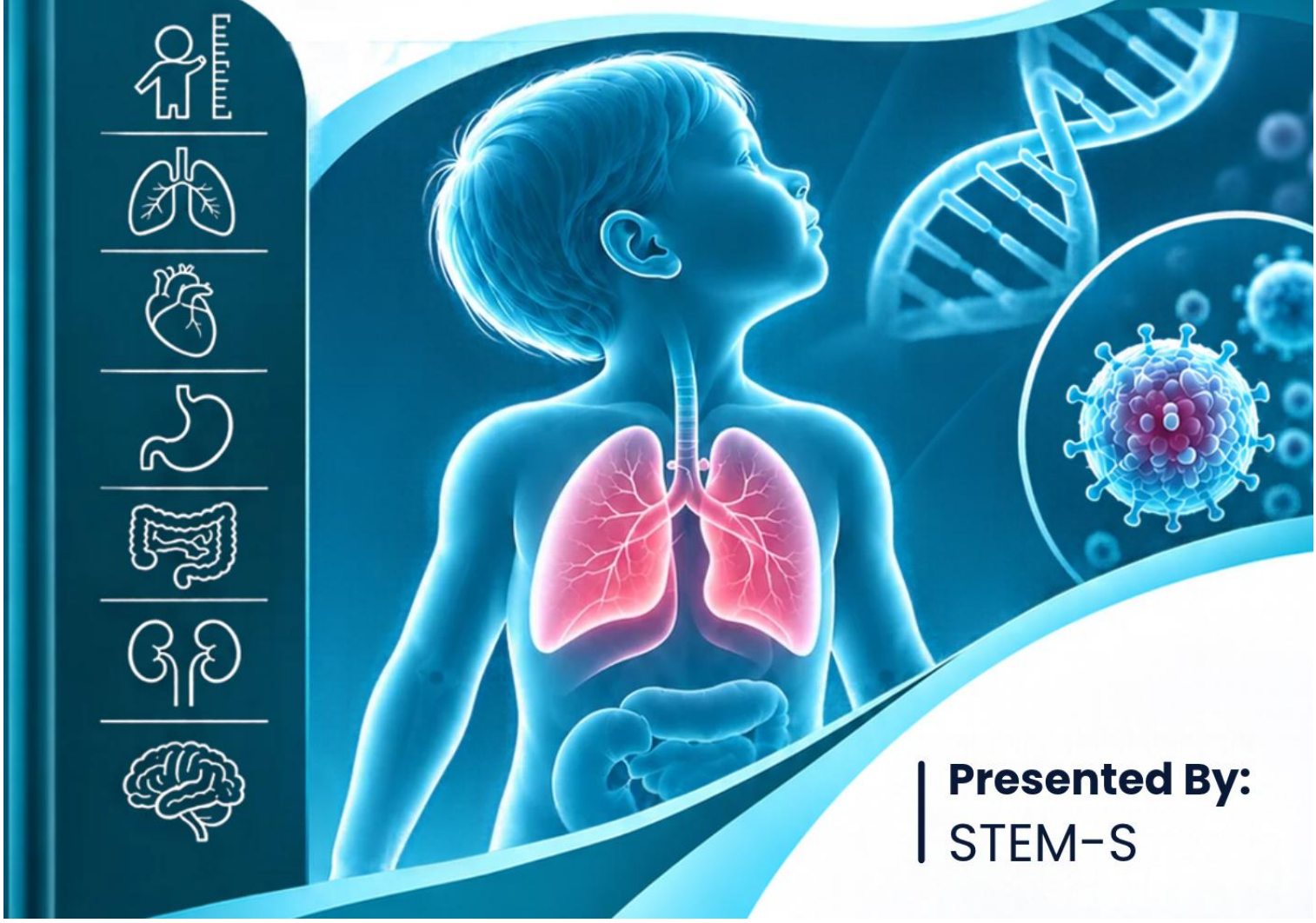
NEET - SS

PEDIATRIC GROUP

LMR | Notes

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INFECTIOUS DISEASE

TUBERCULOSIS (TB) – PEDIATRIC HIGH-YIELD

Organism, transmission and pediatric pattern

- Mycobacterium tuberculosis: acid-fast bacillus; obligate aerobe; slow-growing; airborne transmission from infectious pulmonary/laryngeal TB.
- Children usually develop primary TB; hilar/mediastinal lymphadenopathy is a hallmark. Cavitation and upper-lobe adult-type disease are less common in young children.
- Highest risk of rapid progression/dissemination: infants, children <5 years, HIV infection and other immunocompromised states.
- Primary complex = parenchymal focus + draining lymphatic involvement + regional lymphadenopathy (Ghon focus/complex terminology).
- Ranke complex = calcified primary complex (Ghon focus + calcified hilar nodes).
- Miliary TB = hematogenous dissemination with innumerable tiny nodules; infants have high risk of TB meningitis and disseminated disease.

★ **Important:** A child with TB is often paucibacillary; negative smear does NOT exclude disease. Contact history + clinical/radiological pattern matter.

TB infection tests – TST versus IGRA

Test	What it detects	High-yield interpretation / trap
TST / Mantoux	Delayed-type hypersensitivity to PPD	Read transverse induration at 48–72 h; erythema is not measured.
IGRA	IFN-γ release after stimulation with M. tuberculosis-specific antigens	BCG does not cause a positive IGRA; useful when BCG-vaccinated.
Both	Evidence of TB infection	Neither distinguishes latent TB infection from active TB; neither rules out active TB when negative.

- TST false-positive: BCG (especially recent/repeated vaccination), environmental nontuberculous mycobacteria.
- TST false-negative: severe/miliary TB, TB meningitis, malnutrition, HIV/immunosuppression, very young age, recent infection (window period), viral illness or technical error.
- Common exam thresholds: ≥5 mm in HIV/immunosuppressed or close contacts; ≥10 mm in many high-risk groups/children; ≥15 mm in persons with no known risk.

