

QUESTION

A 2-year-old infant presents with high conjugated (direct) bilirubin, dark coloured urine and clay-coloured stools. On examination, the infant is visibly jaundiced but is feeding normally. What is the most likely diagnosis?

A Biliary atresia

CORRECT

B Gilbert syndrome

C Crigler Najjar Syndrome

D Physiological jaundice

RIGHT ANSWER

OK A. Biliary atresia

WHY THIS IS RIGHT

A young child with conjugated (direct) hyperbilirubinemia, dark urine, and pale (clay-colored) stools is suggestive of obstructive cholestasis, most commonly due to biliary atresia.

- Conjugated hyperbilirubinemia always indicates pathological jaundice
- Pale stools occur due to absence of bile pigments in the intestine
- Dark urine is due to excretion of conjugated bilirubin
- Biliary atresia presents in early infancy with persistent jaundice and requires early surgical intervention (Kasai procedure)
- Conditions like Gilbert syndrome, Crigler-Najjar syndrome, and physiological jaundice cause unconjugated hyperbilirubinemia

QUESTION

A 6-month-old infant presents with sudden, brief spasms involving flexion of the neck, trunk, and limbs occurring in clusters. EEG reveals a hypsarrhythmia pattern. What is the first-line treatment for this condition?

A ACTH

CORRECT

B Valproate

C Ethosuximide

D Carbamazepine

RIGHT ANSWER

OK

A. ACTH

WHY THIS IS RIGHT

A 6-month-old infant with clustered flexor spasms and a hypsarrhythmia pattern on EEG is characteristic of infantile spasms (West syndrome). This is an age-specific epileptic encephalopathy requiring urgent treatment to prevent neurodevelopmental deterioration.

- Classic triad:
 - Infantile spasms (sudden flexion/extension movements in clusters)
 - Hypsarrhythmia on EEG (chaotic, high-amplitude disorganized pattern)
 - Developmental delay or regression
- Peak onset: 3-8 months of age
- First-line treatment: ACTH (or oral steroids); vigabatrin is preferred in tuberous sclerosis
- Drugs like valproate, ethosuximide, and carbamazepine are not effective for this condition

QUESTION

A newborn born to a diabetic mother, presents after 24 hours of life with a blood glucose level of 35 mg/dL. The baby is symptomatic. What is the next best step in management?

CORRECT

A

Administer IV glucose and recheck blood glucose after 20 minutes

B

Administer IV bolus of Hartmann solution

C

Recheck blood glucose after 20 minutes

D

Start oral feeds and observe

RIGHT ANSWER

OK

A. Administer IV glucose and recheck blood glucose after 20 minutes

WHY THIS IS RIGHT

Neonatal hypoglycemia is a medical emergency, especially in infants of diabetic mothers, and requires immediate correction to prevent neurological injury.

- Infants of diabetic mothers are at risk due to hyperinsulinemia
- Symptomatic hypoglycemia (e.g., lethargy, seizures, poor feeding) requires urgent treatment
- Initial management: IV bolus of glucose (10% dextrose, 2 ml/kg) followed by infusion

Clinical approach:

- Confirm hypoglycemia (<40-45 mg/dL depending on age)
- Give IV glucose immediately
- Recheck blood glucose after 15-20 minutes

Definition: Blood <40mg/dL/Plasma <45mg/dL

Asymptomatic: >20mg/dL - oral [BF] - >40mg/dL

└ <20mg/dL → 40mg/dL → Glycaemic infusion

Symptomatic: iv dextrose bolus 2mL/kg - infusion
[jitteriness; seizure; coma]

QUESTION

A previously healthy child presented with acute-onset dyspnea. A chest X-ray shows unilateral hyperinflation of the lungs. What is true for this patient?

CORRECT

A Focal area of decreased air entry will be suggestive of foreign body

B Flexible bronchoscopy used for removal

C In complete obstruction, ball and valve mechanism causes hyperinflation

D The child has developed acute laryngotracheobronchitis

RIGHT ANSWER

OK

A. Focal area of decreased air entry will be suggestive of foreign body

WHY THIS IS RIGHT

Foreign body aspiration should be suspected in a previously healthy child with sudden onset respiratory distress and unilateral hyperinflation on chest X-ray.

- Partial bronchial obstruction creates a “ball-valve” effect -> air enters but cannot exit -> unilateral hyperinflation
- Clinical sign: focal decreased air entry on the affected side is a key clue
- Most common in young children due to aspiration of small objects (e.g., peanuts)

Management:

- Rigid bronchoscopy is the procedure of choice for both diagnosis and removal
- Flexible bronchoscopy is mainly diagnostic, not preferred for removal

Why other options are incorrect:

- Ball-valve mechanism occurs in partial, not complete obstruction
- Laryngotracheobronchitis (croup) causes bilateral airway involvement, not unilateral hyperinflation

QUESTION

Which of the following children are considered at-risk babies?

1. Baby with a birth weight of 2.5 kg
2. Baby on artificial feeds
3. Baby of working mother/single parent
4. Baby with weight <85% of expected weight
5. Birth order of 3 or more

A 2, 3

CORRECT

B 1, 2, 3, 4

C 4, 5

D 1, 4

RIGHT ANSWER

OK

A. 2, 3

WHY THIS IS RIGHT

At-risk babies” are infants who have higher chances of morbidity and mortality due to biological or social risk factors, requiring closer monitoring.

- Social and feeding-related risks are important.
- Artificial feeding increases risk of infections and malnutrition
- Baby of a working mother or single parent may have inadequate care or feeding issues

Why other options are not included:

- Birth weight of 2.5 kg is normal (low birth weight is <2.5 kg)
- Weight <85% expected suggests malnutrition but applies to older infants/children, not specifically “at-risk baby” category
- Birth order alone is not a direct criterion unless associated with poor care

QUESTION

A child presented with a history of loose stools with an increase in frequency for 4 days. On examination, he is drowsy, unable to feed, and skin on pinching goes back very slowly. According to the integrated management of neonatal and childhood illness (IMNCI), this child will be classified as having:

A Mild dehydration

B Some dehydration

C Severe dehydration

CORRECT

D Moderate dehydration

RIGHT ANSWER

OK C. Severe dehydration

WHY THIS IS RIGHT

IMNCI classifies dehydration in children based on clinical signs into no dehydration, some dehydration, and severe dehydration to guide urgent management.

Parameters	No Dehydration	Some Dehydration
Appearance	Well, alert	Restless, irritable
Eyes	Normal	Sunken
Thirst	Drinks normally, not thirsty	Thirsty, drinks eagerly
Skin pinch	Goes back quickly (<1 second)	Goes back slowly (1 second)

Key concepts for severe dehydration:

- Lethargic or unconscious (drowsy child)
- Unable to drink or drinks poorly
- Skin pinch goes back very slowly

Clinical importance:

- Requires immediate IV fluid therapy (Plan C)
- Rapid recognition prevents shock and death

QUESTION

A 2-month-old infant born to an HIV-positive mother presents with recurrent diarrhea. What is the next best step?

A Test stool for giardia and give antibiotics

B Dried spot sample for HIV DNA PCR

CORRECT

C Antibody tests for HIV

D Aerobic culture

RIGHT ANSWER

OK

B. Dried spot sample for HIV DNA PCR

WHY THIS IS RIGHT

In infants born to HIV-positive mothers, diagnosis of HIV infection is made using virological tests (HIV DNA PCR), not antibody tests.

- Maternal IgG antibodies cross the placenta -> antibody tests remain positive up to 18 months
- Therefore, antibody-based tests are unreliable in infants
- Early infant diagnosis is done using HIV DNA PCR (often via dried blood spot)

Clinical importance:

- Allows early detection and timely initiation of antiretroviral therapy
- Improves survival and reduces disease progression

QUESTION

An 8-day-old newborn was found to have thyroid-stimulating hormone level of more than 100 mIU/L. Which of the following will be the next best investigation?

- A Urine iodine excretion
- B Serum thyroid receptor antibody

C Radiotracer uptake with technetium

CORRECT

- D Perchlorate secretion test

RIGHT ANSWER

C. Radiotracer uptake with technetium

WHY THIS IS RIGHT

A markedly elevated TSH (>100 mIU/L) in a neonate strongly suggests congenital hypothyroidism and requires prompt evaluation of thyroid gland anatomy and function.

- Most common causes: thyroid dysgenesis (agenesis, ectopia) or dyshormonogenesis
- Next step after diagnosis is imaging to determine etiology
- Radionuclide scan (technetium or radioiodine uptake) helps identify:
 - Absent gland (agenesis)
 - Ectopic thyroid
 - Normal gland with impaired hormone synthesis

Clinical importance:

- Treatment with levothyroxine should be started immediately without waiting for investigations
- Early treatment prevents irreversible neurodevelopmental delay

QUESTION

A 1-day-old neonate has not passed urine since birth. What is the next step in management?

CORRECT

A Continue breastfeeding and observe

B Admit to NICU

C Start artificial feeding

D Start intravenous fluids

RIGHT ANSWER

OK **A. Continue breastfeeding and observe**

WHY THIS IS RIGHT

In healthy term neonates, delayed passage of urine in the first 24 hours can be a normal finding and does not always indicate pathology.

- Most neonates void within the first 24 hours, but some may pass urine slightly later
- If the baby is otherwise well (feeding, active, normal exam), observation is appropriate
- Continue breastfeeding to ensure adequate hydration

When to investigate:

- No urine after 48 hours
- Associated signs: poor feeding, abdominal distension, vomiting, or suspected urinary obstruction

QUESTION

An infant is brought to OPD with hepatosplenomegaly and thrombocytopenia. Neuroimaging with CT reveals periventricular calcifications. What is the most likely diagnosis?

- A** Congenital rubella syndrome
- B** Congenital herpes simplex virus infection
- C** Congenital toxoplasmosis
- D** Congenital cytomegalovirus infection

CORRECT

RIGHT ANSWER

OK

D. Congenital cytomegalovirus infection

WHY THIS IS RIGHT

Congenital cytomegalovirus (CMV) infection is the most common congenital infection and is characterized by periventricular intracranial calcifications along with systemic features.

Classic features:

- Periventricular calcifications (hallmark)
- Hepatosplenomegaly
- Thrombocytopenia (-> petechiae, "blueberry muffin" rash)
- Sensorineural hearing loss

Differentiation:

- Toxoplasmosis: diffuse intracranial calcifications, hydrocephalus, chorioretinitis
- Rubella: cataracts, PDA, deafness
- HSV: encephalitis with temporal lobe involvement



QUESTION

A 3-month-old baby is brought to OPD with jaundice and clay-colored stools. Lab work up reveals that the baby has conjugated hyperbilirubinemia. A liver biopsy was done and shows periductal proliferation. What is the most likely diagnosis?

A Crigler-Najjar syndrome

B Rotor syndrome

C Dubin-Johnson syndrome

D Biliary atresia

CORRECT

RIGHT ANSWER

OK

D. Biliary atresia

WHY THIS IS RIGHT

Biliary atresia is a cause of neonatal cholestasis characterized by progressive obliteration of the extrahepatic bile ducts, leading to conjugated hyperbilirubinemia.

- Presents in early infancy with persistent jaundice, clay-colored (acholic) stools, and dark urine
- Lab findings: conjugated hyperbilirubinemia
- Liver biopsy shows bile duct proliferation, portal fibrosis, and bile plugs

Clinical importance:

- Requires early diagnosis and intervention
- Kasai portoenterostomy should be performed ideally before 6-8 weeks for best outcomes

Differentiation:

- Crigler-Najjar: unconjugated hyperbilirubinemia
- Dubin-Johnson & Rotor: conjugated hyperbilirubinemia but no obstructive features like pale stools

QUESTION

A 3-month-old baby comes with complaints of deafness, cataract, and patent ductus arteriosus. Which of the following is the most likely diagnosis?

A Congenital herpes simplex virus infection

B Congenital toxoplasmosis

C Congenital cytomegalovirus infection

D Congenital rubella syndrome

CORRECT

RIGHT ANSWER

OK

D. Congenital rubella syndrome

WHY THIS IS RIGHT

Congenital rubella syndrome (CRS) is characterized by a classic triad of sensorineural deafness, cataracts, and congenital heart defects (most commonly patent ductus arteriosus).

- Caused by transplacental infection with rubella virus, especially during the first trimester

Classic triad:

- Sensorineural hearing loss (most common)
- Eye defects (cataracts, retinopathy)
- Cardiac defects (PDA, pulmonary artery stenosis)

Other features:

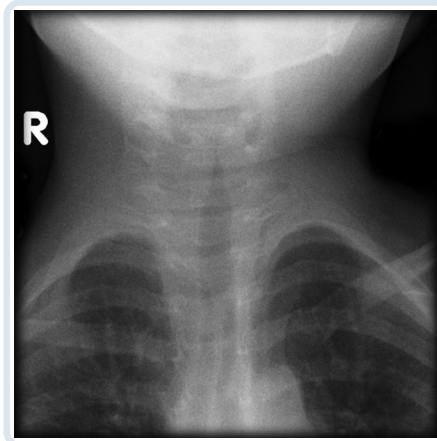
- Blueberry muffin rash
- Microcephaly, developmental delay
- Hepatosplenomegaly

Differentiation:

- CMV: periventricular calcifications, hearing loss
- Toxoplasmosis: hydrocephalus, chorioretinitis, diffuse calcifications
- HSV: skin vesicles, encephalitis

QUESTION

A child is brought to the hospital with features of respiratory distress and biphasic stridor. There is a history of upper respiratory infection. The radiograph is shown below. Mark the correct diagnosis?



- ☐ A Acute epiglottitis
- ☒ B Acute laryngotracheobronchitis
- ☐ C Foreign body in glottis
- ☐ D Laryngomalacia

CORRECT

RIGHT ANSWER

- ☒ OK B. Acute laryngotracheobronchitis

WHY THIS IS RIGHT

Acute laryngotracheobronchitis (croup) is a common cause of upper airway obstruction in children, typically following a viral upper respiratory infection.

- Presents with barking cough, hoarseness, and inspiratory or biphasic stridor
- X-ray (AP neck) shows “steeple sign” due to subglottic narrowing
- Most commonly caused by parainfluenza virus

Management:

- Mild: humidified air
- Moderate to severe: nebulized adrenaline and corticosteroids

Differentiation:

- Epiglottitis: thumb sign on lateral X-ray, toxic child, drooling
- Foreign body: sudden onset without preceding URI
- Laryngomalacia: chronic stridor since infancy

QUESTION

A woman came to OPD with a newborn who has complaints of chest retractions, dyspnea and lethargy. The paediatrician diagnosed the baby with respiratory distress syndrome. This occurs due to the deficiency of:

- A Dipalmitoyl inositol
- B Lecithin**
- C Sphingomyelin
- D Dipalmitoyl phosphatidylethanolamine

CORRECT

RIGHT ANSWER

OK **B. Lecithin**

WHY THIS IS RIGHT

Neonatal respiratory distress syndrome (hyaline membrane disease) is caused by deficiency of pulmonary surfactant, particularly dipalmitoyl phosphatidylcholine (lecithin), produced by type II pneumocytes.

- Lecithin is the major component of surfactant responsible for reducing alveolar surface tension
- Deficiency leads to alveolar collapse (atelectasis), decreased lung compliance, and respiratory distress
- Common in preterm infants due to immature lungs

Clinical relevance:

- L/S ratio (lecithin:sphingomyelin) is used to assess fetal lung maturity
- Antenatal corticosteroids increase surfactant production

QUESTION

A mother delivers a 2.5 kg baby with normal vaginal delivery in a rural area. Which of the following is an incorrect statement?

A Exclusive breastfeeding

B Bath the baby with warm water immediately

CORRECT

C Clean the eyes

D Skin to skin contact

RIGHT ANSWER

OK B. Bath the baby with warm water immediately

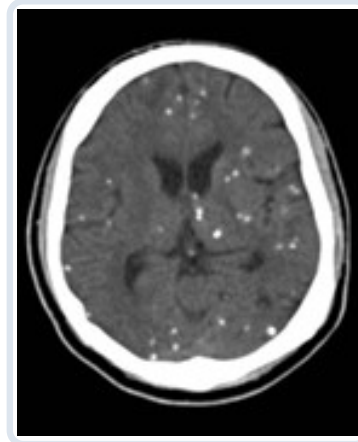
WHY THIS IS RIGHT

Essential newborn care focuses on thermal protection, early breastfeeding, and infection prevention.

- Immediate bathing should be avoided because it can cause hypothermia; bathing is delayed for at least 24 hours
- Early skin-to-skin contact (kangaroo care) helps maintain temperature and promotes bonding
- Exclusive breastfeeding should be initiated within the first hour of life
- Eye care helps prevent neonatal infections

QUESTION

A 3-month-old baby with intracranial diffuse calcifications, chorioretinitis, and hydrocephalus. What is the most likely diagnosis?



A Toxoplasmosis

CORRECT

B CMV

C Zika virus

D Rubella

RIGHT ANSWER

OK

A. Toxoplasmosis

WHY THIS IS RIGHT

Congenital toxoplasmosis classically presents with the triad of chorioretinitis, hydrocephalus, and intracranial calcifications.

- Caused by transplacental infection with *Toxoplasma gondii*
- Intracranial calcifications are typically diffuse (not periventricular)
- Associated features include seizures, developmental delay, and hepatosplenomegaly

Differentiation:

- CMV: periventricular calcifications
- Zika virus: microcephaly with calcifications
- Rubella: cataracts, cardiac defects, deafness

QUESTION

An HIV-positive mother with a viral load of 1200 copies/mL delivers a newborn baby. What is the most appropriate antiretroviral prophylaxis for the newborn?

A Nevirapine for 6 weeks

B Nevirapine + Zidovudine for 12 weeks

CORRECT

C Nevirapine for 12 weeks

D Zidovudine for 4 weeks

RIGHT ANSWER

OK B. Nevirapine + Zidovudine for 12 weeks

WHY THIS IS RIGHT

In infants born to HIV-positive mothers, the choice and duration of antiretroviral prophylaxis depend on the risk of transmission, primarily determined by maternal viral load.

- High-risk exposure (maternal viral load >1000 copies/mL or unknown) requires combination prophylaxis
- Recommended regimen: dual therapy with Nevirapine + Zidovudine for extended duration (e.g., 12 weeks)
- Low-risk exposure (viral load <1000 copies/mL, good ART adherence) may receive single-drug prophylaxis

Clinical importance:

- Early initiation (within hours of birth) significantly reduces vertical transmission
- Adherence to prophylaxis and follow-up testing is essential

QUESTION

A 6-month-old infant presents with recurrent infections and failure to thrive. Laboratory investigations reveal a deficiency of adenosine deaminase (ADA). Which of the following immunodeficiency disorders is most likely associated with this condition?

A Severe Combined Immunodeficiency (SCID)

CORRECT

B Hypogammaglobulinemia

C DiGeorge Syndrome

D Wiskott-Aldrich Syndrome

RIGHT ANSWER

OK

A. Severe Combined Immunodeficiency (SCID)

WHY THIS IS RIGHT

Adenosine deaminase (ADA) deficiency is a cause of severe combined immunodeficiency (SCID), a life-threatening primary immunodeficiency characterized by defective T-cell and B-cell function.

- ADA deficiency leads to accumulation of toxic metabolites (deoxyadenosine) -> lymphocyte destruction
- Results in profound deficiency of both cellular (T-cell) and humoral (B-cell) immunity
- Presents in infancy with recurrent severe infections, failure to thrive, and opportunistic infections
- Requires urgent management such as hematopoietic stem cell transplantation
- Enzyme replacement therapy and gene therapy are also options

Contrast:

- Hypogammaglobulinemia: primarily B-cell defect
- DiGeorge syndrome: thymic aplasia -> T-cell deficiency
- Wiskott-Aldrich syndrome: triad of eczema, thrombocytopenia, and recurrent infections

QUESTION

At 6 weeks of life, a child developed a fever after DPT vaccination. And now child is 10 weeks due for vaccination. What should be done next?

- A** Give DPT Vaccination
- B** Give DT
- C** Don't give DPT
- D** Don't give any vaccination

CORRECT

RIGHT ANSWER

- OK** **A. Give DPT Vaccination**

WHY THIS IS RIGHT

Mild adverse events following immunization, such as low-grade fever after DPT vaccination, are common and not a contraindication to subsequent doses.

- DPT vaccine can cause minor reactions like fever, irritability, and local pain
- These reactions do not warrant withholding or changing future doses
- True contraindications include severe allergic reaction (anaphylaxis) or encephalopathy after a previous dose

Clinical application:

- The child should continue the routine immunization schedule
- Subsequent doses of DPT should be given as planned

QUESTION

INSURE technique is used in which condition?

- A** Meconium aspiration syndrome
- B** Transient tachypnea of newborn
- C** **Hyaline membrane disease**
- D** Neonatal jaundice

CORRECT

RIGHT ANSWER

- OK** **C. Hyaline membrane disease**

WHY THIS IS RIGHT

The INSURE technique (Intubation-Surfactant-Extubation) is used in preterm neonates with hyaline membrane disease (neonatal respiratory distress syndrome) to deliver surfactant while minimizing duration of mechanical ventilation.

- Indicated in surfactant deficiency seen in preterm infants
- Procedure: brief intubation -> administer exogenous surfactant -> early extubation to CPAP
- Reduces complications of prolonged ventilation such as bronchopulmonary dysplasia

Clinical significance:

- Improves oxygenation and lung compliance
- Decreases need for prolonged mechanical ventilation

QUESTION

| All of the following are true about neonatal cholestasis, except?

A Biliary atresia is the most common cause of neonatal cholestasis

B Kasai procedure should be done within 90 days

CORRECT

C HIDA scan is the best investigation for diagnosis

D Can be differentiated from other differential diagnoses by biopsy

RIGHT ANSWER

OK **B. Kasai procedure should be done within 90 days**

WHY THIS IS RIGHT

Neonatal cholestasis is defined as conjugated hyperbilirubinemia in early infancy and requires prompt evaluation, with biliary atresia being the most important cause to rule out.

- Early diagnosis is critical because Kasai portoenterostomy is most effective when performed before 6-8 weeks of age (60 days is the usual cut-off)
- HIDA scan helps assess bile flow but is not definitive
- Liver biopsy is a key investigation to differentiate biliary atresia from other causes (e.g., neonatal hepatitis)

QUESTION

Which among the following is false about breast milk jaundice:

- a. Breast feeding should be completely stopped
- b. Unconjugated hyperbilirubinemia.
- c. Phototherapy is required to treat most cases
- d. Bilirubin levels are usually more than 10 mg/dL at 3-4 weeks

A a, c and d

B a and c

CORRECT

C a, b and d

D a and d

RIGHT ANSWER

OK B. a and c

WHY THIS IS RIGHT

Breast milk jaundice is a benign, late-onset unconjugated hyperbilirubinemia seen in otherwise healthy, thriving breastfed infants and usually does not require interruption of breastfeeding or aggressive treatment.

- Occurs after the first week of life and may persist up to 3-12 weeks
- Caused by increased enterohepatic circulation due to beta-glucuronidase in breast milk
- Infant is clinically well with normal feeding and weight gain
- Bilirubin levels may remain mildly elevated (often >10 mg/dL) during 3-4 weeks

Why statements a and c are false:

- Breastfeeding should not be completely stopped; it is usually continued
- Phototherapy is not required in most cases unless bilirubin reaches treatment thresholds

QUESTION

| Which of the following statements regarding breastfeeding technique is/are correct?

