Gateway to

PEDIATRICS UWORLD

First Edition
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CONTENTS INCLUDE:

- 735 UWorld Pediatrics Questions
- Consolidated Tables & Diagrams
- Topic-Based Organization

For the purpose of University Pediatrics Final Exam, you can skip the following chapters:

- Chapter 4: ENT
- Chapter 10: Rheumatology & Orthopedics
- Chapter 11: Dermatology
- Chapter 13: Psychiatry
- Chapter 14: Public Health Sciences
- Chapter 15: Toxicology & Environmental Exposure
- Chapter 17: Miscellaneous

Notes:

- Due to huge differences between the Jordanian and U.S. vaccinations program and developmental milestones, **this dossier does NOT include the UWorld questions about those topics. So, study them from an external source.**
- **Sorry if the file is a bit long, I tried to cut it down as much as possible.**
- Please don't hesitate to reach out to me **if you find any mistakes or have any questions**, thanks 😊 !

Good Luck!

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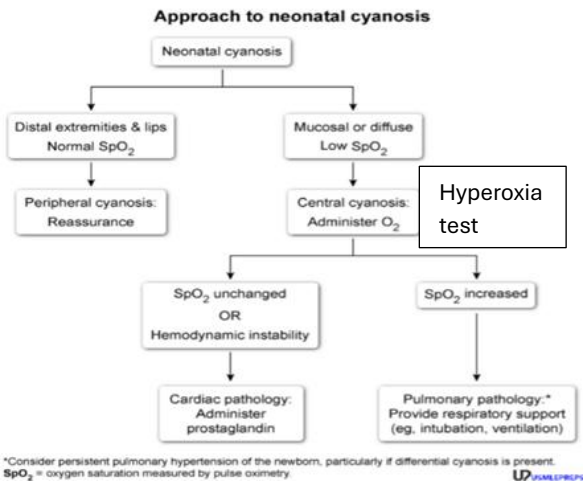
Chapter 1: Cardiology

22q11.2 deletion syndromes (22qDS) [DiGeorge syndrome, velocardiofacial syndrome]	
Pathogenesis	• Loss of genes that control embryonic tissue template transformation (3+4 pharyngeal pouch, secondary heart field) and migration of neural crest cells
Clinical features (CATCH)	• **C**ardiac outflow tract anomalies (TOF, persistent truncus arteriosus, interrupted Aortic arch). **A**nomalous face (ocular hypertelorism, prominent nasal bridge, low-set posteriorly rotated ears, micro/retrognathia). **T**hymic hypoplasia /aplasia → ↓ T-cell immunity (recurrent viral and fungal infx), + bacterial due to reduced B cell activation. **C**left palate. **H**ypoparathyroidism (hypoplastic /aplastic parathyroids) → hypocalcemia
DiGeorge requires thymic involvement (CD3+ T-cell count **<500**) (CTH [cardiac thymus hypothyroid]); velocardiofacial syndrome requires facial abnormalities.	

- Once DGS is suspected, obtain **echocardiography and serum calcium**.
- Hypocalcemia in neonates: tremulousness, **tetany**, **seizures**, and **arrhythmias**.
- DGS: lymphopenia and rarely thrombocytopenia. NO neutropenia or anemia.



Causes of hypoxemia			
	Examples	**A-a gradient**	**Corrects with O_2?**
Reduced PiO_2	High altitude	Normal	Yes
Hypoventilation	CNS depression, morbid obesity	Normal	Yes
Diffusion limitation	Emphysema, ILD	Increased	Yes
V/Q mismatch*	Small PE, lobar pneumonia	Increased	Yes
Large intrapulmonary shunt	Diffuse pulmonary edema	Increased	No
Large dead-space ventilation	Massive PE, right-to-left intracardiac shunt	Increased	No

Benign vs pathologic murmurs		
	Benign	**Pathologic**

History	• Asymptomatic • Normal growth • No significant family history	• Infants: poor weight gain, respiratory distress, difficulty feeding • Older children: exertional fatigue, chest pain, syncope • Family history of SCD or CHD
Murmur characteristics*	• Early or midsystolic • Musical or vibratory • Grade 1-2 intensity • ↓ with standing & valsalva maneuver**	• Holosystolic or diastolic • Harsh • Grade ≥3 intensity • Intensity persists with standing & Valsalva maneuver
Other findings	• Normal vital signs • Normal S1 & S2 • Symmetric pulses	• Central cyanosis • Loud, fixed, or single S2 • Weak femoral pulses • Hepatomegaly
Management	• Reassurance	• ECG & echocardiography
*__Any single pathologic__ feature warrants echo. **Venous hum is a benign murmur exception that increases with standing.		

Still murmur: MC innocent murmur in young children. Systolic murmur due to turbulent left ventricular outflow. ↓ with standing & valsalva maneuver. ↑ when supine. **Reassurance**, resolves by adolescence.

Peripheral pulmonary stenosis, benign, turbulent flow in the **physiologically** narrow and acutely branched pulmonary arteries. Findings include a **soft, low-pitched midsystolic murmur** best heard along the left or right upper sternal border with radiation to the axillae or back. The murmur resolves during **early infancy**, **reassurance**.

❖ The liver edge is normally palpated **up to 3 cm** below the costal margin in newborns.

TOF: Clinical presentation depends on the severity of **RVOT obstruction** resulting from pulmonary stenosis. Severe RVOT obstruction: shortly after birth with central (eg, perioral) cyanosis. Moderate obstruction: Childhood with sudden hypoxic/hypercyanotic "tet"

spells due to right-to-left shunting during exertion or agitation [exertional dyspnea & syncope]. Harsh, **crescendo-decrescendo** systolic ejection murmur over the mid to left upper sternal border due to RVOT obstruction. **Squatting** may ↓ **symptoms** (eg, cyanosis) and increases the intensity of the systolic murmur. The single S2 comprises the normal aortic and the inaudible pulmonary components. TOF typically occurs sporadically.

ToF repair, typically performed in early childhood, involves patch closure of the VSD and **restructuring** (eg, enlarging) of the **narrowed RVOT**.

The eccentric hypertrophy seen in patients with repaired ToF (in whom relief of RVOT obstruction reduces RV pressure load) contrasts with the concentric RV hypertrophy seen in uncorrected ToF (due to persistent, increased RV pressure load).

Acute management involves **knee-chest positioning (kinks femoral A)** /Squats as well as inhaled **oxygen** to stimulate pulmonary vasodilation and systemic vasoconstriction.

Persistent pulmonary hypertension of the newborn	
Pathogenesis	• Abnormal persistence of elevated fetal pulmonary vascular resistance (persistence of fetal circulation). ↓ RV output • Right-to-left shunting across ductus arteriosus • Dysfunctional vascular development or adaptation to insults
Risk factors	• Lung hypoplasia(CDH). Meconium aspiration. Neonatal pneumonia
Examination	• ↓ Postductal relative to preductal oxygen saturation • Respiratory distress & cyanosis + Prominent S2
Treatment	• Oxygenation & ventilation. Inhaled NO. If all failed vecuronium

NO has a half-life of **only a few secs**, limiting vasodilation to the pulmonary vasculature.

(Choice C) NSAIDS closing the ductus in PPHN would further increase right ventricular afterload and worsen right ventricular output.

Transposition of the great vessels (TGV) **cyanosis within the first 24 hours of life**, tachypnea, **single S2** (absent pulmonary component because the aorta is anterior to the pulmonary artery). Murmurs are atypical for TGV but may be heard if a concomitant ventricular septal defect is present. Chest x-ray: narrow mediastinum ("**egg on a string**" appearance), and echocardiogram is diagnostic.

(Choice C) Hypoplastic left heart syndrome may present with tachypnea, cyanosis, no murmur, and a single S2; few days after birth when the ductus arteriosus closes. **Staged palliative repair** should occur in infancy, and **transplantation** may be indicated for heart failure that is **refractory to medical therapy and surgical palliation**.

(Choice E) Isolated ASD is an acyanotic lesion that causes a wide and fixed splitting of S2 and a systolic ejection murmur due to increased blood flow across the pulmonic valve.

Tricuspid atresia: Peaked P wave, single S2(P.stenosis), left axis deviation, normal heart size on x-ray but flattened right heart border + decreased lung markings, VSD ASD.

- **Absent tricuspid valve**: No flow from right atrium (RA) to right ventricle (RV) results in **enlarged RA** (ie, **tall, peaked P waves**) and **hypoplastic RV**. Lack of flow through the pulmonary outflow tract leads to underdevelopment of the pulmonic valve and/or artery (**pulmonary stenosis**).
- **Atrial septal defect** (ASD): Rather than the fixed, widely split S2 characteristic of ASD, patients typically have a single S2 due to pulmonic stenosis. Because the entire systemic VR is delivered to the left side of the heart, **left-sided volume overload** occurs (ie, **left axis deviation**) + central cyanosis in **newborn** period.
- **Ventricular septal defect** (VSD): Ventricular communication is typically present to allow pulmonary blood flow, although the pulmonary stenosis causes pulmonary undercirculation (**decreased pulmonary markings** on chest x-ray).

(Choice C) **TAPVR**: oxygenated blood from the lungs bypasses the LA and drains into the RA, leading to cyanosis. Right-sided volume overload results in RA and RV enlargement (ie, right axis deviation) and pulmonary overcirculation (ie, increased pulmonary markings).

(Choice E) Truncus arteriosus causes cyanosis due to mixing of blood between the aorta and pulmonary artery. A systolic ejection murmur is typical, and x-ray reveals cardiomegaly and increased pulmonary vascular markings.

(Choice C) Symptomatic **Ebstein anomaly**: Tricuspid regurg, accessory conduction pathways, right-sided HF. "Triple or quadruple gallop" (widely split S1 and S2 plus a loud S3 and/or S4) and a holosystolic or early systolic murmur at the left lower sternal border.

Routine newborn care	
Preventive	• IM vit K. Erythromycin eye ointment. Hep B vaccine
Screening	• Newborn screen (metabolic/genetic disorders)

	• Hyperbilirubinemia. Hearing screen • Pre- (Rt arm) & postductal (either leg) pulse oximetry (congenital heart disease). Hypoglycemia (select populations)

Acrocyanosis (blue extremities & circumoral area, pink body): common in the first days of life due to initial peripheral vasoconstriction.

Critical CHD includes heart defects that require early (within 1 year) intervention, such as **ductal-dependent and cyanotic lesions**. One-third of patients with critical CHD are asymptomatic for the first few days of life (due to patent ductus arteriosus).

Echocardiogram is indicated for a positive screen (<90% in either extremity, <95% in both upper and lower extremities, or >3% difference between upper and lower extremities).

(Choice A) Electrocardiogram for bradycardia and suspected neonatal heart block.

Ventricular septal defect		
	Small	**Moderate to large**
Clinical features	• Asymptomatic • Loud, harsh, holosystolic murmur at LLSB	• Respiratory distress, poor feeding, poor growth pHTN • Loud S2 and murmur
Chest x-ray	• Normal	• Cardiomegaly + increased lung markings
Management	• Serial echocardiography • Most spontaneously close at age <2 (No tx needed)	• Diuretics • Defect closure

(Choice C) Conditions that require **SBE prophylaxis** include **unrepaired** cyanotic congenital heart disease and a **recently or partially repaired** (ie, residual shunt) defect.

Moderate or large ventricular septal defect	
Timeline	**Pathophysiologic changes**
In utero	• RV & LV pressures equal. No hemodynamic consequences
Transition	• ↓ PVR, ↑ SVR. Left-to-right shunting through VSD

Infancy	• RV LA & LV volume overload/dilation. Pulmonary overcirculation
Late childhood	• (↑ PVR). Right-to-left shunting **(Eisenmenger)**

LVO increases but cannot keep up with the increased blood return, resulting in **high-output cardiac failure**.. **Tachypnea**, **tachycardia**, **diaphoresis**, and **pallor**, poor feeding, FTT. Bilateral rales (↑ extravascular transudate in the lungs), palpable thrill, hepatomegaly.

LV contractility normal, ↓ afterload , volume overload= eccentric hypertrophy.

As right ventricular pressure increases due to increased flow, the **S2** will become more prominent due to increased pressure closing the pulmonary valve. An apical **diastolic rumble** may also be heard from increased flow across the mitral valve.

PDA maximal intensity is typically at S2, when pressure in the aorta is at its peak.

A small PDA is often asymptomatic, detected incidentally on cardiac auscultation vs larger PDA: respiratory distress, poor feeding, retracted growth within the first year of life, high cardiac output and **hyperdynamic circulation** (eg, accentuated peripheral pulses) due to the left ventricle compensating for the significant reduction in SVR created by PDA shunt.

Coarctation of the aorta	
Pathology	• Thickening of tunica media of aortic arch
Clinical features	• ↑ BP & strong pulses in upper extremities • ↓ BP & weak pulses in lower extremities • Neonates* (severe narrowing): HF + cardiogenic shock (poor feeding, diaphoresis, P.edema, cardiomegaly, ↓UO, lactic acidosis) • Children/adults (mild narrowing): Lower extremity claudication. Palpable pulsations of intercostal vessels (collaterals). Secondary hypertension (upper arms)
Complications	• HF berry aneurysm aortic rupture IE (HBAI)
Treatment	• PGE1 for neonates with severe narrowing + Surgical repair
*After closure of ductus arteriosus. (DA is good here)	

Continuous murmur over the back from flow through compensatory collateral vasculature.

Screening in Turner syndrome (Tx: rGH, Estradiol (age 11), Progestin (at menarche))	
Cardiovascular	• Congenital heart defects*: 4-extremity BP, ECG, echocardiogram • Aortic dilatation/dissection: TTE or cardiac MRI (Dissection tx: Beta blockers, BP control + surgery) • Metabolic syndrome: BP, HbA1c, lipid panel, LFT
Renal	• Horseshoe kidney: renal ultrasound
Musculoskeletal	• Osteoporosis: 25-hydroxyvitamin D level, DXA scan
Vision & hearing	• Strabismus, myopia: ophthalmology evaluation • Recurrent otitis media, hearing loss: audiology testing (anatomic ear malformations)
Autoimmune	• Celiac disease: tissue transglutaminase antibodies • Hypothyroidism: TSH, free T4
*Aortic coarctation, bicuspid aortic valve./ dissection (high risk in pregnancy) / HTN prolonged QT CAD	

Patients also have scattered pigmented nevi, dysplastic nails. Treatment of congenital lymphedema is supportive (eg, compression bandages, skin care), and symptoms generally improve in early childhood.

Chronic pulmonic regurgitation	
Etiologies	• Correction of pulmonic stenosis or TOF (iatrogenic) [MC] • Rheumatic heart disease, infective endocarditis
Pathophysiology	• Progressive RV volume overload → RV dilation (eccentric hypertrophy) & eventual failure
Clinical findings	• Decrescendo, diastolic murmur at LSB, ↑ with inspiration Soft (or sometimes absent) P2 (single S2). Dyspnea & prominent parasternal impulse (once RV dilation develops)
Management	• Serial echocardiography • Valve replacement once symptoms or RV dysfunction develops

(Choice C) Left atrial myxoma often causes transient mitral valve obstruction and may be recognized by a murmur of mitral stenosis (ie, diastolic rumble at apex).

PDA-dependent congenital heart disease (sudden deterioration in normal 1 day old)
Coarctation of the aorta. D-TGA. Hypoplastic left heart syndrome. TAPVR. Tricuspid atresia

PDA begins to close in the first 24 hours of life. **Prostaglandin E1** is a **vasodilator** that prevents PDA closure. It can be administered even after the PDA has closed because it may help reestablish duct patency. Infusion can begin before a definitive diagnosis is made.

(Choice B) Polycythemia: hypoxia, peripheral cyanosis, normal O2 saturation, Tx: partial exchange transfusion.

Viral myocarditis (Coxsackievirus B, adenovirus)	
Clinical features	• Viral prodrome → Heart failure: respiratory distress, dilated cardiomyopathy & S3 gallop with MR murmur, hepatomegaly
Diagnosis	• **Chest x-ray**: cardiomegaly, pulmonary edema. **ECG**: sinus tachycardia • **Echocardiogram**: global hypokinesis + decreased EF • **Biopsy** (**gold standard**): inflammation, necrosis (direct viral injury + autoimmune inflammation → impaired systole and diastole)
Treatment	• Supportive (eg, diuretics, inotropes) + IVIG , monitor in ICU for risk of shock and fatal arrythmia

Hypertrophic cardiomyopathy in infants of diabetic mothers	
Pathogenesis	• DM → fetal hyperglycemia & hyperinsulinemia → Glycogen & fat deposition in interventricular septum → dynamic LVOT obstruction
Clinical findings	• Often asymptomatic/ respiratory distress and/or hypotension • Systolic ejection murmur, cardiomegaly, thickened IVS
Treatment	• Intravenous fluids & beta blockers to increase LV blood volume
Prognosis	• Spontaneous regression by age 1

Other neonatal consequences of maternal diabetes may be present, such as macrosomia (leading to shoulder dystocia/clavicular fx) and plethora (due to polycythemia).

- Beta blockers reduce heart rate (**negative chronotropic effect**), which increases LV diastolic filling time, as well as end-diastolic volume (ie, preload).
- In addition, the **negative inotropic effect** of beta blockers decreases the pressure gradient between the LVOT and aorta, further reducing dynamic LVOT obstruction.

Treatment is rarely required for more than a few weeks because insulin levels quickly normalize, leading to reduced septal thickening and **self-resolution** of HCM.

(Choice A) Dobutamine is primarily a beta-1 agonist with positive chronotropic and inotropic effects. It decreases LV blood volume and increases the pressure gradient between the LVOT and the aorta, likely worsening LVOT obstruction in HCM. Furosemide (diuretic) and nitroprusside (venous and arterial dilator) decrease preload and LV blood volume. These medications are expected to worsen LVOT obstruction in HCM.

(Choice F) Hypoglycemia is often seen in preterm infants or infants of diabetic mothers.

HCM: AD mutation in cardiac sarcomere proteins (myosin-binding protein C, beta-myosin heavy chain), **asymmetric hypertrophy of the IVS** leading to **dynamic LVOT obstruction**. Symptoms: dyspnea, fatigue, chest pain, light-headedness, and syncope; **sudden cardiac arrest** can result from reduced cardiac output or ventricular arrhythmia.

Midsystolic or crescendo-decrescendo systolic murmur best heard at the left sternal border or cardiac apex. *Standing valsalva (strain phase) nitroglycerin* ↑ murmur, and decreased by *squatting(*↑preload and afterload) *leg raise handgrip*.

(Choice D) A sustained handgrip increases afterload to increase LV blood volume and reduce the intensity of the **HCM murmur**. Increased afterload has the same reducing effect on the murmur of **aortic stenosis**, but for a different reason; the pressure gradient across the aortic valve is reduced.

Pericardial effusion is common a **few weeks after cardiac surgery** due to **immune-mediated** inflammatory damage to the pericardium (ie, **postpericardiotomy syndrome**).

Small = asymptomatic. Larger effusions: Infants: decreased appetite and vomiting. Older children: pericardial friction rub and pleuritic chest pain. Fever may be present due to inflammation, and **tachypnea** and **tachycardia** occur in response to **decreased cardiac output**. **Distant, "muffled" heart sounds** on examination.

(Choice C) Postoperative mediastinitis can complicate heart surgery and present with fever and tachycardia. Sternal wound erythema and drainage; mediastinal widening, although this finding is often absent in cases of postoperative mediastinitis (compared to mediastinitis due to esophageal rupture or odontogenic/retropharyngeal infection spread).

Kawasaki disease (KD) is a vasculitis characterized by **≥5 days** of **fever** plus ≥4 of the following findings: (can cause myocarditis in the acute stage)

- **Conjunctivitis**: bilateral, nonexudative, limbus sparing
- **Oral mucosal changes**: erythema, fissured lips, strawberry tongue
- **Polymorphous rash**: often begins in perineal area
- **Distal extremity changes**: erythema, edema, desquamation of the hands and feet/ periungual desquamation
- **Cervical lymphadenopathy**: >1.5-cm node

Laboratory findings	• ↑ Platelets & WBCs; ↓ hemoglobin. ↑ CRP. ↑ AST & ALT. Sterile pyuria. Tx: **IVIG** (within 10 days of symptoms onset) & **aspirin**
Complications	• Coronary A aneurysms (echo for all pts). Ventricular dysfunction (lymphocytic myocarditis). Pericardial effusion → tamponade

Irritability, is also a common feature. Nonspecific gastrointestinal (eg, vomiting, diarrhea) or respiratory (eg, cough, congestion) symptoms may precede the development of mucocutaneous findings. Prolonged fevers (>14 days), delayed treatment with (IVIG), and **age <1** increase the risk for cardiac complications. **Most aneurysms regress over time**, and ventricular dysfunction typically improves with IVIG therapy.

Causes of QT-interval prolongation (Anti ABCDEF+NO) + hypo K Mg Ca + congenital long QT syndrome (syncope and SCD during exercise)**… can lead to torsade de pointes**

Delayed repolarization manifests as a prolonged QT interval (eg, >460 ms) on ECG.

Management: nonselective beta blockers (**propranolol** and **nadolol**). ICD placement or left cardiac sympathetic denervation are also appropriate in some high-risk patients.

Chapter 2: Respiratory

Vascular ring	
Pathophysiology	• MCC: Congenital aortic arch malformation (double aortic arch) • Great vessels encircle & compress trachea &/or esophagus (level of T3 -T4). Associated cardiac defects (eg, VSD) in 50% of patients
Clinical features	• Respiratory symptoms:

	○ Biphasic stridor (improves with neck extension) ○ Recurrent respiratory tract infections • Esophageal symptoms (persistent solid food dysphagia, vomiting, difficulty breathing) -> poor weight gain
Imaging*	• Chest x-ray: right aortic arch, tracheal narrowing • Esophagram: posterior esophageal indentation • CT/MR angiography of the chest (diagnostic)
Treatment	• Surgical correction
*Choice of prediagnostic imaging studies varies depending on presenting symptoms (ie, respiratory vs esophageal).	

Diagnosis can be made with CT scan to delineate the precise anatomy forming the vascular ring and evaluate any associated tracheal abnormalities. Due to possible concurrent cardiac and airway abnormalities, all patients require direct laryngoscopy, bronchoscopy, and echocardiogram. Treatment is surgical division of the structures creating the ring.

Most **CF** cases are identified on **newborn screening**; however, because screening does not detect less common *CFTR* mutations, a minority of cases are not identified until the patient is symptomatic. Detection of an elevated sweat chloride concentration is diagnostic.

Up to one-third have **nasal polyps**, benign outgrowths of inflamed mucosa that form as a result of chronic infection and appear as shiny, grey masses. Polyps obstruct the nasal passages and exacerbate sinusitis; management includes intranasal glucocorticoids (for symptomatic relief) and, in some cases, surgical resection.

Neonates with CF can have meconium plugs in the **gastrointestinal** tract, but chronic sinopulmonary infections and bronchiolar obstruction do not occur in the newborn period.

Imaging reveals **sinus cavity opacification** due to tenacious mucus in almost all CF patients **as early as age 1**, and patients often require **surgical debridement.**

Pulmonary disease is the primary cause of morbidity and mortality in patients with **cystic fibrosis** (CF). Defective mucociliary clearance = **recurrent sinopulmonary infections**.

In **young children** with CF, the **most common** pathogenic organism is ***Staphylococcus aureus***, especially in the setting of concurrent **influenza** infection. Severe pneumonia, frequent hospitalizations, or recurrent skin infections = MRSA = IV Vancomycin (**severe**), oral doxycycline (mild, **only in pts >8 years old** due to teeth discoloration)

Pseudomonas aeruginosa is the most common pathogen in older patients with CF. It should also be covered empirically in young children, with cefepime; fourth-generation cephalosporin, however, it has no activity against MRSA.

(Choice D) Azithromycin is not typically used in acute CF exacerbations, but due to its anti-inflammatory effects, long-term maintenance therapy with azithromycin has been shown to slow lung function decline in patients with CF.

Apnea of prematurity (Virtually affects all preterm infants born <28 weeks)	
Pathophysiology	• Immature central respiratory center in preterm infants
Clinical features	• Intermittent apnea (cessation of respiration for >20 seconds) starting at age 2-3 days (but recurrs on subsequent months) ± Associated bradycardia & desaturation. Well-appearing between episodes
Diagnosis	• Clinical ± Evaluation for (eg, anemia, infection, IVH)
Treatment/prognosis	• Caffeine. Noninvasive ventilation (high flow nasal canula, CPAP). Resolves by expected due date

Chest x-ray is often performed to exclude pulmonary pathology during the evaluation for alternate causes. AoP does not cause abnormalities on x-ray, but concomitant lung disease caused by prematurity may be seen, such as bilateral granular opacities from resolving respiratory distress syndrome. **Causes CP if left untreated.** Caffeine is a **methylxanthine** that chemically stimulates the respiratory drive and is given until the respiratory centers mature, which typically occurs by the expected due date.

Transient tachypnea of the newborn (1st few hours after birth)	
Pathophysiology	• Retained fetal lung fluid
Risk factors	• Cesarean birth - Prematurity - Maternal diabetes
Clinical findings	• Tachypnea, increased work of breathing/ Clear breath sounds (fluids are in interstitial space rather than alveoli) • Chest x-ray: hyperinflation, fluid in fissures
Management	• Supportive (eg, oxygen, nutrition)/ Self-resolution in 1-3 days as passive fluid reabsorption is complete

Delayed resorption and clearance of pulmonary fluid: Normally, alveolar liquid produced during lung development is later absorbed by mature fetal lungs into the interstitial space and the pulmonary vasculature. This resorption is driven by increased catecholamine signals in late gestation and markedly increases during labor.

(Choice C) Chemical irritation and airway obstruction occur with meconium aspiration syndrome, which presents at delivery with respiratory distress {tachypnea, increased WOB, cyanosis}, meconium-stained amniotic fluid. CXR: Hyperinflated lungs and streaky, linear densities. (full term infant, resolves within the 1st week of life).

(Choice F) Pulmonary sequestration: nonfunctional segment of abnormal lung tissue, respiratory distress in newborns, dense mass on CXR (not bilateral infiltrates).

Congenital diaphragmatic hernia	
Pathogenesis	• Incomplete fusion of pleuroperitoneal folds. Herniation of bowel into chest. Pulmonary hypoplasia & hypertension
Clinical features	• Respiratory distress (within hrs of birth). Absent breath sounds ipsilateral to defect. Concave abdomen; barrel-shaped chest
Diagnosis	• Chest x-ray: intrathoracic bowel loops, displaced cardiac silhouette
Management	• Endotracheal intubation (1st step). Gastric decompression (eg, nasogastric tube). Surgical correction

- CDH may be diagnosed on prenatal ultrasound or shortly after birth.
- Mechanical ventilation should be used **cautiously** with limited pressures to avoid barotrauma to the hypoplastic lungs.
- Bag-and-mask ventilation is **contraindicated** because it pumps air into the gastrointestinal tract, leading to further lung compression.

Bronchopulmonary dysplasia	
Pathogenesis	• Premature arrest of pulmonary development (alveolarization) • Alveolar hypoplasia with ↓ septation. Impaired vasculogenesis
Clinical diagnosis	• ***Premature*** *infant with continued supplemental oxygen requirement* ***≥28 days from birth**** [chronic hypoxia]
Chest x-ray	• Mild: diffuse hazy infiltrates, low/normal lung volumes

	• Severe: fibrocystic changes, hyperinflation • Coarse lung markings with cystic changes
Treatment	• Supportive (eg, oxygen, nutrition, fluid restriction/diuretics), improve over 2-4 months [new alveoli develop]
Complications	• HTN, PAH, Recurrent respiratory infections
*Some definitions include an oxygen requirement at 36 weeks postmenstrual age (ie, gestational + chronologic age).	

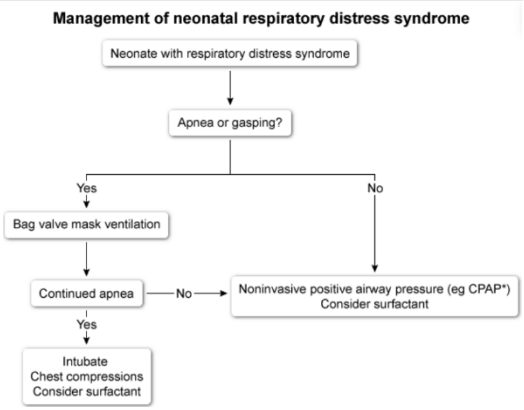
- ❖ Dexamethasone + albuterol for OSA caused by severe bronchopulmonary dysplasia.
- ❖ Air dissecting from the alveoli into the interstitial space: pulmonary interstitial emphysema = worsening hypoxia in premature infants on MV (few days after birth).

Common causes of neonatal respiratory distress			
Diagnosis	**Transient tachypnea of the newborn**	**Respiratory distress syndrome**	**Persistent pulmonary hypertension**
Pathophysiology	• Inadequate alveolar fluid clearance at birth	• Surfactant deficiency • Alveolar collapse & diffuse atelectasis	• High pulmonary vascular resistance • Right-to-left shunt
Clinical features	• Tachypnea shortly after birth • Resolves by day 2 of life	• Prematurity • Severe respiratory distress & cyanosis	• Tachypnea & severe cyanosis
Chest x-ray	• Bilateral, perihilar linear streaking + fluid within fissures	• Diffuse, ground-glass appearance with low lung volumes • Air bronchograms	• Clear lungs with decreased pulmonary vascularity

Neonatal respiratory distress syndrome (surfactant production at 20 weeks gestation)	
Pathophysiology	• Surfactant deficiency in premature infants. ↑ Alveolar surface tension → diffuse atelectasis. Ventilation/perfusion mismatch causes **right-to-left intrapulmonary shunting**
Clinical features	• Tachypnea, grunting, nasal flaring, retractions, hypoxia, cyanosis, immediately (mins-hrs) after birth + ↓ breath sounds
Chest x-ray	• Diffuse, ground-glass appearance/ Air bronchograms/ ↓ Lung V
Complications	• Bronchopulmonary dysplasia (chronic lung disease) , PPHTN

Tachypnea. **Grunting** (to increase end-expiratory pressure and maintain alveolar volume). **Nasal flaring** (to decrease airway resistance). **Retractions** (due to high intrathoracic pressure). **Hypoxia and cyanosis** (due to atelectasis and ventilation/perfusion mismatch)

Hypoxia also leads to vasoconstriction of pulmonary vasculature, causing ↑ PVR and right-to-left shunting across the ductus arteriosus [DA] or foramen ovale, further worsening cyanosis.



Administration of maternal antenatal glucocorticoids, which stimulate fetal surfactant synthesis, reduces but may not eliminate the risk for RDS. Endogenous surfactant production after birth typically results in symptom improvement within a week.

Steriods after birth have no benefit. The greatest risk factor for RDS is prematurity.

GDM: Maternal hyperglycemia → fetal hyperglycemia → fetal hyperinsulinism → inhibit cortisol → **delayed maturation** of phosphatidylcholine and phosphatidylglycerol.

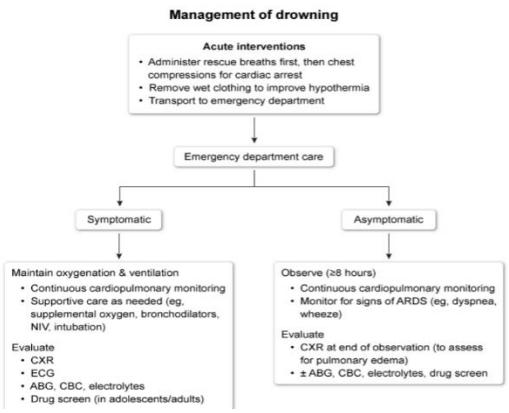
Features of primary ciliary dyskinesia (AR) (no pancreatic insufficiency vs CF)	
Pathophysiology	• Mutations in ciliary dynein arms lead to absent or dysmotile cilia & poor mucociliary clearance
Clinical manifestations	• Recurrent sinopulmonary infections/ Bronchiectasis • ± Situs inversus (Kartagener syndrome)

Diagnosis	• Low nasal NO + Bronchoscopy & electron microscopic visualization of ciliary abnormalities + Genetic testing

(Choice E) Healthy young children who attend day care or have older siblings may experience up to 12 respiratory infections per year. Mostly **viral** and do not require Abs.

Drowning: Respiratory impairment due to submersion in **liquid**	
Risk factors	• Children age <5 & males age 15-25 • Inability to swim &/or inadequate supervision • Concomitant drug/alcohol use
Complications	• Acute respiratory distress syndrome (MC) • Cerebral edema (High ICP) (more common with first 3 poor prognostic indicators) • Arrhythmia (abnormal vitals and PEx) • RARE complications: ATN in hypovolemic shock/ Gastric perforation in prolonged resuscitation + a full, distended stomach (from swallowing liquid) + Abdominal pain and tenderness
Poor prognostic indicators	• Submersion time >5 min • Delay in initiation of cardiopulmonary resuscitation • Prolonged resuscitative efforts • Age >14/ Arterial blood pH <7.1 (metabolic acidosis)

Signs and symptoms of ARDS (eg, dyspnea, wheezing, crackles, cyanosis) typically begin within the **first 8 hours of drowning**, and respiratory insufficiency can **progress insidiously** over 72 hours after the initial event. All patients with a nonfatal drowning event, even with normal vital signs and examination findings, should be admitted to the hospital for **cardiopulmonary monitoring** for at least 8 hours. Then x-ray.



Pneumothorax (Risk factors = prematurity (RDS) + MAS (MC in post-term infants))

	Spontaneous pneumothorax	Tension pneumothorax
Associated features	• Primary: tall thin, young men. 2ndary: COPD CF	• Life-threatening. Often due to trauma or **mechanical ventilation**
Signs & symptoms	• Chest pain, dyspnea • ↓Breath sounds, ↓chest movement • Hyperresonant to percussion	Same as spontaneous plus: • Hemodynamic instability • Tracheal deviation away from affected side
Imaging	• Visceral pleural line • Absent lung markings beyond pleural edge	Same as spontaneous plus: • Contralateral mediastinal shift • Ipsilateral hemidiaphragm flattening
Management	• Small (≤2 cm): observation & oxygen administration • Large & stable: needle aspiration or chest tube	• **Urgent** needle decompression or chest tube placement

Some pts increased brightness on transillumination + displacement of the heart to the rt.

(Choice B) Positive end-expiratory pressure (PEEP) should be avoided.

(Choice C) Chest physiotherapy can be helpful in treating atelectasis due to RDS or meconium plugging. In contrast to pneumothorax, atelectasis does not transilluminate.

Spontaneous pneumomediastinum	
Risk factors	• Asthma exacerbation/Respiratory infx/ Tall, thin, adolescent boy
Clinical features	• Acute chest pain, SOB, cough/ Subcutaneous emphysema • Hamman sign (crunching sound/crepitus over heart)
Diagnosis	• Mediastinal gas on chest x-ray
Treatment	• Rest, analgesics, avoid valsalva maneuvers (resolve in days-wks)

(Choice D) PFTs are contraindicated with SPM because forced expiration increases pulmonary pressure, worsening the air leak.

(Choice E) Stress-induced (takotsubo) cardiomyopathy, a reversible cardiomyopathy likely caused by toxicity from surging catecholamines, can occur after severe physical stress. However, it most commonly occurs in postmenopausal women: chest pain, ECG findings of ischemia, and/or elevated serum troponins.

(Choice F) Uncontrolled pain → HTN, tachycardia, ↑ motor activity in an intubated patient.

Chronic dyspnea in sickle cell disease		
Cause	**Symptoms**	**Diagnostic findings**
Asthma	• Intermittent/chronic wheezing • Worse at night/exercise or URI	• PFT showing reversible airway obstruction
Pulmonary hypertension	• Exertional dyspnea + Rt HF	• Echo first step (Tricuspid regurgitation) + ↑ PAP on catheter + Large PA on xray
Pulmonary fibrosis	• Progressive exertional dyspnea	• Increased reticular markings on xray/ CT+ Transbroncial biopsy confirm the Dx

Features of respiratory failure in acute asthma	
Features	• Absent wheezing/ Accessory muscle use/ Altered mental status
Laboratory findings	• ABG: R. acidosis (unlike mild = R. alkalosis) • Lactate: transient ↑ due to muscle WOB (type A) &/or β-agonist–induced (type B) • K^+: transient ↓ due to β-agonist &/or respiratory alkalosis
Management	• Nebulized albuterol & ipratropium, IV corticosteroids ± IV magnesium • Short trial (<2 hr) of NIPPV, if failed, intubation & invasive MV – if failed tracheostomy

- Children age ≤5 typically cannot perform PFT.
- OTC cough meds are not recommended in young children.

Pediatric obstructive sleep apnea (Tx: Tonsillectomy & adenoidectomy)	
Pathophysiology	• Adenotonsillar hypertrophy (dysphagia, NOT aspiration)

Clinical manifestations	• Night symptoms
	  ○ Loud snoring, pauses in breathing, gasping
	  ○ Enuresis (Elevated BNP), parasomnias (eg, sleepwalking, sleep terrors)
	• Day symptoms
	  ○ Inappropriate naps or falling asleep during school
	  ○ Irritability, inattention, learning problems, behavioral problems/ Mouth breathing, nasal speech

Complications	• Poor growth (ie, failure to thrive)/ Poor school performance • HTN, tachycardia, RVH LVH/ PAH (more in adults)

Conditions mimicking attention deficit hyperactivity disorder		
Diagnosis	**Historical clues**	**Initial evaluation**
Learning disability	• Symptoms predominantly in academic setting	• Neuropsychological testing
Hearing impairment	• Delayed speech	• Audiology evaluation
Mental health conditions (anxiety, depression, ODD)	• Excessive worrying (anxiety), pervasive sadness (depression), defiance (ODD)	• Interview & standardized screening questionnaires
Obstructive sleep apnea	• Snoring, daytime sleepiness/Obesity	• Sleep study
Absence seizure	• Discrete episodes/ unresponsive to stimuli/ Automatisms	• EEG

OSA: In some children, presentation may be limited to **behavioral problems** and symptoms overlapping with ADHD because excessive sleepiness can cause patients to be irritable (eg, fighting with sister), easily frustrated (eg, throwing homework), and impulsive.

Foreign body aspiration [endobronchial obstruction → retention of secretions)	

Clinical features	• ± History of choking event with sudden-onset cough
	• Symptoms depend on location
	  ○ Trachea/main bronchus: acute respiratory distress, cyanosis, stridor (trachea), hemoptysis (bronchial)
	  ○ Lower airway: chronic/recurrent cough

Examination	• Focal wheeze and diminished breath sounds
X-ray findings **(30% normal)**	• Hyperinflation of affected side ± contralateral mediastinal shift • Atelectasis if complete obstruction ± Foreign body
Complications	• Recurrent pneumonia + Bronchiectasis
Management	• Bronchoscopic removal

Foreign body in the upper airway (eg, larynx): stridor + life-threatening, acute respiratory distress. A chronic foreign body in the lower airways (eg, bronchi): recurrent pneumonia that improves symptomatically but still has persistent radiographic changes.

Bronchiolitis (tx: supportive: hydration, saline nasal drops, nasal bulb suction)	
Epidemiology	• Age <2 years. RSV most common cause, rarely Inf, Pinf, Rhinovirus
Presentation **(peak on 5-7d)**	• Antecedent nasal congestion/discharge & cough -> Wheezing, crackles, respiratory distress (tachypnea, retractions, nasal flaring)
Complications	• Apnea (age <2 months)/ RF/AOM/ recurrent wheezes in childhood
Prevention (in children <2 yrs)	• Palivizumab for selected infants:<29 weeks gestation. Chronic lung disease of prematurity. Hemodynamically significant CHD

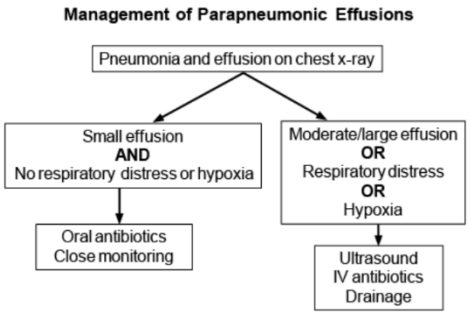
Most patients can be treated as outpatients with **close follow-up** to ensure improvement of symptoms. Premature or young (age <2 months), may require hospitalization for dehydration or severe respiratory symptoms → respiratory support, such as O2 and CPAP.

Parameter	Uncomplicated parapneumonic effusion	Complicated parapneumonic effusion	Empyema
Etiology	Inflammatory fluid from pneumonia → pleural space	Bacterial invasion into pleural fluid	Bacterial colonization → purulent fluid
Pleural fluid analysis	pH = 7.20, ↓/normal glucose, LDH ratio >0.6	pH <7.20, ↓ glucose, LDH ratio >0.6	pH <7.20, ↓ glucose, LDH ratio >0.6
Pleural fluid Gram stain & culture	Negative	Positive or negative	Positive

- Empyema: **pH <7.2** (due to acid production by bacteria), **low glucose** (<40-60 mg/dL due to bacterial/neutrophil utilization), neutrophil-predominant leukocyte counts >50,000/mm^3, and **significantly elevated LDH** (>1,000 IU/L). Tx: broad-spectrum intravenous antibiotics and chest tube drainage.
- Chylothorax typically shows elevated triglyceride and lymphocytes
- Malignant effusion: exudative, ↑ LDH, ↓ glucose; ↓ leukocyte count (<5,000/mm^3).
- *TB* chronic, low glucose and pH, low leukocytes & lymphocytic predominance.

(Choice A) Chest physiotherapy is not recommended in the treatment of pneumonia or pleural effusions as it is not shown to shorten hospital stay and may prolong cough.

(Choice E) Diuretics can be used to decrease effusions in the presence of transudative pleural effusions. Diuretics are not indicated in the treatment of parapneumonic effusions.



Pediatric empyema (S pneumoniae, MRSA/ fever dyspnea pleuritic chest pain, no improvement with routine pneumonia tx, leukocytosis, thrombocytosis)	
Etiology	• Bacterial invasion of pleural space- fibrinopurulent consolidation
Management	• Supportive/ Empiric vancomycin + (certriaxone OR clindamycin) + drainage (ie, chest tube or VATS)/ Intrapleural fibrinolytics (tPA)

- The **thymus** is normal on xray **age <3**. It atrophies after puberty. Infants age <1 normally have a transverse cardiothoracic ratio of ≤60%, vs children age ≥1 and adults ≤50%.

Allergic bronchopulmonary aspergillosis	
Pathogenesis	• Asthma CF. Th2 → allergic inflammation

Clinical features & diagnosis	• Difficult-to-control asthma, thick sputum
	• Chest imaging: fleeting infiltrates, bronchiectasis, mucus plugging (eg, intrabronchial exudates appearing as tubular, finger-in-glove radiodensities)
	• *Aspergillus* sensitization:
	  ○ Elevated serum IgE (usually >1,000 IU/mL)
	  ○ Positive *Aspergillus* skin test &/or IgE
	  ○ Suggestive: eosinophilia, positive *Aspergillus* IgG

Treatment	• Systemic glucocorticoids → ↓ allergic inflammation • Antifungal drugs (eg, voriconazole) → ↓ spore burden • Treatment of underlying asthma (eg, bronchodilators)

ABPA must be suspected if there is an unexplained lung function decline **despite an appropriate antibiotic course** (>1 wk).

(Choice D) Simple (nondisseminated) strongyloidiasis: eosinophilic pulmonary disease (infiltrates with peripheral eosinophilia) and ↑ IgE. In rural tropical and subtropical areas; in addition, because larvae penetrate the skin and are eventually swallowed, patients usually also have dermatologic (roaming urticaria) and GI (obstruction, ileus) manifestations.

(Choice E) Patients with CF bronchiectasis can be colonized and infected with nontuberculous mycobacteria (NTM): chronic (months to years) and presents with nodular, cavitary, and, eventually, fibrotic lung disease.

Croup (laryngotracheitis): (most improve within a few days)	
Pathogenesis	• Parainfluenza viral infection of the larynx & trachea
Epidemiology	• Age 6 months to 3 years. Fall/early winter
Features	• *Inspiratory* stridor /Barking cough /Hoarseness
Treatment	• Mild (no stridor at rest): humidified air ± corticosteroids • Moderate/severe (stridor at rest): steroids + nebulized epinephrine
Prevention	• Handwashing /Decontamination of surfaces /Proper ventilation

- After epinephrine administration, patients are observed for 4 hours because symptoms can recur. Patients needing multiple doses of nebulized epinephrine typically require hospital admission. **Give O2 if sat <92%.**
- Intubation with mechanical ventilation is reserved for those who have failed treatment with corticosteroids and nebulized epinephrine and/or have signs of impending respiratory failure (eg, altered mental status, poor respiratory effort).
- X-ray may reveal subglottic narrowing, or steeple sign.

Epiglottitis: Unimmunized. Stridor, R distress, ill appearing, high fever, drooling, dysphagia dysphonia, hot potato voice, tripod position. **Mx: secure airway THEN vancomycin + ceftriaxone. Croup only 6mo to 3yr, epiglottitis can be >3yr and NO barking cough.**

Initial management is to secure the airway due to the risk of complete airway obstruction in epiglottitis. The patient should be kept calm, and aggravating interventions (eg, detailed oropharyngeal examination) should be minimized because agitation may cause laryngospasm. In children with respiratory distress (stridor, retractions, and cyanosis), **endotracheal intubation** should be performed in a controlled setting (eg, emergency department, operating room) with trained personnel (eg, anesthesiologist) available due to the risk of obstruction. Patients may require a surgical airway (eg, tracheotomy, needle cricothyrotomy) if attempts at intubation fail.

Epiglottitis can cause visible epiglottal enlargement on lateral neck films ("thumb sign").

Chapter 3: Infectious Disease

Acute rheumatic fever (Peak incidence: age 5-15. Twice as common in girls)		
Clinical features	**Major**	• **JONES** [EM: annular nonblanching rash]
	Minor	• Fever Arthralgias Elevated ESR/CRP Prolonged PR interval
Late sequelae	Mitral regurgitation/stenosis [can be decades later]	
Prevention	Penicillin for **group A streptococcal** (*S pyogenes*) pharyngitis	

Diagnosis is made if the patient has 2 major Jones criteria, 1 major and 2 minor criteria, or if either Sydenham chorea or carditis is present.

(Choice A) Coxsackievirus B = MCC of pericarditis and myocarditis.

Sydenham chorea (molecular mimicry between anti-GAS Abs & neuronal antigens in basal ganglia. MC girls age 5-15 and can present months after initial GAS infection)	
Clinical features	• Bilateral progressive involuntary, jerky movements (worse while awake & with action, disappear during sleep). Hypotonia. Emotional lability [irritability, inappropriate laughter], obsessive-compulsive behaviors [frequent hand cleaning]. ± Symptoms of ARF
Evaluation	• Throat culture, ASO & anti-DNAse B titers. Echo, ECG, ESR CRP
Treatment	• Chronic penicillin G. Symptomatic: (antidopaminergics [haloperidol])
Prognosis	• Spontaneous remission within months. Recurrence common. ↑ Risk of rheumatic heart disease

Chorea: jerky, irregular, involuntary contractions of the limbs and face that <u>cannot be suppressed</u>. **Other findings:** Intermittently weakened hand grip (ie, **milkmaid grip**). Tics (eg, repetitive throat clearing that cannot be supressed, vs tics in tourette syndrome [decreased caudate nucleus volume] which can be suppressed).

