Pediatrics 6th Year — Past Questions Study Notes

Organized by System & Topic • All High-Yield Points Extracted from Past Questions

1. HEMATOLOGY & ONCOLOGY

1.1 Leukemia

ALL (Acute Lymphoblastic Leukemia)

- MC malignancy in children; hematopoietic malignancy (ALL) is the MCC of malignancy in children
- ~85% derived from B-cell progenitors (NOT T-cell) (past question)
  - ✗ **WRONG:** Most cases of ALL are derived from T-cell progenitors → **CORRECT:** ~85% are B-cell progenitors; T-cell ALL is a minority
- B-cell markers : CD19, CD10, CD20; T-cell: CD3, CD7
  - ✗ **WRONG:** B-cell marker is CD56 → **CORRECT:** CD56 is NK cell; B-cell markers include CD19
- Staging based on bone marrow biopsy and CSF examination
- Chromosomal abnormalities identified in most cases
- Can present with pallor, thrombocytopenia, lower limb bone pain, leukocytosis
- First test to order if leukemia suspected: Blood film (smear) — NOT flow cytometry or bone marrow first

- Poor prognostic factors in ALL:
  - WBC count > 50,000
  - Age < 1 year
  - Slow response to therapy
  - Abnormal cytogenetics
  - t(9;22) Philadelphia chromosome → WORST prognosis
  - ✗ **WRONG:** t(9;22) has the best prognosis → **CORRECT:** t(9;22) has the WORST prognosis in ALL
  - ✗ **WRONG:** Gender is a poor prognostic factor → **CORRECT:** Gender is NOT a poor prognostic factor in ALL

- Predisposing factors for leukemia:
  - Down syndrome (trisomy 21) — increased risk of ALL and AML
  - Fanconi anemia
  - Ataxia telangiectasia
  - Myelodysplastic syndrome
  - Bloom syndrome
  - ✗ **WRONG:** Bartter syndrome predisposes to leukemia → **CORRECT:** Bartter syndrome does NOT predispose to leukemia
  - ✗ **WRONG:** Hereditary spherocytosis predisposes to hematological tumors → **CORRECT:** Hereditary spherocytosis is NOT a risk for lymphoma/leukemia

- Down syndrome and leukemia: increased susceptibility to ALL; AML associated with trisomy 21 (past question)
  - ✗ **WRONG:** AML most common hematological disorder in Down syndrome → **CORRECT:** ALL (not AML) is the most common
- Exposure to medical radiation → increased incidence of ALL

AML (Acute Myeloid Leukemia)

- Pancytopenia + 9% blasts + t(15;17) → AML (M3)
- Auer rods seen on histology in AML — NOT in neuroblastoma

1.2 Lymphoma

- Hodgkin's lymphoma: painless cervical lymph nodes, dyspnea, weight loss, anterior mediastinal mass on CXR
- Fever, night sweats, weight loss = 'B symptoms'; painless cervical nodes → biopsy
- Anterior mediastinal mass in child → think Hodgkin's or T-ALL
- Reed-Sternberg cells in Hodgkin's

1.3 ITP (Immune Thrombocytopenic Purpura)

- MCC of isolated thrombocytopenia in otherwise healthy children
- Usually follows URTI; antibody-mediated platelet destruction
- Classical presentation: sudden onset purpura; well-appearing child
- Isolated thrombocytopenia, normal Hb, normal WBC, no splenomegaly
  - ✗ **WRONG:** ITP is usually associated with splenomegaly → **CORRECT:** No splenomegaly in ITP
  - ✗ **WRONG:** Steroids decrease chronic ITP cases → **CORRECT:** Prognosis is excellent even without therapy

- First-line mild ITP with platelets ~50k: no treatment (just observe)
- Treatment options: oral prednisolone, IV immunoglobulin, IV anti-D
- Least effective: platelet transfusion (antibodies destroy transfused platelets too)
- Anti-platelet antibodies diagnostic
- Bone marrow exam NOT necessary for diagnosis in typical cases
- Aspirin is CONTRAINDICATED in ITP — causes Reye's syndrome AND inhibits platelet aggregation
  - **(past question)** Child with ITP and fever — aspirin is contraindicated because it may cause Reye's syndrome

1.4 Henoch-Schonlein Purpura (HSP / IgA Vasculitis)

- MCC of non-thrombocytopenic purpura in children
- Most common childhood vasculitis — affects small vessels
- IgA deposits in skin biopsy (leucocytoclastic vasculitis)
- Normal platelet count, normal coagulation profile (PT/PTT normal)

- Classic tetrad: palpable purpura (required), arthritis/arthralgia, abdominal pain/bleeding, renal involvement
- Most defining and required feature: palpable purpuric RASH (not renal involvement)
- Rash: symmetrical lower limb and buttock purpura

- Complications: intussusception, nephritis (hematuria/proteinuria), chronic renal failure (rare)
  - ✗ **WRONG:** Nephritis is not a known complication of HSP → **CORRECT:** Nephritis IS a known complication
  - ✗ **WRONG:** Arthritis is a rare manifestation of HSP → **CORRECT:** Arthritis is COMMON in HSP
  - ✗ **WRONG:** C3 is low in HSP → **CORRECT:** C3 is NORMAL in HSP (unlike MPGN or SLE)

- Treatment: ibuprofen for joints; steroids for abdominal pain (preferred over ibuprofen)

- Follow-up test: urinalysis (monitor renal involvement)
  - **(past question)** HSP case — most important follow-up test: urinalysis

1.5 Hemolytic Uremic Syndrome (HUS)

- Triad: microangiopathic hemolytic anemia (schistocytes), thrombocytopenia, acute renal failure
  - ✗ **WRONG:** Coombs-positive hemolytic anemia in HUS → **CORRECT:** Coombs is NEGATIVE in HUS (mechanical, not immune hemolysis)
- Post-diarrheal HUS (typical) — caused by E. coli O157:H7 (Shiga toxin)
- Best treatment of post-diarrheal HUS: appropriate fluid balance
- Platelet transfusion is NOT used in HUS
- Other findings: colitis, hematuria/proteinuria, jaundice, seizures
  - ✗ **WRONG:** Uveitis can be seen in HUS → **CORRECT:** Uveitis is NOT seen in HUS
- One-year-old with pallor, Hb 7, platelets 30k, elevated urea/creatinine, schistocytes → HUS

1.6 Sickle Cell Disease

- Sickle cell disease is normocytic (NOT microcytic)
  - ✗ **WRONG:** Sickle cell is microcytic hypochromic → **CORRECT:** Sickle cell is NOT microcytic — it is normocytic

- Types of crisis:
  - Aplastic crisis: sudden severe anemia + reticulocytopenia (caused by parvovirus B19)
  - Sequestration crisis: sudden severe anemia + splenomegaly
  - Vaso-occlusive (painful) crisis: most common
  - Hemolytic crisis: increased hemolysis

- Stroke in sickle cell: exchange transfusion; chronic transfusion to prevent subsequent strokes
- Newly diagnosed infant: start prophylactic penicillin (not transfusions, not iron chelation first)

1.7 G6PD Deficiency

- X-linked recessive — affects males primarily; females can be affected as carriers
  - ✗ **WRONG:** Since gene is on X chromosome, females are not affected → **CORRECT:** Females CAN be affected (homozygous or X-inactivation)
- Triggered by: fava beans, naphthalene (moth balls), antimalarials, sulfonamides, nitrofurantoin, infections
  - ✗ **WRONG:** Penicillin is avoided in G6PD → **CORRECT:** Penicillin is SAFE in G6PD; avoid sulfonamides, antimalarials
- Presents 24–48 hours after oxidant exposure: pallor, jaundice, dark urine (hemoglobinuria)
- Blood film: Heinz bodies (denatured Hb)
- Elevated reticulocyte count during acute hemolysis
- G6PD levels may be falsely normal during acute hemolysis (reticulocytes have higher G6PD)
- Definitive diagnosis: G6PD enzyme assay (NOT Hb electrophoresis)
  - ✗ **WRONG:** Hb electrophoresis is the definitive diagnostic test for G6PD → **CORRECT:** G6PD enzyme assay is the definitive test
- Treatment: supportive; avoid triggers
- G6PD can cause neonatal jaundice

    - ✗ **WRONG:** G6PD does not cause neonatal jaundice → **CORRECT:** G6PD CAN cause neonatal jaundice
- Patient with total bilirubin 10, direct bilirubin 4 (indirect-predominant) — G6PD is NOT in DDx (G6PD causes unconjugated hyperbilirubinemia)

1.8 Iron Deficiency Anemia (IDA)

- MCC: poor dietary intake (most common cause in children)
- Microcytic hypochromic anemia
- Lab findings: low ferritin, low serum iron, HIGH TIBC (total iron binding capacity), low MCV, elevated RDW
    - ✗ **WRONG:** TIBC is decreased in IDA → **CORRECT:** TIBC is INCREASED in IDA
- Not common before 6 months (protected by maternal stores)
    - ✗ **WRONG:** IDA is common at 4–6 months of age → **CORRECT:** IDA is NOT common before 6 months of age

- Iron supplementation for exclusively breastfed infants: starts at 4–6 months (NOT at birth)
    - ✗ **WRONG:** Iron supplementation of breastfed babies should start at birth → **CORRECT:** It starts at 4–6 months, not at birth

- First response after iron supplementation: reticulocytosis (within days)
    - ✗ **WRONG:** First change after iron therapy is normalization of Hb → **CORRECT:** First change is reticulocytosis
    - ✗ **WRONG:** First primary response is increase in Hb level → **CORRECT:** First response is reticulocytosis, then improved mood/appetite, then Hb

- After discontinuation of iron therapy: decrease milk to 2–3 cups/day (excessive milk is a risk factor for IDA)
- PICA is associated with IDA
- Manifestations: pallor, irritability, PICA, developmental/intellectual effects
- Basophilic stippling on blood film → test for lead toxicity (additional cause)
    - **(past question)** Child with IDA and basophilic stippling — should test for lead

- 12-month-old with Hb 9, MCV 65, RDW 20%, reticulocytes 1.2% — predisposing factor: poor dietary intake

1.9 Thalassemia

Beta-Thalassemia Major

- Presents ~6 months when HbF switches to HbA
- Severe anemia, Hb very low, HbF elevated on electrophoresis, HbA absent/very low, HbA2 variable
- Requires chronic transfusion therapy
- Complications of iron overload: requires chelation
- Splenectomy indication: hypersplenism (progressive splenomegaly, increasing transfusion needs)
    - **(past question)** 12-year-old on chronic transfusions with progressive splenomegaly and constipation/dyspnea — management: splenectomy

- Electrophoresis: A1=3%, A2=2.4%, F=94.6% at 8 months → Beta-thalassemia major (HbF predominant)
- Diagnosis confirmed by: high HbF level
- Complication of thalassemia major: cholecystitis, Yersinia gastroenteritis (iron overload + chelation), appendicitis, pancreatitis

- ✗ **WRONG:** Abdominal veno-occlusive crisis is a complication of thalassemia major → **CORRECT:** Veno-occlusive crisis is a sickle cell complication, NOT thalassemia

Beta-Thalassemia Minor (Trait)

- Mild anemia, low MCV, normal/elevated RDW, elevated HbA2 on electrophoresis
  - ✗ **WRONG:** Extramedullary hematopoiesis causes atypical facies in thalassemia minor → **CORRECT:** This is a feature of MAJOR, not minor
- Features: high HbA2, mild anemia, normal or increased Fe, microcytic hypochromic
- Case: Hb 9.2, microcytosis, normal RDW, elevated HbA2 → Beta-thalassemia trait
- Case: MCV 63, RDW 13%, Hb 10, reticulocytes 2.1% → Thalassemia minor (normal RDW distinguishes from IDA)

Alpha-Thalassemia

- 4 alpha gene deletion → Hydrops fetalis (Hb Bart's disease) — incompatible with life

1.10 Hereditary Spherocytosis

- Autosomal dominant inheritance
- Extravascular hemolysis: anemia, jaundice, splenomegaly, family history
- Blood film: spherocytes
- Diagnosis: osmotic fragility test
  - ✗ **WRONG:** Splenectomy improves but does not eliminates hemolysis in hereditary spherocytosis → **CORRECT:** Splenectomy eliminates hemolysis in hereditary spherocytosis
- Individuals may be asymptomatic without anemia
- Newborn: may present with anemia and hyperbilirubinemia
- High risk for aplastic crisis with parvovirus B19 infection (not vaso-occlusive)
- Complication of splenectomy: thrombosis
- Father had splenectomy + child with spherocytes + reticulocytes 5% + Hb 8 → osmotic fragility test

Autoimmune Hemolytic Anemia (AIHA)

- Coombs-positive hemolysis
- Spherocytes + polychromasia + nucleated RBCs + positive Coombs = AIHA
- Triggered by febrile illness
- Key distinction from hereditary spherocytosis: Coombs positive in AIHA

1.11 Hemophilia

Hemophilia A (Factor VIII Deficiency)

- X-linked recessive
- Presents with hemarthrosis, prolonged PTT, normal PT, normal bleeding time, normal platelet count
  - ✗ **WRONG:** Hemophilia A increases bleeding time → **CORRECT:** Bleeding time is NORMAL in hemophilia (platelet plug forms)
  - ✗ **WRONG:** Hemophilia A is deficiency of factor 9 → **CORRECT:** Hemophilia A = factor 8 deficiency; Hemophilia B = factor 9
- Factor VIII < 1%: spontaneous bleeding

- Treatment: factor VIII concentrate (or cryoprecipitate if concentrates unavailable — for SEVERE hemophilia A)
- DDAVP: used for mild hemophilia A (releases vWF and factor VIII) — NOT for severe or hemophilia B
  - ✗ **WRONG:** DDAVP is treatment of choice for hemophilia B → **CORRECT:** DDAVP is NOT effective in hemophilia B
- Carrier female: 50% chance each male child is affected
- Development of inhibitors: most common reason for breakthrough bleeding on prophylaxis
  - **(past question)** Child on prophylactic factor VIII with spontaneous hemarthrosis → likely developed factor inhibitors

Hemophilia B (Factor IX Deficiency / Christmas Disease)

- X-linked recessive, clinically identical to hemophilia A
- PTT prolonged, normalizes when mixed with normal plasma (factor deficiency, not inhibitor)
- Bleeding time normal

Conditions Causing Prolonged PT

- Prolonged PT (extrinsic pathway): liver disease, vitamin K deficiency, warfarin, cystic fibrosis (fat-soluble vitamin malabsorption)
  - ✗ **WRONG:** Hemophilia causes prolonged PT → **CORRECT:** Hemophilia causes prolonged PTT only (not PT)

1.12 von Willebrand Disease (vWD)