- A** The infant's lower lip should be inverted
- B** The infant's cheeks should not appear full during feeding
- C** Upper areola should be covered more than lower
- D** The infant's chin should touch the breast

CORRECT

RIGHT ANSWER

- OK** D. The infant's chin should touch the breast

WHY THIS IS RIGHT

Correct breastfeeding technique (proper latch) is essential for effective milk transfer and prevention of maternal nipple problems.

Key features of proper latch:

- Infant's chin should touch the breast
- Mouth wide open with lips everted (not inverted)
- More of the lower areola is inside the mouth than the upper
- Cheeks appear full (not sunken) during feeding

Why others are incorrect:

- Lower lip should be everted, not inverted
- Cheeks should be full, not sunken
- More lower areola (not upper) should be covered

QUESTION

A 3-month-old infant presents with prolonged jaundice, dark urine, and pale stools. The child is exclusively breastfed and otherwise well. HIDA scan is non-excretory. What is the next best step in management?

- A** Rule out galactosemia using enzyme assay.
- B** The child has breast milk jaundice and start formula feeding after 3 weeks.
- C** Immediate liver biopsy to confirm diagnosis. CORRECT
- D** Perform a hepatic ultrasound to look for causes like biliary atresia.

RIGHT ANSWER

- OK** **C. Immediate liver biopsy to confirm diagnosis.**

WHY THIS IS RIGHT

Biliary atresia should be strongly suspected in an infant with prolonged jaundice, pale (acholic) stools, dark urine, and a non-excretory HIDA scan. The next best step is liver biopsy to confirm the diagnosis and differentiate it from other causes of neonatal cholestasis.

- Conjugated hyperbilirubinemia presenting after 2 weeks of life is always pathological
- Pale stools and dark urine indicate obstructive cholestasis
- Non-excretion on HIDA scan suggests biliary obstruction but is not definitive
- Liver biopsy shows bile duct proliferation, portal fibrosis, and bile plugs, helping confirm biliary atresia

Why other options are incorrect:

- Galactosemia presents with systemic illness (vomiting, hypoglycemia, sepsis), not isolated cholestasis
- Breast milk jaundice causes unconjugated hyperbilirubinemia with normal stool and urine color
- Ultrasound is useful but not the next best step after a non-excretory HIDA; biopsy provides definitive diagnosis

QUESTION

A term neonate delivered by cesarean section develops tachypnea and mild respiratory distress shortly after birth. Chest x-ray shows prominent vascular markings with fluid in the interlobar fissure and a normal cardiac shadow. The symptoms dissolve completely within 48 hours. What is the likely diagnosis?

- A RDS
- B Meconium aspiration syndrome
- C Pneumonia

D Transient tachypnea of newborn

CORRECT

RIGHT ANSWER

D. Transient tachypnea of newborn

WHY THIS IS RIGHT

The source quiz contains the placeholder “\$19” here and does not include a written explanation for this item.

QUESTION

A fetus with intrauterine growth restriction was born prematurely with jaundice, hepatosplenomegaly, microcephaly, and diffuse petechiae at birth. A brain CT was done which is shown below. What is the best method for the diagnosis of the etiological agent?



A Urine examination

CORRECT

B Liver biopsy

C Blood examination

D CSF examination

RIGHT ANSWER

OK **A. Urine examination**

WHY THIS IS RIGHT

The brain CT shows periventricular calcifications, which are characteristic of congenital cytomegalovirus (CMV) infection. CMV is a common cause of intrauterine infection leading to IUGR, microcephaly, hepatosplenomegaly, jaundice, and petechiae at birth. The best method for diagnosing congenital CMV infection is by detecting CMV in the urine within the first 2 to 3 weeks of life. Urinary examination detects CMV viral DNA or pp65 antigen, which is a reliable marker for infection.

Why not other options:

- B. Liver biopsy: Invasive and not required for diagnosing congenital CMV.
- C. Blood examination: CMV may be detected, but urine PCR/culture is more sensitive.
- D. CSF examination: Used if CNS infection suspected, but not the best diagnostic test.

QUESTION

A 10-year-old boy is brought to the physician with ataxia, myoclonus, and visual problems. His parents say that he began acting strangely and having difficulty with school work several months ago. After the appropriate workup, a brain biopsy is obtained and an RNA virus containing hemagglutinin is cultured from the tissue sample. Which of the following is the most likely diagnosis?

- A** Guillain-Barre syndrome
- B** Multiple sclerosis
- C** Progressive multifocal leukoencephalopathy
- D** Subacute sclerosing panencephalitis

CORRECT

RIGHT ANSWER

- OK** **D. Subacute sclerosing panencephalitis**

WHY THIS IS RIGHT

Subacute sclerosing panencephalitis (SSPE) is a chronic progressive neurodegenerative disease caused by persistent infection with a mutated measles virus in the brain.

- Occurs years after measles infection
- Progressive cognitive decline
- Behavioral changes
- Myoclonus
- Ataxia and visual disturbances

Measles virus is an RNA virus with hemagglutinin (H) glycoprotein, which helps the virus attach to host cells. Persistent viral infection leads to progressive inflammation and demyelination of the brain.

QUESTION

TIM1 and TAM-XL mutations are associated with:

- A** Turner syndrome
- B** Noonan syndrome
- C** Congenital Zika virus infection
- D** Congenital CMV infection

CORRECT

RIGHT ANSWER

- OK** C. Congenital Zika virus infection

WHY THIS IS RIGHT

TIM1 (T-cell immunoglobulin mucin-1) and TAM receptors (Tyr03, AXL, Mer), particularly AXL, are host cell surface receptors that facilitate entry of certain viruses into cells.

In Zika virus infection:

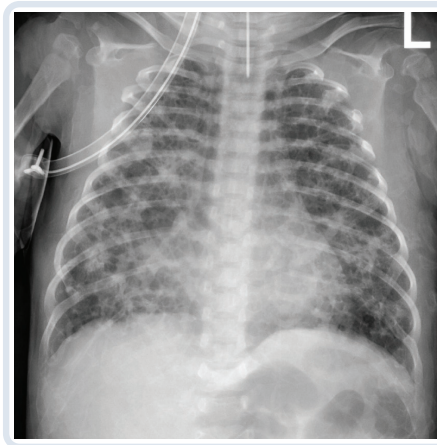
- The virus uses AXL receptors on neural progenitor cells to enter the cells.
- Infection of these developing brain cells leads to impaired neurodevelopment.

Clinical consequence in congenital infection:

- Microcephaly
- Intracranial calcifications
- Developmental delay
- Ocular abnormalities

QUESTION

A 7-week-old boy is evaluated for persistent hypoxia. He has been in the neonatal intensive care unit since birth. He was born at 29 weeks gestation due to placental abruption and weighed 1150g. Six hours after delivery, the patient developed tachypnoea, grunting, and cyanosis, a chest x-ray revealed ground-glass opacities. Surfactant was administered and mechanical ventilation was initiated. He remained on mechanical ventilation for the first 3 weeks of life. Ever since ventilation was discontinued, the patient has remained hypoxic and continues to require oxygen administration by nasal cannula. Chest radiograph is shown below. What is the most likely diagnosis for this patient's current presentation?



- A** Bronchiectasis
- B** Bronchopulmonary dysplasia
- C** Neonatal pneumonia
- D** Persistent pulmonary hypertension

CORRECT

RIGHT ANSWER

- OK** **B. Bronchopulmonary dysplasia**

WHY THIS IS RIGHT

Bronchopulmonary dysplasia (BPD) is a chronic lung disease seen in premature infants who require prolonged oxygen therapy and mechanical ventilation, usually after neonatal respiratory distress syndrome (RDS). Leads to lung injury, inflammation, and impaired alveolar development

Chest X-ray findings:

- Patchy atelectasis
- Hyperinflation
- Cystic changes (“sponge-like” or bubbly lungs)

Why not others:

- A. Bronchiectasis: Chronic disease in older children, not premature neonates.
- C. Neonatal pneumonia: Acute infection, occurs early after birth, not persistent weeks later.
- D. PPHN: Immediate neonatal hypoxia, not after prolonged ventilation.

QUESTION

A 35-year-old primigravida comes to the labor and delivery unit at 36 weeks gestation with abdominal pain and blood-tinged vaginal discharge. She was diagnosed with gestational diabetes mellitus at 27 weeks but has not been compliant with diet or insulin therapy. Her family history is significant for type 2 diabetes mellitus, hypertension, coronary artery disease, and stroke in both maternal and paternal relatives. Monitoring shows regular uterine contractions and a normal fetal heart rate. Blood pressure is 135/86 mm Hg and pulse is 100/min. BMI is 40 kg/m². Cervical examination shows 6 cm dilation and 100% effacement. Her blood glucose at admission is 290 mg/dL and is started on insulin. The patient has a spontaneous vaginal delivery of a boy. Which of the following examination findings would most likely be present in this neonate?

A Heart murmur

B Limb defects

C Respiratory distress

CORRECT

D Small for gestational age

RIGHT ANSWER

OK C. Respiratory distress

WHY THIS IS RIGHT

Infants born to mothers with poorly controlled diabetes (especially gestational diabetes) are at increased risk of neonatal respiratory distress syndrome (RDS).

Mechanism:

- Maternal hyperglycemia → fetal hyperglycemia → fetal hyperinsulinemia.
- High insulin levels inhibit surfactant production by type II pneumocytes.
- This results in delayed lung maturation, increasing the risk of RDS, even in late preterm or term infants.

Other common findings in infants of diabetic mothers:

- Macrosomia
- Neonatal hypoglycemia
- Polycythemia
- Hypertrophic cardiomyopathy

QUESTION

| Which of the following is not true about breast milk jaundice?

- A** Seen in 3-4% of exclusively breastfed children
- B** Phototherapy is rarely needed
- C** Exclusive breastfeeding with total bilirubin more than 10 mg/dl after 3-4 weeks of birth
- D** **Conjugated hyperbilirubinemia** CORRECT

RIGHT ANSWER

- OK** **D. Conjugated hyperbilirubinemia**

WHY THIS IS RIGHT

Breast milk jaundice is a form of prolonged unconjugated hyperbilirubinemia seen in otherwise healthy exclusively breastfed infants, usually appearing after the first week of life and may persist for several weeks.

Key features:

- Occurs in about 3-4% of exclusively breastfed infants.
- Caused by substances in breast milk (e.g., β -glucuronidase) that increase enterohepatic circulation of bilirubin.
- Infants are well, feeding normally, and gaining weight.
- Phototherapy is rarely required because bilirubin levels are usually mild to moderate.

QUESTION

| Which of the following is not an indicator of fetal lung maturity?

- A Lecithin-sphingomyelin ratio > 2.0
- B Positive shake test
- C Increased phosphatidyl glycerol
- D Nile blue test showing >50% of blue cells**

CORRECT

RIGHT ANSWER

OK

D. Nile blue test showing >50% of blue cells

WHY THIS IS RIGHT

Assessment of fetal lung maturity is important to determine the risk of neonatal respiratory distress syndrome (RDS) before delivery.

Common indicators of fetal lung maturity include:

- Lecithin-sphingomyelin (L/S) ratio > 2.0 -> indicates adequate surfactant production.
- Positive shake (foam stability) test -> presence of sufficient surfactant in amniotic fluid.
- Presence or increased levels of phosphatidylglycerol -> a late surfactant component indicating mature lungs and low risk of RDS.

The Nile blue sulfate test assesses the percentage of orange-staining fat cells in amniotic fluid, reflecting fetal skin maturity, not lung maturity.

QUESTION

A 4-day old baby is active and feeding well. He has yellowish staining of face and upper trunk. What is the next step in the management of the baby?

- A Exchange transfusion
- B Stop breastfeeding and start phototherapy
- C IV fluids and phototherapy

D Continue breastfeeding

CORRECT

RIGHT ANSWER

D. Continue breastfeeding

WHY THIS IS RIGHT

Physiological jaundice is common in healthy newborns and usually appears after 24 hours of life, peaks around day 3-5 and resolves spontaneously.

Typical features include:

- Mild jaundice involving the face and upper trunk
- Infant is active and feeding well
- Unconjugated hyperbilirubinemia within safe limits

Management in such cases is reassurance and continuation of breastfeeding, as frequent feeds help increase bilirubin excretion through stool.

QUESTION

A 18-year-old boy is brought to the clinic due to fatigue, malaise, fever, and sore throat for the past 6 days. He does not use tobacco, alcohol, or illicit drugs. Temperature is 38.3°C (101°F), blood pressure is 115/70 mmHg, pulse is 90/min, and respirations are 18/min. Physical examination shows pharyngeal hyperaemia with exudates along with cervical and axillary lymphadenopathy. Breath sounds are normal. Mild hepatosplenomegaly is present. Joints have a normal range of motion. There is no skin rash. A rapid streptococcal antigen test is negative. Which of the following is the most appropriate management for this patient?

- A Administer corticosteroids
- B Initiate antibacterial therapy
- C Prescribe antiviral treatment

D Refrain from sports for at least 3-4 weeks

CORRECT

RIGHT ANSWER

OK D. Refrain from sports for at least 3-4 weeks

WHY THIS IS RIGHT

The patient's symptoms of fever, sore throat with exudates, lymphadenopathy, and hepatosplenomegaly are characteristic of infectious mononucleosis, most commonly caused by Epstein-Barr virus (EBV). A major complication of infectious mononucleosis is splenic enlargement, which increases the risk of splenic rupture, especially with physical trauma. Therefore, patients are advised to avoid contact sports and strenuous physical activity for at least 3-4 weeks or until splenomegaly resolves.

QUESTION

A newborn girl is evaluated in the delivery room for respiratory distress. The patient was born 15 minutes ago by spontaneous vaginal delivery at 37 weeks gestation to a mother who received no prenatal care. Temperature is 37°C (98.6° F), pulse is 176/min, and respirations are 70/min. Pulse oximetry is 82% on room air. Physical examination shows grunting; subcostal and suprasternal retractions; and cyanosis of the lips and tongue. The chest appears rounded and the abdomen appears flat. Auscultation reveals absent breath sounds on the left and clear breath sounds on the right. Heart sounds are loudest in the right chest. Which of the following is the best next step in management of this patient?

- A Chest tube placement
- B Endotracheal intubation**
- C Pericardiocentesis
- D Prostaglandin E1 administration

CORRECT

RIGHT ANSWER

- B. Endotracheal intubation**

WHY THIS IS RIGHT

This neonate with severe respiratory distress, absent breath sounds on the left side, mediastinal shift (heart sounds louder on the right), and a scaphoid/flat abdomen is most consistent with congenital diaphragmatic hernia (CDH).

- A defect in the diaphragm allows abdominal organs to herniate into the thoracic cavity.
- This compresses the lungs and causes pulmonary hypoplasia and severe respiratory distress immediately after birth.

Immediate management:

- Endotracheal intubation with mechanical ventilation to stabilize the airway and oxygenation.
- Bag-mask ventilation must be avoided because it can inflate the stomach and intestines, worsening lung compression.

QUESTION

A 4-day-old, full-term boy is brought to the OPD for his first visit after an uncomplicated vaginal delivery. The patient's mother, gravida 1 para 1, has blood type A positive. Birth weight was 3.4 kg and length was 48.5 cm. He has been exclusively breastfed since birth and nurses for 10 minutes on each breast every 4 hours. The infant passed several dark brown, sticky, meconium stools during the first 2 days of life, but his last stool was yesterday and dark green. He has 2 wet diapers each day but has not voided today. The neonate's current weight is 3. He has scleral icterus and jaundice of the face, chest, and abdomen. The rest of his physical examination is normal. Laboratory results are as follows:

Total bilirubin: 14 mg/dL

Direct bilirubin: 0.9 mg/dL

Which of the following is the most likely cause of this infant's hyperbilirubinemia?

A Alloimmune hemolytic disease

B Biliary atresia

C Breast milk jaundice

D Breastfeeding jaundice

CORRECT

RIGHT ANSWER

OK

D. Breastfeeding jaundice

WHY THIS IS RIGHT

Breastfeeding jaundice occurs in the first week of life due to inadequate breast milk intake, leading to dehydration and decreased bilirubin excretion.

Key features in this case:

- Day 4 of life (early onset)
- Poor feeding frequency (every 4 hours)
- Few wet diapers and decreased urine output -> indicates dehydration
- Weight loss
- Unconjugated hyperbilirubinemia (total bilirubin elevated, direct bilirubin normal)

Mechanism:

- Inadequate milk intake -> reduced intestinal motility
- Increased enterohepatic circulation of bilirubin
- Leads to indirect (unconjugated) hyperbilirubinemia

QUESTION

A 10-day-old neonate with hypoxic-ischemic encephalopathy (HIE) is hyperalert and shows hyperactive tendon reflexes, mydriatic pupils, clonus. Based on the clinical findings, what stage of HIE does this neonate currently have?

A Stage 2

B Stage 1

CORRECT

C Stage 4

D Stage 3

RIGHT ANSWER

OK

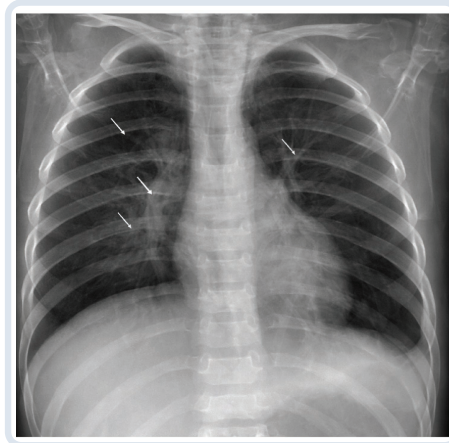
B. Stage 1

WHY THIS IS RIGHT

The source quiz contains the placeholder “\$1a” here and does not include a written explanation for this item.

QUESTION

A 10-month-old infant presents to the OPD with a 3-day history of lower respiratory tract symptoms accompanied by expiratory wheezing. A chest X-ray is shown below. Which of the following statements is incorrect regarding this condition?



A The mainstay of treatment is antibiotics

CORRECT

B The risk increases in infants born to young mothers or those exposed to maternal smoking during pregnancy

C There is a greater risk of asthma

D Breastfeeding is protective

RIGHT ANSWER

OK

A. The mainstay of treatment is antibiotics

WHY THIS IS RIGHT

The clinical picture (infant with URI symptoms, expiratory wheeze, and hyperinflation on chest X-ray) is typical of bronchiolitis, most commonly caused by respiratory syncytial virus (RSV). It is a viral lower respiratory tract infection affecting infants, especially <2 years of age. Chest X-ray often shows hyperinflation, peribronchovascular thickening, and patchy atelectasis. Management is mainly supportive, including oxygen, hydration, and nasal suctioning.

Important facts:

- Antibiotics are not the mainstay of treatment because the disease is viral.
- Risk factors include maternal smoking, young maternal age, prematurity, and crowded living conditions.
- Infants with bronchiolitis may have a higher risk of recurrent wheezing or asthma later in childhood.
- Breastfeeding is protective due to passive immunity.

QUESTION

A newborn boy is brought to the nursery for evaluation after delivery. The mother received no prenatal care but reports that the pregnancy was uncomplicated and she was healthy. The infant was born via spontaneous vaginal delivery and required no resuscitation. On examination, the infant is below the 3rd percentile for weight, 25th percentile for length, and 50th percentile for head circumference.

Hepatosplenomegaly is present on examination. Over the next 48 hours, the infant develops jaundice, clear rhinorrhea, and a maculopapular rash on the feet and buttocks that later desquamates. Which of the following congenital infections is most likely in this patient?

A Cytomegalovirus

B HIV

C Rubella

D Syphilis

CORRECT

RIGHT ANSWER

OK D. Syphilis

WHY THIS IS RIGHT

Congenital syphilis results from transplacental transmission of *Treponema pallidum* from an infected mother to the fetus. Typical early features (within the first weeks of life) include:

- Intrauterine growth restriction (IUGR)
- Hepatosplenomegaly
- Jaundice
- "Snuffles" (profuse mucopurulent rhinorrhea) due to nasal mucosal inflammation
- Maculopapular rash involving palms, soles, buttocks, often followed by desquamation

These findings are characteristic of early congenital syphilis.

QUESTION

A 2-hour-old boy is evaluated in the newborn nursery due to cyanosis. The boy was born at 38 weeks gestation via spontaneous vaginal delivery to a primigravida mother whose pregnancy and delivery were uncomplicated. When the mother attempted to breastfeed in the first hour of life, the patient latched and sucked well but began to appear blue around the lips and face. He began crying and turned pink when he was removed from the breast. He has voided and passed meconium. Heart rate and 4-extremity blood pressures are normal. Examination shows a non-dysmorphic neonate with mild cyanosis at rest. Which of the following is the most likely diagnosis in this patient?

A Choanal atresia

CORRECT

B Laryngomalacia

C Tetralogy of Fallot

D Tracheoesophageal fistula with esophageal atresia

RIGHT ANSWER

OK **A. Choanal atresia**

WHY THIS IS RIGHT

Newborns are obligate nasal breathers during the first few months of life. Therefore, nasal obstruction can cause cyanosis that worsens during feeding and improves when the infant cries.

In choanal atresia, the posterior nasal choanae are blocked (by bone or membrane), preventing airflow through the nose.

- During feeding: the infant attempts to breathe through the nose while sucking -> cyanosis develops.
- During crying: the mouth opens, allowing oral breathing -> cyanosis improves.

This pattern of cyclical cyanosis relieved by crying is characteristic of choanal atresia.

QUESTION

A 22-year-old primigravida develops fever and pruritic vesicular rash one day after delivering a full-term baby. The mother is diagnosed with varicella infection. What is the appropriate management for the neonate?

A Acyclovir therapy should be started immediately

B Wait and monitor the neonate for one week

C Administer varicella-zoster immunoglobulin

CORRECT

D Immediate vaccination against varicella

RIGHT ANSWER

OK **C. Administer varicella-zoster immunoglobulin**

WHY THIS IS RIGHT

If a mother develops varicella infection from 5 days before delivery to 2 days after delivery, the newborn is at high risk of severe neonatal varicella because transplacental maternal antibodies have not yet been transferred.

In this situation, the recommended management for the neonate is administration of varicella-zoster immunoglobulin (VZIG) to provide passive immunity and reduce the severity of infection.

Why other options are incorrect:

- Acyclovir -> used if the neonate develops clinical varicella infection, not as first-line prophylaxis.
- Observation alone -> not appropriate due to high risk of severe disease.
- Varicella vaccine -> contraindicated in neonates and used only after 12 months of age.

QUESTION

An 8-day-old neonate presents with jaundice. Which of the following is least likely to be the cause of jaundice after the first week of life?

- A Breast milk jaundice
- B Erythroblastosis fetalis**
- C Cystic fibrosis
- D Congenital biliary atresia

CORRECT

RIGHT ANSWER

- B. Erythroblastosis fetalis**

WHY THIS IS RIGHT

The timing of neonatal jaundice is important in determining the cause.

- Erythroblastosis fetalis (hemolytic disease of the newborn) causes severe hemolysis immediately after birth, leading to early-onset jaundice within the first 24-72 hours of life.

Jaundice appearing after the first week is more commonly due to other conditions such as:

- o Breast milk jaundice - prolonged unconjugated jaundice starting around day 5-7.
- o Cystic fibrosis - may cause neonatal cholestasis.
- o Congenital biliary atresia - presents with persistent conjugated jaundice after 1-2 weeks.

