II)	alpha-L-iduronidase; iduronate sulphatase	coarse face, dysostosis; **Hurler has corneal clouding, Hunter (X-linked) does not**

- Clues to a metabolic cause in a sick newborn: well at birth, then poor feeding, vomiting, lethargy and seizures after feeds start, unusual odour, unexplained acidosis, hypoglycaemia or hyperammonaemia; a "sepsis" that does not respond. Send ammonia, blood gas, lactate, glucose, and stop protein and galactose until the cause is known.

Endocrine emergencies of infancy

- **Congenital hypothyroidism**: the **commonest preventable cause of intellectual disability**; most often **thyroid dysgenesis** (ectopic gland commonest), then dyshormonogenesis; in iodine-deficient regions, endemic cretinism. Most babies look normal at birth: prolonged jaundice, constipation, large tongue, umbilical hernia, open posterior fontanelle, hypotonia. **Newborn screening (heel-prick TSH, at 48-72 hours)**; start **levothyroxine 10-15 ug/kg/day within the first 2 weeks** (European consensus 2020); the outlook for intelligence depends on early treatment.
- **Congenital adrenal hyperplasia**: **21-hydroxylase deficiency** in 90-95% (autosomal recessive, CYP21A2): girls are virilised (ambiguous genitalia with normal ovaries and uterus, 46,XX); boys with the salt-wasting form present at 1-3 weeks with vomiting, hyponatraemia, hyperkalaemia and shock. **Raised 17-hydroxyprogesterone** is diagnostic. Treat crisis with IV saline, glucose and hydrocortisone; lifelong hydrocortisone with fludrocortisone. 11-beta-hydroxylase deficiency:

virilisation with **hypertension** (excess 11-deoxycorticosterone).

- **Neonatal hypoglycaemia**: an operational threshold of a blood glucose **below 45 mg/dL (2.5 mmol/L)** for treatment; at risk: preterm, small and large for gestational age, infants of diabetic mothers, asphyxia, sepsis. Feed early; IV 10% dextrose 2 mL/kg then infusion for symptoms or persistence. Persistent hyperinsulinaemic hypoglycaemia (high glucose need, more than 8 mg/kg/min) needs diazoxide.

Type 1 diabetes and DKA in children (ISPAD 2022)

- DKA: glucose above 200 mg/dL (11 mmol/L), **pH below 7.3 or bicarbonate below 18 mmol/L** (ISPAD 2022; older criterion 15), ketonaemia or ketonuria.
- **Fluids first**: one or more boluses of 0.9% saline, usually 10-20 mL/kg each, **over 20-30 minutes, repeated until the circulation is restored** (ISPAD 2022); in frank shock the bolus is given faster, 20 mL/kg in most endocrine protocols, while the UK BSPED 2021 and European Resuscitation Council 2021 give 10 mL/kg aliquots with reassessment; then the deficit replaced over 24-48 hours; **insulin infusion 0.05-0.1 unit/kg/hour started an hour or more after fluids begin, never as a bolus**; potassium in the fluids once urine is passed (or from the start if it is low or normal); bicarbonate almost never.
- **Cerebral oedema is the commonest cause of death in childhood DKA** (headache, slowing pulse, rising blood pressure, falling consciousness): treat with **mannitol 0.5-1 g/kg or 3% saline** and reduce the fluid rate.

Liver disease, thyroid and other gaps (curriculum)