- Most common inherited bleeding disorder
- Autosomal dominant
- Prolonged aPTT, normal PT, normal platelet count, low factor VIII (vWF carries factor VIII)
- Presents: epistaxis, menorrhagia, easy bruising
  - ✗ **WRONG:** vWD causes bleeding into joints and muscles → **CORRECT:** Joint/muscle bleeding is hemophilia; vWD causes mucosal bleeding
- Female with menorrhagia + epistaxis + low factor VIII + normal PT + normal platelets → vWD (not hemophilia)

1.13 Solid Tumors

Wilms Tumor (Nephroblastoma)

- MCC abdominal mass in children; embryonal tumor of the kidney
- Median age: ~3–4 years (NOT 15 years)
- Presents: incidental non-tender abdominal mass, hematuria, hypertension
- Associated syndromes: WAGR (Wilms, Aniridia, Genitourinary anomalies, intellectual Retardation), Beckwith-Wiedemann, Denys-Drash
- Bilateral Wilms tumor: NOT rare (especially in WAGR/Beckwith-Wiedemann)
  - ✗ **WRONG:** Bilateral Wilms tumor is very rare → **CORRECT:** Bilateral Wilms occurs in ~5–10% of cases, especially with syndromes
- Calcifications are NOT commonly seen on imaging (unlike neuroblastoma)
  - ✗ **WRONG:** Calcifications are commonly seen in Wilms tumor imaging → **CORRECT:** Calcifications are common in NEUROBLASTOMA, not Wilms
- Hematuria: common. Hypertension: common (due to renin secretion)

- ✗ **WRONG:** BP is always normal in Wilms tumor → **CORRECT:** Hypertension IS common in Wilms tumor
- ✗ **WRONG:** Wilms tumor can present in mediastinum → **CORRECT:** Wilms tumor does NOT present in mediastinum
- ✗ **WRONG:** Neuroblastoma presents with hematuria → **CORRECT:** Hematuria is a feature of WILMS tumor, not neuroblastoma
- **(past question)** 12-month-old boy, left upper quadrant mass, hematuria → Wilms tumor
- **(past question)** Newborn with bilateral aniridia → associated with Wilms tumor
- **(past question)** Fundoscopic finding associated with Wilms: aniridia (WAGR syndrome)

Neuroblastoma

- Arises from neural crest cells / sympathetic nervous system (NOT metanephric blastoma)
  - ✗ **WRONG:** Neuroblastoma arises from metanephric blastoma → **CORRECT:** Neuroblastoma arises from neural crest/sympathetic nervous system
- Abdominal mass + hypertension (catecholamine production or renal artery compression)
- Metastasizes to: bones, bone marrow, skin, liver, orbits
- Clinical features: proptosis/periorbital ecchymosis (raccoon eyes), opsoclonus-myoclonus (dancing eyes and feet), watery diarrhea (VIP), large liver, abnormal eye movements
- Opsoclonus-myoclonus (dancing eyes/feet) → associated with neuroblastoma
- Horner syndrome with supra-mediastinal mass → neuroblastoma (paraspinal sympathetic ganglion involvement)
  - ✗ **WRONG:** Auer rod bodies on histology in neuroblastoma → **CORRECT:** Auer rods are in AML, NOT neuroblastoma

Retinoblastoma

- Presents with leukocoria (white pupillary reflex / absent red reflex)
- Hereditary retinoblastoma: bilateral, autosomal dominant, penetrance ~90%
- Sporadic: unilateral, somatic mutation
  - ✗ **WRONG:** Most cases of retinoblastoma are bilateral and non-hereditary → **CORRECT:** Most HEREDITARY cases are bilateral; most OVERALL cases are sporadic (unilateral)
  - ✗ **WRONG:** Penetrance of hereditary retinoblastoma is 100% → **CORRECT:** Penetrance is ~90%

Other Solid Tumors

- Most common solid tumor in childhood: brain tumor
- Osteosarcoma: sunburst pattern on X-ray, pathological fractures
- Ewing sarcoma: small round blue cell tumor
- Rhabdomyosarcoma: most common pediatric soft tissue sarcoma
- Hepatoblastoma: elevated AFP
- Craniopharyngioma: no role for chemotherapy as primary treatment
  - **(past question)** 3-year-old with ataxia and abdominal mass → neuroblastoma

1.14 Tumor Lysis Syndrome (TLS)

- Electrolyte abnormalities: hyperkalemia, hyperphosphatemia, hyperuricemia, hypocalcemia
  - ✗ **WRONG:** TLS causes hypoglycemia → **CORRECT:** TLS does NOT cause hypoglycemia (causes hyper-K, hyper-PO4, hyper-uric acid, hypo-Ca)
  - ✗ **WRONG:** TLS causes hyponatremia → **CORRECT:** Hyponatremia is NOT a feature of TLS

1.15 Coagulation Quick Reference

Condition	PT	PTT	Bleeding Time	Platelets
Hemophilia A/B	Normal	Prolonged	Normal	Normal
ITP	Normal	Normal	Prolonged	Low
vWD	Normal	Prolonged (often)	Prolonged	Normal
DIC	Prolonged	Prolonged	Prolonged	Low
Liver disease	Prolonged	Prolonged	Normal/Prolonged	Normal/Low
Vit K deficiency	Prolonged	Prolonged	Normal	Normal

⚡ HIGH-YIELD — HEMATOLOGY & ONCOLOGY

- MCC malignancy in children: ALL (B-cell, ~85%)
- First test for suspected leukemia: blood film (smear)
- MCC isolated thrombocytopenia in healthy child: ITP — no splenomegaly, normal Hb
- Least effective ITP treatment: platelet transfusion
- HSP: normal platelets + normal coagulation + normal C3; rash is required feature
- HUS: Coombs NEGATIVE — mechanical hemolysis with schistocytes
- IDA: high TIBC (not low); reticulocytosis is first response to iron therapy
- G6PD: Heinz bodies; Coombs negative; penicillin is SAFE
- Thalassemia minor: normal RDW distinguishes from IDA
- Hemophilia A: prolonged PTT only; bleeding time NORMAL
- vWD: most common inherited bleeding disorder; autosomal dominant
- TLS: hyper-K, hyper-PO4, hyper-uricemia, hypo-Ca — NOT hypoglycemia
- Wilms tumor: aniridia, no calcifications, hematuria, HTN
- Neuroblastoma: opsoclonus-myoclonus, raccoon eyes, catecholamines
- Retinoblastoma: white reflex (leukocoria), penetrance ~90%

2. NEUROLOGY

2.1 Headache

Migraine

- MCC of recurrent headaches in children
- Positive family history in 60–77%
- Common migraine (without aura): more common, bilateral in children, throbbing, photophobia, phonophobia, nausea/vomiting, relieved by sleep
- Classical migraine (with aura): preceded by aura (visual, sensory)
  - ✗ **WRONG:** Migraine in children is always unilateral → **CORRECT:** In children, bilateral is more common than unilateral
  - ✗ **WRONG:** Presence of aura is characteristic of common migraine → **CORRECT:** Aura is in CLASSICAL migraine; common migraine = without aura

    - ✗ **WRONG:** Symptom-free intervals are rare in migraine → **CORRECT:** Symptom-free intervals are COMMON
    - ✗ **WRONG:** EEG is a necessary investigation in migraine → **CORRECT:** EEG is NOT routinely needed for migraine
- Prevalence: 3–14% in school children
- Migraine prophylaxis: propranolol, amitriptyline, cyproheptadine, topiramate, valproate
    - ✗ **WRONG:** Sumatriptan is used for migraine prophylaxis → **CORRECT:** Sumatriptan is SYMPTOMATIC treatment only, NOT prophylaxis

Tension Headache

- MCC headache in children
- Band-like pressing tightness, bilateral, anywhere in cranium or suboccipital region
- Mild to moderate intensity; worsens at end of day; increases with age
- NOT associated with photophobia (usually)
    - ✗ **WRONG:** Tension headache is throbbing in nature → **CORRECT:** Tension headache is pressing/tightening, NOT throbbing
    - ✗ **WRONG:** Tension headache is associated with photophobia → **CORRECT:** Photophobia is a migraine feature, NOT typical of tension headache
    - ✗ **WRONG:** Tension headache is more in the morning → **CORRECT:** It worsens at end of day
    - ✗ **WRONG:** Tension headache can be induced by mild exercise → **CORRECT:** Exercise is not a trigger; stress/fatigue is
- Not aggravated by routine physical activity

Increased ICP / Red Flags

- Red flags: increasing severity/frequency, occipital location, waking from sleep, exclusively morning + vomiting, progressive
    - ✗ **WRONG:** Duration of 1–2 years is a red flag for headache → **CORRECT:** Duration alone (1–2 years) without progression is NOT a red flag
- Classic sign of increased ICP: early morning headache (most common presenting sign)
- Cushing reflex: bradycardia + hypertension + abnormal breathing in ICP
- Papilledema + normal CT + worsening headache → Lumbar puncture with opening pressure (pseudotumor cerebri)

2.2 Muscle Diseases

Spinal Muscular Atrophy (SMA)

- Autosomal recessive, SMN1 gene deletion , High frequency of gene carriers in the Middle East
- Best diagnostic test: DNA testing (SMN1 gene)
- Least useful investigation: CPK (normal or mildly elevated)
    - ✗ **WRONG:** CPK is elevated in SMA → **CORRECT:** CPK is NOT significantly elevated in SMA (unlike muscular dystrophies)

- **SMA Type 1 (Werdnig-Hoffmann):**
    - Presentation: hypotonia, absent deep tendon reflexes, tongue fasciculations, pectus excavatum, paradoxical breathing, cannot support head
    - Cranial nerve involvement: NOT in first/second year (affects extremities first)
    - Progressive respiratory depression → respiratory failure
    - Worst prognosis of all SMAs

    - ✗ **WRONG:** Hyperreflexia is a feature of SMA type 1 → **CORRECT:** SMA has HYPOREFLEXIA/areflexia
    - ✗ **WRONG:** Cranial nerve palsy appears in first or second year in SMA type 1 → **CORRECT:** Cranial nerve palsy appears LATER
- Tongue fasciculations: present in SMA type 0/1 (NOT in Duchenne)
    - ✗ **WRONG:** Tongue fasciculations are found in Duchenne muscular dystrophy → **CORRECT:** Tongue fasciculations are in SMA/anterior horn disorders, NOT Duchenne

Duchenne Muscular Dystrophy (DMD)

- X-linked recessive (NOT chromosomal, NOT autosomal)
    - ✗ **WRONG:** Duchenne is autosomal → **CORRECT:** Duchenne is X-linked recessive
    - ✗ **WRONG:** Duchenne is chromosomal inheritance → **CORRECT:** Duchenne is single-gene X-linked recessive
- High CPK (markedly elevated)
- Pseudohypertrophy of calves
- Positive Gowers sign (uses arms to stand up from floor)
- Cognitive delay may be present

Becker Muscular Dystrophy

- X-linked recessive, same gene as Duchenne but partial function — BEST prognosis of dystrophies

Myotonic Dystrophy

- Autosomal dominant — only AD muscle disease in the list
- Triplet repeat expansion disorder (CTG repeats)
- Distal weakness, myotonia, cataracts, cardiac arrhythmias

Guillain-Barré Syndrome (GBS)

- Ascending flaccid paralysis, areflexia
- Elevated CSF protein (with normal cell count = cytoalbuminous dissociation)
- Autonomic instability
- Treatment: IVIG (NOT steroids)
    - ✗ **WRONG:** Steroids are the treatment for GBS → **CORRECT:** IVIG is the treatment for GBS, not steroids
- Favors GBS over polio: symmetric paralysis, ascending pattern, elevated CSF protein
    - ✗ **WRONG:** Sharp sensory level favors GBS → **CORRECT:** Sensory level suggests SPINAL CORD lesion, NOT GBS
    - ✗ **WRONG:** Sensory deficit favors polio over GBS → **CORRECT:** Sensory deficit is more consistent with GBS (polio is purely motor, CSF shows cells)

Myasthenia Gravis

- Fatigability is the hallmark
- Acetylcholine receptor antibodies: positive in autoimmune MG; NOT in congenital myasthenic syndromes
    - ✗ **WRONG:** AChR antibodies are positive in congenital myasthenic syndromes → **CORRECT:** AChR antibodies are positive in autoimmune MG only
- Treatment: acetylcholine esterase INHIBITORS (pyridostigmine), steroids, IVIG, plasma exchange, thymectomy

    - ✗ **WRONG:** Acetylcholine esterase AGONISTS are treatment → **CORRECT:** The treatment is INHIBITORS (pyridostigmine inhibits esterase → more ACh)
- Drug used: pyridostigmine
- EMG: decremental response on repetitive stimulation

Botulism

- DESCENDING paralysis (from head down)
    - ✗ **WRONG:** Botulism causes ascending paralysis → **CORRECT:** Botulism causes DESCENDING paralysis

Peripheral Neuropathies

- Best prognosis: vitamin B12 deficiency (correctable)
- Anterior horn cell disorders: SMA types, Werdnig-Hoffmann, Poliomyelitis, Arthrogryposis, Transverse myelitis (Note : in the past questions transverse myelitis is listed as anterior horn cell disorder, but actually its spinal cord disorder (demyelinating/inflammatory) , if its in the choices check all choices to be sure whats the answer is)
    - ✗ **WRONG:** Wilson disease is an anterior horn cell disorder → **CORRECT:** Wilson disease is a copper metabolism disorder, NOT anterior horn cell
- Lower motor neuron disorders: anterior horn cell, peripheral neuropathy, NMJ, myopathy
    - ✗ **WRONG:** Spinal cord disorders are LMN disorders → **CORRECT:** Spinal cord (transverse myelitis) is UPPER (or mixed) motor neuron, NOT LMN

Transverse Myelitis

- Sensory level (key feature)
- Motor paralysis below lesion
    - ✗ **WRONG:** Facial problems are seen in transverse myelitis → **CORRECT:** Facial involvement is NOT typical

2.3 Cerebral Palsy

- Spastic CP: UMN signs — hyperreflexia, spasticity, clonus
- SMA is NOT a cause of spasticity/clonus (LMN)
- Case of birth asphyxia (abruption → floppy at birth) → spastic lower limbs at 18 months → CP; motor development will mostly improve with time

2.4 Epilepsy / Seizure Syndromes

- Landau-Kleffner syndrome: acquired aphasia + epileptic EEG activity, previously healthy → stuttering then aphasia
- West syndrome: infantile spasms + hypsarrhythmia + developmental regression (first year of life)
- Lennox-Gastaut: multiple seizure types + intellectual disability + abnormal EEG
- Tuberous sclerosis (ash-leaf spots) + West syndrome → tuberous sclerosis