★ **Important:** A positive TST/IGRA means infection, not necessarily active TB. Always exclude active disease before TB preventive treatment.

PEDIATRIC HEMATOLOGY–ONCOLOGY

INHERITED BLEEDING DISORDERS

1. Hemophilia A and B

- Hemophilia A = factor VIII deficiency; hemophilia B = factor IX deficiency. Both are X-linked recessive; F8 and F9 are located near the end of the long arm of the X chromosome.
- Approximate frequency: 1 in 5,000 males; ~80% hemophilia A and ~20% hemophilia B. About one-third arise from a de novo pathogenic variant.
- Severe hemophilia A: intron 22 inversion in F8 is a classic molecular lesion (around 45–50% of severe HA in many series). Genetic testing identifies the causative variant in the great majority and helps carrier/prenatal testing.
- Female carriers can bleed. Measure factor levels in all known/potential carriers; women with factor activity <40% are considered to have hemophilia and should be managed accordingly. Symptomatic carriers may bleed even with levels above 40%.

Severity classification – repeatedly tested

Severity	Factor VIII/IX activity	Bleeding phenotype
Severe	<1%	Frequent spontaneous bleeding; hemarthroses and deep-muscle bleeds
Moderate	1–5%	Bleeding after mild trauma
Mild	>5% to 40%	Usually after major trauma/surgery; diagnosis may be delayed

- Characteristic bleeding = hemarthrosis, especially ankles early; later knees and elbows. Recurrent bleeding produces a target joint.
- Early joint bleed clue in an older child: warm/tingling sensation before obvious swelling. Repeated hemarthrosis → synovial hypertrophy, cartilage erosion and chronic hemophilic arthropathy.
- Iliopsoas hematoma: groin/lower abdominal pain, hip held flexed (often internally rotated), inability/pain on hip extension; can cause major occult blood loss and mimic appendicitis. Confirm with US/CT/MRI.
- Life-threatening sites: CNS, upper airway, GI tract, iliopsoas or major external hemorrhage. Give factor replacement immediately; if head trauma warrants imaging, factor replacement should precede radiologic evaluation.
- Neonatal clue: neither FVIII nor FIX crosses placenta. Intracranial hemorrhage occurs in ~1–4% of neonates. Absence of bleeding after circumcision does NOT exclude hemophilia.

Laboratory diagnosis

Test	Expected finding	Interpretation / trap
Platelet count	Normal	Primary hemostasis intact
PT	Normal	Extrinsic pathway intact
aPTT	Prolonged, often 2–3× ULN in severe disease	May be normal in mild hemophilia depending on reagent
Mixing study	Correction after 1:1 normal plasma	Factor deficiency; failure/time-dependent loss of correction suggests inhibitor

Congenital hypothyroidism: high-yield facts

- Most common cause of permanent congenital hypothyroidism: thyroid dysgenesis (80–85%); includes agenesis, hypoplasia and ectopic thyroid (lingual thyroid).
- Approximately 30% of congenital hypothyroidism is due to dyshormonogenesis or other causes. Goitre is more suggestive of dyshormonogenesis than dysgenesis.
- Genetic associations : NKX2-1—brain-lung-thyroid syndrome (thyroid dysgenesis, respiratory distress, chorea/ataxia); FOXE1—Bamforth–Lazarus syndrome (thyroid dysgenesis, spiky/curly hair, cleft palate ± choanal atresia); PAX8—thyroid dysgenesis with renal/ureteric anomalies.
- Central hypothyroidism: low free T4 with TSH that is low, normal or inappropriately mildly elevated. Evaluate for other pituitary hormone deficiencies; TSH-only newborn screening can miss central hypothyroidism.
- Primary hypothyroidism: low free T4 with raised TSH. Newborn screening strategy matters—TSH-first detects primary disease; T4-first with reflex TSH can also identify some central disease and TBG deficiency.
- Acquired hypothyroidism: Hashimoto thyroiditis is the commonest cause in iodine-sufficient settings; anti-TPO and anti-thyroglobulin antibodies support diagnosis. Goitre is common but not obligatory.
- Drug causes worth recognizing: excess iodine/amiodarone, lithium, antithyroid drugs, anticonvulsants and selected immune/oncology therapies.

- Levothyroxine: congenital disease requires immediate treatment once confirmed; the supplied LMR already lists 10–15 µg/kg/day. Do not duplicate this dose here. Central hypothyroidism requires free T4-guided monitoring, not TSH alone.

9 Overgrowth / tall stature: endocrine versus non-endocrine

- True pituitary gigantism: accelerated linear growth with elevated IGF-1; distinguish from familial tall stature, obesity (rapid childhood growth with advanced bone age but usually normal adult height), hyperthyroidism and syndromic overgrowth.
- Beckwith–Wiedemann syndrome: macroglossia, omphalocele/umbilical hernia, lateralized overgrowth, macrosomia, neonatal hyperinsulinism/hypoglycaemia; 11p15 imprinting abnormality. Tumour risk includes Wilms tumour and hepatoblastoma.
- Sotos syndrome: NSD1; postnatal overgrowth, macrocephaly/dolichocephaly, prominent forehead, developmental delay and seizures; GH/IGF-1 usually normal—do not label as GH excess.
- Simpson–Golabi–Behmel: GPC3, X-linked; coarse facies, macroglossia, supernumerary nipples and congenital anomalies.
- Marfan vs homocystinuria: both can be tall with arachnodactyly; homocystinuria is associated with developmental disability and lens dislocation (classically downward), while Marfan has aortic root dilatation and lens subluxation (classically upward).