Wilson disease + huntington chorea onset is gradual vs **sydenham chorea (sudden)**

Antibiotic prophylaxis for secondary prevention of rheumatic fever (JONES)

Severity	Duration of therapy following last attack
Uncomplicated rheumatic fever	5 years or until age 21*
With carditis but no valvular disease	10 years or until age 21*
With carditis & valvular disease	10 years or until age 40*

*Whichever duration is longer. Intramuscular penicillin G benzathine every 3-4 weeks is preferred.

Neonatal group B *Streptococcus* infection	
Pathogenesis	• <u>Most common cause of neonatal sepsis, pneumonia & meningitis</u> • Early (age <1 week) = maternal genital tract (sepsis meningitis pneumonia). Late (age >1 week) = exposure to colonized contacts (IAP no benefit) (sepsis meningitis cellulitis, age 4-5 weeks)
Clinical findings	• Sepsis, pneumonia, meningitis: ○ fever, hypothermia. Irritability, lethargy, poor feeding. Tachycardia, hypotension. Respiratory distress, jaundice • Focal infection (late onset): Septic arthritis + Cellulitis
Prevention	• Intrapartum penicillin at 36 weeks

Empiric treatment of pneumonia in a newborn is **ampicillin + gentamicin**. If **GBS** is isolated from the blood, coverage can be narrowed to **penicillin G** alone.

Group B *Streptococcus* pneumonia presents with respiratory distress shortly after birth, x-ray shows diffuse alveolar infiltrates and possible pleural effusions.

Maternal chorioamnionitis → Aspiration of infected amniotic fluid → bacterial pneumonia → Respiratory distress + crackles and an infiltrate on chest x-ray

Neonatal sepsis (GBS/ Ecoli/ ***L monocytogenes*** **(age <7 days)**)	
Evaluation	• CRP, ANC, procalcitonin. Blood, urine, CSF cultures*
Treatment	• **Parenteral** antibiotics (ampicillin & gentamicin) VS Vancomycin + Ceftriaxone in children > 1 month
* **Limited evaluation** (without CSF studies) & outpatient management may be considered in well-appearing, febrile neonates age >21 days.	

Neonates with meningitis do not have nuchal rigidity or positive Kernig and Brudzinski signs. Instead, neonates are **irritable**, **lethargic**, **hypotonic** (eg, weak suck, decreased muscle tone), full fontanelle. Difficult to console and feeding poorly (fewer wet diapers). **Jaundice** (due to sepsis-associated cholestasis), respiratory distress, seizure.

Antibiotic administration before **obtaining** cultures should be avoided when possible because antibiotics can sterilize cultures and make definitive diagnosis difficult.

(Choice B) Cerebral herniation due to infection does not occur in neonates due to their open fontanelles that relieve ICP. **No need for CT before LP.**

Acute otitis media (*S pneumoniae/* Nontypeable *H influenzae/ M catarrhalis*)	
Risk factors	• **Age** (6-18 months, most important RF; narrower straighter eustachian tubes that drain poorly)/ **Lack of breastfeeding** • **Day care** attendance/ **Smoke** exposure (↑bacterial colonization of the nasopharynx)
Clinical features	• Bulging TM, Middle ear effusion **plus** TM inflammation (eg, fever, otalgia, erythema) Infants: difficulty sleeping, poor feeding
Treatment (ORAL)	• Initial: amoxicillin. **2nd-line or if pt took amoxicillin last 30 days**: **amoxicillin-clavulanate** • **SEVERE** penicillin allergy: clindamycin or azithromycin

Complications: **TM perforation** (otorrhea, decreased otalgia, conductive hearing loss, most heal within days)/ **hearing loss/ meningitis/ mastoiditis**. Observation can be considered for afebrile children age ≥6 months with unilateral disease and no/mild pain.

Otoscopy shows a bulging TM and/or a middle ear effusion (poor TM mobility on insufflation) plus erythema(inflammation) or pale, yellow, opaque TM (**purulent effusion**).

Cephalosporins (eg, cefdinir) are **avoided** in patients with **severe penicillin allergy** due to the potential risk for beta-lactam cross-reactivity but may be prescribed for those with a history of mild penicillin reaction (without anaphylaxis, bronchospasm, angioedema, SJS).

(Choice E) Pressure changes during ascent and descent while flying → barotrauma (hemotympanum and TM perforation) → ear pain, ear discharge, hearing loss, and tinnitus.

B pertussis (G – coccobacilli, more contagious in catarrhal stage)	
Clinical phases	• Catarrhal [absent in infants] (weeks 1-2): mild cough, rhinitis • Paroxysmal (weeks 2-8): severe coughing spells ± Inspiratory whoop, posttussive emesis, gagging cyanosis seizures apnea RF (infants); posttussive syncope (older patients)/ subconjunctival hemorrhage • Convalescent (weeks 8+): symptoms resolve gradually
Diagnosis **(start Abs before testing)**	• Clinical: cough + **paroxysmal** symptoms &/or pertussis exposure ○ Supportive findings: low-grade/no fever, leukocytosis with lymphocytosis, normal/nonspecific chest x-ray • Confirmatory: pertussis **PCR (sensitive) & culture (specific)**

Pertussis postexposure prophylaxis (highly contagious, respiratory droplets)	
Indications (regardless of vaccination history)	• Close contact (eg, household members, direct contact with secretions) with symptomatic patient **in the last 21 days** • High-risk patients, even with limited exposure (eg, pregnant, infant, immunodeficient)
Treatment	• Age <1 month: azithromycin • Age ≥1 month: azithromycin, clarithromycin, or erythromycin

First-line **treatment for pertussis infection** is with a macrolide antibiotic (eg, azithromycin, clarithromycin) within the first 3 weeks of illness. Antibiotic administration

during the catarrhal stage can shorten the course of illness and decrease transmission risk, whereas treatment in the early paroxysmal stage reduces the risk for transmission only.

DTaP vaccine at 2, 4, 6, and 15-18 months and at 4-6 years, with booster doses in adolescence and adulthood. **Contraindications**: Anaphylaxis. Encephalopathy (coma, decreased level of consiousness, prolonged seizures). Unstable neurological disorders (infantile spasms, uncontrolled epilepsy). **Within 1 week** of adminstering the DTaP vaccine. As a result, diphtheria and tetanus toxoids should be administered **without aP.**

Seizure, triggered by **fever** or by the pertussis **vaccine component**, is **rare** and is typically short (**<5 minutes**) and **self-limited**. Patients with a family history of febrile seizures or epilepsy **may be at increased risk**. However, neither personal nor family history of seizures is a contraindication to immunization. Specifically, uncomplicated seizure following vaccine administration is **not a contraindication** to future vaccination.

Cervical lymphadenitis in children (MC <5 years old, submandibular)		
	Pathogen	**Key clinical findings**
Unilateral	*S aureus, S pyogenes*	• **Acute**/MC/Suppuration (fluctuance) common
	Anaerobic bacteria (eg, *Prevotella* spp)	• **Acute**/ Hx of periodontal disease or dental caries
	Francisella tularensis	• **Acute**/ History of contact with infected rabbit
	Mycobacterium avium	• **Chronic**/ Nontender, violaceous
	Bartonella henselae	• **Chronic**/ Papule at site of cat scratch or bite
Bilateral	Viral	• **Acute** (eg, adenovirus) associated with self-limited URI. **Subacute/chronic** (eg, EBV, CMV) associated with mononucleosis symptoms

Unilateral: Empiric **clindamycin**/amoxicillin-clavulanate + I&D for abscess. Observe if afebrile + minimal tenderness/LN enlargement. **Third-generation cephalosporins** may be used to treat infection caused by ***S pyogenes*** but not *S aureus* or anaerobes.

Lymph node features		
	Reassuring (observation)	**Worrisome**
Palpation	• Soft Mobile	• Firm or hard Immobile

	• <2 cm (normal: <1 cm)	• >2 cm
Location	• Localized	• Generalized or supraclavicular
Fever + WL	• Absent	• Present

Antibiotic therapy is warranted for lymphadenitis: tender, enlarged, warm nodes.

Meningococcal meningitis (MC in young children and young adults) (Tx: ceftriaxone)	
Clinical features	• Nonspecific fever, headache, vomiting, myalgia, sore throat. **Within 12-24 hr**: petechiae/purpura, meningeal signs, AMS
Complications	• Shock - DIC - Adrenal hemorrhage - Purpura fulminans (less common complication of meningococcal meningitis, presents with gangrenous necrosis of purpuric lesions)
Prevention	• Droplet precautions/ Chemoprophylaxis* for close contacts
*Rifampin, ciprofloxacin, or ceftriaxone.	

DIC: oozing at an IV line site, GI hemorrhage, **purpura**; thrombocytopenia, elevated PT and PTT, and **low fibrinogen**. Management is supportive (eg, hemodynamic stabilization).

The mortality rate of N meningitidis is up to 15% even with antibiotic therapy.

(Choice A) Early disseminated Lyme disease can present with meningitis. Symptoms develop over many days (not hours), and shock is atypical. In addition, CSF testing shows a lymphocytic (not neutrophilic) predominance. **Tx = amoxicillin**

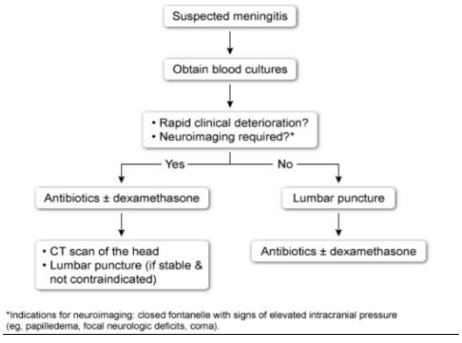
Neurosyphilis: pleocytosis with lymphocytic predominance, elevated protein, low glucose.

Listeria can cause subacute to chronic meningitis with a lymphocytic predominance, low glucose, and high protein in the CSF. Particularly in neonates, the elderly, or pregnant women. **Meningitis <1month old: GBS (MC), Ecoli, Listeria, HSV. Meningitis >1month old: S pneumoniae + Neisseria.**

Cerebrospinal fluid analysis

Diagnosis	WBC count (mm^3)	Glucose (mg/dL)	Protein (mg/dL)
Normal	0-5	40-70	<40
Bacterial meningitis	>1,000	<40	>250
Tuberculous meningitis	100-500	<45	100-500
Viral meningitis	10-500	40-70	<150
Guillain-Barré syndrome	0-5	40-70	45-1,000

Management of bacterial meningitis in children age >1 month



***Neisseria meningitidis* postexposure prophylaxis**	
Recommended populations (regardless of vaccination status)	❖ **Household** members ❖ **Roommates** or intimate contacts ❖ **Childcare** center workers ❖ Persons **directly exposed** to respiratory or oral secretions ❖ Person seated next to affected person for **≥8 hr (eg. flight)**
Prophylaxis	• Rifampin/Ceftriaxone/**Ciprofloxacin (adults only)**

N meningitidis has risk of **epidemic** spread, and most secondary cases develop within 10 days of the initial patient's diagnosis. Risk of transmission occurs from **7 days prior to symptom onset until 24 hours after initiating appropriate antibiotic therapy.**

Antibiotics **as soon as possible** after exposure, ideally within a day of diagnosis but up to 2 weeks. *N meningitides* vaccination prevents infection from **serogroups A, C, Y, and W-135**.

Adolescents should receive the **quadrivalent** meningococcal conjugate vaccine at **age 11-12 with a booster at age 16**. Meningococcal **serogroup B vaccine** at **age 16-18**.

Bacterial meningitis in children	
Clinical features	• Fever. Age <1: bulging fontanelle, irritability, poor feeding. Age >1: headache, vomiting, nuchal rigidity
Diagnosis	• Cerebrospinal fluid culture
Treatment	• Vancomycin + ceftriaxone (or cefotaxime) ± Dexamethasone
Complications	• Intellectual/behavioral disabilities, SNHL, CP, epilepsy

Risk factors for complications: younger age, prolonged seizures, high bacterial load at dx.

SNHL: MC after ***S pneumoniae*** meningitis, due to **inflammation** of the **cochlea**/labyrinth → Fibrosis & ossification (within wks/months) → permanent, profound hearing loss.

All patients with bacterial meningitis should undergo **audiologic testing** as soon as possible (ideally, before hospital discharge).

Tuberculous meningitis (2-6 mo after primary infx)	
Pathophysiology	• **Hematogenous** spread to subarachnoid space

Clinical manifestations	• Stage 1: several weeks of fever, headache, drowsiness • Stage 2: overt meningitis with lethargy, nuchal rigidity, vomiting, **cranial nerve palsies**, or other focal neurologic signs • Stage 3: coma, herniation, death
Diagnosis	• CSF: lymphocytic pleocytosis, ↑ protein, ↓ glucose • Positive acid-fast bacillus culture or PCR test from CSF • TST or IGRA may be falsely negative
Treatment	• Combination antimycobacterial therapy. Glucocorticoids to reduce CNS swelling

(Choice B) Encephalitis caused by arboviruses (eg, West Nile virus), gradual onset several days (not hrs) of prodromal symptoms (fatigue, headache, V) prior to severe encephalitis.

Viral meningitis: Supportive tx (antipyretics, pain control), self limiting within 1-2 weeks.

Acute bacterial rhinosinusitis (Amoxicillin ± clavulanate) (Acute is <12 weeks duration)	
Features	• Cough, nasal discharge. Fever. Face pain/headache
Diagnostic criteria **(1 of 3)**	• Persistent symptoms ≥10 days without improvement • Severe onset (fever ≥39 C [102.2 F] + drainage) ≥3 days • Worsening symptoms following initial improvement

- **Nontypeable *H influenzae*** (~40%-50%), ***S pneumoniae*** (~20%-25%), and ***M catarrhalis*** (~25%) are the MC implicated bacteria. Since the advent of the 13-valent pneumococcal vaccine, *S pneumoniae* infection has become less prevalent.
- Sinus tenderness is rare in young children but often found in adults with ABRS.
- The most common risk factor for ABRS is **viral upper respiratory infection** (URI); up to 10%. Thickened secretions and mucosal inflammation from the viral infection prevent sinus drainage and impair mucociliary clearance of contaminating bacteria, which can lead to secondary bacterial infection. **2nd most common risk factor is allergic rhinitis.**

***Mycoplasma* pneumonia** (droplets, dorms/military recruits, fall/winter)	
Clinical features	• Slow onset headache, malaise, fever, persistent dry cough • Pharyngitis (nonexudative). Macular/vesicular rash

Diagnostic findings	• Normal leukocyte count. Subclinical hemolytic anemia (cold agglutinins). Bilat reticulonodular or patchy infiltrates (like PCP)
Treatment	• Usually empiric **macrolide** or respiratory **fluoroquinolone**

(Choice E) *PCP:* treated with **TMP-SMX**, common in HIV, subacute presentation.

Community-acquired pneumonia in school-aged children		
	Lobar	**Bilateral**
Etiology	• *S pneumoniae*	• *Mycoplasma/ Chlamydia*.Viruses (rare)
Clinical features	• Abrupt onset of fever, cough, chest pain • ↑ WOB. Focal crackles	• Fever, malaise, headache, sore throat. Prolonged, gradually worsening cough. Patient can continue normal activities • Bilateral crackles, wheezing
Treatment	• Oral amoxicillin (outpatient) or IV ampicillin or ceftriaxone (if hospitalized)	• Macrolide (eg, azithromycin)

High-dose oral amoxicillin provides coverage for *S pneumoniae* = drug of choice for the outpatient treatment of community-acquired pneumonia in otherwise healthy children.

*Most children with uncomplicated **community-acquired pneumonia** typically improve within 48-72 hours of appropriate antibiotic therapy (Ampicllin Ceftriaxone for S.pneumoniae + Azithriomycin ONLY before culture results)*. If pneumonia & **fever persist** despite 48 hours of appropriate antibiotics → **repeat xray** to assess for complications: parapneumonic effusion, abscess, or necrotizing pneumonia.

(Choice E) Viral pneumonia in children <5yo = RSV. Older than 5 yrs = influenza.

Retropharyngeal abscess (between pharynx and prevertebral fascia)	
Epidemiology	• Polymicrobial (group A *Strep*, S *aureus* & anaerobes)
Symptoms	• Fever - Odynophagia/dysphagia - Neck pain - Drooling - muffled "hot potato" voice - trismus
Examination	• Retropharyngeal bulge/ Limited neck extension

Diagnosis	• Lateral neck x-ray (increased prevertebral thickening) • CT neck with contrast
Management	• Airway protection/ IV ab (eg, ampicillin-sulbactam, clindamycin) ± Surgical drainage

It is most often preceded by URTI, but can result from direct spread of local bacterial infection (eg, pharyngitis, tonsillitis). MC in children **age 2-4**. The incidence decreases after age 4 due to retropharyngeal LN regression and fewer viral URIs.

Osteomyelitis in children (hematogeneous spread to bone, fever, irritability, limp, bony tenderness, swelling. Investigations: WBC ESR CRP, blood culture, X-ray(often normal)/MRI. Definitive: Bone biopsy/culture)

Patient population	**MC organisms**	**Empiric antibiotic therapy**

Healthy children	• *S aureus*	• Low likelihood of MRSA
		  ○ Nafcillin/oxacillin OR cefazolin
		• High likelihood of MRSA
		  ○ Clindamycin OR vancomycin

Children with SCD	• *Salmonella* spp • *S aureus*	• As above PLUS • ceftriaxone, cefotaxime

- ❖ Patients with SCD are at increased risk for osteomyelitis because impaired blood flow of sickled cells through narrow metaphyseal vessels causes microinfarctions that act as a nidus for infection. In addition, splenic infarctions render patients with SCD functionally **asplenic** and more susceptible to infection with encapsulated organisms.
- ❖ Neither salmonella nor S aureus is a common cause of sepsis.
- ❖ *Pseudomonas aeruginosa* is associated with osteomyelitis of the foot after a puncture wound on the plantar surface. + bacteremia in burn wounds/neutropenia.
- ❖ **Limp/knee pain/monoarticular knee joint effusion** ddx: trauma (ligament or meniscus tear), septic arthritis (fever redness hotness), [Hemophilia (Male), SSA vasoocclusive crisis](severe). Rare: late lyme disease (mild, monoarticular/spreading annular rash), juvenile arthritis (high fever polyarticular, evanescent, macular, salmon-colored rash), and serum sickness (high fever polyarticular,pruritic rash).

Pediatric septic arthritis (joint drainage & debridement + IV abs) [JIA is a risk factor]	
Pathogenesis	• Age <3 months: *S aureus,* GBS, gram-negative bacilli • Age ≥3 months: *S aureus*, group A *Streptococcus*
Clinical features	• Fever ≥38.5 C (101.3 F), fussiness, poor feeding . Acute-onset joint pain, swelling, limited motion, **irritability with repositioning (eg, diaper changes)**. Refusal to bear weight. • **Lack of movement** of the affected side (pseudoparalysis), affected hip flexed, abducted, and externally rotated
Diagnosis	• ↑ WBC, ESR, CRP. Blood culture. Joint aspiration (synovial WBC count of >50,000/mm^3). Effusion on ultrasound/MRI. Tx: IV Vancomycin, if failed, ceftriaxone (for *Kingella kingae*)

- If the patient remains febrile or fails to improve after arthrocentesis and 48 hours of appropriate antibiotic therapy, MRI should be performed to evaluate for concomitant osteomyelitis. A delay in treatment can lead to femoral head necrosis, hip dislocation or leg-length discrepancy. Severe joint destruction may require total hip arthroplasty.

Tularemia (poorly staining gram – coccobacilli, highly virulent, require cysteine for growth, evades most immune defenses and replicates within neutrophils/macrophages)	
Epidemiology	○ Wild animal (eg, hare, rabbit) hunting/skinning ○ Tick or mosquito bite/ Bioterrorism agent
Common manifestations	• Fever, malaise, pneumonia [inhalation/sepsis], purulent conjunctivitis [introduction of infected material into an eye (eg, splashing, rubbing with a contaminated hand)] (oculoglandular disease: unilateral pain, redness, photophobia, purulent ulcers) • Ulceroglandular disease (**Periauricular**, cervical,submandibular) ○ Single papuloulcerative lesion (sometimes absent) ○ Tender, suppurative regional lymphadenopathy

Most cases arise after cutaneous inoculation.

DDx of unilateral conjunctivitis with ipsilateral preauricular lymphadenopathy **(Parinaud oculoglandular syndrome): F tularensis,** *B henselae* and HSV.

(Choice B) *Candida albicans*: prolonged neutropenia, recent eye trauma or surgery, or indwelling central catheters.

Neonatal tetanus (poor cord hygiene [swollen stump/pus], tetanospasmin)	
Clinical features	• Difficult feeding due to trismus, stridor and RF • Spasms & hypertonicity: clenched hands, dorsiflexed feet, opisthotonus
Treatment	• Supportive care. Metronidazole & tetanus immunoglobulin
Prevention	• Vaccination. Hygienic delivery & cord care

Infant botulism (Tx: Antitoxin therapy (botulism immunoglobulin) while waiting for tests)	
Pathogenesis	• Ingestion of *C botulinum* spores (eg, environmental dust/soil, honey) • Spores colonize the immature GI tract & produce toxin -> inhibits presynaptic acetylcholine release
Clinical presentation	• Age <12 months. Constipation, poor feeding, hypotonia • Oculobulbar palsies (eg, absent gag reflex, ptosis, ↓ eye movement) • Symmetric descending flaccid paralysis/weakness • Autonomic dysfunction (↓ salivation, fluctuating HR/BP). NO fever
Diagnosis	• Clinical; supported by abnormal EMG findings (eg, ↓ CMAP) • Confirmation by stool *C botulinum* spores or toxins

(Choice C) In neonates with mothers with MG, oculobulbar weakness and hypotonia within hours to days of birth can occur due to transplacental transfer of Ach receptor Abs.

(Choice D) Omphalitis is a potentially severe cutaneous infection that begins at the umbilical stump with erythema, swelling, and pus and can lead to sepsis.

Preseptal cellulitis: break in the periorbital skin (eg, **abrasion, insect bite**), eyelid erythema, swelling, chemosis, normal VA. *S aureus* or *S pyogenes*, oral clindamycin.

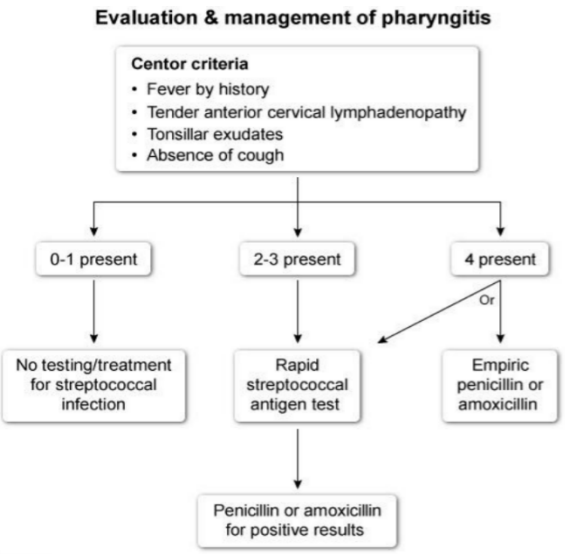
Orbital cellulitis: contiguous spread from another source of infection (eg, **sinusitis, dental abscess, preseptal cellulitis**). Symptoms of preseptal cellulitis **PLUS** Pain with EOM, proptosis &/or ophthalmoplegia with diplopia. IV vancomycin + cerftriaxone ±

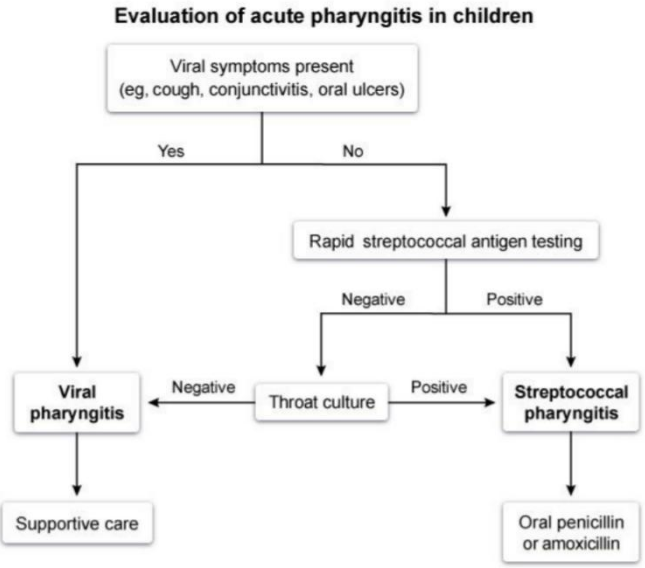
Surgical drainage. Severe complications include permanent visual impairment (eg, optic nerve involvement) and intracranial extension (eg, infection, cavernous sinus thrombosis).

If not treated, gonococcal conjunctivitis leads to corneal ulceration, scarring, blindness.
The majority of infants with gonococcal conjunctivitis are born to mothers who:

- Were not screened during pregnancy
- Had negative screening early in pregnancy but were infected later
- Had positive screening and were either not treated or treated and re-infected
- Had home delivery or refused postpartum prophylaxis with topical erythromycin

All infants, regardless of maternal screening results, should receive **topical erythromycin.**





- In children, tonsillar erythema/exudates, tender anterior cervical nodes, palatal petechiae both GAS & viral pharyngitis. Viral: cough, rhinorrhea, congestion, conjunctivits, oral ulcers.
- Because the risk of **acute rheumatic fever** is much higher in untreated children than in adults, no need for throat culture if negative rapid streptoccocal antigen test in adults.
- Antistreptolysin O antibodies peak approximately a month after streptococcal infection and are not helpful in diagnosing acute pharyngitis.

Nonbullous impetigo (S aureus [MC],GAS/ can lead to PSGN)	
Features	• Painful itchy papules & pustules with honey-crusted lesions. Face or extremities in young children. Local lymphadenopathy, NO fever.
Treatment	• **Topical** mupirocin if localized/ **oral** cephalexin if extensive

Predisposing factors: warm & humid climate, poor hygiene, preexisting skin trauma/inflammation (eg, scabies, atopic dermatitis). **Nonbullous** impetigo is more common than bullous impetigo (ie, flaccid bullae containing yellow fluid, tx with **oral** ABs).

Scarlet fever (S pyogenes, rapid antigen testing + throat culture. Oral amoxicillin)	
Clinical features (Pt >3 years old)	• Fever & pharyngitis headache (before rash)/ Tonsillar erythema & exudates. Tender shooty anterior cervical nodes/ Strawberry tongue. Sandpaper rash (trunk axilla groin -> desquamation). Circumoral pallor. Erythrogenic exotoxin

(Choice E) Roseola presents with high fever followed by a morbilliform rash that typically erupts once the patient has defervesced. In contrast, rash + still has fever, roseola unlikely.

Infective endocarditis (Acute Staph aureus, subacute S. viridans)	
Risk factors	• CHD or prosthetic valve/ Previous endocarditis. Intravascular catheters/ Intravenous drug use
Physical examination	• Fever, New regurgitant murmur. Janeway lesions, Osler nodes. Roth spots, splinter hemorrhages. Splenomegaly ± embolic phenomenon
Diagnostic testing	• Hematuria/proteinuria/ RBC casts (glomerulonephritis, NO biopsy) [immunologic sequelae], leukocytosis, thrombocytosis, anemia. • Positive blood cultures. TEE > TTE for detecting vegetation
Treatment	• Acute: Empiric vancomycin . Subacute: Tx based on culture results

Lyme disease prevention	
Tick removal	• Grasp with small forceps as close to skin as possible • Pull firmly upwards without twisting
Prophylaxis criteria (must meet all 5)	• *Ixodes scapularis* (deer tick) identified • Tick attached for ≥36 hours or engorged • Prophylaxis started within 72 hours of tick removal • Local *B burgdorferi* infx rate ≥20% (eg, New England) • No contraindications to doxycycline (eg, pregnancy)

Antimicrobial prophylaxis	• **Single-dose** doxycycline

Ixodes scapularis (deer tick) is endemic to the northeastern United States and is a vector for borrelia, babesia, anaplasma. Anaplasmosis and babesiosis are transmitted soon after attachment; *Borrelia* requires 48-72 hrs of feeding before salivary gland migration.

The body of the tick should not be squeezed or crushed because this can expel secretions into the wound and increase the risk of transmission. Surrounding erythema during tick attachment is typically due to local skin irritation from the tick saliva, not infection.

Antimicrobial prophylaxis is not effective against anaplasmosis and babesiosis.

(Choice A) Lyme serology cannot diagnose infection at the time of the tick bite because antibodies develop at least 1-4 weeks after transmission.

Stage	Lyme disease clinical features
Early localized (days to 1 month)	• Erythema migrans. Fatigue, headache. Myalgia, arthralgia
Early disseminated (weeks to months)	• **Multiple** erythema migrans. Bell's palsy. Meningitis. AV block. Migratory arthralgia
Late (months to yrs)	• Arthritis. Encephalitis. Peripheral neuropathy

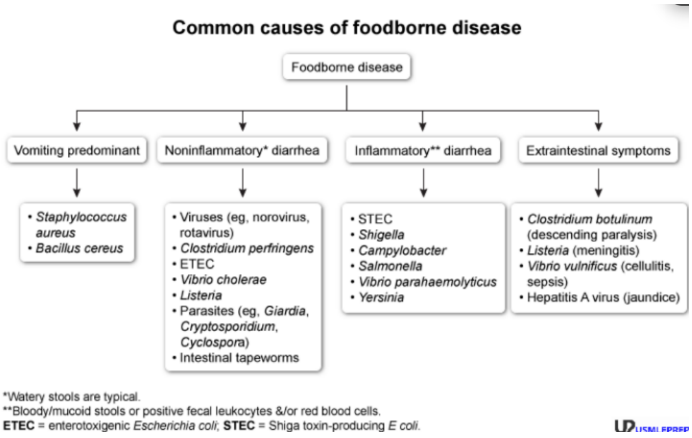
Arthritis: acute monoarticular arthritis of the **knee**, or asymmetric oligoarticular arthritis. **Large effusions** occur but are **minimally painful**. **Systemic symptoms absent**. Synovial fluid analysis: leukocyte count of **10,000-25,000/mm^3** (vs gonorrhea >25,000 & S aureus >50,000) with negative Gram stain and culture. Detection of Lyme **antibodies** in the **serum** (ELISA and Western blot testing) helps confirm the diagnosis. **Oral doxycycline** is the first-line treatment. Lyme arthritis: recurrent knee pain +swelling + **elevated ESR**, the hip is rarely involved.

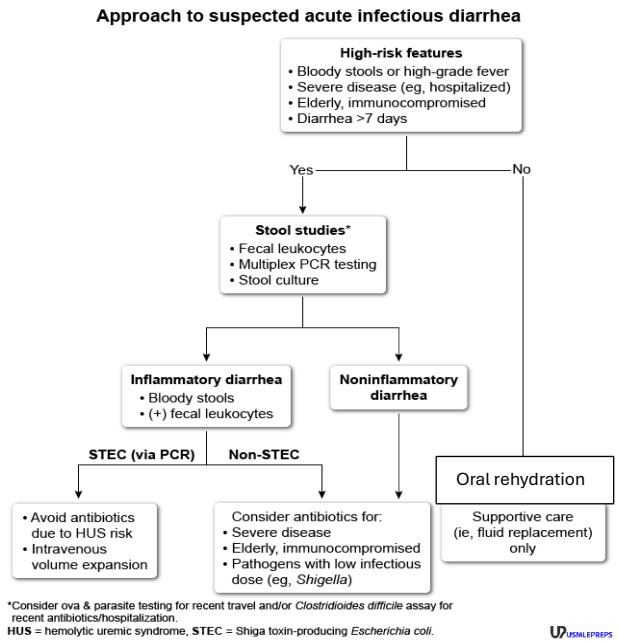
Brain abscess: fever, severe nocturnal positional headache (MC symptom, unresponsive to meds, ↑ ICP when supine [sleep]) AMS, morning vomiting, neck pain focal neurologic deficits. Risk factors: OM, sinusitis, mastoiditis, TOF: (right to left shunt, bypass lung which filters bacteria). Next step CT with contrast/MRI brain.

S aureus enterotoxin: **Diagnosis** is **clinical** (ie, no testing required), and management is fluid repletion. person-to-person spread does not occur, and contact precautions are unnecessary, **self-limited**, within 24-48 hours.

(Choice E) Stool toxin assay is for *Clostridioides difficile*. Blood culture is for shigella.

Tx: Needle aspiration/surgical excision, FOLLOWED BY IV antibiotics.





Acute gastroenteritis: MCC norovirus.

(Choice F) Loperamide, an antimotility drug, is not recommended in children due to its potential adverse effects (eg, paralytic ileus, toxic megacolon). Decreased intestinal motility can also prolong fecal shedding. The risk of HUS is increased with its use in STEC.

Infectious bloody diarrhea			
Organism	**History**	**Treatment**	**Complications**
Shiga-toxin–producing *E coli*	• Ingestion of undercooked beef • Absence of high fever	• Supportive care	• HUS
Shigella	• Ingestion of contaminated food/water (high fever LLQ pain tenesmus) • Outbreaks (eg, day care) **(low ID)**	• Supportive. Abs if severe	• Less commonly HUS +Seizure (in children)
Campylobacter	• Ingestion of raw or undercooked meat	• Supportive. Abs if severe	• GBS
Salmonella	• Undercooked chicken • Contact with reptiles	• Supportive. Abs if severe	• Bacteremia

In contrast to *STEC* diarrhea, in shigellosis, antibiotics do not increase the risk of HUS.

***Campylobacter* gastroenteritis (undercooked poultry)**	
Clinical features	• Fever, abdominal pain, diarrhea (mucoid ± blood) • Pseudoappendicitis (RLQ pain due to acute ileocecitis)
Treatment	• Supportive care (symptoms usually self-limited <7 days) • Antibiotics(Amoxicillin, **doxycycline >8yrs**) in **severe cases***
Complications	• Guillain-Barré syndrome + Reactive arthritis
*Duration >7 days; bloody stools; high fevers; patients who are pregnant, immunocompromised, or elderly.	

Salmonella	**Nontyphoidal**	**Typhoidal**
Epidemiology	Major cause of gastroenteritis worldwide (including United States) Associated with undercooked poultry/eggs	Most common in resource-limited regions with poor sanitation (eg, unvaccinated travelers) Associated with contaminated food or water
Clinical	Vomiting Diarrhea ± blood Fever Invasive disease rare	Fever & bacteremia Abdominal pain & rose spots Late findings: HSM, intestinal perforation
Diagnosis	Stool culture	Blood culture
Outcome & treatment	Usually self-limited Antibiotics rarely needed	Potentially fatal Antibiotics (eg, ceftriaxone) Drug resistance common

Salmonella enteritidis is carried by reptiles (including turtles) and can cause **severe mesenteric adenitis** in conjunction with enteritis.

Rocky Mountain spotted fever (Rickettsia, tick-borne, grassy woods in summer)	
Clinical features	• Nonspecific fever, headache, myalgia, arthralgia, abd pain • Macular & petechial rash on wrists/ankles • Complications: encephalitis, pulmonary edema, bleeding, shock
Labs	• ↓ Platelets/↓ Sodium/ ↑ AST & ALT
Diagnosis	• *Rickettsia* serology + Skin biopsy

	• Tx: Doxycycline, **including in children <8yrs** [dental staining is minimal with the short courses (eg, 5-7 days) used for RMSF]

CSF findings usually include normal glucose and leukocytes <100/mm^3.

Primary pulmonary TB (chronic cough, fever, weight loss FTT, unilateral wheezing)	
Risk factors	• Immunosuppression (eg, HIV). Travel from endemic area • Infected household contact. Resident/employee of prison, homeless shelter, health care facility
Diagnosis	• IGRA, NAAT, TST. Positive AFB smear & mycobacterial culture • Chest x-ray: hilar adenopathy, effusion, consolidation, cavitation. NO upper lobe cavitation (It is present in secondary TB only)

- ❖ Although **chronic cough is generally defined as >8 weeks in adults and >4 weeks in children**, even shorter durations (eg, >2 weeks in adults, >3 weeks in children) can raise suspicion for TB in the appropriate epidemiologic and clinical setting.
- ❖ Younger children who cannot generate sufficient cough: dx by early-morning **gastric lavage**, which recovers *M tuberculosis* from swallowed tracheal secretions.
- ❖ **Miliary TB**: Fever FTT cough lethargy RF. Lymphohematogenous dissemination of *M tuberculosis* from the lungs to other organs → micronodular lesions in the lungs, liver, and spleen. Hematogeneous dissemination back to the lung causes millet-seed appearance. It is most common among **infants and immunocompromised** hosts with poor T-cell function and is often associated with **false-negative IGRA & TST**. Definitive diagnosis requires culture of *M tuberculosis* from lungs, blood, or tissue biopsy.
- ❖ Tuberculous lymphadenitis is the MC extrapulmonary manifestation of tuberculosis.

Catscratch disease (*B henselae*, fastidious gram-negative bacilli, cat scratch/bite)	
Clinical manifestations	• Papule at scratch/bite site - Regional adenopathy ± Fever of unknown origin (≥14 days)
Diagnosis	• Usually clinical ± Serology. Tx: self limiting/azithromycin

- ❖ Painful nonsuppurative regional lymphadenitis that **fails to respond** to initial clindamycin therapy (for staph and strep) should raise suspicion for **catscratch disease**. Deeper spread to the eye, liver, and spleen occasionally occurs.
- ❖ HIV + toxoplasma + disseminated Histoplasma = bilateral generalized, painless lymphadenopathy.

Congenital syphilis (Dx: VDRL, RPR. Darkfield microscopy. Tx: Penicillin)	
Clinical features	**Early:** Jaundice, hepatosplenomegaly, growth restriction, "blueberry muffin" spots. Snuffles: copious clear, purulent, or serosanguineous **rhinorrhea**. Maculopapular rash on palms soles buttocks with **desquamation** & hyperpigmentation. Bilateral and symmetric **metaphyseal erosions and periosteal inflammation** of long bones. Pathologic fx. Pain swelling limited ROM of affected extremities. Skin **fissures** adjacent to orifices (eg, anal, oral, nasal) **Late (>2 years):** Saddle nose, hutchinson teeth, saber shins (Anterior tibial bowing), SNHL
Prevention	• First-trimester maternal screening; repeat if high risk (eg, other prenatal STI). Prenatal penicillin

(Choice B) Radionucleotide bone scanning to diagnose multifocal osteomyelitis - pathologic fractures. Fever, prematurity, history of a central line.

Infection control isolation precautions	
Airborne	• Bacterial (tuberculosis). Viral (varicella, SARS, measles) [aerosols <5 microns, negative pressure rooms, N95 masks]
Contact	• MRSA, VRE. *C difficile*, *E coli* O157:H7, scabies, RSV [gown & gloves]
Droplet	• *N meningitidis* HiB, *M pneumoniae,* influenza, adenovirus [particles >5 microns, surgical mask within 3-6 feet]

- Common **neutropenic precautions**: strict hand hygiene, private rooms with high-efficiency air filtration, no live plants or animals, and avoidance of intrusive probes (eg, rectal thermometers, enemas).
- **Standard precautions** should be followed in the care of all patients. Hand hygiene before and after contact, use of safety gear when there is possible contact with bodily fluids, respiratory hygiene/cough etiquette, and safe injection practices. Standard precautions are included within all other precautions.
- The **(MMR)** vaccination is recommended at age 1 and age 4. For planned international travel, an additional dose between age 6 and 11 months is also recommended. Can cause fever + maculopapular rash **within 1-3 weeks** of immunization (3%-5%). Reassurance, no airborne isolation, only avoid immunocompromised individuals.

Measles virus (rubeola) [tx supportive + vit A for hospitalized patients (promotion of antibody-producing cells + regeneration of epithelial cells (eg, in gut, lungs, retina))]	
Clinical presentation	• Prodrome (eg, fever >40C, cough, coryza, conjunctivitis, Koplik spots [small white lesions on buccal mucosa opposite the molars, do not appear in all patients but are **pathognomonic** of measles]) -> classic maculopapular rash (cephalocaudal & centrifugal spread spares palms & soles) [symptoms 1-3 wks after exposure] • The rash subsequently coalesces and may become nonblanching with a dark, reddish-brown color. Vit A deficiency is a risk factor for complications: pneumonia meningitis + SSPE (fatal, ranging from personality changes to dementia and death over months to years).

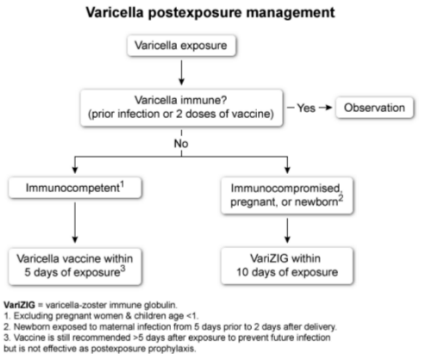
❖ Postexposure prophylaxis is for **influenza, varicella, HIV, rabies (passive immunization)**

Rubella (German measles) [Dx: Serology. Tx: supportive]	
Clinical presentation	• **Congenital (1st trimester):** (deafness, cataracts, PDA blueberry muffin baby, symmetric FGR, hepatosplenomegaly) • **Children:** (fever, maculopapular rash cephalocaudal lasts <3 days, **suboccipital/ posterior auricular/** posterior cervical **lymphadenopathy**, petechiae and erythematous papules on soft palate **[Forchheimer spots]**) • **Adolescents/Adults:** Same as children + arthralgias (can last up to 1 month. Other symptoms few days only)

Infectious mononucleosis (fever, pharyngitis ± exudates, posterior/diffuse cervical lympadenopathy, significant fatigue, ± hepatosplenomegaly ± amoxicillin rash)	
Diagnostic findings	• Positive heterophile antibody (Monospot) test (25% FN rate during 1st week of illness). Atypical lymphocytosis/ Transient hepatitis
Management	• Avoid **sports for ≥3 weeks** (**contact sports ≥4 weeks**) due to the risk of **splenic rupture** within 3 weeks (**Abd pain & anemia**)
Complications	• Splenic rupture, AIHA, thrombocytopenia, acute airway obstruction (severe tonsillar enlargement, treated by corticosteroids to ↓ edema)

Amoxicillin rash: Delayed hypersensitivity. It is **not** a true drug **allergy**, and most patients can subsequently receive the same antibiotic without reaction.

Children age <1 (outside of the immediate newborn period) cannot receive the VZV vaccine and are low risk for severe infection, and therefore do not require PEP.



Neonates: fever, rash, pneumonia, hepatitis, meningoencephalitis. Prevention: Isolate infant + IG. Tx: Acyclovir

Chickenpox (varicella) : [direct contact or aerosols, clinical dx + PCR of skin/blood/CSF in atypical (eg, mild or breakthrough disease in partially vaccinated patients) or severe (eg, immunocompromised patients) cases]	
Epidemiology	• **High risk group**: adolescents, adults, immunocompromised individuals, pregnant women, newborns
Clinical features	• **Prodrome** (eg, fever, malaise) -> Pruritic macules, papules & vesicles in successive crops at <u>different stages that later crust</u> • Complications: <u>skin infection, pneumonia, encephalitis</u>
Treatment	• Supportive (eg, **antipyretics, antihistamines**) in young healthy children. **Acyclovir** for high-risk patients & complicated disease
Prevention	• **2 doses of VZV vaccine (age 1 & 4)**

Classic triad of **herpes zoster oticus** (Ramsay Hunt syndrome, reactivation of VZV in the geniculate ganglion): **Ear pain, facial weakness, vesicular rash in external auditory canal** (which is innervated by facial N).

Subsequent spread to the (CN VIII) can lead to auditory (eg, **hearing loss, hyperacusis, tinnitus**) and vestibular (eg, **vertigo, nausea/vomiting**) disturbances. Treatment is with <u>corticosteroids, ± antiviral medications, artificial tears, lubricating ointment</u>. Early initiation **(<3 days)** is associated with improved recovery of facial nerve function; however, even with treatment many patients have residual facial weakness.

VZV vaccine (live attenuated)	
Contraindications	• Immunocompromised (eg, chemotherapy, T-cell immunodeficiency, active/untreated tuberculosis)

	• Pregnancy/ Allergy to vaccine component (neomycin, gelatin), causing **type 1 or 4** hypersensitivity reaction
Adverse effects	• Redness/soreness at inj site + Mild maculopapular/**vesicular** rash (<10 lesions, less pruritic) (**VZV vaccine strain**) **1-3 weeks** postvaccination, NO fever

Congenital CMV (MC congenital infection. Main risk factor: caring for young children. Findings: Periventricular{germinal matrix bleeds}/intrahepatic calcifications, microcephaly, ventriculomegaly, FGR, hydrops fetalis. SNHL (MC sequelae of congenital CMV infection). Hepatosplenomegaly, thrombocytopenia. Developmental delay, learning disabilities. Dx: PCR, viral culture of urine/saliva. Tx: valgancyclovir if symptomatic)

CMV infection is common in children age 1-4 (who attend day care), and pregnant women typically acquire CMV through contact with **body fluids** (eg, urine, **saliva**) of infected children. Maternal infection is often asymptomatic but may present as a nonspecific, mild viral illness. 1st trimester primary maternal infection is associated with higher infant morbidity as compared with recurrent maternal infection or infection late in pregnancy.

Molluscum contagiosum (poxvirus, direct contact + fomites (towels). Clinical diagnosis (ie, firm, round pink or flesh colored domed, papule with central umbilication, often shiny but may be itchy or inflamed). Symptoms resolve within 6-12 months.

	Children	**Adults**
Typical location	• Trunk, axillae, face (including eyelids)	• Lower abdomen, genitals, upper thighs
Evaluation	• No further evaluation	• Genital lesions: STI testing • Extensive lesions: HIV testing
Treatment	• Reassurance	• Cryotherapy. Curettage. Topical (cantharidin, podophyllotoxin) [can be painful and cause scarring]

Lesions in atypical regions, even the genital area, in children are typically due to autoinoculation and usually not sexual abuse.

Human rabies (bite from infected **bats, racoons,** skunks, foxes, or dogs)

Clinical	**Encephalitic**	• Prodrome: fever, sore throat, malaise, wound numbness

Features **(1-3 mo after exposure)**		• Hydrophobia & aerophobia (due to pharyngeal spasm) - Autonomic instability - Agitation & AMS - Spasticity (grimacing opisthotonos) - hypersalivation
	Paralytic	• Ascending flaccid paralysis
Postexposure prophylaxis*	• Rabies immunoglobulin + Rabies vaccine series	
Prognosis	• Coma, respiratory failure & death within weeks	
*In cases of a high-risk animal that cannot be tested or observed.		

❖ Because bat teeth are small and needle shaped, many bites may go unnoticed, particularly if the patient is asleep. Pts can open their mouths vs tetanus (trismus)

❖ Rabies within 1-3 mo, tetanus within 3 weeks, botulism within 2 weeks of exposure.

Viral gastroenteritis (feco-oral, fever vomiting watery diarrhea abd pain, clinical dx)	
Epidemiology	• Norovirus: MC among all ages/ Rotavirus: unvaccinated age <2

Treatment	• Fluid repletion (symptoms resolve within 2-3d)
	  ○ Oral for mild dehydration (eg, normal vital signs, dry mucous membranes). Antiemetics can be considered for vomting that prevents ORS)
	  ○ Intravenous for severe dehydration (eg, weak/rapid pulse, marked oliguria, ↑ capillary refill time)
	• Regular diet (with limited fats & simple sugars) as tolerated

Bacterial/parasitic: grossly bloody stools, persistent fever, travel, farm animals.	