- Epilepsy + meningitis: avoid meropenem (lowers seizure threshold)
- Seizure + metabolic disorder: if improved with NS → likely IEM
- Non-ketotic hyperglycinemia → seizures as major metabolic sign

2.5 Red Reflex / Eye Findings in Newborn

- Absent red reflex (bilateral): congenital cataract, retinoblastoma, vitreous hemorrhage, retinal detachment
  - ✗ **WRONG:** Conjunctivitis causes absent red reflex → **CORRECT:** Conjunctivitis does NOT cause absent red reflex
  - ✗ **WRONG:** Infantile glaucoma is the most common cause of absent red reflex in premature babies → **CORRECT:** In premature infants, absent red reflex is more commonly congenital cataracts or ROP
- Abnormal red reflex: rubella, retinoblastoma, ROP (congenital cataracts cause absent reflex)

2.6 Increased ICP / Pseudotumor Cerebri

- 1-year-old with irritability, abducent nerve palsy, bilateral papilledema, normal CT → pseudotumor cerebri (benign intracranial hypertension)
- Risk factors: steroid withdrawal, vitamin A toxicity, URTI
  - ✗ **WRONG:** Pseudotumor cerebri does not need follow-up → **CORRECT:** It requires follow-up (can cause vision loss)
- Treatment: acetazolamide, prednisolone, LP
- Acetazolamide is a carbonic anhydrase inhibitor

2.7 Movement Disorders

- Dystonia: sustained twisting, prolonged muscle contraction
- Myotonia: slow relaxation of muscle after contraction
- Myokymia: continuous muscle fiber activity

2.8 PRES (Posterior Reversible Leukoencephalopathy Syndrome)

- Features: seizures, headache, visual disturbances, altered consciousness
  - ✗ **WRONG:** Pinpoint pupils are a feature of PRES → **CORRECT:** Pinpoint pupils are NOT a feature of PRES

⚡ HIGH-YIELD — NEUROLOGY

- MCC recurrent headache in children: migraine; MCC headache overall: tension headache
- Sumatriptan is symptomatic only, NOT prophylactic
- Best diagnostic test for SMA: DNA testing; CPK is not helpful
- GBS: IVIG (not steroids); ascending; elevated CSF protein; no sensory level
- DMD: X-linked recessive; NOT chromosomal; markedly elevated CPK
- Myotonic dystrophy: only AD muscle dystrophy
- Botulism: DESCENDING paralysis
- Absent red reflex: conjunctivitis does NOT cause it
- Landau-Kleffner: acquired aphasia + epileptiform EEG
- Tongue fasciculations: SMA type 1, NOT Duchenne

3. NEPHROLOGY

3.1 Nephrotic Syndrome

- Triad: massive proteinuria (>3.5g/day), hypoalbuminemia, edema
- Labs also show: hyperlipidemia, hyponatremia (dilutional)
- Minimal change disease (MCD): most common in children; responds to steroids; normal complement
  - ✗ **WRONG:** MCD is associated with low complement → **CORRECT:** Complement is NORMAL in MCD
- Microscopic hematuria: can occur in MCD (mild)

- Complications: thromboembolism (loss of antithrombin III), SBP (spontaneous bacterial peritonitis), infection, hyponatremia, hypocalcemia
  - ✗ **WRONG:** Sagittal sinus thrombosis is NOT a complication of nephrotic syndrome → **CORRECT:** It IS a complication (hypercoagulable state)
  - ✗ **WRONG:** Hypocalcemic seizure is NOT a complication of NS → **CORRECT:** Hypocalcemia is listed as a complication in some sources; answer varies
  - ✗ **WRONG:** Polyuria is seen in nephrotic syndrome → **CORRECT:** Polyuria is NOT typical; oliguria is more common
- Most serious complication: peritonitis

- NOT used in acute treatment of MCD: ACE inhibitors
- Treatment: prednisolone + furosemide + albumin (when severe)
- IV albumin indication: albumin ≤1.6–2 mg/dL and severe edema resistant to diuretics (or scrotal edema)
- Periorbital swelling only: start diuretic first; do NOT give albumin first
- Outcome of MCD at 16 years: full renal recovery (excellent long-term prognosis)
  - ✗ **WRONG:** MCD leads to chronic renal failure → **CORRECT:** MCD has excellent prognosis with steroid response
  - **(past question)** 3-year-old with NS, NOT at risk for: chronic renal failure

- Congenital nephrotic syndrome treatment: does NOT use steroids (steroid-resistant)
- NS on prednisone — stopped abruptly → adrenal insufficiency → check cortisol, electrolytes, glucose (NOT ACTH level)

3.2 Glomerulonephritis (GN)

PSGN (Poststreptococcal GN)

- Follows throat infection (2–4 weeks) or skin infection (3–6 weeks) with GABHS
- Hematuria (cola/brown urine), HTN, edema, oliguria
- Low C3 (normalizes in 6–8 weeks), C4 normal (alternative pathway activation)
- Treatment: fluid restriction, salt restriction, diuretics, antihypertensives — NO steroids
  - ✗ **WRONG:** Steroids are used in PSGN → **CORRECT:** Steroids are NOT used in PSGN treatment
- Proteinuria: present but NOT heavy (unlike nephrotic syndrome)
  - ✗ **WRONG:** Heavy proteinuria is a typical hallmark of PSGN → **CORRECT:** Heavy proteinuria is NOT a feature of PSGN
- C3 persists low → think MPGN (not resolving PSGN)
- Resolving PSGN: normal C3, microscopic hematuria, no proteinuria, normal/improving creatinine

IgA Nephropathy

- Gross hematuria CONCURRENT with or 1–2 days after URTI (very short interval)
- Normal complement (C3 and C4 normal) — key differentiator
- RBC casts on microscopy
- Episodes of gross hematuria last 1–2 weeks
  - ✗ **WRONG:** Low C3 indicates IgA nephropathy → **CORRECT:** IgA nephropathy has NORMAL complement

MPGN (Membranoproliferative GN)

- Persistently LOW C3 and C4 for >8 weeks — persistent hypocomplementemia differentiates from PSGN
  - **(past question)** C3 still low after PSGN resolves → MPGN

Lupus Nephritis / SLE Nephritis

- Low C3 AND C4 (classic pathway activation)
- Positive ANA, anti-dsDNA

Alport Syndrome

- Hereditary nephritis: X-linked dominant (COL4A5 mutation)
- Persistent microscopic hematuria + sensorineural hearing loss + ocular abnormalities
- Family history of CKD and dialysis
- Painless hematuria
  - **(past question)** 2-year-old with persistent hematuria, father on dialysis → Alport syndrome

GN Differentials — Key Complement Chart

Condition	C3	C4	Key Feature
PSGN	LOW (normalizes)	Normal	Follows strep infection 2–4 weeks, RBC casts
IgA nephropathy	Normal	Normal	Hematuria with/days after URTI
MPGN	Persistently LOW	LOW or normal	Persistent hypocomplementemia >8 wks
SLE nephritis	LOW	LOW	ANA+, anti-dsDNA+
HSP nephritis	Normal	Normal	IgA deposits, purpura
HUS	Normal	Normal	Schistocytes, thrombocytopenia, AKI

3.3 Hematuria

- RBC casts → glomerular origin (most important clue for GN)
- Extra-glomerular hematuria: normal RBC shape, no casts, clots present, bright red urine, no significant proteinuria
  - ✗ **WRONG:** Protein +3 suggests extraglomerular hematuria → **CORRECT:** Heavy proteinuria (3+) suggests GLOMERULAR, not extraglomerular source
- Painful gross hematuria (no RBC casts) → complement testing NOT needed

- Isolated proteinuria: first step is to repeat first morning void urinalysis (orthostatic proteinuria)
- GFR reaches adult levels (120 mL/min/1.73m²) by age 2 years

3.4 Acute Kidney Injury (AKI)

Prerenal AKI

- Low urine Na (<20 mEq/L), low FeNa (<1%), high urine osmolality (>500), high BUN/Cr ratio
- Causes: dehydration, hemorrhage, burns
- Treatment: IV fluid resuscitation
- BUN/Cr rapidly corrects with hydration

Acute Tubular Necrosis (ATN)

- High urine Na (>40), high FeNa (>2–4%), low urine osmolality, granular casts
  - ✗ **WRONG:** High urine osmolality (>500) is seen in ATN → **CORRECT:** High urine osmolarity is prerenal; ATN has LOW osmolarity
  - ✗ **WRONG:** Urine Na <20 is found in ATN → **CORRECT:** ATN has HIGH urine Na (>40); low Na is prerenal
- Causes: asphyxia (Apgar 2 at 1 min), nephrotoxic drugs (gentamicin)
- Preterm with Apgar 2/1min, high urine Na, normal C-reactive protein, normal U/S → ATN from asphyxia

Tubulointerstitial Nephritis (TIN)

- Drug-induced (NSAIDs, beta-lactams like cefaclor, penicillins)
- Urine: eosinophils, pyuria, RBCs, NO proteinuria
- Normal complement, normal kidney U/S
  - **(past question)** Child given cefaclor for pharyngitis → renal impairment + eosinophils in urine → TIN

AKI — Electrolyte Changes

- AKI findings: metabolic acidosis, hyperkalemia, hyperphosphatemia, hypocalcemia, hyponatremia (dilutional)
  - ✗ **WRONG:** Hypernatremia is found in acute renal failure → **CORRECT:** AKI causes HYPOnatremia (dilutional), NOT hypernatremia
  - ✗ **WRONG:** Anemia is expected in AKI (acute) → **CORRECT:** Anemia is a feature of CHRONIC kidney disease, not acute

3.5 Chronic Kidney Disease (CKD)

- Features: anemia (normochromic normocytic → EPO deficiency), FTT, hyperkalemia, metabolic acidosis, hyperphosphatemia, hyperparathyroidism, renal osteodystrophy
- CKD medications: EPO (anemia), GH (short stature), NaHCO3 (acidosis), 1-alpha hydroxyvitamin D (hyperparathyroidism), calcium carbonate WITH meals (as phosphate binder)
  - ✗ **WRONG:** 25-OH Vitamin D is given in CKD → **CORRECT:** 1-alpha hydroxyvitamin D is given (kidney cannot perform 1-alpha hydroxylation)
  - ✗ **WRONG:** Calcium carbonate with meals treats hypocalcemia in CKD → **CORRECT:** Calcium carbonate is given WITH MEALS as a PHOSPHATE BINDER, not just for hypocalcemia
- NOT seen in CKD: alopecia
- Highly suggestive of CKD: hyperkalemia (in conjunction with other features)

- CKD = small echogenic kidneys (chronic) vs large kidneys in acute
  - ✗ **WRONG:** Large echogenic kidneys suggest chronic cause of renal impairment → **CORRECT:** Large kidneys suggest ACUTE process; small kidneys = chronic
- NOT a cause of CKD: neurogenic bladder (causes UTI/obstruction but not direct CKD cause listed — answer may vary)

- CKD bone disease: low 1,25-OH Vit D → high PTH → osteitis fibrosa cystica → aplastic bone disease (with over-suppression of PTH)
  - ✗ **WRONG:** High phosphate levels increase IGF-23 that stimulates phosphate excretion → **CORRECT:** High phosphate INCREASES FGF-23, which DECREASES phosphate reabsorption (not IGF-23)
  - ✗ **WRONG:** Growth hormone levels are low in CKD → **CORRECT:** GH levels are often normal or HIGH, but there is GH RESISTANCE in CKD

- Target Hb in CKD: 11–13 g/dL
- Ca x PO4 product should be <55–65 to prevent calcification

3.6 Fluid & Electrolyte Disorders

Hyperkalemia Treatment

- Treatments: calcium gluconate (membrane stabilization), insulin + glucose (IC shift), salbutamol/Ventolin (IC shift), sodium bicarbonate, kayexalate, dialysis
  - ✗ **WRONG:** Calcium is NOT used to treat hyperkalemia → **CORRECT:** Calcium gluconate IS used (stabilizes cardiac membrane)
  - ✗ **WRONG:** Oral binding resins are used for ACUTE hyperkalemia in renal failure → **CORRECT:** Oral resins are for maintenance, NOT acute treatment
- Salbutamol + insulin/glucose: increase K shift INTO cells (intracellular)

Causes of Hypokalemia

- GI losses: diarrhea, vomiting, nasogastric suction
- Renal losses: hyperaldosteronism, RTA type I and II, Bartter syndrome, Gitelman syndrome, diuretics (furosemide)
  - ✗ **WRONG:** Rhabdomyolysis causes hypokalemia → **CORRECT:** Rhabdomyolysis causes HYPERkalemia (cell lysis releases K)
  - ✗ **WRONG:** 21-hydroxylase deficiency causes hypokalemia → **CORRECT:** CAH (21-OH deficiency) causes HYPERkalemia (aldosterone deficiency type)
  - ✗ **WRONG:** Congenital adrenal hyperplasia causes hypokalemia → **CORRECT:** CAH (salt-wasting) causes HYPERkalemia

Bartter Syndrome

- Loop of Henle transport defect → similar to furosemide effect
- Hypokalemia, metabolic alkalosis, low/normal BP, normal/high urine chloride
- Nephrocalcinosis is associated
- Low urinary chloride suggests extra-renal loss (vomiting, CF); HIGH urine chloride in Bartter
  - ✗ **WRONG:** Low urinary chloride is seen in Bartter syndrome → **CORRECT:** Bartter has HIGH urine chloride (renal chloride wasting)
- 2-month-old: hypokalemia, low serum Cl, metabolic alkalosis pH 7.55, PCO2 50, HCO3 32, high urine Cl → Bartter syndrome

Gitelman Syndrome

- Distal tubule transport defect (similar to thiazides)
- Hypokalemia, metabolic alkalosis, hypomagnesemia, LOW blood pressure, normal/high urine Cl
- 13-year-old male, muscle weakness, constipation, hypokalemia, normal BP, elevated urinary K → Gitelman
- 18-month-old: K 2.3, Na 123, HCO3 30, Cl 76 (low), urine Cl HIGH, Mg normal → Bartter

Renal Tubular Acidosis (RTA)

Type	Defect	Urine pH	Kalemia	Nephrocalcinosis	Treatment
Type 1 (Distal)	Cannot acidify urine	>5.5	Hypokalemia	YES	Bicarbonate + K
Type 2 (Proximal)	Cannot reabsorb HCO3	<5.5 (initially)	Hypokalemia	Rare	Large bicarb doses
Type 4	Aldosterone deficiency/resistance	≤5.5	Hyperkalemia	No	Fludrocortisone