- **Neonatal cholestasis**: conjugated bilirubin above 1 mg/dL (or over 20% of the total) at any age beyond 2 weeks needs urgent work-up; biliary atresia is the surgical cause not to miss (see Neonatology). Hepatitis A in children is usually mild and anicteric; hepatitis B acquired at birth becomes chronic in about 90%.
- **Portal hypertension in children**: commonest cause in South Asia is **extrahepatic portal vein obstruction** (in the West, cirrhosis from biliary atresia leads) (umbilical sepsis, catheterisation); presents with **variceal haemorrhage and splenomegaly with normal liver function**. Treatment: endoscopic band ligation, beta-blocker, and a shunt (meso-Rex) in suitable cases. Cirrhosis in a child suggests Wilson disease (low caeruloplasmin, Kayser-Fleischer rings, copper chelation), biliary atresia, alpha-1 antitrypsin deficiency or autoimmune hepatitis.
- **Hypothyroidism**: juvenile (acquired) hypothyroidism is mainly **Hashimoto thyroiditis** (anti-TPO antibodies), with short stature, delayed bone age, weight gain and a goitre; it is treated with levothyroxine. Endemic goitre and cretinism come from iodine deficiency.
- **Tracheo-oesophageal fistula (surgical emergency to refer)**: with the commonest type (oesophageal atresia with a distal fistula) air passes down the fistula into the stomach, so the **abdomen shows gas**; isolated atresia gives a gasless abdomen; an **H-type fistula** presents later with coughing and recurrent chest infections on feeds and the tube passes freely. Do not feed, keep the upper pouch drained and the baby head up, then operate. **Congenital diaphragmatic hernia** (left Bochdalek, with pulmonary hypoplasia): scaphoid abdomen, bowel loops in the chest on X-ray, respiratory distress at birth; avoid bag and mask ventilation, intubate and pass a nasogastric tube. **Imperforate anus**: no meconium, no anal opening; high lesions need a colostomy first, and a **cross-table lateral film at 24 hours** or later shows the level.

Haematology and oncology

Anaemias

- **Iron deficiency** is the commonest nutritional deficiency and the commonest anaemia of childhood; peak at 6 months to 2 years (exclusive cow's milk, late weaning, prematurity). Microcytic hypochromic film with pencil cells, **low ferritin**, raised TIBC and RDW. **Mentzer index** (MCV divided by red cell count) above 13 favours iron deficiency, below 13 thalassaemia trait. Treat with **elemental iron 3-6 mg/kg/day** (oral, between meals) for 2-3 months after the haemoglobin normalises. Response in order: less irritability and better appetite within 12-24 hours (the earliest clinical sign), marrow response at 36-48 hours, **reticulocytosis from 48-72 hours peaking at 5-7 days (the earliest laboratory sign)**, and a haemoglobin rise of about 1-2 g/dL over the next 4 weeks. Failure to respond: poor adherence, ongoing loss (hookworm) or wrong diagnosis.
- **Hereditary spherocytosis**: autosomal dominant (ankyrin, spectrin, band 3 defects); jaundice, splenomegaly, **raised MCHC**, spherocytes, negative Coombs test, positive **eosin-5-maleimide binding** (older test: osmotic fragility); splenectomy after 5-6 years for severe disease. **G6PD deficiency**: X-linked, haemolysis after fava beans, infection or oxidant drugs (primaquine, dapsone, nitrofurantoin, rasburicase), **Heinz bodies and bite cells**; enzyme assay done after the acute attack.
- **Aplastic anaemia**: pancytopenia with a hypocellular marrow and no splenomegaly or abnormal cells; causes are idiopathic, drugs (chloramphenicol), viral hepatitis and inherited **Fanconi anaemia** (chromosome breakage with diepoxybutane or mitomycin, short stature, radial ray and skin pigment anomalies). Treatment: **matched sibling stem cell transplant**, or immunosuppression (antithymocyte globulin, ciclosporin, with eltrombopag) if no donor. Diamond-Blackfan anaemia is a pure red cell aplasia (macrocytic, thumb anomalies, ribosomal protein genes) treated with steroids.