Norovirus low ID. Feco-oral: person-to-person spread (MC), ingestion of contaminated food (shellfish, salad, and fruit). Symptoms within 1-2 d of exposure

The diagnosis is generally clinical. However, detection of norovirus in the stool may be performed in outbreaks or atypical cases (eg, symptoms >7 days). Fecal leukocytes, mucus or blood are not seen

Because norovirus is not killed by alcohol-based sanitizer or standard cleaning solutions, Prevention= soap and water + cleansing of contaminated surfaces with bleach.

(Choices A and E) *B cereus* and *S aureus* no diarrhea, symptoms within 6 hours of eating the contaminated food. Vs norovirus 36 hrs

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- ❖ Mumps: School-age children. Fever myalgia fatigue -> unilateral or bilateral parotitis. Complications (due to systemic spread of the virus): Orchitis, pancreatitis, aseptic meningitis (benign) SNHL (transient, but can lead to deafness)
- ❖ Myositis, particularly of the calves or thighs, can be a complication of influenza, often with lower respiratory tract infection. Primary HIV & EBV can cause parotitis.

Herpes simplex virus (HSV) encephalitis (Tx: Empiric IV acyclovir while awaiting tests)	
Clinical findings	• Fever Headache Seizure AMS (eg, confusion, agitation) • ± Focal neurologic findings (eg, hemiparesis, CN palsies, ataxia)

Diagnostic findings	• CSF analysis:
	  ○ ↑ WBCs (↑ lymphocytes), ↑ RBCs
	  ○ ↑ Protein, normal glucose, HSV DNA on PCR
	• Brain MRI: temporal lobe hemorrhage/edema

Spread of primary infection likely occurs via the olfactory bulb to the olfactory cortex (within the temporal lobe) whereas reactivated HSV spreads from the trigeminal ganglion (where HSV typically remains dormant) to the adjacent temporal lobe.

Neonatal HSV: Vertical transmission (Intrauterine, perinatal, postnatal), can be disseminated (Sepsis, hepatitis, pneumonia). Dx: Viral surface culture/PCR. Neonatal encephalitis in 2nd/3rd week of life, full fontanelle

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Perinatal hepatitis B virus infection (prevention: HB vaccine + IG within 12 hrs of birth)	
Epidemiology	• 90% risk of vertical transmission without prophylaxis, <2% risk after prophylaxis. Chronic infection in 90% of perinatally infected infants
Risk factors	• High maternal viral load. Maternal HBeAg+
Transmission	• Genital secretions (MC), transplacental (rare), NOT breastfeeding

The majority of HepB vertical transmissions occur during the passage of the fetus through the birth canal. HIV is the **only absolute infectious contraindication** to breastfeeding in the developed world.

Herpangina vs herpetic gingivostomatitis		
	Herpangina (NO rash)	**Herpetic gingivostomatitis**
Etiology	• Coxsackievirus A	• Herpes simplex virus type 1
Patient age	• 3-10 years	• 6 months to 5 years
Seasonality	• Late summer/early fall	• None
Clinical features	• Fever & pharyngitis • Gray vesicles-> fibrin coated ulcers on posterior oropharynx	• Fever & pharyngitis • Clusters of vesicles/ulcers on anterior oral mucosa & lips • Erythematous & edematous gingiva
Treatment	• Supportive(oral hydration, analgesia), resolve in 1 wk	• Oral acyclovir

- **Herpangina**:The oral enanthem is located in the posterior oropharynx on the posterior **soft palate**, anterior **palatine pillars**, **tonsils**, and **uvula**. Vs HSV :anterior oral cavity (buccal mucosa, tongue, gingiva, hard palate) and lips.
- **Ludwig angina**, a potentially fatal cellulitis of the submandibular space, is caused by bacterial spread from dental abscess. Inadequate dental hygiene is a RF. Symptoms: fever, drooling, muffled voice, stridor, dysphagia. Tender bilateral induration of the submandibular area, often with elevation of the floor of the oropharynx
- **Acquired subglottic stenosis**, complication from ETT, hoarseness + stridor.

Acute HIV infection: young, sexually active adults, 2-4wks after transmission: unexplained fever, **generalized lymphadenopathy**, **maculopapular rash**, aseptic meningitis (mild, 25% of cases).

HIV in infancy (Dx: HIV DNA or RNA PCR, Tx: combined ART)	
Risk factors	• High maternal viral load (MC)(eg, insufficient prenatal care, lack of antiretroviral therapy). Breastfeeding by infected mother
Clinical features	• FTT. Chronic diarrhea. Lymphadenopathy, hepatosplenomegaly • *Pneumocystis* pneumonia. Prolonged/refractory candidiasis

Because B cell and CD8+ T cell production persists, total absolute lymphocyte count is often normal. Therefore, lymph node enlargement commonly occurs as the immune

system responds to high viral loads. In contrast, patients with SCID (eg, adenosine deaminase deficiency) have profound lymphopenia and the absence of lymphoid tissue.

Trasmitted during delivery (MC) or transplacental. Antibody testing is not performed at age <18 months as maternal antibodies may cause false positives.

PCP: **age 3-6 months**. low-grade fever, tachypnea, poor feeding, **progressive dyspnea**. Generalized adenopathy, hepatomegaly, **severe hypoxia**, **bilateral perihilar reticulonodular infiltrates** that become more diffuse as the illness progresses. Because the pathogen cannot be cultured, the diagnosis is established when the yeast-like organism is identified in pulmonary fluid (eg, induced sputum, bronchiolar lavage). CD4 (**1,000-1,200/mm^3**); vs adults <200/mm^3, cause of thymus in neonates.

First-line treatment is **TMP-SMX**, a short course of corticosteroids if PaO_2 <70 or A-a gradient of >35. Following resolution, PJP prophylaxis is required until CD4 counts improve past age-related risk levels (eg, >1,500/mm^3 for patients age 1-11 months vs ≥200/mm^3 [for ≥3 months] in adults). Antiretroviral therapy should be initiated within a few weeks.

(Choice D) Streptomycin is sometimes used for IE, tularemia, or brucellosis.

Congenital toxoplasmosis (Central Africa. Tx: Pyrimethamine, sulfadiazine, folinic acid)	
Maternal risk factors	• Raw or **undercooked meat**, Unwashed produce (ie, contaminated soil), Handling of cat feces
Clinical features	• Classic triad: Chorioretinitis, hydroceph, diffuse intracranial calcif • Microcephaly (brain atrophy), Seizures • Jaundice, hepatosplenomegaly, rash, growth restriction
Diagnosis	• *Toxoplasma* serology or PCR

Chorioretinitis both toxoplasma & CMV, hydrocephalus only toxoplasma.

***E vermicularis* (pinworm) [Perianal pruritis, especially at night]**	
Treatment	Albendazole OR pyrantel pamoate for pt & all household contacts

School age children, contact with contaminated objects (eg, bedding, clothing) or unwashed hands after scratching the perianal area. Other symptoms may include nausea, vomiting, abdominal pain, and vulvovaginitis. Dx: eggs on scotch tape.

Prolonged (≥2 weeks) travelers' diarrhea (Bacterial usually <2 weeks)

Pathogen	Clinical features
E histolytica	• RLQ pain. Bloody/mucoid diarrhea. Liver abscess. NO fever
Giardia	• Steatorrhea. Bloating, fat malabsorption, weight loss
Cryptosporidium or Cystoisospora	• Watery diarrhea in immunosuppressed patients
Cyclospora	• Watery diarrhea ± blood. Waxing/waning symptoms

Chapter 4: ENT

Choanal atresia (Dx: catheter, CT, nasal endoscopy. Tx: Oral airway, surgical repair)	
Clinical findings	• Unilateral (most common): Chronic nasal discharge. Symptomatic during childhood • Bilateral: Cyanosis that worsens with feeding & improves with crying. Noisy breathing (stertor). Symptomatic shortly after birth • May be associated with CHARGE syndrome

CHARGE syndrome	
Characteristic features	• Coloboma [hole in one of the eye structures]. Heart defects (TOF, VSD). Atresia choanae. Retardation of growth/development. Genitourinary anomalies [cryptorchidism]. Ear abnormalities (eg, hearing loss & dysplastic ears [short and wide with no earlobes]) + Anosmia, cleft lip/palate, hypotonia
Diagnosis	• Clinical. *CHD7* gene testing

Initial management of infants with CHARGE syndrome includes placement of an **oral airway** for respiratory support in patients with choanal atresia + **Screening echocardiogram and renal ultrasound**. Children with CHARGE syndrome require **lifelong, multispecialty care due to delays in development, puberty, and growth.**

Causes of stridor in infants & toddlers

Acute	
Croup (6mo-3 yr age)	• Parainfluenza virus, most cases in fall/winter • Inspiratory or biphasic stridor (subglottic edema), "barky" cough, infectious symptoms
Foreign body aspiration	• ± Choking episode • Inspiratory stridor &/or wheeze, focally diminished breath sounds
Chronic	
Laryngomalacia (normal xray, MCC of stridor in infants, flexible laryngoscopy [omega shaped epiglottis], GER)	• "Floppy" supraglottis, prominent age 4-8 months • Inspiratory stridor worsens when feeding, crying, or supine; improves when prone, self-limiting in 18 mo • Reassurance with close follow up. GER tx. Supraglottoplasty if severe
Vascular ring	• Great vessels encircle & compress trachea • Biphasic stridor that improves with neck extension and worsens with feeding/crying
Airway hemangioma	• Hemangiomas enlarge in the first few weeks of life (not immediately at birth) • **Worsening** biphasic stridor, concurrent skin hemangiomas ("beard distribution")

Laryngomalacia: Inspiration -> faster airflow -> ↓ intralaryngeal pressure -> turbulent flow & **inspiratory stridor**. VS **tracheomalacia: expiratory stridor.** The subglottic airway is less flexible because it is supported by a complete ring of cartilage (the cricoid); as a result, there is minimal change in subglottic airway caliber in inspiration compared with expiration. Therefore, conditions that result in subglottic narrowing (eg, subglottic stenosis) often lead to **biphasic stridor.**

Bullous myringitis, uncommon complication of AOM, TM bullae.

Cholesteatoma: Abnormal growth of keratinized squamous epithelium in the middle ear, congenital (age 5) or aquired (MC). Risk factors: recurrent OM (cleft palate), chronic middle ear effusion, and tympanostomy tube placement. Recurrent painless otorrhea, CHL, pearly

white mass or a retraction pocket in the anterosuperior quadrant of the TM. Complications: CHL, CN palsy, meningitis, brain abscess.

Chronic suppurative otitis media Polymicrobial (*S aureus, P aeruginosa, Aspergillus*)	
Symptoms	• Chronic (>6 wk), purulent otorrhea, hearing loss, no ear pain
Examination	• TM perforation. Otorrhea within normal-appearing EAC.
Treatment	• Ototopical fluoroquinolone drops (eg, ofloxacin)
Complications	• Mastoiditis, meningitis, intracranial abscess. Permanent HL

Risk factors: Young children (acute otitis media), negative middle ear pressure (ET dysfunction, cholesteatoma) -> weaker TM -> perforation. Clinical dx

CSOM does not typically respond to oral antibiotics, because the middle ear is poorly vascularized due to chronic inflammation and scarring. Of note, **cholesteatoma** can also cause chronic ear drainage, and should be considered **when the condition does not respond to topical therapy for presumed CSOM**.

Otitis externa (*P aeruginosa (MC). S aureus*. Tx: Remove debris, topical fluoroquinolone ± topical glucocorticoid ± wick placement to facilitate medication delivery)	
Risk factors	• Water exposure (MC, loss of antibacterial cerumen/ raised pH/ skin maceration/ water introduces pathogenic bateria). Ear canal trauma (eg, cotton swabs, ear candling)/ occlusion by foreign material (hearing aid, headphones) • Dermatologic conditions (eg, eczema, contact dermatitis)
Clinical manifestations	• Otalgia (worse with auricle manipulation), pruritus, HL, discharge • Ear canal erythema, edema, debris. Normal TM

(Choices B and F) Fungal otitis (otomycosis), after treatment for acute otitis externa, chronically moist ear canals (hearing aids, warm/humid climates). EAC shows whitish fungal debris with fruiting bodies or spores. Mx is debridement and topical clotrimazole.

Otitis media with effusion (middle ear fluid **without TM inflammation**[bulging erythema fever]) RF: age 6-24 mo, viral infx or post AOM. Typically asymptomatic but can cause ear tugging/pulling, CHL. Air fluid levels posterior to TM and poor TM mobility with NO bulging/ erythema. **Effusion is nonpurulent unlike AOM**.

Mx: Observe and follow up because **chronic** OME (>3 months) can cause **speech delay** and **long-term hearing loss**. Tympanostomy tube placement is warranted for chronic OME with associated hearing loss.

Mastoiditis: **MC suppurative complication of AOM**, fever, ear pain, protrusion of the outer ear (outward/vertical displacement of the auricle), mastoid (postauricular) erythema and tenderness, bulging TM. Children age ≤2. *S pneumoniae (MC)*, *S pyogenes*, *S aureus*. *P aeruginosa* (recurrent infections or recent antibiotic use). Clinical dx (CT for suspected meningitis/brain abscess/neurological deficits, toxic appearance, not responsive to tx). **Tx: IV Ab therapy** + **Drainage** with tympanostomy (± ear tube placement) or mastoidectomy.

Complications	• Extracranial extension (subperiosteal abscess, facial nerve palsy, hearing loss, labyrinthitis) • Intracranial extension (meningitis, brain abscess)

(Choice D) Osteoma: Benign, solitary area of bony overgrowth. Osteomas are more common in adults and in external ear, can lead to hearing loss.

(Choice E) Otosclerosis: bony overgrowth of the stapes footplate, CHL, normal TM.

Tympanocentesis and culture during tympanostomy tube placement should be considered for multiple episodes of AOM (eg, ≥3 episodes in 6 months) or persistent (>3 months) middle ear effusion with hearing loss.

Acute rhinosinusitis in children	
Clinical features	• Nasal congestion, purulent drainage, facial pain (leaning forward) • ± Fever, cough, headache, loss of smell, ear pain
Etiology	• Viral ○ No fever or early resolution of fever, well-appearing, mild facial pain, improvement & resolution by day 5-10 • Bacterial (1 of 3 criteria)
Treatment	• Supportive: Intranasal saline, saline irrigation, NSAIDs • Antibiotics if bacterial

(Choice D) Parotitis: fever, painful preauricular and postauricular swelling. Elderly post-op.

(Choice E) Perichondritis, painful, erythematous ear limited to the cartilaginous portions.

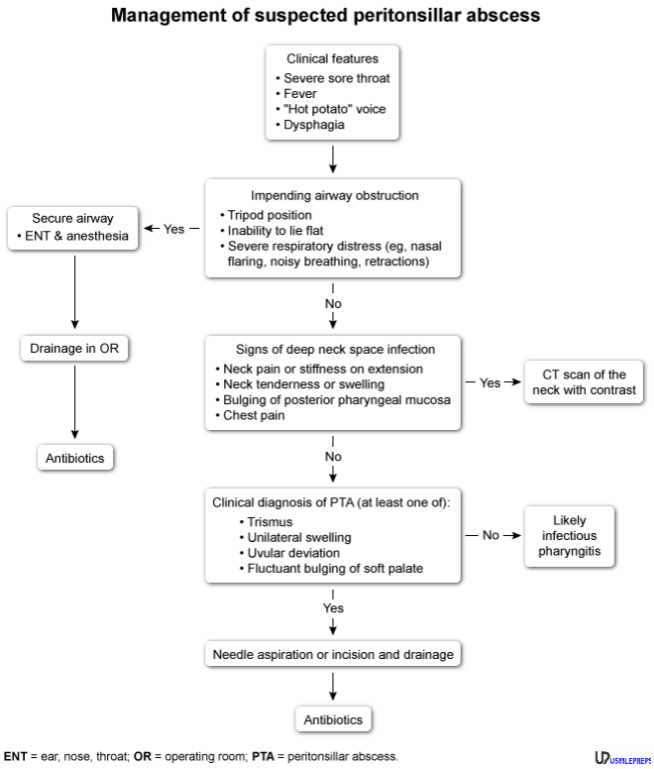
Laryngeal papillomas due to **recurrent respiratory papillomatosis (RRP)**. These lesions can be finger-shaped, warty, or grapelike, and as with skin papillomas, have dark-red punctate areas corresponding to blood vessels.

HPV 6 11 **vertical transmission** prior to delivery (because neither vaginal nor cesarean delivery prevent transmission), hoarseness of voice, airway obstruction, repeated operative interventions. Tx: surgical debridement

(Choice A) Laryngeal web: Anomalous airway development, hoarseness from birth.

(Choice B) Adenoid hypertrophy: MCC of nasal obstruction in children.

Peritonsillar abscess (PTA), an acute bacterial infection of the region between the tonsil and the pharyngeal muscles. The infection begins as **persistent** tonsillitis/**pharyngitis** (eg, 2 weeks of sore throat) and **progresses** (eg, fever, worsening sore throat) to cellulitis/phlegmon, with pus collecting into an abscess within a few days of symptom onset. PTA is most common in older **adolescents and young adults; drug or alcohol use increases the risk.**



Severe (often unilateral) sore throat and fever, **muffled ("hot potato") voice**, **trismus**, dysphagia, unilateral swelling of the soft palate with **uvular deviation**, earache, lymphadenopathy, and pooling of saliva. GAS + anaerobes

(Choice C) If lymph node swelling persists (eg, >3-4 weeks), biopsy may be indicated to rule out certain conditions (eg, malignancy).

Interpretation of Rinne & Weber tests		
	Rinne result	**Weber result**
Normal	AC > BC in **both** ears	Midline
Conductive hearing loss	BC > AC in **affected** ear, AC > BC in **unaffected** ear	Lateralizes to **affected** ear

Sensorineural hearing loss	AC > BC in **both** ears	Lateralizes to **unaffected** ear, **away** from affected ear
Mixed hearing loss	BC > AC in **affected** ear, AC > BC in **unaffected** ear	Lateralizes to **unaffected** ear, **away** from affected ear

MCC of nonhereditary SNHL in children is **congenital (CMV) infection**. SNHL due to noise exposure is rare at age ≤5.

Nasal foreign body (age 1-6 yo)	
Clinical manifestations	• Inorganic substance (eg, toy): mild pain/discomfort • Organic substance (eg, food): unilateral, foul-smelling, purulent, bloody discharge/ sneezing & epistaxis • Button battery: epistaxis, purulent or black discharge
Treatment	• Positive pressure expulsion (eg, forceful exhalation with unaffected naris occluded). Mechanical extraction
Complications	• Local irritation, sinusitis, aspiration, nasal septal perforation (button battery/multiple magnets), periorbital cellulitis

Imaging is unhelpful because most objects are radiolucent. A visualized object can usually be safely removed in the **office**. However, if the object cannot be visualized because of its location or surrounding edema or if removal could cause further trauma (eg, penetrating objects), referral to otolaryngology for **nasal endoscopy** is appropriate.

TEF with esophageal atresia	
Clinical features	• Coughing, choking, drooling, vomiting with feeding, VACTERL, respiratory distress and coarse breath sounds (particularly on the right side) due to aspiration
Diagnosis	• Inability to pass enteric tube beyond (10-15 cm) + coiling in proximal esophagus
Management	• Surgical correction + echocardiography, renal ultrasound/ Contrast enema (to detect anal atresia) and limb radiographs may also be performed if clinical suspicion for VACTERL association is high.

MCC of hearing impairment in children: CHL due to repeated ear infections. Hearing impairment can be **mistaken** for other behavioral or pervasive **developmental**

disorders. Although this child's 3-year audiometry screening 1 year ago was normal, he has had **multiple ear infections** in the last year, making repeat **audiometry** testing necessary to assess for acquired hearing loss. Can lead to **poor language development and social isolation unlike ADHD**.

A formal speech and language assessment **should be done only after a hearing assessment has ruled out hearing loss.**

(Choice E) An **EEG** can be used to rule out **Landau-Kleffner syndrome**, an **epileptic** condition that presents with regression in language milestones after normal development.

Eustachian tube dysfunction	
Physiology	• Eustachian tubes normally open and close to: ○ Equalize middle ear pressure. Drain middle ear. Prevent reflux of nasopharyngeal secretions
Pathophysiology	• Inflammation (eg, infection, allergies, environmental irritation)
Signs & symptoms	• Ear fullness/discomfort. Tinnitus. CHL. "Popping" sensation when yawning/swallowing. Retracted TM (due to negative pressure in middle ear)
Management	• Treat underlying cause (eg, antibiotics for ABRS. Antihistamines for allergic rhinitis)

Complications: AOM, permenant HL, TM rupture, cholesteatoma.

Thyroglossal duct cyst	
Clinical presentation	• Midline cystic neck mass • Moves superiorly with swallowing or tongue protrusion • Often present **after upper respiratory tract infection** (secondary infection -> erythema and tenderness). • Associated with ectopic thyroid tissue
Management	• Confirm presence of normal thyroid tissue • Surgical resection of cyst, associated tract & central portion of hyoid bone (**sistrunk procedure**) [due to risk of recurrent infx]

Chapter 5: Endocrine

Clinical features of hypothyroidism	
Symptoms + Signs	• Psychomotor depression (declining school performance), delirium, psychosis (myxedema madness), bradycardia, muscle weakness & myalgia.... • Declining growth velocity, short stature, delayed puberty.

Congenital hypothyroidism: Familial or sporadic. MCC is **thyroid dysgenesis** (i.e., aplasia, hypoplasia, or ectopic gland). Other causes include **dyshormogenesis** (10%), and transplacental maternal thyrotropin-receptor blocking **antibodies** (5%). Infants **appear normal at birth** but can have delayed meconium passage (moderate amounts of **maternal hormones** in the infant's circulation → **mandatory screening** by ↓ free T4 ↑ TSH)

Weeks - months after birth: Apathy, weakness, hypotonia, large tongue, sluggish movement, abdominal bloating, and an umbilical hernia. Signs: pathologic jaundice, difficult breathing, noisy respiration, hypothermia, and refractory macrocytic anemia. Constipation, dry skin, puffy face, lethargy, poor feeding, enlarged fontanelle.

Treatment	• Levothyroxine (Avoid soy products, iron, or calcium)
Prognosis	• No deficits if treatment started in neonatal period • Untreated disease is associated with neurocognitive dysfunction (↓ IQ)

Craniopharyngioma (CT/MRI: Calcified cystic supracellar mass, surgery + radiation)	
Epidemiology	• Benign, slow growing. Derived from remnants of Rathke pouch • Bimodal age distribution (age 5-14 & 50-75),
Clinical features	• Optic chiasm compression → bitemporal hemianopsia • Pituitary stalk compression → endocrinopathies ○ Growth failure in children (↓ TSH or ↓ GH). Pubertal delay in children or sexual dysfunction in adults (↓ LH & FSH). Diabetes insipidus (↓ ADH)
Most common childhood supratentorial tumor	

DKA in children (URI precipitates DKA due to increased catecholamine, cortisol, and glucagon release. Complication: cerebral edema)	
Clinical features	• Polyuria/nocturia. Polydipsia, polyphagia. Vomiting, abd pain • Weight loss, fatigue, dehydration. Kussmaul respirations (deep, rapid breathing)
Laboratory findings	• Glucose >200 mg/dL. Bicarbonate <15 mEq/L. pH <7.3. AG >14 Serum/urine ketones • Glucosuria, osmotic diuresis (increased U excretion of Glc, ketones, Na, K, phosphate). Concentrated urine, depleted total body K+ stores but normal or ↑ K+ level (due to ↓ insulin). ↑FFA. Hyperosmolarity (Glc). Hypovolemia -> ↑ BUN Cr, ↑RAAS, ↑ADH
Management	• **10 mL/kg isotonic** fluid bolus **over 1 hour** • **Insulin** infusion + isotonic fluids with **potassium (to prevent hypokalemia)**

Exercise while taking insulin (Continued Glc muscle uptake [↑insulin sensitivity] + changes in skin perfusion that lead to erratic insulin absorption -> **hypoglycemia**. Reduce **premeal** insulin (if exercise within 3 hours of eating) + **basal** insulin in prolonged exercise (marathon) + **increase carbohydrate intake** (particularly if training sessions >60 mins)

MC complication in infants of diabetic mothers: hypoglycemia.

Differentiating type 1 from type 2 diabetes mellitus		
	Type 1	**Type 2**
Associated conditions	• Other autoimmune disorders (thyroiditis, celiac disease), thin	• Dyslipidemia, hypertension, **acanthosis nigricans, obesity**
C-peptide levels	• Low	• Usually elevated
Autoantibodies*	• Usually present	• Usually absent
*For example, glutamic acid decarboxylase.		

Excess adiposity/ ↑ GH in puberty contribute to **insulin resistance**. Glucokinase mutations cause monogenic diabetes (MODY), in which mildly defective glucose sensing results in

relatively low insulin secretion for any given glucose level. Unlike in T2DM, hyperglycemia is mild (<140 mg/dL), and no signs of insulin resistance.

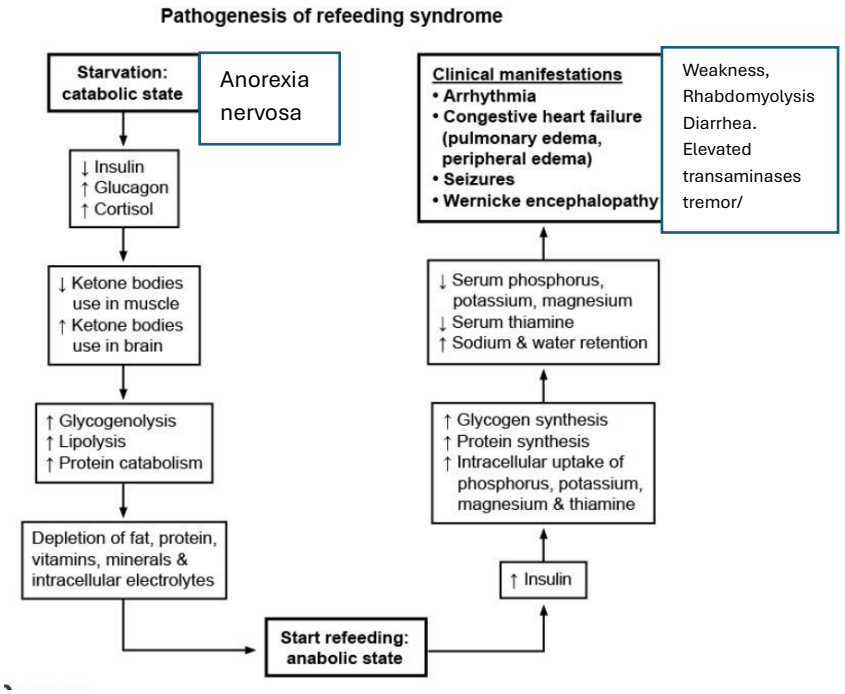
MEN2B: Medullary thyroid (prophylactic thyroidectomy in infancy), pheochromocytoma (metanephrines), marfanoid habitus, mucosal neuroma (painless rubbery tongue nodules).

Refeeding syndrome can occur when nutrition is reintroduced following prolonged **mal**nutrition (not TPN).

PO_4 is the most deficient (required for ATP). ↓ K Mg = cardiac arrythmia in an atrophic heart (due to malnutrition)

Aldoesterone elevated in starvation. Glucagon decreased when feeding.

Anorexia nervosa: euthyroid hypothyroxinemia, normal TSH and normal to decreased T3 T4



Gaucher disease (AR, presents at any age. Tx: glucocerebrosidase replacement)	
Clinical features	• Hepatomegaly, splenomegaly(Severe), pancytopenia, osteoporosis, avascular necrosis, bone crises. FTT delayed puberty
Notes	• Most common Lysosomal storage disease, lipid laden macrophages

Glucose-6-phosphatase deficiency (type I glycogen storage disease, von Gierke disease) age 3-4 months: Severe fasting **hypoglycemia** (**seizures**), hepatomegaly (protuberant abdomen), renomegaly, **lactic acidosis** (due to buildup in the liver) ↑ triglycerides, uric acid. PEx: **Doll-like** face with rounded cheeks, thin extremities, short stature, and a protuberant abdomen due to **hepatomegaly**. The spleen and heart are not involved.

MCAD: Episodes of hypoketotic hypoglycemia during fasting states (eg, illness).

Classic Galactosemia (1st few days of life. ↓ **RBC** GALT, AR, milk, Tx: soy-based formula)	
Clinical findings	• FTT (vomiting), jaundice hepatomegaly infantile cataracts intellectual disability ↑ E.coli sepsis (↓leukocyte function and superoxide release)

Laboratory findings	• ↑ **Unconjugated** or conjugated bilirubin, AST, ALT. ↓ Glucose (seizures). Metabolic acidosis (renal tubular damage). + Urine reducing substance.

With treatment, end-organ dysfunction typically resolves, and the prognosis is good.

Phenylketonuria (AR, Phe hydroxylase. Tx: avoid protein rich foods, supplemental/ dietary tyrosine (soy products, chicken, fish, milk)	
Features	• Severe intellectual disability, developmental delay, microcephaly , seizures, hypopigmentation (skin, hair, eyes), eczema, musty body odor
Diagnosis	• ↑ Phenylalanine levels: Universal newborn screening (day 2-3 of life) (on tandem mass spectrometry). Serum AA analysis
Prognosis	• **Preventable** disease manifestations with early treatment • Delayed diagnosis/treatment → **irreversible** neurologic injury

Constitutional delay of growth and puberty (IMPORTANT!)	
Clinical features	• Family history of "late bloomers" • Short stature; **normal growth velocity** • Delayed puberty. **Delayed bone age**
Management	• **Reassurance**; watchful waiting ± Hormone therapy
Prognosis	• Puberty onset correlates with family members. Normal expected adult height

Linear growth velocity starting at **age ≥3**. Affected patients typically have a **normal birth weight and height** but show a **slowed linear growth velocity between age 6 months to 3 years**. They **subsequently (eg, age ≥3) grow at a normal rate** (4-6cm/yr), so their growth curve remains **below but parallel to the lowest percentile line** (ie, linear growth curve).

Familial short stature: Short **Mid Parental Height**. **Bone age = chronologic age**, puberty is **not** delayed. Normal linear growth velocity.

(Choice A) Celiac disease: Short stature, WL (Type 2 FTT) delayed bone age.

(Choice D) GH deficiency: **Short stature, delayed puberty, delayed bone age. ↓ linear growth velocity** (declining height percentiles crossing 2 major percentiles).

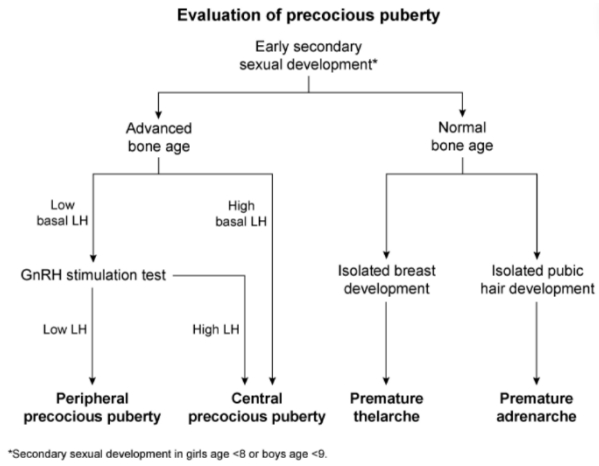
(Choice F) Hypothyroidism: Short stature, **delayed** puberty, delayed bone age, ↓ linear growth velocity, **fatigue, constipation, or cold intolerance**.

(Choice E) Hypogonadism: delayed puberty, **normal bone age**, gynecomastia.

Kallman: X-LR; but can be sporadic, AD, AR. Short stature, '**Rhinencephalon hypoplasia'**

(Choice D) Patients with a **47,XXX** genotype typically have **normal** secondary sexual characteristic development and **tall stature**. **FSH and LH levels are normal**.

Nonclassic CAH: Peripheral precocious puberty (**pubic/axillary hair**, **cystic acne**, accelerated growth -> **tall in childhood, short in adulthood**), sufficient glucocorticoid and mineralocorticoid levels are maintained -> **normal electrolytes** (no salt wasting).



McCune-Albright syndrome: **Irregular** café-au-lait macules [confined to 1 side of the body], polyostotic fibrous dysplasia [recurrent bone fx], peripheral precocious puberty. Can lead to thyrotoxicosis (TSH), acromegaly (GH), and Cushing syndrome (ACTH). NF1 café au lait are more numerous (≥6) with regular borders.

Central PP next step: MRI brain, if excluded, diagnosis is idiopathic precocious puberty, tx: **GnRH agonist** (prevents premature epiphyseal plate fusion & ↑ adult height potential).

Isolated **premature adrenarche**: Early activation of **adrenal androgens**. Typical manifestations: body odor, oily skin, **acne**, **pubic and axillary hair**. Mildly ↑ DHEA, normal estrogen and testosterone levels -> NO breast development, testicular enlargement, or clitoromegaly. Generally benign but a risk factor for **developing PCOS, type 2 DM, and metabolic syndrome**, especially in obese patients.

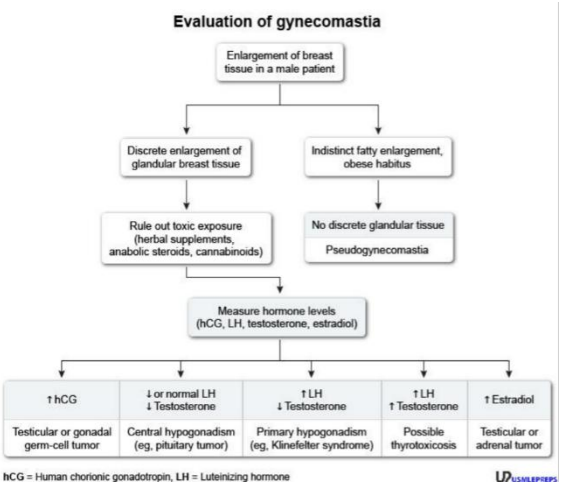
Classic congenital adrenal hyperplasia	
Clinical presentation	• Ambiguous genitalia in girls. Salt-wasting syndrome* (Affects most girls & boys: Hypotension, dehydration & vomiting)
Laboratory findings	• ↓ Na, ↑ K, ↓ glucose. ↑ 17-Hydroxyprogesterone
Treatment	• Glucocorticoids & mineralocorticoids. High-salt diet. Psychosocial support
*Clinical symptoms & electrolyte abnormalities **develop at age 1-2 weeks**. Before that only **ambiguous genitalia, increased testosterone and hypoglycemia**	

In the first few days of life, serum electrolytes are normal due to the presence of **maternal adrenal hormones.**

(Choice E) Sertoli-Leydig cell tumors in **early adulthood**, not the newborn period.

↑ **hCG** by testicular germ cell tumors → ↓ **testosterone** production in Leydig cells → ↑ **aromatase activity** → conversion of **androgens to estrogens**→ ↑ **estrogen/androgen ratio**.

Physiologic gynecomastia: during pubertal **age 12-14** in midpuberty. **Tanner stage 3-4**, develops **gradually**, **resolves within a year**, no discrete glandular tissue, well-circumscribed **small firm uni or bilateral subareolar mass**, no nipple discharge, axillary LN, systemic illness.



Anabolic steroids, marijuana, alcohol can cause gynecomastia that is always **bilateral**.

Chapter 6: Reproductive

8 yo, unilateral, firm, tender mass posterior to the nipple = physiologic **thelarche**, the **first sign of puberty** in most girls (breast buds: enlarged areola, elevated papilla, small mound of breast tissue). Rarely, adrenarche precedes thelarche. Pubertal changes in girls typically begin around age 10 and **as early as age 8**. Reassurance. Menarche 2.5 years later.

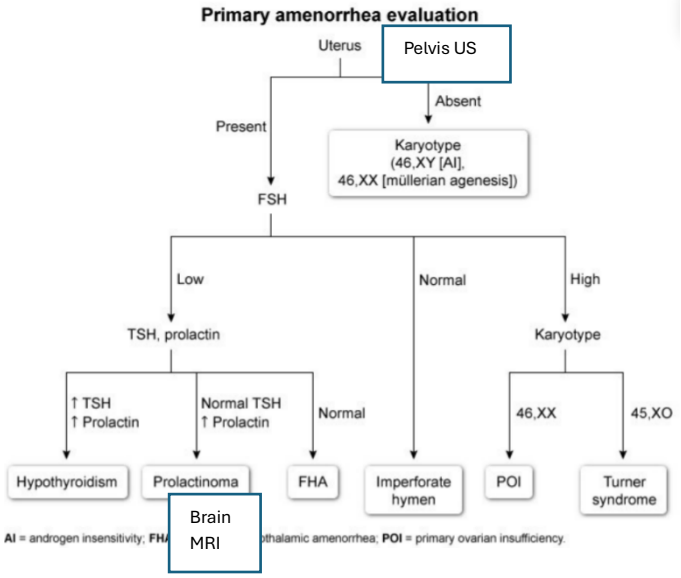
- Breast fat necrosis **firm, irregular, nontender** mass
- Fibrocystic change = cyclic premenstrual tender breast

Fibroadenoma (Adolescent girls & women age <30)	
Clinical features	• Unilateral, firm, mobile, well-circumscribed, nontender mass in UO quadrant. • Cyclic changes with menses (eg, premenstrual breast tenderness, size change)
Management	• Observation & repeat examination in adolescents • US in adults or patients with persistent/enlarging mass

Adolescents **benign & Self limited**. Adults higher baseline risk of malignancy

Breast self-examination is no longer recommended for breast cancer screening in any age group; it increases the rate of false-positive results and unnecessary procedures (eg, biopsy) and does not affect outcomes.

Primary amenorrhea is the absence of menarche at **age ≥13** in girls with **no secondary sexual characteristics** or age ≥15 with secondary sexual characteristics.



In patients **who do not have a uterus**, **serum testosterone levels** are measured to evaluate for **androgen insensitivity syndrome** (46,XY and **high** [ie, normal male] testosterone) and **müllerian agenesis** (46,XX and **low** [ie, normal female] testosterone).

TS does not affect adrenal glands testosterone production → **normal adrenarche** occurs → develop pubic and axillary hair. Next step: **Karyotyping**, if normal, do **FISH** to detect mosaicism. (Low testosterone → short stature, some pts have learning disorder/ADHD)

Androgen insensitivity syndrome (X-linked)	
Clinical features	• Genotypically male (46,XY karyotype). Phenotypically female • Breast development. Absent or minimal axillary & pubic hair. Female external genitalia. Absent uterus, cervix, & upper one-third of vagina. Cryptorchid testes
Management	• Gender identity/assignment counseling • Gonadectomy (malignancy prevention)

Timing of the procedure is based on possible current malignancy, symptoms (eg, pain), and the potential benefits of androgen-stimulated puberty (eg, attainment of adult height).

Neonatal withdrawal bleeding (MCC of vaginal discharge in neonates)	
Physiology	• In utero: maternal estrogen → fetal **endometrial proliferation** • After delivery: withdrawal of maternal hormones → endometrial sloughing in neonate (**light vaginal bleeding** in the 1st 2 weeks of life),

	physiologic leukorrhea (thickening of vaginal epithelial cells), **labial swelling** (vulvar vasocongestion), **breast hypertrophy** (↑ mammary gland proliferation), **galactorrhea**. Clinical dx, **reassurance**, self limiting within days.

Primary dysmenorrhea (↑PG production, Tx: NSAIDS[inactive], COCP[sexually active])	
Risk factors	Age <30, BMI <20, menarche age <12, heavy/long menstrual periods, smoking, sexual abuse
Clinical features	Crampy bilateral lower abd pain, N V, diarrhea. Normal pelvic examination begins 1-2d prior to menses, and resolves 2-3d after **onset** of menses

Abnormal uterine bleeding (AUB), defined during adolescence as menstrual bleeding <21 days or >45 days apart. HPO immaturity: **1st few years post menarche**, **anovulatory** cycles + **painless, irregular, heavy bleeding**.

AUB mx: Hemodynamically stable: High dose OCP, unstable PRBC + D&C.

Vaginal foreign bodies in children (prepubertal, MC= toilet paper: friable white object)	
Clinical features	• Vaginal spotting. Malodorous/purulent vaginal discharge. No signs of trauma (eg, lacerations). Vulvovaginitis
Management	• Topical anesthetic & warmed fluid irrigation + removal (Small, easily visualized) • Vaginoscopy under anesthesia (can't be visualized, large, button battery causing burns, anxiety, lack of cooperation, age <5)

When a vaginal foreign body is suspected, an external pelvic examination is performed with the patient in a knee-chest or frog-leg position.

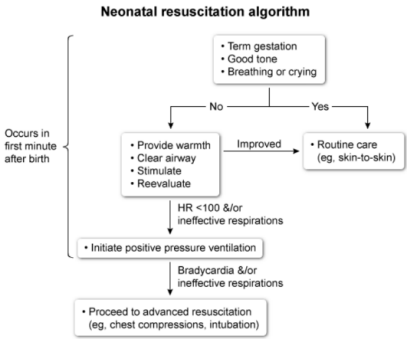
(Choice B) A vaginal foreign body can be the initial presentation of sexual abuse. However, the presence of what is likely toilet paper does not immediately raise concern for abuse, particularly in an otherwise asymptomatic patient (eg, no behavior changes, no signs of vulvar trauma) whose mother sought medical care on the first day of symptom onset.

(Choice D) Speculum examination should not be performed in a prepubertal girl. It can result in significant discomfort and trauma due to the narrow vaginal introitus and sensitive hymenal tissue from a low estrogen level at this age. Even when under anesthesia, prepubertal girls should be evaluated with vaginoscopy, not speculum examination.

FGR management (monitor/ treat complications)	• Hypoglycemia: **frequent screening** and frequent feedings • Hypothermia: **skin to skin** with mother, examinations in **incubator** • **Polycythemia and hypocalcemia**: screen if symptoms develop (eg, poor feeding, vomiting, jitteriness) • **Perinatal asphyxia and meconium aspiration**

(Choice C) Magnesium freely crosses the placenta; therefore, the fetus's serum concentration equals the mother's serum concentration. Infants born to mothers receiving magnesium sulfate for either seizure prevention or fetal neuroprotection have hypermagnesemia at birth, resulting in respiratory depression and hypotonia.

Newborns who do not need resuscitation should be **placed on the mother's chest** for at least 1 hour of **skin-to-skin contact** to provide warmth and promote early initiation of breastfeeding, after 1 hr give VitK and erythromycin. HB vaccine in the 1st 24hrs.



(Choice C) Oropharyngeal suctioning may be necessary for excessive airway secretions; however, **endotracheal suctioning is not recommended** in routine neonatal resuscitation due to the risk for vagal stimulation and subsequent **apnea or bradycardia**.

Fetal hydantoin syndrome: (**phenytoin**, carbamazepine, valproate) : **cleft lip and palate**, microcephaly, wide anterior fontanelle, developmental delay, NTD, **distal phalange hypoplasia**, and cardiac anomalies (eg, pulmonary stenosis, aortic stenosis).

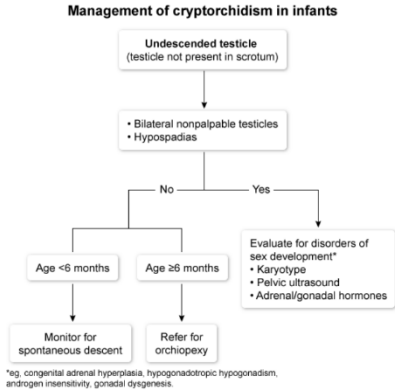
Neonatal displaced clavicular fracture	
Risk factors	• Maternal DM, post term, vacuum/forceps, shoulder dystocia
Clinical features	• Crying/pain with passive motion of affected extremity. Crepitus over clavicle. Asymmetric Moro reflex. Dx: X-ray
Treatment	• Reassurance. Gentle handling. Analgesics (self limiting 7-10d) • Place affected arm in a long-sleeved garment & pin sleeve to chest with elbow flexed at 90 degrees

- Shoulder dislocation, closed reduction, NO clavicular crepitus.
- Traumatic clavicular fx: Displaced fractures: surgically reduced. Nondisplaced fractures: figure-of-8 splint.

- MC risk factor for shoulder dystocia: fetal hyperglycemia (due to maternal DM

Because **concomitant anterior urethral injury** can occur with penile trauma, any of the following should prompt evaluation for urethral injury (retrograde urethrography, if normal, oral analgesics and outpatient follow-up):

- Blood at the urethral meatus
- **Hematuria** (eg, pinkish-red urine)
- Difficulty voiding (due to disruption of urethral continuity)



(Choice B) Posterior urethral injury: pelvic fx, high-riding prostate on exam (prostate exam rarely done in children)

(Choice F) Testicular rupture: significant testicular pain/tenderness, swelling, impaired cremasteric reflex. The most common indication for testicular ultrasound.

Hypospadias	
Management	• Defer circumcision. **Urologic** evaluation for surgical repair • ± **Karyotype** (DSD: 10% of cases associated with cryptorchidism: undervirilization in males, virilization in females. Even if testis is present (Y chromosome), do karyotyping in severe hypospadias to evaluate for mosaicism), **pelvic ultrasound** (if severe)

(Choice E) **Prenatal** screening for neural tube defects (NTDs) includes a maternal serum alpha-fetoprotein (**AFP**). However, **ultrasound** (not AFP) is performed in the **newborn** with findings concerning for NTDs (eg**, deep/large sacral dimple without a base**). A sacral dimple with intact skin is a benign finding that does not require further workup.

(Choice F) Renal ultrasound for hypospadias + aniridia (WAGR Syndrome)

Cryptorchidism (Empty poorly rugated scrotum on affected side(s) + inguinal fullness/mass. Complications: Inguinal hernia, infertility, testicular torsion, testicular CA)	
Risk factors	• Prematurity. Small for gestational age
Management	• Surgical referral at age ≥6 months. Orchiopexy before age 1 (complications reduced to the **same rate as the general population** except seminoma [in both testes])

Noncommunicating hydrocele is formed if the PV **obliterates normally** but fluid collected within the tunica vaginalis **does not reabsorb**. **Irreducible and constant size**.

Communicating hydrocele: **Painless** unilateral or bilateral **scrotal swelling**. The swelling may **fluctuate** with crying or straining (increased intra-abdominal pressure).

Both hydroceles +transillumination and resolve spontaneously by 1 year. If persists, surgery

Balanitis & balanoposthitis (glans + foreskin) : erythema pruritis discharge, uncircumcised children age (2-5 yo)	
Etiologies	• Poor hygiene (MC), contact dermatitis. Candida, bacterial flora, STI. Aggressive foreskin retraction
Evaluation	• Consider KOH microscopy for suspected *Candida* infection (eg, thick, white discharge). STI screen if urethral discharge present
Management	• Foreskin hygiene, sitz baths. Topical antifungal/ab. DM screen for candida without risk factor (diaper dermatitis, recent ab use)

Chapter 7: Renal

Primary nocturnal enuresis (age ≥5, **never achieved** ≥6 months of **dryness** overnight)	
Pathogenesis	• Delayed maturation of bladder control/ ↑ Nocturnal urine output (eg, ↑ evening fluids, ↓ ADH)/ ↓ Bladder capacity
Risk factors	• Family history (greatest RF). **Boys (x2 more likely than girls)** age 5-8, ADHD (Minor, due to delayed brain maturation + exacerbation of inattentive and hyperactive behaviors due to sleep disturbance)
Evaluation	• **Urinalysis** (to exclude other causes). Voiding diary
Management	• Treatment of constipation, restrict evening fluids → enuresis alarm → Desmopressin therapy → If all failed can give imipramine

(Choice C) Emotional stressors (eg, parental conflict, bullying) are often associated with **secondary enuresis** (ie, bed wetting after establishing nighttime continence).