- Distal RTA: hyperchloremic NAGMA, urine pH >5.5, hypokalemia, hypercalciuria, nephrocalcinosis, positive urine AG
  - ✗ **WRONG:** Hypophosphatemia is a finding in distal RTA → **CORRECT:** Hypophosphatemia is in FANCONI syndrome/proximal RTA; distal RTA has normal phosphate
- NAGMA + glucosuria + aminoaciduria + phosphaturia + normal glucose → FANCONI syndrome (proximal tubule generalized dysfunction)
- Fanconi syndrome: glucosuria with normal blood glucose, metabolic acidosis, hypophosphatemia, rickets; urine pH can be <5.5
- MCC of Fanconi syndrome in childhood: cystinosis
- FTT + hypokalemia + acidosis → Fanconi syndrome
- High anion gap metabolic acidosis (HAGMA) causes :, organic acidemia, DKA, renal failure, lactic acidosis, toxins
  - ✗ **WRONG:** Urea cycle defects causes HAGMA → **CORRECT:** Urea cycle defects causes high ammonia (alkalosis tendency sometimes)

Metabolic Alkalosis

- Chloride-responsive (low urine Cl): pyloric stenosis, vomiting, diuretics (after stopping), cystic fibrosis
- Chloride-resistant (high urine Cl): Bartter, Gitelman, hyperaldosteronism, Liddle syndrome
  - ✗ **WRONG:** Bartter is a cause of chloride-responsive metabolic alkalosis → **CORRECT:** Bartter is chloride-RESISTANT (high urine Cl)

Hyperphosphatemia

- Causes: renal failure, hypoparathyroidism, tumor lysis syndrome, excess phosphate intake
  - ✗ **WRONG:** Hyperparathyroidism causes hyperphosphatemia → **CORRECT:** Hyperparathyroidism causes HYPOphosphatemia (PTH promotes phosphate excretion)
- Associated with nephrocalcinosis: distal RTA, hyperoxaluria, hypercalciuria

Hypernatremia Causes

- Diabetes insipidus, salt poisoning, gastroenteritis (free water loss)
  - ✗ **WRONG:** Psychogenic polydipsia causes hypernatremia → **CORRECT:** Psychogenic polydipsia causes HYPOnatremia

Enuresis

- Primary nocturnal enuresis: fluid restriction is NOT used
- 7-year-old with daytime incontinence → MCC: overactive bladder (not enuresis, which is nocturnal)

Urine Anion Gap

- UAG = urine Na + urine K − urine Cl
- Positive UAG: impaired NH4 excretion (distal RTA, CKD)
- Negative UAG: extra-renal losses (diarrhea, CF), proximal RTA

⚡ HIGH-YIELD — NEPHROLOGY

- PSGN: low C3, normal C4, short interval after strep; no steroids; NOT heavy proteinuria
- IgA nephropathy: normal complement; hematuria concurrent/days after URTI
- MPGN: persistently low C3 (>8 weeks)
- RBC casts = glomerular disease
- ATN: high urine Na, low urine osmolality; Prerenal: low urine Na, high osmolality
- MCD: steroid-responsive, excellent prognosis, normal complement
- Bartter: high urine Cl, metabolic alkalosis, hypokalemia, normal BP
- Distal RTA: urine pH >5.5, hypokalemia, nephrocalcinosis
- Fanconi: glucosuria + aminoaciduria + phosphaturia + NAGMA
- Hyperkalemia treatment: calcium gluconate → insulin/glucose → salbutamol → dialysis

4. CARDIOLOGY

4.1 Resuscitation

- Neonatal resuscitation: PPV is first step when HR <60 bpm
- Chest compressions start when HR <60 after 30 seconds of adequate PPV
- Neonatal CPR ratio: 3:1 (3 compressions : 1 breath)
- Pulse check site in neonate: umbilical artery (NOT carotid or radial)
- IV route preferred for medications in neonatal resuscitation
  - ✗ **WRONG:** Umbilical artery is not the site for pulse in neonatal resuscitation → **CORRECT:** Umbilical artery IS the preferred site for neonatal pulse
- DO NOT use ambu bag in congenital diaphragmatic hernia (causes bowel herniation into chest to expand more)
- Cardiac arrest with asystole: resume chest compressions immediately (no shock for asystole/PEA)
- V-fib and pulseless V-tach: DEFIBRILLATE
- PEA (pulseless electrical activity): NO shock — CPR + treat cause
- Stop CPR to check rhythm (not to check pulse every 30 sec)
  - ✗ **WRONG:** Stop chest compressions every 30 seconds to check pulse → **CORRECT:** Do NOT stop every 30 sec — check rhythm as needed, resume immediately

4.2 Arrhythmias

- SVT: most common tachyarrhythmia in children
  - Narrow complex QRS, rate 250–300 bpm, no visible P waves
  - Infant: irritability, pallor, tachypnea, poor feeding, gallop rhythm
  - Treatment: if hemodynamically stable → IV adenosine; if unstable → synchronized cardioversion
  - NOT a cause of sudden cardiac death
- Long QT syndrome: associated with sudden cardiac death, syncope
  - **(past question)** 3-year-old, consanguineous parents, hearing loss, syncope, FHx sudden deaths → Long QT syndrome (Jervell-Lange Nielsen syndrome)
- HOCM: cause of sudden cardiac death
- Pulmonary stenosis: NOT associated with sudden cardiac death
  - ✗ **WRONG:** Pulmonic stenosis is associated with sudden cardiac death → **CORRECT:** Pulmonic stenosis is NOT associated with sudden cardiac death

4.3 Shock

- MCC of pediatric shock: hypovolemic shock
- MCC of shock requiring ICU in children: septic shock
- Decreased CO in hemorrhagic shock: decreased preload
- Shock can be diagnosed before hypotension develops; low BP is a LATE sign
  - ✗ **WRONG:** Decrease in BP is required to diagnose shock → **CORRECT:** Low BP is a LATE sign; shock diagnosed before BP drops
- Compensated shock signs: tachycardia, prolonged CRT, cool extremities, normal BP
- Septic shock treatment: antibiotics, fluid, vasopressors, nutritional support, mechanical ventilation
  - ✗ **WRONG:** Diuretics help in septic shock to increase urine output → **CORRECT:** Diuretics are NOT given in septic shock

4.4 Cardiomyopathy

- Dilated cardiomyopathy: systolic dysfunction (correct)
- Most important prognostic factor: improvement in ejection fraction (EF)
- Indicators of improvement: increased UOP, decreased CRT, decreased HR, improved BP, decreasing lactate
  - ✗ **WRONG:** Decrease of HCO3 from 24 to 18 indicates improvement in dilated cardiomyopathy → **CORRECT:** Decreasing HCO3 means worsening acidosis, NOT improvement
  - ✗ **WRONG:** UOP from 0.5 to 1.2 mL/kg/hr is NOT an indicator of improvement → **CORRECT:** Increased UOP IS an indicator of improvement

4.5 Pericarditis / Myocarditis

- Pericarditis: sharp midsternal pain, friction rub, low-grade fever → first step: ECG + CXR
- Myocarditis: after viral illness, gallop, cardiomegaly on CXR, tachycardia, respiratory distress

4.6 Rheumatic Fever

- Caused by Group A beta-hemolytic streptococcus (GABHS) pharyngitis
- Evidence of GABHS required for diagnosis: positive throat culture or elevated ASO titer
- Jones Criteria — Major: carditis, polyarthritis, chorea, erythema marginatum, subcutaneous nodules

    - ✗ **WRONG:** Fever is a major criterion of rheumatic fever → **CORRECT:** Fever is a MINOR criterion
    - ✗ **WRONG:** Janeway lesion is a major criterion of RF → **CORRECT:** Janeway lesions are in infective endocarditis, NOT RF
    - ✗ **WRONG:** Cervical lymphadenopathy is a criterion of RF → **CORRECT:** Not a Jones criterion
- Arthritis: migratory, self-limiting, resolves completely with no deformity
- Affected valve: mitral valve most common (MR first, then MS in chronic)
    - ✗ **WRONG:** Aortic valve is the most likely affected in rheumatic carditis → **CORRECT:** Mitral valve is most commonly affected
- Mitral stenosis murmur: apical diastolic with presystolic accentuation
- Prophylaxis: long-acting penicillin
    - No carditis: 5 years or until age 21
    - With carditis (no damage): 10 years or until age 21
    - With persistent valve disease: lifelong
    - ✗ **WRONG:** Prophylaxis for 5 years is correct when there is cardiac involvement → **CORRECT:** With cardiac involvement, prophylaxis should be for 10 years (not 5)

4.7 Murmurs

- Innocent murmur: systolic, grade 1–2/6, disappears with standing/Valsalva, no radiation
- NEVER innocent: diastolic murmur, grade 3+, radiating, thrill
    - ✗ **WRONG:** Diastolic murmur can be innocent → **CORRECT:** Diastolic murmurs are NEVER innocent
- Mitral stenosis: DIASTOLIC murmur at apex with presystolic accentuation
- Systolic murmur that INCREASES with Valsalva: HOCM
- Wide pulse pressure causes: AR, PDA, truncus arteriosus, large AVM
    - ✗ **WRONG:** Mitral stenosis causes wide pulse pressure → **CORRECT:** Mitral stenosis causes NARROW pulse pressure

4.8 Infective Endocarditis (IE)

- Staphylococcus aureus is the overall most common cause of infective endocarditis, while Streptococcus viridans is classically associated with subacute IE and dental procedures.
- Work-up: 3 blood cultures from different sites before starting antibiotics

4.9 Hypertension

- Definition in children/adolescents: average BP ≥95th percentile (not 140/90 as in adults)
- Single BP measurement 140/85 in 12-year-old: needs repeat measurement first
- MCC renovascular HTN in childhood: fibromuscular dysplasia
- HTN crisis: IV nitroprusside (drug of choice)
- HTN with proteinuria: ACE inhibitor first line
- BP cuff: large cuff → falsely LOWER readings
    - ✗ **WRONG:** Large cuff gives falsely higher readings → **CORRECT:** Large cuff gives falsely LOWER BP readings
- Causes of HTN in children: renal artery stenosis, pheochromocytoma, coarctation of aorta, PSGN
    - ✗ **WRONG:** MCD causes HTN → **CORRECT:** MCD does NOT cause HTN (normal BP in nephrotic syndrome usually)

4.10 Anaphylaxis

- First-line treatment: IM epinephrine (intramuscular, NOT IV)
- Also used: antihistamines, inhaled beta-agonists, corticosteroids
  - ✗ **WRONG:** Theophylline is used in anaphylaxis → **CORRECT:** Theophylline is NOT used in anaphylaxis

4.11 Congenital Rubella — Cardiac

- PDA most commonly associated with congenital rubella
- Cardiac defects occur with first trimester infection (not third trimester)
  - ✗ **WRONG:** Subclinical maternal rubella does not lead to congenital infection → **CORRECT:** Even subclinical infection CAN cause congenital rubella

⚡ HIGH-YIELD — CARDIOLOGY

- Neonatal pulse check: umbilical artery
- PEA: no shock; asystole: no shock — resume CPR
- SVT treatment: adenosine (stable) → cardioversion (unstable)
- Shock: low BP is LATE sign; diagnose before hypotension
- Rheumatic fever: mitral valve most affected; with carditis → 10 years prophylaxis
- Innocent murmur: NEVER diastolic
- HTN in children: defined as ≥95th percentile; single reading → repeat first
- Anaphylaxis: IM epinephrine (not IV)
- Large cuff → falsely LOW BP

5. PULMONOLOGY & RESPIRATORY

5.1 Asthma

- Asthma: spirometry is the diagnostic tool (not CXR)
- Asthma does NOT cause weight loss
- Asthma exacerbation: salbutamol 3 times → if improved but PO2 only slightly increased → add systemic steroids
- Preschool wheezes: penicillin allergy is NOT a risk factor for persistence

5.2 Cystic Fibrosis

- AR inheritance; 25% recurrence risk for siblings
- Can cause: metabolic alkalosis with hypokalemia and hyponatremia (sweat chloride loss)
- Associated findings: recurrent pulmonary infections, pancreatic insufficiency, FTT, meconium ileus
- CF with ascites: Hb least affected (platelets, ammonia more affected by liver disease)
- Kartagener syndrome (bronchiectasis + dextrocardia + sinusitis): best diagnostic test = ciliary biopsy

5.3 Respiratory Distress in Newborn

- Term neonate with retractions and dyspnea → start CPAP
- 2-month infant with decreased air entry one side, tachypnea, decreased O2 sat → CPAP

- Do NOT ambu bag in congenital diaphragmatic hernia
- Maternal chorioamnionitis: may PREVENT respiratory distress syndrome (RDS) by stimulating surfactant production
- Best method to improve hypoxia: increase O2 saturation (not Hb from 8 to 10)

5.4 Empyema

- Characteristics: low pleural fluid pH, high LDH, high protein
- MCC: Streptococcus pneumoniae (NOT Haemophilus influenzae type b)
  - ✗ **WRONG:** Haemophilus influenzae type b is MCC of empyema → **CORRECT:** S. pneumoniae is MCC of lung empyema
- Treatment: drainage + antibiotics

5.5 TB

- Most common type of primary TB in children: endobronchial TB with lymph node enlargement
- PPD 17mm induration in 2-year-old → next step: chest X-ray (not repeat test)

5.6 Hypoxemia Causes

- Causes: alveolar hypoventilation, V/Q mismatch, diffusion block, intrapulmonary shunt
  - ✗ **WRONG:** Hyperinflation causes hypoxemia → **CORRECT:** Hyperinflation is NOT a cause of hypoxemia

5.7 Otitis Media

- More common in <3 years, bottle-fed infants, allergic rhinitis
- Erythema of TM alone is NOT sufficient to diagnose acute otitis media
  - ✗ **WRONG:** Erythema of TM is sufficient to diagnose acute otitis media → **CORRECT:** Additional signs (bulging, loss of landmarks) required
  - ✗ **WRONG:** Clindamycin is first-line for otitis media → **CORRECT:** Amoxicillin/amoxicillin-clavulanate is first-line
  - ✗ **WRONG:** Haemophilus influenzae type b is very common cause of otitis media → **CORRECT:** Strep pneumoniae and non-typeable H. influenzae are common; Hib vaccine reduced Hib cases
- MCC otitis media: Strep pneumoniae > non-typeable H. influenzae
  - ✗ **WRONG:** Staph aureus is MCC otitis media → **CORRECT:** Strep pneumoniae is MCC
- Otitis media with effusion: tympanometry is the test

5.8 Allergic Rhinitis

- Features: pale/blue-tinged nasal mucosa, clear discharge, otitis media risk
  - ✗ **WRONG:** Red nasal mucosa is seen in allergic rhinitis → **CORRECT:** Allergic rhinitis shows PALE nasal mucosa, not red