QUESTION

An ultrasound shows a fetus with intrauterine growth restriction with head circumference at 75th percentile. All of the following could cause this pattern except:

- A Poor maternal nutrition
- B Pregnancy-induced hypertension
- C TORCH infections**
- D Placental insufficiency

CORRECT

RIGHT ANSWER

OK
C. TORCH infections

WHY THIS IS RIGHT

Intrauterine growth restriction (IUGR) with normal or relatively preserved head circumference (e.g., head circumference at higher percentile than body size) suggests asymmetric IUGR, also called “head-sparing” growth restriction. In conditions causing uteroplacental insufficiency, the fetus redistributes blood flow preferentially to vital organs such as the brain, heart, and adrenals, preserving head growth while abdominal circumference and body weight decrease.

Common causes of asymmetric IUGR:

- Poor maternal nutrition
- Pregnancy-induced hypertension (PIH)
- Placental insufficiency

TORCH infections typically cause symmetric IUGR, because infection occurs early in gestation, affecting overall cell division and growth, resulting in proportionately small head and body.

QUESTION

| Blueberry muffin” rash in an infant most likely represents:

A Dermal erythropoiesis

CORRECT

B Palpable purpura

C Hepatic metastasis

D Viral lesion

RIGHT ANSWER

OK

A. Dermal erythropoiesis

WHY THIS IS RIGHT

A “blueberry muffin” rash in a newborn represents dermal extramedullary hematopoiesis (dermal erythropoiesis) caused by congenital infections-most commonly TORCH infections (especially congenital rubella and CMV)-leading to blue-purple papules due to hematopoietic cell infiltration of the skin.

Why not other options:

- B. Palpable purpura - Due to vasculitis (e.g., IgA vasculitis); lesions are purpuric from vessel inflammation, not dermal hematopoiesis.
- C. Hepatic metastasis - Can occur in neuroblastoma, but classic blueberry muffin rash is from extramedullary hematopoiesis.
- D. Viral lesion - Viruses cause vesicles/macules, not the nodular bluish lesions of dermal erythropoiesis.

QUESTION

| Which of the following is seen in hyaline membrane disease?

- A $V/Q > 1$ and low FRC
- B $V/Q = 1$ and normal FRC
- C $V/Q < 1$ and low FRC
- D $V/Q > 1$ and increased FRC

CORRECT

RIGHT ANSWER

OK

C. $V/Q < 1$ and low FRC

WHY THIS IS RIGHT

Hyaline membrane disease (neonatal respiratory distress syndrome) occurs due to surfactant deficiency, most commonly in premature infants.

- Surfactant deficiency -> alveolar collapse (atelectasis)
- Decreased lung compliance
- Reduced functional residual capacity (FRC)
- Poor ventilation of perfused alveoli -> V/Q mismatch

Because perfusion is relatively maintained but ventilation is reduced, this leads to:

- Low V/Q ratio (<1)
- Decreased FRC

QUESTION

A 2-month-old former 28-week preterm infant is admitted with cough, wheezing, and feeding difficulty. Examination shows moderate respiratory distress with nasal flaring and intercostal retractions. Nasopharyngeal swab is positive for RSV. The infant has a history of bronchopulmonary dysplasia and oxygen requirement at discharge. Which of the following is the most appropriate antiviral therapy?

- A Nirsevimab
- B Ribavirin aerosol therapy**
- C Oral oseltamivir
- D Palivizumab intramuscular injection

CORRECT

RIGHT ANSWER

- OK **B. Ribavirin aerosol therapy**

WHY THIS IS RIGHT

Severe RSV infection in high-risk infants (e.g., premature infants, bronchopulmonary dysplasia, congenital heart disease) may be treated with aerosolized ribavirin, an antiviral that inhibits viral RNA synthesis. It is reserved for severe RSV disease in hospitalized high-risk patients.

Key distinctions:

- Palivizumab: Monoclonal antibody used for RSV prophylaxis, not treatment
- Nirsevimab: Long-acting monoclonal antibody for RSV prevention
- Oseltamivir: Antiviral for influenza, not RSV

QUESTION

Absence of the Landau reflex in an infant older than 6 months is suggestive of:

A Benign familial macrocephaly

B Spastic cerebral palsy

CORRECT

C Infantile colic

D Autism spectrum disorder

RIGHT ANSWER

OK **B. Spastic cerebral palsy**

WHY THIS IS RIGHT

The Landau reflex appears at around 3-4 months of age and persists until about 12-24 months. When an infant is held in prone suspension, the normal response is extension of the head, trunk, and legs. Absence of the Landau reflex after 6 months suggests abnormal motor development, commonly seen in spastic cerebral palsy, due to impaired central motor control.

QUESTION

A 32-week preterm neonate on enteral feeds develops abdominal distension, vomiting, and bloody stools on day 5 of life. Abdominal X-ray shows pneumatosis intestinalis. What is the most appropriate initial management?

- A** Immediate laparotomy
- B** Stop feeds, start broad-spectrum antibiotics, gastric decompression CORRECT
- C** Continue feeds and observe
- D** Give steroids to reduce inflammation

RIGHT ANSWER

- OK** **B. Stop feeds, start broad-spectrum antibiotics, gastric decompression**

WHY THIS IS RIGHT

Necrotizing enterocolitis (NEC) is a serious gastrointestinal emergency seen mainly in preterm infants. The classic radiologic finding is pneumatosis intestinalis (gas within the intestinal wall).

Initial management:

- Stop enteral feeds (NPO)
- Nasogastric/gastric decompression
- Start broad-spectrum IV antibiotics
- Provide IV fluids and supportive care

Surgery is reserved for complications such as intestinal perforation or clinical deterioration.

QUESTION

A late preterm, asymptomatic neonate has a blood glucose of 18 mg/dL at 1 hour of life. What is the most appropriate next step?

A Feed orally and recheck in 1 hour

B Give 40% dextrose gel and recheck

C Start IV 10% dextrose infusion immediately

CORRECT

D Give a glucagon bolus only

RIGHT ANSWER

OK C. Start IV 10% dextrose infusion immediately

WHY THIS IS RIGHT

Severe neonatal hypoglycemia (<25 mg/dL in the first 4 hours of life) requires immediate treatment with intravenous glucose, even if the infant is asymptomatic, because of the risk of acute neurologic injury and seizures.

Management:

- Severe hypoglycemia (<25 mg/dL in first 4 hours) -> IV 10% dextrose bolus/infusion immediately
- Mild/moderate hypoglycemia -> feeding or dextrose gel may be used

QUESTION

A term IDM (infant of a diabetic mother) presents with tachypnea and a harsh systolic murmur. Echocardiography reveals asymmetric septal hypertrophy with no outflow obstruction. What is the underlying mechanism?

- A** Excess fetal glucagon
- B** Fetal hyperinsulinemia acting as a growth factor
- C** Maternal insulin crossing the placenta
- D** Fetal thyroid hormone excess

CORRECT

RIGHT ANSWER

- OK** **B. Fetal hyperinsulinemia acting as a growth factor**

WHY THIS IS RIGHT

In infants of diabetic mothers (IDM), maternal hyperglycemia leads to fetal hyperglycemia, which stimulates the fetal pancreas to produce excess insulin.

Fetal hyperinsulinemia acts as a growth factor, causing myocardial hypertrophy, particularly asymmetric septal hypertrophy (hypertrophic cardiomyopathy). This may present with tachypnea and a systolic murmur in the neonate.

QUESTION

In prenatal evaluation of congenital diaphragmatic hernia, which parameter is used to predict the severity of pulmonary hypoplasia and overall prognosis?

- A Crown-rump length
- B Head circumference percentile
- C Lung-to-Head Ratio**
- D Biparietal diameter

CORRECT

RIGHT ANSWER

- OK
- C. Lung-to-Head Ratio**

WHY THIS IS RIGHT

In congenital diaphragmatic hernia (CDH), the major determinant of prognosis is the degree of pulmonary hypoplasia caused by herniation of abdominal contents into the thoracic cavity. The Lung-to-Head Ratio (LHR) measured on prenatal ultrasound estimates the size of the contralateral fetal lung relative to head circumference, helping predict lung development and survival.

- Low LHR -> severe pulmonary hypoplasia -> poor prognosis
- Higher LHR -> better lung development -> improved survival

QUESTION

| Failure of which mechanism is central to TTN?

- A** Surfactant production
- B** ENaC-mediated alveolar sodium absorption
- C** Type II pneumocyte proliferation
- D** Pulmonary vasodilation

CORRECT

RIGHT ANSWER

- OK** **B. ENaC-mediated alveolar sodium absorption**

WHY THIS IS RIGHT

Transient Tachypnea of the Newborn (TTN) occurs due to delayed clearance of fetal lung fluid after birth. Normally, during labor and shortly after delivery, epithelial sodium channels (ENaC) in the alveolar epithelium actively reabsorb sodium and water from the alveoli into the pulmonary circulation and lymphatics. Failure of ENaC-mediated sodium absorption leads to retained lung fluid, causing tachypnea and mild respiratory distress shortly after birth.

Why not the other options?

- A. Surfactant production: Deficiency causes Respiratory Distress Syndrome (RDS) in premature infants, not TTN.
- C. Type II pneumocyte proliferation: Related to surfactant production; not the mechanism of TTN.
- D. Pulmonary vasodilation: Failure causes Persistent Pulmonary Hypertension of the Newborn (PPHN).

QUESTION

A macrosomic infant of a poorly controlled diabetic mother develops lethargy and seizures on day 2 of life. Labs show hematocrit of 72%. Which complication is most likely due to fetal hyperinsulinemia?

A

Renal vein thrombosis

CORRECT

B

Adrenal hemorrhage

C

Intracranial calcifications

D

Periventricular leukomalacia

RIGHT ANSWER

OK

A. Renal vein thrombosis

WHY THIS IS RIGHT

Infants of poorly controlled diabetic mothers often develop fetal hyperinsulinemia, which leads to macrosomia and chronic intrauterine hypoxia. Hypoxia stimulates increased erythropoietin production, resulting in polycythemia (hematocrit >65%). Severe polycythemia causes hyperviscosity of blood, increasing the risk of thrombotic complications, most classically renal vein thrombosis in the neonate.

Why not the other options?

B. Adrenal hemorrhage: Usually due to birth trauma, asphyxia, or difficult delivery, not polycythemia from hyperinsulinemia.

C. Intracranial calcifications: Seen in congenital infections (TORCH), not in infants of diabetic mothers.

D. Periventricular leukomalacia: Occurs mainly in premature infants due to hypoxic-ischemic injury, not due to neonatal polycythemia/hyperviscosity.

QUESTION

A 6-month-old infant is being evaluated for delayed motor milestones. On examination, the child has poor head control and generalized hypotonia. Birth and immediate neonatal period were uncomplicated. Family history is unremarkable. The pediatric neurologist considers several differential diagnoses that can present with hypotonia in infancy.

Which of the following conditions is least likely to present with hypotonia during infancy?

A Periventricular leukomalacia

CORRECT

B Prader-Willi syndrome

C Congenital myopathy

D Congenital hypothyroidism

RIGHT ANSWER

OK **A. Periventricular leukomalacia**

WHY THIS IS RIGHT

Hypotonia (“floppy infant”) in early infancy is commonly seen in genetic, neuromuscular, and endocrine disorders, such as Prader-Willi syndrome, congenital myopathies, and congenital hypothyroidism, due to impaired muscle tone or neuromuscular function.

In contrast, periventricular leukomalacia (PVL), a form of white matter injury seen in premature infants, typically evolves into spasticity and hypertonia (spastic cerebral palsy) rather than persistent hypotonia. Therefore, PVL is least likely to present with hypotonia in infancy.

QUESTION

A 12-year-old girl is brought by her mother, who asks about the normal sequence of puberty. The girl is noted to have Tanner stage 2 breast development and Tanner stage 2 pubic hair. What is the next physiological event expected in her pubertal progression?

A Axillary hair

B Peak growth in height

CORRECT

C Menarche

D Start of growth spurt

RIGHT ANSWER

OK

B. Peak growth in height

WHY THIS IS RIGHT

In normal female puberty, breast development (thelarche, Tanner stage 2) is the first sign, followed by a sequence of predictable events culminating in menarche.

- Thelarche (breast budding, Tanner stage 2)
- Pubarche (pubic hair)
- Peak height velocity (maximum growth spurt)
- Menarche (typically occurs about 2-2.5 years after thelarche)

Application:

- At Tanner stage 2 for breast and pubic hair, the next expected event is peak growth in height

QUESTION

A 12-year-old child presents with short stature. On evaluation, bone age is found to be 10 years, and the upper segment to lower segment (US:LS) ratio is 1.1:1. There are no dysmorphic features, and the child is otherwise healthy. What is the most likely diagnosis?

A Achondroplasia

B Familial short stature

C Constitutional delay of growth and puberty

CORRECT

D Chondrodysplasia

RIGHT ANSWER

OK

C. Constitutional delay of growth and puberty

WHY THIS IS RIGHT

Constitutional delay of growth and puberty (CDGP) is a common cause of short stature characterized by delayed bone age with otherwise normal body proportions and health.

- Bone age is delayed compared to chronological age
- Normal upper segment to lower segment ratio indicates proportionate short stature
- No dysmorphic features or systemic illness
- Growth velocity is usually normal, but puberty is delayed

Differentiation:

- Familial short stature: bone age matches chronological age
- Achondroplasia/chondrodysplasia: disproportionate short stature with abnormal body ratios

QUESTION

Which of the following is the correct sign indicating adequate growth in an infant with a birth weight of 3 kg?

- A** Increase in length of 25 cm in the first year
- B** Weight gain of 300 grams per month till 1 year
- C** Anterior fontanelle closure by 6 months of age
- D** Weight under 75th percentile and height under 25th percentile

CORRECT

RIGHT ANSWER

- OK** A. Increase in length of 25 cm in the first year

WHY THIS IS RIGHT

Normal infant growth follows predictable patterns, and linear growth (length gain) is a reliable indicator of adequate growth.

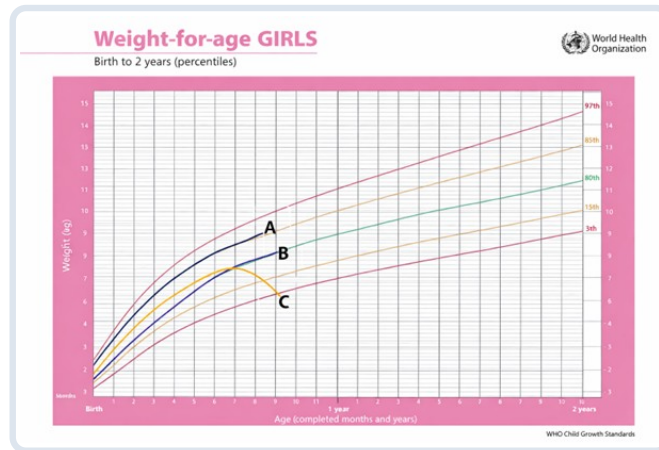
- Length increases by about 25 cm in the first year (≈50 cm at birth → ~75 cm at 1 year)
- Weight typically triples by 1 year, but monthly gain is not constant throughout the year
- Anterior fontanelle normally closes between 9-18 months (not by 6 months)
- Growth assessment depends on tracking along percentiles, not absolute percentile positions

Why option A is correct:

- Increase of ~25 cm in length in the first year is a standard normal growth parameter

QUESTION

Identify the child whose growth is compromised:



A Line A

B Line B

C Line C

CORRECT

D Both B and C

RIGHT ANSWER

OK C. Line C

WHY THIS IS RIGHT

Growth monitoring using WHO growth charts is essential to identify growth faltering, with particular attention to trends rather than single measurements.

- A child's growth should follow a consistent percentile channel
- Crossing down of percentiles or a falling growth curve indicates growth faltering
- Line C shows a downward trend crossing percentiles -> indicates compromised growth
- Lines A and B follow expected growth trajectories without significant deviation

Clinical significance:

- Growth faltering suggests underlying issues such as malnutrition, chronic illness, or malabsorption
- Requires further evaluation and intervention

QUESTION

Maximum brain growth occurs at which year of life?

A 2 years

CORRECT

B 6 years

C 10 years

D 12 years

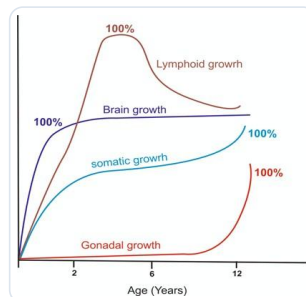
RIGHT ANSWER

OK

A. 2 years

WHY THIS IS RIGHT

The brain undergoes rapid growth in the first 2 years of life, reaching about 80% of adult brain size. This period is critical for synaptogenesis and myelination, making it highly sensitive to nutrition and environmental factors. Hence, maximum brain growth occurs by 2 years of age.



QUESTION

Calculate the Downe score of a newborn who has cyanosis which improves on giving oxygen, mild to moderate retractions, grunting heard on naked ear, air entry markedly decreased, RR 40 per minute?

A 4

B 5

C 6

CORRECT

D 7

RIGHT ANSWER

OK C. 6

WHY THIS IS RIGHT

The Downe score is used to assess the severity of respiratory distress in newborns.

1 (cyanosis) + 1 (retractions) + 2 (grunting) + 2 (air entry) + 0 (RR) = 6

A score of 6 in this case indicates moderate respiratory distress, characterized by cyanosis that improves with oxygen, mild to moderate retractions, grunting heard on naked ear, markedly decreased air entry, and a normal respiratory rate.

Parameter	0	1	2
Respiratory Rate	< 60/min	60–80/min	> 80/min
Cyanosis	None	In room air	Even with 40% O ₂
Air Entry	Normal	Decreased	Barely audible
Grunting	None	Audible with stethoscope	Audible without stethoscope
Retractions	None	Mild	Moderate–severe

QUESTION

| Which of the following does not play a role in fetal growth?

A Growth hormone

CORRECT

B Insulin-like growth factors

C Thyroxine

D All of the above

RIGHT ANSWER

OK

A. Growth hormone

WHY THIS IS RIGHT

Fetal growth is primarily regulated by insulin-like growth factors (IGF-1, IGF-2) and maternal-placental factors, not by growth hormone.

- IGFs are the main drivers of fetal growth and are influenced by fetal insulin and nutrient supply
- Thyroxine plays a supportive role in overall growth and development
- Growth hormone has minimal to no role in intrauterine growth and becomes important only after birth

QUESTION

A 4-year-old child with weight for height less than -1 SD & height for age is -2 SD. What is the best possible assessment?

- A Acute malnutrition
- B Chronic malnutrition**
- C Acute on chronic malnutrition
- D None of the above

CORRECT

RIGHT ANSWER

- OK
- B. Chronic malnutrition**

WHY THIS IS RIGHT

Different anthropometric indices help distinguish between acute and chronic malnutrition in children.

- Height-for-age reflects long-term nutritional status (stunting) -> indicator of chronic malnutrition
- Weight-for-height reflects current nutritional status (wasting) -> indicator of acute malnutrition

Application to the question:

- Height-for-age = -2 SD -> indicates stunting -> chronic malnutrition
- Weight-for-height = -1 SD -> within normal/mild range, does not indicate acute malnutrition

QUESTION

What is the rate of increment of head circumference in the first three months of life?

A 3 cm per month

B 2 cm per month

CORRECT

C 1 cm per month

D 0.5 cm per month

RIGHT ANSWER

OK B. 2 cm per month

WHY THIS IS RIGHT

Head circumference is a key indicator of brain growth in early infancy, with the most rapid increase occurring in the first few months of life.

Key growth pattern:

- 0-3 months: increases by about 2 cm per month
- 3-6 months: increases by about 1 cm per month
- 6-12 months: increases by about 0.5 cm per month

Clinical importance:

- Rapid head growth reflects normal brain development
- Deviations may indicate conditions like microcephaly or hydrocephalus

QUESTION

Treatment non-responsive Severe Acute Malnutrition (SAM) includes all except:

- A** Appetite not increased by day 4
- B** No edema reduction by day 4
- C** **No Complete absence of edema by day 20**
- D** Weight gain less than 5 gm/kg/day for 3 consecutive days

CORRECT

RIGHT ANSWER

OK

C. No Complete absence of edema by day 20

WHY THIS IS RIGHT

Non-responsive severe acute malnutrition (SAM) is identified when a child fails to show expected clinical or nutritional improvement despite appropriate treatment, prompting evaluation for underlying causes or complications.

Criteria suggesting non-response:

- Poor appetite not improving by day 3-4
- Failure of edema to start reducing by day 4
- Inadequate weight gain (<5 g/kg/day) over consecutive days

Why option C is incorrect:

- Complete resolution of edema is expected by around 2-3 weeks, but not necessarily by exactly day 20
- Lack of complete edema disappearance by day 20 alone is not used as a standard criterion for non-response

QUESTION

Which among the following is not included in the severe acute malnutrition definition?

A Weight for height

B Height for age

CORRECT

C MUAC <11.5

D Pedal oedema

RIGHT ANSWER

OK B. Height for age

WHY THIS IS RIGHT

Severe acute malnutrition (SAM) is defined using indicators of acute nutritional status, not chronic growth failure.

Diagnostic criteria for SAM:

- Weight-for-height/length < -3 SD
- Mid-upper arm circumference (MUAC) < 11.5 cm (6-59 months)
- Presence of bilateral pitting edema

Why height-for-age is not included:

- Height-for-age reflects chronic malnutrition (stunting), not acute malnutrition
- SAM focuses on wasting and edema, which indicate recent or severe nutritional deficiency

QUESTION

Which of the following statements is true regarding Marasmus in children?

- A Seen due to calories deficiency
- B Protein loss is responsible for edema in this patient
- C Subcutaneous fat is preserved
- D Voracious appetite is seen

CORRECT

RIGHT ANSWER

- OK D. Voracious appetite is seen

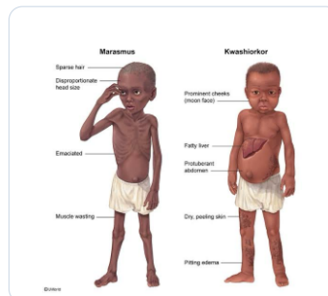
WHY THIS IS RIGHT

Marasmus is a form of severe acute malnutrition caused by deficiency of total caloric intake (both protein and energy), leading to severe wasting without edema.

- Marked loss of subcutaneous fat and muscle mass -> “skin and bones” appearance
- No edema (distinguishes it from kwashiorkor)
- Child is typically alert with a voracious appetite
- Growth failure with significant weight loss

Why others are incorrect:

- A is incomplete: marasmus involves deficiency of both calories and protein
- B describes kwashiorkor (protein deficiency -> edema)
- C is incorrect: subcutaneous fat is markedly reduced



QUESTION

Which of the following condition best explains the development of hypophosphatemia and hypomagnesemia in a malnourished child following the initiation of nutritional rehabilitation?

CORRECT

A Mechanism of refeeding syndrome

B Insulin mediated uptake of electrolytes

C Urinary loss of phosphates

D None of the above

RIGHT ANSWER

OK

A. Mechanism of refeeding syndrome

WHY THIS IS RIGHT

Refeeding syndrome occurs when nutrition is reintroduced in a severely malnourished child, leading to rapid metabolic and electrolyte shifts driven by a sudden increase in insulin secretion.

- Starvation state: low insulin, high glucagon -> depletion of intracellular electrolytes (phosphate, magnesium, potassium)
- On refeeding (especially carbohydrates): insulin release increases -> drives glucose and electrolytes (phosphate, magnesium, potassium) into cells
- This results in hypophosphatemia, hypomagnesemia, and hypokalemia in the blood
- Hypophosphatemia is the hallmark and can lead to muscle weakness, respiratory failure, cardiac dysfunction, and hemolysis
- Hypomagnesemia can cause arrhythmias and neuromuscular irritability

Why others are incomplete:

- Insulin-mediated uptake is part of the mechanism but does not fully define the syndrome
- Urinary loss is not the primary cause

QUESTION

A three-year-old child presents with cough, wheeze, and recurrent infection for which he has been taking oral medications. He now presents with excessive weight gain, a round face, hirsutism, and hair growth in the forehead. Developmental milestones were normal, child's BMI is more than +2 standard deviation, but height for age is below -2 standard deviation. What is the next best step in management?