Diagnosis – specimens and tests

Investigation	Best use / exam point
Xpert MTB/RIF or Xpert Ultra	Rapid molecular detection of MTB and rifampicin resistance; Ultra improves sensitivity in paucibacillary samples.
Truenat MTB / MTB-RIF Dx	Chip-based real-time PCR platform used in Indian TB diagnostic pathways.
Culture (liquid MGIT / solid LJ)	Microbiological confirmation and drug-susceptibility testing; slower but important.
Gastric aspirate / gastric lavage	Useful in young children unable to expectorate; early-morning sample, often on consecutive days per protocol.
Induced sputum	Can improve microbiological yield in children able to undergo procedure.
Stool Xpert MTB/RIF Ultra	May be used in children as a non-invasive specimen in appropriate algorithms.
TST / IGRA	Support TB infection; not a microbiological test for active TB.

- Always send samples for molecular testing and culture/DST when feasible, especially in suspected drug resistance or treatment failure.
- CXR: hilar/mediastinal lymphadenopathy, segmental/lobar collapse or consolidation, miliary mottling; CT is more sensitive for lymphadenopathy/airway compression.
- Primary TB may cause airway compression by enlarged nodes → air trapping, hyperinflation, collapse or obstructive pneumonia.

★ **Important:** A negative Xpert in a child does not rule out TB; clinical diagnosis remains important in paucibacillary childhood disease.

Anti-TB drugs – doses, toxicity and interactions

Drug	Usual daily pediatric dose range*	Classic adverse effect / trap
Isoniazid (H)	10 mg/kg (range 7–15; max commonly 300 mg)	Hepatitis, peripheral neuropathy; give pyridoxine to at-risk children.
Rifampicin (R)	15 mg/kg (range 10–20; max commonly 600 mg)	Orange-red body fluids; potent CYP inducer; major ART/contraceptive/anticoagulant interactions.
Pyrazinamide (Z)	35 mg/kg (range 30–40)	Hepatotoxicity, hyperuricemia, arthralgia.
Ethambutol (E)	20 mg/kg (range 15–25)	Optic neuritis; red-green colour discrimination impairment.
Streptomycin	Avoid routine use where alternatives exist	Ototoxicity, nephrotoxicity; injectable-associated toxicity.

*Dose ranges are reference ranges; follow current weight-band NTEP pediatric FDC dosing and adjust to formulation/age/organ function.

- Pyridoxine: commonly 0.5–1 mg/kg/day (usual max 25–50 mg/day in routine supplementation; higher therapeutic doses for neuropathy under supervision); indicated particularly in malnutrition, HIV, breastfeeding infants on INH, pregnancy/adolescence and neuropathy risk.
- M. bovis is intrinsically resistant to pyrazinamide.
- Rifampicin causes orange discoloration of urine, tears and sweat; can stain contact lenses.

★ **Important:** Ethambutol → optic neuritis; isoniazid → neuropathy/hepatitis; rifampicin → enzyme induction; pyrazinamide → hyperuricemia/hepatotoxicity.

Class	Examples	High-yield adverse effects / traps
NNRTI	Efavirenz (EFV), nevirapine (NVP), rilpivirine	EFV → vivid dreams/neuropsychiatric effects; NVP → rash/hepatotoxicity; HIV-2 intrinsically NNRTI resistant.
INSTI	Dolutegravir (DTG), raltegravir (RAL), bictegravir (BIC)	Cation-containing antacids/iron/calcium can reduce absorption; separate dosing as advised.
Protease inhibitors	Lopinavir/ritonavir (LPV/r), atazanavir	GI/metabolic effects; ritonavir is a potent CYP3A inhibitor; atazanavir → indirect hyperbilirubinemia.

- ART goals: durable viral suppression, immune reconstitution, prevention of transmission and normal growth/development.
- Virologic failure requires adherence assessment, repeat viral load and resistance testing where available; do not add a single drug to a failing regimen.
- Rifampicin interactions: markedly lowers levels of many ARVs; DTG dose adjustment or alternative rifamycin/ART strategy may be required under guideline.

★ **Important:** Dolutegravir + polyvalent cations (iron/calcium/magnesium) → reduced absorption; separate administration. TDF is more nephrotoxic than TAF.

Opportunistic infections – CD4 pattern

Immune status (classic adult threshold)	Think of	Pediatric note
CD4 <200 cells/μL	Pneumocystis jirovecii pneumonia (PJP)	In children, use age-specific CD4 percentage/count thresholds for prophylaxis.
CD4 <100 cells/μL	Toxoplasma encephalitis (seropositive), cryptococcosis	Toxoplasma risk depends on serostatus and immune suppression.
CD4 <50 cells/μL	CMV retinitis, disseminated MAC	Classic advanced immunodeficiency association.

PJP / PCP

- Subacute fever, dry cough, progressive dyspnea; hypoxemia may be disproportionate to chest signs; bilateral diffuse ground-glass/interstitial infiltrates.
- Diagnosis: induced sputum/BAL microscopy or PCR; serum β-D-glucan often elevated but nonspecific.
- Treatment: TMP-SMX, TMP component 15–20 mg/kg/day divided q6–8h for 21 days; adjust for renal function.
- Add corticosteroid in moderate-severe hypoxemia (adult criterion PaO_2 <70 mmHg or A–a gradient ≥35 mmHg); pediatric decision uses oxygenation/clinical severity.

★ **Important:** TMP-SMX is first-line treatment and also the preferred prophylaxis for PJP; it also protects against toxoplasmosis.

Toxoplasma encephalitis

- Reactivation in Toxoplasma-IgG-positive severe immunosuppression; classically multiple ring-enhancing lesions, often basal ganglia, with edema.
- Treatment: pyrimethamine + sulfadiazine + leucovorin; TMP-SMX is an alternative in selected settings.
- Steroids only for clinically significant mass effect/edema; anticonvulsants only if seizures occur.

★ **Important:** Multiple ring-enhancing lesions in advanced HIV → toxoplasmosis; solitary lesion or atypical response raises concern for primary CNS lymphoma.

- Liver hypnozoites occur in P. vivax and P. ovale only → relapse. Recrudescence = persistence of blood-stage parasites.
- P. falciparum infects RBCs of all ages → high parasitemia; P. vivax/ovale prefer reticulocytes; P. malariae prefers older RBCs.
- Classic periodicity: P. vivax/ovale tertian (48 h); P. malariae quartan (72 h); falciparum often irregular; know periodicity is not always evident early.