⚡ HIGH-YIELD — RESPIRATORY

- Asthma: spirometry for diagnosis; systemic steroids if not improving
- CF: AR; metabolic alkalosis with hypokalemia

- Empyema MCC: Strep pneumoniae
- TB in children: lymph node enlargement is most common type
- PPD ≥15mm → chest X-ray next
- Erythema of TM alone does NOT diagnose AOM
- Allergic rhinitis: pale mucosa (not red)

6. INFECTIOUS DISEASES

6.1 Neonatal Sepsis

- MCC: Group B Streptococcus (GBS) and E. coli
- Most indicative lab finding: elevated I:T ratio (immature/total neutrophil ratio) ≥0.2 (40% highly abnormal)
- Sepsis workup includes: CBC diff, blood culture, CSF culture, CXR (if respiratory symptoms)
  - ✗ **WRONG:** Urine culture is indicated in 1-day-old with neonatal sepsis → **CORRECT:** Urine culture is NOT indicated in first few days (day 1 neonatal sepsis)
  - ✗ **WRONG:** Stool culture is part of sepsis workup in neonate → **CORRECT:** Stool culture is NOT routine in neonatal sepsis workup
  - ✗ **WRONG:** CXR is always part of initial septic workup → **CORRECT:** CXR only if respiratory symptoms

6.2 Viral Exanthems

Virus	Features	Rash Type	Notes
Measles (Rubeola)	Cough, coryza, conjunctivitis (3Cs) + Koplik spots, high fever	Maculopapular, head→trunk	Vitamin A treatment; no vesicles
Rubella	Mild; cervical LAP; rash trunk first	Maculopapular, fine	No antiviral; supportive only
Roseola (HHV-6)	High fever 3 days → fever breaks → maculopapular rash trunk	Maculopapular	Very young children
Varicella (VZV)	Vesicular crops in different stages; trunk first	Vesicular/pustular	Acyclovir for adults/immunocompromised
Hand Foot Mouth (Coxsackie A)	Vesicles: mouth, palms, soles	Vesicular	No specific treatment
Erythema infectiosum (Parvo B19)	Slapped-cheek; lacy extremity rash	Maculopapular	Aplastic crisis in sickle cell

- Measles is NOT caused by vesicular rash
  - ✗ **WRONG:** Varicella rash is maculopapular → **CORRECT:** Varicella is VESICULAR
  - ✗ **WRONG:** Measles rash is vesicular → **CORRECT:** Measles rash is MACULOPAPULAR

- ✗ **WRONG:** Measles has no long-term complications → **CORRECT:** Measles can cause SSPE (subacute sclerosing panencephalitis), encephalitis
- ✗ **WRONG:** Encephalitis after measles leads to permanent damage in majority → **CORRECT:** Only a MINORITY develop permanent neurologic damage

- MCC: 10-month-old with 4 days fever → roseola most likely HHV-6 (NOT measles — look for rash AFTER fever breaks)

6.3 Mumps

- Orchitis: usually unilateral, more common post-pubertally; does NOT commonly lead to infertility
  - ✗ **WRONG:** Orchitis following mumps commonly leads to infertility → **CORRECT:** It is usually unilateral and does NOT commonly cause infertility
- Pancreatitis: rare; more common in children (not adults)
  - ✗ **WRONG:** Pancreatitis is more common in adults than children with mumps → **CORRECT:** Pancreatitis is rare and occurs MORE in children
- Aseptic meningitis: common complication
- Nerve deafness: rare complication
- No specific antiviral treatment; acyclovir is NOT effective for mumps
  - ✗ **WRONG:** Acyclovir is the drug of choice for mumps meningitis → **CORRECT:** Acyclovir is for herpesviruses, NOT mumps
- Mumps vaccine (MMR): contraindicated in children receiving >2 mg/kg/day or >20 mg/day prednisolone (immunosuppressive dose)
- Live vaccines are contraindicated in immunosuppressed patients

6.4 Varicella (Chickenpox)

- Virus characteristics: latency (reactivates as herpes zoster)
- Infectious until all lesions crusted
- Incubation period: 10–21 days
- Rash appears first on trunk
- Complications more common in: adults, immunocompromised, neonates
  - ✗ **WRONG:** Complications of varicella are greatest in children 1–12 years → **CORRECT:** Complications are greatest in adults and immunocompromised
  - ✗ **WRONG:** Chickenpox is milder in adults → **CORRECT:** Chickenpox is WORSE in adults
  - ✗ **WRONG:** Transplacental varicella usually leads to hydrocephalus → **CORRECT:** Congenital varicella causes limb hypoplasia, NOT hydrocephalus typically
- Acyclovir: indicated for adults, immunocompromised, neonates, and severe cases
- Varicella vaccine: useful in post-exposure prophylaxis (within 72–120 hours)
- SSPE is NOT a complication of varicella (it's a measles complication)

6.5 Congenital Infections (TORCH)

Infection	Key Features
CMV	MCC congenital infection overall; hepatitis, microcephaly, periventricular calcifications, sensorineural deafness; blueberry muffin rash
Rubella	Cataracts, PDA, sensorineural deafness, blueberry muffin rash; intracranial calcifications NOT common; DM later

Infection	Key Features
Toxoplasma	Hydrocephalus, chorioretinitis, cerebral calcifications (basal ganglia), IUGR; NO cataracts
Syphilis	Saddle nose, snuffles, periostitis, blueberry muffin, skin rash
HSV	Vesicular lesions; encephalitis; often peripartum
GBS	Early neonatal sepsis (within 7 days)

    - ✗ **WRONG:** Toxoplasma causes periventricular calcifications → **CORRECT:** Toxoplasma causes BASAL GANGLIA calcifications; CMV causes periventricular
    - ✗ **WRONG:** Toxoplasma causes cataracts → **CORRECT:** Cataracts are in RUBELLA, not toxoplasma
- Blueberry muffin rash (extramedullary hematopoiesis): CMV, rubella, congenital syphilis
- Rubella: cataracts + PDA + deafness (classic triad); DM type 1 later in life
- CMV: MCC acquired sensorineural hearing loss + other abnormalities
- Congenital infections transmissible as maternal IgG via placenta: MG, ITP, thyrotoxicosis, Rh iso-immunization
    - ✗ **WRONG:** HIV is transmitted via maternal IgG crossing the placenta → **CORRECT:** HIV is NOT transmitted as IgG — it crosses via blood/virus during delivery/breastfeeding

6.6 Streptococcal Infections

- GABHS (Group A): causes pharyngitis, rheumatic fever, PSGN, impetigo
- Rheumatic fever follows respiratory infection ONLY (not skin)
- PSGN can follow both respiratory AND skin GABHS infection
- Penicillin-resistant GABHS has NOT been described
    - ✗ **WRONG:** Penicillin-resistant GABHS should be treated with vancomycin → **CORRECT:** GABHS has NO penicillin resistance; erythromycin for penicillin allergy
- GBS intrapartum prophylaxis: ampicillin (first choice)
- Sore throat with GABHS: first-line = amoxicillin; if type 1 penicillin allergy → azithromycin

6.7 Other GI Infections

- Salmonella: acquired from poultry/eggs; incubation 6–72 hours (not <6 hours); bacteremia rare in immunocompetent
    - ✗ **WRONG:** Salmonella incubation period is <6 hours → **CORRECT:** Salmonella incubation is 6–72 hours
    - ✗ **WRONG:** Salmonella bacteremia is common → **CORRECT:** Bacteremia is rare in normal older children/adults
- Shigella: person-to-person transmission is COMMON; antibiotic treatment indicated; bacteremia rare; febrile seizures possible
    - ✗ **WRONG:** Shigella person-to-person transmission is uncommon → **CORRECT:** Person-to-person transmission is VERY common in shigella
- Pseudomembranous colitis (C. difficile): treat with metronidazole or vancomycin (oral)

6.8 Immunodeficiency

Deficiency	Organisms	Notes
B-cell / Agammaglobulinemia	Encapsulated bacteria (Pneumo, H. flu, Staph)	IVIG treatment; NOT associated with staph recurrent infections or fungal infections
T-cell (CMI) deficiency	Fungi, viruses, intracellular organisms, PCP	Onset early infancy; predispose to systemic candidiasis
Combined (SCID)	All of the above	Severe, presents early infancy
CGD (Chronic Granulomatous Disease)	Catalase-positive (Staph, Aspergillus)	NBT test abnormal; recurrent abscesses, osteomyelitis
Complement deficiency	Neisseria (Encapsulated)	C1 esterase inhibitor deficiency → hereditary angioedema
Phagocyte defect (LAD)	Staphylococcus, gram-negatives	Delayed cord separation, recurrent infections; flow cytometry diagnosis

- MCC primary immune deficiency: selective IgA deficiency
  - ✗ **WRONG:** SCID is the most common primary immune deficiency → **CORRECT:** Selective IgA deficiency is the MCC
- NBT (nitroblue tetrazolium) test: abnormal in CGD
- T-cell deficiency: predisposes to systemic fungal infections
  - ✗ **WRONG:** B-cell deficiency predisposes to systemic fungal infections → **CORRECT:** T-cell (CMI) deficiency, and neutropenia predispose to fungal infections
  - ✗ **WRONG:** Splenectomy predisposes to systemic fungal infections → **CORRECT:** Splenectomy predisposes to encapsulated bacteria, NOT fungi
  - ✗ **WRONG:** Recurrent staph infections = agammaglobulinemia → **CORRECT:** Recurrent staph infections suggest CGD, not B-cell immunodeficiency
- Recurrent abscesses + delayed umbilical cord separation → LAD; diagnostic test: flow cytometry
  - **(past question)** 15-month-old with recurrent abscesses + delayed cord separation: most useful test → flow cytometry (LAD)
- PCP/Pneumocystis pneumonia: associated with T-cell (cellular) deficiency, NOT B-cell
- PPD skin test: used to test T-cell immunity
- Selective IgA deficiency: autosomal (NOT AR as sometimes thought)
- X-linked agammaglobulinemia (Bruton): treatment = IVIG

6.9 Antibiotic Guide

Drug	Key Facts
Amoxicillin	Streptococcal pharyngitis; drug of choice for most strep infections
Azithromycin	For penicillin type 1 allergy in strep pharyngitis; first-line neonatal pertussis; anti-Pseudomonal? NO

Drug	Key Facts
Ceftriaxone	Long half-life; good CSF penetration; CONTRAINDICATED in neonates (displaces bilirubin); NOT effective vs Pseudomonas
Meropenem	Avoid in epileptics; NOT for MRSA; NOT anti-Pseudomonal in some contexts
Vancomycin	For MRSA; does NOT discolor teeth; increase interval between doses if level high; NOT for intra-abdominal sepsis alone; NOT for Pseudomonas
Clindamycin	Safe with penicillin angioedema; for MRSA (some); can cause C. difficile colitis
Metronidazole	C. difficile colitis treatment
Ceftazidime	Anti-Pseudomonal cephalosporin with minimal toxicity; preferred for Pseudomonas UTI in children
Beta-lactams general	Inhibit cell wall synthesis (PBPs); bactericidal; cleared by kidneys; wide therapeutic index; NOT safe intrathecally

- Drugs that cross BBB: ampicillin, cefotaxime, ceftriaxone, meropenem, vancomycin
  - ✗ **WRONG:** Gentamicin crosses the blood-brain barrier → **CORRECT:** Gentamicin does NOT reliably cross the BBB
- Hepatotoxic: paracetamol, carbamazepine, isoniazid, fluconazole
  - ✗ **WRONG:** Vancomycin is hepatotoxic → **CORRECT:** Vancomycin is nephrotoxic/ototoxic, NOT primarily hepatotoxic
- Nephrotoxic: amikacin, cyclosporine, vancomycin, captopril
  - ✗ **WRONG:** Ceftriaxone is nephrotoxic → **CORRECT:** Ceftriaxone is NOT nephrotoxic
- Anti-Pseudomonal: imipenem, piperacillin-tazobactam, meropenem, cefepime, ceftazidime, ciprofloxacin
  - ✗ **WRONG:** Ceftriaxone is anti-Pseudomonal → **CORRECT:** Ceftriaxone is NOT active against Pseudomonas
- MRSA antibiotics: vancomycin, linezolid, clindamycin, TMP-SMX, daptomycin
  - ✗ **WRONG:** Meropenem is active against MRSA → **CORRECT:** Meropenem is NOT active against MRSA

⚡ HIGH-YIELD — INFECTIOUS DISEASES

- MCC neonatal sepsis: GBS + E. coli
- Neonatal sepsis workup: NO stool Cx, NO urine Cx (day 1), CXR only if respiratory symptoms
- Measles: maculopapular (NOT vesicular); encephalitis in MINORITY
- Roseola (HHV-6): fever → breaks → rash appears
- Varicella: worse in adults; 10–21 day incubation; trunk first
- CMV: periventricular calcifications; Toxoplasma: basal ganglia
- Rubella: cataracts + PDA + deafness; NO cataracts in toxoplasma
- MCC primary immunodeficiency: IgA deficiency
- T-cell deficiency → fungi; B-cell deficiency → encapsulated bacteria
- LAD (delayed cord separation): flow cytometry for diagnosis
- Ceftriaxone: contraindicated in neonates; NOT anti-Pseudomonal
- Gentamicin: does NOT cross BBB

7. GASTROENTEROLOGY

7.1 GERD

- Best diagnostic tool: 24-hour pH study (or pH impedance probe)
- Best way to diagnose GERD in an infant: history and physical examination
- Treatment in infants: thicken formula, burping after feeds, elevated head position, cessation of parental smoking
  - ✗ **WRONG:** H2 blockers are used for GERD in 5-month-old → **CORRECT:** H2 blockers are NOT recommended as routine in infants; behavioral interventions first
  - ✗ **WRONG:** PPI is least appropriate in neonatal GERD → **CORRECT:** Correct — PPI is NOT first-line in neonates
- GERD vs physiologic reflux: FTT, recurrent pneumonia, apnea, excessive irritability → GERD concern

7.2 Pyloric Stenosis

- Projectile non-bilious vomiting, 3–6 weeks of age, male:female 4:1 (NOT equal)
  - ✗ **WRONG:** Pyloric stenosis equally affects boys and girls → **CORRECT:** Male predominance 4:1
- Metabolic alkalosis + hypokalemia + hypochloremia + low urine Cl → pyloric stenosis (OR GERD-like persistent vomiting)
- Diagnostic test: ultrasound (NOT CT)
- Treatment: surgical pyloromyotomy (Ramstedt)
- ECG showing flattened T wave in projectile vomiting patient → hypokalemia