- A** Checking for congenital hypothyroidism with T3, T4, TSH.
- B** Checking for early morning cortisol and history of exogenous cortisol administration. CORRECT
- C** Advise exercise and dietary changes for the child.
- D** Evaluate for genetic causes of obesity.

RIGHT ANSWER

- OK** **B. Checking for early morning cortisol and history of exogenous cortisol administration.**

WHY THIS IS RIGHT

Exogenous Cushing syndrome should be suspected in a child with excessive weight gain, growth failure (short stature), and cushingoid features such as moon face, hirsutism, and facial fullness, especially with a history of recurrent respiratory illness suggesting possible prolonged corticosteroid use.

- Weight gain with decreased height velocity is a red flag for endocrine pathology (not simple obesity)
- Features like moon face, hirsutism, and truncal obesity point toward hypercortisolism
- Most common cause in children is exogenous steroid use (e.g., repeated oral steroids for wheeze/asthma)
- Initial step is to take a detailed drug history and check early morning cortisol (suppressed in exogenous cause)

Contrast:

- Simple obesity: weight gain with normal or increased height
- Hypothyroidism: weight gain is mild with lethargy, constipation, and delayed milestones
- Genetic obesity: usually early onset with normal or increased height initially

QUESTION

A 7-year-old child presents with short stature (height is -5 SD). The child is eating well and gaining weight. On examination, upper segment to lower segment ratio is 1.4:1 and systemic examinations are within normal. What is the most likely diagnosis?

A Spondyloepiphyseal dysplasia

B Achondroplasia

CORRECT

C Growth hormone deficiency

D Severe malnutrition

RIGHT ANSWER

OK B. Achondroplasia

WHY THIS IS RIGHT

Achondroplasia is a skeletal dysplasia characterized by disproportionate short stature with relatively normal trunk length and shortened limbs (rhizomelic shortening). A key clinical clue is an increased upper segment to lower segment ratio, reflecting short limbs compared to the trunk.

- Disproportionate short stature with normal weight gain and nutrition
- Increased upper segment to lower segment ratio
- Normal intelligence and systemic examination
- Due to mutation in FGFR3 causing impaired endochondral ossification

Contrast:

- Growth hormone deficiency: proportionate short stature with delayed growth and often poor weight gain
- Severe malnutrition: both height and weight are reduced
- Spondyloepiphyseal dysplasia: primarily affect

QUESTION

| All of the following are contraindications of breastfeeding except:

- A Active TB
- B Congenital lactose intolerance
- C Metastatic malignancy in mother**
- D Maternal HIV

CORRECT

RIGHT ANSWER

- OK
- C. Metastatic malignancy in mother**

WHY THIS IS RIGHT

Breastfeeding is contraindicated in certain maternal or infant conditions due to the risk of infection transmission or metabolic intolerance.

Absolute contraindications include:

- Maternal HIV infection (in settings where safe alternatives are available).
- Untreated active tuberculosis (breastfeeding may begin after treatment initiation and precautions).
- Congenital lactose intolerance / classic galactosemia in the infant because breast milk contains lactose.

Metastatic malignancy in the mother is not a direct contraindication to breastfeeding unless the mother is receiving chemotherapy, radiotherapy, or contraindicated medications.

QUESTION

A child with rickets not responding to treatment is being evaluated. Investigations show a serum calcium level of 9 mg/dL, phosphate level of 2 mg/dL, elevated alkaline phosphatase, and normal parathyroid hormone levels with normal arterial blood gas levels. What is the diagnosis?

A Vitamin D resistant rickets type 2

B Hypophosphatemic rickets

CORRECT

C Vitamin D resistant rickets type 1

D Nutritional rickets

RIGHT ANSWER

OK

B. Hypophosphatemic rickets

WHY THIS IS RIGHT

Hypophosphatemic rickets (most commonly X-linked hypophosphatemic rickets) is characterized by renal phosphate wasting, leading to defective bone mineralization.

- Normal serum calcium
- Low serum phosphate
- Elevated alkaline phosphatase
- Normal or mildly increased PTH
- Normal acid-base status

Rickets that does not respond to vitamin D therapy should raise suspicion for hypophosphatemic rickets.

QUESTION

28-year-old postpartum woman complains of sore nipple. A lactation specialist evaluates and tells the patient that the baby's attachment is poor. All of the following are signs of good attachment during breastfeeding except?

A Baby's chin is touching the breasts

B Baby's lower lip is everted

C Baby's mouth is wide open

D Lower areola more visible than upper

CORRECT

RIGHT ANSWER

OK

D. Lower areola more visible than upper

WHY THIS IS RIGHT

Proper latch (good attachment) during breastfeeding is essential to ensure effective milk transfer and prevent nipple pain or trauma.

Signs of good attachment include:

- Baby's mouth wide open
- Chin touching the breast
- Lower lip everted (turned outward)
- More areola visible above the baby's mouth than below

If the lower areola is more visible than the upper areola, it indicates poor latch, meaning the baby is not taking enough of the lower breast tissue into the mouth.

QUESTION

A 3-week-old girl is brought to the emergency department due to lethargy. The patient was breastfeeding well until this morning when she became increasingly difficult to rouse. She was born at term to a 35-year-old woman who had a spontaneous vaginal delivery at home. The patient did not receive any vaccinations or medications after birth due to parental preference. Head circumference is at the 99th percentile. Weight and length are at the 25th percentile. Temperature is 37° C (98.6° F). Physical examination shows a large, bulging anterior fontanelle. The eyes are driven downward, and the patient does not appear to be able to look upward. No scalp swelling is present. Intracranial hemorrhage is confirmed on CT scan of the head. Which of the following is the most likely underlying cause of this patient's condition?

A Germinal matrix fragility

B Inherited hemophilia

C Trauma during birth

D Vitamin deficiency

CORRECT

RIGHT ANSWER

OK D. Vitamin deficiency

WHY THIS IS RIGHT

Newborns are at risk of vitamin K deficiency bleeding (VKDB) because they have:

- Low vitamin K stores at birth
- Poor placental transfer of vitamin K
- Sterile intestines (lack of gut flora to synthesize vitamin K)
- Low vitamin K content in breast milk

Infants who do not receive prophylactic vitamin K injection at birth are at risk of late VKDB (2-12 weeks of age), which commonly presents with intracranial hemorrhage.

Why not other options:

- A. Germinal matrix fragility: Occurs in premature infants with IVH in the first few days of life. This baby is term and 3 weeks old.
- B. Inherited hemophilia: X-linked, mainly affects males; this patient is female.
- C. Birth trauma: Bleeding appears immediately after birth, not after 3 weeks.

QUESTION

An 11-year-old girl is brought to the physician for a physical examination prior to participating in sports. She is at the 50 percentile for height and weight. Physical examination shows a normal female body habitus, a slight increase in breast areolar diameter with nipple protrusion, and normal-appearing external genitalia with sparse straight hair on the labia majora. The physician concludes that the patient's breast and external genitalia are in the same stage of sexual development. Which of the following terms best describes the sexual development of this patient?

A Tanner stage 1

B Tanner stage 2

CORRECT

C Tanner stage 3

D Tanner stage 5

RIGHT ANSWER

OK B. Tanner stage 2

WHY THIS IS RIGHT

Tanner staging is used to assess sexual maturation during puberty based on breast development and pubic hair growth in girls.

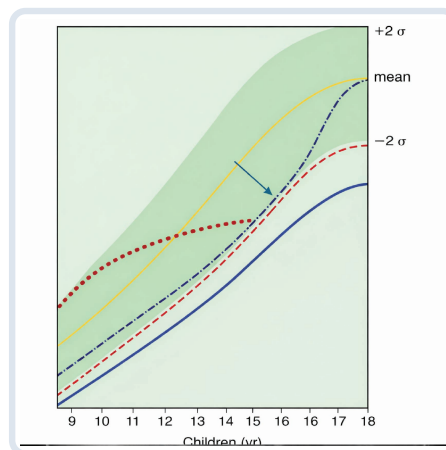
Tanner Stage 2 (early puberty) features:

- Breast bud stage: small breast mound with enlargement of the areola and nipple.
- Pubic hair: sparse, lightly pigmented, straight hair along the labia majora.

This stage represents the earliest visible sign of puberty in girls, typically occurring around 10-11 years of age.

QUESTION

Identify the correct cause of the marked graph



CORRECT

A A child whose bone age is less than the chronological age and the growth velocity is normal.

B A child with a pituitary tumor presenting with growth hormone deficiency.

C A child diagnosed with Down's syndrome.

D A child with a family history of short stature.

RIGHT ANSWER

OK **A. A child whose bone age is less than the chronological age and the growth velocity is normal.**

WHY THIS IS RIGHT

The growth chart shows a child who is initially below the average height percentile, followed by a delayed but rapid catch-up growth during adolescence, eventually approaching the normal mean height. This pattern represents a delayed growth spurt with eventual normal adult height.

This is characteristic of constitutional delay of growth and puberty (CDGP).

- Bone age is delayed compared to chronological age
- Normal growth velocity
- Delayed onset of puberty
- Late growth spurt with catch-up growth
- Final adult height usually normal

QUESTION

Anthropometric measurements for a 1-year-old child and corresponding values at birth are given below. Which of the following parameters indicates abnormal growth?

1. Weight - 6 kg; Birth weight - 3 kg
2. Length - 75 cm; Birth length - 50 cm
3. Head circumference (HC) - 47 cm; Birth HC - 34 cm
4. Chest circumference (CC) - 47 cm; Birth CC - 31 cm

A 3,4

B 1

CORRECT

C 1,3

D 2,4

RIGHT ANSWER

OK B. 1

WHY THIS IS RIGHT

Evaluation of given parameters:

1. Weight: 6 kg (birth 3 kg) -> only doubled, not tripled -> abnormal growth
 2. Length: 75 cm (birth 50 cm) -> increase of 25 cm -> normal
 3. Head circumference: 47 cm (birth 34 cm) -> normal
 4. Chest circumference: 47 cm (birth 31 cm) -> normal (equals head circumference at 1 year)
- Therefore, the only abnormal parameter is weight.

QUESTION

A mother of a 2-month-old infant is concerned about her baby's slow weight gain. The baby is exclusively breastfed. The baby passes urine 6-8 times a day. What is the most likely explanation in this case?

- A Lactose intolerance
- B Insufficient intake of hindmilk**
- C Insufficient intake of foremilk
- D Galactosemia

CORRECT

RIGHT ANSWER

- B. Insufficient intake of hindmilk**

WHY THIS IS RIGHT

In an exclusively breastfed infant with slow weight gain but adequate urine output (6-8 times/day), the most likely cause is insufficient intake of hindmilk.

- Foremilk (initial milk during feeding) is low in fat and high in lactose, mainly satisfying thirst.
- Hindmilk (milk released later during feeding) is rich in fat and calories, which is essential for weight gain.

If the infant switches breasts too early or feeds for a short duration, they may receive mostly foremilk and not enough hindmilk, leading to poor weight gain despite adequate hydration.

QUESTION

A 6-year-old child is brought with complaint of short stature. The height of the child corresponds to parent's height and the chronological age of the child corresponds to bone age. What is the likely condition?

A Constitutional delay

B Familial short stature

CORRECT

C GH deficiency

D Normal

RIGHT ANSWER

OK B. Familial short stature

WHY THIS IS RIGHT

Familial short stature is characterized by short height consistent with the genetic potential of the parents, with normal growth velocity and normal bone age.

- Height below average but appropriate for parental height (target height)
- Normal growth velocity
- Bone age equal to chronological age
- Normal onset of puberty

This distinguishes it from constitutional growth delay, where bone age is delayed compared to chronological age, and puberty is also delayed.

QUESTION

A father brings his child with a distended abdomen and poor feeding. Laboratory investigations show markedly low serum albumin levels. Which of the following findings are not consistent with this condition?

A Simian facies

CORRECT

B Flag sign

C Flaky paint dermatosis

D Fatty liver

RIGHT ANSWER

OK

A. Simian facies

WHY THIS IS RIGHT

The clinical picture of distended abdomen, poor feeding, and markedly low serum albumin suggests kwashiorkor, a form of severe protein malnutrition.

Characteristic features of kwashiorkor include:

- Edema due to hypoalbuminemia
 - Fatty liver from impaired lipoprotein synthesis
 - Skin changes such as “flaky paint” dermatosis
 - Hair changes like the “flag sign” (alternating bands of depigmented hair due to intermittent protein deficiency)
- Simian facies, however, is typical of marasmus, which is caused by severe calorie deficiency and leads to loss of subcutaneous fat giving an “old man” or monkey-like appearance.

QUESTION

| Which of the following statement is false in a 1-year-old child?

A Chest circumference is equal to head circumference

B Birth weight is quadrupled

CORRECT

C Arm span is lesser than height

D Head circumference is approximately 46-47 cm

RIGHT ANSWER

OK

B. Birth weight is quadrupled

WHY THIS IS RIGHT

Normal growth parameters at 1 year of age include characteristic changes in weight and body proportions.

Key developmental growth facts at 1 year:

- Birth weight is tripled (not quadrupled).
- Head circumference $\approx$ 46-47 cm.
- Chest circumference becomes approximately equal to head circumference around 1 year.
- Arm span is slightly less than height in infancy and early childhood.

Therefore, the statement "birth weight is quadrupled" is false because birth weight normally quadruples by about 2 years of age, not at 1 year.

QUESTION

| What is the expected growth velocity in a child after 4 years?

A 6 cms/year

CORRECT

B 3 cms/year

C 8 cms/year

D 10 cms/year

RIGHT ANSWER

OK

A. 6 cms/year

WHY THIS IS RIGHT

Normal growth velocity varies with age in children. After the early childhood growth spurt, the rate of growth becomes relatively steady.

Typical growth velocities:

- Infancy (0-1 year): ~25 cm/year
- 1-2 years: ~10-12 cm/year
- 2-4 years: ~7-8 cm/year
- After 4 years until puberty: ~5-6 cm/year

Thus, the expected average growth velocity after 4 years is about 6 cm per year.

QUESTION

| Which of the following statements is incorrect about breastmilk composition?

- A** Polyunsaturated fatty acids help nerve myelination.
- B** Lactoferrin provides antimicrobial action.
- C** Breastmilk is deficient in vitamin C
- D** Exclusive breastfeeding may cause vitamin D deficiency.

CORRECT

RIGHT ANSWER

OK

C. Breastmilk is deficient in vitamin C

WHY THIS IS RIGHT

Human breast milk provides adequate amounts of vitamin C, so it is not deficient in vitamin C. Vitamin C in breast milk helps with iron absorption and immune function in the infant.

Important components of breast milk include:

- Polyunsaturated fatty acids (DHA, ARA) -> essential for brain development and nerve myelination.
- Lactoferrin -> an iron-binding protein with antimicrobial properties, inhibiting bacterial growth.
- Vitamin D -> present in low amounts, which is why exclusively breastfed infants require vitamin D supplementation.

QUESTION

Regarding vitamin D supplementation in an exclusively breast-fed newborn, which of the following is recommended?

- A Immediately, initially 200 IU and after 6 months 400 IU
- B To be started after breastfeeding is stopped
- C To be started 400 IU at the time of complementary feeding

D Immediately 400 IU

CORRECT

RIGHT ANSWER

D. Immediately 400 IU

WHY THIS IS RIGHT

Exclusively breast-fed infants should receive vitamin D supplementation of 400 IU/day starting soon after birth.

- Human breast milk contains insufficient vitamin D ($\approx 20\text{-}40$ IU/L) to meet the infant's requirement.
- Therefore, guidelines recommend 400 IU of vitamin D daily from the first few days of life, regardless of maternal vitamin D status.
- Supplementation should continue until the infant consumes adequate vitamin D from fortified formula or diet (≥ 400 IU/day).

QUESTION

| Which of the following criteria is common to both acute and chronic malnutrition?

A Weight for height

B Weight for age

CORRECT

C Height for age

D Mid-arm circumference

RIGHT ANSWER

OK B. Weight for age

WHY THIS IS RIGHT

Acute malnutrition -> assessed by weight for height and mid-arm circumference (reflect wasting)

Chronic malnutrition -> assessed by height for age (reflect stunting)

Weight for age is influenced by both weight (acute) and height (chronic), so it becomes a combined indicator, making it common to both conditions.

QUESTION

Delayed puberty is seen in all of the following conditions except:

- A** Growth hormone deficiency
- B** Constitutional delay in growth and puberty
- C** Turner syndrome
- D** **Familial short stature**

CORRECT

RIGHT ANSWER

OK

D. Familial short stature

WHY THIS IS RIGHT

Delayed puberty occurs when there is delayed activation of the hypothalamic-pituitary-gonadal (HPG) axis or gonadal failure.

Conditions associated with delayed puberty:

- Growth hormone deficiency -> impaired growth and delayed sexual maturation
- Constitutional delay of growth and puberty (CDGP) -> temporary delay in puberty with eventual normal development
- Turner syndrome -> gonadal dysgenesis causing primary ovarian failure

Not associated:

- Familial short stature -> children are short but have normal timing of puberty and bone age.

QUESTION

A 2-year-old child with severe marasmus is admitted for nutritional rehabilitation. The WHO cost sheet for management includes therapeutic feeds (F-75/F-100), antibiotics, vitamin A, zinc, potassium, magnesium, and folate supplements. Which of the following is not included, and for good physiologic reason?

- A** Zinc supplementation
- B** Therapeutic milk feeds
- C** Potassium supplementation
- D** Insulin administration

CORRECT

RIGHT ANSWER

- OK** D. Insulin administration

WHY THIS IS RIGHT

Insulin is not given in initial management of severe PEM because:

- Children are in a low-insulin catabolic state
- Refeeding -> endogenous insulin rises naturally
- Exogenous insulin can precipitate refeeding syndrome -> hypophosphatemia, hypokalemia, arrhythmias, seizures
- WHO protocols focus on slow, safe anabolic transition rather than forcing insulin-driven anabolism

QUESTION

A 12-month-old infant presents with severe wasting, loss of subcutaneous fat, and no edema. The mother states she diluted cow's milk due to poverty. Which statement best explains the metabolic state?

A Adequate protein but deficient carbohydrate

B Adequate protein but deficient fat

C Deficient protein with adequate calories

D Deficient total calories (protein + calories)

CORRECT

RIGHT ANSWER

OK

D. Deficient total calories (protein + calories)

WHY THIS IS RIGHT

Marasmus is a form of severe protein-energy malnutrition caused by deficiency of total calories (both protein and energy). It typically occurs due to prolonged inadequate intake, such as feeding diluted milk.

- Severe wasting
- Loss of subcutaneous fat
- No edema
- "Skin and bones" appearance
- Growth failure and irritability

QUESTION

A child with rickets has the following profile:

Serum calcium: Normal

Serum phosphate: Low

PTH: Normal

1,25-dihydroxyvitamin D: Low-normal

Urinary phosphate excretion: Increased

Which therapy directly targets the primary pathogenic mechanism?

- A** Ergocalciferol
- B** Oral phosphate supplementation
- C** Burosumab
- D** Teriparatide

CORRECT

RIGHT ANSWER

OK **C. Burosumab**

WHY THIS IS RIGHT

Rickets with normal calcium, low phosphate, normal PTH, low-normal 1,25-dihydroxyvitamin D and increased urinary phosphate excretion suggests FGF23-mediated hypophosphatemic rickets, most commonly X-linked hypophosphatemia. In this disorder, excess Fibroblast growth factor (FGF23) causes Renal phosphate wasting -> hypophosphatemia and reduced renal 1- α hydroxylase activity -> decreased production of 1,25-dihydroxyvitamin D

The targeted therapy is Burosumab, a monoclonal antibody against FGF23 that:

- Inhibits excess FGF23 activity
- Increases renal phosphate reabsorption
- Raises 1,25-dihydroxyvitamin D levels
- Improves bone mineralization and rickets

Why not the other options?

- Ergocalciferol (Vitamin D₂): Used for nutritional vitamin D deficiency rickets, not FGF23 excess.
- Oral phosphate supplementation: Traditional therapy but does not address the underlying FGF23 excess.
- Teriparatide: PTH analog used in osteoporosis; unrelated to phosphate-wasting rickets.

QUESTION

A teenage boy, who was previously raised as a girl, presents with primary amenorrhea and absence of secondary sexual characteristics. On examination, he is tall, has small testes, and gynecomastia. Serum testosterone levels are within the normal range. Which of the following is the most likely diagnosis?

A 45, X

B 46, XX

C 47, XXY

CORRECT

D 47, XYY

RIGHT ANSWER

OK

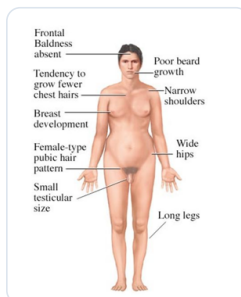
C. 47, XXY

WHY THIS IS RIGHT

Tall stature, small testes, gynecomastia, and primary amenorrhea-like presentation with normal-range testosterone point to Klinefelter syndrome (47, XXY).

It causes primary testicular failure with low fertility and often learning/behavioral issues.

Labs typically show $\uparrow$ FSH/ $\uparrow$ LH with relatively low-normal testosterone.



QUESTION

A young boy is brought to the clinic for developmental assessment. On examination, he has a prominent epicanthal fold, a single transverse palmar crease, hypotonia and intellectual disability. Facial features appear flat with upslanting palpebral fissures. What is the most likely diagnosis?

A Down Syndrome

CORRECT

B Patau Syndrome

C Edward Syndrome

D Fragile X Syndrome

RIGHT ANSWER

OK

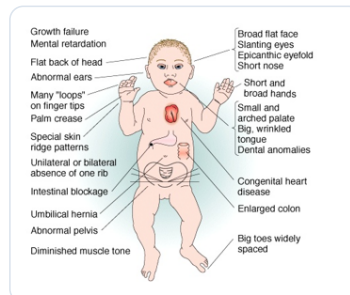
A. Down Syndrome

WHY THIS IS RIGHT

The findings-flat facial profile, upslanting palpebral fissures, epicanthal folds, hypotonia, intellectual disability, and single transverse palmar crease-are classic for Down syndrome (Trisomy 21).

Common associated issues include AV septal defect, duodenal atresia, and hypothyroidism.

Early screening for cardiac, hearing, vision, and thyroid problems is essential.



QUESTION

Which genetic syndrome results from a deletion of the short arm of chromosome 5 (5p deletion)?

A Edward syndrome

B Patau syndrome

C Cri du chat syndrome

CORRECT

D Turner Syndrome

RIGHT ANSWER

OK C. Cri du chat syndrome

WHY THIS IS RIGHT

Cri du chat syndrome is caused by deletion of the short arm of chromosome 5 (5p deletion) and is characterized by a distinctive high-pitched “cat-like” cry in infancy.