★ **Important:** P. knowlesi has a 24-hour cycle and may progress rapidly; do not dismiss a high parasitemia in a traveler from Southeast Asia.

Clinical features and severe malaria

- Incubation (approximate): falciparum 9–14 days; vivax 12–17 days (can be delayed); ovale 16–18 days; malariae 18–40 days. Partial immunity/chemoprophylaxis may prolong presentation.
- Children may not show classic chills/rigors or periodic fever; symptoms include fever, headache, vomiting, diarrhea, cough, lethargy and poor feeding.
- Common laboratory findings: anemia, thrombocytopenia, normal/raised WBC; splenomegaly/hepatomegaly.

WHO severe-malaria feature	High-yield threshold / definition
Impaired consciousness / coma	GCS <11 in adults or Blantyre coma score <3 in children.
Prostration	Generalized weakness: unable to sit, stand or walk without assistance.
Multiple convulsions	More than 2 episodes within 24 h.
Acidosis	Base deficit >8 mEq/L, bicarbonate <15 mmol/L or venous lactate ≥5 mmol/L; respiratory distress may be the clinical clue.
Hypoglycemia	Glucose <40 mg/dL (<2.2 mmol/L).
Severe malarial anemia	Hb <5 g/dL or Hct <15% in children <12 y with parasitemia >10,000/μL; older patients use age-specific criteria.
Renal impairment	Creatinine >3 mg/dL (adult criterion); interpret pediatric AKI using age/height-based criteria.
Jaundice	Bilirubin >3 mg/dL with high parasite burden (classic WHO criteria include parasitemia >100,000/μL).
Pulmonary edema / ARDS	Hypoxemia, respiratory distress, radiographic pulmonary edema.
Shock	Compensated/decompensated shock with impaired perfusion; assess age-specific BP and perfusion.
Significant bleeding	Recurrent/prolonged bleeding, hematemesis or melena.
Hyperparasitemia	P. falciparum parasitemia >10% (threshold varies by guideline/setting).

C. Diagnosis

- Thick smear: more sensitive for detecting low parasitemia; thin smear: species identification and percentage parasitemia.
- Giemsa-stained thick and thin smears are standard; examine both. If initial smear negative but suspicion remains high, repeat smears every 12–24 h for 3 sets.
- RDT: HRP-2 detects P. falciparum; pLDH/aldolase may detect other species. HRP-2 may remain positive after parasite clearance; gene deletions can cause false-negative HRP2 tests.
- PCR is more sensitive and useful for species confirmation/mixed infection but often too slow for acute treatment decisions.
- Falciparum smear clues: multiple ring forms/RBCs, appliqué forms, double chromatin dots; mature trophozoites/schizonts often sequestered and absent in peripheral blood.

- Emicizumab loading is followed by weekly, every-2-week or every-4-week maintenance schedules; because it distorts aPTT-based assays, use appropriate chromogenic assays for FVIII/inhibitor assessment.

Inhibitors and chronic complications

- Neutralizing alloantibodies to FVIII/FIX are a major complication, especially in severe hemophilia A. Suspect with poor clinical response or unexpectedly low post-infusion factor recovery.
- Inhibitor titre is measured by Bethesda/Nijmegen-Bethesda assay. Low-responding vs high-responding inhibitor status guides bypassing-agent choice and immune-tolerance strategy.
- High-titre inhibitor bleed: bypassing agents (recombinant activated factor VII or activated prothrombin complex concentrate); emicizumab is prophylaxis, not acute hemostasis. Avoid/coordinate aPCC in patients on emicizumab because of thrombotic microangiopathy/thrombosis risk.
- Immune tolerance induction is used to eradicate inhibitors, particularly in hemophilia A. Inhibitor management in hemophilia B can be complicated by allergic reactions and nephrotic syndrome.
- Chronic complications: target-joint arthropathy, muscle contractures, pseudotumour, and historical transfusion-transmitted HBV/HCV/HIV risk (greatly reduced with modern recombinant/safer products).
- Avoid aspirin and NSAIDs that impair platelet function. Encourage safe physical activity, joint-strengthening physiotherapy, vaccination and comprehensive hemophilia-centre care.

2. von Willebrand disease (VWD)

- VWF mediates platelet adhesion to subendothelium and carries/stabilizes FVIII. VWD therefore causes mucocutaneous bleeding and may lower FVIII.

Classification – learn the distinctions

Type	Defect	Typical laboratory/clinical clue
Type 1	Partial quantitative deficiency; commonest (60–80%)	VWF antigen and activity decrease proportionately; mucocutaneous bleeding
Type 1C	Increased VWF clearance	Desmopressin response may be short-lived/ineffective; VWF-containing concentrate may be needed
Type 2A	Qualitative defect with loss of high-molecular-weight multimers	Reduced platelet-dependent activity; abnormal multimer pattern
Type 2B	Increased affinity for platelet GPIb	Thrombocytopenia/platelet aggregation; desmopressin generally avoided
Type 2M	Qualitative platelet-binding defect without loss of multimers	Low activity relative to antigen; multimers relatively preserved
Type 2N	Defective FVIII binding	Low FVIII; can mimic mild hemophilia A; autosomal recessive pattern
Type 3	Virtually complete VWF deficiency; rare/severe	VWF antigen and activity nearly absent, FVIII very low; deep-tissue/joint/CNS bleeding may occur

- Type 1 VWD: VWF:Ag <30 IU/dL supports diagnosis; 30–50 IU/dL is often termed low VWF and interpretation depends on bleeding phenotype/family history. Activity and antigen are usually proportionately reduced.
- VWF activity:antigen ratio <0.7 raises suspicion for type 2 qualitative VWD; multimer analysis/subtype testing differentiates 2A/2B/2M/2N.
- Type 3: VWF antigen/activity essentially zero; FVIII may be <10 IU/dL. Desmopressin is ineffective; treat with VWF-containing concentrate.