Bedwetting is normal before age 5. Mastery of nighttime continence can take **months to years**, and **boys generally achieve this milestone later than girls.**

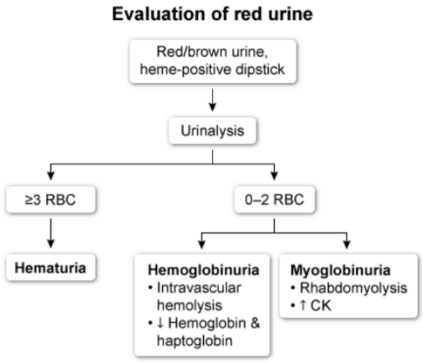
Medical conditions causing enuresis [*Often a clinical diagnosis.]		
Etiology	**Symptoms**	**Findings**
Constipation*	• Infrequent & hard stools • Encopresis	• ± Palpable stool mass. Tx: laxatives
Bladder dysfunction*	• Daytime incontinence. Weak stream, urgency, **straining**	• ± Recurrent urinary tract infections
Urinary tract infection	• Dysuria, urgency, frequency • Abdominal pain	• Positive urine culture
Chronic kidney disease	• Daytime incontinence. **Weight loss, fatigue**, recurrent UTI • **MCC in children with PUV**	• Hypertension. Proteinuria, hematuria. Next step: Check Creatinine
Diabetes mellitus	• Polyuria, polydipsia, polyphagia. **WL, fatigue**	• Glucosuria. Type 1 DM onset ages 4-6 & 10-14. DKA = fruity breath
Diabetes insipidus	• Polyuria, polydipsia	• Low urine specific gravity
OSA	• Snoring. Hyperactivity, inattention	• Adenotonsillar hypertrophy

Posterior urethral valves are the MCC of **urinary tract obstruction in newborn** boys AND **CKD in children**.

Nocturnal enuresis in children		
	Primary	**Secondary**
Definition	• Nighttime incontinence at age ≥5 without prior prolonged period of continence	• Nighttime incontinence at age ≥5 after prolonged period of continence
Causes	• Brain maturation delay + Genetics (eg, family history)	• Underlying medical condition + Psychological stressors

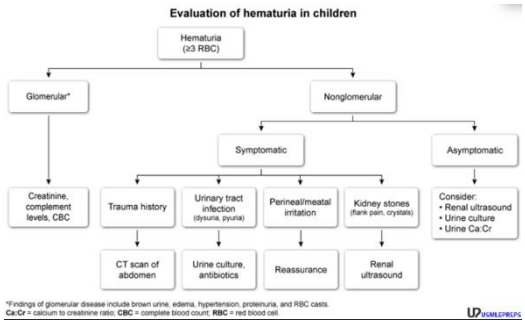
Initial evaluation	• Urinalysis	• Urinalysis
Management	• Reassurance. Behavioral modifications (eg, evening fluid restriction). Bedwetting alarm	• Treatment of underlying condition+ Behavioral modification

Significant exertion (eg, **long-distance running**) can lead to **dark urine** due to the following:



- Benign hematuria from bladder trauma (repeated **collision** of the **bladder wall** and **base** while running, or the bladder and the perineum during a bumpy bike ride)
- Myoglobinuria from SM injury (ie, **rhabdomyolysis**)
- March hemoglobinuria (**mechanical (RBC) damage**/ hemolysis in the vasculature of the plantar surface vasculature of the feet) (rare)

Testing not needed in classic history and no (hypertension, proteinuria, elevated creatinine). **Self limiting within a week of discontinuing the triggering exercise**.



Prosthetic heart valve: red/brown urine that is **heme (+) but (-) for RBC**-> do **CBC** to evaluate prescence and severity of **anemia**.

Isolated gross hematuria in children: meatal irritation, which is usually painful and may be visible on external examination, and **nephrolithiasis (in young children. Older children have flank/abd pain,** urinalysis may be otherwise unrevealing because characteristic crystals are not always present, **DO renal and bladder US)**.

A major risk factor for nephrolithiasis is **hypercalciuria**, which may be suspected because of a previous history of **microscopic hematuria** and confirmed with an **elevated spot urine calcium/creatinine ratio**.

Primary vesicoureteral reflux in children	
Dx	• Renal US: hydronephrosis. VCUG: ureteral filling ± dilated collecting system
Mx	• Mild (grade I-II)*: observation ± prophylactic ABs, resolution by age 5

	• Severe (grade III-IV)*: prophylactic antibiotics ± surgery. Renal scarring and CKD if untreated
*Plus screening/tx of concomitant bladder & bowel dysfunction (stool softeners/laxatives).	

POTTER sequence causes weight gain in the 1st few days of life (retained urine), instead of the normal weight loss **(<10% weight change at day 2, regaining it by day 10-14**).

Minimal change disease (Kidney bx only in atypical cases, Tx: steroids)	
Epidemiology	• Most common cause of nephrotic syndrome in children
Pathogenesis	• T-cell–mediated injury to podocytes (IL-13)→ ↑ permeability to albumin
Clinical features	• Edema (eg, periorbital, scrotal, generalized). Fatigue headache ± Abdominal pain (nonspecific symptoms)
Prognosis	• Most achieve remission, but relapse is common • Low risk of CKD if episodes are steroid responsive

PSGN (**supportive** tx, self limiting within several weeks in children, adults more likely to develop CKD ± **furosamide ± nifedipine ±hemodialysis** in refractory cases) (Tea or cola colored urine [vs pink/red in lower urinary tract bleeding], can be asymptomatic)	
Laboratory findings	Positive **streptozyme test** (antistreptolysin O, anti-DNAse B, anti–NAD, antihyaluronidase, antistreptokinase).

1. **ICs** are deposited between the (GBM) and the mesangium (ie, subepithelial).
2. **Complement activation** causes leukocytic infiltration and

	IgA nephropathy	APSGN
Onset	Several days after upper respiratory tract infection or spontaneous Most common in young adults	Weeks after group A *Streptococcus* infection (eg, pharyngitis, impetigo) Most common in school-aged children
Pathogenesis	IgA-based immune complex deposition in mesangium	Immune complex deposition along GBM (ie, subepithelial)
Clinical features	Gross hematuria (often recurrent) ± Flank pain, low-grade fever, hypertension May be asymptomatic	Gross hematuria Edema (periorbital or generalized) Hypertension May be asymptomatic
Laboratory findings	Urinalysis: + protein, RBCs, RBC casts Serum: ○ Normal C3 & C4 ○ ↑ Creatinine	Urinalysis: + protein, RBCs, RBC casts Serum: ○ ↓ C3 & CH_{50} ○ ↓ Or normal C4 ○ ↑ Creatinine

inflammation, resulting in a **thickened GBM**, decreased GFR -> elevated Cr **edema**, **hypertension**.

(Choice D) Renal vein thrombosis: hematuria proteinuria severe flank pain nausea, rare in children; risk factors: **nephrotic syndrome, central venous catheters, and malignancy.**

Initial **evaluation** of **glomerulonephritis** includes **serum complement (C3, C4) levels.** Low C3: PSGN & lupus nephritis -> do ANA/ASO, if unrevealing, do kidney biopsy.

Membranous nephropathy (Adults: common/primary. Children: rare/secondary)	
Prognosis	• Partial/full remission common (spontaneous or treatment induced) • End-stage kidney disease rare

HBV: Membranous nephropathy is caused by immune complexes formed from **(HBeAg) and anti-HBeAg**. Evaluating all patients with MN with HBV serologies is recommended.

Common causes of neonatal acute kidney injury (oliguria)	
Prerenal disease	• Hypovolemia. Cardiac disease. Sepsis
Intrinsic renal disease	• Acute tubular necrosis (eg, ischemic injury) • Nephrotoxins (eg, maternal NSAID use, aminoglycosides) • Renal vascular disease (eg, **renal vein thrombosis**) • Glomerular & cystic disease (eg, polycystic kidney disease) • Congenital anomalies (eg, **renal agenesis**)
Postrenal disease	• Obstructive uropathy (eg, posterior urethral valves)

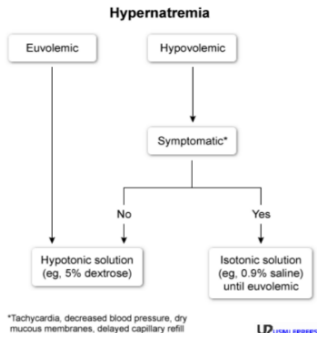
Normal UO in a neonate is defined as ≥1 void per day of life. All neonates with oliguria **require renal and bladder US**. If pre-renal give IV NS bolus. However, establishing normal renal anatomy is required first because in neonates with obstructive uropathy or renal dysplasia, a fluid bolus could lead to volume overload.

NV myalgia fatigue. Anemia (eg, fatigue, pallor), thrombocytopenia (eg, ecchymoses), and leukocytosis with immature myeloid forms = acute myelogenous leukemia (AML).

Concurrent hyperkalemia, hyperuricemia, hyperphosphatemia (tumor cells have >3 times more intracellular phosphate than healthy cells), ↑ (LDH), (AKI, from **stones obstructing renal tubules**) (with resultant acidosis) raises suspicion for **spontaneous** (TLS). Although

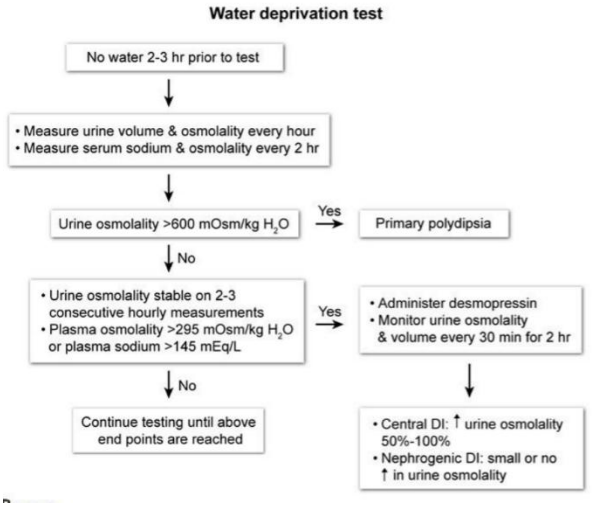
most cases of TLS occur following initiation of cytotoxic chemotherapy, some **hematologic malignancies** (eg, AML) with high tumor cell burdens or rapid replication rates can cause TLS in the absence of chemotherapy due to massive, spontaneous tumor cell lysis.

Tx: **Aggressive IV hydration**, **uric acid reduction** (rasburicase, allopurinol), and **treatment of electrolyte abnormalities** is required to prevent long-term sequalae and death.



Infants are particularly at risk for developing **hypernatremia with a GI illness** due to greater insensible losses (↑ TBSA) and the inability to communicate thirst. **Signs of hypernatremia**: irritability, muscle weakness, ↓ DTR, and seizures in severe cases.

Dilute urine (urine osmolality <300 mOsm/kg H_2O, urine specific gravity <1.006): DI (MCC in young children = hereditary nephrogenic diabetes insipidus) and primary polydipsia (psychiatric or CNS disorders) -> **water deprivation test**.



The water deprivation test is performed under hospital supervision in older children and should not be used in very young children due to the risk of severe hypernatremia.

Central DI: polyuria, hyposthenuria, **impaired thirst mechanism, ↑ serum Na**

Primary (psychogenic) polydipsia: hyposthenuria, **↓ serum Na, improves on fluid restriction.** Primary polydipsia can be diagnosed **without additional testing** in patients with both **polyuria and hyponatremia**. However, **water deprivation test** mandatory if **normal serum Na**. Treatment consists of decreasing water intake (ie, "drink to thirst" only).

ADH/AVP-related causes of polyuria (UO>3L/day) & polydipsia (All low UNa)			
	Primary polydipsia	**AVP deficiency (central DI)**	**AVP resistance (nephrogenic DI)**
Defect	• ↑ Water intake	• ↓ ADH release from pituitary gland	• Renal ADH resistance (normal or ↑ ADH)

Etiology	• Antipsychotic use, anxiety, psychiatric condition	• Idiopathic. Trauma. Pituitary surgery. Ischemic encephalopathy	• Lithium, hypercalcemia. Hereditary (*AVPR2* mutation)
Clinical features	• Low serum Na	• High-normal or elevated serum Na*	• Normal or mildly elevated serum Na*
*Na is elevated if thirst mechanism is impaired (eg, hypothalamic involvement).			

Classification of hyponatremia			
Serum osmolality	**ECV**	**Urine findings**	**Causes**
Hypotonic (<275 mOsm/kg)	Hypovolemic	U_{Na} <40 mEq/L	• Nonrenal salt loss (eg, vomiting, diarrhea)
		U_{Na} >40 mEq/L	• Renal salt loss (eg, diuretics, primary adrenal insufficiency)
	Euvolemic	U_{Osm} <100 mOsm/kg	• Primary polydipsia • Malnutrition (eg, beer potomania)
		U_{Osm} >100 mOsm/kg & U_{Na} >40 mEq/L	• SIADH*
	Hypervolemic	Variable	• Heart failure, cirrhosis, nephrotic syndrome
Isotonic	Variable		• Pseudohyponatremia (eg, paraproteinemia, hyperlipidemia)
Hypertonic (>295 mOsm/kg)			• Translocational hyponatremia (eg, hyperglycemia, exogenous solutes such as mannitol)

*Hypothyroidism & secondary adrenal insufficiency should be ruled out.

- MDMA (ectasy) can cause SIADH.
- Normal AG: 10-14 mEq/L

Urinary tract infection (UTI) in children	
Risk factors	• Female sex. Uncircumcised male. VUR, constipation

Management	• Antibiotics (eg, 3rd generation cephalosporin) for 1-2 wks
	• **First febrile** UTI:
	  ○ **Age <2**: **RBUS**, followed by VCUG if abnormal
	  ○ **Age ≥2**: **observation** alone
	• **Recurrent febrile UTIs**: RBUS & VCUG

❖ **Pyuria** (white blood cells ≥5/hpf) + **bacteriuria** (≥50,000 colony-forming units/mL from a catheterized specimen) are diagnostic of UTI.

- **Indications for VCUG: ≥2 febrile UTIs; abnormal renal ultrasound; fever ≥39 C with bacteria other than *E coli*; signs of chronic kidney disease (poor growth, HTN).**
- The ultrasound should be performed after fever and symptoms have resolved to minimize false positive results from acute inflammation. If the patient has **persistent /worsening** symptoms, or doesn't improve with 2 days of Antibiotics: an **ultrasound** should be performed **immediately** to assess for renal **abscess** +broader spectrum Ab.
- In **UTI with hematuria**, **repeat urinalysis** a few weeks after completion of Abs. UTI without initial hematuria can typically be followed clinically for resolution of symptoms.
- **Repeat CBC and urine culture** should be performed only in children **who fail to improve after 2-3 days** of appropriate antibiotics. .

Antibiotic administration before obtaining urine studies is **not recommended** due to the risk of antibiotic resistance and the potential for unnecessary therapy.

(Choice A) Addison: generalized hyperpigmentation (most prominent in pressure- or sun-exposed areas such as the elbows and knees) + anorexia, weight loss

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Functional constipation -> urine stasis -> recurrent cystitis.

The definitive diagnosis of VUR is made by contrast VCUG. Renal ultrasound is performed to screen for hydronephrosis. Renal scintigraphy with DMSA is the preferred modality for long-term evaluation for renal scarring. Renal function should be followed by serial creatinine. Patients should be monitored closely for complications of **chronic renal insufficiency**, such as hypertension and anemia.

(Choice F) Most patients with a unilateral renal agenesis are asymptomatic.

Wilms tumor (nephroblastoma): (MC renal malignancy in children, peak age (2-5), arises from metanephros, 90% survival rate with tx. 1st step: **Abd US THEN CT or MRI**)	
Epidemiology	• Usually sporadic but may be associated with: ○ **WAGR** (Chromosome 11, bilateral, screening abd US every 3 months in infancy & early childhood for early detection & tx). **Beckwith-Wiedemann syndrome**. **Denys-Drash syndrome**
Clinical features	• Usually asymptomatic. Unilateral abdominal mass ± Abd pain, hypertension (renin secretion by tumor/ compression of renal vessels), hematuria
Treatment	• Surgical excision. Chemotherapy ± Radiation therapy

MC site of metastatic spread: lungs, but children rarely have pulmonary symptoms.

(Choice A) Neuroblastoma: children age <2, neural crest cells (eg, adrenal glands).

Beckwith-Wiedemann syndrome (Wilms tumor/hepatoblastoma, macrosomia (rapid growth until late childhood) macroglossia hemihyperplasia organomegaly omphalocele/ umbilical hernia, hypoglycemia). **Overexpression of IGF-2 (WT2 mutation)**	
Surveillance	• **Abdominal US** and **AFP** level testing should occur every 3 months from birth to age 4 and **renal ultrasound** every 3 months from age 4-8.

Varicocele		
	Primary	**Secondary**
Pathophysiology	• Compression of **left renal vein** between SMA and aorta • **Incompetent venous valves**	• Extrinsic compression (renal or retroperitoneal mass) of **IVC (HTN)** • **Venous thrombus**
Clinical features	• "Bag of worms" mass • **Pubertal** onset • **Left**-sided • **Decompresses** when supine	• "Bag of worms" mass • **Prepubertal** onset • **Right**-sided • **Persists when supine**
Initial mx	• Reassurance and observation	• Abdominal **ultrasound**

Chapter 8: Gastrointestinal

Neonatal weight trends	
Natural course	• Weight loss in first 5 days of life (excretion of excess fluid, delayed lactogenesis). Weight regained (back to birth weight) by age 2 weeks
Management	• ≤10% of birth weight lost: Provide reassurance & continue to monitor • >10% of birth weight lost &/or signs of dehydration (poor urine output, sunken fontanelle): Assess for **breastfeeding complications** (eg, tongue-tie). **Measure weights daily**. Consider **formula**

	supplementation. Consider **serum electrolytes & glucose measurements**

The purpose of the initial **well-child visit** is to assess overall **newborn** health and provide anticipatory guidance. Specific aspects of newborn health include the following:

- **Feeding:** Newborns **should be fed on demand** but typically ≥**8 times a day**. **Exclusively breastfed** neonates feed more frequently (**sometimes every hour**) compared to formula-fed infants due to **more rapid digestion of breast milk**. Each breastfeeding session is **usually ≥15 minutes per breast**. Parents may need to gently rouse the patient (tickle feet) midfeed.
- **Elimination: The number of daily wet diapers should be at least the newborn's age** in days (eg, ≥3 wet diapers for a 3-day-old patient) **through the first week**. Stool frequency may vary, but bowel movements should transition **from tarry meconium to yellow, seedy stools in the first few days.**
- **Weight: Loss ≤10% of birth weight** is **physiologic** and likely due to excretion of excess fluid. Weight loss may be more pronounced in **exclusively breastfed** newborns due to delayed lactogenesis; normally, **maternal milk production increases by day 3-4 postpartum but may take longer**. In the first few days after birth, breast milk is **colostrum, an antibody-rich fluid that is low in volume.**

(Choice B) Breast milk fortification may be recommended for premature or low-birth-weight newborns who require higher-caloric-density feeds to gain weight.

Almost all newborns on **days 2-4 of life** have **physiologic jaundice** due to **indirect hyperbilirubinemia**. The following physiologic differences in bilirubin metabolism account for this finding:

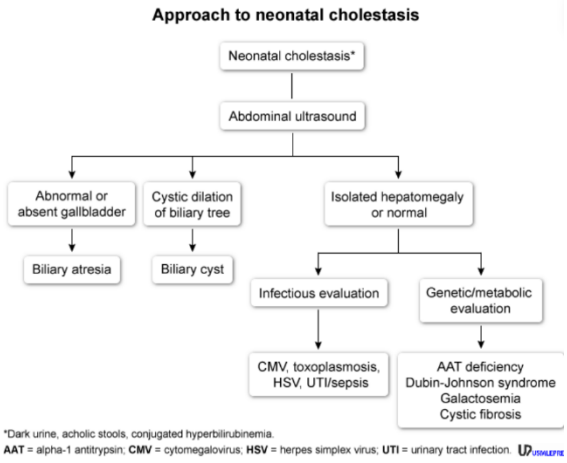
1. At birth, **fetal (RBCs)** are increased (**hematocrit 50%-60%**) with a shortened life span (**90 days**), resulting in **high RBC turnover** and **increased bilirubin production.**
2. **Hepatic bilirubin clearance is decreased** because UDP glucuronosyltransferase (**UGT**) activity does not reach adult levels **until age 2 weeks**. This hepatic enzyme **conjugates** bilirubin. **Eastern Asian** newborns have decreased UGT .
3. **Enterohepatic recycling is increased** because **low bacterial load** in the newborn gut results in **slower conversion of bilirubin to urobilinogen** for fecal excretion.

Physiologic jaundice of the newborn is **benign** and resolves by **age 1-2 weeks**. Frequent **feeding** promotes **gut colonization and fecal excretion**. **Phototherapy** may be indicated for **rapidly rising levels of bilirubin to prevent kernicterus**.

Neonatal indirect hyperbilirubinemia (1st line tx: phototherapy)	
Cause	**Examples**
↑ Bilirubin production	• Immune-mediated hemolysis (Coombs positive): Rh isoimmunization. ABO incompatibility • Nonimmune-mediated hemolysis (Coombs negative): Spherocytosis, G6PD • **Cephalohematoma.** Polycythemia
↓ Bilirubin clearance	• Gilbert syndrome. Crigler-Najjar syndrome
↑ Enterohepatic circulation	• Lactation failure jaundice. Breastmilk jaundice

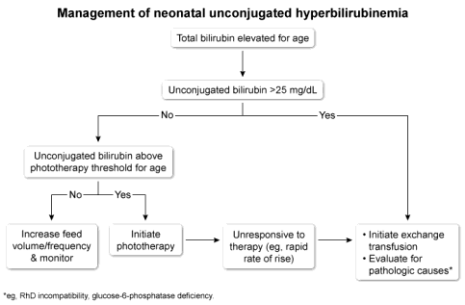
Risk factors for **severe hyperbilirubinemia**: **prematurity**, exclusive breastfeeding, **cephalohematoma** (self resolves within weeks), bruising, **jaundice at age <24 hours, ABO incompatibility**, spherocytosis, East Asian ethnicity, sibling who received phototherapy

Prematurity: immaturity of the liver (which results in decreased bilirubin conjugation) as well as decreased enteral feeds (which increase enterohepatic circulation).



Lactation failure jaundice vs breast milk jaundice			
Diagnosis	**Timing**	**Pathophysiology**	**Clinical features**
Lactation failure jaundice	Age <1 week	Insufficient intake of breast milk: • ↓ Bilirubin elimination • ↑ Enterohepatic circulation	• Suboptimal breastfeeding + Signs of **dehydration & excess WL. May benefit from cow milk based formula**
Breast milk jaundice	Age >1 wk (peaks at 2 wks)	↑ β-glucuronidase in breast milk: • ↑ Deconjugation of intestinal bilirubin. ↑ Enterohepatic circulation	• Adequate breastfeeding. **Well-hydrated** • **Patients should continue breastfeeding**

Normally, with frequent (**every 2-3 hr**) breastfeeding, maternal milk production increases by **day 5** after delivery. In contrast, **inadequate breast milk intake** can develop in the presence of **infrequent feeding (every 4 hours)** and **delayed milk production (day 6)**. **Exclusive breastfeeding** and **ineffective latch** are other risk factors.



Wet diapers typically equal a neonate's age in days (eg, ≥4 diapers on day 4, increasing to >6 daily), whereas this patient has **inadequate urine output** with only 3 wet diapers on day 5. In addition, bowel movements should transition from **meconium to frequent, yellow stools** in the first few days, and **delayed transition** can be another sign of dehydration. Tx: increase frequency of feeds with close follow-up

- **Infants require 8-12 feeds a day** (**every 2-3 hours**) for the **first few weeks of life**.
- Indications for **exchange transfusion: bilirubin level ≥25 mg/dL**, bilirubin-induced neurologic dysfunction (eg, **change in tone, lethargy**), unresponsive to phototherapy.

Biliary atresia (MCC of cholestasis/ direct hyperbilirubinemia in neonates)	
Clinical findings	• Asymptomatic at birth. Infants age 2-8 weeks: Jaundice, acholic stools, dark urine. Hepatomegaly
Diagnostic evaluation	• Direct hyperbilirubinemia • **Ultrasound**: Absent/abnormal gallbladder &/or CBD, triangular cord sign (fibrous remnants seen above the porta hepatis). • Liver **biopsy**: **Intrahepatic** bile duct **proliferation**. Portal tract **inflammation & edema**. **Extrahepatic** BD **Fibrosis** • Intraoperative **cholangiography** (gold standard): Biliary obstruction
Treatment	• Surgical hepatoportoenterostomy (Kasai procedure)+Liver transplant

Transaminases are often normal but may be elevated with advanced disease.

A (+) direct antiglobulin test confirms isoimmune hemolytic disease (ABO incompatibility)

(Choice E) Alpha-1 antitrypsin (AAT) deficiency is a rare cause of neonatal cholestasis due to the **intracellular** accumulation of AAT, but **elevated transaminases** would be expected.

Biliary cyst (Dx: Ultrasound ± CT scan or MRCP)	
Presentation	• May be incidental finding on imaging

	• Classic triad in children/adults: Abdominal pain. RUQ mass. Jaundice • Neonates: jaundice, acholic stools, dark urine, hepatomegaly • ± Nausea, vomiting
Complications	• Cholangiocarcinoma. Acute cholangitis. **Pancreatitis**. Stones
Treatment	• Cyst resection (At the time of dx to ↓ risk for malignancy) ± Roux-en-Y hepaticojejunostomy (to allow for biliary drainage)

Anomalous pancreaticobiliary junction: Abnormally long, common channel connecting the pancreatic duct & CBD outside the duodenum. **Obstruction → pancreatitis/cholestasis**

Reye syndrome	
Pathophysiology	• Aspirin use in children during viral infection (eg, influenza, varicella). Microvesicular fat deposits in the liver. **Generalized cerebral edema without focal findings**
Clinical features	• Acute liver failure (Hepatomegaly without jaundice). Rapidly progressive encephalopathy (Vomiting, lethargy, seizure, coma)
Laboratory findings	• ↑ AST, ALT, ammonia. ↑ PT, INR, PTT. ↓ Glucose. Metabolic acidosis. Normal bilirubin. Tx: supportive

Toxic-metabolic encephalopathy* & cerebral edema	
Diagnosis	**Precipitating factors**
Hypoxic-ischemic encephalopathy	• Near-drowning event. Choking episode
Diabetic ketoacidosis	• Poorly controlled diabetes mellitus
Hyponatremia	• SIADH. Rapid correction of hypernatremia
Liver failure	• Viral or autoimmune hepatitis, acetaminophen
Reye syndrome	• Aspirin administration for viral illness in children
*Acute cerebral dysfunction/AMS in the absence of primary CNS pathology.	

Aspirin is contraindicated in children except in Kawasaki disease and JIA

Hyperglycemia (DKA) and hyperammonemia can cause cerebral edema, not hypoglycemia.

Meckel diverticulum (MC GI congenital anomaly)	
Pathogenesis	• Ectopic gastric mucosa → intestinal ulceration & bleeding
Epidemiology	• Presentation often by age 2 (but can occur at any age) • Usually <2 inches long. 2 feet of ileocecal valve
Presentation	• May be asymptomatic (incidental finding) • Painless rectal bleeding ± iron deficiency anemia • Acute abdominal pain due to **complications**: Intussusception (recurrent, atypical). Diverticulitis (mimics appendicitis). Bowel obstruction, perforation (peritoneal signs)
Diagnosis	• Technetium-99m pertechnetate scan detects gastric mucosa

- Abdominal pain, diarrhea, and vomiting are uncommon.
- CT scans cannot reliably differentiate the diverticulum from other loops of bowel.

Intussusception (Bowel edema → ischemia & necrosis). Children draw up their legs	
Risk factors	• Hypertrophy of intestinal Peyer patches (eg, recent viral illness) • Pathologic lead point (eg, Meckel diverticulum (MC), HSP, tumor)
Clinical presentation	• Sudden, severe intermittent abdominal pain & V. Sausage-shaped mass in right abdomen. Currant jelly stools. Lethargy or AMS
Diagnosis & management	• Ultrasound: target sign. Air (pneumatic) or saline enema. Surgery for failed enema reduction or signs of peritonitis

Pathologic lead point suspected if: **Recurrent** episodes. **Atypical location** (small bowel into small bowel). Atypical age. Persistent rectal bleeding despite reduction

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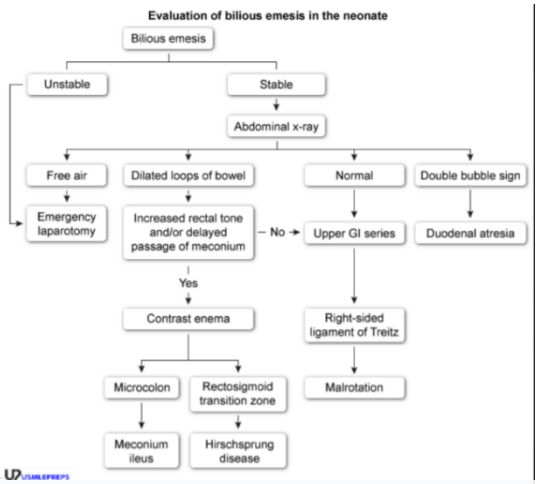
Malrotation with midgut volvulus [Most common in infancy (usually age <1 month)]	
Pathogenesis	• Narrow mesenteric base allows for ↑ small bowel mobility → Twisting of small bowel around SMA → gut ischemia & necrosis
Clinical features	• Acute: bilious emesis (MC), abdominal distension • Chronic: intermittent abd pain & bilious vomiting, FTT, irritability

	• Untreated: hematochezia, peritonitis & shock
Diagnosis	• Abd x-ray: ± dilated bowel, air-fluid levels, pneumoperitoneum • **Upper gastrointestinal series (gold standard)**: ligament of Treitz on right side; corkscrew, or bird's-beak, duodenum
Treatment	• Emergency laparotomy (to relieve volvulus) • Ladd procedure (to reposition malrotated bowel)

Malrotation: Associated Hx of omphalocele/CDH/ heterotaxy syndrome. Examination may be normal between episodes.

An immature LES: GER (ie, spit-up) commonly seen in infants. Spit-up **resembles formula (or breastmilk) and is never bilious**. **Introduction of solid foods is at age 6 months.**

Initial management of **clinically stable** patients (eg, normal vital signs) with bilious emesis includes **NPO, NG tube decompression, IV fluids, diagnostic imaging**.



Pediatric functional constipation (MCC of constipation in children)	
Risk factors	• Multiple transitional events: Initiation of solid food & cow's milk. Toilet training. School entry (stool withholding behaviour)
Symptoms	• Painful/hard bowel movements. Stool withholding. Encopresis
Alarm signs	• Delayed passage of meconium. Fever/vomiting. "Ribbon" stools. Poor growth. Severe abd distension. Displaced anus. Tuft at gluteal cleft
Complications	• Anal fissure. Hemorrhoids. Enuresis/UTI
Treatment	• ↑ Dietary fiber & water intake/ prune juice. Limit cow's milk (<24 oz/day). Laxatives (eg, polyethylene glycol). Age-appropriate toileting guidance. ± Enemas/suppositories

If symptoms become **chronic**, the rectum can **dilate** and **fecal impaction** (**stool ball** on examination) can occur. Tx: **enema followed by long-term laxative**. However, in children with **anal fissures**, enemas cause more rectal trauma and should be **limited**.

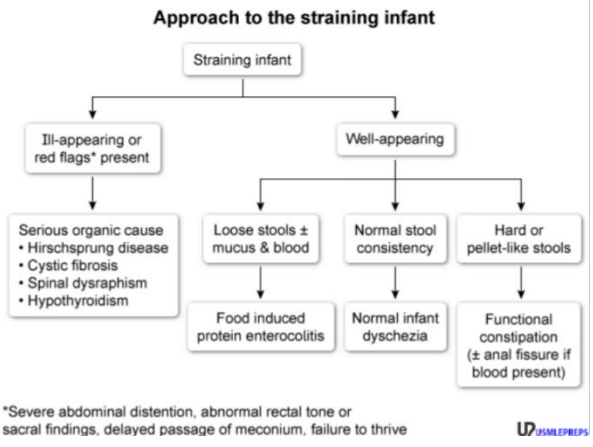
Excessive intake of cow's milk >24 oz/day can worsen constipation because it provides **fat and protein without fiber**, leading to **slow transit time** and causing **satiety without room in the diet for higher fiber foods.**

(Choice D) Abdominal x-rays can be considered for patients with signs of severe constipation (eg, **abdominal pain, vomiting, abnormal PEx**) because severe fecal impaction can mimic intestinal obstruction.

(Choice E) Juvenile polyps present in children age 2-10 with painless rectal bleeding.

Hirschsprung disease (congenital aganglionic megacolon)	
Pathology	• Failed neural crest cell **migration** to distal colon
Symptoms	• Neonates: Delayed passage of meconium (in **long segment**), **bilious** vomiting, ± enterocolitis. Children/adolescents: Chronic constipation, FTT (**short segment**)
Physical examination	• Abd distention + tenderness. **Tight anal sphincter**. Absence of stool in rectal vault. Forceful stool expulsion on rectal examination (**squirt sign**)
Diagnosis	• Rectal suction biopsy (gold standard). Anorectal manometry, contrast enema (adjuncts)
Management	• Surgical resection of aganglionic segment

Thin caliber stools due to failure of the distal colon to relax. **Alarm signs** will be present. **Anal fissures are uncommon**. Straining in infants due to lack of internal sphincter relaxation.



Pathogenesis of **infant dyschezia** involves:

- Failure to **coordinate** increased intraabdominal pressure with relaxation of the pelvic floor muscles
- Inadequate abd muscle tone to produce effective Valsalva maneuver

Crying, turning red in the face, straining for greater than 10 mins, followed by passage of a soft, nonbloody stool. Well-appearing & normal PEx. Self limiting by age 9 months (gut nervous system matures). Reassurance.

Infant constipation		
	Functional	**Pathologic causes**
Risk factors	• Introduction of solid foods. ↓ Water intake. ↓ Fiber diet	• Down syndrome. Displaced anus, tuft at gluteal cleft
Clinical features	• Infrequent defecation. Hard, painful stools. Large-caliber or pellet-like stools. ± Anal fissure	• Delayed passage of meconium. Fever or vomiting. Ribbon stools. Poor growth. Severe abd distension
Management	• Add undigestable, osmotically active carbohydrate (prune or apple juice/puree). Polyethylene glycol if refractory	• Workup for serious organic cause: Hirschsprung disease (barium enema). CF (sweat chloride test). Spinal dysraphism (MRI)

STEC (Tx: aggressive fluids, avoidance of Abs & antidiarrheals)	
Pathogenesis	• Contaminated/undercooked beef or contact with farm animals • Invasion of intestinal epithelial cells. Production of Shiga toxin
Clinical features	• **Watery → bloody diarrhea within 3 days**. No high fever (Low-grade temperature of <38.5 C may occur).
Diagnosis	• Multiplex stool PCR testing. Stool Shiga toxin assay. Stool culture
Complications	• HUS (may develop 1-2 weeks after diarrhea onset)

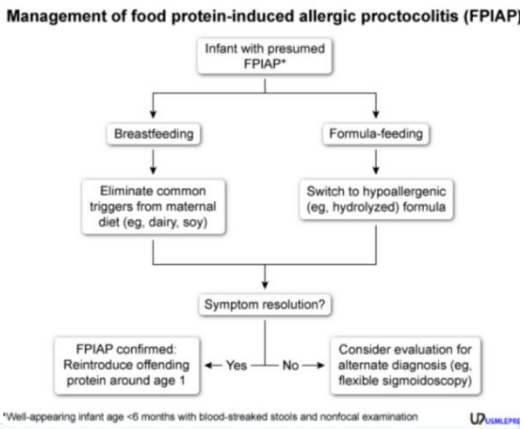
(Choice A) ***C perfringens*** **Gastroenteritis**: watery diarrhea that resolves within 1-2 d.

IgE- & non–IgE-mediated food allergies				
	Example	**Age**	**Symptom onset**	**Clinical features**
IgE mediated	Anaphylaxis	Any	Immediate (<1 hr)	• Urticaria, V, wheezing. Angioedema, hypotension

Non–IgE mediated	Food protein–induced allergic proctocolitis	<6 months	Insidious	• Painless, bloody loose stools. Well appearing
	Food protein–induced enterocolitis syndrome	<12 months	Within hours	• Profuse vomiting, diarrhea (± blood), dehydration, lethargy. Ill appearing

Food protein–induced allergic proctocolitis (onset insidious, casein/whey)	
Pathophysiology	• Non–IgE-mediated reaction. Eosinophilic inflammation of rectosigmoid colon. Common triggers: cow's milk & soy proteins
Risk factors	• Eczema. Family history of food allergies
Clinical features	• Age 1-4 weeks (up to 6 months). Well appearing. painless bloody loose/mucus streaked stools, **normal growth**.
Prognosis	• Tolerance of offending protein by age 1

Symptoms suggestive of a different diagnosis include failure to thrive, profuse diarrhea, and forceful vomiting. For formula-fed infants, switching to an **extensively hydrolyzed formula** (predigested cow's milk proteins and is free of potentially cross-reacting soy proteins) is ideal. In rare cases of persistent bleeding, an **amino acid–based (elemental) formula** may be required. Symptoms resolve in **days to weeks**.



Meconium ileus (Inspissated stool causes obstruction at terminal ileum)	
Features	• Failure to pass meconium within 24 hr of birth (ileal obstruction). Abdominal distension ± Bilious emesis. No stool in rectal vault
Workup	• X-ray: dilated loops of small bowel. Contrast enema: microcolon • Diagnostic evaluation for CF (eg, sweat test)
Treatment	• Hyperosmolar enema ± Surgical management

Delayed passage of meconium		
	Hirschsprung disease	**Meconium ileus**
Pathophysiology	• Failure of neural crest cell migration	• Obstruction by inspissated stool
Level of obstruction	• Rectosigmoid	• Ileum
Rectal examination	• Increased tone • Positive squirt sign	• Normal tone • Negative squirt sign
Meconium consistency	• Normal	• Inspissated
Imaging	• Dilated proximal colon ± small bowel • Narrow rectosigmoid	• Dilated small bowel • Microcolon
Associated disorder	• Down syndrome	• Cystic fibrosis

(Choice E) SNHL can develop in a minority of patients with CF due to frequent treatment with aminoglycosides for Pseudomonas but would not be expected in the newborn period.

Pseudoappendicitis (**infectious ileocecitis**): acute-onset fever, (RLQ) pain, leukocytosis, **profuse watery → mucoid/bloody diarrhea**, sick contacts. Y entercolitica/C jejuni.

Ultrasound, CT scan: ileocecal inflammation + mesenteric lymphadenopathy with sparing of the appendix. **Stool studies (multiplex PCR, culture) are diagnostic**. Self-resolves, may require antibiotics in severe cases. No surgery needed vs acute appendicitis.

Duodenal atresia: MC GI anomaly with down syndrome, **bilious vomiting** in the first 2 days of life. Double bubble sign. Abdominal distension is absent because gas cannot pass the duodenum, polyhydramnios. Management: NPO, NGT decompression, **surgical repair**.

(Choice B) Karyotype confirms trisomy 21 but is **not urgent** in children with characteristic dysmorphic features and signs of **duodenal atresia**. Surgery is more important.

Jejunal atresia: Vascular accident in utero. Risk factors: Vasoconstrictive medications, **cocaine**, tobacco (NOT chromosomal anomalies [vs duodenal atresia])

Necrotizing enterocolitis (weeks after birth)	
Pathogenesis	• Gut mucosal wall invasion by gas-producing bacteria • Intestinal inflammation, necrosis
Risk factors	• Prematurity/ Very low birth weight (<1.5 kg [3 lb 4 oz]). Enteral feeding. Cyanotic CHD and hypotension
Clinical findings	• Nonspecific: apnea, lethargy, vital sign instability • Gastrointestinal: Tense distended abdomen with abd wall erythema/ Feeding intolerance/ Bilious emesis/ Bloody stools
X-ray findings	• Pneumatosis intestinalis (air in bowel wall)/ Pneumoperitoneum (intestinal perforation, indication for surgery)/ portal vein gas
Complications	• Early: Sepsis, DIC// Late: strictures, short-bowel syndrome// Advanced: thrombocytopenia and metabolic acidosis

Moderate dehydration capillary refill (2-3 secs). Severe dehydration (>3 secs)

Management of necrotizing enterocolitis	
Immediate interventions	• Discontinuation of enteral feeds. NG decompression. **Blood cultures THEN empiric antibiotics**. IV fluid repletion
Monitoring	• Serial CBC, electrolytes, abd examinations & imaging
Indications for surgery	• Bowel perforation (**pneumoperitoneum**, NOT pneumatosis intestinalis). Clinical deterioration [vital sign instability, persistent acidosis] despite medical management (suggestive of bowel necrosis)

Giardiasis	
Risk factors	• Contaminated food or water. Hiking in lakes • Fecal incontinence & crowding (day care, nursing home) • Immunodeficiency (eg, CVID, IgA deficiency, CF, HIV)
Pathogenesis	• Villous blunting, disruption of epithelial tight junctions, loss of brush border enzymes → malabsorption

Clinical features **(1-2 wks after exposure)**	• Subacute (<4 weeks) or chronic (months). Loose, oily, nonbloody stools. Bloating, flatulence. Weight loss, decreased linear velocity (children), vitamin deficiencies
Diagnosis	• Stool antigen/PCR (↑ sensitivity). Stool microscopy (↓ sensitivity)
Treatment	• First-line: tinidazole or nitazoxanide/ Alternate: metronidazole (children)/ Pregnancy (first trimester): paromomycin • Refractory/recurrent symptoms: evaluation for immunodeficiency

In high-resource settings, the most common risk factor is exposure to cysts shed by infected mammals into rivers and lakes.

Lactose intolerance [worsening diarrhea after gastroenteritis with no fever/vomiting]	
Pathogenesis	• Primary: Acquired, persistent lactase deficiency. Onset in **early childhood** • Secondary: SI epithelial damage due to infection (gastroenteritis) or inflammation (celiac disease). Often transient (weeks to months)
Clinical features	• Crampy periumbilical abdominal pain. Flatulence/bloating. Watery diarrhea after dairy intake. Bulky/malodorous stools
Diagnosis	• Resolution of symptoms on dairy-restricted diet. Lactose breath hydrogen test
Management	• Dietary restriction of lactose. Lactase replacement if dairy ingested

Most patients with secondary lactase deficiency do not need a diagnostic hydrogen breath test, because symptoms usually resolve within weeks as the intestinal mucosa heals.

Breastfeeding contraindications
• MATERNAL: Active untreated TB. HIV infection*. Herpetic breast lesions. Active varicella infection. Chemotherapy or radiation therapy. Active substance use disorder. // FETAL: Galactosemia
*In developed countries where formula is readily available.

- Active tuberculosis until no longer contagious (**2 weeks** of antituberculin therapy)
- Active substance use disorder without enrollment in a **methadone** (low concentration in breastmilk) **or buprenorphine** treatment program

(Choice A) Maternal blood alcohol level (eg, legal limit of 0.08) closely matches the level of alcohol in breastmilk, which translates to a generally low percentage (eg, 0.08%) of alcohol ingested by the breastfeeding newborn. Therefore, **casual, moderate alcohol use is considered safe and not a contraindication to breastfeeding.**

Vegan diet	
Benefits	• Lower blood glucose & cholesterol levels. ↓ heart disease, stroke
Potential nutritional deficiencies	• Common: vitamin B_2 B_{12}, vitamin D, calcium • Possible*: iron, zinc
*Increased risk: children (rapid growth requires higher zinc/iron) & young women (menstrual blood loss requires higher iron).	

Pellagra (niacin deficiency) [meat fish legumes, absorbed in stomach and SI]	
Associated conditions	• Alcoholism. Anorexia nervosa. Malabsorptive disease (eg, Crohn disease). Dietary niacin deficiency
Symptoms	• Diarrhea (watery), dementia ,dermatitis (photosensitivity, erythematous symmetric well demarcated rash, hyperpigmented/thickened skin), depression, distraction, death. Atrophic glossitis (beefy red tongue)

Water-soluble vitamins		
Vitamin	**Source**	**Deficiency**
B_1 (thiamine)	Whole grains, meat, fortified cereal, nuts, legumes	• Beriberi (peripheral neuropathy, HF) • Wernicke-Korsakoff syndrome
B_2 (riboflavin)	Dairy, eggs, meat, green vegetables	• Angular cheilosis, stomatitis, glossitis. **Normocytic** anemia • **Seborrheic** dermatitis
B_3 (niacin)	Meat, whole grains, legumes	• Pellagra

B_6 (pyridoxine)	Meat, whole grains, legumes, nuts	• Cheilosis, stomatitis, glossitis, seborrheic dermatitis. Irritability, confusion, depression
B_9 (folate, folic acid)	Green leafy vegetables, fruit, meat, fortified cereal/grains	• Megaloblastic anemia • Neural tube defects (fetus)
B_{12} (cobalamin)	Meat, dairy	• Megaloblastic anemia • Neurologic deficits (confusion, paresthesias, ataxia)
C (ascorbic acid)	Citrus fruits, strawberries, tomatoes, potatoes, broccoli	• Scurvy (punctate hemorrhage, gingivitis, corkscrew hair)

- **Vitamin B_2 deficiency:** underdeveloped countries, anorexia nervosa, low-dairy diet (eg, lactose intolerance), and malabsorptive syndromes (eg, celiac sprue).
- Riboflavin has no known toxic effects as excess amounts are excreted in urine.
- Hypervitaminosis A: neuropsychiatric symptoms and cerebral edema.

Vitamin C deficiency (scurvy) [Low income countries]	
Pathogenesis	• Insufficient dietary intake (lack of citrus fruits/vegetables): Chronic alcohol/substance use, anorexia, autism, restricted diet
Manifestations	• Mucocutaneous: Coiled hair, perifollicular hemorrhage. Petechiae, ecchymoses. Impaired wound healing. Bleeding gums • MSK (IM + subperiosteal hemorrhage): arthralgia, limp, leg pain • Constitutional: malaise, depression, vasomotor instability
Laboratory findings	• Anemia (normocytic or microcytic). Normal platelet count & coagulation studies
Treatment	• Oral/injectable vitamin C (improvement within days to weeks) • Toxicity (Abd pain, diarrhea) with excessive supplementation

Rickets (Histopathology: Excess unmineralized osteoid. Tx: Ca and VitD supplementation)	
Risk factors	• Exclusive breastfeeding. Inadequate sun exposure

	• Dark skin (melanin partially blocks ultraviolet rays from reaching the skin). Absence of fortified dairy (dairy alternative) foods
Clinical manifestations	• Decreased muscle tone & delayed development • Delayed fontanelle closure, frontal bossing, **craniotabes** (skull bones that depress with pressure) • Widening of epiphyses (widened wrists, growth plate enlargement) • Hypertrophy of costochondral joints (rachitic rosary) • Short stature, femoral & tibial bowing (genu varum, symmetric or asymmetric) + UL bowing
Laboratory findings	• Low 25-hydroxyvitamin D. Low/normal Ca. Low urine Ca. Low PO4. Elevated AlkP, PTH.