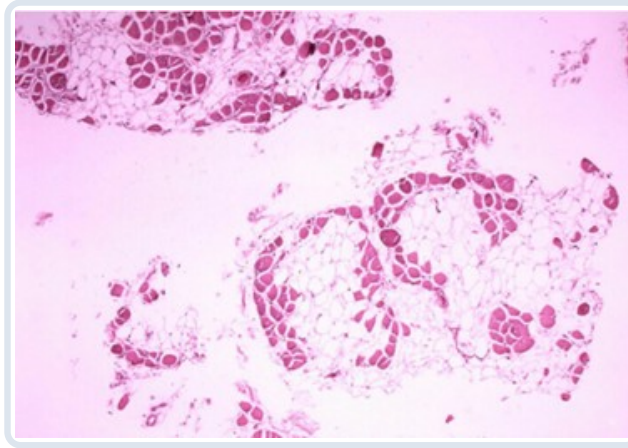
- Genetic defect: deletion of 5p
- Clinical features:
 - High-pitched cry
 - Microcephaly
 - Intellectual disability
 - Hypertelorism and facial dysmorphism

Differentiation:

- Edward syndrome: trisomy 18
- Patau syndrome: trisomy 13
- Turner syndrome: monosomy X (45,X)

QUESTION

An 8-year-old child has difficulty walking and getting up from a squatting position. A muscle biopsy was done and is as shown in the image. Which of the following is true about this condition?



A Death occurs in 3rd decade

CORRECT

B Previous history of viral prodrome

C It is a mitochondrial storage disorder

D Early treatment has excellent prognosis

RIGHT ANSWER

OK

A. Death occurs in 3rd decade

WHY THIS IS RIGHT

Duchenne muscular dystrophy (DMD) is an X-linked recessive disorder caused by mutation in the dystrophin gene, leading to progressive muscle degeneration.

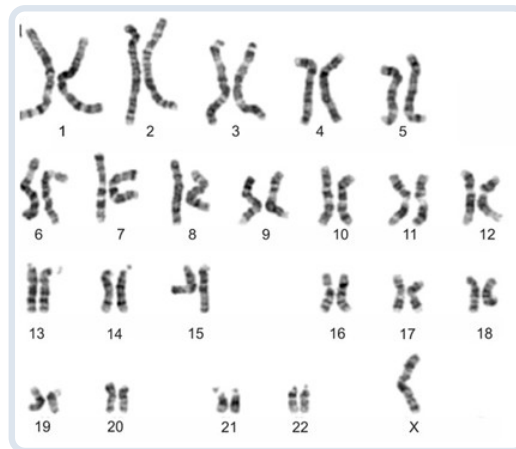
- Presents in early childhood with proximal muscle weakness and Gowers' sign (difficulty getting up from squatting)
- Muscle biopsy shows muscle fiber degeneration with replacement by fat and connective tissue
- Serum creatine kinase (CK) is markedly elevated
- Progressive weakness leading to loss of ambulation by adolescence
- Death usually occurs in the second to third decade due to respiratory or cardiac failure

Differentiation:

- Not associated with viral prodrome (rules out myositis)
- Not a mitochondrial disorder

QUESTION

A 15-year-old girl with wide-spaced nipples, short height, and the following karyotype.



A Turner syndrome

CORRECT

B Klinefelter syndrome

C Down syndrome

D Partial mole

RIGHT ANSWER

OK

A. Turner syndrome

WHY THIS IS RIGHT

Turner syndrome is a chromosomal disorder characterized by monosomy X (45,X), leading to gonadal dysgenesis and characteristic physical features.

- Clinical features: short stature, webbed neck, shield chest with widely spaced nipples, primary amenorrhea
- Karyotype: 45,X (absence of one X chromosome)
- Streak gonads → infertility

Associated conditions:

- Congenital heart defects (e.g., coarctation of aorta)
- Renal anomalies
- Hypothyroidism

Differentiation:

- Klinefelter syndrome: 47,XXY males
- Down syndrome: trisomy 21
- Partial mole: triploid karyotype

QUESTION

An abnormally tall patient presents with lens subluxation, long limbs, and arachnodactyly. A mutation in which of the following genes is most commonly associated with this condition?

A Fibrillin-1 gene (FBN1)

CORRECT

B Collagen Type I gene

C Elastin gene

D Dystrophin gene

RIGHT ANSWER

OK

A. Fibrillin-1 gene (FBN1)

WHY THIS IS RIGHT

Marfan syndrome is an autosomal dominant connective tissue disorder caused by mutation in the FBN1 gene encoding fibrillin-1, leading to abnormal elastic fiber formation and increased TGF- β activity

Clinical features: tall stature, long limbs (dolichostenomelia), arachnodactyly, lens subluxation (typically upward), and aortic root dilation

- Risk of aortic aneurysm and dissection (most serious complication)
- Requires regular cardiovascular monitoring

Contrast:

- Collagen type I defect -> osteogenesis imperfecta
- Elastin gene defect -> supravalvular aortic stenosis (e.g., Williams syndrome)
- Dystrophin defect -> Duchenne muscular dystrophy

QUESTION

| Which of the following is not seen in down syndrome?

A Hypothyroidism

B Hearing loss

C Short stature

D Caudal regression syndrome

CORRECT

RIGHT ANSWER

OK D. Caudal regression syndrome

WHY THIS IS RIGHT

Down syndrome (Trisomy 21) is associated with multiple systemic abnormalities affecting endocrine, auditory, and growth parameters.

- Common associations include hypothyroidism, hearing loss, and short stature
- Other features: intellectual disability, characteristic facies, congenital heart defects (e.g., AV septal defect)

Why caudal regression syndrome is not seen:

- Caudal regression syndrome is a separate congenital anomaly associated with maternal diabetes, not Down syndrome

QUESTION

A tall 10-year-old boy presents with Sexual Maturity Rating 5 and hyperpigmentation. His BP is 150/100 mm Hg. What is the likely diagnosis?

A 11 β -Hydroxylase deficiency

CORRECT

B 21 β -Hydroxylase deficiency

C 17 β -Hydroxylase deficiency

D SCC deficiency

RIGHT ANSWER

OK

A. 11 β -Hydroxylase deficiency

WHY THIS IS RIGHT

11 β -Hydroxylase deficiency in congenital adrenal hyperplasia causes hypertension due to salt retention and precocious puberty due to excess androgen production.

| Enzyme Deficiency | Mineralocorticoids | Glucocorticoids | Androgens | Symptoms |

| ----- | ----- | ----- | ----- | ----- |

| 11 β -hydroxylase | $\uparrow^*$ | $\downarrow$ | $\uparrow$ | • Hypertension, $\downarrow$ K $\square$

• Ambiguous genitalia in girls |

| 17 α -hydroxylase | $\uparrow$ | $\uparrow^{**}$ | $\downarrow$ | • Hypertension, $\downarrow$ K $\square$

• Ambiguous genitalia in boys •

Absent puberty |

| 21-hydroxylase | $\downarrow$ | $\downarrow$ | $\uparrow$ | • Salt wasting (vomiting, hypotension, $\downarrow$ Na $\square$, $\uparrow$ K $\square$)

• Ambiguous genitalia in girls |

QUESTION

| Which of the following is not an example of an aneuploidy disorder?

CORRECT

A Bloom syndrome

B Trisomy 21

C Trisomy 13

D Klinefelter syndrome

RIGHT ANSWER

OK **A. Bloom syndrome**

WHY THIS IS RIGHT

Aneuploidy refers to an abnormal number of chromosomes due to gain or loss of whole chromosomes, typically caused by nondisjunction.

- Trisomy 21, Trisomy 13, and Klinefelter syndrome (47,XXY) are classic examples of aneuploidy
- These conditions involve numerical chromosomal abnormalities

Why Bloom syndrome is not aneuploidy:

- Bloom syndrome is a chromosomal instability disorder caused by mutation in the BLM gene
- It involves defective DNA repair and increased sister chromatid exchanges, not a change in chromosome number

QUESTION

Which of the following is not seen in Turner syndrome?

A Widely spaced nipples

B Short stature

C Prominent occiput

CORRECT

D Barr body

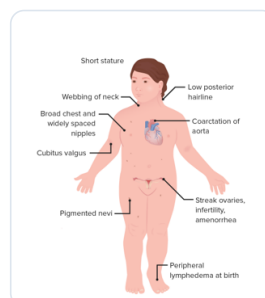
RIGHT ANSWER

OK C. Prominent occiput

WHY THIS IS RIGHT

Turner syndrome is a chromosomal disorder (45,XO) characterized by gonadal dysgenesis and distinctive somatic features, with absence of Barr body due to monosomy X.

- Common features include short stature, webbed neck, shield chest with widely spaced nipples, and streak gonads
- Other findings: coarctation of aorta, lymphedema at birth, primary amenorrhea
- Barr body is absent because there is only one X chromosome
- Prominent occiput is not a feature of Turner syndrome



QUESTION

| Which of the following has autosomal recessive pattern of inheritance?

- A Achondroplasia
- B Huntington's disease
- C Cystic fibrosis**
- D Familial Hypercholesterolemia

CORRECT

RIGHT ANSWER

OK **C. Cystic fibrosis**

WHY THIS IS RIGHT

Cystic fibrosis is an autosomal recessive disorder caused by mutations in the CFTR gene, which encodes a chloride channel involved in fluid and electrolyte transport across epithelial cells.

Defective CFTR leads to thick, viscous secretions in multiple organs, resulting in:

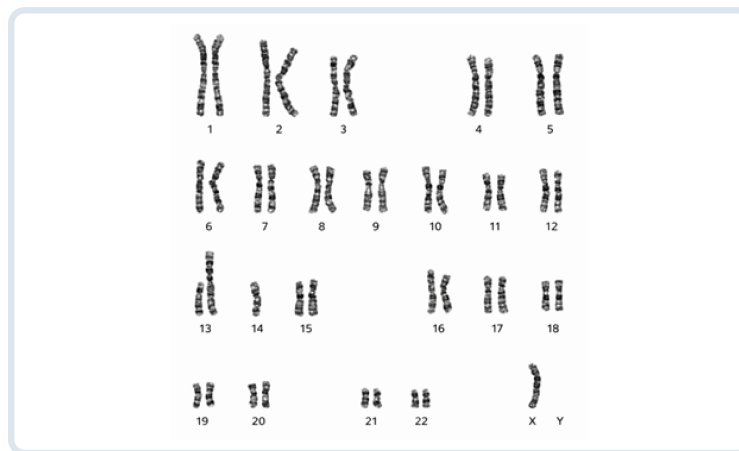
- Chronic lung infections
- Pancreatic insufficiency and malabsorption
- Meconium ileus in newborns

Why other options are incorrect:

- Achondroplasia: Autosomal dominant (FGFR3 mutation).
- Huntington's disease: Autosomal dominant (trinucleotide repeat disorder).
- Familial hypercholesterolemia: Typically autosomal dominant (LDL receptor defect).

QUESTION

A female patient with infertility presented to the clinic and underwent karyotyping. What is the likely diagnosis in the karyotype shown?



A Down syndrome

B Turner syndrome

CORRECT

C Klinefelter syndrome

D Edward syndrome

RIGHT ANSWER

OK

B. Turner syndrome

WHY THIS IS RIGHT

The karyotype shows monosomy X (45,X), which is diagnostic of Turner syndrome, a common chromosomal cause of female infertility.

- Absence of one X chromosome -> 45, X karyotype
- No Barr body present

Clinical features:

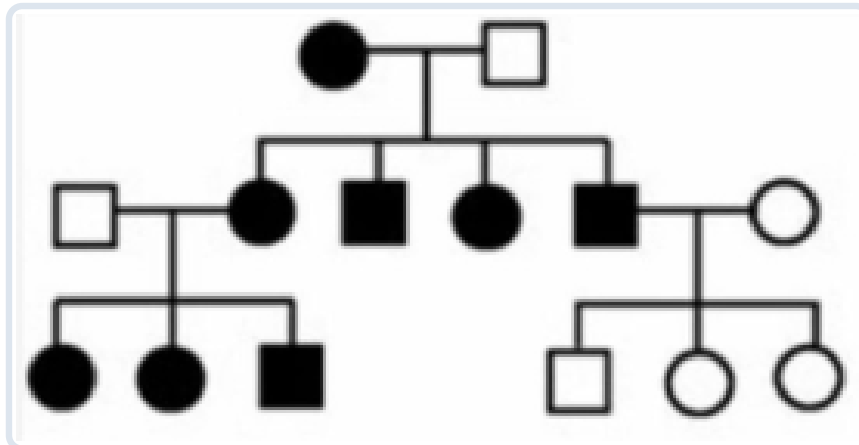
- Primary amenorrhea & infertility
- Streak ovaries (gonadal dysgenesis)
- Short stature
- Webbed neck
- Shield chest with widely spaced nipples

Why other options are incorrect:

- Down syndrome: Trisomy 21
- Klinefelter syndrome: 47,XXY (male infertility)
- Edward syndrome: Trisomy 18

QUESTION

Which of the following diseases is shown by the pedigree chart?



A Prader-Willi Syndrome

B Achondroplasia

C **Kearns-Sayre Syndrome**

CORRECT

D Williams syndrome

RIGHT ANSWER

OK

C. Kearns-Sayre Syndrome

WHY THIS IS RIGHT

The pedigree shows maternal transmission, where an affected mother passes the disease to all offspring, while affected fathers do not transmit it.

This inheritance pattern is characteristic of mitochondrial disorders.

Kearns-Sayre syndrome is a classic mitochondrial disease caused by mtDNA deletions.

Classic Features (Triad)

- Progressive external ophthalmoplegia
- Pigmentary retinopathy
- Cardiac conduction defects

Pattern	Key Feature	Example
-----	-----	-----
Mitochondrial	Maternal only	Kearns-Sayre
Autosomal dominant	Every generation	Achondroplasia
Imprinting	Parent-specific	Prader-Willi
Microdeletion	Sporadic	Williams

QUESTION

Match the following:

Column A	Column B
A. Mitochondrial inheritance	1. Spinobulbar dystrophia
B. XLR	2. Cystic fibrosis
C. AR	3. Leber's disease
D. Trinucleotide repeat	4. Duchene's muscular dystrophy

A A-3, B-4, C-2, D-1

CORRECT

B A-1, B-2, C-4, D-3

C A-2, B-3, C-1, D-4

D A-4, B-1, C-3, D-2

RIGHT ANSWER

OK A. A-3, B-4, C-2, D-1

WHY THIS IS RIGHT

Mitochondrial inheritance is classically seen in Leber's hereditary optic neuropathy due to mtDNA mutations. Inherited only from mother.

X-linked recessive disorders include Duchenne muscular dystrophy caused by dystrophin gene deletion. Features:

- Gowers' sign
- Calf pseudohypertrophy

Autosomal recessive inheritance is typical of cystic fibrosis due to CFTR mutation. Features:

- Thick mucus
- Lung infections
- Pancreatic insufficiency

Spinobulbar muscular atrophy is caused by trinucleotide (CAG) repeat expansion. Also called: Kennedy disease

Similar concept seen in:

- Huntington disease
- Fragile X

QUESTION

| All of the following are characteristic features of Turner syndrome, except:

A Prominent occiput

CORRECT

B Short 4th metacarpal

C Webbed neck

D Wide spaced nipples

RIGHT ANSWER

OK **A. Prominent occiput**

WHY THIS IS RIGHT

Turner syndrome (45,X) shows classic somatic features like webbed neck, shield chest with wide-spaced nipples, and short 4th metacarpal due to SHOX gene haploinsufficiency.

Classic Phenotype

- Short stature
- Webbed neck
- Shield chest
- Wide spaced nipples
- Streak ovaries -> primary amenorrhea

Skeletal abnormalities and lymphatic dysgenesis are characteristic.

Prominent occiput is not a recognized feature of Turner syndrome and is more associated with certain craniofacial anomalies in other syndromes.

A. Prominent occiput- Incorrect statement

- Not a feature of Turner syndrome
- Seen more in:
- Trisomy 18 (Edwards syndrome)
- Some neural tube defects

B. Short 4th metacarpal

- Gives knuckle dimpling when making a fist
- Also seen in:
- Pseudohypoparathyroidism

C. Webbed neck

- Due to cystic hygroma in fetal life
- Leaves behind lateral neck folds

QUESTION

A 16-year-old girl presents with polyuria, polydipsia, progressive vision loss, and a history of recurrent urinary tract infections. Further evaluation reveals optic atrophy, poorly controlled diabetes mellitus, and diabetes insipidus. Genetic testing confirms a diagnosis of DIDMOAD syndrome. Which of the following genes is most commonly associated with this condition?

A WFS1

CORRECT

B FOXP3

C HNF1A

D PDX1

RIGHT ANSWER

OK A. WFS1

WHY THIS IS RIGHT

- DIDMOAD syndrome (also known as Wolfram Syndrome) is a rare genetic disorder characterized by Diabetes Insipidus, Diabetes Mellitus, Optic Atrophy, and Deafness.
- It is most commonly caused by mutations in the WFS1 gene, which encodes wolframin, a protein involved in endoplasmic reticulum homeostasis.
- The syndrome typically presents in childhood or adolescence with diabetes mellitus and progressive optic atrophy.

Why not others (very brief):

- FOXP3: Causes IPEX syndrome (autoimmune disease).
- HNF1A: Causes MODY type 3 diabetes.
- PDX1: Causes pancreatic agenesis / MODY type 4.

QUESTION

A 12-year-old boy is brought to the clinic for evaluation of muscle weakness. The patient has a history of seizures. Examination shows decreased strength in the upper and lower extremities on the left compared to the right. Laboratory evaluation shows increased serum lactate levels both after exercise and at rest. His older sister is affected by the same disorder, but she displays very few symptoms. Which of the following is the most likely pedigree chart in the following?

A https://corebtr-assets-production.s3.ap-south-1.amazonaws.com/questions_image/option/01KMFBRYC8N53KXDX9E53ZPYJN.jpg

B https://corebtr-assets-production.s3.ap-south-1.amazonaws.com/questions_image/option/01KMFBRYGWP HH0SDJQ451C69MR.jpg **CORRECT**

C https://corebtr-assets-production.s3.ap-south-1.amazonaws.com/questions_image/option/01KMFBRYMVJ6JDGVJBHFQ7VQQ1.jpg

D https://corebtr-assets-production.s3.ap-south-1.amazonaws.com/questions_image/option/01KMFBRYRGSJG9E5BTREE94N61.jpg

RIGHT ANSWER

OK B. https://corebtr-assets-production.s3.ap-south-1.amazonaws.com/questions_image/option/01KMFBRYGWP HH0SDJQ451C69MR.jpg

WHY THIS IS RIGHT

The clinical features (muscle weakness, seizures, elevated lactate levels) suggest a mitochondrial disorder (e.g., mitochondrial myopathy such as MELAS). Mitochondrial diseases follow maternal inheritance because mitochondrial DNA (mtDNA) is transmitted only from the mother.

Pedigree characteristics of mitochondrial inheritance:

- Affected mother -> all children inherit the mutation
- Affected father -> none of the children are affected
- Variable severity among siblings due to heteroplasmy.

QUESTION

Which of the subsequent options is non-essential in the evaluation of mosaic Turner syndrome?

A Anti-nuclear antibodies

CORRECT

B Glucose tolerance test

C Audiometry

D Echocardiography

RIGHT ANSWER

OK A. Anti-nuclear antibodies

WHY THIS IS RIGHT

Evaluation of Turner syndrome (including mosaic Turner syndrome) focuses on identifying common associated systemic complications, as the condition can affect multiple organs.

Important investigations include:

- Echocardiography -> to detect congenital heart defects such as coarctation of the aorta or bicuspid aortic valve.
- Audiometry -> due to increased risk of sensorineural and conductive hearing loss.
- Glucose tolerance test -> because patients have a higher risk of insulin resistance and type 2 diabetes mellitus.

Anti-nuclear antibodies (ANA) testing is not routinely required in the evaluation of Turner syndrome, although autoimmune disorders can occur.

QUESTION

A 6-year-old child with known Down syndrome presents for routine follow-up. The mother reports poor weight gain and chronic bloating. Examination shows midface hypoplasia and a large tongue. Which of the following conditions is NOT associated with an increased risk in Down syndrome?

- A Celiac disease
- B Infantile spasms
- C Obstructive sleep apnea

D NF1

CORRECT

RIGHT ANSWER

OK

D. NF1

WHY THIS IS RIGHT

Children with Down syndrome (Trisomy 21) have an increased risk of several medical conditions due to immune dysregulation, craniofacial abnormalities, and genetic factors.

Common associations in Down syndrome:

- Celiac disease -> autoimmune condition causing malabsorption, poor weight gain, bloating
- Infantile spasms -> increased risk of epilepsy
- Obstructive sleep apnea -> due to macroglossia, midface hypoplasia, and hypotonia

Not associated:

- Neurofibromatosis type 1 (NF1) -> caused by NF1 gene mutation on chromosome 17, unrelated to trisomy 21.

QUESTION

All of the following genes have known associations with Down syndrome-related phenotypes EXCEPT:

A GATA1

B CRELD1

C APP

D RB1

CORRECT

RIGHT ANSWER

OK
D. RB1

WHY THIS IS RIGHT

Down syndrome (Trisomy 21) results from an extra copy of chromosome 21, leading to overexpression of several genes that contribute to its characteristic clinical features.

Genes associated with Down syndrome phenotypes:

- APP (Amyloid precursor protein) -> Located on chromosome 21; responsible for early-onset Alzheimer disease seen in Down syndrome.
- GATA1 -> Mutations associated with transient myeloproliferative disorder and acute megakaryoblastic leukemia in Down syndrome.
- CRELD1 -> Associated with congenital heart defects (especially atrioventricular septal defects) seen in Down syndrome.

Not associated:

- RB1 -> Located on chromosome 13; tumor suppressor gene linked to retinoblastoma, not Down syndrome.

QUESTION

A 6-month-old infant presents with recurrent seizures, irritability, poor feeding, and progressive microcephaly. Neuroimaging shows intracranial calcifications and white-matter abnormalities. CSF examination reveals elevated interferon- α levels. Genetic testing identifies a mutation in the TREX1 gene. Which of the following diagnoses best fits this clinical and genetic profile?

A Aicardi-Goutières syndrome

CORRECT

B Leigh syndrome

C Krabbe disease

D Zellweger syndrome

RIGHT ANSWER

OK **A. Aicardi-Goutières syndrome**

WHY THIS IS RIGHT

Aicardi-Goutières syndrome (AGS) is a rare genetic interferonopathy caused by mutations in genes involved in nucleic acid metabolism (commonly TREX1). It presents in infancy with features resembling congenital viral infection (TORCH-like syndrome).

- Early infancy onset
- Recurrent seizures and irritability
- Progressive microcephaly
- Intracranial calcifications and white-matter abnormalities on neuroimaging
- Elevated interferon- α in CSF (hallmark finding)

Why not the other options?

B. Leigh syndrome: Mitochondrial disorder with lactic acidosis and basal ganglia lesions, not intracranial calcifications or $\uparrow$ CSF interferon- α .

C. Krabbe disease: Lysosomal leukodystrophy causing irritability, stiffness, and demyelination, but no intracranial calcifications or interferon- α elevation.

D. Zellweger syndrome: Peroxisomal disorder with hypotonia, craniofacial dysmorphism, liver dysfunction, not TORCH-like calcifications with TREX1 mutation.

QUESTION

A 5-year-old child presents with exertional dyspnoea and a history of cyanotic spell. On examination, there is clubbing and a harsh systolic murmur heard at left upper sternal border. Chest X-ray is shown below. What is the most likely diagnosis?



A Tetralogy of Fallot

CORRECT

B TAPVC

C TGA

D COA

RIGHT ANSWER

OK

A. Tetralogy of Fallot

WHY THIS IS RIGHT

Tetralogy of Fallot is the most common cyanotic congenital heart disease beyond infancy, presenting with exertional dyspnea, cyanotic spells, clubbing, and a systolic murmur.