Thalassaemia and sickle cell disease

- **Beta-thalassaemia major**: no beta chains (HBB mutations; chromosome 11); symptoms start after 3-6 months when HbF falls. Severe anaemia, hepatosplenomegaly, growth failure, **frontal bossing, chipmunk facies, hair-on-end skull** from marrow expansion. **Electrophoresis or HPLC: raised HbF (up to 90%) with absent or very little HbA; HbA2 variable.** Beta-thalassaemia trait: HbA2 above 3.5%, mild microcytic anaemia, normal ferritin. Alpha-thalassaemia: four genes on chromosome 16; four deleted gives **Hb Bart (hydrops fetalis)**, three deleted gives **HbH disease**.
- Treatment of major: regular transfusion to keep the pre-transfusion haemoglobin at 9.5-10.5 g/dL (TIF 2021; 11-12 g/dL with heart disease), with **iron chelation** (deferasirox or deferiprone orally; desferrioxamine by infusion) guided by ferritin and cardiac and liver MRI; the cause of death is **cardiac siderosis**. Splenectomy for hypersplenism; **allogeneic stem cell transplantation from a matched sibling is the established cure** (best before iron overload). Newer, for transfusion-dependent disease in rich countries: **luspatercept** (an erythroid maturation agent, US FDA 2019, adults) lowers transfusion needs, and gene therapies for age

12 or more: **betibeglogene autotemcel** (a lentiviral beta-globin gene, US FDA 2022) and **exagamglogene autotemcel** (CRISPR editing of the BCL11A enhancer to switch HbF back on; UK November 2023, US January 2024).

- **Sickle cell disease**: HbS from a **glutamic acid to valine substitution at beta-6**; autosomal recessive; sickling in hypoxia, acidosis and dehydration. Presents after 4-6 months, when HbF falls: **dactylitis (hand-foot syndrome)** first, then pain crises, **acute chest syndrome**, splenic sequestration, stroke, priapism. **Functional asplenia by early childhood** means infection by capsulated organisms (pneumococcus, Salmonella osteomyelitis): **penicillin prophylaxis from 2 months**, pneumococcal and meningococcal vaccination. **Hydroxyurea** (raises HbF) lowers crises and is advised from 9 months; **transcranial Doppler screening** (age 2-16) finds children at risk of stroke, who are transfused. Electrophoresis: HbS with no HbA in disease, HbA with HbS in trait. **Hydroxyurea remains the main disease-modifying drug** (the exam answer and current practice). Newer drugs and their status: **voxelotor** (an HbS polymerisation inhibitor, US FDA 2019) was **withdrawn from all markets by Pfizer in September 2024** after more deaths and pain crises than placebo; **crizanlizumab** (anti-P-selectin) lost its European licence in 2023 when the STAND trial showed no fewer crises, though it is still licensed in the US; L-glutamine is licensed in the US only (2017). **Gene therapies for age 12 or more with recurrent crises (US FDA December 2023): exagamglogene autotemcel** (CRISPR, raises HbF) and **lovotibeglogene autotemcel** (a lentiviral anti-sickling beta-globin); matched sibling stem cell transplant is the established cure.

Bleeding disorders

- **Haemophilia A** (factor VIII) and **B** (factor IX, Christmas disease) are X-linked recessive; haemarthrosis (knees, elbows, ankles) and muscle haematomas, bleeding after circumcision. **Prolonged aPTT, normal PT, normal platelet count and bleeding time**, because factor VIII sits in the intrinsic pathway; a mixing study corrects. Treatment: replacement of the factor on demand and, for severe disease, **regular prophylaxis** (WFH 2020) with factor concentrate or **emicizumab** (a subcutaneous bispecific antibody bridging factors IXa and X, so mimicking factor VIII, used in haemophilia A with or without inhibitors); about a third of severe haemophilia A patients develop **inhibitors** (alloantibodies to factor VIII; bypassing agents or emicizumab, immune tolerance induction); adult gene therapy is licensed (etranacogene dezaparvovec for haemophilia B, US FDA 2022); tranexamic acid for mucosal bleeds, and **desmopressin for mild haemophilia A**. Severity by factor activity (WFH 2020): **severe below 1%** (spontaneous joint bleeds), moderate 1-5%, mild 5-40%. Cryoprecipitate no longer the first choice. Avoid intramuscular injections and aspirin.
- **Von Willebrand disease**: the commonest inherited bleeding disorder; mucocutaneous bleeding, **prolonged bleeding time** and PFA-100 closure time, **often a prolonged aPTT** (vWF carries factor VIII and protects it from clearance, so factor VIII falls too), normal platelet count and PT, low vWF antigen and ristocetin cofactor activity; desmopressin for type 1.
- **Immune thrombocytopenia (ITP)**: commonest cause of thrombocytopenia in a well child, 1-6 weeks after a viral infection or vaccine; isolated thrombocytopenia with normal haemoglobin and white cells, **normal or increased megakaryocytes**, no splenomegaly. Most recover within 6 months. Observe if no or only skin bleeding, whatever the count (ASH 2019); for mucosal or worse bleeding a **short course of prednisolone (7 days or less) is first choice, with IVIG** when a rapid rise is needed, or anti-D (Rh-positive, non-splenectomised, Coombs-negative); bone marrow is not needed in typical ITP; persistent ITP is 3-12 months, chronic when over 12 months (eltrombopag, romiplostim, rituximab, splenectomy as late options). Beware of leukaemia if the examination or film is not typical.