- Diagnosis: lymphocyte chromosome-breakage test with diepoxybutane (DEB) or mitomycin C (MMC). Increased breaks, rearrangements, gaps, endoreduplication and chromatid exchanges are characteristic.
- Somatic mosaicism occurs in ~10–15%; peripheral lymphocyte breakage may be falsely less impressive. If suspicion remains high, test skin fibroblasts and/or perform molecular testing.
- Genetic diagnosis: NGS panel of FA genes; pathogenic variants in one of 22 FA genes found in >95% of cases in the supplied source. If negative, consider copy-number analysis and broader genomic testing.
- After diagnosis: minimize ionizing radiation; prefer MRI over CT when possible. Screen congenital organ abnormalities (abdominal ultrasound, echocardiography), growth/endocrine function, renal/liver/thyroid/metabolic/immune systems.
- Surveillance in the supplied text: CBC every 3 months; marrow surveillance approximately yearly (more often if counts decline); monitor glucose and thyroid yearly; careful annual solid-tumour examination.
- HSCT is the only curative therapy for hematologic disease. Use reduced-intensity/radiation-free conditioning because FA cells are hypersensitive to DNA-damaging agents. Outcomes are best before multiple transfusions and before MDS/AML; HLA typing of patient, parents and siblings early.
- Androgens: response in ~70%; reticulocytosis/Hb rise in 1–2 months, WBC then platelets may improve later. Danazol and oxymetholone are commonly used; monitor hepatic adenoma, peliosis, virilization and lipid/LFT effects.

5. Shwachman–Diamond syndrome (SDS)

- Ribosomopathy; classic triad = exocrine pancreatic insufficiency + bone-marrow dysfunction (especially neutropenia) + metaphyseal dysplasia.
- Genes: SBDS (chromosome 7q11; ~80–90% in supplied text), DNAJC21 and EFL1; SRP54 variants may cause a severe congenital neutropenia phenotype without pancreatic insufficiency/metaphyseal dysplasia.
- Pancreatic phenotype: fatty malabsorption from birth; steatorrhea may be subtle. ~50% may improve pancreatic enzyme secretion with age. Pancreatic fatty replacement is characteristic.
- Skeletal clues: metaphyseal dysplasia, osteopenia, delayed secondary ossification centres, short/flared ribs and thoracic dystrophy. Short stature commonly remains below 3rd centile.
- Hematology: neutropenia, anemia and/or thrombocytopenia; recurrent bacterial/fungal infection. Risk of clonal cytogenetic abnormalities, MDS and AML.
- Diagnosis: consider genetic testing in unexplained hypoplastic/aplastic marrow, exocrine pancreatic insufficiency or metaphyseal dysplasia. Genetic testing for SBDS, DNAJC21 and EFL1 is diagnostic in most cases.
- Differentials: Pearson syndrome (sideroblastic anemia, marrow vacuoles, mitochondrial DNA deletion, often severe transfusion-dependent anemia in infancy); Fanconi anemia (positive DEB/MMC breakage); dyskeratosis congenita (short telomeres/mucocutaneous signs).
- Treatment: pancreatic enzyme replacement + fat-soluble vitamins; monitor CBC every 3 months and marrow every 1–3 years (more often if deterioration); G-CSF for profound neutropenia; transfusion support as needed.
- Allogeneic HSCT is the only curative option for severe marrow failure/advanced MDS or leukemia. Conventional myeloablative HSCT has high treatment-related mortality; reduced-intensity fludarabine-based regimens are safer. Graft failure is a concern.

6. Dyskeratosis congenita / telomere biology disorders

- Classic mucocutaneous triad: nail dystrophy, lacy reticular pigmentation of upper chest/neck, oral leukoplakia. Usually nail/skin changes appear in first decade; leukoplakia may appear later.
- Hematologic disease is common: marrow failure in ~90%; severe aplastic anemia in ~50%. Initial abnormality may be thrombocytopenia, anemia or both, progressing to pancytopenia; macrocytosis and elevated HbF.
- Telomeres <1st percentile for age are hallmark. Flow-FISH has ~97% sensitivity and ~91% specificity in the supplied text.
- Genes: DKC1 (X-linked), TINF2, TERT, TERC, RTEL1, ACD/TPP1, PARN, WRAP53/TCAB1, CTC1, POT1, RPA1, NOP10, NHP2. DKC1 most common in classic X-linked form.

1. Acute Lymphoblastic Leukemia (ALL)

Genetics that change prognosis

Finding	Association
B-ALL	~85% of childhood ALL; commonest lineage.
T-ALL	~15%; often adolescent male, high WBC and mediastinal/thymic mass; CNS disease is adverse.
Hyperdiploidy	Favourable-risk cytogenetics.
ETV6::RUNX1, t(12;21)	Favourable-risk B-ALL.
TCF3::PBX1, t(1;19)	Recognized B-ALL genetic subtype; assess risk in protocol context.
BCR::ABL1, t(9;22)	Philadelphia-positive ALL; adverse biology; add a BCR-ABL TKI to chemotherapy.
KMT2A (MLL) rearrangement	Adverse, especially infant ALL.
Hypodiploidy / complex karyotype	Adverse-risk features.
DUX4 rearrangement / ERG deletion	Recognized genomic subtype; generally favourable biology in source table.
IKZF1 deletion / Ph-like profile	Adverse-risk association; Ph-like ALL may activate kinase pathways.
iAMP21	Adverse-risk subtype; protocol-specific intensified therapy.

- Predisposition syndromes: Down syndrome, Fanconi anemia, ataxia-telangiectasia, Bloom syndrome and Nijmegen breakage syndrome.