- Components (PROV):
- Pulmonary stenosis
- Right ventricular hypertrophy
- Overriding aorta
- Ventricular septal defect

Clinical features:

- Cyanotic (tet) spells due to increased right-to-left shunting
- Harsh systolic murmur at left upper sternal border
- Squatting relieves symptoms

Chest X-ray finding: "Boot-shaped heart" due to right ventricular hypertrophy

Differentiation:

- TAPVC: snowman sign
- TGA: egg-on-string appearance
- Coarctation: rib notching

QUESTION

| Which of the following is not a component of Tetralogy of Fallot (TOF)?

A ASD, left ventricular hypertrophy

CORRECT

B Pulmonary stenosis

C Overriding of aorta

D Right ventricular hypertrophy

RIGHT ANSWER

OK

A. ASD, left ventricular hypertrophy

WHY THIS IS RIGHT

Tetralogy of Fallot is a cyanotic congenital heart disease characterized by four structural abnormalities resulting in right-to-left shunting. Right ventricular outflow obstruction leads to right-to-left shunt and cyanosis

Severity of pulmonary stenosis determines clinical presentation

Components (remember: PROV):

- Pulmonary stenosis
- Right ventricular hypertrophy
- Overriding aorta
- Ventricular septal defect (VSD)

Why option A is incorrect:

- ASD is not a component of TOF
- Left ventricular hypertrophy is not seen; instead, right ventricular hypertrophy occurs

QUESTION

A one-month-old child presents with recurrent pneumonia and no cyanosis. On examination, a continuous murmur is heard below the left clavicle and a bounding pulse is present. What is the most likely diagnosis?

A ASD (Atrial Septal Defect)

B VSD (Ventricular Septal Defect)

C PDA (Patent Ductus Arteriosus)

CORRECT

D Coarctation of Aorta

RIGHT ANSWER

OK

C. PDA (Patent Ductus Arteriosus)

WHY THIS IS RIGHT

Patent ductus arteriosus is a congenital left-to-right shunt in which the ductus arteriosus fails to close after birth, leading to continuous blood flow from the aorta to the pulmonary artery.

- Continuous “machinery” murmur best heard below the left clavicle
- Bounding pulses and wide pulse pressure due to runoff of blood into pulmonary circulation
- Recurrent respiratory infections and signs of heart failure due to increased pulmonary blood flow
- Usually no cyanosis initially (acyanotic congenital heart disease)

Pathophysiology:

- Persistent connection between aorta and pulmonary artery -> left-to-right shunt
- Increased pulmonary blood flow -> pulmonary congestion -> recurrent pneumonia

Contrast:

- ASD: fixed split S2, no continuous murmur
- VSD: pansystolic murmur at left lower sternal border
- Coarctation of aorta: radio-femoral delay, upper limb hypertension, weak lower limb pulses

QUESTION

A 10-month-old baby is brought to the emergency room with bluish discoloration of the skin and rapid breathing. His mother reveals a history of congenital heart disease in the baby and adds that this episode was provoked by a fall leading to crying of the child. Which of the following is least likely to be used for managing this child?

- A Subcutaneous morphine
- B Intravenous sodium bicarbonate
- C Intravenous propranolol

D Intravenous nitroglycerin

CORRECT

RIGHT ANSWER

D. Intravenous nitroglycerin

WHY THIS IS RIGHT

A child with cyanosis triggered by crying in the setting of congenital heart disease suggests a hypercyanotic ("Tet") spell, commonly seen in Tetralogy of Fallot.

- Sudden increase in right ventricular outflow tract obstruction
- Increased right-to-left shunting across the VSD
- Reduced pulmonary blood flow -> acute hypoxia and cyanosis

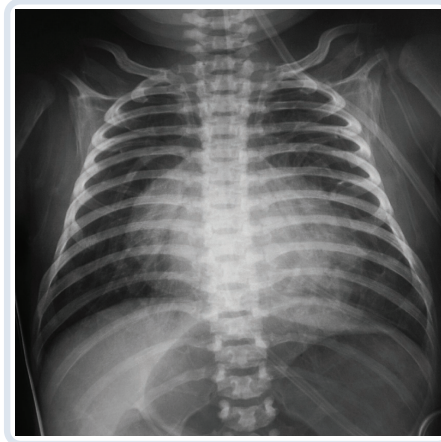
Management of a Tet spell includes:

- Knee-chest position -> increases systemic vascular resistance (SVR)
- Oxygen administration
- Morphine -> reduces agitation and catecholamine surge
- Propranolol -> relieves RV outflow tract spasm
- Sodium bicarbonate -> corrects metabolic acidosis

Nitroglycerin is least likely to be used because it decreases systemic vascular resistance, which would increase right-to-left shunting and worsen cyanosis.

QUESTION

Which of the following, all are findings of the condition in the below chest X-ray except:



A Ventriculo-arterial discordance

B Narrow vascular pedicle

C Septum-dependent

D Kerley lines

CORRECT

RIGHT ANSWER

OK

D. Kerley lines

WHY THIS IS RIGHT

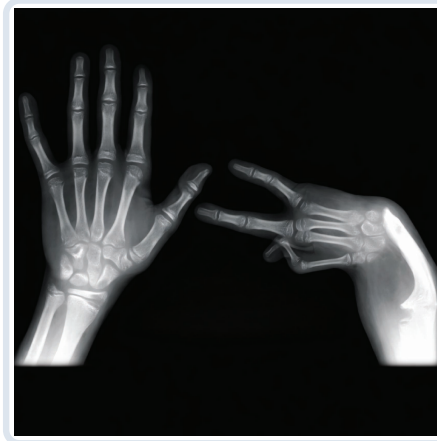
The chest X-ray shown demonstrates the classic “egg-on-a-string” appearance, which is characteristic of transposition of the great arteries (TGA).

- Ventriculo-arterial discordance: the aorta arises from the right ventricle and the pulmonary artery from the left ventricle.
- Narrow vascular pedicle (string sign) due to the anteroposterior relationship of the great vessels.
- Septum-dependent circulation: survival requires mixing through ASD, VSD, or PDA.

Kerley lines, however, represent interstitial pulmonary edema typically seen in left-sided heart failure or pulmonary venous hypertension, and are not a feature of TGA.

QUESTION

A 4-year-old child presents with an upper limb deformity as given in the image below. Examination shows a wide split and fixed second heart sound through all phases of respiration. Identify the heart disease likely to be present in this child.



A ASD

CORRECT

B Transposition of great vessels (TGA)

C Patent ductus arteriosus (PDA)

D VSD

RIGHT ANSWER

OK
A. ASD

WHY THIS IS RIGHT

The image shows an upper limb deformity consistent with radial ray defect (absent or hypoplastic radius/thumb). This limb anomaly is classically associated with Holt-Oram syndrome, an autosomal dominant disorder caused by TBX5 gene mutation.

Holt-Oram syndrome features:

- Upper limb abnormalities (especially radial ray defects)
- Congenital heart defects, most commonly atrial septal defect (ASD) or sometimes VSD

The clinical clue of a wide, fixed splitting of the second heart sound (S2) is characteristic of atrial septal defect (ASD) due to increased right-sided blood flow and delayed pulmonary valve closure.

QUESTION

| Which cardiac lesion is most commonly associated in this patient?



- A Tetralogy of Fallot
- B Endocardial cushion defect**
- C Coarctation of aorta
- D TGA

CORRECT

RIGHT ANSWER

- B. Endocardial cushion defect**

WHY THIS IS RIGHT

The image shows a child with features suggestive of Down syndrome (Trisomy 21), such as flat facial profile, upslanting palpebral fissures, epicanthal folds, and hypotonia.

Children with Down syndrome commonly have congenital heart defects, the most frequent being endocardial cushion defect (atrioventricular septal defect, AVSD).

Failure of fusion of the superior and inferior endocardial cushions leads to defects in:

- Atrial septum
- Ventricular septum
- Atrioventricular valves

This results in a complete AV canal defect, causing left-to-right shunting and early heart failure if untreated.

QUESTION

A 2-month-old boy is brought to the emergency department for evaluation of cyanosis. He became fussy and sweaty while taking his bottle, and his lips "turned blue" for several minutes during the feeding. The infant has had similar episodes during feeding and crying, but these resolved quickly. The patient was born at 39 weeks gestation, and his birth weight was average for gestational age. His weight gain has been slow, currently at the 5th percentile. On examination, the infant is ill-appearing, agitated, cyanotic, and tachypneic. Cardiac auscultation reveals a grade 2/6 crescendo-decrescendo systolic ejection murmur at the left upper sternal border and a single second heart sound. The patient is placed in a knee-chest position immediately. This maneuver improves the patient's condition predominantly by which of the following mechanisms?

A Decreased pulmonary vascular resistance

B Decreased systemic vascular resistance

C Increased systemic vascular resistance

CORRECT

D Increased systemic venous return

RIGHT ANSWER

OK C. Increased systemic vascular resistance

WHY THIS IS RIGHT

This infant's episodic cyanosis during feeding or crying that improves with the knee-chest position is characteristic of a hypercyanotic ("tet") spell seen in Tetralogy of Fallot.

During a tet spell:

- Increased right-to-left shunting across the ventricular septal defect occurs due to right ventricular outflow obstruction.
- This allows deoxygenated blood to bypass the lungs and enter systemic circulation, causing sudden cyanosis.

Knee-chest position mechanism:

- Compresses the femoral arteries -> increases systemic vascular resistance (SVR).
- Higher SVR reduces right-to-left shunting across the VSD.
- More blood is directed toward the pulmonary circulation for oxygenation, improving cyanosis.

QUESTION

A 9-month-old infant with known Tetralogy of Fallot suddenly becomes irritable and deeply cyanotic while crying. During the episode, pulse oximetry shows a drop in saturation and on auscultation the systolic murmur becomes faint. Which of the following is a clinical marker of a cyanotic spell?

- A improvement in SpO₂
- B disappearance of murmur.**
- C increase in S₂.
- D improvement in cyanosis.

CORRECT

RIGHT ANSWER

- OK
- B. disappearance of murmur.**

WHY THIS IS RIGHT

A cyanotic (hypercyanotic or “tet”) spell in Tetralogy of Fallot occurs due to sudden increase in right-to-left shunting across the VSD from worsening right ventricular outflow tract obstruction.

During a tet spell:

- Cyanosis worsens
- Oxygen saturation falls
- The systolic murmur becomes faint or disappears because less blood flows through the pulmonary outflow tract, reducing turbulence.

QUESTION

A term neonate becomes cyanotic shortly after birth. Saturations do not improve with 100% oxygen (hyperoxia test negative). Which lesion most likely benefits from maintaining ductal patency with prostaglandins?

- A** Total anomalous pulmonary venous return (TAPVR) obstructed
- B** Tetralogy of Fallot
- C** Transposition of great arteries (TGA) CORRECT
- D** Tricuspid atresia

RIGHT ANSWER

- OK** **C. Transposition of great arteries (TGA)**

WHY THIS IS RIGHT

A neonate with severe cyanosis that does not improve with 100% oxygen (negative hyperoxia test) suggests a cyanotic congenital heart disease with right-to-left shunt.

In Transposition of the Great Arteries (TGA), the systemic and pulmonary circulations run in parallel rather than in series, so survival depends on mixing of oxygenated and deoxygenated blood through the ductus arteriosus, ASD, or VSD.

Maintaining ductal patency with prostaglandin E1 (PGE1) allows mixing between the circulations and improves systemic oxygenation until definitive surgical correction (arterial switch operation).

Why not others?

- A. Obstructed TAPVR: Requires urgent surgery; prostaglandin does not help much.
- B. Tetralogy of Fallot: Cyanosis due to RV outflow obstruction; PDA not essential for survival.
- D. Tricuspid atresia: PGE₁ may help but not the classic duct-dependent mixing lesion.

QUESTION

A child presents with recurrent sinusitis and repeated lung infection. Genetic testing shows a mutation in CFTR gene. Which test is considered gold standard to confirm the diagnosis?

A Sweat Chloride Test

CORRECT

B Lung Biopsy

C Chest X-ray

D Nasal Swab Culture

RIGHT ANSWER

OK

A. Sweat Chloride Test

WHY THIS IS RIGHT

A child with recurrent sinopulmonary infections and a CFTR gene mutation is suspected to have cystic fibrosis. The gold standard test for confirming the diagnosis is the sweat chloride test, which demonstrates elevated chloride levels due to defective chloride transport.

- Cystic fibrosis is caused by mutation in the CFTR gene leading to thick, viscous secretions
- Common features include recurrent sinusitis, chronic lung infections, malabsorption, and failure to thrive
- Sweat chloride test:
 - ≥ 60 mmol/L is diagnostic
 - 30-59 mmol/L is intermediate
- Newborn screening and genetic testing support diagnosis but confirmation is by sweat test

QUESTION

A 4-month-old infant is brought to the emergency department with a respiratory rate of 66 breaths per minute, along with chest indrawing and nasal flaring. There are no other significant findings. What is the most likely diagnosis?

A Pneumonia

CORRECT

B Severe Pneumonia

C Severe Asthma

D Common Cold

RIGHT ANSWER

OK A. Pneumonia

WHY THIS IS RIGHT

According to IMNCI guidelines, a 2-12 month old infant with fast breathing (respiratory rate $\geq 50/\text{min}$) is classified as pneumonia. In this case, a respiratory rate of 66/min indicates fast breathing for age.

- Age-specific fast breathing:
 - <2 months: $\geq 60/\text{min}$
 - 2-12 months: $\geq 50/\text{min}$
 - 1-5 years: $\geq 40/\text{min}$
- Chest indrawing in infants may be present due to compliant chest wall and does not always indicate severe disease
- Severe pneumonia is diagnosed when there are danger signs such as inability to feed, lethargy, convulsions, or central cyanosis

QUESTION

| In which disease among the following, Chloride level in sweat is evaluated?

- A Phenylketonuria
- B Cystic fibrosis**
- C Gaucher's disease
- D Osteogenesis imperfecta

CORRECT

RIGHT ANSWER

- B. Cystic fibrosis**

WHY THIS IS RIGHT

Sweat chloride test is the gold standard diagnostic test for cystic fibrosis, reflecting defective chloride transport due to CFTR mutation.

- CFTR dysfunction -> impaired chloride reabsorption in sweat glands -> increased chloride concentration in sweat
- Sweat chloride level >60 mmol/L is diagnostic (in appropriate clinical setting)

Clinical relevance:

- Used in children with recurrent respiratory infections, failure to thrive, or steatorrhea
- Helps confirm diagnosis early and guide management

QUESTION

| Choose the true statement regarding CF (Cystic Fibrosis) :

A Chloride secretion is decreased in sweat and increased in pancreatic fluid

B Chloride secretion is increased in sweat and decreased in pancreatic fluid

CORRECT

C Chloride secretion is decreased in both sweat and pancreatic fluid

D Chloride secretion is increased in both sweat and pancreatic fluid

RIGHT ANSWER

OK

B. Chloride secretion is increased in sweat and decreased in pancreatic fluid

WHY THIS IS RIGHT

Cystic fibrosis is caused by CFTR channel dysfunction leading to abnormal chloride transport in different tissues, producing opposite effects in sweat glands and exocrine organs.

- In sweat glands: defective reabsorption of chloride -> increased chloride in sweat (basis of sweat chloride test)
- In pancreas and other exocrine glands: decreased chloride secretion -> thick, viscous secretions causing ductal obstruction

Clinical correlation:

- High sweat chloride is diagnostic
- Pancreatic insufficiency leads to malabsorption and failure to thrive

QUESTION

A 2-year-old child is brought to the emergency department with fever, hoarse voice, and a barking cough for 2 days. On examination, the child has inspiratory stridor that worsens at night. There is no drooling, and the child is able to swallow.

Which of the following is the most likely causative organism?

- A** Haemophilus influenzae
- B** Streptococcus
- C** Parainfluenza virus
- D** Corynebacterium diphtheriae

CORRECT

RIGHT ANSWER

OK C. Parainfluenza virus

WHY THIS IS RIGHT

- Barking cough + stridor + hoarseness -> classic triad
- Age: 6 months - 3 years
- No drooling -> rules out epiglottitis
- Most common cause of croup = Parainfluenza virus (Type 1)
- X-ray: Steeple sign
- Treatment: Steroids (dexamethasone) ± nebulized adrenaline

QUESTION

| All are seen in Cystic fibrosis except:

A Biliary atresia

CORRECT

B Meconium ileus

C Nasal polyp

D Bleeding diathesis

RIGHT ANSWER

OK A. Biliary atresia

WHY THIS IS RIGHT

Cystic fibrosis is an autosomal recessive disorder caused by CFTR gene mutation leading to thick, viscous secretions affecting multiple organs.

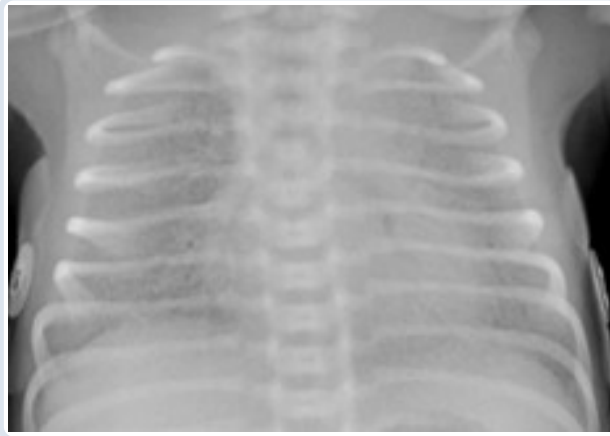
- Gastrointestinal: meconium ileus in neonates, pancreatic insufficiency causing malabsorption
- Respiratory: recurrent infections, bronchiectasis, nasal polyps
- Hepatobiliary: can cause biliary cirrhosis but not biliary atresia
- Hematological: fat-soluble vitamin (vitamin K) deficiency -> bleeding tendency

Why biliary atresia is not seen:

- Biliary atresia is a separate obstructive condition of the extrahepatic bile ducts, not related to CFTR dysfunction (bile duct obstruction is seen in CF, not atresia)

QUESTION

A 34-week newborn presents with breathing difficulty that improves with FiO_2 at 30%. What is the most likely diagnosis?



A Hyaline membrane disease

CORRECT

B Transient tachypnea of newborn

C Meconium aspiration syndrome

D None of the above

RIGHT ANSWER

OK

A. Hyaline membrane disease

WHY THIS IS RIGHT

Hyaline membrane disease (neonatal respiratory distress syndrome) occurs due to surfactant deficiency (produced by type II pneumocytes), most commonly in preterm infants, leading to alveolar collapse and impaired gas exchange.

- Seen in preterm or late preterm infants (like 34 weeks)
- Presents with respiratory distress soon after birth that improves with supplemental oxygen
- Chest X-ray shows diffuse reticulogranular (ground-glass) appearance with air bronchograms and reduced lung volumes
- Managed with oxygen, CPAP, and exogenous surfactant if needed
- Antenatal steroids reduce risk in preterm delivery

Differentiation:

- Transient tachypnea of newborn: hyperinflation, fluid in fissures, milder disease
- Meconium aspiration syndrome: patchy infiltrates, hyperinflation, term/post-term

QUESTION

An 8-year-old child presents with prolonged cough for 7 months, streaky hemoptysis for 1 month and fever for 4 days. Chest X-ray is shown below. What is the diagnosis?



A Congenital lobar hyperinflation

B Lung abscess

C **Round pneumonia**

CORRECT

D Congenital thoracic malformation

RIGHT ANSWER

OK

C. **Round pneumonia**

WHY THIS IS RIGHT

Round pneumonia is a form of bacterial pneumonia seen mainly in children, characterized by a round or oval opacity on chest X-ray, which can mimic a lung mass.

Why it occurs in children:

- Children have poorly developed collateral ventilation (pores of Kohn and canals of Lambert).
- Infection therefore spreads centrifugally, producing a well-circumscribed round consolidation.

Clinical features:

- Fever, cough
- Sometimes hemoptysis
- Round homogeneous opacity, usually in the posterior lower lobes on CXR.

QUESTION

A 5-year-old boy presents with a persistent productive cough for the past 3 days, producing thick yellow sputum. He has a history of recurrent sinus infections and failure to thrive since infancy. Previous investigations revealed high sweat chloride content and pancreatic insufficiency. Which of the following findings is most consistent with this patient's underlying disorder?

A Abnormal post-translational processing of a transmembrane protein

CORRECT

B Decreased transcription of a transmembrane protein

C Increased conductivity of a transmembrane chloride channel

D Presence of a truncated transmembrane protein on the cell surface

RIGHT ANSWER

OK **A. Abnormal post-translational processing of a transmembrane protein**

WHY THIS IS RIGHT

No explanation was provided in the source quiz.

QUESTION

A 6-year-old male child was presented with a past of persistent cough and foul-smelling stools. His sibling, who had experienced comparable symptoms, passed away a few years earlier. Tests including the sweat chloride test and CFTR gene analysis were conducted and cystic fibrosis was diagnosed. What will happen to the transport of Chloride ions in sweat gland and pancreatic ducts respectively?

A Decrease, decrease

CORRECT

B Decrease, increase

C Increase, decrease

D Increase, increase

RIGHT ANSWER

OK A. Decrease, decrease

WHY THIS IS RIGHT

Cystic fibrosis is caused by mutations in the CFTR (cystic fibrosis transmembrane conductance regulator) gene, which encodes a chloride channel present in epithelial cells.

Defective CFTR leads to impaired chloride transport in multiple organs:

- Sweat glands: Normally, CFTR reabsorbs chloride (and sodium) from sweat. In cystic fibrosis, chloride reabsorption is decreased, resulting in high chloride levels in sweat (basis of the sweat chloride test).
- Pancreatic ducts: CFTR normally secretes chloride and bicarbonate into the ductal lumen to keep secretions fluid. In cystic fibrosis, chloride secretion is decreased, leading to thick pancreatic secretions and duct obstruction, causing malabsorption and steatorrhea.

QUESTION

| Which of the following is not seen in a child with cystic fibrosis?

- A Sweat chloride test chloride conc of 70 mEq/L
- B Increase immunoreactive trypsinogen level
- C Hyperkalaemia**
- D Contraction alkalosis

CORRECT

RIGHT ANSWER

- OK **C. Hyperkalaemia**

WHY THIS IS RIGHT

Cystic fibrosis (CF) is caused by mutation of the CFTR gene, leading to defective chloride transport across epithelial cells. This results in thick secretions in the lungs, pancreas, and other organs and characteristic electrolyte abnormalities.

Typical findings in CF include:

- Elevated sweat chloride (>60 mEq/L) on the sweat chloride test (diagnostic).
- Increased immunoreactive trypsinogen (IRT) on newborn screening due to pancreatic injury.
- Electrolyte loss in sweat (Na^+ and Cl^-) leading to hyponatremic, hypochloremic dehydration and metabolic alkalosis (contraction alkalosis).

Because of salt loss, patients usually develop hypokalemia, not hyperkalemia.

QUESTION

| Δ F508 mutation has a functional defect on the CFTR channel. It leads to:

- A** Defective regulation: CFTR not activated by adenosine triphosphate or cyclic adenosine monophosphate
- B** Lack of protein production
- C** Reduced chloride transport through CFTR at the apical membrane
- D** Defect in protein trafficking with ubiquitination and degradation in the endoplasmic reticulum/Golgi body CORRECT

RIGHT ANSWER

- OK** **D. Defect in protein trafficking with ubiquitination and degradation in the endoplasmic reticulum/Golgi body**

WHY THIS IS RIGHT

The Δ F508 mutation is the most common mutation causing cystic fibrosis. It results in misfolding of the CFTR protein, leading to defective trafficking from the endoplasmic reticulum to the cell membrane. The abnormal protein is recognized as defective, ubiquitinated, and degraded in the ER/Golgi, preventing it from reaching the apical membrane.