7.3 Hirschsprung Disease

- Caused by absent ganglion cells (aganglionic segment) — typically rectosigmoid
- Presents: failure to pass meconium in first 24–48 hours, constipation in neonates
  - ✗ **WRONG:** Passage of meconium in first 24 hours rules out Hirschsprung disease completely → **CORRECT:** Does NOT completely rule it out (some pass meconium)
- Empty rectum on digital exam
- Boys > girls
- Contrast enema: shows transition zone (contracted distal segment, dilated proximal)
  - ✗ **WRONG:** Enema shows dilated aganglionic area in Hirschsprung → **CORRECT:** Enema shows NARROW/contracted aganglionic area and dilated normal segment — transition zone
- Diagnosis confirmed: rectal biopsy (absence of ganglion cells)
- Down syndrome association
- Treatment: surgery (colostomy if urgent, then pull-through)

7.4 Intestinal Malrotation

- Best diagnostic test: upper GI barium series

7.5 Constipation

- MCC constipation in 3-year-old: functional (NOT Hirschsprung)
- Functional constipation: rarely associated with organic cause; fecal impaction can occur

    - ✗ **WRONG:** Functional constipation is rarely associated with fecal impaction → **CORRECT:** Fecal impaction IS common in functional constipation
- Maintenance treatment: osmotic laxatives, fiber, increased fluids, proper toilet routine
- NOT for long-term: stimulant laxatives (bisacodyl, senna)
    - ✗ **WRONG:** Stimulant laxatives can be safely used long-term for constipation maintenance → **CORRECT:** Stimulant laxatives are for SHORT-TERM use only
- Initial workup for constipation: urine analysis and blood tests; stool tests NOT routine initially

7.6 GI Bleeding

- Lower GI bleeding causes by age:
    - Infants: cow's milk protein allergy, NEC, intussusception, Meckel's
    - Toddlers (2–5 years): juvenile polyps, Meckel's, intussusception
    - School age: juvenile polyps, Meckel's, IBD
    - ✗ **WRONG:** Celiac disease is a common cause of lower GI bleeding in 4-year-old → **CORRECT:** Celiac disease causes malabsorption, NOT lower GI bleeding
    - ✗ **WRONG:** IBD is a common cause of GI bleeding in infants → **CORRECT:** IBD is uncommon in infants
- MCC hematochezia in pediatrics: anal fissure
- Most children with lower GI bleeding do NOT need urgent colonoscopy
- Juvenile polyp: painless bright red blood per rectum in preschool children; rectal mass on exam; diagnosis = colonoscopy
- Meckel's diverticulum: painless LGIB, mostly <2 years; diagnosed by technetium scan
- Esophageal varices: cause of upper GI bleeding (portal HTN)
- Mallory-Weiss tear: upper GI bleed with blood streaks in vomitus
- 2-week-old with 2 episodes of bloody vomiting → maternal blood swallowing (not peptic ulcer)

7.7 H. Pylori

- Causes: gastritis, gastric ulcer, iron deficiency anemia, nodular stomach on endoscopy
    - ✗ **WRONG:** H. pylori causes celiac disease → **CORRECT:** H. pylori does NOT cause celiac disease
- Recurrent abdominal pain: serum H. pylori antibodies NOT used (low sensitivity)

7.8 Wilson Disease

- Autosomal recessive copper metabolism disorder
- Caused by: inadequate biliary excretion of copper
    - ✗ **WRONG:** Wilson disease is caused by poor absorption of copper → **CORRECT:** Caused by INADEQUATE BILIARY EXCRETION of copper
- Two screening tests: low serum ceruloplasmin + elevated 24-hr urinary copper
- Low ceruloplasmin is characteristic
- Kayser-Fleischer rings in cornea

7.9 Hepatitis

- Hepatitis A: fecal-oral; IgM anti-HAV for acute diagnosis; asymptomatic in young children
    - ✗ **WRONG:** Hepatitis A can be transmitted from cat's feces → **CORRECT:** Hepatitis A is human-to-human only (fecal-oral), not from animals

- Indications for admission in Hep A: hypoglycemia, hyperammonemia, high INR, low O2 sat
  - ✗ **WRONG:** High ALT alone is an indication for hospital admission in Hep A → **CORRECT:** High ALT alone is NOT an indication for admission
  - ✗ **WRONG:** Jaundice alone is an indication for admission in Hep A → **CORRECT:** Jaundice alone is NOT an admission indication
- Hepatitis B chronic: increased risk of hepatocellular carcinoma
- HBsAg + HBeAg + IgM anti-HBc → active hepatitis B infection
- Mother with HBsAg + HBeAb → give newborn HBIG + vaccine within 12 hours

7.10 NASH

- ALT usually higher than AST
- Risk factors: obesity, DM, hypertension, dyslipidemia
  - ✗ **WRONG:** Most NASH patients are symptomatic → **CORRECT:** Most NASH patients are asymptomatic
  - ✗ **WRONG:** N-acetylcysteine is used to treat NASH → **CORRECT:** N-acetylcysteine is NOT used for NASH

7.11 Biliary Atresia

- Management: special formula, special vitamins (fat-soluble: A, D, E, K)
- Cannot send home without close follow-up
- Kasai procedure (hepatoportoenterostomy): done if <3 months
- Vitamin E deficiency in biliary atresia: cerebellar ataxia, muscle weakness, areflexia, acanthocytes on blood smear

7.12 Malnutrition

- Marasmus: most common form of protein-energy malnutrition
- Kwashiorkor: edema starting in lower limbs
- Severely malnourished: do NOT aggressively refeed immediately (risk of refeeding syndrome: low PO4, K, Mg, Ca)
  - ✗ **WRONG:** Aggressively nutritional supplement immediately in severe malnutrition → **CORRECT:** Aggressive early feeding causes refeeding syndrome

⚡ HIGH-YIELD — GASTROENTEROLOGY

- GERD in infant: history + PE is best diagnostic approach; thicken formula
- Pyloric stenosis: male 4:1; metabolic alkalosis + hypokalemia; diagnosis by ultrasound
- Hirschsprung: absent ganglion cells; empty rectum; biopsy confirms; associated with Down syndrome
- MCC constipation in children: functional
- Juvenile polyp: painless bright red PR bleeding; diagnosis = colonoscopy
- Meckel's: mostly <2 years; Tc scan
- Wilson: low ceruloplasmin + high 24-hr urine copper; inadequate biliary excretion
- Hep A admission: NOT for jaundice or high ALT alone — for coagulopathy, hyperammonemia, hypoglycemia

8. ENDOCRINOLOGY

8.1 Rickets / Vitamin D

- Vitamin D deficiency rickets: low Ca, low PO4, high PTH, high ALP, low 25-OH VitD, low/normal urine Ca/Cr
  - ✗ **WRONG:** High urine Ca/Cr ratio is seen in Vitamin D deficiency rickets → **CORRECT:** Urine Ca/Cr is LOW in VitD deficiency rickets
  - ✗ **WRONG:** Low PTH is found in Vitamin D deficiency → **CORRECT:** PTH is HIGH (secondary hyperparathyroidism)
- Ca 7, PO4 2, PTH 100, ALP 1000 → Vitamin D deficiency rickets (NOT hypophosphatemic)
- Hypophosphatemic rickets (X-linked dominant): normal/high PTH, normal Ca, LOW PO4, normal/low VitD
- High PTH + normal Ca + low PO4 + normal VitD → Hypophosphatemic rickets
- Hypoparathyroidism: low Ca, HIGH PO4, low PTH
  - ✗ **WRONG:** Low serum phosphate is seen in primary hypoparathyroidism → **CORRECT:** Hypoparathyroidism causes HIGH phosphate
- Pseudohypoparathyroidism: low Ca, HIGH PO4, HIGH PTH (PTH resistance)
- Vitamin D functions: increase Ca and PO4 absorption; stimulates 1-alpha hydroxylase
- PTH functions: increase Ca reabsorption from bone and kidney; DECREASE renal PO4 reabsorption; stimulate 1-alpha hydroxylase
  - ✗ **WRONG:** PTH increases renal tubular absorption of phosphate → **CORRECT:** PTH DECREASES phosphate reabsorption (promotes phosphaturia)

- In CKD: NOT hypercalcemia (CKD causes hypocalcemia due to decreased 1-alpha hydroxylation)
  - ✗ **WRONG:** Hypercalcemia is present in CKD → **CORRECT:** CKD causes HYPOcalcemia, NOT hypercalcemia

8.2 DKA (Diabetic Ketoacidosis)

- Criteria: glucose >180–200, pH <7.3, HCO3 <15, ketonuria/ketonemia
  - ✗ **WRONG:** DKA is defined as pH <7.35 with glucose >180 and ketones → **CORRECT:** DKA requires pH <7.30 (not 7.35) and more stringent criteria

- Severity: mild pH 7.2–7.3; moderate 7.1–7.2; severe <7.1
- Initial step: normal saline bolus 20 mL/kg (not insulin first, not NaHCO3 first)
  - ✗ **WRONG:** First step in DKA is insulin bolus → **CORRECT:** First step is IV normal saline bolus
  - ✗ **WRONG:** Insulin as IV bolus then infusion → **CORRECT:** Insulin should be given as CONTINUOUS INFUSION only — NOT bolus
- NaHCO3: NOT routinely used; only for life-threatening hyperkalemia or pH <6.9
- Potassium: always add to IV fluids (start when urine output is confirmed and K <5.5 (or <5.0))
- When glucose drops to 240–300 mg/dL but still DKA: add dextrose 5% (do not stop insulin)
- Most common cause of mortality in DKA: cerebral edema
- Cerebral edema treatment: decrease fluid rate, elevate HOB, mannitol or hypertonic saline — NOT CT scan first
  - **(past question)** Child in ICU for DKA with seizure, glucose 180, Na 136 → suspect cerebral edema → mannitol, not CT scan first

- Labs needed for DKA management: electrolytes, pH, glucose (NOT lipid profile)
  - ✗ **WRONG:** Serum lipid profile is necessary to guide DKA management → **CORRECT:** Lipid profile is NOT needed in DKA management

8.3 Hypoglycemia

- NOT a cause: PKU, mucopolysaccharidosis, lipid storage disease
- Causes: sepsis, glycogen storage disease, fatty acid oxidation defect, urea cycle defect (??), Reye syndrome, MSUD, insulin excess
  - ✗ **WRONG:** PKU causes hypoglycemia → **CORRECT:** PKU does NOT cause hypoglycemia
- Neonatal hypoglycemia causes: IDM (infant of diabetic mother), LGA, SGA, hyperinsulinism, preterm
  - ✗ **WRONG:** LGA newborns are NOT predisposed to hypoglycemia → **CORRECT:** LGA infants ARE prone to hypoglycemia
  - ✗ **WRONG:** Term (appropriate for gestational age) is a risk factor for neonatal hypoglycemia → **CORRECT:** Term AGA is NOT a risk factor
  - **(past question)** Not a risk for neonatal hypoglycemia: breast milk jaundice, term AGA

- Down syndrome does NOT predispose to hypoglycemia
  - ✗ **WRONG:** Down syndrome newborns may develop significant hypoglycemia → **CORRECT:** Down syndrome is NOT a cause of neonatal hypoglycemia

8.4 Pituitary

- Panhypopituitarism: low ACTH, low cortisol, low IGF-1, low FSH/LH, low TSH/FT4, low GH → also low urine osmolality (DI from ADH deficiency)
  - ✗ **WRONG:** High urine osmolality is seen in panhypopituitarism → **CORRECT:** Panhypopituitarism includes DI → LOW urine osmolality (dilute urine)
  - ✗ **WRONG:** High prolactin is found in panhypopituitarism → **CORRECT:** Prolactin may be LOW or normal (loss of stimulatory TRH), not high

8.5 SIADH

- Features: hyponatremia, elevated urine Na, HIGH urine osmolality, euvolemic, absence of adrenal/renal/thyroid disease
  - ✗ **WRONG:** SIADH has low urine osmolality → **CORRECT:** SIADH has HIGH urine osmolality (concentrated urine despite hyponatremia)
- Causes: meningitis, pneumonia, brain tumors, drugs (carbamazepine, cyclophosphamide)

8.6 Precocious Puberty

- Central (GnRH-dependent): all secondary sexual characteristics develop including gonadal size
- Peripheral (GnRH-independent): adrenal or gonadal source — gonads remain PREPUBERTAL size

- Female, 6 years, breast stage 3, pubic hair stage 1 → Central (breast development first, gonads stimulated) → Central hamartoma
- Male, 6–8 years, large testes (pubertal size) + pubic hair → Central precocious puberty → MRI brain mandatory
- Male, 8 years, pubic hair stage 3, small/prepubertal testes → Peripheral (adrenal source)
  - If <10 years with testes 3cc: non-classical CAH most likely if bilateral, adrenal carcinoma if virilization is severe (males)
  - Male with prepubertal testes + adrenarche → adrenal tumor most likely if severe

- β-hCG producing tumors: cause precocious puberty in MALES only (not females)

- ✗ **WRONG:** β-hCG is investigated for isolated premature pubarche in females → **CORRECT:** β-hCG is NOT indicated in females with premature pubarche
- NOT a cause of central precocious puberty: anorexia nervosa (can cause delayed puberty)
- NOT a cause of peripheral precocious puberty: suprasellar arachnoid cyst (central CNS lesion → central PP)
- Hypothyroidism: can cause precocious puberty (rare mechanism)

⚡ HIGH-YIELD — ENDOCRINOLOGY

- VitD deficiency: low Ca, low PO4, HIGH PTH, HIGH ALP, low urine Ca/Cr
- Hypoparathyroidism: low Ca, HIGH PO4, low PTH
- DKA first step: normal saline bolus (NOT insulin bolus)
- DKA insulin: continuous infusion (NOT bolus)
- Cerebral edema in DKA: mannitol/hypertonic saline; decrease fluids
- SIADH: high urine osmolality (concentrated urine despite low serum Na)
- Panhypopituitarism: low urine osmolality (DI component)
- Central PP: large gonads; peripheral PP: prepubertal gonads
- hCG tumor → precocious puberty in MALES only

9. RHEUMATOLOGY & DERMATOLOGY

9.1 Juvenile Idiopathic Arthritis (JIA/JRA)

Type	Key Features
Oligoarticular (Pauciarticular) type 1	Most common; peak age 1–3 years; FEMALES; ANA+ in 50%; large joints (knee); uveitis risk (asymptomatic)
Oligoarticular type 2	HLA-B27+; males; older; enthesitis
Polyarticular RF-negative	5+ joints; any age; females
Polyarticular RF-positive	RF+; adolescent females; symmetric small joints; poor prognosis
Systemic JIA (Still's)	Males = females; quotidian fever; salmon rash; hepatosplenomegaly; ANA usually negative; uveitis absent