All exclusively breastfed infants should be started on **400 International Units of vitamin D daily within the first month of life.**

Celiac disease	
Risk factors	• First-degree relative. Down syndrome. Autoimmune disorders
Symptoms	• ± Abdominal pain, distension, bloating, diarrhea, nonbillious vomiting, FTT, WL, short stature, delayed puberty/menarche, enamel hypoplasia, atrophic glossitis, IDA, peripheral neuropathy, anxiety, depression, arthritis, rickets/osteomalacia
Diagnosis	• ↑ Tissue transglutaminase IgA antibody • Proximal intestinal biopsy (villous atrophy, crypt hyperplasia, intraepithelial lymphocytosis)
Treatment	• Gluten-free diet. Dapsone for dermatitis herpetiformis

Pureed fruits and vegetables should be introduced first followed by **pureed proteins such as meats**. There is no evidence suggesting that early introduction of highly allergenic foods such as **eggs** is associated with an increased risk of allergies; these foods can be introduced **any time after age 6 months**.

(Choice B) For families who choose to offer juice, **no more than 4-6 ounces per day should be given (after age 6 months) and it should never be given in a bottle** given the significantly increased risk **of dental caries**.

Cyclic vomiting syndrome (clinical dx, requires labs & imaging to r/o other causes, pathogenesis unknown. Tx: triptans, antiemetics, rehydration, most children have gradual resolution of symptoms during adolescence)	
History	• Personal or family history of migraines • Episodes often have identifiable trigger (eg, infection, stress)

Symptoms	• Stereotypical vomiting episodes
	  ○ Acute onset of nausea, abdominal pain, headache, vomiting. Self-limited, lasting 1-2 days, erosive caries from vomiting
	• **Between episodes**
	  ○ Usually **asymptomatic**. Often regular intervals (eg, 2-4 weeks)

Recurrent, predictable pattern of acute and frequent vomiting that resolves spontaneously with no symptoms between episodes. Growth, examination, eating patterns are normal.

Differential diagnosis of regurgitation & vomiting in infants		
Diagnosis	**Clinical features**	**Management**
Gastroesophageal reflux	• Physiologic (asymptomatic, happy spitter) [spit up, normal weight gain, no pain/back arching]	• Reassurance • Positioning therapy
	• Pathologic (GERD) (FTT, significant irritability, sandifer syndrome (intermittent opisthotonic posturing))	• Thickened feeds (oatmeal). Antacid. If severe, esophageal pH probe monitoring & upper endoscopy
Milk protein allergy	• Regurgitation/vomiting. Eczema. Bloody stools	• Elimination of dairy & soy protein from diet
Pyloric stenosis	• Projectile nonbilious vomiting • Olive-shaped abdominal mass • Dehydration, WL	• Abdominal ultrasonography • Pyloromyotomy

Gastroesophageal reflux is **extremely common,** affects more than 50% of infants. Shorter esophagus, incomplete closure of the LES, greater time spent in the supine position.

Smoking exposure is a risk factor, peak symptoms at age 4 months. Aspiration risk is **not** greater in the supine position due to an intact gag reflex. GER complicated by poor weight gain, feeding refusal, or irritability in an infant is referred to as (GERD).

Tx: **frequent, small-volume feeds + burping during feeds**; **hold the infant upright** for 20-30 minutes after feeds; and place the infant **prone when awake**. Activities that **increase intraabdominal pressure (eg, fastening the diaper too tight, bringing the knees to the stomach) should be avoided**. Regurgitation usually **improves around age 6 months** (when the infant can sit unsupported) and **resolves by age 1 year**.

An infant should be placed only on the **back (supine) during sleep** as the risk of sudden infant death syndrome with prone sleeping outweighs the benefit of reflux reduction.

(Choice F) Cow milk protein–based formula (which is different from cow's milk) is standard infant formula; it provides infants with necessary nutrients. **Goat milk is deficient in folate and would result in a macrocytic anemia.**

Eosinophilic esophagitis in children (Th2, comorbid atopy)	
Presentation	• Toddler: feeding difficulties (solid food refusal [unable to advance diet from purees to solids]), weight loss, normal swallow study • School-aged: abdominal pain, vomiting • Adolescent: dysphagia, heartburn, food impaction
Diagnosis	• **Endoscopy & esophageal biopsy** (eosinophils: ≥15/hpf) • Exclusion of alternate diagnoses (eg, achalasia, infection)
Treatment	• Elimination diet (1st line), PPI, topical glucocorticoid, feeding therapy

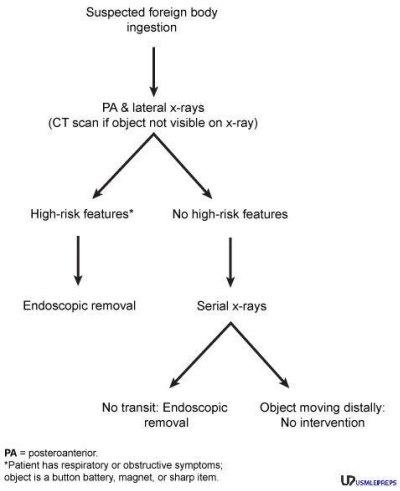
Infantile hypertrophic pyloric stenosis (firstborn male, formula, macrolide exposure)	
History	• Age 3-6 weeks. Projectile nonbilious emesis. Immediately postprandial. Followed by interest in refeeding ("hungry vomiter")
Examination	• Palpable, olive-shaped epigastric mass. Visible peristalsis. ± Signs of dehydration (eg, sunken fontanelle) + WL
Laboratory/ imaging	• Hypochloremic, hypokalemic metabolic alkalosis (may be normal if diagnosed early) ± Indirect hyperbilirubinemia + thickened, elongated pylorus on US

Treatment	• IV fluids + correction of electrolytes **before** pyloromyotomy

- Metoclopramide has no role in the treatment of pyloric stenosis as the emesis is a result of a fixed gastric outlet obstruction rather than poor gastric motility.

Button batteries: tissue **corrosion**, liquefaction **necrosis**, pressure necrosis, esophageal ulceration **perforation/** stricture/stenosis, hemorrhagic shock, and death. Asymptomatic/ N V anorexia chest pain, **fever hematemesis, shock (life threatening perforation)**

Radiographic features that distinguish the bilaminar structure of a button battery from a flat coin include: a **halo** or double-ring around the circular object on AP view/ a step-off or beveled edge on lateral view (occasionally but not always seen).



Emergency endoscopic removal + **honey** soon after ingestion in an asymptomatic patient may provide a partial protective barrier but should not delay definitive management.

Foreign body ingestions are common in children **age 6 months to 3 years** due to increased oral exploratory behavior. **≥2** Magnets lead to **necrosis/ perforation/ fistula**. **Endoscopic removal even if asymptomatic**. Most fish bones are radiolucent and visualized only on CT scan.

If a sharp foreign body is distal to the proximal duodenum + asymptomatic, observation with repeat x-ray in 12-24 hrs

(Choice C) Barium esophagography may be performed **several months** after ingestion of a **corrosive** agent (eg, cleaning product) to assess for **stricture** formation. Contrast is **avoided in the acute management** of foreign body ingestion because it can **obscure visualization and hinder removal of the object.**

Colic: Symptoms peak at age 6 weeks, resolve by age 3-4 months. The cause of colic is unknown but may be related to gut immaturity or suboptimal feeding techniques (excessive swallowing of air). **Diagnosis of exclusion**.

Mx: **reassurance**, **review soothing techniques** (include using a pacifier; holding, rocking, or swaddling the baby; and minimizing environmental stimuli (eg, dark room)). Use of swings, carriers, and strollers can also be calming and allows parents to rest from active soothing. In addition, adjusting feeding techniques (eg, upright feeding position in bottle-fed babies) reduce air swallowing and relieve colic.

Crying in young infants	
Diagnosis	**Key features**
Normal	• Intermittent, consolable, **<3 hr/day**
Colic	• **≥3 hr/day (usually evening), ≥3 days/week** • **Healthy infant age <3 months**
GERD	• Frequent spit-up. Back-arching after feeding, feeding difficulty
Infection	• Acute otitis media: bulging tympanic membrane, ± fever • Meningitis: fever, lethargy, bulging fontanel • Septic arthritis: fever, **limited extremity movement** • UTI: fever, vomiting, poor feeding
Intussusception	• Episodic irritability with legs drawn to abdomen ± Bilious emesis, bloody stools
Torsion	• Testicular swelling or abdominal distension (ovarian)
Trauma	• **Hair tourniquet**: hair accidentally wrapped around digit • **Corneal abrasion**: tearing, photophobia; + fluorescein test • **Abuse/fracture**: bruising, laceration, asymmetric movements

MCC of splenic rupture: high velocity blunt trauma

Atraumatic splenic rupture	
Risk factors	• Leukemia, lymphoma, CMV, EBV, malaria, SLE, pancreatitis, cirrhosis, pregnancy, anticoagulation, G-CSF
Clinical presentation	• Diffuse or LUQ abdominal pain, peritonitis. Referred left shoulder pain (Kehr sign). Hemodynamic instability
Diagnosis	• Acute anemia. Intraperitoneal free fluid on imaging
Treatment	• Catheter-based angioembolization (stable patients) • Emergency splenectomy (unstable patients)

Rotavirus vaccine (Rotavirus is the MCC of gastroenteritis in infants & children worldwide)	
Specifications	• Live attenuated oral vaccine. Series administration between age 2 & 6 months
Contraindications	• Allergy to vaccine ingredients. SCID. Hx of intussusception

(Choice D) Live virus vaccinations can be **safely administered to household contacts of pregnant women** because transmission of vaccine-derived infection is exceedingly rare.

(Choice E) Vaccines can be administered safely during **minor illnesses** (URI, otitis media). They should be **postponed until recovery from moderate or severe illnesses** (eg, **high fever**) because manifestations of the underlying illness can be incorrectly attributed to the vaccine or vice versa.

Gastroschisis: **full-thickness** abdominal wall defect **rt to the umbilicus**. Prenatal US: free-floating intestinal loops within the amniotic sac, ↑ **AFP** , **isolated** defect with no extraintestinal anomalies, **FGR, oligohydramnios**.

Continued exposure of the intestines to **amniotic fluid** can cause **chronic inflammation and edema**, resulting in **intestinal thickening** and **reduced bowel motility**. In some cases, this may ultimately lead to **bowel obstruction**, resulting in **polyhydramnios**.

After delivery, the lower half of the infant is placed in a **sterile plastic bag** to minimize insensible heat and fluid losses, and a **nasogastric or orogastric tube** is placed to decompress the stomach. Definitive management is **surgical repair.**

(Choice D) Prune belly syndrome is due to a **defect in abdominal musculature**. Intestinal loops may be seen through the thin abdominal wall, causing the prune appearance, but are **covered by skin**.

Chapter 9: Hematology

Diamond-Blackfan anemia (Congenital pure red cell aplasia)	
Clinical findings	• Short stature. Craniofacial abnormalities. Triphalangeal thumbs. ↑ risk of malignancy. Cleft palate, webbed neck
Laboratory findings	• **Macrocytic** anemia. Reticulocytopenia. Normal plt, WBCs

Treatment	• Corticosteroids + RBC transfusions in refractory cases

➢ Normal lymph nodes: (several 1-cm anterior cervical lymph nodes)

Folate deficiency anemia (Tx: folic acid supplements)	
Etiology	• Chronic hemolysis (**SCD**). **Poor dietary intake**. Malabsorption (eg, **gastric bypass**). **Methotrexate, trimethoprim, phenytoin**
Features	• Dyspnea, fatigue, pallor, weakness
Laboratory findings	• Macrocytic anemia. Poor reticulocyte response (low to normal). Hypersegmented neutrophils. Low serum folate

(Choice B) Parvovirus: severe, acute (<2 weeks) normocytic anemia with decreased RC.

Common causes of macrocytosis	
Increased reticulocytes	• Hemolytic anemia. Bone marrow recovery
Impaired RBC maturation	• Vit B_{12}/folate deficiency, hydroxyurea, methotrexate
Bone marrow disorders	• MDS. Sideroblastic anemia. Multiple myeloma
Lipid abnormalities	• Liver disease. Hypothyroidism

Hydroxyurea: High doses cause **myelosuppression** (pancytopenia) with ↓ **RC** due to cell cycle arrest of rapidly dividing hematopoietic cells. [The primary dose limiting side effect]

Acute splenic sequestration	
Pathophysiology	• Complication of SCD seen in <u>early childhood</u> • Vasoocclusion within spleen -> trapping of RBCs & platelets
Clinical features	• Abd pain, **tender** splenomegaly, anemia, hypotensive shock
Labs	• Acute drop in hemoglobin. Reticulocytosis. <u>Thrombocytopenia</u>
Treatment	• **Isotonic** fluid resuscitation . **RBC** transfusion. ± **Splenectomy**

A **packed RBC** should be administered cautiously (eg, **half of normal volume**) because **RBCs in the spleen may return to circulation**, resulting in a **rapid increase in hemoglobin** and complications such as **hyperviscosity syndrome.**

(Choice C) Splenectomy is **not typically performed in the acute setting and is often unnecessary** due to the natural history of splenic autoinfarction in patients with sickle cell disease. **Functional asplenia** occurs by **age 5** in patients with SCD.

Sickle cell disease (AR, Glu6Val, ↓Hct, ↑RC, ↑ HbS, ↓ HbA) (common in Nigeria)	
Mx	• **Maintenance**: Pneumococcal vaccine/ **Penicillin (until age 5)** /Folic acid supplementation/ Hydroxyurea (for pts with recurrent vaso-occlusive crises) • **Acute pain crises**: Hydration + Analgesia (NSAIDS & opioids) ± Transfusion

Despite vaccination, ***S pneumoniae*** remains by far the most common cause of sepsis in patients with SCD, usually from **non-vaccine serotypes**.

Stroke in sickle cell disease (pain crises is a risk factor, anticoagulants are useless)	
Pathogenesis	• Ischemic stroke: Intimal hyperplasia & stenosis (vasculopathy). Adhesion of sickled RBCs to vasculature • Hemorrhagic stroke: Weakened cerebral vessels. Aneurysm rupture
Presentation	• Hemiparesis, seizure, AMS. Dx: MRI (ischemic), CT (hemorrhagic)
Treatment	• Exchange transfusion. Simple transfusion if unavailable
Primary prevention	• Transcranial Doppler screening during childhood. Hydroxyurea ± Chronic transfusions

Pneumococcal vaccination plus penicillin prophylaxis (twice daily until age 5) can prevent almost all cases of pneumococcal sepsis in patients with sickle cell disease.

Most patients with sickle cell trait lead normal, healthy lives. Painless hematuria is the MC complication THEN hyposthenuria THEN UTI (particularly in pregnancy)

G6PD: Heinz bodies & bite cells on **crystal violet** stain, dark urine, splenomegaly rare.

Electrophoresis patterns in sickle cell syndromes

	HbA	HbA2	HbF	HbS	HbC
Normal	++++	+	+	None	None
Sickle cell trait	+++	+	+	+++	None
Sickle cell anemia (SCA)	None	+	+	++++	None
SCA on hydroxyurea	None	+	++	+++	None
Hemoglobin SC disease	None	+	+	+++	+++

Sickle cell trait (usually asymptomatic, no change in overall life expectancy)	
Laboratory findings	• **Normal** hemoglobin, reticulocyte count, RBC indices & morphology • Hemoglobin electrophoresis: **Hb A > Hb S**
Complications	• Hematuria/papillary necrosis, **hyposthenuria** • Splenic infarction (especially at higher altitudes), VTE, priapism. **Exertional rhabdomyolysis**

Hyposthenuria in SS: Inability of the kidneys to concentrate urine (Damage to vasa recta due to sickled RBCs → renal medulla damage). Polyuria nocturia despite fluid restriction, low specific gravity (<1.010), low Uosm, normal serum Na (intact ADH), normal urinary diluting capacity (intact superficial loop of henle, which is not supplied by the vasa recta).

Mild hyposthenuria due to SC**T** requires **no treatment**. In patients with SC**D**, **RBC transfusions** often improve urine-concentrating ability and provide relief of symptoms.

Acute, severe (<6mg/dl) anemia in sickle cell disease		
Cause	**Reticulocytes**	**Key features**
Aplastic crisis	↓ (<1%)	• Transient arrest of erythropoiesis. Parvovirus B19. NO splenomegaly. Tx: blood transfusions. **Platelet count normal**
Splenic sequestration crisis	↑	• Splenic vasoocclusion → rapidly enlarging spleen. Occurs in children prior to autosplenectomy. **Thrombocytopenia** & hypovolemic shock

(Choice A) Acute chest syndrome is caused by **pulmonary vasoocclusion or infection**, and is characterized by **fever, chest pain, and a new infiltrate on chest radiograph**.

(Choice B) Aplastic anemia: pancytopenia, vs aplastic crisis (only RBCs affected).

Differential diagnosis of bone pain in sickle cell disease			
Cause	**Vaso-occlusive crisis**	**Osteomyelitis**	**Avascular necrosis**
Clinical features	• **Acute**, severe pain • **Pain >1 site** (eg, dactylitis)	• **Acute or subacute** pain	• **Chronic**, progressive hip pain with weight bearing

	• +/- Low-grade fever • Erythema & warmth • May be preceded by trigger (eg, dehydration)	• Focal pain at 1 site (eg, long bone) • Prolonged fever • Erythema & warmth • **(+) blood culture**	• **Absence of fever** • **Absence of warmth or erythema**

Dactylitis (hand-foot syndrome) can be the **earliest manifestation** of **vaso-occlusion** in **sickle cell disease** (SCD). Age 6 months to 4 years with an acute onset of pain and **symmetric swelling of the hands and feet**. +/- Low-grade fever. Soft tissue swelling on x-ray. Tx: **hydration and pain control**.

Common etiologies of pediatric stroke: SCD. Prothrombotic disorders. CHD. Bacterial meningitis. Vasculitis. Focal cerebral arteriopathy. Head/neck trauma

(Choice E) Further noninvasive workup (eg, serum lactate/pyruvate, creatine kinase) must be performed **before muscle biopsy** if MELAS is suspected.

Hereditary spherocytosis [AD, MC hemolytic anemia in Northern Europe, classic triad: hemolytic anemia, jaundice (refractory to phototherapy), splenomegaly]	
Laboratory findings	• ↑ MCHC (membrane loss and cellular dehydration). Negative Coombs test. ↑ **Osmotic fragility** on acidified glycerol lysis test • Abnormal **eosin-5-maleimide binding** test
Treatment	• Folic acid supplementation. Blood transfusion. Splenectomy

PNH: hemolytic anemia, cytopenias, and hypercoagulability.

Drug-induced AIHA (NSAIDS, penicillin, cephalosporins)	
Pathogenesis	• Drug coats erythrocytes (creates hapten for IgG attachment) → IgG binding → splenic destruction of RBCs (**extravascular** hemolysis) • Drug triggers immune complexes → complement-mediated destruction of RBCs (**intravascular** hemolysis)
Manifestations	• Sudden onset (within hours of exposure). Anemia: fatigue, pallor, dyspnea. Hemolysis: jaundice, dark urine, abdominal or back pain, ↑ **Reticulocytes/indirect bilirubin & LDH**, ↓ Haptoglobin, Spherocytes, + Direct Coombs test (anti-IgG, anti-C3)

Treatment	• Discontinue offending drug. Transfusion (if severe) ± Glucocorticoids, IVIG

Rare in newborn (No Ab production)

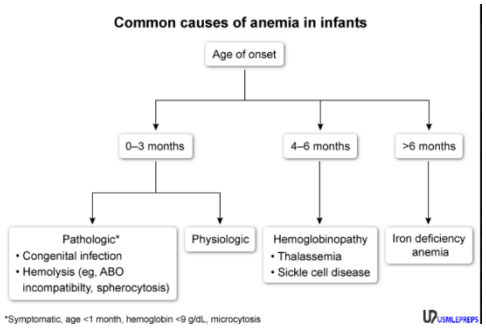
Medications that often trigger hemolysis in G6PD deficiency	
Avoid	Dapsone, primaquine, nitrofurantoin, rasburicase, isobutyl nitrite
Use with caution	Acetaminophen, aspirin, Chloramphenicol, Chloroquine, Colchicine, Diphenhydramine, Glyburide, Isoniazid, L-Dopa, Quinine, TMP-SMX, Vit K

Iron deficiency anemia in young children (Tx: empiric trial of iron supplementation)	
Risk factors	• Prematurity. Lead exposure • Age <1: Delayed introduction of solids (ie, exclusive breastfeeding after 6 months). Cow's, soy, or goat's milk • Age >1: >24 oz/day of cow's milk. <3 servings/day of iron-rich foods
Diagnosis	• Universal Screening Hb at age 1. **Hb <11 g/dL**, ↓ MCV, ↑ RDW
Notes	• MC nutritional deficiency in young children, often asymptomatic and may not have telltale symptoms such as pica, fatigue, or pallor. Aniosocytosis is the 1st finding in peripheral blood smear

Empiric oral iron therapy: Hb should be rechecked in 4 weeks; if the Hb level has risen 1 g/dL, the oral iron therapy should be continued for 2-3 months after the Hb normalizes to replete iron stores. **Low erythrocyte count (vs thalassemia) & pencil cells**

(Choice E) Iron deficiency: restless legs syndrome, fidgety movements at night.

Blood transfusions are rarely necessary in the treatment of dietary iron deficiency anemia, even when the hemoglobin is as low as 4 g/dL. Iron therapy should be sufficient.



Term infants usually have adequate iron stores for the first 6 months of life. **Cow's milk** has **low iron content** + **low bioavailability** because the **calcium and caseino phosphopeptide** found in cow's milk interfere with intestinal **absorption** of dietary iron + **replaces the normal intake of iron-rich foods**.

Alpha thalassemia		
Genotype	**Disorder**	**Clinical features**
1 gene loss (αα/α−)	Alpha thalassemia minima	Asymptomatic, silent carrier
2 gene loss (αα/−−) or (α−/α−)	Alpha thalassemia minor	Mild microcytic anemia
3 gene loss (α−/−−)	Hemoglobin H disease	**Chronic hemolytic** anemia
4 gene loss (−−/−−)	Alpha thalassemia major, hemoglobin Barts disease	**Hydrops fetalis**: anasarca, high output HF, death in utero

Alpha- and beta-**thalassemia minor** are often asymptomatic and found incidentally on universal screening for anemia around age 1. **Mentzer index (MCV/RBC) <13** vs IDA >13

Beta-thalassemia: Patients around **age 6-12 months** develop fatigue and pallor. **Splenic hemolysis** of RBCs can cause jaundice, dark urine, and **splenomegaly**. If left untreated, patients can develop skeletal abnormalities due to extramedullary hematopoiesis.

Beta-thalassemia major is **transfusion-dependent**. The additional iron from transfused RBCs increases the risk for iron overload. **Chelation therapy** is required.

Iron deficiency anemia (↓RC) **& thalassemias** (↑RC, compensatory BM response)			
Parameter	**IDA**	**α-Thalassemia minor**	**β-Thalassemia minor**
MCV	↓	↓	↓
RDW	↑	Normal	Normal
RBCs	↓	Normal/↑	Normal/↑
Peripheral smear	Microcytosis, hypochromia	Target cells/teardrop	Target cells/teardrop
Serum iron studies	↓ Iron & ferritin ↑ TIBC	Normal/↑ iron & ferritin (RBC turnover)	Normal/↑ iron & ferritin (RBC turnover)
Response to iron	↑ Hemoglobin	No improvement	No improvement
Hemoglobin electrophoresis	Normal	**Normal**	↑ Hemoglobin A_2

Iron studies are performed prior to hemoglobin electrophoresis in patients with microcytic anemia suspicious for iron deficiency (heavy menses, low erythrocyte count, reactive thrombocytosis) and concomitant thalassemia (Greek origin, family history).

Iron studies in microcytic anemia					
Cause	**MCV**	**Iron**	**TIBC**	**Ferritin**	**Transferrin saturation (Iron/TIBC)**
Iron deficiency	↓	↓	↑	↓	↓
Thalassemia	↓↓	↑	↓	↑	↑↑
Anemia of chronic disease	Normal/↓	↓	↓	Normal/↑	Normal/↓

Sideroblastic anemia is characterized by **increased serum iron levels and normal TIBC**.

Aplastic anemia (Pancytopenia, normal PT PTT, no hepatosplenomegaly)	
Pathogenesis	• Multipotent hematopoietic stem cells are destroyed by cytotoxic T cells or direct cytotoxic injury → bone marrow aplasia/hypoplasia → lack of circulating peripheral blood cells
Common triggers	• Autoimmune, chemotherapy, immunosuppressants, idiosyncratic reactions, Ionizing radiation & toxins, viral hepatitis, HIV
Diagnosis	• BM biopsy: hypocellular marrow with abundance of stromal & fat cells

(Choice A) Bone marrow biopsy should be considered in patients with **more than 2 abnormal cell lines** to evaluate for acute leukemia or aplastic anemia.

Fanconi anemia (**AR** inherited bone marrow failure due to **defective DNA repair**, **MC congenital cause of aplastic anemia**): Short stature, café au lait, hypoplastic thumbs/ polydactyly/ flat thenar eminence, developmental delays. Predisposition to malignancy, pancytopenia, (+) chromosomal breakage test (following DNA exposure to interstrand crosslinking agent). Tx: HSC transplant

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Anemia of prematurity	
Pathogenesis	• **↑ Oxygenation at birth** → ↓ EPO production. **Impaired transition from hepatic to renal EPO in 3rd trimester**

	• Exacerbating factors in premature infants: ↓ RBC life span. **Frequent blood draws in NICU. Iron depletion**. Lower initial Hct. ↓ capacity to product EPO
Features	• Often asymptomatic. Tachycardia, **poor weight gain**, apnea, hypoxia
Labs	• Normocytic, normochromic anemia. Inadequate RC response
Management	• RBC transfusion if severe or symptomatic • Minimize exacerbating factors (iron supplements, limit blood draws)

After birth, increased oxygenation -> ↓ (EPO) production by the liver and kidney -> mild **transient anemia** that reaches a nadir of **around 9-11** g/dL **at age 2-3 months** in term infants (physiologic anemia of infancy). VS preterm: **erythrocyte nadir** tends to be **more severe** (~**7** g/dL) and occur **earlier** (eg, 1-2 months), resulting in AOP.

Physiologic anemia of infancy	
Pathogenesis	• ↑ Tissue oxygenation at birth → down-regulation of EPO
Clinical features	• **Asymptomatic term** infant age 2-3 months • Normocytic anemia (hemoglobin 9-11). Low to normal RC
Treatment/prognosis	• Reassurance. Resolves with **↑ EPO drive** after age 3 months

Neonatal polycythemia (Tx: IV fluids, Glc, partial exchange transfusion)	
Definition	• Hematocrit >**65%** in **term** infants
Causes	• ↑ erythropoiesis from intrauterine hypoxia: maternal diabetes, hypertension (pre-eclampsia), or smoking; IUGR • Erythrocyte transfusion: delayed cord clamping, TTTS • Genetic/metabolic disease: hypo/hyperthyroidism, trisomy (13 18 21)
Clinical presentation	• Asymptomatic (MC). **Ruddy skin**. Hypoglycemia, hyperbilirubinemia. R. distress, cyanosis, apnea. Irritability, jitteriness. Abd distension

(Choice C) In the **first 2 days of life**, **dehydration** (↑Hct) **is rare** in term neonates, as they are born with **excess extracellular water.**

Causes of pancytopenia	
Bone marrow aplasia	• Aplastic anemia. Infection (parvovirus, HIV, viral hepatitis). Nutritional deficiency (eg, vitamin B_{12}/folate). Hydroxyurea
Bone marrow infiltration	• Cancer (eg, hematologic, metastatic). Myelofibrosis. Infection (eg, tuberculosis, fungal infection)
Mature blood cell destruction	• Intravascular (eg, DIC, TTP). Extravascular (eg, hypersplenism)

Immunologic blood transfusion reactions			
Reaction	**Onset**	**Cause**	**Key features**
Anaphylactic	Within secs to minutes	Recipient **anti-IgA antibodies** directed against donor blood IgA	• Angioedema, hypotension, R distress/wheezing,shock IgA-deficient recipient
Acute hemolytic	Within 1 hr	ABO incompatibility	• Fever, flank pain, hemoglobinuria, DIC • Positive Coombs test
Febrile nonhemolytic (most common reaction)	Within 1-6 hr	**Cytokine** accumulation during blood storage	• Fever & chills
Urticarial	Within 2-3 hr	Recipient IgE against blood component	• Urticaria
Transfusion-related acute lung injury (tx: respiratory support alone)	Within 6 hr	Donor **anti-leukocyte antibodies**	• R distress, hypotension • Noncardiogenic pulmonary edema with bilateral infiltrates
Delayed hemolytic	Within days to weeks	Anamnestic antibody response	• Often asymptomatic, hemolytic anemia. (+) Coombs test, (+) new antibody screen

Graft versus host (skin, biliary, GI, no lymphadenopathy)	Within weeks	Donor T lymphocytes	• Rash, fever, GI symptoms, pancytopenia

AHTR Mx: immediate cessation of transfusion, aggressive IV fluids, and supportive care.

Transfusion-associated circulatory overload (<6 hr following transfusion initiation)	
Risk factors	• Age <3 and >60. Underlying cardiac or renal condition/ chronic anemia. Large transfusion volume/fast infusion rate
Clinical features	• Respiratory distress, ↑HR BP P. edema (rales) S3 gallop. ↑ JVP
Management	• Oxygen and furosamide

Transfusion reactions		
	Anaphylactic	**Urticarial (common, mild)**
Mechanism	• Anti-IgA antibodies (IgG or IgE) in IgA-deficient patient against donor blood IgA	• Preformed recipient IgE antibodies against soluble allergen in donated plasma
Onset	• Seconds to minutes	• Hours
Findings	• R distress/wheeze. Angioedema, hypotension, hives	• Hives. Itching
Treatment	• Immediate cessation of transfusion+ Epinephrine • Antihistamines, oxygen, fluids & vasopressors	• Immediate cessation of transfusion + Antihistamines (extensive urticaria) • Resume transfusion if patient is otherwise asymptomatic

Urticarial: Diagnosis is confirmed **if symptoms improve and no signs of anaphylaxis develop**. The **transfusion** can then be **resumed without additional evaluation**.

(Choice C) Transfusion-transmitted bacterial infection (**TTBI**) : fever, tachycardia, and hypotension around 30 minutes after transfusion completion. Occurs with **platelet transfusions** because platelets are stored at room temperature, increasing their susceptibility to bacterial growth. Cultures obtained from the recipient & from the transfused product to identify the pathogen and potential source of infection, respectively.

Hemophilia A & B (X- linked recessive, hemosiderin deposition & fibrosis)	
Clinical features	• Delayed/prolonged bleeding after mild trauma: Hemarthrosis, intramuscular hematomas, GI/GU bleeding, intracranial hemorrhage, dental extraction. **Complications: hemophilic arthropathy**
Laboratory findings	• ↑ Activated PTT. Normal PT & PC. Absent or ↓ factor VIII (hemophilia A) or factor IX (hemophilia B) activity
Treatment	• **Factor replacement. Desmopressin** for mild hemophilia A

Although severe hemophilic arthropathy may be visible on x-ray, MRI allows for earlier detection and characterization of the degree of joint damage. **Early prophylaxis** with factor concentrates can significantly **reduce the risk** of developing **arthropathy**.

Inhibitor development occurs in approximately **25%** of patients with **severe hemophilia A (frequent, spontaneous bleeds)** as a complication of treatment (recombinant factor VIII infusions **through a port**): **Increased bleeding frequency** or hemorrhage **refractory** to tx.

Regular screening for inhibitor development is performed when patients with **severe hemophilia receive factor infusions** as well as when an **inhibitor is clinically suspected.** Tx: (**recombinant activated factor VII, activated PCC**); such agents work downstream in the coagulation cascade to promote clotting **without the need for factor VIII**.

Bone marrow evaluation is **contraindicated** in severe hemophilia due to **bleeding risk**.

Vitamin K deficiency (Easy bruising, mucosal bleeding, GI bleeding) ↑ PT, ↑ PTT if severe	
Risk factors	• Malnutrition. CF, Biliary atresia. Celiac disease, IBD. Decreased production by bacterial flora (frequent antibiotic use)

TTP: Reduced activity of ADAMTS13, the enzyme that cleaves von Willebrand factor, acute and rapidly progressive disorder, abd pain, confusion, acute bleeding. (no chronic bruising)

Infantile vitamin K–deficient bleeding (not received prior medical care)	
Pathophysiology	• Low vitamin K stores (poor placental transfer, sterile gut, low content in breast milk). Inefficient vitamin K use by immature liver
Clinical features	• Day 2-7 of life (can occur up to 6 months): Easy bruising, umbilical, mucosal, GI bleeding, intracranial hemorrage
Labs	• ↑ PT. ↑ PTT (if severe). Normal platelet count/ **thrombin time (TT)**

Prevention	• Intramuscular vitamin K at birth

Afibrinogenemia is a rare, **AR** bleeding disorder characterized by **absent fibrinogen production**. Severe bleeding **at the site of the umbilical cord** after birth. ↑PT, PTT, **TT**

Immune thrombocytopenia (Chronic ITP: platelets <100,000/mm^3 **for >1 year**, evaluate for HIV, HepC, CMV, SLE, thyroid disorders)	
Etiology	• Platelet autoantibodies. Preceding viral infection
Clinical findings	• Petechiae, ecchymosis, epistaxis, hematuria
Laboratory findings	• Isolated thrombocytopenia <100,000/mm^3. Few platelets (size normal to large) on peripheral smear

Treatment (spontaneous recovery within 3 months)	• Children (MC in age 2-5)
	  ○ Observe if cutaneous symptoms only. Glucocorticoids, IVIG, or anti-D if bleeding
	• Adults
	  ○ Observation if cutaneous symptoms AND platelets ≥30,000/mm^3
	  ○ Glucocorticoids, IVIG, or anti-D if bleeding or platelets <30,000/mm^3

2nd line: rituximab, TPO receptor agonist, splenectomy (↑ life span & quantity of platelets)

(Choice E) Platelet transfusion is the tx for Bernard-Soulier syndrome: very large platelets on peripheral smear. Platelet transfusion is considered in severe cases of ITP (intracranial bleeding) but is typically avoided as it can lead to further platelet destruction.

Hemolytic uremic syndrome (STEC > Shigella, vascular damage + microthrombi)	
Clinical features	• Preceding bloody diarrhea. 1 week later: Fatigue, pallor. Bruising, petechiae. Oliguria, edema. AKI (↑BUN, Cr), thromobcytopenia, MAHA (schistocytes, ↑ bilirubin)
Treatment	• Fluid & electrolyte management. Blood transfusions. Dialysis

HUS: injury to small BV endothelium → **thrombotic microangiopathy** and **renal vascular occlusion** (afferent arteriole, glomerular capillaries) → **intrinsic AKI** (BUN/creatinine ratio <20:1, oliguria, hypertension, hematuria, proteinuria). Most children recover completely.

Platelet dysfunction in chronic kidney disease	
Pathogenesis	• ↑ urea → ↑NO → ↓ plt adhesion, activation & aggregation
Features	• Easy bruising, epistaxis, GI hemorrhage
Labs	• Normal platelet count. Normal coagulation studies
Treatment	• Desmopressin (↑ vWF secretion from endothelial cells): Only required if having bleeding symptoms or upcoming procedure

Peutz-Jeghers syndrome (Mx: upper & lower endoscopy + annual anemia screening)	
Etiology	• AD. Tumor suppressor gene mutation -> unregulated tissue growth
Clinical features	• Pigmented macules (eg, lips, buccal mucosa, palms/soles) • ≥2 gastrointestinal hamartomatous polyps: Abdominal pain due to obstruction or intussusception. Anemia. Rectal prolapse • ↑ Cancer risk (GI, breast, genital tract). (+) FHx. Dx: Genetic testing

Polyps are monitored every few years, and polypectomy is indicated if lesions are **large, symptomatic, or malignant.**

Posttransplantation lymphoproliferative disorder	
Pathogenesis	• **Immunosuppression (wks-months)** following **solid-organ or SC transplant** → suppressed **CD8** immunosurveillance → unchecked viral replication → **immortalized** lymphocytes or plasma cells • **EBV (95%)**, but other herpesviruses (**HHV8**) can trigger the disease
Manifestations	• Fever. Leukopenia. Lymphadenopathy & hepatosplenomegaly. Masses in nonlymphatic tissue
Diagnosis	• High viral **titers. Biopsy:** lymphoid or plasma cell proliferation
Treatment	• Reduce immunosuppression. Rituximab. EBV titer monitoring

(Choice E) Heart transplant rejection is generally marked by significant echocardiogram abnormalities (eg, reduced left ventricular ejection); lymphadenopathy is atypical.

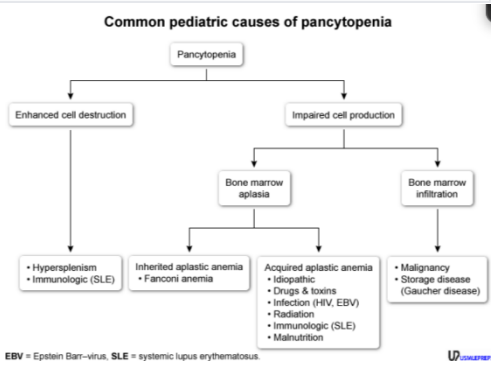
Acute lymphoblastic leukemia (MC childhood cancer. Peak age 2-5 years)

Clinical features	• Fever, WL, anemia, thrombocytopenia, bone pain (at night, femur & tibia), lymphadenopathy, headache, testicular enlargement • Mediastinal mass (T cell lineage): airway compression &/or SVC syndrome (↑JVP, cough, difficulty breathing) • Leptomeningeal spread: (eg, cranial nerve deficits, meningismus)
Evaluation & diagnosis	• CBC*. BM biopsy (>20% blasts is diagnostic) with flow cytometry • Lumbar puncture to evaluate for CNS involvement
***≥2 cytopenias (leukocytes may be ↓ or ↑), ± blasts on peripheral smear.	

With treatment (ie, multidrug chemotherapy), prognosis is favorable in children, with a 5-year survival rate of >85%.

In contrast to infectious causes of **lymphadenopathy**, malignant nodes are typically firm and **nontender**.

Aplastic anemia no bone pain (vs ALL)



Chapter 10: Rheumatology & Orthopedics

Legg-Calvé-Perthes disease (Idiopathic avascular necrosis of the femur)	
Clinical features	• **Boys age 3-12 (peak age 6)**. Insidious chronic hip pain (worse with activity, may be referred to thigh/knee/groin), limping. Limited ROM (Restricted hip abduction, internal rotation). Proximal thigh atrophy. Positive Trendelenburg sign, **antalgic gait** (avoids weight bearing on the affected side due to pain)
Diagnosis	• X-ray: Early: normal. Later stages: femoral head **flattening, fragmentation, sclerosis** (alternating regions of lucency and density, reflecting replacement of necrotic bone by new bone) • MRI: avascular/necrotic femoral head
Treatment	• Non–weight bearing. Splinting, possible surgical repair

(Choice A) Osteomyelitis affects the **metaphyses** of long bones (humerus, femur) and classically presents with **fever, point tenderness, and localized edema**.

Slipped capital femoral epiphysis (SCFE): displacement of the proximal femur relative to the femoral head due to weakening of the **femoral growth plate** (physis). **Obese**, adolescent (age **10-14**) **boys**. **Hypothyroidism** (↓ **ossification** of the growth plate, earlier age of onset **(<10)**, more likely to develop **bilateral** disease).

Symptoms: **insidious** onset of hip, thigh, or knee pain (worse with activity/minor trauma) + limping. **Limited internal rotation** of the hip, and the thigh externally rotates during passive hip flexion, **atrophy** of the **quadriceps** and **gluteus muscles** due to chronic disuse. Complications: AVN, osteoarthritis. Unilateral SCFE: antalgic gait/ bilateral: waddling gait.

1st step: bilateral hip x-ray: **posteriorly displaced femoral head on frog-leg lateral hip radiograph is diagnostic**. Patients should be prescribed **non-weight–bearing status** and may require **bedrest if SCFE is bilateral**. Definitive treatment is prompt **surgical screw fixation** to stabilize the physis and prevent further slippage. Closed reduction is **not typically recommended** because it can lead to **avascular necrosis**.

Trendelenburg sign: (DDH, SCFE, avascular necrosis of femoral head)

(Choice F) Stress fractures are **uncommon** in children and usually develop after **repeated tensile stress**. X-ray findings include **periosteal elevation and cortical thickening.**

Total hip replacement for avascular necrosis & chronic pain despite surgical screw fixation.

(Choice A) Iliopsoas bursitis: due to **overuse or trauma**. Patients **have hip pain and limited range of motion**, palpable click with manipulation of the hip.

Osteogenesis imperfecta (AD, COL1A1)	
Clinical features	• Mild to moderate (type I): Frequent fractures. Blue sclera. CHL. Short to normal stature. Dentinogenesis imperfecta. Joint hypermobility (ligamentous laxity) • Lethal (type II): In utero and/or neonatal fractures. Pulmonary failure

Dentinogenesis imperfecta: an opalescent blue-gray or yellow-brown discoloration of the teeth caused by discolored dentin shining through translucent and weak enamel.

Child neglect: poor dental hygiene: dental caries (**well-defined, painful areas of decay**)

(Choice C) Ehlers-Danlos syndrome: CTD caused by defective type V collagen. Joint hypermobility (eg, **subluxation, dislocation**) **but not recurrent fractures.**

Congenital clubfoot (talipes equinovarus): Deformity caused by **abnormal development** of the **talus bone**. Mostly **isolated** without other structural abnormalities. **Multifactorial**: **genetic**, **oligohydramnios**, underlying **chromosomal** or neuromuscular disorders (eg, **spina bifida, myotonic dystrophy**).

Plantar-flexed foot with an **adducted** and **internally rotated forefoot**. Congenital clubfoot is a **fixed deformity**, which can be seen on imaging as an affected foot in the same plane as the tibia and fibula **even with fetal movement**.

Tx: **gentle manipulation and stretching** with **serial molding casts**, ideally beginning in the neonatal period. Once an optimal position is achieved by casting, **long-term bracing** is typically required; **surgical correction** is reserved for refractory cases or recurrence of clubfoot after nonsurgical treatment. If untreated, clubfoot leads to an **abnormal gait with weight bearing on the lateral aspects of the feet.**

(Choice E) Positional clubfoot is a **flexible** condition in which the foot can be passively repositioned to a neutral or normal position postnatally. Tx: **reassurance and observation**. Can appear similar to congenital clubfoot on a single prenatal US image, but the foot would **not stay in the same plane as the tibia and fibula during fetal movement**.

Metatarsus adductus vs clubfoot		
	Metatarsus adductus	**Clubfoot**
Clinical features	• Medial deviation of forefoot • Neutral position of hindfoot • Flexible positioning typical	• **Medial/upward** deviation of forefoot **& hindfoot** • **Hyperplantar flexion** of foot • Rigid positioning typical (congenital clubfoot)

Metatarsus adductus (MA), MC congenital foot deformity, bilateral, first-born infants (crowded positioning in a smaller, primigravid uterus). Clinical dx. Reassurance

For the minority of cases of MA that do not passively correct to a neutral position, stretching exercises and/or serial casting can be considered.

Congenital muscular torticollis (CMT): neck mass, ipsilateral **head tilt**, contralateral **chin deviation. Postural deformity** in which the **sternocleidomastoid** muscle is tight and contracted, due to intrauterine crowding (breech, multiple gestation, oligohydramnios).

CMT is evident from **birth** but typically comes to medical attention at **age 1-6 months** with a head preference to one side. When an infant lies down with the head facing the preferred

side, **positional plagiocephaly** **often occurs**, which entails **flattening of the head on that side** as well as **anterior displacement of the ear and forehead ipsilateral to the flattening**. **Limited ROM of the neck**. **SCM thickening/well-circumscribed mass** from muscle fibrosis may be palpable.

Treatment strategies include positioning (eg, increased tummy time), passive stretching, and physical therapy. **Missed or delayed diagnosis** can lead to craniofacial asymmetry.

(Choice A) Cystic hygromas: **fluctuant mass that transilluminates**.

Growing pains (idiopathic nocturnal pains of childhood) MCC of MSK pain in age 3-12	
Clinical features	• **Episodic, non progressive bilateral lower extremity** (eg, thighs, calves, shins) pain: Occurs primarily at **night**, after increased daytime physical exertion. No systemic symptoms, joint involvement, or activity limitations. Normal physical examination
Treatment	• Parental education & reassurance. Massage, heat, analgesics, or stretching exercises
Course	• May wax/wane for months to years. Self-resolves by adolescence

Growing pains are a **clinical diagnosis**; pathologic causes should be considered for fever, weight loss, joint inflammation, limp, progressive or unilateral pain.

Strength training (resistance training) in children	
Benefits	• ↑ Muscle strength & joint stability. ↑ Endurance & coordination. ↑ Muscle mass (**if pubertal/postpubertal**)
Preparticipation assessment	• Cardiac and musculoskeletal **examination** (NOT imaging). Assessment of patient maturity (eg, ability to follow directions), typically **age ≥8**
Safety guidelines	• Proper supervision & equipment. Avoid excessive load
Contraindications	• HOCM, pulmonary hypertension, uncontrolled hypertension

Children should engage in **≥1 hour** of **daily moderate to vigorous** physical activity, which typically consists of **aerobic exercise**, such as hiking, running, jumping, or swimming, **with or without** strength training.

Radial head subluxation (nursemaid's elbow) (age 1-5)

Mechanism	• **Axial traction** on forearm with elbow extended (child pulled, lifted, or swung by arm)
Physical findings	• Arm held extended & pronated (attempted forearm supination will be resisted and cause the child to cry out in pain) • No swelling, deformity, or focal tenderness
Treatment	• Hyperpronation of forearm OR Supination of forearm **&** flexion of elbow

No post-reduction films are needed when the patient resumes full use of the extremity. A **pop** may also be heard on successful reduction. Clinical dx as **radiographs** are often **normal**. Elbow radiographs may be obtained to exclude fractures in **children age >5 with elbow pain** (in whom radial head subluxation would be uncommon) and in **children <5 with a high-force injury, focal swelling, or deformity**.

(Choice A) Lateral epicondylitis ("tennis elbow") is caused by repetitive contraction of the extensor muscles at the lateral epicondyle. Tenderness is elicited over the lateral epicondyle, and pain is felt with **passive wrist flexion and resisted wrist extension**.

(Choice B) Medial epicondylitis ("golfer elbow") manifests as localized tenderness over the medial epicondyle, pain with resisted wrist flexion, and pain with passive wrist extension.

➢ **Panner disease**: **osteochondrosis of the capitulum**, adolescent actively engaged in throwing sports. Chronic dull pain, crepitation, loss of pronation & supination.

DDH (abnormal development of femoral head & acetabulum)	
Risk factors	• Breech. FHx. Female. White ethnicity. Excessively tight swaddling
Clinical features	• Red flags: (+) Ortolani test. Dislocated hip. Limited hip abduction • Supportive findings: (+) **leg-length discrepancy** (eg, Galeazzi test). Asymmetric gluteal/inguinal/thigh creases
Management	• Red flags: refer to orthopedic surgery • Supportive findings or risk factors: Age <4 mo: hip US. if >4 mo x-ray

Because many patients have no risk factors, **all infants** should have serial hip **examinations** from birth **until age 1**. Infants with abnormal examination should undergo bilateral hip **US** or x-ray. Confirmed DDH is treated with a Pavlik harness, a splint that holds the hip in flexion and abduction while preventing extension and adduction.