Why not the other options?

- A. Defective regulation: Seen in Class III CFTR mutations (gating defect), not Δ F508.
- B. Lack of protein production: Seen in Class I mutations due to nonsense/frameshift mutations.
- C. Reduced chloride transport at membrane: Seen in Class IV mutations (conductance defect).

QUESTION

A 10-year-old child presents with diarrhea and weight loss. On examination, the height and weight are lesser than expected. Laboratory investigations were positive for class II HLA-DQ2. Which of the following will you advise the child?

A Fat-free diet

B Lactose-free diet

C Low carbohydrate diet

D **Gluten-free diet**

CORRECT

RIGHT ANSWER

OK **D. Gluten-free diet**

WHY THIS IS RIGHT

Celiac disease is an autoimmune disorder triggered by gluten in genetically predisposed individuals, commonly associated with HLA-DQ2 (or DQ8), leading to malabsorption and growth failure.

- Presents with chronic diarrhea, weight loss, and short stature
- Associated with HLA-DQ2/DQ8 positivity
- Pathology: villous atrophy in small intestine

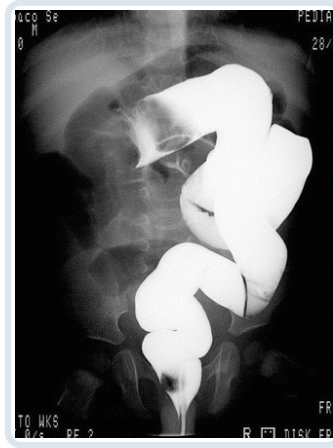
Management:

- Lifelong strict gluten-free diet (avoid wheat, barley, rye)
- Leads to symptom resolution and catch-up growth

Clinical importance: Early dietary intervention prevents complications like malnutrition and delayed growth

QUESTION

A baby presented with abdominal pain. On examination, a mass is palpated in the right lumbar region. A barium enema is done, and the image is given below. What is the likely diagnosis?



A Intussusception

CORRECT

B Volvulus

C Duodenal atresia

D Intestinal obstruction

RIGHT ANSWER

OK

A. Intussusception

WHY THIS IS RIGHT

Intussusception is the most common cause of intestinal obstruction in infants and young children, caused by telescoping of one segment of bowel into another.

- Presents with intermittent abdominal pain, vomiting, and a palpable abdominal mass (often right sided)
- May have "red currant jelly" stools (blood + mucus)
- Barium or air enema shows characteristic "coil spring" or "claw" sign

Clinical significance:

- Barium/air enema is both diagnostic and therapeutic
- Early diagnosis prevents complications like bowel ischemia and perforation

Differentiation:

- Volvulus: bilious vomiting, corkscrew appearance
- Duodenal atresia: double bubble sign in neonates

QUESTION

1-month-old baby came with complaints of projectile vomiting with an olive-shaped mass. X-ray abdomen was done which is shown. What is the diagnosis?



A Pyloric stenosis

CORRECT

B Intestinal obstruction

C Duodenal atresia

D Jejunal atresia

RIGHT ANSWER

OK **A. Pyloric stenosis**

WHY THIS IS RIGHT

Congenital hypertrophic pyloric stenosis is a common cause of non-bilious projectile vomiting in early infancy due to hypertrophy of the pyloric muscle causing gastric outlet obstruction.

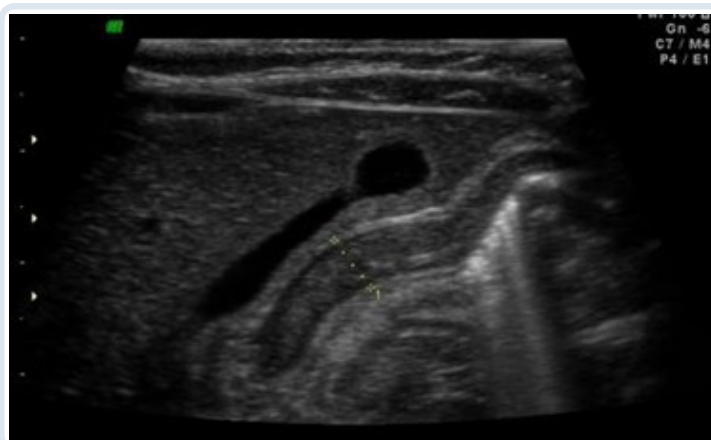
- Presents at 2-6 weeks with projectile non-bilious vomiting and an “olive-shaped” mass
- X-ray may show a distended stomach with paucity of distal bowel gas
- Ultrasound is the investigation of choice (elongated, thickened pylorus)

Clinical significance:

- Leads to hypochloremic, hypokalemic metabolic alkalosis
- Management requires fluid and electrolyte correction followed by pyloromyotomy

QUESTION

A 1-month-old baby presents with an olive-shaped mass and recurrent vomiting. An ultrasound (USG) was performed, as shown below. What is your diagnosis?



A CHPS (Congenital Hypertrophic Pyloric Stenosis)

CORRECT

B Intussusception

C Meckel's Diverticulum

D None of the above

RIGHT ANSWER

OK **A. CHPS (Congenital Hypertrophic Pyloric Stenosis)**

WHY THIS IS RIGHT

Congenital hypertrophic pyloric stenosis is a common cause of gastric outlet obstruction in early infancy, characterized by hypertrophy of the pyloric muscle.

- Presents at 2-6 weeks with non-bilious projectile vomiting and an "olive-shaped" palpable mass
- Infant is typically hungry after vomiting
- Ultrasound is the investigation of choice showing elongated and thickened pylorus (target or donut sign)
- Hypochloremic, hypokalemic metabolic alkalosis due to repeated vomiting

Management:

- Initial stabilization with fluids and electrolyte correction
- Definitive treatment is pyloromyotomy

QUESTION

A 24-hour-old neonate presents with severe respiratory distress and is admitted to the Intensive Care Unit (ICU). A chest X-ray of the neonate is provided. What is the most probable diagnosis?



A Congenital Pulmonary Airway Malformation (CPAM)

B Congenital Diaphragmatic Hernia (CDH)

CORRECT

C Congenital Lobar Emphysema

D Neonatal Pneumonia

RIGHT ANSWER

OK B. Congenital Diaphragmatic Hernia (CDH)

WHY THIS IS RIGHT

Congenital diaphragmatic hernia is a neonatal surgical emergency characterized by herniation of abdominal contents into the thoracic cavity, leading to lung compression and severe respiratory distress.

- Presents soon after birth with respiratory distress, scaphoid abdomen, and decreased breath sounds
- Chest X-ray shows bowel loops in the thorax with mediastinal shift
- Most commonly left-sided (Bochdalek hernia)

Clinical importance:

- Causes pulmonary hypoplasia and pulmonary hypertension
- Immediate management includes endotracheal intubation and gastric decompression (NG tube)
- Bag-mask ventilation is contraindicated

QUESTION

A 1-day old neonate presented with scaphoid abdomen, respiratory distress and mediastinal shift. All of the following can be done except?

- A O2 by mask
- B Positive pressure ventilation by ventilator
- C ET intubation
- D Bag and mask ventilation

CORRECT

RIGHT ANSWER

- OK
- D. Bag and mask ventilation

WHY THIS IS RIGHT

Congenital diaphragmatic hernia presents with respiratory distress, scaphoid abdomen, and mediastinal shift due to herniation of abdominal contents into the thoracic cavity causing lung compression.

- Immediate management focuses on preventing further gastric distension and lung compression
- Bag and mask ventilation is contraindicated because it forces air into the stomach, worsening respiratory compromise
- Endotracheal intubation with controlled ventilation is preferred
- Nasogastric tube should be inserted for gastric decompression

QUESTION

| What is the fluid of choice in pediatric patients?

A RL

B DNS

CORRECT

C N/2 saline

D 5% dextrose

RIGHT ANSWER

OK

B. DNS

WHY THIS IS RIGHT

In pediatric patients, maintenance fluid therapy should provide both water and glucose to prevent hypoglycemia and ketosis; hence dextrose-containing fluids are commonly used.

- DNS (dextrose normal saline) provides glucose along with sodium, making it suitable for many pediatric maintenance needs
- Children have limited glycogen stores and are prone to hypoglycemia during illness or fasting
- Pure crystalloids like normal saline or Ringer lactate do not provide glucose
- 5% dextrose alone lacks electrolytes and is not appropriate for routine use
- In emergency resuscitation (e.g., shock), isotonic fluids like normal saline are preferred initially

QUESTION

Which of the following is the best indicator of some dehydration?

A Skin pinch

B Sunken eyeballs

C Urine output

D Irritable status

CORRECT

RIGHT ANSWER

OK

D. Irritable status

WHY THIS IS RIGHT

Some dehydration" is diagnosed when ≥ 2 of the following are present:

- Restless / irritable (early & sensitive sign)
- Sunken eyes
- Thirsty, drinks eagerly
- Skin pinch goes back slowly

Why Irritability is BEST indicator

- Reflects early CNS response to dehydration
- Appears before severe signs
- More reliable clinically than isolated physical signs

Parameters	No Dehydration	Some Dehydration
Appearance	Well, alert	Restless, irritable
Eyes	Normal	Sunken
Thirst	Drinks normally, not thirsty	Thirsty, drinks eagerly
Skin pinch	Goes back quickly (<1 second)	Goes back slowly (1 second)

QUESTION

A 3-week-old infant presents with non-projectile, non-bilious vomiting and a normal abdominal examination. What is the most likely diagnosis?

A GERD

CORRECT

B CHPS

C Cow milk protein allergy

D Intestinal obstruction

RIGHT ANSWER

OK
A. GERD

WHY THIS IS RIGHT

Key Clinical Reasoning

- Age: 3 weeks (very early infancy)
- Vomiting:
- Non-projectile
- Non-bilious
- Abdominal exam: Normal

This pattern is classic for physiological reflux / GERD in infants

Why NOT others?

B. CHPS (Congenital Hypertrophic Pyloric Stenosis)

- Projectile vomiting (key feature)
- Usually presents at 3-6 weeks
- May have palpable olive, visible peristalsis

C. Cow milk protein allergy

- Associated with diarrhea, blood in stool, eczema
- Not isolated vomiting

D. Intestinal obstruction

- Bilious vomiting
- Abdominal distension

QUESTION

| All of the following statements regarding Hirschsprung disease are true except:

- A** Failure of migration of ganglion cells.
- B** Colonic segment proximal to the affected area is constricted. CORRECT
- C** Failure of passage of meconium in the postnatal period.
- D** Loss of coordinated peristalsis.

RIGHT ANSWER

- OK** **B. Colonic segment proximal to the affected area is constricted.**

WHY THIS IS RIGHT

Hirschsprung disease is caused by failure of neural crest cell migration, leading to absence of ganglion cells in the distal bowel (aganglionosis). This results in loss of coordinated peristalsis and functional obstruction.

Key concepts:

- The affected distal segment is spastic and constricted due to aganglionosis
- The proximal segment becomes dilated due to accumulation of intestinal contents (megacolon)
- Newborns typically present with failure to pass meconium within the first 48 hours
- There is absence of the rectoanal inhibitory reflex

QUESTION

A boy born to a normal father and mother had Hemophilia. Detailed history taking revealed that the mother's side had a family history of similar affected individuals. The children born to this boy are likely to have which of the following?

- A All boys and girl children equally affected
- B 50% boys affected and no girl children unaffected
- C No boy affected and 50% girl children are carrier
- D 25% boy affected and 25% girl children are carrier

CORRECT

RIGHT ANSWER

- OK
- C. No boy affected and 50% girl children are carrier

WHY THIS IS RIGHT

Hemophilia is an X-linked recessive disorder affecting males who inherit the mutant X chromosome. An affected male transmits his X chromosome to all daughters and Y chromosome to all sons. Hence, none of the sons are affected as they receive the father's Y chromosome.

All daughters receive the mutant X, making 100% daughters carriers; effectively, 50% of total children are carrier females.

Genotype setup:

- Affected male (boy): X^hY
- Assume wife is normal: X^HX^H
- Punnett Square:

Mother

$X^H X^H$

Father $X^h X^HX^h X^HX^h$

$Y X^HY X^HY$

- Interpretation:
- Daughters (X^HX^h) -> All are carriers (100%)
- Sons (X^HY) -> All are normal (0% affected)
- Final conclusion:
- No male child affected
- All female children are carriers

QUESTION

A 1-month-old infant, born at 35 weeks gestation, presents with blood-streaked, mucous-filled diarrhea after taking a standard cow's milk-based formula. Symptoms resolved upon switching the formula. If a biopsy was performed during the symptomatic period, which of the following histological findings would most likely be observed?

- A Neutrophilic crypt abscesses in the colon
- B Eosinophilic infiltration in the distal colon**
- C Hemorrhagic necrosis of the intestinal wall
- D Presence of heterotopic gastric mucosa in the distal ileum

CORRECT

RIGHT ANSWER

- OK **B. Eosinophilic infiltration in the distal colon**

WHY THIS IS RIGHT

A young infant with blood-streaked, mucous-filled stools after ingestion of cow's milk formula that improves after eliminating cow's milk is characteristic of food protein-induced allergic proctocolitis (cow's milk protein allergy).

- It is a non-IgE-mediated hypersensitivity reaction to cow's milk protein.
- The immune response leads to inflammation of the distal colon and rectum.

Histological finding:

- Eosinophilic infiltration of the colonic mucosa, particularly in the distal colon, often with mucosal edema and mild inflammation.

QUESTION

| Which of the following is not a feature of severe dehydration in children?

A Thirsty child

CORRECT

B Drowsy child

C Skin pinch retracts slowly

D Sunken fontanelle

RIGHT ANSWER

OK A. Thirsty child

WHY THIS IS RIGHT

Assessment of dehydration in children is based on clinical signs and is classified as no dehydration, some dehydration, and severe dehydration.

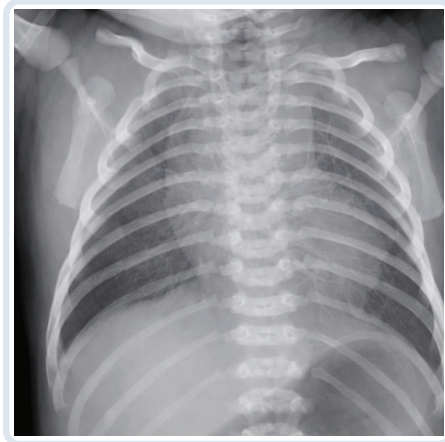
Features of severe dehydration include:

- Lethargy or drowsiness
- Very sunken eyes or sunken anterior fontanelle
- Skin pinch returning very slowly (>2 seconds)
- Poor drinking or inability to drink

A thirsty child who drinks eagerly is typically a sign of some (moderate) dehydration, not severe dehydration.

QUESTION

A newborn baby presents with continuous dribbling of saliva and choking while feeding. CXR is shown here. Identify the type of TEF:



A A

B B

C C

CORRECT

D E

RIGHT ANSWER

OK C. C

WHY THIS IS RIGHT

A newborn with excessive salivation, choking during feeds, and inability to swallow suggests esophageal atresia with tracheoesophageal fistula (TEF).

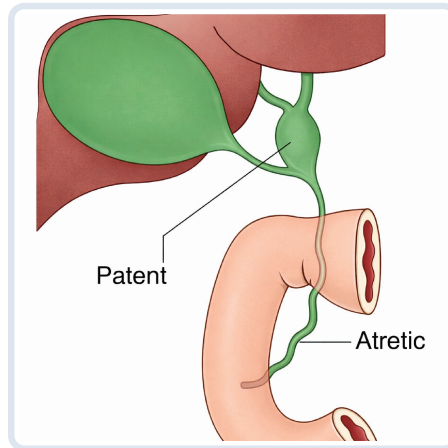
On chest X-ray, the key diagnostic clues are:

- Nasogastric/orogastric tube coiled in the upper esophageal pouch, indicating proximal esophageal atresia.
- Presence of gas in the stomach and intestines, indicating a distal tracheoesophageal fistula that allows air from the trachea to enter the gastrointestinal tract.

This combination corresponds to Type C TEF (proximal esophageal atresia with distal tracheoesophageal fistula), which is the most common type (~85-90%).

QUESTION

What is the Kasai grade of EHBA depicted in the image?



A Grade 1

CORRECT

B Grade 2a

C Grade 2b

D Grade 3

RIGHT ANSWER

OK A. Grade 1

WHY THIS IS RIGHT

The Kasai classification of extrahepatic biliary atresia (EHBA) is based on the level of obstruction in the extrahepatic biliary tree.

- Type/Grade 1: Atresia of the common bile duct, while the proximal bile ducts (common hepatic duct and intrahepatic ducts) remain patent.
- Type/Grade 2: Atresia of the common hepatic duct.
 - o 2a: Common hepatic duct atresia with patent cystic duct and common bile duct.
 - o 2b: Atresia of common hepatic duct, cystic duct, and common bile duct.
- Type/Grade 3: Atresia at the porta hepatis with complete obstruction of extrahepatic ducts (most common type).

In the image, the common bile duct is atretic while proximal ducts are patent, corresponding to Kasai Grade 1 EHBA.

QUESTION

A newborn infant fails to pass meconium for the first 48 hours. On examination, the abdomen is distended and emesis has occurred overnight. What should be the next diagnostic step?

A Anorectal manometry

B Contrast imaging of the lower GI tract

CORRECT

C Genotype of the child for cystic fibrosis

D Sweat chloride assay

RIGHT ANSWER

OK

B. Contrast imaging of the lower GI tract

WHY THIS IS RIGHT

Failure to pass meconium within the first 48 hours of life with abdominal distension and vomiting suggests neonatal intestinal obstruction. The initial diagnostic step is contrast imaging of the lower gastrointestinal tract (contrast enema).

- It helps differentiate common causes of distal neonatal obstruction, such as:
 - o Hirschsprung disease (transition zone between dilated proximal bowel and narrow distal aganglionic segment)
 - o Meconium ileus (microcolon with inspissated meconium; often associated with cystic fibrosis)
 - o Small left colon syndrome
- It is both diagnostic and sometimes therapeutic (e.g., helps evacuate meconium in meconium ileus).

Why not the other options initially?

- Anorectal manometry -> used later to confirm Hirschsprung disease by demonstrating absence of the rectoanal inhibitory reflex.
- CF genotype testing or sweat chloride test -> performed if meconium ileus is suspected after imaging findings.

QUESTION

A 9-month-old infant presents with 2 days of profuse diarrhea. On exam the child is lethargic, has delayed capillary refill, cool extremities, and weak pulses. The child has vomited multiple times in the ER and is noted to have a palpable sausage-shaped abdominal mass and currant-jelly stools. Which of the following clinical scenarios would still be an indication for ORS rather than a contraindication?

- A Hypovolemic shock with poor perfusion
- B Depressed level of consciousness with aspiration risk
- C Suspected intussusception with bowel obstruction

D Mild dehydration with preserved thirst and oral intake

CORRECT

RIGHT ANSWER

OK D. Mild dehydration with preserved thirst and oral intake

WHY THIS IS RIGHT

Oral rehydration solution (ORS) is the treatment of choice for mild to moderate dehydration when the child is alert and able to drink, as intestinal sodium-glucose cotransport allows effective fluid absorption even during diarrhea.

ORS is appropriate when:

- Mild dehydration
- Child is conscious and able to drink
- No intestinal obstruction
- Vomiting is mild and not persistent

Contraindications to ORS:

- Hypovolemic shock
- Severe dehydration requiring IV fluids
- Depressed consciousness/aspiration risk
- Intestinal obstruction (e.g., intussusception)
- Persistent severe vomiting preventing intake

QUESTION

A 3-month-old infant presents with persistent jaundice, pale stools, and pruritus. Examination reveals a systolic murmur and butterfly-shaped thoracic vertebrae on X-ray. Which diagnosis is most likely?

- A** Biliary atresia
- B** Alagille syndrome
- C** Progressive familial intrahepatic cholestasis
- D** Neonatal hepatitis

CORRECT

RIGHT ANSWER

- OK** **B. Alagille syndrome**

WHY THIS IS RIGHT

Alagille syndrome is an autosomal dominant disorder (JAG1 mutation) characterized by paucity of intrahepatic bile ducts, leading to chronic cholestasis in infancy.

Key Clinical Features (Classic clues):

- Persistent jaundice with pale stools and pruritus (cholestasis)
- Congenital heart defects (commonly pulmonary artery stenosis -> systolic murmur)
- Butterfly vertebrae on X-ray
- Characteristic facies (broad forehead, deep-set eyes, pointed chin)

QUESTION

A 4-year-old child is brought to the clinic with a low grade fever, sore mouth and painful vesicular rash on the hands, feet and inside the mouth as shown in image below. The child attends daycare and similar cases have been reported among classmates. What is the most likely causative organism?



A Coxsackie A16

CORRECT

B EBV

C VZV

D Coxsackie B

RIGHT ANSWER

OK

A. Coxsackie A16

WHY THIS IS RIGHT

A child with low-grade fever, painful oral ulcers, and vesicular lesions on the hands and feet in a daycare setting is characteristic of hand, foot and mouth disease caused most commonly by Coxsackievirus A16.

- Spread is via feco-oral and respiratory routes, leading to outbreaks in children
- Typical features include oral ulcers, vesicles on palms and soles, and mild fever
- Illness is self-limiting and resolves within about a week

Differentials:

- Varicella: lesions in different stages, predominantly on trunk
- Epstein-Barr virus: pharyngitis with lymphadenopathy, no vesicular rash
- Coxsackie B -> Myocarditis, pleurodynia

QUESTION

A 8-year-old child presents with enuresis. The parents mention the child had a fall from bed once but has no other significant medical history. There are no signs of urinary tract infections or neurological deficits. What is the best next step in management?

A Behavioural modification

CORRECT

B Start anticholinergic drugs

C Prescribe Oxybutinin

D Start desmopressin therapy

RIGHT ANSWER

OK

A. Behavioural modification

WHY THIS IS RIGHT

Primary nocturnal enuresis in children is most often a benign, functional condition and is managed initially with non-pharmacological measures.

- Diagnosis is clinical after ruling out UTI, diabetes, and neurological causes
- Most children have normal bladder function and outgrow the condition

First-line management:

- Behavioral modification:
- Limit evening fluid intake
- Timed voiding before sleep
- Positive reinforcement (reward system)
- Enuresis alarm (most effective long-term)

Pharmacological therapy:

- Desmopressin or anticholinergics are reserved for refractory cases or specific indications

QUESTION

A 10-year-old presents with edema and anasarca. A diagnosis of minimal change disease is made. Which of the following is true about this condition?

A Light microscopy shows effacement of podocytes

B Good response to steroids

CORRECT

C Most common in adults

D Non-selective proteinuria

RIGHT ANSWER

OK **B. Good response to steroids**

WHY THIS IS RIGHT

Minimal change disease is the most common cause of nephrotic syndrome in children and is characterized by excellent response to corticosteroid therapy.

- Pathology:
- Light microscopy: appears normal
- Electron microscopy: effacement of podocyte foot processes
- Proteinuria is selective (mainly albumin)
- Most common in children, not adults
- Presents with edema, proteinuria, hypoalbuminemia
- Steroid-sensitive with good prognosis

Why other options are incorrect:

- Podocyte effacement is seen on electron microscopy, not light microscopy
- It is most common in children
- Proteinuria is selective, not non-selective

QUESTION

A male child presented with arthralgia and abdominal pain. On examination, there was palpable purpura over the lower limbs. There is a past history of upper respiratory tract infection prior to the onset of presenting symptoms. Which of the following is the treatment for this condition?