Acute lymphoblastic leukaemia

- **ALL is the commonest childhood malignancy** (about 75-80% of leukaemias; peak 2-5 years; boys more; Down syndrome raises the risk), mostly **precursor B-cell (CD10-positive)**. Presents with pallor, fever, bleeding and bone pain, lymphadenopathy and hepatosplenomegaly; the film shows blasts, the **bone marrow with 20% or more blasts** diagnoses it (WHO; many paediatric protocols use 25%), and flow cytometry types it. An anterior mediastinal mass is typical of **T-cell ALL**.
- **Prognosis** (the exam favourite): **favourable**: age 1-10 years, white cell count below 50,000/μL at diagnosis, hyperdiploidy (above 50 chromosomes), **t(12;21) ETV6-RUNX1**, rapid response to induction and negative minimal residual disease. **Poor**: age under 1 year or over 10, white count above 50,000/μL, **Philadelphia chromosome t(9;22) BCR-ABL**, KMT2A (MLL) rearrangement, hypodiploidy, T-cell lineage, CNS disease at diagnosis and persistent residual disease after induction. Minimal residual disease is now the strongest predictor.
- Treatment: **induction** (vincristine, corticosteroid, asparaginase, with an anthracycline in higher-risk disease; remission in over 95%), consolidation, **CNS prophylaxis with intrathecal methotrexate**, delayed intensification, then **maintenance for about 2 years** (daily mercaptopurine and weekly methotrexate; some protocols longer for boys). Philadelphia-positive ALL adds **imatinib or dasatinib**. Relapse sites: bone marrow, CNS, testes. For relapsed or refractory disease: **blinatumomab, inotuzumab and CD19 CAR-T cells** (tisagenlecleucel); adding blinatumomab to first-line chemotherapy improved survival in standard-risk B-ALL (COG AALL1731, NEJM 2024). Tumour lysis syndrome (high uric acid, potassium and phosphate, low calcium) is prevented by hydration and allopurinol or **rasburicase**. About 90% of children are now cured. **AML** is the second commonest leukaemia; Auer rods, and acute promyelocytic leukaemia (t(15;17), PML-RARA; DIC is the early danger) is treated with all-trans retinoic acid, now with arsenic trioxide in standard-risk disease.

Lymphomas

- **Hodgkin lymphoma**: adolescents; painless firm cervical or supraclavicular nodes, mediastinal mass, B symptoms (fever, night sweats, weight loss); **Reed-Sternberg cells** (CD15 and CD30 positive); EBV link; nodular sclerosis is the commonest type in adolescents, mixed cellularity more in young children in low-income countries; children are treated on risk-adapted paediatric protocols (for example OEPA/COPDAC, or brentuximab with AVEPC, AHOD1331, NEJM 2022), with radiotherapy only for slow responders on PET; ABVD is the adult regimen; cure over 90%. Staging by the Ann Arbor system with PET scan.
- **Burkitt lymphoma**: B-cell non-Hodgkin lymphoma, **t(8;14) with MYC**, the fastest-growing human tumour (starry-sky

pattern). Endemic (African) form: **jaw and facial bones, EBV**; sporadic form: abdomen (ileocaecal region, intussusception lead point). High-risk of tumour lysis; treated with short intensive chemotherapy and rituximab; excellent cure rate.