Transient synovitis vs septic arthritis		
	Transient synovitis	**Septic arthritis**
Clinical presentation	• Well-appearing. Afebrile or low-grade fever. Able to bear weight	• Ill-appearing. Febrile. Non–weight-bearing
Diagnosis	• Normal or mildly elevated WBCs, ESR, CRP • Unilateral/**bilateral** joint effusion on ultrasound. Dx of exclusion	• Moderately elevated WBCs, ESR, CRP. ± Positive blood culture • **Unilateral** joint effusion on ultrasound. Synovial fluid WBCs >50,000/mm^3
Treatment	• Conservative	• Joint drainage & antibiotics

Transient synovitis, well appearing, boys **age 3-8** with **hip pain** (midly restricted ROM) **and limp/ referred knee pain following a viral illness/** posttraumatic (eg, gymnastics class)**, lasts <4 wks**. **X-ray may be normal or show uni/bilateral hip effusion**. Conservative management with **rest and analgesics** (NSAIDS)**, self limiting in 1-2 wks,** recurrence rare (<15%). Patients tend to hold the affected hip in flexion, slight abduction, and external rotation (to maximize the joint space) and resist extension and internal rotation.

Sometimes patients can have **overlapping clinical features**, i.e. low-grade fever and mildly elevated ESR (suggestive of transient synovitis) but with severe pain, inability to ambulate, and leukocytosis with neutrophil predominance (suggestive of septic arthritis).

When the dx is unclear: **bilateral hip ultrasound** . If unilateral effusion, do arthrocentesis.

Nonaccidental trauma (MCC of death in child abuse: brain injury)	
Red flags	• Injury inconsistent with developmental stage. Delay in seeking care. Conflicting historical details
Clinical features	• Bruises: patterned appearance (eg, belt buckle), occurring in nonmobile child or over noninjury-prone area (eg, ear, trunk) • Burns: linear, well demarcated. • Fractures: Multiple & in various stages of healing. Femur fx in nonambulatory child. Posterior rib fx. Metaphyseal corner fx (**forcible pulling or twisting** of an extremity)

	• Seizure, disproportionately large HC, full fontanelle, papilledema, retinal hemorrhages, AMS (from subdural hemorrhages). Malnutrition. Sudden behavioral or scholastic changes, **bedwetting**, and/or **academic difficulties**.
Management	• Disposition: ensure immediate safety of the child (inpatient mx, child protective services). Document objective findings • Evaluation: **skeletal survey**, CT scan of the head, funduscopy

Infants are particularly susceptible because they have a relatively large, heavy head; weak neck musculature; immature brain myelination; and a soft brain with high water content.

The **most common mechanism** of inflicted injury in infants: **violent shaking** → repetitive acceleration-deceleration forces → subdural bleeding due to **shearing of the bridging veins** → coup-contrecoup injury as the brain impacts the skull.

Features of possible child abuse	
Risk factors	**Caregiver background** • Young or single parents. Lower education levels. Substance use disorder. Psychiatric conditions (depression, impulse control disorders). History of childhood abuse/neglect **Home environment** • Unstable family situation (eg, divorce, conflict). Financial difficulties, job loss. Lack of social support. Domestic violence **Children** • Physical, intellectual, or emotional disabilities. Unplanned pregnancy/unwanted child. Children with chronic medical problems/ congenital defects, premature babies
Medical child abuse (Munchausen syndrome by proxy)	
When to suspect	• **Persistent or recurrent unexplainable illness**: Nonphysiologic symptom clusters. Inconclusive diagnostic testing. Ineffective medical therapies • Reported signs/symptoms incongruous with clinical findings or **seen only in presence of specific caregiver**

	• Siblings who have died or have unexplained illness • Seeking care from multiple facilities or specialists • **Reluctance to accept less severe diagnosis**
Management	• Report to Child Protective Services. Case-specific medical evaluation for child abuse (eg, urine toxicology screen)
Communication strategies	• Use direct, honest & specific language (eg, "**I suspect abuse**") • Inform family of legal mandate to report. Focus on best interest/safety of child. Avoid assigning blame

Osgood-Schlatter disease is a common, benign cause of knee pain caused by overuse in **young adolescents**. During periods of rapid growth (typically age 13-14 for boys and 10-12 for girls), the quadriceps muscles pull on the insertion site of the patellar tendon. Pain occurs at this site, which is located on the apophysis of the tibial tubercle. This **traction apophysitis** may lead to elevation and chronic avulsion of the tibial tubercle.

Patients have **progressive unilateral pain** over the tibial tuberosity that worsens **with activities** involving repetitive running, jumping, or kneeling (eg, soccer, basketball, gymnastics) and improves with rest. Pain can be reproduced by squatting or extending the knee against resistance. Clinical dx. However, x-ray may be indicated for atypical findings (eg, erythema, pain unrelated to activity) and may reveal soft tissue swelling anterior to the tibial tubercle. Treatment is supportive: NSAIDS, activity as tolerated, and physical therapy. Symptoms self-resolve on ossification of the tibial growth plate in adolescence.

- **Patellar tendinitis**: overuse injury due to repetitive jumping (basketball, volleyball). Ant knee pain that worsens with activity, point tenderness at the inferior pole of the patella.
- **Patellofemoral pain**: overuse injury, progressive knee pain localized to the patella in the setting of running and squatting. Sensation of instability or "buckling" at the knee.
- **Prepatellar bursitis**: direct, chronic trauma (eg, wrestling) of the anterior knee. Pain with direct pressure and **superficial swelling over the patella**.

Supracondylar fracture (MC type of pediatric elbow fx, age 2-7)	
Features	• High-impact fall onto an outstretched arm with the elbow hyperextneded. Pain, swelling, limited range of motion
Diagnostic findings	• X-ray with wide anterior fat pad (normally narrow or absent) and a posterior fat pad (occult). Fx line, or displacement of humerus

Treatment	• Nondisplaced: long arm splint & sling • Displaced: surgical reduction & pinning
Complications	• Neurovascular injury. Compartment syndrome (**displaced fracture** or concomitant forearm fracture are at increased risk, Features: **increasing swelling and pain** that is unresponsive to escalating analgesics, Tx: removal of any bandages, measurement of compartment pressures, and emergency fasciotomy)

(Choice A) Avascular necrosis of the humeral physis is a **rare** complication that presents with **insidious pain and decreased ROM months to years** after a supracondylar fracture with vessel injury.

Spiral fractures of the **distal tibia (toddler's fractures)**: **ambulatory** children **age ≤3**, **twisting injury** during a low-impact fall in the early walking years, pain (elicited with ankle dorsiflexion or twisting of the knee and ankle in opposite directions), limp, refusal to bear weight. Fracture site tenderness and leg swelling. X-ray may be **normal** initially but usually shows a **hairline fracture**. **Tx**: immobilization and pain control with **no additional workup**.

Distal forearm fractures: common in children, **fall onto an outstretched hand**. Patients have pain, swelling, and limited range of motion of the wrist.

Greenstick fracture: Because the periosteum surrounding the bone is thick and strong in children, the fracture may involve only one side of the bony cortex rather than extending through the width of the bone. The **opposite side** appears to have a **deformation or bend without a break in the cortex**. **Tx**: Prompt **reduction** (if displaced) and **immobilization** of the forearm (in unstable fxs due to the potential for refracture or further displacement if improperly treated). **Repeat x-rays** should be performed **prior to cast removal** to confirm **bony union**. Once the fracture is fully healed, **no long-term complications** are expected.

(Choice C) Fracture involving the **growth plate** (physis) can lead to **LL discrepancy**.

(Choice D) Long-term limited range of wrist supination and pronation is a complication of **malunion after a severely displaced radial fracture**.

Characteristics of physiologic genu varum: Symmetric bowing. Normal stature. No LL discrepancy. No lateral thrust when walking. Clinical dx, reassurace, self-limiting by age 2.

- Blount disease: Pathologic bowlegs due to abnormal cartilage growth. Features: LL discrepancy, asymmetrical bowing, lateral thrust with ambulation. Tx: surgical.

Calcaneal apophysitis (Sever disease) [repetitive microtrauma to the heel growth plate]	
Risk factors	• Running/jumping sports. Growth spurts (age 8-12). Athletic cleat use or footwear without heel padding (flip flops)
Clinical features	• Chronic heel pain (50% bilateral). Pain with palpation at the base of the heel, calcaneal compression test. Decreased gastrocnemius/ soleus flexibility + tight achilles tendon → limited ankle dorsiflexion
Treatment	• NSAIDs, ice. Activity limitation, stretching, heel cup insert

Symptoms typically improve within a couple of months.

(Choice A) Pain associated with Achilles tendinopathy is exacerbated by running and jumping. However, tenderness is over the Achilles tendon (posterior ankle), not the heel.

Postural kyphosis is not due to a bony abnormality, the curvature is not rigid, and the kyphosis corrects in the supine and prone positions. Tx: reassurance

Vs **Scheuermann kyphosis** (abnormal anterior vertebral wedging, pain **worse** with flexion/activity & improves with rest. PEx: **hyperkyphosis** of the thoracic spine and compensatory **hyperlordosis** of the lumbar spine). Diagnosis of Scheuermann kyphosis requires lateral spine x-rays demonstrating kyphosis >40 degrees and >5 degrees of anterior wedging in 3 or more adjacent vertebrae. **Tx**: back strengthening and stretching exercises + avoidance of activities that incite pain or involve prolonged flexion. In addition, patients are **monitored routinely during their teenage years**; if kyphosis progresses to >50-60 degrees, bracing and surgical interventions may be warranted.

Adolescent idiopathic scoliosis: Asymptomatic children age ≥10. Rib hump, asymmetric scapulae, unilateral thoracic/lumbar prominence on forward bend test. **Spine x-ray** (AP and lateral) is used to assess skeletal maturity, measure the degree of curvature (ie, Cobb angle), and rule out other deformities. Cobb angle >10 degrees is consistent with scoliosis.

Management of scoliosis is based on the risk of **curve progression**. The greatest increase in curvature occurs during early adolescence at the time of the pubertal growth spurt, after which progression slows and subsequently **stops** once **bone ossification** is **complete**.

Risk factors for curve progression: Female. Age <12. Early pubertal status (premenarchal, should be monitored until 2 yrs after menarche). Skeletal immaturity. Severe curvature (Cobb angle ≥25 degrees).

BMI at the ≥85th percentile is associated with increased **severity** of scoliosis at diagnosis (possibly due to delayed detection) but is not a risk factor for curve progression.

(Choice B) A positive family history increases the **likelihood of developing scoliosis** but does not predict the severity of disease or increase the risk for curve progression.

(Choice E) Left thoracic curves may be associated with (**tumors**) in **children age <10**.

Spondylolysis: unilateral or bilateral **fatigue fracture** in the **pars interarticularis**, **overuse injury** in athletes who perform repetitive back extension and rotation. Central low back **pain with lumbar extension** ± radicular pain. Flexion offloads the fracture site and improves the pain. Pain is **worse with activity** and improves with rest, nighttime pain is rare. Deep palpation is required to elicit tenderness at the fracture site.

AP and lateral views are diagnostic; the oblique view is not recommended due to the added radiation exposure and limited sensitivity. Tx: **limit activity** (ie, abstain from sports) and continue conservative management (eg, analgesics, ice/heat) for **90 days** to allow the fx to heal. After 90 days, patients who improve can gradually go back to their activities.

Spondylolisthesis [age 10-19 yrs, due to increased physiologic lumbar lordosis (exposing L5 vertebra) and decreased bone mineralization]	
Pathophysiology	• **Bilateral** pars interarticularis defects (spondylolysis) → **anterior slippage of vertebral body**. Most common: **L5 slips over S1**
Risk factors	• Repetitive back extension & rotation (eg, gymnasts, divers). Adolescent growth spurt
Clinical features	• Low back pain that is worsened by extension. Radiculopathy as slippage progresses. **Palpable step-off** may be present
Diagnosis	• Lumbar x-rays (typically visible on lateral views)
Management	• Activity modification (avoid inciting sports), pain control • **Neurologic deficits or symptoms >90 days**: obtain MRI of spine & surgical consultation

- Lumbar disc herniation (pain worse with **flexion**) and lumbosacral strain (**nonspecific pain**) are common causes of back pain in adults.

Down syndrome comorbidities	
Neurologic	• Intellectual disability. Early-onset Alzheimer disease (age 40-60)
Cardiac	• Complete atrioventricular septal defect (MC). VSD. ASD

Gastrointestinal	• Duodenal atresia, Hirschsprung disease
Endocrine	• Hypothyroidism. Type 1 DM. Obesity. Short stature
Other	• ALL. Atlantoaxial instablity. LBW. Hypotonia (floppy baby, protruding tongue)

❖ **Complete atrioventricular septal defect** is the most common heart defect in Down syndrome. Can hear **ASD VSD MR** murmurs. **Diaphoresis and dyspnea with feeds and crackles at age 6 weeks** as PVR falls. increased pulmonary markings and cardiomegaly

❖ In contrast to infants with normal tone who have **extremities flexed** toward their bodies when held vertically, those with low tone may **slip through the examiner's hands** with **arms and legs extended** and floppy.

Atlantoaxial instability: >10% of patients. Excessive laxity in the posterior transverse ligament increases mobility between the atlas (C1) and the axis (C2). **Mostly asymptomatic**. Weakness, gait changes, urinary/fecal incontinence, dizziness, vertigo, imbalance, and diplopia, spasticity, hyperreflexia, and (+) Babinski sign. **Diagnosis**: lateral x-rays of the cervical spine in flexion, extension, & neutral position. Open-mouth x-rays can be helpful in visualizing the odontoid. **Tx:** surgical fusion of C1 to C2.

- Trisomy 21 is associated with umbilical hernia (**abdominal wall** defect covered by **skin**) and duodenal atresia, which is characterized by a "double bubble" and polyhydramnios on ultrasonography. In addition, AFP is reduced in trisomy 21.

Unicameral bone cyst (fluid filled)	
Epidemiology & pathophysiology	• Skeletally **immature** children. **Solitary**, benign cyst with fibrous lining in **medullary** cavity of long bones (eg, humerus, femur)
Clinical features	• Usually **asymptomatic** & found incidentally on imaging ± Localized pain or pathologic fracture (medullary expansion)
Diagnosis	• X-ray: **well-defined lucent** lesion arising from **metaphysis** with **narrow transition zone & no periosteal reaction** • MRI: **homogeneous cyst with enhancing rim**
Treatment & prognosis	• **Observation** with serial x-rays until spontaneous resolution (typically by the time of physeal closure)

(Choice A) Giant cell tumor can be asymptomatic and found incidentally on x-ray as a **radiolucent** lesion with a "soap bubble" appearance (bony trabeculae within the lesion)

and a **narrow zone of transition**. Epiphyses, skeletally mature patients, age 20-40 females. Locally aggressive, often causing marrow edema and extending into soft tissue.

(Choice C) Osteomyelitis: radiolucent lytic lesion with a periosteal reaction + marrow edema and cortical destruction. Histopathology: inflammatory cells, necrotic bone, and irregular bony trabeculae.

(Choice D) Osteosarcoma: MC bone malignancy in children, boys age (13-16). Localized pain (tender soft tissue mass), constitutional symptoms absent, ↑AlkP LDH (turnover of damaged osteocytes; high levels may correlate with adverse prognosis), ill-defined lesion of radiodense and radiolucent areas with an aggressive periosteal reaction.

Ewing sarcoma (2nd MC pediatric bone malignancy, white males age <15)	
Clinical features	• Chronic, localized pain & swelling/erythema/warmth (↑ at night/ activity, rapidly progressive). Long-bone diaphyses & pelvis. ± fever, WL, leukocytosis, ↑ESR. Early metastasis (lung, bone, bone marrow)
X-ray findings	• Central poorly defined lytic lesion surrounded by concentric layers of new bone "Onion skinning" (lamellated periosteal reaction). "Moth-eaten"/mottled appearance (soft tissue extension). Codman triangle. Tx: **surgery + chemoradiotherapy**

- **Biopsy** is **diagnostic** and shows sheets of uniform, small, round blue cells with scant cytoplasm. Fibrous septa separating the sheets of cells are typical.

(Choice B) Fibrosarcoma: rare, malignant spindle cell neoplasm, painful mass in patients age >30. Osteolytic lesion with **well-defined or ragged, "moth-eaten" margins**.

Langerhans cell histiocytosis (Bone most commonly affected)	
Clinical findings	• Lytic bone lesions (skull, jaw, femur). Skin lesions (purplish papules, eczematous rash). Lymphadenopathy, hepatosplenomegaly. Pulmonary cysts/nodules. Central DI. Langerhans cells on bone/skin biopsy
Treatment	• Chemotherapy (prednisone ± vinblastine). Curettage • Desmopressin for diabetes insipidus

Bone lesions may be asymptomatic or associated with local pain/tenderness. X-ray shows characteristic "punched-out" **lytic lesions**; biopsy is required to confirm the diagnosis.

Osteochondroma: static through adulthood. Mx: **observation as the risk of malignant transformation** in adulthood is very low (1%) for a solitary lesion, but patients should be

counseled on worrisome signs (growth after physeal closure). Asymptomatic or firm mass but can rarely lead to pathologic fractures (break in the stalk of the pedunculated lesion).

	Clinical features*	X-ray findings	Natural course
Osteochondroma (most common)	Hard, palpable nodule on long bone Usually painless	Bony spur extending from metaphysis & away from joint Contiguous with medullary cavity & cortex	Growth until physeal closure
Osteoid osteoma	Nocturnal pain in long bone or spine relieved by NSAID	Small, round radiolucency with sclerotic margins	Spontaneous resolution
Unicameral bone cyst	Asymptomatic or localized pain in long bone	Well-defined radiolucency with narrow transition zone in metaphysis	
Chondroblastoma	Joint pain/swelling involving long bone	Well-defined radiolucency with thin sclerotic margin in epiphysis	Locally aggressive with extension into growth plate/soft tissue
Giant cell tumor	Joint pain/swelling involving long bone Age >20	Well-defined radiolucency with soap-bubble appearance in epiphysis	

*Classically present at age 10-20 unless otherwise noted.

Osteoid osteoma (Benign, **bone-forming** tumor, males <25 years)	
Clinical features	• Diaphysis & cortex of proximal femur (MC), tibia and spine. Pain: Worse at night. Relieved by NSAIDs. Unrelated to activity. No systemic symptoms
X-ray	• Small, round well-circumscribed lucency with sclerotic margins ± central ossification
Tx	• NSAIDs. Self limiting over several years. Serial x-rays. Surgery for refractory symptoms

Osteoid osteoma contains a small, osteoid nidus that produces high levels of **prostaglandins**, which cause localized inflammation and pain. Examination is often **normal**, although localized tenderness/ deformity/ swelling may be present.

(Choice C) Lumbosacral paraspinal muscle strain is one of the most common causes of **low** back pain in adolescents. Paraspinal tenderness on examination, rarely lasts 12 wks.

Differential diagnosis for nontraumatic joint swelling			
	Infectious	**Inflammatory**	**Neoplastic**

Onset	Acute	Subacute/chronic	Subacute/chronic
Timing of pain	Constant	Worse in morning	Worse in evening/night
Able to bear wt?	No	Yes	Variable
Multiple joints?	Uncommon	Common	Variable
Laboratory findings	↑ WBCs/platelets ↑ Inflammatory markers	↑ WBCs/platelets, ↓ RBCs ↑ Inflammatory markers	↓ WBCs/platelets

Juvenile idiopathic arthritis (MCC of arthritis in children, oligoarticular is the MC form)

Subtype	Frequency	Age of onset	Clinical features	Sex ratio
Systemic	10%	Age <18	• **Arthritis** (worse in morning) in ≥1 joint for ≥6 weeks. Quotidian fever for ≥2 weeks. Evanescent rash (pink/macular, worsens during fever). HSM. Lymphadenopathy	F = M
Polyarticular	40%	Age 2-5, 10-14	• Arthritis ≥5 joints (6 mo) ± uveitis	F > M
Oligoarticular	50%	Age 2-4	• Arthritis <5 joints. ± uveitis	F > M

Polyarticular: Symmetric involvement of elbows, wrists, knees, and ankles ± small joints of the hands and feet (in older children). **No fever/rash**. **Tx: NSAIDS (1st line), DMARDs**

Oligoarticular: **Painless** joint swelling (**knees, ankles**), no fever, rash. ANA is a risk factor for uveitis (regular ophthalmology screening). Normal leukocyte count, hemoglobin, ESR. **Tx: NSAIDS, intraarticular glucocorticoids, DMARDs**

Disseminated gonococcal infection (Tx: IV ceftriaxone) [synovial fluid leukocyte count **<50,000. Negative culture**]

Manifestations	• Purulent monoarthritis OR Triad of migratory polyarthralgia, tenosynovitis, **pustular** dermatitis
Diagnosis	• Detection of *Neisseria gonorrhoeae* in urine, cervical, or urethral sample (NAAT). Culture of blood, synovial fluid (less sensitive)

IgA vasculitis (Henoch-Schönlein purpura)	
Pathogenesis	• Perivenular leukocytoclastic (neutrophils & monocytes) vasculitis • Deposition of IgA, C3 & fibrin in small vessels

Clinical manifestations	• Classic findings*:
	  ○ **MC: Nonblanching** palpable purpura/petechiae on lower extremities. The rash **coalesces** and darkens over days.
	  ○ Arthritis/arthralgia (lower limbs -> refusal to walk)
	  ○ **Intestinal edema** -> Colicky Abd pain, N V GI bleeding, **intussusception (ileoileal)**
	  ○ Renal disease (similar to **IgA nephropathy**)
	• Other findings: scrotal pain & swelling. Blood flow to the testicles is normal or increased, and scrotal symptoms typically self-resolve.

Laboratory findings	• **Normal platelet count & coagulation studies**. Normal to ↑ creatinine. Hematuria ± RBC casts &/or proteinuria
Management	• Supportive care (hydration & NSAIDs) for most patients • Hospitalization & systemic glucocorticoids for severe symptoms
*Clinical diagnosis requires purpuric rash plus ≥2 additional classic findings.	

In **pediatric** patients with **classic** findings, **no additional diagnostic testing** is required. In adults (in whom IgA vasculitis is rare) and in children with an **atypical** or incomplete presentation, **skin biopsy** is the next diagnostic step, revealing neutrophilic infiltration with IgA, C3, and fibrin deposition within postcapillary venules. ↑ IgA and low C3 may be seen in some patients, are nonspecific. **Serial screening urinalyses should be performed**.

Chapter 11: Dermatology

Miliaria rubra (heat rash): Risk factors: Pt dressed in multiple layers of clothing (onesies, pajamas) and spends most of the day and night swaddled in a fleece blanket. Due to **delayed development/patency** of eccrine sweat glands -> sweat accumulation -> inflammation

Clinical manifestations include a fine (2-4 mm), erythematous, papular rash. It is typically located in **intertriginous areas** (eg, groin, axilla, anterior neck) or in areas where the skin is **occluded** (eg, back, head) by clothing, hats, or adhesive bandages. The rash is typically asymptomatic but may be pruritic, and infants are otherwise well appearing.

(Choice A) High-potency topical corticosteroids are **not recommended** in neonates due to the risk of systemic absorption enhanced by the thin stratum corneum in this age group.

Benign neonatal rashes

Diagnosis	Onset	Clinical features	Management/resolution
Erythema toxicum neonatorum	Birth to age 3 days	Pustules with erythematous base on trunk & proximal extremities	Observation Resolves within a week
Milia	Birth	Firm, white papules on face	Observation Resolves within a month
Miliaria rubra	Any age, but not present at birth	Erythematous, papular rash on occluded & intertriginous areas	Avoid overheating (eg, cool environment, thin/cotton clothing) If severe, topical corticosteroid
Neonatal pustular melanosis	Birth	Nonerythematous pustules → evolve into hyperpigmented macules with collarette of scale Diffuse, may involve palms & soles	Observation Pustules resolve within days Hyperpigmentation may last months
Neonatal cephalic pustulosis	Around age 3 weeks	Erythematous papules & pustules on face & scalp only	Observation Resolves in weeks to months If severe, topical corticosteroid or ketoconazole

Neonatal cephalic pustulosis inflammatory reaction to the ***Malassezia*** species that colonize the skin. Because androgen stimulation of sebaceous glands is not involved, "neonatal cephalic pustulosis" has replaced the term "neonatal acne" used previously to describe this condition. Comedones (hair follicles occluded by keratin) and nodulocystic lesions are not present. Patients are asymptomatic and well-appearing.

Most cases of neonatal cephalic pustulosis are mild and can be managed with daily cleansing of the affected area with **gentle soap and water**. **Oil-containing lotions or emollients should be avoided**.

(Choice C) Staphylococcal pustulosis: erythematous pustules, vesicles, or bullous lesions that rupture to form erosions and honey-colored crusts in neonates. Classic distribution = **diaper area** or a prior wound (eg, **circumcision site**). Tx: **topical mupirocin**

Congenital dermal melanocytosis (CDM, "Mongolian spots"): benign, flat, **blue-gray patches** in infants over the **lower back** and **buttocks**. Due the presence of melanocytes within the skin's dermis. Most infants of **African**, **Asian**, and **Hispanic** ethnicity have CDM at birth. No tx required as the hyperpigmentation usually **fades spontaneously** during the 1st decade of life. CDM should be documented as these patches can be mistaken for bruises, potentially raising concern for **coagulopathy or child abuse**.

Erythema toxicum neonatorum (ETN): innate immune response to skin bacteria penetrating **hair follicles**, anywhere **except the palms and soles** (where no hair follicles are present), but more likely in trunk and proximal extremities. **Features that should raise suspicion of an alternate diagnosis** include involvement of the palms and soles, lesions with crusting or a vesicular appearance, and systemic symptoms (eg, fever, irritability).

Sun-protective measures (**Sun avoidance** is the best method of photoprotection)	
Exposure	• Sun avoidance, especially age <6 months. ↓ exposure 10:00AM–4:00PM
Sunscreen	• **SPF ≥30**. Apply **15-30 minutes prior** to sun exposure (Not immediately before sun exposure). Reapply **every 2 hours** & **after** swimming
Clothing	• Long sleeves, broad-brim hats. Tight weave, **dark color**

(Choice C) Sunscreens with **SPF >50** provide a negligible increase in UV protection as compared to sunscreens with SPF 30-50 and are **not routinely recommended**.

Alopecia areata (Autoimmune attack on hair bulb cells. Genetic predisposition)	
Clinical presentation	• **Painless, patchy, nonscarring** hair loss (develops over wks, no erythema/scaling). Dermatoscope: Narrowing of hair shafts close to skin (exclamation point hairs). (+) hair pull test (>5-6 hairs extracted)
Management	• **Mild/moderate hair loss:** topical or intralesional corticosteroids • **Extensive hair loss:** topical immunotherapy (eg, diphenylcyclopropenone), oral corticosteroids

Circular, smooth, patches completely devoid of hair. A recurring course is common, but **most patients have regrowth over time**.

(Choice B) DLE: MC form of chronic, cutaneous lupus erythematosus. **Discrete, erythematous, well-demarcated scaling plaques**. Hypo- or hyperpigmented lesions, scarring, and photosensitivity. + additional lesions on the face or extremities.

(Choice C) Pediculosis capitis (head lice infestation): pruritus of the scalp and visible nits. It is not typically associated with hair loss.

(Choice D) Telogen effluvium: diffuse thinning of the hair. **It results from premature shedding** and is often triggered by stress (eg, major illness or surgery, malnutrition, pregnancy, endocrine disorders, emotional trauma).

(Choice E) Traction alopecia: hair loss in the frontal and temporal scalp, from braiding.

(Choice F) Androgenetic alopecia in females (also known as female-pattern hair loss) occurs in adult women, progressive hair thinning **at the vertex and frontal hairline.**

Tinea corporis (ringworm) [Dermatophyte infection, most commonly T rubrum]	
Risk factors	• Athletes who have skin-to-skin contact. Humid environment. Contact with infected animals (eg, rodents)
Presentation	• Scaly, erythematous, pruritic patch with centrifugal spread • Subsequent central clearing with raised, annular border
Treatment	• First-line/localized: topical antifungals (clotrimazole, terbinafine) • Second-line/extensive: oral antifungals (terbinafine, griseofulvin)

Individuals with **HIV infection or diabetes mellitus** may have multiple, widespread lesions. The diagnosis is apparent on inspection and can be confirmed with **potassium hydroxide** examination of skin scrapings in severe, atypical, or refractory cases.

(Choice D) Subacute cutaneous lupus erythematosus causes annular plaques (numerous lesions in sun-exposed areas of the upper body). Itching is uncommon.

Tinea pedis (*Trichophyton* species (most common type of dermatophyte infection))	
Clinical features	• **Acute:** pruritus, burning pain, erythematous vesicles/bullae • **Chronic:** pruritus, erythema, interdigital scales/fissures/erosions with **hyperkeratotic rash** extending onto the sole, side (**moccasin pattern**), or dorsum of the foot
Treatment	• Topical clotrimazole miconazole terbinafine, oral if unresponsive • Keep feet dry & dispose of old footwear

Risk factor: (Athletic facilities, swimming pools) while barefoot.

Tinea capitis [African American children, direct contact or fomite (eg, shared combs)]	
Clinical features	• Scaly, erythematous patch with hair loss on scalp ± Black dots in affected area ± Tender lymphadenopathy
Management	• **Oral** griseofulvin or terbinafine

The most common form in the United States is black dot TC (BDTC), which is due to ***Trichophyton tonsurans***. Patients develop an initial **scaly, erythematous plaque** on

the scalp that can progress to **patchy alopecia** with a residual black dot (**broken hair**). Other findings can include **inflammation, pruritus, tender occipital or postauricular lymphadenopathy, and scarring**. TC is often diagnosed clinically but can be confirmed with potassium hydroxide (KOH) examination of **hair stubs**. As dermatophyte carriers can be asymptomatic, many experts recommend that household contacts be treated with selenium sulfide or ketoconazole shampoo.

(Choice D) Pressure-induced (postoperative) alopecia is due to prolonged pressure on the scalp during surgical procedures. Transient hair loss develops a few weeks after the surgery, followed by regrowth without significant residual alopecia.

Tinea versicolor (pityriasis versicolor)	
Pathogenesis	• *M globosa* **skin flora** grows in exposure to hot & humid weather
Clinical features	• Hypopigmented, hyperpigmented, or mildly erythematous lesions (face in children, trunk & upper extremities in adolescents & adults) ± Fine scale ± Pruritus
Diagnosis	• KOH preparation shows hyphae & yeast cells in a "spaghetti & meatballs" pattern
Treatment	• Topical ketoconazole, terbinafine, or selenium sulfide

(Choice B) Irritant contact dermatitis might occur in a child using **new skin products and swimming in a chlorinated pool**. The rash is typically **erythematous** due to inflammation and may progress to dry, cracked skin. Discomfort and burning are common.

(Choice G) Staphylococcal scalded skin (exfoliatin) superficial flaccid bullae -> extensive exfoliation of the skin. It is most common in infancy and rarely occurs beyond age 5.

Staphylococcal scalded skin syndrome (exfoliative toxin, infants & young children)	
Clinical features	• Fever, irritability, skin tenderness. Generalized erythema -> blisters (axilla/groin). Epidermal shedding (Nikolsky sign). Perioral crusting (but intact mucosal surfaces)
Management	• Nafcillin, vancomycin. Wound care

In neonates, the source of infection is often the **umbilicus or circumcision site**, whereas in older children, there may be **nasopharyngeal colonization or a primary skin lesion** (eg, pustule under the naris). Cultures from the blood and potential infectious sources are

obtained; however, the diagnosis is generally clinical, and intact bullae are usually **sterile**. The mortality rate is low in children, and symptoms typically **resolve in 1-2 weeks**.

Scabies (close direct contact, incubation period 3-6 wks)	
Clinical features	• Burrows (thin serpiginous lines <1cm, female mites burrowing into the epidermis to lay eggs) may not be visible but pathognomonic if present. Extremely pruritic, small, erythematous papules/vesicles • Distribution: interdigital web spaces, flexor wrists, extensor elbows, axillae, feet, umbilicus & genitalia
Diagnosis	• Clinical. Skin scraping with microscopy is confirmatory
Treatment	• Topical 5% permethrin OR **oral** ivermectin • **Treat household members & close personal contacts** • Environmental measures (eg, launder clothing/bedding)

(Choice B) Crusted scabies: erythematous patches with **scale** in the **scalp, hands, and feet** in patients who are **immunodeficient**.

(Choice E) Topical corticosteroids (hydrocortisone) can relieve symptoms of **poison ivy dermatitis** (intensely pruritic linear lesions) and **bed bug infestations** (pruritic erythematous papules on exposed areas).

Perianal streptococcal dermatitis: perianal **pruritus** and pain, particularly with stooling, perirectal fissures and blood-streaked stools, constipation from withholding (due to **pain** while stooling). Although patients often do not have concomitant streptococcal pharyngitis, a close contact may have had a recent streptococcal infection (eg, cellulitis, pharyngitis). Dx: clinical, perianal bacterial culture.

Irritant diaper dermatitis presents with erythema and skin breakdown in the diaper area due to **prolonged exposure to urine or stool**, in the setting of **diarrhea**. Conservative measures to prevent irritant contact diaper dermatitis include: frequent diaper changes, diaper-free periods, and gentle cleansing with soap and water (or fragrance-/alcohol-free wipes). Low-potency topical corticosteroid ointment (hydrocortisone) for refractory cases.

Candidal dermatitis: Risk factors: **Diarrhea** (fecal bacteria increase the local pH), **recent antibiotic use, presence of oral thrush. Langerhans cell histiocytosis** can present with a rash similar to candida diaper dermatitis + **lytic bone lesions**.

Perianal dermatoses			
Diagnosis	**Irritant contact diaper dermatitis**	***Candida* diaper dermatitis**	**Perianal *Streptococcus***
Epidemiology	Most common diaper rash in infants	Second most common diaper rash in infants	Infants through school-aged children
Examination	Erythematous papules, plaques Spares skinfolds	Beefy-red, confluent plaques Involves skinfolds Satellite lesions	Bright, sharply demarcated erythema of perianal/perineal area
Treatment	Topical barrier (eg, petrolatum, zinc oxide)	Topical antifungal (eg, nystatin)	Oral antibiotics (eg, amoxicillin)

Impetigo [bacterial invasion through a break in the epidermis (eg, wound, eczema, burn)]		
Type	**Nonbullous**	**Bullous**
Microbiology	• *S aureus, GAS*	• *S aureus*–producing exfoliative toxin A
Clinical features	• Papules & pustules with **honey**-crusted lesions	• Enlarging, flaccid bullae with yellow fluid. Ruptured lesions with **collarette scale** at periphery
Treatment	• Limited: topical mupirocin • Extensive: oral cephalexin	• Oral antibiotics (eg, cephalexin)

Atopic dermatitis (eczema) [FHx of atopy, filaggrin gene mutation]	
Clinical features	• Acute: pruritic, erythematous patches & papules. Infant: extensor surfaces, trunk & face. Child/adult: flexural creases • Chronic: lichenified plaques
Treatment	• Topical emollients. First line: topical corticosteroids • Second line: topical calcineurin inhibitors (eg, pimecrolimus)
Prognosis	• Chronic with intermittent flares in early childhood

	• Usually resolves by adulthood

Lack of improvement after therapy raises suspicion for an **infectious complication**.

(Choice E) Wet wrap therapy uses wet clothing to cover skin treated with emollients or topical corticosteroid to enhance hydration and medication absorption. Used **for widespread or severe atopic dermatitis** & is **contraindicated with concurrent infection**.

Infectious complications of atopic dermatitis		
Diagnosis	**Pathogen**	**Presentation**
Impetigo (MC)	• *S aureus* • *S pyogenes*	• Papules & pustules with honey-crusted adherent coating ± Pain or pruritus
Eczema herpeticum	• HSV1	• Fever, irritability, lymphadenopathy. **Painful** vesicular rash with an erythematous base. Punched-out erosions & hemorrhagic crusting. Tx: immediate systemic acyclovir
Molluscum contagiosum	• Poxvirus	• Skin-colored papules with central umbilication
Tinea corporis	• *T rubrum*	• Pruritic circular patch with central clearing • Raised, scaly border

Pityriasis rosea (common, adolescence to young adulthood)	
Clinical features	• ± Viral prodrome. Annular, pink **herald patch** on trunk -> increases in size & develops scaling around the edge-> Within a week, **oval lesions in "Christmas tree" pattern**. ± Pruritus
Management	• Reassurance (spontaneous resolution) • Treatment of pruritus (eg, antihistamines, topical steroids)

(Choice B) Nummular eczema: chronic rash characterized by dry, erythematous, and intensely pruritic patches on the extremities.

Keratosis pilaris (KP): benign, retained **keratin plugs** in the **hair follicles**. It presents with small, painless follicular papules, a roughened skin texture, and mottled **perifollicular erythema**. KP can occur anywhere on the body but is most common on the **posterior**

surface of the upper arm. It is usually **asymptomatic**, although **pruritus** may sometimes occur; exacerbations are common in **cold, dry weather**.

KP can occur alone or in association with **atopic dermatitis or ichthyosis vulgaris** (an inherited disorder characterized by chronically rough, scaly skin). Clinical dx. **Treatment**, when necessary, includes **emollients** (eg, creams containing petrolatum or mineral oil) and **topical keratolytics** (eg, salicylic acid, urea).

(Choice D) Pseudofolliculitis presents with small, painful papules caused by growth of the hair shaft into the perifollicular skin. Individuals with tightly curled hair (beard).

(Choice E) Papulopustular rosacea: small papules, pustules, and skin sensitivity typically on the convex surfaces of the central face. Symptoms are aggravated by hot or spicy foods, alcohol, sun exposure, or high temperatures. Tx: topical metronidazole

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Seborrheic dermatitis in infants (resolves spontaneously)	

Clinical features	• Scalp (cradle cap), face (eyebrows), retroauricular areas
	  ○ Yellow, greasy scales ± erythematous, scaly plaques
	• Intertriginous regions (eg, neck folds, axillae, diaper area), umbilicus
	  ○ Glistening without active oozing, confluent erythema
	• ± Mild pruritus; no systemic symptoms

Treatment	• First-line: emollients, nonmedicated shampoos • Second-line: topical antifungals or low-potency corticosteroids

The incidence peaks in the **first year of life** (usually by age 3 months) and again in adolescence/adulthood. Parents can be instructed to apply topical emollients and gently cleanse the area with a soft brush to remove scales if desired.

(Choice C) Candidal intertrigo: moist erythema in opposing skin surfaces (eg, neck folds, axillae) + satellite papules/pustules with peripheral scales.

Infantile hemangioma (Appears days to weeks after birth)	
Natural history	• Proliferation (age 0-6 mo): growth of bright red, soft, raised plaque • Involution (age >6 mo): deep red/violet lesion that regresses in size
Evaluation	• Clinical diagnosis

	• Special considerations:
	  ○ ≥5 cutaneous lesions → liver ultrasound
	  ○ Facial/segmental → echocardiography & MRI of the head (ie, PHACE)
	  ○ Cervicofacial (beard distribution) → laryngoscopy
	  ○ Lumbosacral → spinal ultrasound

Management	• Observation for most lesions
	• **Oral** propranolol for high-risk features:
	  ○ Large, facial, segmental, &/or rapidly growing (ulceration/scarring)
	  ○ Periorbital (visual impairment, strabismus [eyelid])
	  ○ Hepatic (high-output heart failure)
	  ○ Subglottic (airway obstruction)

PHACE = posterior fossa anomalies, **h**emangioma, **a**rterial, **c**ardiac, **e**ye anomalies.	

(Choice C) A nevus flammeus, or port wine stain, is a capillary malformation that appears as a unilateral blanchable, red-purple erythematous patch. Present from birth and does not regress, does not grow in infancy. **Tx: pulsed dye laser.**

(Choice D) A nevus simplex (eg, macular stain, salmon patch, stork bite, angel kiss) is a blanching, pink patch on the eyelid, glabella, and nape of neck. It is present at birth and fades spontaneously by age 1-2, although **neck lesions may persist with no sequelae.**

Congenital melanocytic nevus (CMN): 1st few months of life, **hyperpigmented, solitary,** lesions with ↑ density of **overlying dark, coarse hairs**. Initially flat homogenous hyperpigmentation → grow during infancy → heterogeneously pigmented and raised.

The risk of transformation to melanoma increases (up to 5%) with increasing size of a CMN, and large lesions are often removed surgically to reduce risk. Small lesions may be removed for cosmetic reasons given their low risk of melanoma transformation.

Scald injuries account for the majority of burns in pediatric patients. Intentional: **stocking or glove burn distribution** with **sharp lines of demarcation, uniform depth, lack of splash marks, and spared flexural creases.**

Unintentional scald injuries are characterized by the presence of splash marks, nonuniform burn depth, and poorly defined and asymmetric wound margins. Most commonly following **hot liquid spill from an elevated surface** & results in facial, proximal upper extremity, and superior trunk burns.

Bedbugs are **small, red-brown, blood-feeding insects** that hide in clusters within the surrounding environment (folds of mattress, cushions) and feed at night. Transmission occurs within a physical living space or via colonized objects (eg, luggage); **person-to-person transmission is rare**.

Bites are typically **painless** and located on **exposed areas** (eg, neck, arms). The local inflammatory reaction causes **2- to 5-mm pruritic, erythematous papules**. A characteristic appearance of clustered, linear lesions is often described as a "breakfast, lunch, and dinner" pattern, presumably from bedbugs searching for superficial vessels to feed. Salivary proteins of the bedbug can inhibit coagulation; bites often present with a **central hemorrhagic punctum**. Household contacts with similar symptoms support the diagnosis, but confirmation requires **visualization of bedbugs**.

Common insect bites/infestation

Insect	Clinical features*
Bed bug 	Pruritic, small puncta & maculopapules in linear groups ("breakfast, lunch, dinner" pattern) on unclothed skin
Pediculosis 	Widespread itching of hair, body, or genitalia with visible louse
Scabies 	Pruritic burrows or hemorrhagic crusts in intertriginous areas
Spider 	Solitary papule, pustule, or wheal +/- pruritus
Tick 	Painless red papule +/- pruritus during the spring & summer

*Some patients have no symptoms

Tx: **supportive** (eg, **topical corticosteroids, oral antihistamines**), resolve within a week. However, bedbugs can live for up to a year without feeding. Therefore, **definitive treatment** requires **eradication** of bedbugs, such as **insecticides and high heat**. Contaminated **objects** can be **frozen or laundered on high heat** as well.

(Choice B) Chiggers are small mites that cause intensely pruritic, erythematous papules on exposed areas (eg, ankles, waistline). Found outdoors (eg, grassy fields).

Chapter 12: Neurology

NF1: Multiple, large (>0.5 cm) café au lait macules by age 1, skinfold freckling age 5, lisch nodules age 6. **Optic pathway glioma** (OPG) is the MC intracranial lesion and manifests during toddlerhood. Although many OPGs are asymptomatic, a growing tumor can cause **headache** and **decreased visual acuity**. Headache is often **worse in the morning** because intracranial pressure increases overnight while supine. Patients with NF1 are also at increased risk for **astrocytomas and brainstem gliomas**, even into adulthood.

Tuberous sclerosis complex	
Surveillance	• **Tumor screening**: Regular skin & eye examinations. Serial MRI of the brain & kidney. Baseline echocardiography & serial ECG • Baseline EEG. Neuropsychiatric screening (Learning disabilities, autism)

Subependymal giant cell tumor is the classic CNS lesion in TSC. This **slow-growing**, **periventricular** tumor is usually **asymptomatic** but may cause **obstructive hydrocephalus** (eg, headache, vomiting, focal neurologic deficits). Refractory epilepsy (eg, infantile spasms, tonic-clonic) is the most common presenting feature of TSC, with typical onset in infancy or early childhood.

Sturge-Weber syndrome (Mutation in *GNAQ* gene) [Dx: brain MRI]	
Clinical features	• Port-wine stain (trigeminal nerve [CN V1/V2] distribution) • Leptomeningeal capillary-venous malformation • Seizures (originate focally with subsequent generalization) ± hemiparesis, ID, VF defects, glaucoma (congenital anterior chamber **angle anomaly** and ↑ episcleral VP from episcleral hemangioma. Onset at birth but can occur into adulthood).
Management	• Laser therapy. Antiepileptic drugs. IOP reduction

VHL (AD) disease mx involves routine tumor surveillance. **Laser therapy** is the preferred treatment for **retinal hemangioblastomas** (decreased visual acuity due to edema and/or distortion of the retina. Bleeding from the tumor → RD, glaucoma, and **vision loss**).

Cerebral palsy (prematurity <37 wks is the greatest risk factor, Dx established by age 2)	
Etiology*	**Imaging findings**
Prematurity/LBW [MC]	• Periventricular leukomalacia [ischemia & necrosis]. IVH [friable germinal matrix vessels]
Hypoxic-ischemic injury	• Watershed injury. Basal ganglia/thalamic lesions
Perinatal stroke	• Vascular distribution
Intrauterine infection	• Calcifications (CMV, toxoplasmosis)

*Etiology is often multifactorial & the underlying cause may not be identified.

CP: UMN signs, **nonprogressive** motor dysfunction due to neurologic injury. Patients are at increased risk for foot deformities (eg, talipes equinovarus)

The following early signs should raise suspicion: Motor delay (not rolling over at age 4 months corrected). **Early hand preference** (age <1 year). Persistent neonatal reflexes (eg, tonic neck reflex at age >6 months). Abnormal tone.

Infants initially hypotonic → spasticity/hyperreflexia/clonus develop with time. Subtypes:

- **Spastic (MC): diplegia** (**MC**, "Commando crawling": pulling self with arms while dragging legs), hemiplegia (unilateral MRI findings, early hand preference), quadriplegia, motor delay, hypertonia, contractures.
- **Dyskinetic** (eg, choreoathetotic, dystonic): involuntary movements, dysarthria
- **Ataxic**: hypotonia, incoordination, jerky speech

Intraventricular hemorrhage	
Pathophysiology	• Rupture of fragile germinal matrix vessels
Risk factors	• Prematurity. Very low birth weight
Clinical findings	• 1st few days of life: Often asymptomatic (MC). Seizures or apnea, hypotonia, decreased movements. Full fontanel. Rapid ↑ in HC. Acute anemia, tachycardia. Long term sequelae: CP, ID
Diagnosis	• Cranial ultrasonography*: Hyperechoic material (blood) within germinal matrix ± extension to ventricles or parenchyma
*Performed if symptomatic or as routine screening if <32 weeks gestational age.	