- ✗ **WRONG:** Oligoarticular JIA affects small joints → **CORRECT:** Oligoarticular JIA primarily affects LARGE joints (knees, ankles, hips)
- ✗ **WRONG:** Polyarticular JRA has the best prognosis → **CORRECT:** Oligoarticular (pauciarticular) type 1 has the best prognosis
- ✗ **WRONG:** Iridocyclitis (uveitis) is very common in polyarticular JRA → **CORRECT:** Uveitis is most common in OLIGOARTICULAR type 1 JRA (ANA+)
- ✗ **WRONG:** In systemic JIA, ANA is positive → **CORRECT:** Systemic JIA typically has NEGATIVE ANA
- ✗ **WRONG:** Males who have polyarticular type develop chronic uveitis → **CORRECT:** Uveitis is most associated with ANA+ oligoarticular type 1, not polyarticular males

- First-line treatment for ALL JIA types: NSAIDs
- Uveitis in JIA: asymptomatic (does NOT present with eye pain/redness in children — requires slit lamp screening)

    - ✗ **WRONG:** Uveitis associated with JIA is usually symptomatic → **CORRECT:** JIA uveitis is usually ASYMPTOMATIC — requires active screening
- Positive RF found in most patients with oligoarticular JIA: WRONG — RF is negative in oligoarticular
    - ✗ **WRONG:** Rheumatoid factor positive in most patients with oligoarticular JRA → **CORRECT:** RF is NEGATIVE in oligoarticular JRA

9.2 SLE

- Criteria include: malar rash, discoid rash, photosensitivity, oral ulcers, arthritis, serositis, renal disease, neurological, hematological, ANA, anti-dsDNA, anti-Sm
    - ✗ **WRONG:** High complements are found in SLE → **CORRECT:** SLE causes LOW complement (C3 and C4)
- Photophobia is NOT an SLE criterion
    - ✗ **WRONG:** Photophobia is a feature of SLE → **CORRECT:** Photosensitivity (skin) is a criterion; photophobia (eyes) is not
- SLE patients with lupus anticoagulant: prolonged PTT but tend toward THROMBOSIS (not bleeding like hemophilia)
    - ✗ **WRONG:** SLE with lupus anticoagulant has hemorrhagic tendencies similar to hemophilia → **CORRECT:** Lupus anticoagulant causes THROMBOSIS, not hemorrhage
- Neonatal SLE: most serious complication = congenital heart block (maternal anti-Ro/SSA antibodies)

9.3 Dermatomyositis

- Heliotrope rash, Gottron's papules
- PROXIMAL muscle weakness (NOT distal)
    - ✗ **WRONG:** Distal muscle weakness is in pediatric dermatomyositis → **CORRECT:** Dermatomyositis causes PROXIMAL weakness
- Elevated muscle enzymes (CK, aldolase, AST)

9.4 Atopic Dermatitis

- IgE-mediated sensitization
- Pruritic (intense itching is hallmark)
- Distribution: infants → face/extensor surfaces; children/adults → flexor surfaces
    - ✗ **WRONG:** Diaper area is usually affected in atopic dermatitis → **CORRECT:** Diaper area is typically SPARED in atopic dermatitis
    - ✗ **WRONG:** Extensor surfaces in infants are spared → **CORRECT:** Extensor surfaces are AFFECTED in infants; flexors affected in older children
- Predisposition to asthma and allergic rhinitis (atopic march)
- Atopic vs seborrheic dermatitis: pruritus is the key feature favoring atopic

9.5 Septic Arthritis vs Transient Synovitis

- Septic arthritis: fever, inability to bear weight, raised CRP/ESR, raised WBC, leukocytes in synovial fluid
- MCC organism: Staphylococcus aureus
- Treatment: IV antibiotics (methicillin-sensitive Staph → oxacillin/nafcillin)
    - ✗ **WRONG:** During osteomyelitis treatment, CRP decreases more slowly than ESR → **CORRECT:** CRP normalizes FASTER than ESR in osteomyelitis treatment
- If hip effusion not drained → aseptic necrosis

9.6 Osteomyelitis

- MCC in children: Staphylococcus aureus
- Most common site: metaphysis of long bones

9.7 DDH (Developmental Dysplasia of Hip)

- Multifactorial inheritance
- More common in females
- Normal exam at birth does NOT exclude DDH
  - ✗ **WRONG:** Normal exam at birth excludes DDH → **CORRECT:** DDH can develop after birth and normal neonatal exam does NOT rule it out
- Hip X-ray is NOT reliable in newborns (unossified femoral head) — ultrasound preferred
- NOT a risk factor for DDH: polyhydramnios
- Harness treatment can cause avascular necrosis if not done properly

9.8 Behcet Disease

- Features: oral ulcers, genital ulcers, uveitis, DVT, vasculitis
  - ✗ **WRONG:** Malar rash is present in Behcet disease → **CORRECT:** Malar rash is in SLE, NOT Behcet

9.9 Skin Conditions in Newborns

- Erythema toxicum neonatorum: benign, eosinophils in pustules, common in term newborns
- Hemangiomas: respond to propranolol
- Mongolian spot, milia, harlequin phenomenon, acrocyanosis: all benign
- Coloboma (gap in eye structure): SERIOUS — not benign
- Vesicular rash in newborn: ABNORMAL (unlike common benign findings)
  - **(past question)** Which skin finding is abnormal in newborns: vesicular rash

⚡ HIGH-YIELD — RHEUMATOLOGY

- Oligoarticular JIA type 1: ANA+, females, large joints, ASYMPTOMATIC uveitis
- Systemic JIA: equal sex, ANA usually negative, uveitis absent
- First-line JIA: NSAIDs
- SLE: low complement; lupus anticoagulant → thrombosis (NOT bleeding)
- Dermatomyositis: PROXIMAL weakness
- Atopic dermatitis: diaper area SPARED; extensor in infants
- MCC septic arthritis: Staph aureus; if untreated → avascular necrosis
- CRP normalizes faster than ESR in osteomyelitis

10. PSYCHIATRY & NEURODEVELOPMENT

10.1 Autism Spectrum Disorder (ASD)

- Core deficit: impaired reciprocal SOCIAL COMMUNICATION and interaction
- DSM-5 criteria: social communication deficits + restricted/repetitive behaviors
- Rett syndrome was removed from DSM-5 ASD criteria (no longer included)
  - ✗ **WRONG:** Rett syndrome is included in DSM-5 ASD → **CORRECT:** Rett syndrome is NOT included in DSM-5 ASD
- Incidence is increasing globally
- Onset: recognized before age 3 years
- ASD can coexist with hyperactivity (does NOT rule it out)
  - ✗ **WRONG:** Hyperactivity rules out autism → **CORRECT:** Hyperactivity does NOT rule out autism
- Diagnosis: clinical; neuroimaging (MRI/CT) is NOT routinely used for diagnosis
  - ✗ **WRONG:** Most cases of autism are diagnosed by neuroimaging → **CORRECT:** Autism is diagnosed clinically, not by neuroimaging
- EEG: only if seizures suspected (not routinely required)
- No evidence for dietary intervention benefit
- Recurrence risk: NOT negligible (siblings have ~10–20% risk)
  - ✗ **WRONG:** Recurrence risk in ASD is negligible if no cause identified → **CORRECT:** Recurrence risk for siblings is ~10–20%, NOT negligible
- No relation to immunizations
- Screening: recommended at 18–24 months
- Drug treatment: risperidone (for irritability/behavioral issues in ASD)

10.2 ADHD

- Criteria (DSM-5): inattention and/or hyperactivity-impulsivity symptoms
- Symptoms present in MORE THAN ONE setting
- Onset: symptoms before age 12 years (DSM-5 changed from age 7)
  - ✗ **WRONG:** Symptoms must appear before age 7 in ADHD (DSM-5) → **CORRECT:** DSM-5 changed the onset criterion to BEFORE AGE 12 (not 7)
  - ✗ **WRONG:** Symptoms of ADHD last at least 2 years → **CORRECT:** Symptoms must be present for at least 6 months
  - ✗ **WRONG:** Most patients show improvement by age 18 → **CORRECT:** Many patients continue to have symptoms into adulthood
  - ✗ **WRONG:** ADHD is more common in females → **CORRECT:** ADHD is more common in MALES
  - ✗ **WRONG:** ADHD is mostly diagnosed in the first year → **CORRECT:** ADHD onset is typically school-age
- NOT included in DSM-5: Asperger syndrome (merged into ASD); ADHD symptoms before age 18 (criterion is before age 12)
- NOT a diagnostic criterion: stereotyped behavior (this is in ASD)
- Onset: typically evident in preschool/early school years (NOT absent in preschool years)
  - ✗ **WRONG:** Onset of ADHD symptoms is usually not evident in preschool years → **CORRECT:** Symptoms are often evident in PRESCHOOL years

- Treatment: behavioral therapy + methylphenidate (first-line pharmacotherapy)
- Other drugs: atomoxetine (NRI), clonidine
  - ✗ **WRONG:** Phenobarbital is treatment for ADHD → **CORRECT:** Phenobarbital is NOT used for ADHD

- ✗ **WRONG:** Tricyclic antidepressants are first choice → **CORRECT:** Methylphenidate + behavioral therapy is first-line

10.3 Eating Disorders

- Anorexia nervosa: hypokalemia, hypomagnesemia, HIGH cortisol (stress response)
  - ✗ **WRONG:** Low cortisol in anorexia nervosa → **CORRECT:** Cortisol is HIGH (elevated) in anorexia nervosa
- Low gonadotropins (amenorrhea)

⚡ HIGH-YIELD — PSYCHIATRY/NEURODEVELOPMENT

- ASD core: social communication deficit; risperidone for irritability
- Rett syndrome: NOT in DSM-5 ASD
- ADHD onset: before age 12 (not 7); must be in 2+ settings
- ADHD: not more common in females — more common in males
- Hyperactivity does NOT rule out autism
- ASD: neuroimaging not used for diagnosis; EEG only if seizures

11. METABOLIC DISORDERS & GENETICS

11.1 Inborn Errors of Metabolism (IEM)

- General presentations: encephalopathy, acidosis, hypoglycemia, hyperammonemia
  - ✗ **WRONG:** Alkalosis is a presentation of IEM → **CORRECT:** IEM causes ACIDOSIS (not alkalosis); alkalosis is NOT a typical IEM presentation
  - ✗ **WRONG:** Cyanotic heart disease is a likely presentation of IEM → **CORRECT:** Cyanotic heart disease is NOT typical of IEM
- HAGMA causes in IEM: organic acidemia, urea cycle defects (some), fatty acid oxidation defects
- Hyperammonemia: urea cycle defects (venous blood gas normal, normal CBC + elevated ammonia)
- Disorders with organomegaly: Gaucher, Niemann-Pick, galactosemia, tyrosinemia, glycogen storage disease
  - ✗ **WRONG:** PKU is associated with organomegaly → **CORRECT:** PKU is NOT associated with organomegaly
- Disorders with abnormal urine smell: PKU (musty), MSUD (maple syrup), isovaleric acidemia (sweaty feet), methylmalonic acidemia, branched-chain amino acid disorders
  - ✗ **WRONG:** Fructosemia is associated with abnormal urine smell → **CORRECT:** Fructosemia does NOT characteristically cause abnormal smell

11.2 PKU (Phenylketonuria)

- Autosomal recessive; aminoacid disorder (NOT lipid storage, NOT urea cycle)
- Missense mutation in PAH gene
- Features: fair skin/blond hair, musty urine smell, intellectual disability, seizures
- Neonatal screening: part of routine screen in Jordan
- Treatment: phenylalanine-restricted diet; breastfeeding is NOT contraindicated

    - ✗ **WRONG:** Breastfeeding is contraindicated in PKU → **CORRECT:** Breastfeeding is NOT contraindicated in PKU (breast milk has lower Phe than cow's milk); supplemented with Phe-free formula
    - ✗ **WRONG:** Early dietary restriction has minimal benefit in PKU → **CORRECT:** Early treatment prevents intellectual disability
    - ✗ **WRONG:** Acute intermittent course is characteristic of PKU → **CORRECT:** PKU is CHRONIC, not episodic
- PKU is NOT a cause of hypoglycemia

11.3 Galactosemia

- AR; galactose-1-phosphate uridyltransferase deficiency
- Presents in newborn: jaundice, cataracts, E. coli sepsis, vomiting/diarrhea, liver disease
    - ✗ **WRONG:** Pseudomonas sepsis is characteristic of galactosemia → **CORRECT:** E. COLI sepsis is characteristic of galactosemia
- Treatment: galactose-free diet (NOT sucrose-free)
    - ✗ **WRONG:** Sucrose-free diet is treatment for galactosemia → **CORRECT:** GALACTOSE-free (lactose-free) diet is treatment
- Jaundice: appears early in neonatal period (NOT second year of life)
    - ✗ **WRONG:** Jaundice in galactosemia appears in second year of life → **CORRECT:** Jaundice appears in NEONATAL period
- Diagnosis: reducing substance in urine
- NOT associated with galactosemia: metabolic alkalosis (causes acidosis)

11.4 MSUD (Maple Syrup Urine Disease)

- Branched-chain amino acid (leucine, isoleucine, valine) metabolism defect
- Characteristic urine smell: maple syrup
- Neonatal presentation: poor feeding, encephalopathy, seizures

11.5 Mucopolysaccharidoses (MPS)

- Features: coarse facies, hepatosplenomegaly, bone deformities, developmental regression
- Hurler syndrome (MPS I): alpha-L-iduronidase deficiency, AR
- Hunter syndrome (MPS II): iduronate sulfatase, X-linked recessive
- MPS does NOT cause acute neurological manifestations (unlike organic acidemias)
    - ✗ **WRONG:** MPS presents with acute neurological manifestations → **CORRECT:** MPS causes chronic progressive decline, NOT acute neurological crisis
- MPS does NOT cause hepatic dysfunction (mucopolysaccharides accumulate but not hepatotoxic)
    - ✗ **WRONG:** MPS is associated with hepatic dysfunction → **CORRECT:** MPS causes hepatosplenomegaly but NOT hepatic dysfunction/fibrosis like tyrosinemia

11.6 Mitochondrial Disorders

- Leigh disease is a mitochondrial DNA disorder
- High serum very long chain fatty acids: Zellweger disease (peroxisomal disorder)
- Treatment: Coenzyme Q10
- Presents: regression, lactic acidosis, multisystem

11.7 Other IEM

- Wilson disease: copper; low ceruloplasmin; allopurinol NOT used (used for Lesch-Nyhan)
- Lesch-Nyhan syndrome: uric acid metabolism disorder (HGPRT deficiency); allopurinol treatment
- Acrodermatitis enteropathica: zinc deficiency (perioral/perianal rash, diarrhea, alopecia)
  - ✗ **WRONG:** Acrodermatitis enteropathica is normal growth rate → **CORRECT:** Growth is impaired in acrodermatitis enteropathica