- Lymphoblastic lymphoma is the T-cell mediastinal disease treated as ALL.

Neuroblastoma and brain tumours

- **Neuroblastoma**: the commonest extracranial solid tumour and the commonest malignancy under 1 year; arises from neural crest sympathetic tissue, adrenal medulla commonest. Hard, irregular, **abdominal mass that may cross the midline**, with calcification on imaging; metastases to bone marrow and bone (periorbital ecchymoses, "raccoon eyes"), skin (blueberry muffin), and **opsoclonus-myoclonus**; hypertension and diarrhoea (VIP) may occur. **Urinary VMA and HVA raised** (catecholamine metabolites; 5-HIAA is for carcinoid). **MYCN amplification** (chromosome 2p) and age over 18 months mean poor outcome; opsoclonus-myoclonus goes with a better tumour outlook but often leaves neurological deficits. Spontaneous regression: **stage 4S** (INSS: infants **under 12 months**, a small primary with spread limited to liver, skin and under 10% marrow) or its successor **stage MS** (INRG staging 2009: under 18 months) often regresses on its own. Treatment by risk: observation, surgery, chemotherapy, high-dose chemotherapy with stem cell rescue, anti-GD2 antibody (dinutuximab), isotretinoin. **Against Wilms tumour** (details under Renal): Wilms is a smooth flank mass that does not usually cross the midline, rarely calcifies, causes haematuria and renin-driven hypertension, and has normal VMA and HVA, at a mean age of 3-4 years.
- **Brain tumours** are the commonest solid childhood tumours and the second commonest malignancy after leukaemia; in children over 1 year most are **infratentorial** (headache with morning vomiting, ataxia, papilloedema, cranial nerve palsies). **Pilocytic astrocytoma** (cerebellum; cystic with a mural nodule; best prognosis) is the commonest overall; **medulloblastoma** (cerebellar vermis, midline, obstructive hydrocephalus; small blue cells with rosettes; spreads by CSF drop metastases; radiosensitive; molecular groups WNT, SHH, group 3 and 4, WNT best) is the commonest malignant one. **Craniopharyngioma** (suprasellar, calcified, bitemporal hemianopia, growth failure and diabetes insipidus, from Rathke pouch remnants; adamantinomatous type, CTNNB1) is the commonest suprasellar and commonest non-glial brain tumour of childhood (some exam books call it the commonest supratentorial tumour, but supratentorial low-grade gliomas are commoner in registry data, CBTRUS); under 1 year supratentorial tumours predominate; ependymoma fills the fourth ventricle; diffuse midline glioma (brainstem, H3 K27M mutation) has a very poor outlook.