Subgaleal hemorrhage: shearing of veins between the dural sinuses and scalp due to scalp traction during delivery. Blood accumulates between the periosteum and galea aponeurotica, causing **diffuse, fluctuant** scalp swelling that shifts with movement and **expands over 2-3 days**. Rapid hemorrhage expansion -> hypovolemic shock, DIC, and death. Pts have signs of anemia unlike **cephalohematoma** (**firm, nonfluctuant** swelling, slow bleeding -> hours after birth, reassurance – reabsorb spontaneously in a month)

Neonatal scalp swelling			
	Caput succedaneum	**Cephalohematoma**	**Subgaleal hemorrhage**

Location	• Subcutaneous. Involves portion of the head presenting during vertex delivery	• Subperiosteal (between skull & periosteum)	• Between periosteum & gala aponeurotica
Clinical features	• Present at birth • Soft, boggy • Crosses sutures • Overlying skin is normal	• Present hours after birth • Firm, nonfluctuant • Does not cross sutures • Overlying skin is normal	• Can expand over days • Soft, fluctuant • Diffuse, crosses sutures • ± Overlying bruising
Prognosis	• Self-resolves in days	• ↑ Hyperbilirubinemia risk • Resorbs within a month	• Can cause life-threatening blood loss

Lesch-Nyhan syndrome (XLR, HGPRT deficiency, ↑ hypoxanthine and uric acid)	
Clinical features	• Delayed milestones & hypotonia in infancy ○ Early childhood: ID. Age 3 -> Extrapyramidal symptoms (eg, dystonia, chorea) + Pyramidal symptoms (eg, spasticity, hyperreflexia). Self-mutilation. Late: Gout

The development of **dopaminergic** pathways appears to be particularly affected, which may explain the progressive neuropsychiatric manifestations of the condition.

Friedreich ataxia (FA): AR, most common hereditary form of ataxia: dysarthria, limb weakness, loss of DTR, progressive gait and limb ataxia in **adolescence**. **Loss of position and vibratory senses**. Death from HCM, imaging may reveal cervical spinal cord atrophy. **Dx**: **genetic testing**. **Management** is **supportive** (eg, physical therapy, psychological support), as **no disease-modifying therapies are available**. **Prenatal genetic testing** and counselling are available for future pregnancies for confirmed cases in siblings or if both parents are known carriers. NO increased risk for cancer (vs ataxia-telangiectasia). Mean age of death is 30-40 [survival rate rarely exceeds 20 years after disease onset].

Muscular dystrophies

Diagnosis	Duchenne	Becker	Classic myotonic
Genetics	X-linked recessive Deletion of dystrophin gene		Autosomal dominant Trinucleotide repeat in *DMPK* gene
Onset	Age 2-3	Age 5-15	Age 12-30*
Clinical presentation	Proximal muscle weakness (eg, Gowers sign) Calf pseudohypertrophy	Milder proximal weakness relative to Duchenne	Facial & distal muscle weakness Grip (hand) myotonia
Associated findings	Cardiomyopathy, arrhythmias Scoliosis	Cardiomyopathy, arrhythmias	Cardiomyopathy, arrhythmias Dysphagia Cataracts Testicular atrophy
Prognosis	Wheelchair-dependent by adolescence Death at age 20-30 from respiratory or heart failure	Death at age 40-50 from heart failure	Death at age 45-55 from respiratory or heart failure*

*Severe forms can present in infancy & early childhood & are associated with earlier death.

Duchenne muscular dystrophy (X-linked recessive. Tx: Glucocorticoids)	
Clinical presentation	• Onset age 2-3. Delayed walking, toe walking (due to tight heel cords), waddling gait, ↓ endurance. Proximal muscle weakness (Gower sign, calf pseudohypertrophy). Dilated cardiomyopathy. Scoliosis
Diagnosis	• **↑ Serum CK** [screening tool, drops later as more muscle is replaced by fat & fibrosis]. **Genetic testing**: dystrophin deletion, if inconclusive, do **muscle biopsy**: fibrosis, fat, muscle degeneration

Patients with DMD usually do not **walk** independently until age **>18 months** and often have difficulty running/jumping/climbing stairs, frequent falls, fatigue with activity.

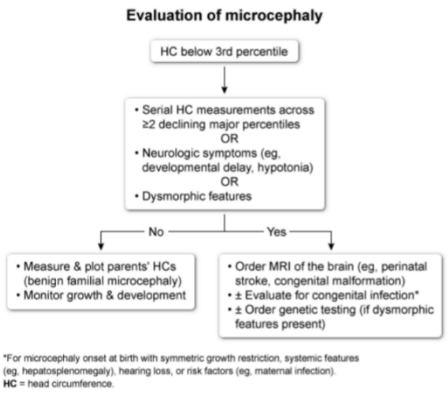
Myotonic dystrophy (AD CTG trinucleotide repeat in DMPK gene)	
Presentation	• **Classic** (adult): myotonia & weakness (eg, face, hands, ankles) • **Childhood** {by age 10}: cognitive [intellectual impairment] & behavioral [ADHD, mood disorder] difficulties (classic symptoms develop over time) • **Infantile**: hypotonia, respiratory failure, inverted V–shaped upper lip
Associated findings	• Arrhythmias, cardiomyopathy, dysphagia, constipation, pharyngeal weakness, hypoventilation, insulin resistance, hypogonadism, cataracts, frontal balding, excessive daytime sleepiness

Dx & Tx	• Genetic testing. Supportive management

+ Muscle **atrophy** with ptosis, **temporal wasting**, emaciated extremities/sustained thumb abduction after thenar eminence percussion [percussion myotonia]).

(Choice A) Brain imaging should be considered in patients **global developmental delay**.

The first step in evaluation of microcephaly is a thorough physical examination. **Benign familial microcephaly**: Patients are monitored clinically, and no further work-up is required in the absence of pathologic signs.



Chiari I malformation: the most common and mildest subtype. **Asymptomatic** through childhood. May be diagnosed incidentally on imaging or present in **adolescence** or early adulthood with **occipital headache** and/or neck pain. Dizziness and worsening pain with **physical activity** or **Valsalva** maneuvers (eg, cough, straining) due to pressure of the cerebellar tonsils on the foramen magnum. And ↑ ICP, cranial neuropathies from brainstem compression (dysarthria), cerebellar dysfunction (ataxia).

Fragile X syndrome	
Pathogenesis	• Trinucleotide (CGG) repeat expansion of *FMR1* • Gene methylation prevents transcription & protein production • X-linked dominant inheritance
Features	• Macroorchidism (age>8), long face with large jaw/macrocephaly, large everted ears, autism/ADHD/anxiety/ID/Developmental delay, MVP, hypermobile joints, self-mutilation
Diagnosis	• *FMR1* DNA analysis (PCR or Southern blot)

FMR = fragile X **messenger ribonucleoprotein**. Normal life expectancy

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Risk factors for sudden infant death syndrome	
Maternal/antenatal	• Substance use (cigarettes, alcohol, recreational drugs). Maternal age <20. Inconsistent prenatal care
Infant	• Prematurity or low birth weight. Smoke exposure

	• Sleep environment: Prone-/side-sleep position. Soft sleep surface, loose bedding. Bed sharing

Sudden infant death syndrome (SIDS) is the leading cause of mortality in infants age 1 month to 1 year in the United States. It is defined as a **sudden, unexpected** death that **cannot be explained by history or postmortem examination**. Pathogenesis is uncertain but may involve **delayed brain maturation** resulting in inadequate CNS arousal responses to hypoxia and hypercarbia in the setting of an inciting event (airway obstruction).

Although prone positioning (tummy time) is recommended for gross motor development and positional plagiocephaly prevention, it should occur only **during awake, observed periods**. **Side-sleep** positioning is a less significant risk factor but should also be avoided due to an increased ability to roll into the prone position. Instead, infants should be placed to **sleep** in a **supine position** **on a firm, flat surface with no other objects (pillow, stuffed animals) in the crib/bassinet.**

- **Pacifier** use during sleep is associated with a **↓ risk of SIDS**. (**increased arousability**)
- **Periodic breathing**: a normal newborn respiratory pattern has **brief (5-10 sec) pauses.**
- **Cranial molding helmets** for **positional** plagiocephaly (abnormal skull flattening), which is caused by **supine positioning of young infants** (not newborns). **Congenital** plagiocephaly may be due to abnormal closure of sutures (ie, craniosynostosis).

Infants develop **object permanence** by **age 9 months** and realize when specific people leave their presence (seperation anxiety) or objects are taken away (can find a toy covered by a hand). **Language** development begins in infancy with cooing and babbling, and most children say their first word (eg, "mama," "dada") by age 12 months. By **age 2**, children should be able to form **2-word phrases**, and by age 2.5, they should say ≥50 words (although many can say over 200). In addition, a stranger should be able to **understand half of the child's speech** at age 2. Children age 2-3 are also in the process of learning pronouns and will frequently misuse "me" instead of "I."

By age 2, children can also run, throw a ball overhand, kick a ball, sort and match objects & understand the use of familiar objects (imitate truck noises while playing). Children at this age often show defiance as they become more independent and confident with new skills. Two-year-old children commonly engage in **parallel play** (play alongside sibling) rather than **cooperative play**, in which children play together (typically age ≥3).

Speech delay and language disorders are the **most common** developmental disabilities in children. Patients with **language disorders** have difficulty **understanding** others (eg, receptive language) and/or **communicating** thoughts (eg, expressive language). In

contrast, **isolated speech delay** is characterized by problems with: **articulation** (eg, mispronunciation), **fluency** (eg, stuttering), **vocal quality** (eg, abnormal volume or pitch)

The most common cause of hearing impairment in young children is **conductive hearing loss from recurrent acute otitis media**. The first step in evaluation of **all patients with a speech or language disorder**, even without a history of frequent ear infections or abnormalities on otoscopy, is a **hearing test**. Speech 100% intelligible by age 4

Fetal alcohol syndrome is characterized by ≥2 of the following pathognomonic facial dysmorphisms: **Smooth philtrum. Thin vermilion border. Small palpebral fissures.**

Midface hypoplasia, microcephaly, **poor growth, developmental delay**, **ID**, cognitive impairment (memory issues, learning disabilities) poor adaptive functioning.
ADHD (**Hyperactivity**, inattention), **poor social skills** (eg, decreased eye contact, disinterest in peer play), and seizures are also common. **Height, weight & head circumference <10th percentile.**

Women who are pregnant or trying to conceive are advised to **abstain from alcohol completely** because there is **no known safe amount of prenatal alcohol consumption**. Early diagnosis is critical so that affected children can **maximize benefit from speech, physical, and occupational therapies.**

Glaucoma in children	
Pathophysiology	• Optic neuropathy ± ↑ IOP • Causes: Impaired drainage of intraocular fluid (MC). Primary anatomic abnormality (angle dysgenesis). Sturge-Weber syndrome. Tumor, trauma, infection of the angle. Corticosteroids
Key features	• Tearing, photophobia, blepharospasm. Conjunctival erythema. Enlarged cloudy cornea and/or globe. Optic nerve cupping. **Tonometry**: ↑ IOP
Management	• Surgery ± Pressure-reducing eye drops

Rett syndrome (Rare, greater incidence in girls, onset 6-18 mo)	
Key features	• Initially, normal development followed by regression in motor verbal cognitive abilities, ataxia seizures scoliosis stereotypic hand-wringing (rocking back & forth) + loss of **purposeful hand movements** (eg, twisting of fingers). Microcephaly, breathing

	abnormalities, sleep disturbance, autistic features. MECP2 gene mutation on chromosome X causing deceleration of brain growth.
Prognosis	• Middle-aged life expectancy; reduced mobility, seizures, respiratory difficulties

Alternating episodes of hyperventilation and hypoventilation tend to occur during periods of heightened emotion and are often associated with stereotypical movements.

Differential diagnosis of Marfanoid body habitus		
Diagnosis	**Overlapping features**	**Distinguishing features**
Marfan syndrome	• Pectus deformity • Tall stature; ↑ Arm: height ratio. ↓ Upper: lower segment ratio • Arachnodactyly. Joint hyperlaxity. Skin hyperelasticity • Scoliosis. Crowded teeth, high arch palate, iridodonesis (rapid contraction & dilation of the iris), myopia (from elongation of the globe)	• Autosomal dominant • Normal intellect • Aortic root dilation • Upward lens dislocation
Homocystinuria **Tx: B6 9 12, antiplatelets /anticoagulants**		• AR • ID/ Developmental delay • Thrombosis/ CVA • Downward lens dislocation • Megaloblastic anemia. Fair complexion (fair hair & eyes)

(Choice D) Congenital contractural arachnodactyly AD mutation of **fibrillin-2** gene (vs **marfan fibrillin-1**). These patients have tall stature, arachnodactyly, and **multiple contractures** involving large joints. Ocular and cardiovascular symptoms are not present.

Hydrocephalus in children	
Causes	• Impaired CSF circulation (obstructive/noncommunicating MC)/ absorption. Excessive CSF production
Clinical manifestations	• Rapidly enlarging HC (cross 2 percentiles) &/or macrocephaly. Prominent scalp veins. Full anterior fontanelle (if open) • Behavioral changes &/or developmental delay (axonal damage)

	• Lower extremity weakness & spasticity • Signs of increased ICP*: irritability, headache, vomiting. Papilledema, impaired upward gaze (pressure on the midbrain, poor eye contact). Hypertension, bradycardia
Evaluation	• Neuroimaging: US (if fontanelle is open) or ultrafast MRI. CT scan for signs of acutely increased ICP
*May not be present if the anterior fontanelle is open. (1st 12-15 months)	

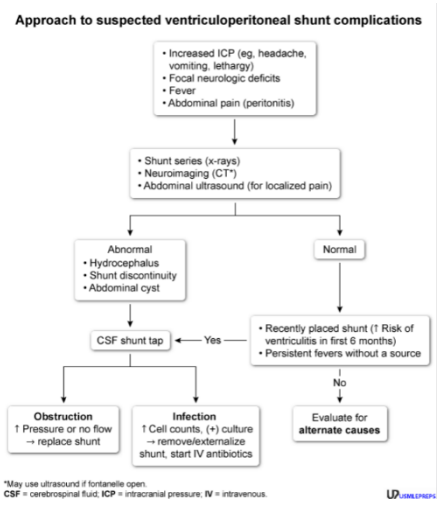
Lead toxicity: peripheral neuropathy, irritability, hyperactivity, fatigue, abd pain. [infants]

MCC of macrocephaly is **benign familial macrocephaly** due to megaloencephaly (increased brain parenchyma volume), a condition characterized by a large head in **both** an otherwise **normal patient and a first-degree relative**. Normal US. Reassurance

Examination findings for **benign macrocephaly** include: Normal development. No syndromic features. No signs of increased ICP (bulging fontanelle). No fever, lethargy

(Choice D) MRI of the brain is performed in patients with macrocephaly **and** either an **abnormal neurologic examination** or **abnormal findings on head ultrasound**.

IVH can result in permanent obstruction of cerebrospinal fluid (CSF) flow, resulting in persistent hydrocephalus. In these patients, a ventriculoperitoneal (VP) shunt is placed as a long-term solution to drain excess CSF into the abdomen. Although it is a life-saving treatment, **shunt malfunction** (eg, catheter obstruction, shunt migration, split tubing) or infection → recurrence of hydrocephalus. Shunt complications occur at any time after placement but usually in the **first 6 months** (and most commonly within the first month).



(Choice D) Cerebral venous sinus thrombosis can increase ICP by blocking CSF absorption by arachnoid granulations.

Myelomeningocele (Failure of neural tube closure at week 4 of development)	
Clinical features	• Membranous sac over lumbosacral region. Severe neurologic deficits distal to lesion (paralysis, bowel/bladder incontinence)

Associated abnormalities	• **Chiari II malformation**: Inferior displacement of medulla & cerebellum through foramen magnum. **Obstructive hydrocephalus**

(Choice A) Arteriovenous malformation at the vein of Galen is a rare, **isolated** condition that most commonly causes neonatal heart failure due to high-pressure venous blood flow.

(Choice B) Choroid plexus papilloma: ↑ CSF production. Resection if large/symptomatic.

Diffuse leptomeningeal enhancement: meningitis due to ↑ permeability of the meninges.

Vitamin K deficiency -> intracranial hemorrhage -> **obstructive hydrocephalus**.

Classic VKDB presents in the **first week of life** with easy bruising and mucosal and gastrointestinal bleeding. Late-onset VKDB occurs **between age 2 weeks and 6 months** and is more commonly associated with **intracranial hemorrhage** (layered, hyperdense fluid collection in the posterior ventricles on imaging due to supine positioning).

(Choice A) Congenital atresia of the foramina of Luschka and Magendie prevents CSF outflow into the subarachnoid space, causing **hydrocephalus**. This congenital anomaly is associated with **Dandy-Walker syndrome**.

(Choice C) Generalized cerebral edema can occur with **untreated, worsening hydrocephalus** because increased hydrostatic pressure allows CSF to flow to the parenchyma. Imaging would show **flattened gyri and sulci**.

(Choice E) In children, **nontraumatic** intraparenchymal hemorrhage is typically caused by a **ruptured arteriovenous malformation** and presents acutely with **focal neurologic deficits and signs of increased intracranial pressure.**

Focal seizure	
Onset	• Neuronal discharge begins in **1 cerebral hemisphere.** Symptoms may be motor, sensory, or autonomic. Underlying structural abnormality (eg, tumor) more likely than with a generalized seizure
Categories	• No impairment of awareness (**remains localized** to 1 hemisphere) • Impairment of awareness (**spreads to the other hemisphere**). Often associated with **automatisms** (Repetitive semipurposeful movements such as **chewing, picking**)
Diagnosis	• EEG. Brain MRI (may identify a triggering, structural lesion)

Due to the impact on cognition, school-aged children with uncontrolled seizures or subtle seizure manifestations may also have a history of **decline in academic performance**.

During a focal seizure, electroencephalogram (EEG) reveals abnormal electrical activity arising from a specific region in the brain, whereas **interictal** EEG is often normal.

(Choice A) Absence seizures are **generalized** (ie, originating from both cerebral hemispheres) and present with staring spells, with or without automatisms, lasting only 10-20 seconds. Easily provoked by hyperventilation and not associated with a postictal period.

(Choice C) Juvenile myoclonic epilepsy: adolescents with **myoclonic jerks immediately on wakening**. Absence and generalized tonic-clonic seizures may also be seen.

(Choice D) Lennox-Gastaut syndrome typically presents by **age 5 with** intellectual disability and severe seizures of varying types (eg, atypical absence, tonic). **Interictal** EEG demonstrates a **slow spike-and-wave pattern**.

(Choice E) Tic disorders present as sudden, brief movements (eg, grimacing) or vocalizations. **Consciousness is not impaired** by tic disorders.

- **Status epilepticus**: mortality from cerebral anoxia, aspiration, respiratory failure.
- **VSD**: **LOC and pt goes limp + no postictal confusion**.

	Seizure	Vasovagal syncope
Triggers	Lack of sleep Flashing light Emotional stress Alcohol withdrawal Idiopathic	Prolonged standing Physical/emotional stress Heat
Clinical clues	Preceding aura (eg, olfactory hallucinations) Can occur with sleeping/sitting Tonic/clonic movements Rapid, strong pulses Tongue biting Incontinence Perioral Cyanosis Open eyes	Preceding lightheadedness (ie, presyncope) Unlikely to occur while sleeping/sitting Uncommon to have clonic jerks (can occur with prolonged cerebral hypoperfusion) Weak, slow pulses Pallor & diaphoresis
Sequelae	Delayed return to baseline (postictal drowsiness or confusion)	Immediate return to baseline

Although the **incidence** of epilepsy is highest in early childhood and late adulthood, a **first-time seizure** can occur at any age and time, including during sleep. **Tonic clonic movements <3 mins. Cardiogenic syncope:** triggered by exertion/dehydration, sudden

LOC without prodrome, immediate return to baseline, hx of HCM or arrhythmia (pre-excitation syndrome).

Acute causes of hemiplegia in children		
Cause	**Historical clues**	**Key findings**
Ischemic stroke	• Prothrombotic disorder (eg, ATIII deficiency) • Cardiac disease (PFO). Vasculopathy (SSD)	• Focal infarct on brain imaging
Intracranial hemorrhage	• Recent trauma. Bleeding disorder (hemophilia). ↑ ICP (vomiting, bradycardia)	• Hemorrhage on brain imaging
Seizure (Todd paralysis)	• Symptoms preceded by limb jerking or LOC + postictal confusion	• Symptoms self-resolve
Hemiplegic migraine	• Adolescence, (+) FHx, migraine with aura. No LOC	• Symptoms self-resolve

Todd paralysis: Focal **flaccid** weakness or paralysis of 1 or both extremities **on the same side of the body** that occurs in the **postictal** period after a **focal-onset seizure**, which may become secondarily generalized. The pathophysiology likely involves neuronal exhaustion. Clinical Dx. Normal imaging. Supportive tx, self limiting within 36 hrs.

(Choice B) Conversion disorder is uncommon before adolescence and can present with a range of neurologic deficits (eg, nonepileptic seizure, weakness/paralysis, visual changes) without an underlying physiologic abnormality.

Syncope occurs due to global cerebral hypoperfusion. In contrast, **psychogenic pseudosyncope** (PPS), a type of conversion **disorder** is an **apparent, transient LOC without impaired cerebral perfusion**. Features:

- **Prolonged LOC** (eg, 20 min): PPS episodes typically last many minutes to hours versus approximately 1-2 minutes in syncope.
- **Normal vital signs, no pallor/sweating.**
- Patient's **reports of symptoms**/events that **occurred during the episode** ("I felt my head throbbing after it hit the floor"): This awareness rules out true LOC. Symptoms are often reported in a detached or disassociated manner (la belle indifférence).

- Witness present, high frequency of episodes (>1 episode/day, >50 episodes/year), history of underlying psychiatric disorder

(Choice E) Serum prolactin levels, which are usually elevated immediately following seizure, are used primarily to differentiate seizure from PPS **in adults**.

(Choice A) Cataplexy (daytime sleepiness) + congenital long QT syndrome (abnormal ECG) + vasovagal syncope (nausea prodrome) are brief (<2 mins).

Breath-holding spell (BHS) vs seizure			
	Cyanotic BHS	**Pallid BHS**	**Seizure**
Epidemiology	• Age 6 months to 2 years • ↑ Risk with IDA, FHx		• Any age. ↑ Risk with hx of febrile seizure or developmental delay
Triggers	• Crying/frustration	• Minor trauma • Pain or fear	• Often unprovoked • Sleep deprivation
Clinical features	• Apnea & cyanosis → LOC • Rapid return to baseline	• Bradycardia, apnea & pallor → LOC • ± Brief (<5 min) confusion or sleepiness	• LOC → tonic-clonic movements • Prolonged (>5 min) postictal confusion

Pallid BHS (less common): **Pain** or **fear** causes the heart rate to slow and the child to hold the breath and become **pale, diaphoretic**, and limp. NO crying

LOC during a BHS is typically <1 minute; **diagnosis** is **clinical** and requires **no further testing** when presentation is classic. However, screening for associated IDA (CBC, ferritin) is recommended because IDA patients improve with iron therapy. Tx: reassurance, iron supplementation if deficient, resolution by age 5 with no sequelae.

(Choice A) Patients with atypical BHS (eg, multiple daily episodes, duration >1 min) or a family history of cardiac disease or sudden death should undergo an **ECG** to evaluate for arrhythmias (eg, prolonged QT syndrome).

(Choice C) TOF cyanosis differentiated from cyanotic BHS by **rapid and deep breathing** (vs breath-holding).

Febrile seizure	
Risk factors	• Fever ≥38 C (typically with viral illness). Family history of seizures
Diagnostic criteria	• Age 6 months to 5 years. No previous afebrile seizure. No signs of CNS infection. No acute metabolic cause (eg, hypoglycemia)
Management	• Abortive therapy (≥5 min), antipyretics, reassurance
Prognosis	• No long-term sequelae. Recurrence (30%). ↑ risk of epilepsy (~1%)

Risk factors include high fever (viral illness), recent immunization, FHx of febrile seizures.

Simple febrile seizures are characterized by **generalized** tonic-clonic movements lasting **<15 minutes & do not recur within 24 hours**. Postictal drowsiness but **return to baseline** within mins. Neurologic examination is normal. Diagnosis is clinical, and **no further evaluation** is required. Antipyretics do not reduce the risk of future febrile seizures.

- Do **brain imaging** for patients with febrile seizure who have **focal neurologic signs**.

(Choice C) Hospitalization and observation are required in a **patient who has not returned to neurologic baseline following a seizure.**

Acute ischemic stroke in **young children** (eg, age <6): **headache** (25%-50%), generalized or focal **seizures** (15%-25%), AMS (**lethargy**), focal neurologic deficits. Next step: **MRA** (more sensitive than CT). Obtaining an MRI/MRA is particularly important in pediatric patients because stroke mimics (eg, Todd paralysis, complicated migraine) are common and the **etiology of stroke** is more varied than in adults.

Risk factors for pediatric ischemic stroke	
Cardiac	• Congenital heart disease (eg, causing paradoxical embolism, thrombus). Bacterial endocarditis
Vascular	• Arterial dissection (eg, head/neck trauma). Vasculitis (eg, Takayasu arteritis, SLE). Infection (eg, bacterial meningitis)
Hematologic	• SSD. Hypercoagulable states (eg, protein C or S deficiency)

(Choice D) Todd paralysis, which requires no additional testing, can manifest as transient focal deficits (eg, aphasia, unilateral weakness) following a seizure. However, it is a diagnosis of exclusion that requires first ruling out acute ischemic stroke (with MRI).

(Choice C) Isolated pontine infarction can cause locked-in syndrome in which patients have complete quadriplegia and cannot speak; however, they typically retain consciousness, eye opening, and vertical eye movements.

Hemorrhagic stroke in children	
Risk factors	• Vascular malformations: AVM (isolated or associated with HHT). Aneurysm. Hematologic abnormalities (eg, hemophilia, SSD)
Features	• Headache, vomiting. Seizure. Focal neurologic deficits. AMS. HTN
Imaging	• Head CT scan: intraparenchymal, intraventricular, or subarachnoid hyperdense fluid collection
Management	• Supportive (eg, antiepileptics) • Reduction of ICP (eg, elevate head of bed, surgical decompression)

Concussion	
Clinical features	• Headache, dizziness, disorientation, anterograde/retrograde amnesia, difficulty concentrating after mild TBI. Transient abnormalities in coordination (eg, stumbling, falls), speech (eg, slurring), attention (eg, poor focus), or emotion (eg, lability), vomiting and LOC. No structural intracranial injury.
Management	• Remove from same-day physical play. Neurologic evaluation. **Rest for ≥24 hr**. Symptomatic management: NSAIDS, ondansetron. **Gradual** return to normal activity if symptoms do not worsen: ○ **Physical**: light aerobic exercise → noncontact sports → contact sports ○ **Neurocognitive**: limited screen time, school accommodations (eg, frequent breaks, shortened days)

Concussions are common in **children and adolescents** who participate in contact sports (eg, **hockey, football**). Pathogenesis involves **axonal shearing from rotational acceleration** of the brain after a fall or strike to the head.

Head trauma (typically due to blunt force) → widespread neuron depolarization → ↓ CBF → localized lactic acidosis → **transient disturbance of normal neuronal function**.

The predominant finding in patients with concussions is **headache**, shortly after the head trauma. Triggered by physical or cognitive exertion, visual tasks (eg, rapid eye movement between two points), light and/or **noise**.

Neuroimaging is **not required** for diagnosis but may be performed to exclude structural intracranial injury (**contusion, hematoma**) in pts with high-risk features **(vomiting, LOC)**

Physical and cognitive rest for **24-48 hours**. THEN **gradual return-to-play** protocol (over a minimum of 5 days) that involves **increasing intensity** of physical activity (light exercise → noncontact sport → competitive play). Patients should remain **symptom-free** before progressing to the next level. Most patients resume normal activities within a month.

Postconcussion syndrome	
Risk factors	• Nonadherence to gradual return-to-play protocol after concussion • History of multiple concussions
Clinical features	• Prolonged (>4 weeks) concussion symptoms after mild TBI: Headache (MC symptom). Dizziness. Sleep disturbance. Mood changes. Cognitive impairment. No structural intracranial injury
Management	• Symptomatic care (improves within 3 mo): amitriptyline (headache), CBT (insomnia/anxiety), **reassurance alone (if symptom severity is mild)**. Activity as tolerated

(Choice B) Most **posttraumatic seizures** occur **within 24 hours of injury** and are **generalized tonic-clonic movements**.

Recurrent head injury from any contact sport **during the initial recovery period** can lead to **second impact syndrome**: **cerebral edema**, **can be fatal**.

(Choice D) Perilymphatic fistula: rare complication of head trauma, fluid leakage from the cochlea and semicircular canals, hearing loss and vertigo triggered by the Valsalva maneuver; riding in an elevator; or sudden, loud sounds.

Epidural hematoma	
Pathogenesis	• Trauma to sphenoid bone with tearing of MMA

Clinical features	• Brief LOC -> **lucid interval** (mins-hrs) -> rapid hematoma expansion: ↑ ICP (impaired consciousness, headache, nausea/vomiting) + **Uncal herniation** (ipsilateral pupillary dilation & contralateral hemiparesis [due to compression of the ipsilateral cerebral peduncle, may later cause compression of the contralateral cerebral peduncle -> ipsilateral hemiparesis])
Diagnosis	• CT scan of the head: **biconvex** (lens-shaped) hyperdensity that does not cross suture lines
Treatment	• Urgent surgical evacuation for symptomatic patients

In **adolescents and adults**, EDH most commonly occurs following **high-impact head trauma** (eg, motor vehicle collision) vs **low-velocity falls** in **young children**.

(Choice A) Cerebral contusion: microhemorrhages ("bruising") within the superficial brain parenchyma due to coup-contrecoup injury. Symptoms of increased ICP may develop (over hrs-days, vs EDH within mins) if significant cerebral edema occurs.

Pediatric traumatic brain injury (PECARN rule)	
High-risk features age <2	• AMS (fussy behavior). LOC. Severe mechanism of injury (fall >0.9m, high impact, MVC). Nonfrontal scalp hematoma. Palpable skull fx
High-risk features age ≥2-18	• AMS (somnolence, agitation). LOC. Severe mechanism of injury (fall >1.5 m [5 ft], high impact, MVC). Vomiting, severe headache. Basilar skull fracture signs (CSF rhinorrhea)
Management	• Head CT scan without contrast

Observation for 4-6 hours in the ER may be an option for patients with isolated or mild/improved high-risk features (brief LOC, resolved vomiting) but normal mental status and no signs of a basilar skull fracture. Head CT if symptoms worsen.

Patients with **no high-risk features**, **imaging** should be **avoided** , and management should include **reassurance and discharge home** with education regarding symptoms (eg, severe headache) that would require reevaluation.

(Choice E) Urgent neurosurgical evaluation: if an **intracranial bleed** is detected on **neuroimaging** or in an **unstable**, **altered patient with head trauma**.

Traumatic carotid injuries (dissection) (Dx: CT scan or MRA)

Mechanism	• Penetrating trauma. Fall with object in mouth (eg, toothbrush, pencil). Neck manipulation (eg, yoga, sports)
Presentation	• Gradual hemiplegia. Aphasia. Neck pain. "Thunderclap" headache

Migraine (Headache may be bifrontal in children)	
Pathogenesis	• Abnormal neuronal activation of trigeminal ganglion afferents. ↑ (CGRP), a neuropeptide involved in pain signaling. • Triggers: sleep deprivation, stress, menses, fasting
Epidemiology	• M=F until puberty. ↑ Risk in adolescent girls & women
Clinical features	• Pulsatile/throbbing, unilateral headache lasting hours to 3 days • N V Photophobia & phonophobia ± Preceding aura (eg, scintillating scotoma). Normal neurologic examination. Autonomic symptoms: facial sweating, tearing, nasal congestion (children ONLY)
Treatment	• Acetaminophen, NSAIDS, oral/intranasal triptans (refractory), antiemetics (promethazine/ prochlorperazine), dihydroergotamine

Diagnosis is **clinical**. **Neuroimaging** (eg, CT scan of the head, brain MRI) is indicated for **features suggesting an alternate, life-threatening diagnosis, including**: Abnormal neurologic examination (eg, weakness, ataxia). Signs of increased ICP (eg, headache waking patient from sleep, papilledema). Rapidly increasing headache frequency.

Funduscopic examination may elicit pain due to exposure to bright light.

Idiopathic intracranial hypertension	
Risk factors	• Obese women of childbearing age. VitA, tetracycline, GH
Pathophysiology	• Impaired CSF resorption & intracranial venous hypertension
Clinical features	• Throbbing headache (worse when flat/morning, improves when standing) nausea/vomiting • Visual changes (transient obscurations; vision loss; blurred vision; diplopia due to [CN VI] palsy). Pulsatile tinnitus (ie, "whooshing" sound) • Retrobulbar pain/neck pain/back pain

Diagnosis	• Papilledema & enlarged blind spots. MRI to rule out mass lesions/ hydrocephalus. MRV to rule out venous thrombosis. LP: ↑ opening pressure (>250 mm H_2O)
Treatment	• Weight loss, including bariatric surgery • Carbonic anhydrase inhibitor (acetazolamide), topiramate

In the prepubertal population, headache may be a less obvious finding; vision abnormalities may be predominant.

Medulloblastoma (MC **malignant** pediatric brain tumor). Leptomeningeal spread	
	• Posterior fossa tumor (from cerebellum, compresses 4th ventricle)

Clinical features	• Cerebellar dysfunction
	  ○ Truncal/gait ataxia (cerebellar vermis)
	  ○ Dysmetria, intention tremor (cerebellar hemispheres)
	• Obstructive hydrocephalus (↑ ICP): Headache vomiting papilledema abducens N palsy

Treatment	• Resection, craniospinal radiation, chemotherapy

Neuroblastoma (neural crest origin, involves adrenal medulla/sympathetic chain)	
Clinical features	• MC childhood extracranial solid tumor. Median age <2. Abd mass. Periorbital ecchymoses (orbital metastases). Spinal cord compression from epidural invasion ("dumbbell tumor"). Opsoclonus-myoclonus syndrome. Horner syndrome
Diagnostic findings	• Elevated catecholamine metabolites. Definitive (Biopsy): Small, round blue cells on histology. *N-myc* gene amplification

Tumor involvement of the **cervical paravertebral sympathetic chain** can lead to ipsilateral Horner syndrome (ie, ptosis, miosis, anhidrosis). Although sweating is often difficult to appreciate in young children, absent facial flushing (ie, **"harlequin" sign**) can be observed in anhidrotic areas.

(Choice C) Glioblastoma is an **aggressive**, **supratentorial** malignant tumor that usually presents with **headache, seizures, and focal neurologic deficits** (eg, motor weakness).

Pinealoma: papilledema headache vomiting ataxia obstructive hydrocephalus (blockage of CSF flow in the aqueduct of Sylvius). Parinaud syndrome (from pressure on the pretectal region of the midbrain near the superior colliculus and the oculomotor nerve (CN III)): limitation of upward gaze, convergence retraction nystagmus, light-near dissociation (pupils reactive to accommodation but not to light), and bilateral upper eyelid retraction (collier sign). Children age 1-12

Central nervous system (CNS) tumors are the second most common pediatric malignancies after leukemia and the most common pediatric solid tumors. Tumors originating in the **cerebral cortex** (ie, supratentorial) classically present with headache, seizures, and focal neurologic deficits. Changes in speech, memory, and personality (eg, irritability, declining academic performance) can also occur with supratentorial masses, and **hemiparesis** and **hyperreflexia** are common with **parietal lobe** involvement.

The **most common** brain tumor in **children** is **low-grade astrocytoma**. Astrocytomas are derived from astrocytes (the dominant glial cell in the brain) and can be low or high grade. Low-grade astrocytomas (eg, pilocytic astrocytoma, diffuse astrocytoma) are typically slow-growing and benign, causing symptoms to develop over many months.

vs High-grade astrocytomas (glioblastoma), malignant, much less common, symptoms over days to weeks.

(Choice B) MC intracranial site for ependymoma is 4th ventricle.

(Choice E) Retinoblastoma is an intraocular tumor in children age <5. A trilateral retinoblastoma consists of bilateral retinoblastoma and a pineal gland tumor.

Closed spinal dysraphism (spina bifida occulta)	
Pathophysiology	• Failure of **posterior vertebral arch** fusion at L5-S1 • ± Spinal cord anomalies (eg, **lipoma, cyst**) • Stretch-induced distal spinal cord dysfunction (**tethered cord**)
Clinical features	• May be asymptomatic. Cutaneous, lumbosacral anomalies (hemangioma, pit, nevus, hair tuft) • Tethered cord syndrome (tight filum terminale): ○ Neurologic: Abnormal gait. LMN signs (weakness, hypotonia, hyporeflexia, loss of sensation)

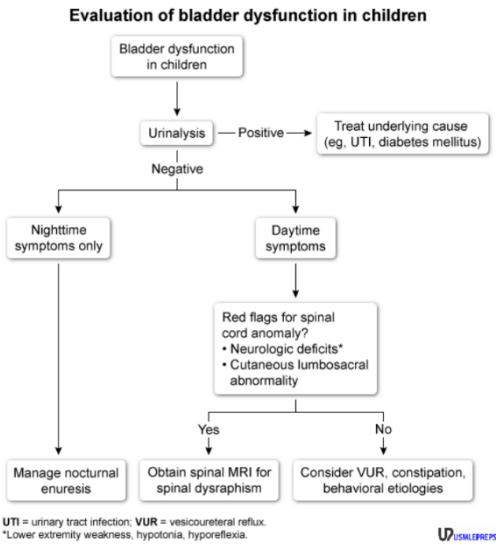
	○ Urologic: incontinence/retention; recurrent UTI + chronic constipation ○ Orthopedic: back pain [exacerbated by bending forward, can radiate to buttocks], scoliosis, foot deformities. Neuromuscular (secondary) hip dysplasia [in nonambulatory patients]
Management	• MRI of the spine. Surgical detethering of cord if symptomatic

The presence of upper motor neuron (UMN) signs (eg, hypertonia, hyperreflexia) argues against the diagnosis of tethered cord syndrome.

Patients may be asymptomatic for years before the insidious onset of symptoms. Age of onset & organ system involvement are variable.

The spinal cord can abnormally attach to surrounding tissues and undergo traction (stretching), particularly with rapid growth in childhood and repetitive movement of the spine → tethered cord syndrome

Foot deformities in tethered cord include: pes cavus [peroneal muscle weakness] & hammer toe [tight foot ligaments]



Chronic back pain in adolescents	
Diagnosis	**Clinical features**
Malignancy	• Nocturnal pain. Systemic symptoms. Neurologic findings if spinal cord is involved. New-onset scoliosis
Tethered cord (spinal dysraphism)	• Back/leg pain worse with activity. Neurologic findings (eg, weakness, hyporeflexia). New-onset scoliosis. Lumbosacral cutaneous abnormality
Chronic multifocal osteomyelitis	• Nocturnal pain. Systemic symptoms
Spondylosis & spondylolisthesis	• Occurrence following repetitive trauma (eg, gymnastics). Pain with extension

Ankylosing spondylitis	• Morning stiffness. Pain worse at rest. Sacroiliac joint tenderness

Guillain-Barré syndrome [motor involvement more common than sensory/autonomic]	
Pathophysiology	• Immune-mediated demyelinating polyneuropathy • Preceding GI (*Campylobacter* = ***worse prognosis***) or respiratory infection, CMV, recent immunization/surgery/trauma
Clinical features	• Paresthesia, neuropathic pain. Symmetric, ascending weakness. Decreased/absent DTR. Autonomic dysfunction (eg, arrhythmia, ileus). Respiratory compromise
Diagnosis	• Clinical [symptoms develop over weeks] • Supportive findings: Cerebrospinal fluid: ↑ protein, normal leukocytes. Abnormal EMG & nerve conduction. MRI: normal or enhancement of anterior nerve roots/cauda equina
Management	• Monitoring of autonomic & respiratory function (**spirometry**) • **IVIG or plasmapheresis**

Once GBS is suspected in a **hemodynamically stable** patient, the next step in management is serial PFT (eg, spirometry) of FVC and negative inspiratory force.

Indications for elective/emergency endotracheal intubation in GBS: decline in FVC (≤20 mL/kg), tachypnea/accessory muscle use, severe **dysautonomia** (HR/BP instability), widened pulse pressure. With early diagnosis and cardiopulmonary monitoring and support, most young patients experience spontaneous recovery.

GBS can lead to aspiration pneumonia in the setting of bulbar muscle weakness.

Bilirubin-induced neurologic dysfunction (total bilirubin ≥30 mg/dL)	
Pathophysiology	• Excess levels of free, unconjugated bilirubin cross the BBB → Deposition of bilirubin in basal ganglia & brainstem nuclei → Neuronal damage, necrosis & atrophy
Risk factors	• Prematurity. Hemolysis (G6PD deficiency). Birth trauma (cephalohematoma). Exclusive breastfeeding with excessive WL

Acute encephalopathy (may be subtle and overlooked)	• ± Reversible (treatment: phototherapy, exchange transfusion) • **Clinical findings**: Lethargy or high-pitched, inconsolable crying. Hypotonia (early) or hypertonia (late). Apnea/respiratory failure, feeding difficulties, seizures.
Chronic encephalopathy (kernicterus)	• Irreversible. Clinical findings (after early infancy): Developmental delay, SNHL, **choreoathetoid movements** (dystonic movements: involuntary twisting/writhing), upward gaze palsy

(Choice D) Physiologic chorea of infancy: benign, chorea-like movements in normal newborns, **resolves by age 8 months.**

Chapter 13: Psychiatry

Attention deficit hyperactivity disorder	
Clinical features	○ **Inattentive symptoms:** difficulty focusing, distractible, does not listen or follow instructions, disorganized, forgetful, loses/misplaces objects ○ **Hyperactive/impulsive symptoms:** fidgety, unable to sit still, "driven by a motor," hypertalkative, interrupts, blurts out answers in class • Several symptoms present **before age 12. Dx established after age 4**
Treatment	• Methylphenidate, behavioural therapy, atomoxetine (NE reuptake inhibitor, if **personal or family history of drug use** or **parent preference** against medications with abuse potential), clonidine, guanfacine, imipramine, buproprion (if atomoxetine failed)

Subtypes: predominantly inattentive, predominantly hyperactive/impulsive, combined.

ADHD is twice as common in **boys**, with **girls** more likely to have predominantly inattentive.

Diagnosis requires 6 months of ≥6 inattentive **or** ≥6 hyperactivity/impulsivity symptoms in **2 or more settings**. **If symptoms are at home only, obtain behavioural rating scales from teachers.** If the collateral information helps establish an ADHD diagnosis, the next step would be **discussing medication options**. However, if an ADHD diagnosis is **not supported**, behavioral interventions (eg, **to-do lists, daily schedules**) combined with parental support (eg, **Parent Management Training**) would be indicated.

Autism spectrum disorder (x4 more common in males)	
Clinical features	• **Deficits in social communication** with onset in **early childhood**: Impaired social engagement (back-and-forth conversation). Impaired nonverbal communication (eye contact/ gesturing). Difficulty developing/understanding relationships (interest in peers) • **Restricted, repetitive patterns of behavior**: Repetitive movements or speech. Insistence on sameness/routines. Intense fixated interests. Adverse responses to sensory input (eg, textures) • May occur with or without **language & intellectual impairment**
Assessment & management principles	• Early diagnosis & intervention • Comprehensive, multimodal treatment (eg, speech & behavioral therapy, educational services, occupational therapy) • Adjunctive pharmacotherapy for psychiatric comorbidities

Patients with **milder** autism frequently have normal or near-normal language and intellectual ability that may lead to a **delay in diagnosis** (whereas **more severe ASD is usually diagnosed by age 5**).

The diagnosis of ASD is a clinical one, but it should ideally involve a comprehensive evaluation, including structured assessments of social, language, and intellectual development in addition to hearing, vision, and genetic (eg, fragile X syndrome) testing.

When parents have differing levels of concern, physicians should take a **sensitive approach** that considers **both** parents' views, **educates** them about the spectrum of developmental disorders, and **encourages further evaluation**.

(Choice C) Separate parent interviews may be helpful if a parent is quiet or not contributing during the interview.

Fragile X commonly associated with ADHD and autism.

Diagnosis of ID requires **standardized testing** and is **not given to patients age <5.**

Tourette syndrome: multiple motor and **one or more** vocal tics that present before age 18; more common in males. The tics occur many times a day (frequently in bouts) nearly every day or at regular intervals for at least a year. Motor tics frequently observed include grimacing, shoulder shrugging, eye blinking, nose twitching, head jerking, sniffing, tongue protrusion. Vocal tics include barking, yelling, grunting, throat clearing, squeaking,

coughing. In a minority of cases, patients experience **coprolalia** (uttering of obscene words). Symptoms are exacerbated by stress and tend to subside during sleep. **Associated with OCD** (within 3-6 yrs after tics first appear, peak in late adolescence or early adulthood when the tics are waning) **and ADHD**

Less common comorbid conditions include anxiety disorders; depression; ASD; learning disorders; and disruptive, impulse-control, and conduct disorders.

Tx: **psychoeducation**, **behavioural therapy** (habit reversal training), **tetrabenazine** (dopamine depleter), **antipsychotics** (dopamine receptor antagonists), **alpha 2 agonists**. (most effective are antidopaminergic agens: tetrabenazine, risperidone, haloperidol).

Language disorder (LD): a common developmental disability of **childhood** that occurs in 5%-10% of children. LD is characterized by persistent difficulties in **comprehension (receptive)** and/or **production (expression)** of spoken and written language. It may involve the rules (grammar, syntax, morphology), content (vocabulary), and/or functional use of language. Typical symptoms include a markedly limited vocabulary, errors in tense, and difficulty producing sentences with developmentally appropriate length and complexity.

LDs interfere with academic functioning and **increase the risk for specific learning disorders** (eg, reading and writing disabilities). Early intervention includes **structured language therapy and stimulating language development at school and at home**.

- **Childhood-onset fluency disorder** (stuttering):, is an impairment in the fluency of speech production. **Speech sound disorder** is an impairment in speech articulation.
- **Social (pragmatic) communication disorder:** persistent difficulty in the social use of verbal and nonverbal communication **in multple settings**.

Stranger anxiety, part of normal development, starts at age 6 months, **peaks at 8-9 months**, and **resolves** by age **2 years**. Children with stranger anxiety typically **cry** when an **unfamiliar person** approaches, **even in the presence of the primary caregiver**.

(Choice E) Separation anxiety is also **part of normal development** and typically **resolves by age 2**. Separation anxiety disorder is diagnosed in older children who exhibit extreme and persistent anxiety with separation and excessive worry about losing major attachment figures. It occurs only when separating from the parent, **unlike stranger anxiety in which the distress occurs when the primary attachment figure is still present**.

Normal creative and healthy behaviors for young children: imaginary friends (age 3-6), pretend play, dress-up, storytelling with fanciful details.

Selective mutism tx: **CBT, family and play therapy, SSRI**

Wilson disease	
Pathogenesis	• AR mutation of *ATP7B* → hepatic copper accumulation → leak from damaged hepatocytes → deposits in tissues (basal ganglia, cornea)
Clinical findings	• Hepatic (acute liver failure, chronic hepatitis, cirrhosis) • Neurologic (**parkinsonism**, gait disturbance, dysarthria) • Psychiatric (depression, personality changes, academic decline, **psychosis**)
Diagnosis	• ↓ Ceruloplasmin & ↑ urinary copper excretion. Kayser-Fleischer rings on slit-lamp examination. ↑ Copper content on liver biopsy
Treatment	• Chelators (D-penicillamine, trientine) • Zinc (interferes with copper absorption)

Parkinsonism due to **wilson disease**: tremor, hypertonia, facial stiffness, diminished expressivity, soft voice, and pooled saliva.

Neuropsychiatric symptoms often precede hepatic symptoms in teenagers and may be **mistakenly attributed to normal adolescence or primary psychiatric illness**.