11.8 Genetics — Key Concepts

Chromosomal Disorders

- Down syndrome (trisomy 21): trisomy due to nondisjunction; associated with ADVANCED MATERNAL AGE
  - ✗ **WRONG:** Increased incidence with fathers over 35 → **CORRECT:** Down syndrome correlates with MATERNAL (not paternal) age
- Down syndrome features: hypotonia, short stature, single palmar crease, Brushfield spots, epicanthic folds, mental retardation, endocardial cushion defect (most common cardiac defect), small macro ears (not 'malformed micro')
  - ✗ **WRONG:** Postaxial polydactyly is a feature of Down syndrome → **CORRECT:** Polydactyly is NOT a feature of Down syndrome
  - ✗ **WRONG:** Hydrocephalus is a common finding in Down syndrome → **CORRECT:** Hydrocephalus is NOT common in Down syndrome
- Down syndrome associations: leukemia (ALL), celiac, congenital hypothyroidism, Hirschsprung, Alzheimer, cataracts
  - ✗ **WRONG:** Cystic fibrosis is associated with Down syndrome → **CORRECT:** CF is NOT associated with Down syndrome
  - ✗ **WRONG:** DM is NOT associated with Down syndrome → **CORRECT:** Type 1 DM can occur (autoimmune) but is not a classic association
  - ✗ **WRONG:** DI is associated with Down syndrome → **CORRECT:** DI is not associated with down
- Karyotyping: used for chromosomal disorders (Down, Turner, Klinefelter)
- Turner syndrome (45,X): does NOT correlate with advanced maternal age; normal IQ
  - ✗ **WRONG:** Turner syndrome is associated with low IQ → **CORRECT:** Turner syndrome has NORMAL IQ generally
- Klinefelter (47,XXY): tall, small testes, infertility, gynecomastia
- Trisomy 18 (Edwards): rocker-bottom feet, micrognathia, cardiac defects, low birth weight; MICROCEPHALY (not macrocephaly)
  - ✗ **WRONG:** Macrocephaly is seen in Edwards syndrome → **CORRECT:** Edwards syndrome causes MICROCEPHALY
- Trisomy 13 (Patau): microcephaly, scalp defects, small eyes, polydactyly, cardiac + renal malformations

Genetic Inheritance

- Autosomal dominant with anticipation (triplet repeat): Huntington, Myotonic dystrophy, Fragile X (but Fragile X is X-linked)
- Imprinting disorders: Prader-Willi syndrome (paternal deletion 15q), Angelman syndrome (maternal deletion 15q)
  - ✗ **WRONG:** Kearns-Sayre syndrome is an imprinting disorder → **CORRECT:** Kearns-Sayre is a mitochondrial disorder
- Y-linked: only males affected; ALL sons of affected father are affected; father to son transmission

    - ✗ **WRONG:** X-linked dominant is transmitted from affected father to all sons → **CORRECT:** Father-to-son transmission is Y-LINKED, not X-linked (fathers give Y to sons)
- Autosomal recessive: G6PD (NO — X-linked), 21-OH deficiency (YES), CF (YES), ataxia telangiectasia (YES)
    - ✗ **WRONG:** G6PD is autosomal recessive → **CORRECT:** G6PD is X-LINKED RECESSIVE
- X-linked recessive NOT: Vitamin D-resistant rickets (X-linked DOMINANT), Rett syndrome
    - ✗ **WRONG:** Vitamin D-resistant rickets is X-linked recessive → **CORRECT:** Hypophosphatemic rickets is X-linked DOMINANT
- Males with autosomal dominant disorders transmit to sons (male-to-male transmission): differentiates AD from X-linked

- Fragile X syndrome: X-linked; large ears, long face, macro-orchidism (large testes — in adolescents/adults; Normal at birth/childhood) , hyperextensible finger joint , autistic behavior , intellectual disability
    - ✗ **WRONG:** Small testes is a feature of Fragile X in children → **CORRECT:** Macro-orchidism appears post-pubertally , in children testes has normal size
    - ✗ **WRONG:** Small testes are a characteristic of Fragile X at 3 years old → **CORRECT:** Small testes are not a specific Fragile X feature at this age
- Chromosome instability syndromes: ataxia telangiectasia, Bloom syndrome, Fanconi anemia, xeroderma pigmentosum
    - ✗ **WRONG:** Klinefelter is associated with chromosome instability → **CORRECT:** Klinefelter is NOT a chromosome instability syndrome

- McCune-Albright syndrome: café-au-lait spots, polyostotic fibrous dysplasia, autonomous endocrine hyperfunction; FEMALE predominance; G-protein missense mutation
    - ✗ **WRONG:** McCune-Albright usually presents in males → **CORRECT:** McCune-Albright usually presents in FEMALES

11.9 IgE Mediated Allergy

- IgE-mediated allergy = Type I hypersensitivity (immediate)
- Elevated IgE: parasitic infestation, extrinsic asthma, allergic rhinitis, hyper-IgE syndrome
    - ✗ **WRONG:** Ataxia telangiectasia causes elevated IgE → **CORRECT:** Ataxia telangiectasia causes LOW IgE (immunodeficiency)

⚡ HIGH-YIELD — METABOLIC & GENETICS

- PKU: aminoacid disorder; dietary treatment; breastfeeding allowed; chronic course
- Galactosemia: E. coli sepsis + cataracts + jaundice; galactose-free diet
- MSUD: maple syrup urine smell
- MPS: coarse features, hepatosplenomegaly; chronic NOT acute neurological
- Down: maternal age; endocardial cushion defect; ALL leukemia; no polydactyly
- Turner: normal IQ; not maternal age dependent
- Trisomy 18: microcephaly (NOT macrocephaly), rocker-bottom feet
- Y-linked: father-to-ALL-sons; Prader-Willi = imprinting
- G6PD: X-linked recessive (not AR)

12. GENERAL & MISCELLANEOUS

12.1 Poisoning & Toxicology

- Paracetamol toxicity: antidote = N-acetylcysteine; liver injury most feared; level taken at 4 hours post ingestion; ipecac NOT recommended
  - ✗ **WRONG:** Ipecac should be given for paracetamol overdose → **CORRECT:** Ipecac is NO LONGER recommended for any poisoning
- Organophosphate poisoning: cholinergic syndrome (DUMBBBELLSS); features: miosis, bradycardia, bronchospasm, diarrhea, lacrimation, salivation, urination
  - ✗ **WRONG:** Hyperglycemia is a feature of organophosphate poisoning → **CORRECT:** Organophosphate causes cholinergic symptoms including HYPOGLYCEMIA possibly; NOT hyperglycemia
  - ✗ **WRONG:** Dry mouth is a feature of organophosphate poisoning → **CORRECT:** Organophosphate causes EXCESS secretions (wet: lacrimation, salivation, bronchorrhea)
- Organophosphate antidote: atropine + pralidoxime
- Iron overdose: abdominal X-ray to see remaining tablets; deferoxamine for CHRONIC/severe poisoning; BAL is NOT used for iron
  - ✗ **WRONG:** BAL (British anti-lewesite) is antidote for iron poisoning → **CORRECT:** BAL is for heavy metals (arsenic, mercury); iron antidote is deferoxamine
- Cyanide: antidote = sodium nitrite + sodium thiosulfate
- Opioids: antidote = naloxone
- Atropine: antidote = physostigmine
- Salicylate overdose: uncoupling of oxidative phosphorylation; causes respiratory alkalosis then metabolic acidosis
  - ✗ **WRONG:** Aspirin causes metabolic alkalosis → **CORRECT:** Aspirin causes respiratory alkalosis (early) + metabolic acidosis (late); NOT metabolic alkalosis
- Corrosive ingestion (acid/alkali): next step = esophagoscopy (NOT lavage, NOT nasogastric tube)
- Severely constricted pupils in comatose child: drug poisoning (opioids, organophosphates)

12.2 Familial Mediterranean Fever (FMF)

- Autosomal recessive; Mediterranean populations (NOT restricted to Mediterranean)
  - ✗ **WRONG:** FMF affects people living in the Mediterranean area only → **CORRECT:** FMF can affect people of any origin
- Features: recurrent febrile episodes <5 days, abdominal pain (MCC), arthritis (large joints), pleuritis, skin rash
  - ✗ **WRONG:** Pleuritis is more common than arthritis in FMF → **CORRECT:** Arthritis is more common than pleuritis
- Arthritis affects LARGE joints (not small joints)
  - ✗ **WRONG:** Recurrent arthritis of small joints is seen in FMF → **CORRECT:** FMF arthritis affects LARGE joints
- Amyloidosis: major long-term complication (renal)
- Treatment: colchicine (prevents attacks AND amyloidosis)
- Colchicine side effects: diarrhea, myopathy; does NOT cause irreversible azoospermia
  - ✗ **WRONG:** Colchicine causes irreversible azoospermia → **CORRECT:** Colchicine does NOT cause irreversible azoospermia

12.3 Child Abuse

- Most likely presentation: bruises (MCC physical sign)
- Sign of child abuse: presence of associated injuries
- Most likely age group physically abused: toddlers
  - ✗ **WRONG:** Infants are the most likely age group physically abused → **CORRECT:** Toddlers are the most commonly physically abused age group
- Child neglect: NOT the least common (it's actually the most common form of child maltreatment)
  - ✗ **WRONG:** Child neglect is the least common form of abuse → **CORRECT:** Neglect is the MOST common form of child maltreatment
- Subdural hematoma, spiral fracture, hot water burns are also indicators

12.4 Steroid Side Effects

- Side effects: psychosis, HTN, weight gain, hyperglycemia, osteopenia, intracranial hypertension, cataracts (not keratitis), myopathy
  - ✗ **WRONG:** Uveitis/keratitis is a side effect of systemic steroids → **CORRECT:** Steroids cause CATARACTS, not uveitis/keratitis
  - ✗ **WRONG:** Cyclosporine causes hypotension → **CORRECT:** Cyclosporine causes HYPERTENSION

12.5 Drug Side Effects Summary

Drug	Key Side Effect
Propranolol	Bronchospasm
Prednisolone	Cataracts, osteopenia, HTN, hyperglycemia
Cyclophosphamide	Hemorrhagic cystitis
Cyclosporine	HYPERTENSION (not hypotension), nephrotoxicity
Carbamazepine	Liver toxicity, hyponatremia, SJS
Sodium valproate	Thrombocytopenia, hepatotoxicity, weight gain
Desmopressin	Hyponatremia
Clonazepam	CNS depression
Diazepam	Respiratory depression
Aspirin	Reye syndrome, GI bleeds, inhibits platelets; causes resp alkalosis + metabolic acidosis (NOT metabolic alkalosis)

  - ✗ **WRONG:** Clonazepam causes decreased salivation and bronchial secretions → **CORRECT:** Clonazepam does not significantly affect salivation or bronchial secretions, it mainly causes CNS depression

Note : Clonazepam is well-known for causing an increase in saliva. However, this is often not because the salivary glands are overproducing. Because clonazepam is a central nervous system depressant and a muscle relaxant, it can slow down the automatic swallowing reflex. This causes normal amounts of saliva to pool in the mouth, which can lead to drooling.

12.6 Vitamin Deficiencies

Vitamin	Deficiency Signs
Vitamin A	Night blindness, xerophthalmia, keratomalacia
Vitamin C	Scurvy: bleeding gums, petechiae, periosteal hemorrhage
Vitamin B12	Megaloblastic anemia, neurological damage
Vitamin E	Cerebellar ataxia, peripheral neuropathy, hemolysis, muscle weakness — NOT hepatosplenomegaly
Vitamin K	Bleeding (hemorrhagic disease of newborn)
Zinc	Acrodermatitis enteropathica, alopecia, growth failure
Riboflavin (B2)	Cheilosis (cracked lips)

    - ✗ **WRONG:** Hepatosplenomegaly is a feature of Vitamin E deficiency → **CORRECT:** Hepatosplenomegaly is NOT caused by Vit E deficiency

12.7 Gestational Diabetes Complications

- IDM: macrosomia, hyperinsulinemia, hypoglycemia, polycythemia, cardiomegaly, RDS, congenital anomalies
    - ✗ **WRONG:** Gestational diabetes is associated with congenital cardiac anomalies → **CORRECT:** Gestational diabetes (GDM) is NOT a major cause of congenital cardiac anomalies (pre-existing DM is, but GDM is less clearly linked)
    - ✗ **WRONG:** Gestational diabetes is associated with oligohydramnios → **CORRECT:** GDM is associated with POLYHYDRAMNIOS

12.8 Miscellaneous Clinical Pearls

- Thyroglossal duct cyst: diagnose with ultrasound
- Pinworm (Enterobius) treatment: mebendazole (drug of choice); treat ALL family members
    - ✗ **WRONG:** Mebendazole is NOT drug of choice for pinworm → **CORRECT:** Mebendazole IS the drug of choice
    - ✗ **WRONG:** Treat only siblings (not all family members) for pinworm → **CORRECT:** ALL family members should be treated
- Most indicative of respiratory failure: altered mental status (not just low O2 sat)
- Least factor contributing to tissue perfusion: PaO2 (oxygen content = Hb × SaO2 + dissolved; Hb and cardiac output matter more)
- NF1 (neurofibromatosis type 1): associated with optic nerve glioma
- William syndrome: high serum calcium (hypercalcemia) + supravalvular aortic stenosis + elfin facies + intellectual disability
    - ✗ **WRONG:** DiGeorge syndrome has hypercalcemia → **CORRECT:** DiGeorge has HYPOcalcemia (absent parathyroids); William syndrome has HYPERcalcemia
- Sanjad-Sakati syndrome: hypoparathyroidism, growth failure, dysmorphic features
- Cherry red spots in retina: storage disorders (Tay-Sachs, Niemann-Pick, Farber disease)
- Vancomycin level high within therapeutic range: increase the INTERVAL between doses
- Neural tube defects : Anencephaly , meningocele , meningomyelocele , spina bifida occulta (Not Hydrocephalous)

HIGH-YIELD — GENERAL

- Paracetamol overdose: N-acetylcysteine; take level at 4 hours; no ipecac
- Organophosphate: cholinergic (DUMBBBELLSS); antidote atropine; NOT dry mouth/hyperglycemia
- Iron antidote: deferoxamine; BAL is for heavy metals ONLY
- FMF: large joints; more arthritis than pleuritis; colchicine prevents amyloidosis
- Child abuse MCC: bruises; most common age: toddlers; neglect is MOST common type
- Aspirin: respiratory alkalosis + metabolic acidosis (NOT metabolic alkalosis)
- Cyclosporine: HYPERTENSION
- DiGeorge: HYPOcalcemia; William: HYPERcalcemia