Neurology

Seizures and status epilepticus

- **Status epilepticus**: a seizure lasting **5 minutes or more**, or repeated seizures without recovery between them (the operational definition; the older one was 30 minutes). It is an emergency: airway, oxygen, **check blood glucose** (treat hypoglycaemia with 2-5 mL/kg of 10% dextrose), calcium and sodium, then drugs on a clock.
- **First line (from 5 minutes, the initial phase to about 20 minutes, AES 2016): a benzodiazepine**: IV lorazepam 0.1 mg/kg or IV diazepam 0.2-0.3 mg/kg, repeated once after 5 minutes. **With no IV access: buccal, intranasal or intramuscular midazolam (IM 0.2 mg/kg, buccal 0.3-0.5 mg/kg; maximum 10 mg)**, or rectal diazepam 0.5 mg/kg; **IM midazolam is at least as effective as IV lorazepam** (RAMPART trial, 2012). Respiratory depression is the risk; watch the airway.
- **Second line (established status, about 20-40 minutes, AES 2016)**: IV phenytoin 20 mg/kg (needs cardiac monitoring, give in saline, as it precipitates in dextrose; extravasation injures tissue), **levetiracetam 40-60 mg/kg** (60 mg/kg in ESETT) or **valproate 40 mg/kg**; the ESETT trial (2019) found all three equally effective, about half stopping seizures. Phenobarbital is an alternative (the first drug in neonates).
- **Refractory status** (not controlled by two drugs): rapid-sequence anaesthesia with thiopental or midazolam or propofol infusion on the intensive care unit with EEG monitoring.
- **Childhood absence epilepsy** (4-10 years, girls more): many brief (5-15 second) daily staring spells with eyelid flutter, **provoked by hyperventilation**, no aura, no post-ictal state; **3 Hz generalised spike-and-wave**. **Ethosuximide** is first line for absences alone (better tolerated than valproate, equally effective, and better than lamotrigine; Glauser, NEJM 2010); valproate when tonic-clonic seizures coexist. **Carbamazepine, oxcarbazepine, phenytoin, vigabatrin and gabapentin worsen absences**. Most remit by adolescence.
- Treat the cause (meningitis, hypoglycaemia, electrolytes, poisoning, withdrawal of antiepileptic drugs). Febrile seizures and the drug of choice for each epilepsy type are under Paediatric infections and emergencies.

Hydrocephalus

- Excess CSF with ventricular enlargement. **Obstructive (non-communicating)**: aqueductal stenosis (the commonest congenital cause), Dandy-Walker malformation, posterior fossa tumours, Chiari II with myelomeningocele. **Communicating**: after intraventricular haemorrhage or meningitis, from reduced absorption at the arachnoid villi; over-production by choroid plexus papilloma is rare.
- Infant: rapidly increasing head circumference, **tense bulging anterior fontanelle, split sutures, "setting-sun" sign (upward gaze failure), scalp vein prominence**, irritability and vomiting. Older child: headache, vomiting, papilloedema, gait disturbance, squint. Cranial ultrasound in infants; MRI otherwise.
- Treatment: **ventriculoperitoneal shunt**, or **endoscopic third ventriculostomy** for obstructive hydrocephalus in suitable cases. The commonest shunt complication is **blockage (malfunction)**; infection (Staphylococcus epidermidis, mostly within 6 months) is the second.

Cerebral palsy and intellectual disability

- **Cerebral palsy** is a non-progressive disorder of movement and posture from a lesion of the developing brain; types and causes are under Growth and development. Spastic cerebral palsy is the commonest; management is multidisciplinary (physiotherapy, spasticity drugs including botulinum toxin, orthoses, treating epilepsy and feeding problems).
- **Intellectual disability** (the older term mental retardation): IQ below 70 with impaired adaptive function from the developmental period; mild 50-69, moderate 35-49, severe 20-34, profound below 20 (ICD-10). ICD-11 calls it disorders of intellectual development, and DSM-5-TR grades severity by

adaptive functioning rather than IQ. Commonest genetic cause: **Down syndrome**; commonest **inherited** cause: **fragile X syndrome**; **iodine deficiency is the world's commonest preventable cause of brain damage** (WHO), and **congenital hypothyroidism the commonest treatable one**, found by newborn screening; other causes are fetal alcohol syndrome, perinatal asphyxia, kernicterus, phenylketonuria and infections. Assess with a developmental test (Denver, Bayley, Gesell) and a standard IQ test; investigate by history, examination, thyroid function, metabolic screen, karyotype, fragile X testing and microarray.