Risk stratification and response

- NCI standard risk: age 1–9 years AND presenting WBC <50,000/μL. Age <1 year, age ≥10 years, or WBC ≥50,000/μL = high risk by NCI criteria.
- Other adverse factors: T-lineage, CNS involvement, adverse cytogenetics/molecular lesions, slow early response and persistent measurable/minimal residual disease (MRD).
- MRD is more sensitive than morphology and is a major relapse predictor. Always interpret against the protocol-specific time point and assay sensitivity.
- Ph-positive ALL: combine multiagent chemotherapy with a BCR-ABL TKI (e.g., imatinib/dasatinib); not TKI monotherapy.
- Treatment phases: induction → consolidation/intensification → maintenance, with CNS-directed therapy. Total pediatric treatment commonly lasts about 2–3 years.

Favourable pattern	Adverse pattern
Age 1–<10 y; B-cell lineage; low WBC; hyperdiploidy; ETV6::RUNX1; rapid response; low/undetectable MRD	Age <1 or ≥10 y; T-cell lineage; high WBC; CNS disease; hypodiploidy; BCR::ABL1; KMT2A; slow response/persistent MRD

caution: demographic associations in prognostic tables are not stand-alone treatment decisions. Genetics, early response and MRD guide practical risk-adapted therapy.

Pediatric Endocrinology

1 Cushing syndrome: diagnostic approach

- Most common overall cause: exogenous glucocorticoid exposure. In children, always ask about oral, inhaled, topical, injected and other steroid preparations.
- Endogenous cause varies with age: infants—functioning adrenocortical tumour is prominent; children >7 years—Cushing disease (pituitary ACTH-secreting microadenoma) is the commonest endogenous cause.
- Most discriminating pediatric clue: weight gain with reduced height velocity / growth failure. Simple obesity usually has preserved or increased linear growth.
- Other clues: centripetal weight gain, facial plethora, hypertension, pubertal arrest, fatigue/depression and broad violaceous striae.

Cause / pattern	Key points
ACTH-dependent	Pituitary Cushing disease; ectopic ACTH is uncommon in children.
ACTH-independent	Adrenal adenoma/carcinoma, bilateral adrenal hyperplasia/nodular disease.
PPNAD	Pigmented nodular adrenocortical disease; association with Carney complex (lentigines, cardiac myxomas, endocrine tumours).
Iatrogenic	Most common overall; can cause hyperglycaemia, hypertension, weight gain, linear growth retardation and osteoporosis.

Screening and localization

1. Screen for cortisol excess with midnight salivary cortisol; confirm with at least one additional test—24-hour urinary free cortisol or overnight dexamethasone suppression test.
2. Once Cushing syndrome is confirmed, measure ACTH (repeat if needed): low ACTH suggests an adrenal source; elevated/inappropriately normal ACTH suggests ACTH-dependent disease.
3. Pituitary MRI is used to localize a corticotropinoma. Pediatric Cushing disease adenomas are usually microadenomas, commonly <5 mm, and may be difficult to visualize.
4. If localization is uncertain / tests are discordant, bilateral inferior petrosal sinus sampling (BIPSS) may be required.

- High-dose dexamethasone suppression: suppression supports pituitary Cushing disease; lack of suppression favours ectopic ACTH or adrenal tumour in the source algorithm.

Treatment pearls

- Pituitary Cushing disease: trans-sphenoidal microsurgery is the treatment of choice. Persistent/recurrent disease may require reoperation or pituitary irradiation.
- Residual transient hypoadrenalism can follow successful surgery; monitor adrenal function. Height deficit may persist and GH deficiency can become apparent after treatment.
- Steroidogenesis inhibitors mentioned in the supplied chapter for preoperative/refractory control: metyrapone, ketoconazole, aminoglutethimide and etomidate.
- Functioning adrenocortical tumours: surgical removal.

Status epilepticus

- Operational definition: ongoing seizure ≥5 minutes OR recurrent seizures without recovery of consciousness between events. Treat at 5 minutes; do not wait for 30 minutes.
- Priorities: ABC, oxygen, cardiorespiratory monitoring, IV/IO access, bedside glucose, temperature and urgent search for cause.
- Investigations guided by context: glucose, electrolytes (Na, Ca, Mg), renal function, CBC, antiseizure drug levels and toxicology; consider infection/encephalitis, trauma, stroke, haemorrhage, metabolic disease and hypoxia.

Step	Management
1. First-line	Give an adequate benzodiazepine dose (e.g., IV lorazepam; alternatives include IV/IM/intranasal/buccal midazolam or diazepam by available route). One repeat may be given; avoid repeated small under-doses.
2. Second-line	If persistent after two benzodiazepine doses: IV fosphenytoin, levetiracetam or valproate have similar efficacy in older children; lacosamide/phenobarbital may be considered. Phenobarbital is commonly used in neonates.
3. Refractory	Continuous infusion such as midazolam or pentobarbital with airway protection, haemodynamic support and continuous/video EEG; prepare for general anaesthesia if ongoing.

In convulsive status, benzodiazepine first; if it persists after two adequate doses, move promptly to a second-line antiseizure drug. Check glucose immediately.

- Ketogenic diet and cannabinoids were already listed in the original LMR; retain the existing section and expand only with a separate evidence-based table if specifically requested.

Weakness and Hypotonia

UMN versus LMN

Sign	UMN lesion	LMN / peripheral motor unit
Tone	Increased/spastic (may be low acutely)	Decreased/flaccid
Deep tendon reflexes	Brisk, clonus may occur	Reduced/absent
Plantar response	Extensor/Babinski may be present	Flexor/absent
Atrophy	Mild/disuse	Prominent
Fasciculations	Absent	May be present

- Acute spinal cord lesion can initially be flaccid/areflexic (spinal shock); later UMN signs may emerge below the lesion.
- Spinal cord disease clues: sensory level, sphincter dysfunction, back pain/tenderness, autonomic dysfunction or neurogenic shock. Acute flaccid paralysis with a sensory level/bladder symptoms requires urgent spinal imaging.

Spinal muscular atrophy (SMA)

- Progressive degeneration of anterior horn cells; usually autosomal recessive SMN1-related disease. Earlier onset generally predicts more severe phenotype.