(Choice A) X-linked adrenoleukodystrophy: **adrenal gland crisis, progressive loss of neurologic function** (weakness ataxia paralysis/academic difficulties and behavioral abnormalities in early childhood or adolescence), **death**. ABCD1 mutation, defective beta oxidation of VLCFA, X-linked recessive.

Pediatric major depression may present with irritability rather than depressed mood. If a patient displays **irritability along with social withdrawal and academic decline**, MDD should be considered. Tx: individual/family therapy, antidepressants.

Childhood depression is often associated with **somatic symptoms** such as stomach aches and headaches because children often have difficulty recognizing or discussing their emotions. **Any child with a change in behavior in the context of family stress and parental substance use should be assessed for child abuse**.

Considerations for inpatient care include: **suicidal ideation**, **poor psychosocial support**, and **lack of psychiatric follow-up** (as an outpatient).

Firearm injury

Risk factors	• Male adolescent. Behavioral or psychiatric problems. Impulsive, violent, or criminal behavior. Low socioeconomic status
Prevention	• **Remove all firearms from the home.** Store firearms unloaded. Lock firearms & ammunition in separate containers

Suicide is a leading cause of mortality in adolescents (age 10-17). Over 40% of these deaths involve **firearms**, and the majority result from access to a gun in the home.

Reactive attachment disorder	
Clinical features **(<5 years)**	• Insufficient care (eg, neglect, abuse, prolonged institutionalization, inconsistent caregiving [frequently moving between foster homes]). • **Does not seek or respond to comfort**, social withdrawal, poor emotional/social responsiveness, limited positive affect • Unexplained irritability/fear/sadness even during safe encounters • **Not crying in response to an unfamiliar person/separation from a parent**
Associated features	• Toileting & sleep difficulties. Anxiety, aggression, hyperactivity/ impulsivity
DDx	• Adjustment disorder, PTSD. Global developmental delay, ID, ASD • Anxiety disorders (eg, social anxiety disorder, selective mutism) • Depressive disorders
Management	• Early intervention promoting: 1- safe, stable & enriching environment. 2- consistent, responsive caregiving/attachment figures • Psychological services (parenting skills, individual/family counseling)

(Choice C) Disinhibited social engagement disorder: overfamiliarity with unfamiliar adults [lack of boundaries when approaching adults (sitting on another person's lap, leaving with a stranger)]. Caused by severe, early neglect.

Obsessive-compulsive disorder (begins in childhood or adolescence)	
Clinical features	• **Obsessions** ○ Recurrent, intrusive, anxiety-provoking thoughts, urges, or images

	• **Compulsions**
	  ○ Response to obsessions with repeated behaviors or mental acts
	  ○ Behaviors not connected realistically with preventing feared event
	• Time-consuming (>1 hr/day)/ significant distress or impairment

Treatment	• SSRI (fluoxetine, fluvoxamine, and sertraline), CBT (exposure & response prevention)

(Choice E) CBT is the psychotherapy of choice for OCD. Psychodynamic psychotherapy, which focuses on **unconscious conflicts and developing insight**, does not include the targeted behavioral exposure techniques aimed at decreasing compulsive behaviors.

Trichotillomania (hair-pulling disorder) [scalp eyebrows eyelids]	
Features	• Recurrent hair pulling resulting in hair loss • Repeated attempts to decrease/stop hair pulling • Not due to a medical/dermatological condition (eg, alopecia areata) • Not due to another mental disorder (eg, body dysmorphic disorder)
Examination findings	• Irregular patches of hair loss. Hair shafts of variable lengths. Noninflammatory, nonscarring
Treatment	• CBT (habit reversal therapy)

Hair pulling may be an automatic behavior (without full awareness) or preceded by tension followed by relief. Prevalence = 1%-2% and is more common in **children and adolescent girls.** It commonly occurs on the patient's dominant side (ie, usually **right**).

Neonatal abstinence syndrome (in utero exposure to opioids)	
Clinical manifestations	• Uncoordinated sucking reflexes (swallowing), irritability, high-pitch crying, tremor, tachypnea, sneezing, diarrhea, seizures. Other findings include: hypertonia, shortened sleep-wake cycles, diaphoresis, yawning, vomiting, FGR
Diagnosis	• Primarily clinical. Confirm: umbilical cord blood, urine, or meconium drug testing

Treatment	• Mild withdrawal: nonpharmacologic
	  ○ Minimize environmental stimuli, swaddling, frequent small feeds
	• Moderate to severe withdrawal: pharmacologic
	  ○ Methadone, morphine, buprenorphine

(Choice E) **Propranolol** for tx of **neonatal Graves disease**: placental transfer of maternal thyroid-stimulating antibodies. Has **similar symptoms to neonatal abstinence syndrome**

Sexual behavior in preadolescents	
Normal	**Abnormal (red flag for abuse)**
Toddler • Exploring one's own or others' genitals • Masturbatory movements • Undressing self or others • Playing "doctor" with other children	• Repeated insertion of objects into vagina or anus • Sex play involving genital-genital, oral-genital, or anal-genital contact • Use of force, threats, or bribes in sex play • Age-inappropriate sexual knowledge, • Preoccupation with masturbation, • excessive or aggressive talk about sexuality
School-age • Increased interest in sex words & play • Asking questions about sex & reproduction • Masturbatory movements (may become more sophisticated)	

Repeated UTIs with no medical cause is a red flag for sexual abuse.

(Choice A) It is common for young children to express frustration through physical aggression. This tends to improve around age 5 or 6.

Pyromania (rare impulse-control disorder, males)	
Clinical features	• Deliberate fire setting >1 occasion. Fascination with fire. Tension/ arousal prior to act; pleasure/relief when setting/witnessing fires. No external motivation (eg, financial gain, political statement, recognition)

Differential diagnosis	• Conduct disorder [rule violation, theft, cruelty toward people/animals], antisocial personality disorder. Mania, psychosis. Impaired judgment (eg, neurocognitive disorder, substance intoxication)

Individuals with this condition tend to be fascinated by fire and anything related to it (eg, **fire stations, firefighters, cigarette lighters**).

(Choice C) Normal adolescence: arguing with parents and testing limits (eg, forging a parent's signature).

Chapter 14: Public Health Sciences

An ANOVA that is **not statistically significant** (eg, $p \geq 0.05$) indicates that all group **means are equal** whereas an ANOVA that is **statistically significant** (eg, $p < 0.05$) indicates only that **at least 1 group mean is different** from the rest. However, an ANOVA does not determine which group means differ—further evaluation is required to determine the specific difference.

Case series is a descriptive observational study design in which a (generally small) group of patients with a similar diagnosis or treatment is described at a point in time or followed over a certain period **with no control group**.

Experimental studies (RCT) can help establish **causal** relationships whereas observational studies (eg, case series) only suggest **associations**. Results of observational studies do not provide enough scientific evidence to make clinical decisions (eg, treating patients with new drugs or therapies), regardless of sample size or study type.

According to the **HFE hierarchy of effectiveness**, strategies that remove or greatly **reduce human effort** for correct action are most effective. Infusion pumps with dosing algorithms and preset limits (ie, **"smart pumps"**) apply the most effective HFE strategies to reduce heparin-associated errors:

- **Computerized automation**: Dosing **algorithm software** automates conversion between infusion protocols, preventing mathematical errors. Automation is appropriate for constant, predictable processes (eg, constant heparin infusion on a hospital floor) but is less suitable for dynamic processes requiring critical analysis and frequent adjustments (eg, administering heparin boluses in an operating room).
- **Forcing functions/preset limits** are **hard stops** in a design or process that eliminate the risk of incorrect action. (Preconfigured dose limits)

HFE strategies requiring greater human effort for correct action are comparatively less effective than forcing functions and automation, including:

- Checklists (eg, reviewing list of sterile precautions prior to central line insertion)
- "Forced pauses" or time-out (eg, prior to surgery to verify procedure and patient)
- Double-checks (eg, requiring 2 providers to calculate an infusion dose)
- Organizational policy change, including promoting shared governance (eg, employee involvement in structuring organizational culture)

Although these strategies are important and do reduce error risk, they are vulnerable to human factors such as **confirmation bias** (eg, tendency to interpret information toward an anticipated outcome, such as believing all items on a checklist are complete) and **workarounds** (bypassing protocols to increase efficiency, such as skipping certain components of a time-out to expedite a procedure).

Human factors engineering strategies

Reliability	Strategy	Description & examples
Highest	**Forcing functions**	Hard stops in design or process to eliminate risk of incorrect use Example: each anesthesia gas fits only one compatible socket & is not interchangeable
	Computerized automation	Automated processes to remove human effort & variations that cause error Example: automated vital signs monitoring
	Environment & physical layout	Workspace design to facilitate correct action & minimize error Example: look-alike drugs stocked in different locations
High	**Standardization & simplification**	Uniform processes to minimize variation, complexity & learning curve Example: every hospital unit follows the same process for heparin administration
	Human-machine redundancy	Repetitive step to confirm correct action in an error-prone process Example: barcode scanning of medications in addition to visual inspection
Medium	**Reminders, alerts & double-checks**	Processes prompting providers to check actions to reduce errors Examples: drug-drug interaction alerts; time-out before procedures

Human factors engineering seeks to reduce error risk by designing systems based on expected human behaviors. Less reliable HFE strategies include trainings, policy changes & education.

Burnout: emotional exhaustion (eg, insomnia, tearfulness), depersonalization (eg, appearing withdrawn), low sense of personal accomplishment (eg, failure at the job), commonly affects physicians in the **ICU** who have frequent exposure to death and severe illness but often receive little support for **grief** associated with caring for dying patients.

Residents are vulnerable to perceiving untimely death as a personal failure, and have few protected opportunities to discuss emotionally distressing episodes. **Interdisciplinary debriefing sessions** ("**grief rounds**"), which are dedicated to discussing psychosocial aspects of patient care from the provider's perspective, are associated with **improved physician well-being** and coping skills.

Grief rounds are conducted in specific settings (ICU), with **protected time** given to residents to attend regularly (biweekly, monthly). They are characterized by the following:

- **Team-based** structure (eg, physician, psychotherapist, chaplain)
- **Interactive format**, which elicits open sharing of experiences and reduces perceived stigma associated with expressing emotional distress
- **Focus on the caregiver's experience** associated with loss and caring for ill patients

(Choice D) Fatigue can increase risk for burnout, and **duty hour and sleep restrictions** are associated with reduced fatigue and improved learning and safety.

The **Plan-Do-Study-Act** (PDSA) and **Plan-Do-Check-Act** (PDCA) paradigms are quality improvement (QI) tools that involve cyclical processes with the goal of continual improvement. Once a QI issue is identified (eg, elevated rate of failed extubations among preterm infants), a 4-step process is put in place to address it, as follows:

- **Plan:** Identify any factors contributing to the issue (eg, lack of standardized extubation strategies), set an objective (eg, reduce failed extubation rate by 30%), and put a plan in place to carry out a cycle (eg, new protocol and checklist development, training in preparation for implementation).
- **Do: Implement** the plan (ie, new protocol) while making sure to collect data and document problems and unexpected observations.
- **Study (or Check):** Using the data gathered, complete a data analysis that **compares the results** obtained to the **objectives** set (eg, evaluate variations in failed extubation rates). Although the terms PDSA and PDCA (which may have developed in different settings) are sometimes used interchangeably, the use of "S" ("Study") in PDSA is felt to better emphasize the need for a full analysis.
- **Act:** Based on these data, identify changes that need to be made and lessons learned, and then begin work on the next cycle if needed to achieve the objective.

Hypoxia, hypotension, and tachypnea immediately following "**flushing**" (injecting a small volume of low-concentration heparin or saline, done to prevent catheter occlusion, dislodging the thrombus [formed at catheter tip] into systemic circulation & lungs) /removal /repositioning of central venous catheter (CVC) -> PE due to **catheter-related thrombus**.

- **CVC** leading risk factor for (VTE) in children. CVCs introduce foreign material (triggering the coagulation cascade), interfere with blood flow, and cause intravascular micro-trauma (thrombogenic)

- Meningococcemia: in **hypercoagulability** due to activated protein C deficiency.

Heparin flushes and DVT prophylaxis (LMWH) do not prevent catheter-related thrombus.

(Choice B) Incorrect CVC placement (tip in bronchus): acute pulmonary edema: hypotension, hypoxia, and tachycardia, immediately following **initial** CVC use.

(Choice D) Incorrect heparin administration (accidentally using high- rather than LMWH) can result in immediate, catastrophic cerebral hemorrhage (eg, seizures, vomiting, abnormal pupillary findings) and systemic bleeding (eg, from catheter site).

Discussions regarding advance care planning for **adolescents** with chronic, life-limiting illnesses (cystic fibrosis) should take a **family-centered approach** and involve the adolescent patient alongside the parents. Discussion should include information about prognosis, care options, and the **patient's and family's** preferences for end-of-life care.

Physicians should encourage parents to include minors in treatment decisions, including advance care planning, **as developmentally appropriate.**

Drug testing **adolescents** without their knowledge is inappropriate. **Parents** requesting surreptitious drug testing should be **engaged to understand their specific concerns**, **educated about the limitations of drug testing**, and **informed about the need for private patient evaluation.**

Parents cannot refuse life-saving treatment for their child in emergency situations (intubation in neonatal RDS, tetanus vaccine after deep puncture wound). If a parent refuses medically necessary care for a child in such a situation, life-saving treatment should proceed without consent. BUT parents can decline elective routine vaccination.

If parents attempt to disrupt emergency care in these situations, **security** may need to assist while **local law enforcement** and **child protective services** are contacted.

Parental consent is required prior to providing nonemergency medical care for a minor who does not qualify for confidential care (eg, pregnant) or is not emancipated (married). **Assent from the child is ideal but not required** to proceed with treatment.

In pediatric patients with a chronic or terminal illness and poor prognosis, **the patient's understanding should first be determined** before making a detailed plan with parents for disclosure.

Disclosure of an HIV diagnosis to a child with perinatally acquired infection should occur by adolescence to **foster patient autonomy, increase medication adherence**, and

prevent transmission. The physician should respect the family's concerns and **offer joint participation in establishing a timeline and plan** for disclosure to the child.

In patients with divorced parents, only parents with **custody** may give consent for medical care. Parents with joint custody are each able to consent, but consent from only **one parent** is all that is necessary to proceed with treating the minor, especially when the decision is clearly in the child's best interests.

Protecting the **adolescent's confidentiality** is important to maintaining a **trusting patient-physician relationship** for discussions of sensitive topics, such as mental health, drug use, and sexuality. **At the initial visit**, physicians should prepare adolescents and guardians for increasing adolescent autonomy in health care decisions while promoting open communication between the parties. This discussion should also include situations in which the physician is ethically or legally bound to breach confidentiality **(conditional confidentiality)**: **imminent risk** (eg, suicidality, homicidality), **threats to safety** (eg, sexually abusive relationships, intoxicated driving), **legal mandates**.

When confidentiality must be breached, it **should be discussed first with the adolescent**, who **should be given the option** to broach the topic with the guardian.

Establishing rapport and maintaining privacy with adolescents supports positive health practices, but there are circumstances that require **disclosure to guardians** (eg, imminent risk, recurrent safety concerns), to **state protective agencies** (eg, abuse, neglect), or to **health departments** (eg, transmissible infections). Keeping everything "between you and me only" (**unconditional confidentiality**) undermines the physician's duties to practice in the patient's best interest (**beneficence**) and to do no harm (**nonmaleficence**).

(Choice E) Although the Health Insurance Portability and Accountability Act (**HIPAA**) addresses provider responsibilities regarding patient confidentiality practices, it **does not ensure unconditional confidentiality for adolescents or expressly prohibit guardians from lawfully accessing sensitive information via record request.**

When transferring care, it is preferable to **provide the medical record directly to the new provider** rather than providing a copy to the parent.

Providing the parent with only nonconfidential information maintains confidentiality but **fails to ensure continuity regarding the entirety of the patient's treatment history**.

Coining: cultural practice from China and Southeast Asia, a coin is rubbed on the body, producing multiple symmetric linear erythema/ecchymoses on the chest/back. Thought to alleviate illness and fever, it is usually **safe**. To provide culturally sensitive care, **physicians should not discourage patients from the practice or report them for misperceived**

abuse. Another common cultural practice is **cupping**, in which a heated cup applied to the skin creates **suction**, resulting in **circular ecchymoses**. These practices can be painful but usually carry little risk. Further management may include offering less painful alternatives (eg, ibuprofen for fever, antibiotics if indicated).

If both parents refuse to consent to **necessary but nonurgent** life-saving treatment (chemotherapy for ALL in a stable patient), the physician should continue to engage the parents, explaining the benefits of treatment and the consequences of withholding diagnostic tests and chemotherapy. The hospital ethics committee, social services, and hospital risk management can also assist. In some cases, this multidisciplinary approach will enable parents to overcome their fears and consent to the proposed treatment. However, **if the parents continue to refuse ->** seek a **court order**

Parents of a child with CP can experience severe **guilt** over the thought of causing their child's condition. It is important for physicians to allow parents to express their emotions and engage compassionately. Physicians should relieve feelings of guilt by providing reassurance that the parents did **not cause the condition** and that they should **not blame** themselves. Counseling is often necessary to help the family deal with the grief and frustration that is to be expected after receiving this diagnosis.

Asking open-ended questions ("Why do you think you are responsible for this condition?") is typically the best approach, but in this case asking the mother why she feels responsible suggests that there may have been direct and preventable actions that caused the child's condition. Instead, the provider should immediately remove blame and provide comfort.

Testing in children is performed based on the **onset of disease**. Tests for genetic conditions with an early onset of disease (CF, PKU) are performed because early diagnosis affects management in childhood. In contrast, genetic testing in children for an **adult-onset condition** (eg, breast cancer) is **typically not recommended** because it usually does not affect patient care (eg, frequency of monitoring) or prognosis. It should be **deferred until adulthood to ensure informed consent**.

Breast cancer monitoring during childhood has no benefit but has **potential psychosocial harm**. Unlike tests used for screening (eg, colonoscopy for colon cancer), the timing of predictive genetic testing for *BRCA1/2* does not depend on the affected relative's age at diagnosis. **If a pathogenic *BRCA1/2* variation is confirmed**, patients typically undergo breast cancer screening (eg, MRI) beginning **at age 25**. Risk-reducing surgery for breast and ovarian cancer (ie, prophylactic mastectomy and salpingo-oophorectomy) is also discussed with adult carriers.

In some cases, parents may wish to **withhold information** due to **concern for potential adverse effects** experienced by their child (HCM dx in an athlete). The physician can help alleviate some concerns by educating parents about **age-appropriate** health care participation. In addition, the physician can open a dialogue about disclosure of difficult information by **explaining the rationale** for involving this adolescent in his health care (eg, "knowing his diagnosis could help the child be aware of potential symptoms").

Medication nonadherence	
Risk factors	• Complex treatment regimen. Adverse drug effects. Expensive drug regimen/limited patient financial resources. Little immediate or apparent benefit of medication. Inadequate physician supervision & written instruction
Interventions to improve adherence	• Integration into daily habits/schedule. **Pill organizers & dispensers**. Simplified treatment regimen. **Automated reminders** (eg, smartphone applications). Frequent telephone contacts & interprofessional (eg, nurse, pharmacist) follow-up. Motivational interviewing. Consolidated refill schedule

Clinical features suggesting nonadherence: sudden or **unexplained increase in seizure frequency**, **subtherapeutic drug levels**, and **recurring missed follow-up appointments**.

(Choice A) Family reminders are sometimes helpful, but they can increase the family care burden, may not always be practical, and can lead to increased parent-child conflict.

Chapter 15: Toxicology & Environmental Exposure

Organophosphate poisoning (DUMBBELSS:+ muscle weakness, paralysis, fasciculations)	
Common exposures	• Pesticide: farmer/field worker, pediatric ingestion, suicide attempt • Nerve agent: multiple patients presenting with similar symptoms
Management	• ABC (Remove patient's clothes + copious **irrigation of the skin** and/or eyes to prevent cutaneous absorption) THEN atropine THEN pralidoxime • Atropine reverses muscarinic symptoms

	• Pralidoxime, a **cholinesterase-reactivating agent,** reverses both nicotinic and muscarinic symptoms

Nicotine poisoning (children age <6 are particularly susceptible, life threatening)	
Epidemiology	• Toxicity dependent on amount & type of exposure*: ○ Ingested (eg, cigarette ends, nicotine gum, concentrated liquid nicotine). Cutaneous (eg, green tobacco sickness from tobacco leaves). Inhaled (eg, e-cigarettes)

Features	• Biphasic response due to action of nicotine at Ach receptors:
	  ○ Stimulatory (early <1hr) phase (agitation N V HTN, tachycardia, seizures, myoclonus): relatively small concentration of nicotine acting as an agonist at the nicotinic receptors. (↑ sympathetic)
	  ○ Inhibitory phase (late ~1-4 hr) (eg, hypotension, bradycardia, coma): larger concentrations of nicotine overwhelm the receptors, resulting in functional inhibition (delayed parasympathetic effects)
	• Muscarinic symptoms (DUMBBELSS)

Treatment	• Primarily **supportive**. **Decontamination** if transdermal exposure. **Benzodiazepines** for seizures. **Atropine** for symptomatic bradycardia & muscarinic symptoms
*Highly concentrated nicotine liquid used in e-cigarettes can be toxic in small amounts.	

(Choice C) Hemodialysis is sometimes used to treat toxic alcohol poisoning, which presents with AMS + vision changes (methanol) or flank pain & hematuria (ethylene glycol).

Large local reactions [higher risk for developing a future systemic reaction (<3%-16%)]	
Epidemiology & pathophysiology	• Bites from *Hymenoptera* species (bees, ants, wasps, yellow jackets). Exaggerated, IgE-mediated, local allergic response • Associated with possible ↑ risk for future systemic reactions
Manifestations	• Significant **(~10 cm or more)** localized swelling, erythema & warmth, **contiguous** with site of the sting • Develop within 24 hr, peak within 24-48 hr, resolve in 5-10 days

Treatment	• Cold compresses. Oral &/or topical steroids. Antihistamines. Anti-inflammatory medications

(Choice A) Cellulitis generally **occurs 3-5 days after the initial bite.**

(Choice D) Lymphangitis occurs when bacteria from a distal site seed the lymphatic channels, causing **erythematous, painful streaks in the skin and fever.**

(Choice F) Toxic reactions: **infants or elderly** who have **sustained envenomation** from **numerous stings** by members of the *Hymenoptera* species. Symptoms (**nausea, vomiting, diarrhea, seizures, fever**).

Heat exhaustion (vs exertional heat stroke: fever >40 C + CNS dysfunction)	
Risk factors	• Strenuous activity during hot & humid weather. Dehydration, poor acclimatization. Lack of physical fitness, obesity. Acute illness. Older age. CF (due to infection + Na Cl lost in sweat) • Medications: anticholinergics, antihistamines, phenothiazines, tricyclics, antipsychotics
Manifestations	• Hyperthermia ≤40 C with **normal mental status.** Profuse sweating. N V. Headache, weakness, dizziness. Tachycardia, tachypnea, hypotension
Management	• Cool patient (eg, air conditioning, cool water shower) • Chilled salt-containing oral fluids

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When **physostigmine** is used for **atropine overdose**, it is administered **slowly and in small doses** to avoid sending the patient into cholinergic toxicity.

Clinical features of caustic ingestion	
Clinical presentation	• Chemical burn or liquefaction necrosis injury. Affects lips, tongue, oral mucosa & esophagus • Patients can develop chest & abdominal pain, vomiting with occasional hematemesis & dysphagia/odynophagia
Management	**Prehospital** • Decontamination (eg, remove contaminated clothing, brush off visible chemical, irrigate exposed skin)

	• Do not induce vomiting (milk, water, activated charcoal, vinegar, or nasogastric lavage) **Emergency department** • Confirm decontamination; **chest & abdominal x-rays** • **Endotracheal intubation** for significant oropharyngeal injury • Consider **gastric lavage if nasogastric tube is placed** **Inpatient** • UGI endoscopy within 12-24 hr if hemodynamically stable & without respiratory distress or perforation (The extent of injury may not be apparent if performed immediately, and delayed endoscopy increases **perforation** risk) • **Serial x-rays** to rule out perforation. **Tube feedings & surgical intervention for severe injury**

(Choice A) All patients with **persistent dysphagia** or **significant esophageal burns** on endoscopy should undergo **barium contrast studies 2-3 weeks after ingestion** to assess for **esophageal strictures** or **pyloric stenosis**.

(Choice C) Vinegar combined with an **alkaline substance** can cause an **exothermic** reaction and burn the mucosa, exacerbating the existing injury.

Acute iron poisoning (accidental ingestion of mother's **prenatal vitamins)**	
Features	• Abd pain, hematemesis/melena, diarrhea. Shock. Liver necrosis
Findings	• AG metabolic acidosis (H^+ production during iron absorption and ↑ lactic acid in shock). ↑ serum iron. Radiopaque pills on abd x-ray (‘**small opacities in the stomach and intestines**’)
Treatment	• Aggressive IV fluids (1st line). Deferoxamine. Whole bowel irrigation. Hemodialysis if deferoxamine failed

Vomitus and stool may appear **black or green** from **disintegrating iron tablets**.

Because iron is rapidly taken up by various organs (eg, myocardial cells, hepatocytes), **deferoxamine must be administered early to be effective**. Severe, untreated acute iron poisoning may lead to **distributive shock** (due to systemic vasodilation), **cardiogenic shock** (due to iron-induced myocardial injury), **hepatic necrosis, and death.**

Clinical presentation of methemoglobinemia	
History	• Oxidizing substances (dapsone, nitrites, local/topical anesthetic)
Clinical examination	• Cyanosis (not improved with O_2). Pulse oximetry saturation ~85% (Because methemoglobin absorbs light at a different wavelength than hemoglobin). Dark chocolate–colored blood
Laboratory findings	• **Saturation gap** (>5% difference between oxygen saturation on pulse oximetry & ABG). **Normal PaO_2. Abnormal co-oximetry test** (analyzes hemoglobin **absorption wavelengths** and can identify **hemoglobin, methemoglobin, and carboxyhemoglobin**)

Ferric iron is unable to bind oxygen + changes the hemoglobin structure and causes ferrous sites to have ↑ affinity for oxygen ("left shift" on oxygen-dissociation curve) → decreased oxygen release in peripheral tissues (ie, **decreased oxygen delivery**).

The antidote for **acquired symptomatic** methemoglobinemia or **high levels** of methemoglobin (as measured by co-oximetry) is **methylene blue**. Methylene blue acts as an **electron acceptor** for NADPH and is reduced to **leucomethylene blue**, which in turn **reduces methemoglobin to hemoglobin**. High-dose (vitamin C) acts as a reducing agent and can be used when methylene blue is unavailable or contraindicated (G6PD).

(Choice B) ↑ plasma osmolal gap: ethanol, methanol, or ethylene glycol ingestion.

Lead poisoning (inhalation/ingestion)	
Risk factors	• Home built before 1978 (especially during renovation). Pica or mouthing behaviors (eg, infants, developmental delay). Sibling with lead poisoning. Low socioeconomic status. Immigrant or international adoptee. Lead piping. Having a parent who works with batteries or pottery
Clinical features	• May be asymptomatic. Abdominal pain/constipation. WL, fatigue, IDA. Cognitive impairment/behavioral problems. Encephalopathy. **Blue "lead lines"** along the gingival margin
Management	• Obtain **venous** sample (if a screening capillary [fingerstick] lead level is ≥5 µg/dL). Environmental surveillance (identify & remove lead sources). Notify public health department • Nutritional counseling. **Chelation** therapy if **lead level ≥45 µg/dL**

Targeted screening of high-risk populations regardless of symptoms is important as most children with lead toxicity are initially **asymptomatic** but can have cognitive and behavioral problems that become apparent after school entry.

(Choice A) Wallpapering over walls painted with lead-based paint is **not an appropriate treatment measure**, as the paint can still loosen underneath the paper and release lead dust. Lead paint should be encapsulated or removed by a professional to avoid exposure.

Dimercaptosuccinic acid (**succimer**) is typically used when lead levels are **45-69** µg/dL. **Dimercaprol** (British anti-Lewisite) plus calcium disodium edetate (**EDTA**) should be administered on an **emergency** basis for **levels ≥70 µg/dL** or **acute encephalopathy**.

Abdominal x-rays: radiopaque lead flecks, paint chips, or lead-paint–covered foreign bodies. Indicated if elevated levels **and** GI symptoms (constipation, abdominal pain, vomiting) **or** suspicion of foreign body ingestion.

(Choice G) Patients with elevated blood lead levels **should be screened for iron deficiency** and prescribed oral ferrous sulfate if deficiency exists. **Comorbid iron deficiency can increase gastrointestinal absorption of lead.**

Chapter 16: Allergy & Immunology

Diagnostic criteria for anaphylaxis (Tx: IM epinephrine)	
Anaphylaxis is likely if there is **rapid symptom onset & any 1** of the following criteria:	
1	Skin/mucosa involvement (eg, hives, lip/tongue swelling) & either hypotension or respiratory distress
2	Involvement **of ≥2 organ systems** after exposure to a **likely** allergen • Skin/mucosa (eg, hives, lip/tongue swelling) • Respiratory (eg, wheezing, stridor, dyspnea) • Cardiovascular (eg, hypotension, tachycardia, syncope) • Gastrointestinal (eg, abdominal pain, vomiting, diarrhea)
3	Hypotension after exposure to a **known** allergen

Management of anaphylaxis	

Immediate management	• Epinephrine (**most important**): [B2 agonist: bronchodilation & ↓ release of inflammatory mediators + Alpha 1 agonist: vasoconstriction, ↑BP, ↓ upper airway edema]
	  ○ IM preferred, **may be repeated (eg, 3 doses)**
	  ○ IV in severe/refractory cases
	• IV crystalloid & Trendelenburg positioning for hypotension. Albuterol for bronchospasm. Early intubation for upper airway obstruction [oropharyngeal swelling, stridor, voice alteration]

Adjunct management	• H_1/H_2 antihistamines. Glucocorticoids. Glucagon for patients on beta blockers (reversal). **Hospital admission** for **severe** initial presentation (eg, shock) or **ongoing** symptoms despite treatment

Symptoms may recur or persist despite treatment, according to the following patterns:

- **Biphasic anaphylaxis** is characterized by an initial anaphylactic reaction followed by an asymptomatic period (typically 1-30 hr) and then a recurrence of symptoms.
- **Protracted anaphylaxis** (less common) is characterized by anaphylactic symptoms that last hours or days despite treatment.

(Choice F) Venom immunotherapy is a highly effective desensitization therapy for patients with anaphylaxis to bee (or other *Hymenoptera*) stings; however, it **takes months to years to be effective** and is not an appropriate treatment for acute anaphylaxis.

Hereditary angioedema (face, limbs, and genitalia are most commonly affected)	
Pathophysiology	• Autosomal dominant (MC) or de novo mutation • C1 inhibitor deficiency/dysfunction • **Excessive bradykinin** → fluid extravasation into skin & mucosal tissues (eg, bowel wall, upper airway)
Clinical features	• Episodic cutaneous swelling (eg, face, extremities, genitalia) ○ No urticaria or pruritis. **No identifiable trigger** (eg, ACE inhibitor, NSAID) • Colicky abd pain, V, diarrhea. Laryngospasm, airway obstruction

Diagnosis	• ↓ C4 level. ↓ C1 inhibitor protein or function
Acute management	• C1 inhibitor concentrate. **Bradykinin antagonist** (eg, icatibant). **Kallikrein inhibitor** (eg, ecallantide)

Attacks are often precipitated by stress or minor trauma (eg, injections). Self-limiting in 2-5 days. NO pruritus, urticaria, bronchospasm, hypotension.

Serum sickness & serum sickness–like reaction

	SS	**SSLR**
Common triggers	Foreign proteins in antivenom, antitoxin, or monoclonal antibody	Medications, particularly cefaclor, penicillin & TMP-SMX
Immune complexes	High titer	Mild or none
Complement activation	Extensive	Minimal or none
Onset	5-14 days after exposure	5-14 days after exposure
Fever	High	Low-grade
Arthralgia	Yes	Yes
Urticaria	Yes	Yes
Resolution	Spontaneous	Spontaneous (+ discontinue drug)
Notes	More severe	Most common in children

Both share fever, mildly pruritic urticarial rash that persists after 24 hrs, arthralgia [significant multiarticular joint pain, NO joint swelling erythema warmth], diffuse lymphadenopathy.

SSLR is due to genetic deficiencies in eliminating the metabolic by-products of certain medications, which leads to hapten-mediated cytotoxic T-cell injury or direct cytotoxicity.

(Choice E) Spontaneous SSLR does not occur; SSLR and SS require recent exposure to medication, vaccine, or foreign protein antigens.

X-linked agammaglobulinemia

Pathophysiology	• *BTK* gene mutation resulting in defective Bruton tyrosine kinase • Impaired B-cell maturation & immunoglobulin production
Clinical manifestations	• Recurrent sinopulmonary & GI (*Salmonella*, *Campylobacter, Giardia*) infections at age >3-6 mo (↓ maternal IG). Chronic enteroviral infection (meningoencephalitis). Absent/small LN (Notably, these tissues are not prominent even in healthy children age <2)
Labs	• ↓ Immunoglobulins & antibody response to vaccines • Flow cytometry: ↓ $CD19^+$ B cells & normal T cells
Treatment	• IG replacement therapy. Prophylactic antibiotics if severe

Live vaccines are contraindicated in XLA; other vaccines are not contraindicated but do not generate an effective antibody response.

Selective IgA deficiency (MC primary immune deficiency)	
Features	• Usually asymptomatic. Recurrent sinopulmonary & gastrointestinal infections. Associated with autoimmune disease (SLE, celiac) & atopy (eg, asthma, eczema). **Anaphylaxis during transfusions**
Diagnosis	• Low or absent IgA. Normal IgG, IgM levels, B cells
Treatment	• Supportive care. Medical alert bracelet for transfusion reactions (for severe deficiency) and receive blood products that are washed of residual plasma or from an IgA-deficient donor.

Patients with severe IgA deficiency: IgE antibodies directed against IgA (anti-IgA abs).

Common variable immunodeficiency (Tx: IVIG)	
Pathophysiology	• Abnormal **differentiation** of B cells into plasma cells → decreased immunoglobulin production
Clinical manifestations	• Symptom onset classically **age 20-40, as early as puberty** • Recurrent sinopulmonary infections (eg, pneumonia, sinusitis, otitis) [encapsulated organisms] • Recurrent GI infections (*Salmonella, Campylobacter, Giardia*)

	• Chronic disease:
	  ○ Autoimmune (eg, RA, thyroid disease)
	  ○ Pulmonary (eg, bronchiectasis, fibrosis)
	  ○ GI (eg, chronic diarrhea, IBD-like conditions), FTT

Diagnosis	• ↓↓ IgG, ↓ IgA/IgM. **No response to vaccination**

Lymphocyte and B-cell concentrations are generally **normal, but plasma cells are decreased**. Patients have ↑ risk of autoimmune disease and lymphoma, despite tx.

Severe combined immunodeficiency	
Etiology	• Gene defect leading to failure of T-cell development • B-cell dysfunction due to absent T cells
Inheritance	• X-linked recessive. Autosomal recessive
Clinical features	• FTT, chronic diarrhea, thrush, recurrent, severe viral, bacterial, fungal, protozoal, or opportunistic (ie, *Pneumocystis*) infections. Absent LNs
Treatment	• **Urgent** stem cell transplant (fatal in early childhood if left untreated)

SCID is included in routine newborn screening in the United States and is detected by the absence of T cell receptor excision circles (circular DNA excreted by developing T cells in the thymus) in dried blood.

(Choice A) Antiviral and antifungal prophylaxis are indicated in patients with impaired cellular immunity (**CGD, SCID**).

Humoral immunodeficiency syndromes					
Condition	**B cell count**	**IgG**	**IgA**	**IgM**	**IgE**
Selective IgA deficiency	Normal	Normal	↓	Normal	Normal
Job syndrome (hyper-IgE syndrome)	Normal	Normal	Normal	Normal	↑
CD40 ligand deficiency (hyper-IgM syndrome)	Normal	↓	↓	↑	↓

Common variable immunodeficiency	**Normal**	↓	↓	↓	↓
X-linked agammaglobulinemia	↓	↓	↓	↓	↓

Hyper-IgM syndrome: infancy, **recurrent sinopulmonary infections** by encapsulated bacteria (due to lack of opsonizing IgG) + **viral + PCP + FTT**. **Lymphocytosis** can occur during active infection, and **neutropenia** is common. **High CD4/CD8 Ratio.** Treatment includes **antibiotic prophylaxis and intravenous immunoglobulin.**

Transient hypogammaglobulinemia of infancy (prolonged IgG physiologic nadir)	
Clinical features	• May be asymptomatic. Mild recurrent respiratory & GI infections in infancy. Atopy (asthma, eczema, food allergies)
Laboratory findings	• ↓ IgG (± ↓ IgA and/or IgM) [**IG levels normalize by age 9-15 mo**]. Normal Ab response to vaccines. Normal B and T lymphocytes
Treatment	• Observation; prophylactic Abs for recurrent infections; self-resolution in early childhood

A physiologic nadir in infant IgG levels occurs around age 3-6 months, at which time infants start to produce their own IgG, and levels begin to rise again. However, in patients with THI, **IgG synthesis is delayed**, and the physiologic nadir is **prolonged beyond age 6 months**.

Hyper-IgE syndrome	
Pathogenesis	• Autosomal dominant. Defective JAK-STAT signaling → impaired Th17 ↓ Neutrophil proliferation/chemotaxis
Clinical features	• Noninflammatory (ie, cold) abscesses (*Staphy, Candida*). Retained primary teeth. Coarse facies (broad nose, prominent forehead). **Severe, chronic eczema (first manifestation of the disease in the 1st few weeks of life)**. Recurrent sinopulmonary infections
Labs	• ↑ IgE. Eosinophilia. Normal leukocyte count with ↓ Th17
Management	• Supportive skin care. Antibiotic prophylaxis & treatment

(Choice D) Cyclic neutropenia: recurrent episodes of **reduced peripheral neutrophils, fever, sore throat, and lymphadenopathy**, commonly occurring **every 3 weeks.**

(Choice E) Wiskott-Aldrich syndrome: X-LR, **microthrombocytopenia** (bleeding postcircumcision/from umbilical stump), eczema, recurrent bacterial (pyogenic) viral infections. B and T cells are classically decreased but may be normal in infancy, and immunoglobulin profile shows low to normal IgG and IgM with **elevated IgA and IgE**.

Primary immunodeficiency syndromes

Classifications	Age of onset	Key features	Laboratory findings	Examples
B-cell disorders	Variable (>4-6 months)	Encapsulated bacteria (recurrent sinopulmonary infection) Enterovirus*	↓ Ig & vaccine response	X-linked agammaglobulinemia Common variable immunodeficiency
T-cell (& combined B- & T-cell) disorders	Early (<4-6 months)	Viral, bacterial & fungal infections (eg, *Candida*, *Pneumocystis*) Failure to thrive	↓ Leukocytes ± ↓ Ig & vaccine response	DiGeorge syndrome (T) Severe combined immunodeficiency (B & T) Wiskott-Aldrich syndrome (B & T) Ataxia-telangiectasia (B & T)
Phagocyte disorders	Early (childhood)	Skin & soft tissue infections Fungal infections Catalase-positive organisms**	Normal Ig Impaired oxidative burst**	Chronic granulomatous disease Leukocyte adhesion defect
Complement disorders	Variable	*Neisseria* infections Autoimmune disease	↓ CH50 Normal leukocytes & Ig	C1q deficiency Terminal complement (C5-C9) deficiency

*Only in X-linked agammaglobulinemia.
**Only in chronic granulomatous disease (eg, *Staphylococcus aureus*, *Burkholderia*, *Serratia*, *Aspergillus*, *Nocardia*).

Chronic granulomatous disease (Nocardia → cavitary pneumonia)	
Pathogenesis	• X-LR (boys), mutation of NADPH oxidase, age <5
Clinical features	• Recurrent infections with catalase-positive* bacteria & fungi • Lungs, skin, liver, LN (suppurative adenitis), bones involvement • Diffuse granulomas (eg, gastrointestinal, genitourinary), IBD
Diagnosis	• Measurement of neutrophil superoxide production: DHR flow cytometry (preferred)/NBT testing. Normal neutrophil count
Treatment	• Prophylaxis: TMP-SMX, itraconazole, immunomodulator (IFN γ) • Active infection: culture-based, antimicrobial therapy • Hematopoietic cell transplant is curative

Patients are predisposed primarily to recurrent **lung infections** (pneumonia, empyema, hilar lymphadenopathy), **abscesses** (skin, liver, lymph node), perirectal infection

Because **breakthrough infection** may involve resistant bacterial or fungal organisms, patients (unless they are critically ill) generally should undergo **biopsy** of areas of active infection **prior to initiating empiric antimicrobial therapy**. In the case of pulmonary infection, **bronchoscopy** with biopsy is typically performed, followed by initiation of broad-spectrum antibacterial and antifungal therapy while awaiting biopsy results.

Live bacterial vaccines such BCG vaccine should be **avoided** in CGD because they can induce severe infection. **Live viral** vaccines, as well as **inactivated** vaccines, are **safe** because patients with CGD do not have a defective immune response to viruses.

CGD can cause **IBD** in children <5. **Pigment-laden histiocytes** are characteristically seen on histology and represent **lipofuscin accumulation** due to **impaired lipolytic activity**.

(Choice C) Primary immunodeficiency in NK cells: severe HSV in infancy.

Leukocyte adhesion deficiency (S aureus & gram negative bacilli are most common)	
Pathophysiology	• AR defect in CD18-containing integrins • Impaired leukocyte adhesion & **endothelial transmigration**
Clinical features	• Delayed umbilical cord separation (age >30 days). Skin & mucosal infxs (cellulitis, omphalitis, gingival erythema/ulceration, periodontitis) without pus formation. Impaired wound healing
Labs	• Leukocytosis & neutrophilia

GI and respiratory tract (eg, pneumonia, pleural effusion without leukocytes) involvement can also occur. Infections may be prolonged and are often life-threatening.

Preterm infants should receive routine immunizations according to **chronologic age** rather than age corrected for gestation. Live virus vaccines are withheld in immunocompromised patients, but **mild intercurrent illness is not a contraindication to vaccination**.

(Choice B) Corrected age, or age adjusted for gestation, is considered when assessing a **premature infant's growth and developmental milestones. The first hepatitis B vaccine is administered when the patient weighs ≥2 kg (4 lb 6 oz), typically at birth.**

Individuals **age ≥15 require 3 doses** of (HPV) vaccine to achieve immunity. In contrast, individuals **age <15, require only 2 doses** administered 6 months apart.

Chapter 17: Miscellaneous

Injury prevention in children	
Water	• **Never leave child unattended** around water. Fence all 4 sides of pool & install self-locking, self-latching gate. Use life jackets. Teach child to swim
Fire	• Install smoke detectors on every level of home • Test smoke detectors & change batteries regularly
Gun	• If present, store guns **unloaded** and locked • Store **ammunition separately** from guns
Home	• Lock medications & toxic household products **out of reach** [NOT open, elevated shelving because children can climb and access these items] • Remove dangling cords & cover power outlets • Install **safety gates on the top & bottom of stairs** • Set **water heater** to **maximum temperature of 49 C** (120 F) • **Mount furniture & TV to wall** • Avoid choking hazards (**whole grapes, raw vegetables, hard candy, uncut hot dogs, nuts, seeds, popcorn,** latex balloons, marbles, coins) until age 4

Whole grapes and hot dogs can be cut **lengthwise** so that they are **no longer cylindrical**. In addition, while eating, children should be **supervised and sitting upright**, avoiding activities such as **running or playing**.

(Choice A) Those age <4, particularly infants, can drown in <2 inches of water and should be within arm's reach when near water, including bathtubs, baby pools, or buckets.

(Choice B) Beginning with development of the first tooth, children should brush **twice a day** with a **small amount of fluoride-containing toothpaste.**

(Choice E) A **rear-facing car seat protects the head and spine** in a collision and should be used from birth **until a child reaches the maximum height or weight limit of the particular car seat.** The transition to a forward-facing car seat **should not occur before age 2,** and many children can remain rear-facing until age 4.

Car seats should never be placed in a front seat with an active airbag because the airbag's force can cause **head injury or death** upon deployment.

Reassurance should be provided to parents who are concerned about leg positioning in a tall child who is in a rear-facing car seat. Legs bent against the back seat are typically not dangerous or uncomfortable to the child, and protecting the brain and spine is the priority.

Intraosseous lines [proximal tibia MC] can be placed rapidly when emergency access is necessary and **peripheral access cannot be obtained**. Intraosseous access can be performed with less required skill and practice than central venous access.

Contraindications to IO placement: infection (cellulitis) overlying the access site, fracture or previous IO attempts in the chosen extremity, bone fragility (osteogenesis imperfecta).

Routine adolescent screening	
Category	**Screening method***
Mental health	• Validated depression questionnaire, **age ≥12**
Sexual health	• Confidential discussion about sexual activity • Gonorrhea & chlamydia testing if sexually active • HIV testing (**once** at age <18)
Substance use	• Confidential discussion about exposure, use, abuse
Dyslipidemia	• Lipid panel **once** between age 17-21 or children age 9-11 [because lipid levels are relatively stable just before and after puberty]
Safety	• Inquiry about bullying. nquiry about seatbelt & helmet use
*Annually, unless otherwise indicated.	

The adolescent well-child visit (around age 14) should include screening for **depression & substance use**, **healthcare maintenance**, **contraceptive counseling** (ideally before 1st sexual encounter) and discussion of **safe sex practices**.

Vanderbilt Assessment Scales: screen for ADHD in patients **with impaired functioning** due to hyperactivity or inattention. Routine ADHD screening is not recommended.

Chlamydia & gonorrhea in women (Dx: NAAT)	
Risk factors	• Age <25. High-risk sexual behavior

Manifestations	• Asymptomatic (MC). Urethritis. Cervicitis. Perihepatitis (Fitz-Hugh–Curtis syndrome)
Treatment	• Empiric: ceftriaxone + doxycycline* • Confirmed chlamydia: doxycycline* • Confirmed gonorrhea: ceftriaxone
Complications	• PID. Infertility. Ectopic pregnancy. Pharyngitis
*Azithromycin in pregnancy.	

Annual screening for **chlamydia and gonorrhea** is recommended in **sexually active women age <25**, as well as those with other risk factors (eg, **multiple sexual partners, illicit drug use**). Patients who test positive should receive antibiotics immediately and refrain from sexual intercourse until treatment is complete. **All sexual partners from the preceding 2 months should also be tested and treated.**

(Choice B) Electrocardiographic screening in asymptomatic children may be indicated if there is a significant family history of premature cardiac death (ie, at age <50).

(Choice D) Even if both parents have type 2 diabetes mellitus, routine screening is not indicated in children without other risk factors (eg, obesity).

Prader-Willi syndrome (the most common syndromic form of **obesity**)	
Clinical features	• Hyperphagia [age 1-6]/obesity. Intellectual disability. Hypogonadism. Hypotonia. Weak suck/feeding problems in infancy. Short stature [hypothalamic/pituitary dysfunction]. Behavioural difficulties (in relation to food seeking), developmental delay. **Dysmorphic facies**: Narrow forehead. Almond-shaped eyes. Downturned mouth
Diagnosis	• Loss of expression on paternal chromosome 15 (q11-q13)
Complications	• Sleep apnea (70%). Type 2 DM (25%). Gastric distension/rupture. Death by choking (8%)

THE END