Guillain-Barré syndrome and acute flaccid paralysis

- **Guillain-Barré syndrome (GBS)**: acute immune-mediated polyradiculoneuropathy, **1-3 weeks after an infection (Campylobacter jejuni, the commonest trigger, which causes the axonal form, AMAN; also CMV, EBV, Zika)**. **Symmetrical ascending flaccid weakness with areflexia**, minimal sensory loss, back pain, facial and bulbar weakness, autonomic instability. Maximum weakness within 4 weeks.
- **CSF: albuminocytological dissociation** (high protein with a normal cell count, under 10 cells/μL), which may be normal in the first week and is typical after it; nerve conduction studies show slowed conduction or conduction block. A raised cell count suggests poliomyelitis, HIV or Lyme disease instead.
- Treatment: **IV immunoglobulin 2 g/kg over 2-5 days or plasma exchange**; **corticosteroids do not help**. Supportive care is vital: monitor forced vital capacity (ventilate if it falls below 15-20 mL/kg or with bulbar failure) and for autonomic disturbance, and give DVT prophylaxis and pain control. Most children recover well. Variants: Miller Fisher syndrome (ophthalmoplegia, ataxia, areflexia; anti-GQ1b).
- **Acute flaccid paralysis (AFP)** surveillance, for polio eradication: a child under 15 with acute flaccid paralysis of any cause, or any age with suspected polio, is reported at once, and **two stool specimens are collected at least 24 hours apart within 14 days of onset**, with a follow-up examination at 60 days. Causes: polio (asymmetric, fever at the onset of paralysis, no sensory loss, CSF pleocytosis, **residual paralysis at 60 days**), GBS (symmetric, ascending, areflexia, albuminocytological dissociation), transverse myelitis (sensory level, sphincter loss), traumatic neuritis after an injection (one limb, an injection site in the gluteus), critical illness neuropathy and hypokalaemic periodic paralysis.

Muscle disease and the unconscious child

- **Duchenne muscular dystrophy** (X-linked, dystrophin): boys of 3-5 years with a waddling gait, **Gowers sign**, calf pseudohypertrophy, very high **creatine kinase**, **dystrophin gene deletion** on multiplex PCR or MLPA (muscle biopsy if negative); steroids (prednisolone or deflazacort) delay loss of walking, and cardiomyopathy and respiratory failure are the causes of death. Becker dystrophy is a milder in-frame form.
- **Unconscious child**: assess with the **Glasgow Coma Scale** (children under 5 use the paediatric version) or AVPU. ABC first, **blood glucose at once**, then examine for meningism, raised intracranial pressure signs (Cushing triad: hypertension, bradycardia, irregular breathing), posture and pupils. Common causes: infection (meningitis, encephalitis, cerebral malaria), head injury, poisoning, hypoglycaemia, metabolic and seizure (post-ictal or non-convulsive status), shock, Reye syndrome. Treat the cause; CT before lumbar puncture if raised intracranial pressure is suspected.