- Diagnosis: genetic testing for SMN1; CK normal or mildly elevated. Early diagnosis is critical.
- Disease-modifying treatments: nusinersen (intrathecal antisense oligonucleotide), onasemnogene abeparvovec (one-time IV gene replacement) and risdiplam (oral SMN2 splicing modifier). Earlier/pre-symptomatic treatment gives best outcomes.
- Support: respiratory surveillance, non-invasive ventilation when needed, nutrition/swallowing assessment, physiotherapy, contracture/scoliosis prevention and infection prevention.

SMA: tongue fasciculations + hypotonia + areflexia with preserved sensation/cognition; confirm SMN1. Do not delay disease-modifying treatment while weakness progresses.

Type	Classic phenotype
SMA 1 (Werdnig–Hoffmann)	Severe infantile onset, profound hypotonia/weakness, poor head control, tongue fasciculations, bell-shaped chest, paradoxical abdominal breathing; respiratory failure risk.
SMA 2	Later infantile onset; sits but does not achieve independent walking; progressive proximal weakness.
SMA 3 (Kugelberg–Welander)	Juvenile onset; ambulant initially, proximal weakness and gradual progression.

Peripheral neuropathies

Guillain–Barré syndrome (GBS)

- Often follows respiratory or GI infection (classically Campylobacter jejuni); acute inflammatory demyelinating polyradiculoneuropathy is the commonest form.
- Symmetric ascending weakness, areflexia, hypotonia, paraesthesia/pain; may involve facial, bulbar and respiratory muscles. Autonomic dysfunction: tachy/bradycardia, BP lability, urinary retention/ileus.
- CSF: albuminocytologic dissociation (raised protein with normal/low cell count); may be normal early. NCS/EMG supports diagnosis; MRI nerve-root enhancement may occur.
- Monitor respiratory function, bulbar function, ECG and BP. Admit moderate/severe or rapidly progressive cases; ICU for respiratory/bulbar compromise or autonomic instability.
- Treatment: IVIG or plasma exchange; steroids are not effective for typical GBS. Rehabilitation and prevention of complications are essential.

CIDP and hereditary motor-sensory neuropathy

- CIDP: progressive or relapsing proximal and distal weakness with sensory symptoms over >8 weeks; diagnosis supported by demyelinating NCS. Main treatments: IVIG, corticosteroids or plasma exchange.
- HMSN/Charcot–Marie–Tooth: inherited progressive distal weakness, foot drop, pes cavus/high arches, frequent tripping and distal wasting; sensory loss variable. CMT1 typically demyelinating; CMT2 axonal.

Tick paralysis

- Acute ascending flaccid paralysis, sometimes with bulbar/ocular weakness, due to tick salivary neurotoxin blocking neuromuscular transmission. Search carefully for attached tick, especially scalp/hair-bearing areas; removal leads to prompt improvement.

Structural airway disorders

• Laryngomalacia: commonest cause of congenital/infant stridor. Inspiratory stridor worsens with feeding, crying, agitation or supine position and improves upright/prone. Peaks around 3–5 months; most resolve by 12–24 months. Flexible nasopharyngolaryngoscopy confirms dynamic supraglottic collapse. Observe if thriving; severe apnea, hypoxaemia, failure to thrive or feeding compromise may require supraglottoplasty.

• Subglottic stenosis: biphasic stridor; congenital or acquired after prolonged/repeated intubation, especially in premature infants. Diagnosis by endoscopic assessment. Mild cases may improve with growth; severe disease may need endoscopic/surgical intervention or tracheostomy.

• Airway haemangioma: early infancy, progressive/biphasic stridor, may mimic recurrent croup. Propranolol is usual first-line systemic treatment for clinically significant infantile haemangioma under specialist care.

• Recurrent respiratory papillomatosis: HPV-6/11; progressive hoarseness/weak cry and stridor. Endoscopic diagnosis; repeated debulking/laser is common. Avoid tracheostomy if possible because of distal airway seeding risk.

• Vocal-cord paralysis: bilateral disease may cause severe airway obstruction; unilateral disease more often causes weak cry/aspiration. Evaluate swallowing (VFSS) if aspiration is suspected. Traumatic recurrent laryngeal nerve palsy may recover over 3–6 months.

• Adenotonsillar hypertrophy: chronic nasal obstruction, mouth breathing, snoring and OSA; may obstruct Eustachian tube and predispose to otitis media. Adenotonsillectomy is most effective when adenotonsillar tissue is the major cause of OSA.

Tracheomalacia, vascular compression and congenital lung lesions

• Tracheomalacia is excessive dynamic collapse due to deficient tracheal cartilage/support, usually most evident during expiration or coughing. Clues: recurrent infections, brassy/barking cough, monophonic wheeze and noisy expiration.

• Dynamic bronchoscopy is the reference assessment; >50% expiratory reduction in airway calibre is a commonly used threshold, interpreted with symptoms. Mild–moderate disease often improves with growth; severe cases may require CPAP, aortopexy/tracheopexy or selected airway intervention.

• Differentiate intrinsic malacia from extrinsic compression. Vascular rings/slings may cause respiratory symptoms plus dysphagia. Barium swallow may show oesophageal indentation; CT angiography/MR angiography defines vascular anatomy. Double aortic arch forms a complete vascular ring.

Lesion	Exam clues	Diagnosis / management
Congenital lobar emphysema	Hyperinflated lobe compresses adjacent lung; neonatal distress or later symptoms; can mimic pneumothorax.	CXR/CT; lobectomy if severe/progressive compromise; observe selected asymptomatic cases.
CPAM	Cystic/solid developmental lesion; antenatal detection or recurrent infection.	Contrast chest CT best defines lesion; symptomatic/large lesions need surgical assessment; selected asymptomatic lesions may be observed.
Pulmonary sequestration	Non-functioning lung tissue with systemic arterial supply; intralobar within pleura, extralobar has own pleura.	CT angiography identifies systemic feeding vessel; resection/embolisation according to symptoms/anatomy.
Pulmonary hypoplasia	Reduced lung/alveolar development; CDH, oligohydramnios, CHD or absent pulmonary artery associations.	Assess underlying cause and pulmonary hypertension; supportive care.
Vascular ring/sling	Stridor, wheeze, recurrent infections, feeding difficulty/dysphagia.	CT angiography/MRA; surgical repair for significant symptomatic compression.