Sources

Nelson Textbook of Pediatrics (21st ed.); Ghai Essential Pediatrics; Park's Textbook of Preventive and Social Medicine; Robbins Pathologic Basis of Disease; Harrison's Principles of Internal Medicine; Williams Obstetrics (infant of a diabetic mother, antenatal steroids); WHO guidelines on severe acute malnutrition (2013, 2023), the pocket book of hospital care for children (2013), IMCI, immunisation position papers and the WHO routine schedule (2023); AAP hyperbilirubinemia 2022; AHA/AAP Neonatal Resuscitation guidelines 2020 and 2025; ILAE neonatal seizure guideline 2023; European Consensus Guidelines on RDS 2022; HELIX trial (Lancet Glob Health 2021); NASPGHAN/ESPGHAN cholestatic jaundice guideline 2017; Kawasaki disease, AHA 2017 and the AHA scientific statement 2024; GINA 2024; WHO consolidated guidelines on tuberculosis, child and adolescent TB (2022); WHO pneumonia and IMCI recommendations (2014); WHO Immunization Data portal, Nepal's WHO/UNICEF Joint Reporting Form 2025 (read 2026-11-26), and WHO Nepal's HPV campaign report (June 2025); WHO summary of routine immunisation recommendations; ACIP influenza recommendations 2023; AAP Down syndrome health supervision 2022; AAP UTI guideline 2011 (reaffirmed 2016); NICE UTI in under 16s (CG54, updated 2022); NICE nocturnal enuresis CG111; IPNA clinical practice recommendations for steroid-sensitive (2023) and steroid-resistant (2020) nephrotic syndrome; KDIGO glomerular diseases 2021; EULAR/PRINTO/PRES criteria for IgA vasculitis (2010); COG and SIOP-RTSG (UMBRELLA) Wilms tumour protocols; ISPAD 2022 (DKA); ESPGHAN coeliac guideline 2020; European Society for Paediatric Endocrinology congenital hypothyroidism consensus 2020; Global Consensus on nutritional rickets 2016; CFF diagnostic consensus 2017 and CFTR modulator guidance. Further sources for the curriculum additions: Nelson Textbook of Pediatrics (21st ed.); Ghai Essential Pediatrics; Harrison's Principles of Internal Medicine; Robbins Pathologic Basis of Disease; Hoffbrand's Essential Haematology; WHO classification of tumours of the central nervous system (2021); Thalassaemia International Federation guidelines (2021); NHLBI sickle cell disease expert panel (2014) and ASH 2019-2020 guidelines; ASH 2019 ITP guideline; WFH guidelines for the management of haemophilia (3rd ed., 2020); Children's Oncology Group and St Jude risk classifications for ALL; International Neuroblastoma Risk Group staging; Nelson Textbook of Pediatrics (21st ed.); Ghai Essential Pediatrics; Harrison's Principles of Internal Medicine; Bradley and Daroff's Neurology in Clinical Practice; American Epilepsy Society status epilepticus guideline (2016) and ESETT trial (NEJM 2019); RAMPART trial (NEJM 2012); GBS consensus (Willison, Lancet 2016); WHO polio AFP surveillance standards; WHO ICD-10 on intellectual disability; AHA 2015 revised Jones criteria and AHA 2009 rheumatic fever statement; AAP 2017 hypertension guideline; modified Duke criteria; WHO malaria and leishmaniasis guidelines; WHO consolidated HIV guidelines (2016, 2021). Review additions: WHO guidelines for the diagnosis, treatment and prevention of leprosy (2018); WHO Expert Committee on the control of the leishmaniases (TRS 949, 2010); WHO pocket book of hospital care for children (2013) on shock in severe malnutrition; Duke-ISCVID infective endocarditis criteria (2023); US FDA and Pfizer notices on voxelotor's withdrawal (September 2024), EMA on crizanlizumab (2023), FDA approvals of exagamglogene and

lovotibeglogene autotemcel (December 2023) and betibeglogene autotemcel (2022), and FDA's Elevidys safety labelling (November 2025); COG AALL1731 (NEJM 2024); AHOD1331 (NEJM 2022); CBTRUS paediatric brain tumour statistics; ICD-11 and DSM-5-TR on intellectual disability. Also: Nelson Textbook of Pediatrics (21st ed.) on milestones, primitive reflexes, breath-holding spells, congenital infection, parvovirus B19 and the duration of meningitis treatment; Cloherty and Eichenwald Manual of Neonatal Care; AAP clinical report on vitamin K prophylaxis (2022); NRP 2020 and 2025 on meconium; WHO guidelines on hepatitis B (2024) on tenofovir in pregnancy; WHO 2013 severe acute malnutrition guideline and pocket book (ReSoMal); ISPAD 2022 and BSPED 2021 on DKA fluids; WFH guidelines 2020 on haemophilia severity; Glauser et al., NEJM 2010 (childhood absence epilepsy); ILAE epilepsy syndrome classification 2022 and neonatal seizure classification 2021-2023; US FDA approvals of nusinersen (2016), onasemnogene abeparvovec (2019) and risdiplam (2020).