Coeliac disease

- Immune-mediated small-bowel injury triggered by gluten (wheat, barley, rye) in genetically susceptible individuals.
- Presentation: chronic diarrhoea, abdominal distension, poor growth, delayed puberty, iron-deficiency anaemia, osteopenia, dental enamel defects, elevated transaminases or dermatitis herpetiformis; may be minimally symptomatic.
- Screen while the child is consuming gluten: tissue-transglutaminase IgA (tTG-IgA) plus total serum IgA. If IgA deficient, use IgG-based testing (e.g. DGP-IgG or tTG-IgG).
- Positive serology generally requires specialist assessment and, in many cases, duodenal biopsy. Selected children may qualify for a no-biopsy pathway under strict guideline criteria (very high tTG-IgA titre plus confirmatory EMA-IgA on a second sample).
- Do not start a gluten-free diet before diagnostic testing; antibodies may fall and biopsy may normalise.
- Treatment: lifelong strict gluten-free diet with dietitian support; monitor growth, symptoms, serology and nutritional deficiencies (iron, folate, B12, calcium, vitamin D).

Exam pearl: tTG-IgA is the preferred first-line serological test, but always check total IgA.

Cow's milk protein allergy (CMPA) / allergic proctocolitis

Phenotype	Typical presentation
IgE-mediated	Minutes to 2 hours: urticaria, angioedema, wheeze, vomiting or anaphylaxis.
Non-IgE-mediated	Delayed GI symptoms: blood/mucus in stool, vomiting, diarrhoea, feeding difficulty or poor growth.
Food protein-induced allergic proctocolitis	Otherwise well young infant with blood-streaked stools; usually improves after eliminating cow's milk protein.

- Diagnosis is based on compatible symptoms, improvement with elimination and recurrence on supervised reintroduction/challenge where appropriate.
- Breastfed infant: continue breastfeeding; maternal dairy elimination may be trialled when symptoms support CMPA, with calcium/vitamin D adequacy.
- Formula-fed infant: extensively hydrolysed formula is first-line for most; amino-acid formula for severe disease or failure to respond.
- Do not confuse lactose intolerance (non-immune carbohydrate malabsorption) with CMPA (immune-mediated protein reaction).

Intussusception

- Most common in infants 6–36 months; ileocolic type is typical. Most cases in this age group are idiopathic and may follow viral illness/lymphoid hyperplasia.
- Older child or recurrent intussusception → search for pathological lead point (Meckel diverticulum, polyp, lymphoma, duplication cyst or HSP).
- Classic triad: episodic colicky abdominal pain, vomiting and red currant-jelly stool; the full triad is not always present. Lethargy may be the only clue.
- Ultrasound: target/doughnut sign on transverse view and pseudokidney sign longitudinally.
- Stable child without perforation/peritonitis: pneumatic (air) or hydrostatic enema reduction is first-line and diagnostic/therapeutic.
- Peritonitis, perforation, shock or failed enema reduction → surgery. Recurrence can occur after successful enema reduction.

Wilson disease – additional clinical points

- Presentation may be hepatic, neurological or psychiatric. Neurological clues include tremor, deteriorating handwriting, dysarthria, declining school performance and behavioural change.
- Kayser–Fleischer rings support the diagnosis but may be absent in isolated hepatic disease, especially in younger children.
- Treatment: copper chelation with D-penicillamine or trientine; zinc reduces intestinal copper absorption and may be used in selected patients/maintenance. Monitor urinary copper and treatment toxicity.
- Fulminant Wilson disease may present with Coombs-negative haemolytic anaemia and severe liver failure; urgent transplant assessment is required.

Autoimmune hepatitis – additional clinical points

- Type 1: ANA and/or ASMA; Type 2: anti-LKM-1 (and sometimes anti-LC1), often in younger children.
- Elevated IgG supports diagnosis; liver biopsy typically shows interface hepatitis with lymphoplasmacytic/plasma-cell-rich infiltrate.
- Treatment is corticosteroid induction (prednisolone) with azathioprine as a steroid-sparing agent when appropriate; monitor transaminases and IgG for biochemical remission.
- Relapse after withdrawal is common enough that treatment cessation should be cautious and specialist-led.

D. Pancreatic disease

Exocrine pancreatic insufficiency

- CF is the leading paediatric cause; other causes include Shwachman–Diamond syndrome, Pearson syndrome and severe malnutrition.
- Bulky, foul-smelling, oily stools; increased appetite with poor weight gain; fat-soluble vitamin deficiency and protein-calorie malnutrition are typical.
- Faecal elastase-1 is a useful non-invasive test; low levels support exocrine insufficiency. Watery stool can dilute the sample and produce a falsely low result.
- Faecal fat may be assessed by Sudan stain or 72-hour quantitative faecal fat. Imaging may show pancreatic structural disease but does not directly quantify exocrine function.
- PERT: use enteric-coated microspheres with meals/snacks; sprinkle capsule contents on a small amount of acidic soft food if needed. Dose is titrated to stool quality, growth and symptoms.

Age / group	Dose quoted in supplied chapter
Infants	2,000–4,000 lipase units per 120 mL formula
Children <4 years	1,000 lipase units/kg/meal
Older children	500 lipase units/kg/meal
Safety ceiling	Avoid >2,500 units/kg/meal; very high doses (>6,000 units/kg/meal) are associated with fibrosing colonopathy

Doses must be individualised to the product and local CF/pancreatic insufficiency protocol; include snack dosing and avoid exceeding recommended daily limits.