A Azathioprine

B Methotrexate

C Cyclosporine

D Glucocorticoids

CORRECT

RIGHT ANSWER

OK

D. Glucocorticoids

WHY THIS IS RIGHT

IgA vasculitis (Henoch-Schönlein purpura) is a small-vessel vasculitis seen in children, typically following an upper respiratory infection, presenting with palpable purpura, arthralgia, abdominal pain, and possible renal involvement.

- Classic tetrad:
- Palpable purpura (non-thrombocytopenic, mainly lower limbs)
- Arthralgia/arthritis
- Abdominal pain
- Renal involvement (hematuria/proteinuria)

Treatment: Usually self-limiting; supportive care is mainstay. Glucocorticoids are used in moderate to severe cases (significant abdominal pain, renal involvement)

Why others are incorrect:

- Azathioprine, methotrexate, cyclosporine are not first-line and used only in severe/refractory cases



QUESTION

Identify the condition:



A Bladder exstrophy

CORRECT

B Omphalocele

C Persistent vitellointestinal duct

D Gastroschisis

RIGHT ANSWER

OK

A. Bladder exstrophy

WHY THIS IS RIGHT

Bladder exstrophy is a congenital defect with absence of the lower anterior abdominal wall, exposing the posterior bladder wall.

It is commonly associated with epispadias, widened pubic symphysis, and urinary incontinence.

Early surgical reconstruction is required to preserve renal function and continence.

QUESTION

A 10-month-old infant was brought with complaints of jerking movement of limbs towards the body. On examination, there is a regression of developmental milestones. Electroencephalogram shows hypsarrhythmia. Which of the following is the drug of choice in this condition?

A Phenytoin

B Adrenocorticotrophic hormone (ACTH)

CORRECT

C Levetiracetam

D Phenobarbitone

RIGHT ANSWER

OK

B. Adrenocorticotrophic hormone (ACTH)

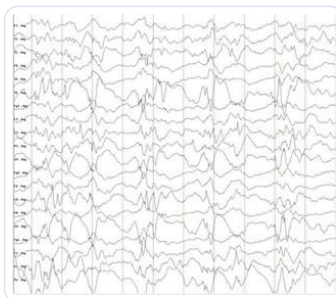
WHY THIS IS RIGHT

Infantile spasms (West syndrome) is an epileptic encephalopathy characterized by the triad of epileptic spasms, developmental regression, and hypsarrhythmia on EEG.

- Age: typically 3-12 months
- Seizures: brief flexor or extensor spasms ("jackknife seizures")
- EEG: chaotic, high-amplitude pattern called hypsarrhythmia

Treatment:

- First-line: ACTH (adrenocorticotrophic hormone)
- Alternative: oral steroids or vigabatrin (especially in tuberous sclerosis)
- Early treatment is crucial to improve neurodevelopmental outcomes
- Delay can lead to long-term cognitive impairment



QUESTION

A 2-year-old child with stable vitals and ECG showing PSVT. What is the appropriate management?

- A** Cardioversion
- B** Adenosine 0.3 mg per kg
- C** Adenosine 0.2 mg/kg
- D** **Adenosine 0.1 mg/kg**

CORRECT

RIGHT ANSWER

- OK** **D. Adenosine 0.1 mg/kg**

WHY THIS IS RIGHT

Paroxysmal supraventricular tachycardia (PSVT) in a hemodynamically stable child is treated first with rapid-acting AV nodal blocking agents, with adenosine as the drug of choice.

- First-line dose of adenosine: 0.1 mg/kg rapid IV bolus (maximum 6 mg)
- If ineffective, second dose: 0.2 mg/kg (maximum 12 mg)
- Must be given rapidly followed by saline flush due to very short half-life

Clinical approach:

- Ensure child is hemodynamically stable
- Attempt vagal maneuvers first (if feasible)
- Use synchronized cardioversion only if unstable or drug therapy fails

QUESTION

A newborn baby is born with a congenital defect. Identify the defect shown in the image:



A Omphalocele

B Gastroschisis

C Bladder Exstrophy

CORRECT

D Meckel's Diverticulum

RIGHT ANSWER

OK

C. Bladder Exstrophy

WHY THIS IS RIGHT

Bladder exstrophy is characterized by absence of the lower anterior abdominal wall, with exposed bladder mucosa. It is associated with epispadias, widened pubic symphysis, and urinary incontinence. This defect differs from omphalocele and gastroschisis, which involve bowel herniation rather than bladder exposure.

QUESTION

2-year-old was brought to the ED following choking on a peanut. The child was unresponsive. What is the next step for managing this patient?

- A** Give 5 back slaps
- B** Give abdominal thrusts
- C** Endotracheal intubation

D Chest compressions

CORRECT

RIGHT ANSWER

OK

D. Chest compressions

WHY THIS IS RIGHT

- Unresponsive child with choking (foreign body airway obstruction)
- Treat as cardiac arrest -> start CPR immediately
- Begin with 30 chest compressions
- Then open airway -> look for visible foreign body
- Give 2 rescue breaths
- Repeat cycles
- Hemlich manoeuvre/ Back slaps / abdominal thrusts -> only if conscious child

QUESTION

A 3-year-old child is brought to the emergency department with a generalized seizure following a high-grade fever. Which of the following is the first-line drug of choice for seizure control in this acute febrile episode?

A Diazepam

CORRECT

B Valproate

C Fosphenytoin

D Doxycycline /Amoxicillin

RIGHT ANSWER

OK

A. Diazepam

WHY THIS IS RIGHT

Acute management of febrile seizures focuses on rapid termination of the seizure using benzodiazepines as first-line therapy.

- Diazepam (IV/rectal) or lorazepam (IV) are first-line drugs for ongoing seizures
- They act by enhancing GABA activity -> rapid seizure control
- Most febrile seizures are self-limiting, but if seizure persists >5 minutes, treatment is required

Clinical approach:

- Ensure airway, breathing, circulation first
- Give benzodiazepine if seizure is prolonged
- Long-term antiepileptic therapy is not indicated for simple febrile seizures

Why other options are incorrect:

- Valproate and fosphenytoin are second-line agents if seizures persist
- Antibiotics have no role unless infection is specifically indicated

QUESTION

The following finding can be seen in:



A Molluscum contagiosum

B Parvovirus B19 infection

CORRECT

C HFMD

D Kawasaki disease

RIGHT ANSWER

OK B. Parvovirus B19 infection

WHY THIS IS RIGHT

Parvovirus B19 infection classically presents with erythema infectiosum showing a "slapped cheek" rash. It may be followed by a lacy rash on the trunk and limbs. The virus can also cause aplastic crisis in hemolytic anemias and hydrops fetalis in pregnancy, adding diagnostic value.

QUESTION

| Causes of non-immune hydrops fetalis are all except:

A ABO incompatibility

CORRECT

B Congestive cardiac failure

C Parvovirus infection

D Chromosomal anomalies

RIGHT ANSWER

OK

A. ABO incompatibility

WHY THIS IS RIGHT

Hydrops fetalis is defined as abnormal accumulation of fluid in two or more fetal compartments and is classified into immune and non-immune causes.

- Immune hydrops is caused by red cell alloimmunization (e.g., Rh incompatibility; ABO incompatibility is also immune-mediated, though usually milder)
- Non-immune hydrops (more common) includes:
 - Cardiac causes (congestive cardiac failure)
 - Infections (e.g., parvovirus B19 causing fetal anemia)
 - Chromosomal anomalies (e.g., Turner syndrome, trisomies)

Why option A is correct:

- ABO incompatibility is an immune cause, not a non-immune cause

QUESTION

A child came to OPD with fever for 6 days with strawberry tongue, conjunctival congestion, and peeling of skin. What should be the ideal treatment?

A IVIG

CORRECT

B Aspirin

C Plenty of fluids

D Cephalosporins

RIGHT ANSWER

OK

A. IVIG

WHY THIS IS RIGHT

Kawasaki disease is an acute vasculitis of childhood characterized by prolonged fever (>5 days), conjunctival congestion, strawberry tongue, mucocutaneous changes, and desquamation.

- Diagnosis is clinical: fever ≥ 5 days + mucocutaneous features
- Risk of coronary artery aneurysms is the most serious complication

Management:

- First-line treatment is intravenous immunoglobulin (IVIG) given early (within 10 days of illness) as it rapidly decreases inflammation and reduces risk of coronary artery aneurysms.
- High-dose aspirin is also used as adjunct therapy

Why others are incorrect:

- Aspirin alone is insufficient
- Fluids are supportive but not definitive
- Antibiotics (cephalosporins) have no role

QUESTION

Cannabinoids are approved in all of the following except:

- A** Dravet Syndrome
- B** Lennox-Gastaut Syndrome
- C** Tuberous Sclerosis

D Rett Syndrome

CORRECT

RIGHT ANSWER

OK **D. Rett Syndrome**

WHY THIS IS RIGHT

Cannabinoids (particularly cannabidiol) are approved for specific treatment-resistant epileptic syndromes in children, where they help reduce seizure frequency. Cannabidiol acts by modulating neuronal excitability and reducing seizure activity. Used as adjunct therapy in refractory epilepsy

Approved indications:

- Dravet syndrome
- Lennox-Gastaut syndrome
- Tuberous sclerosis complex-associated epilepsy

Why Rett syndrome is incorrect:

- Rett syndrome is a neurodevelopmental disorder with seizures, but cannabinoids are not an approved standard indication

QUESTION

| Which of the following statements is not true about febrile seizure?

- A Age 6 months - 5 years
- B Simple febrile seizure lasts for <15 minutes
- C No need for long term antiepileptics

D 54% recurrence

CORRECT

RIGHT ANSWER

OK D. 54% recurrence

WHY THIS IS RIGHT

Febrile seizures are benign, age-dependent seizures occurring in children between 6 months and 5 years in association with fever, without evidence of intracranial infection or metabolic cause.

- Simple febrile seizures are generalized, last less than 15 minutes, and do not recur within 24 hours
- Long-term antiepileptic therapy is not indicated due to the benign nature and low risk of epilepsy
- Recurrence risk is about 30-35%, not as high as 54%

QUESTION

| All of the following are incorrect about resuscitation of a critically sick child, except?

- A** Preferred route of adrenaline intraosseous
- B** Compression to ventilation ratio should be 30:2 with 2 rescuers
- C** Chest compression depth should be one-third of A-P diameter of chest CORRECT
- D** Injection Adenosine 0.01 ml/kg IV or IO (1:10,000)

RIGHT ANSWER

- OK** **C. Chest compression depth should be one-third of A-P diameter of chest**

WHY THIS IS RIGHT

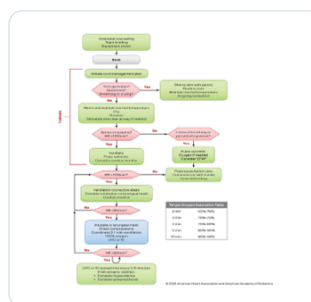
In pediatric resuscitation, effective chest compressions are critical and should be delivered to a depth of one-third of the anteroposterior diameter of the chest to ensure adequate cardiac output.

- Compression depth: one-third of chest A-P diameter (correct statement)
- Compression-ventilation ratio:
 - Single rescuer: 30:2
 - Two rescuers: 15:2 (not 30:2)
- Preferred route for emergency drugs is IV; intraosseous is used when IV access is not available (not preferred primarily)
- Adrenaline dose: 0.01 mg/kg (0.1 ml/kg of 1:10,000), not adenosine

Why other options are incorrect:

- IO is an alternative route, not the preferred first line if IV is available
- 30:2 ratio is for single rescuer, not two rescuers
- Adenosine dose and indication are incorrect in this context (used for SVT, not cardiac arrest)

NRP 2025:



QUESTION

A 2 yr old child presented with fever, thrombocytopenia, hematuria. The mother tells you that the baby also suffered from diarrhea a few days back. Which is the most common organism for likely cause?

A E Coli 0157:H7

CORRECT

B Salmonella

C Shigella

D Yersinia

RIGHT ANSWER

OK

A. E Coli 0157:H7

WHY THIS IS RIGHT

Hemolytic uremic syndrome (HUS) is a common cause of acute kidney injury in young children, classically occurring after a diarrheal illness caused by Shiga toxin-producing *Escherichia coli*, most commonly serotype O157:H7.

- Triad: microangiopathic hemolytic anemia, thrombocytopenia, and acute kidney injury (hematuria)
- Typically follows bloody diarrhea ("D+ HUS")
- Shiga toxin damages endothelial cells -> platelet activation and microthrombi formation
- Management is mainly supportive (fluids, dialysis if needed); antibiotics are generally avoided

Why other options are less likely:

- Shigella can also cause HUS but is less common
- Salmonella and Yersinia are not typical causes of HUS

QUESTION

A child is NS1 antigen positive and presents with fever and warning signs. Lab findings show hematocrit of 41% and platelet count of 50,000 per microliter. What is the most appropriate next step in the management?

- A Platelet transfusion.
- B Start IV fluids with crystalloids at 7 ml/kg/hour.**
- C Start IV fluids with crystalloids at 10-20 ml/kg/hour.
- D Start IV fluids with colloids at 7 ml/kg/hour.

CORRECT

RIGHT ANSWER

OK

B. Start IV fluids with crystalloids at 7 ml/kg/hour.

WHY THIS IS RIGHT

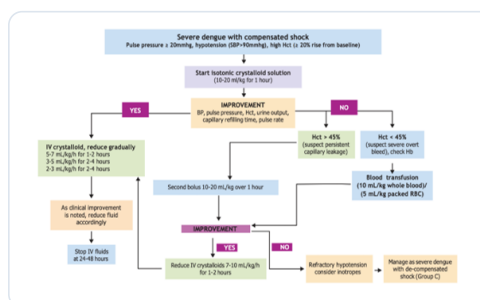
Dengue with warning signs requires careful fluid management using isotonic crystalloids to prevent progression to shock while avoiding fluid overload.

- Warning signs include abdominal pain, persistent vomiting, rising hematocrit, and thrombocytopenia
- Hematocrit of 41% with low platelets suggests plasma leakage phase
- First-line management is controlled IV fluid therapy with crystalloids (normal saline or Ringer lactate) at 5-7 ml/kg/hour. It helps maintain intravascular volume without causing fluid overload

Why others are incorrect:

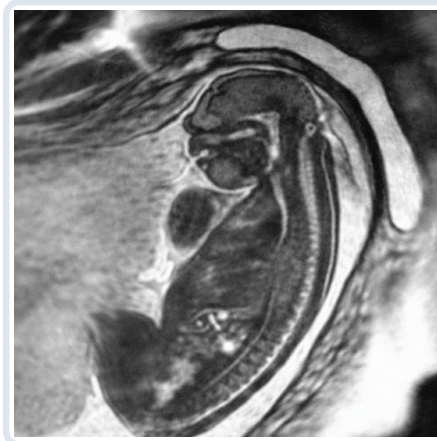
- Platelet transfusion is not indicated unless there is significant bleeding
- 10-20 ml/kg/hour is used in dengue shock (more severe cases)
- Colloids are reserved for patients not responding to crystalloids or with severe shock

Screenshots from national Dengue management guidelines:



QUESTION

| What is the likely diagnosis on the fetal MRI?



A Congenital diaphragmatic hernia

B CPAM

C **Anencephaly**

CORRECT

D Twin to twin transfusion syndrome

RIGHT ANSWER

OK C. **Anencephaly**

WHY THIS IS RIGHT

Anencephaly is a severe neural tube defect caused by failure of closure of the anterior (rostral) neuropore during early embryonic development (around day 25 of gestation). Often detected on prenatal ultrasound or fetal MRI. Associated with polyhydramnios due to impaired fetal swallowing

Risk factors:

- Folate deficiency
- Maternal diabetes
- Certain medications (e.g., valproate)

Periconceptual folic acid supplementation significantly reduces the risk of neural tube defects.

Why not other options:

- A. Congenital diaphragmatic hernia: Shows abdominal organs in the thoracic cavity with mediastinal shift, not absence of cranial vault.
- B. CPAM (Congenital pulmonary airway malformation): Cystic lung lesion in the fetal chest.
- D. Twin-to-twin transfusion syndrome: Occurs in monochorionic twin pregnancies, not a single fetus brain defect.

QUESTION

A 10-minute-old boy is being evaluated in the delivery room. The patient was born at term to a 30-year-old primigravida woman. Pregnancy was unremarkable, but labor was complicated by recurrent late decelerations necessitating vacuum assistance for vaginal delivery. Apgar scores were 6 and 8 at 1 and 5 minutes, respectively. Temperature is 37° C (98.6° F), pulse is 170/min, and respirations are 40/min. Scalp examination shows large, fluctuant swelling at the occiput that extends bilaterally to the ears, superiorly toward the crown, and inferiorly into the nape of the neck. This patient's hemorrhage is most likely located between which of the following structures?

A Arachnoid mater and dura mater

B Skull and periosteum

C Dura mater and periosteum

D Periosteum and galea aponeurosis

CORRECT

RIGHT ANSWER

OK D. Periosteum and galea aponeurosis

WHY THIS IS RIGHT

The newborn has a large, fluctuant scalp swelling that crosses suture lines and spreads widely over the scalp, which is characteristic of a subgaleal hemorrhage.

Subgaleal hemorrhage:

- Often associated with vacuum-assisted delivery
- Blood accumulates in the potential space between the periosteum of the skull and the galea aponeurotica
- The swelling is diffuse and crosses suture lines

Clinical importance:

- This space can hold a large volume of blood, leading to significant blood loss and hypovolemic shock in neonates.

QUESTION

A 13-month-old girl is brought to the OPD by her mother due to concern about bruising. The mother first noticed bruising around her daughter's eyes 2 weeks ago after the patient returned from a weekend at her father's house. She recently started walking and falls frequently; the bruising was attributed to bumping her head on a coffee table. The bruising has since been persistent, and there is no other notable bruising on her body. The patient has no fever, vomiting, diarrhea, cough, or congestion. The girl's parents are divorced, and she spends most weekends with her father. Weight is 9 kg down from 9.5 kg a month ago. On examination, the patient has rapid, jerking movements of both eyes with infraorbital ecchymoses bilaterally. The abdomen is mildly distended with a 6-cm palpable, firm mass anterior to the left flank. Which of the following is the most likely diagnosis?

A Child abuse

B Leukaemia

C Neuroblastoma

CORRECT

D Von Willebrand disease

RIGHT ANSWER

OK C. Neuroblastoma

WHY THIS IS RIGHT

Neuroblastoma is the most common extracranial solid tumor of childhood, arising from neural crest cells of the sympathetic nervous system, often in the adrenal medulla.

- Abdominal mass (often firm and irregular)
- Periorbital ecchymosis ("raccoon eyes") due to orbital metastasis
- Opsoclonus (rapid, chaotic eye movements) from paraneoplastic syndrome
- Weight loss and abdominal distension

Why not other options:

- A. Child abuse: Periorbital bruising could occur, but opsoclonus (rapid jerky eye movements) and a firm abdominal mass strongly suggest a tumor.
- B. Leukemia: Causes pallor, generalized bruising, hepatosplenomegaly, not a localized abdominal mass with raccoon eyes.
- D. Von Willebrand disease: Causes mucocutaneous bleeding, not orbital ecchymosis with abdominal mass.

QUESTION

Which of the following is not a method of administering free-flow oxygen in a neonate?

A Bag and mask ventilation with oxygen inlet

B Face mask

C Tubing near mouth and nose

D Bag and mask ventilation without oxygen inlet

CORRECT

RIGHT ANSWER

OK D. Bag and mask ventilation without oxygen inlet

WHY THIS IS RIGHT

Free-flow oxygen in neonates refers to providing supplemental oxygen without delivering positive pressure ventilation. Common methods of giving free-flow oxygen include:

- Face mask
- Oxygen tubing held near the mouth and nose ("blow-by oxygen")
- Self-inflating bag and mask with an oxygen inlet (without squeezing the bag)

A self-inflating bag and mask without an oxygen inlet cannot deliver oxygen effectively and therefore cannot provide free-flow oxygen.

QUESTION

An unconscious child is brought to the casualty. What is the correct sequence of the management?

1. Assess breathing
2. Assess pulse
3. Assess response
4. Start compressions
5. Bag and mask ventilation

A 3-1-2-5-4

B 1-2-3-4-5

C 3-1-2-4-5

CORRECT

D 1-2-4-3-5

RIGHT ANSWER

OK C. 3-1-2-4-5

WHY THIS IS RIGHT

Initial management of an unconscious child follows the Pediatric Basic Life Support (PBLIS) sequence, focusing on rapid assessment and initiation of resuscitation.

Correct sequence:

1. Assess response -> Check if the child is conscious or responds to verbal/painful stimuli.
2. Assess breathing -> Look for normal breathing.
3. Assess pulse -> Check for a pulse (usually carotid or femoral) for ≤ 10 seconds.
4. Start chest compressions -> If pulse is absent or $< 60/\text{min}$ with signs of poor perfusion.
5. Provide bag-mask ventilation -> Deliver assisted ventilation along with compressions.

QUESTION

Which of the following increase the risk of recurrence of febrile seizures?

1. Age < 1 year
2. Fever more than 102 F
3. Duration of fever < 24 h
4. Duration of fever > 48 h

A 1, 2, 4

B 1, 4

C 1, 3

CORRECT

D 1, 3, 4

RIGHT ANSWER

OK C. 1, 3

WHY THIS IS RIGHT

The risk of recurrence of febrile seizures is influenced by several clinical factors related to the child's age and fever characteristics.

Established risk factors for recurrence include:

- Younger age (<1 year) at the first febrile seizure
- Short duration of fever (<24 hours) before seizure onset
- Low-grade fever at the time of seizure
- Family history of febrile seizures

QUESTION

A 4-year-old male child with a body weight of 15kg and height of 100cm is admitted with renal failure. His blood urea was 100 mg/dL and serum creatinine was 1 mg/dL. What is the closest calculated eGFR in the patient?

A 33 ml/min/1.73 m² BSA

B 40 ml/min/1.73 m² BSA

CORRECT

C 55 ml/min/1.73 m² BSA

D 80 ml/min/1.73 m² BSA

RIGHT ANSWER

OK B. 40 ml/min/1.73 m² BSA

WHY THIS IS RIGHT

In children, glomerular filtration rate (GFR) is estimated using the Schwartz formula:

- k = 0.413 (for children)
- Height is in cm
- Height = 100 cm
- Serum creatinine = 1 mg/dL

Thus, the closest value is 40 ml/min/1.73 m².

$$eGFR = \frac{k \times \text{Height (cm)}}{\text{Serum creatinine (mg/dL)}}$$

$$eGFR = \frac{0.413 \times 100}{1} = 41.3 \approx 40 \text{ ml/min/1.73 m}^2$$

QUESTION

A 12-year-old boy is brought to the emergency department with complaints of sudden jerks in the morning causing him to drop objects. Identify the correct statements regarding this condition.

1. EEG shows generalized 4-5 Hz polyspike and slow-wave discharges
2. CACNB4, CLNC2 and EJM2 genes are mutated
3. Onset is classically during early infancy
4. KCNQ2 and SLC2A1 genes are mutated
5. Precipitated by sleep deprivation

A 1, 3, 4

B 1, 2, 5

CORRECT

C 3, 4, 5

D 1, 2, 3, 5

RIGHT ANSWER

OK B. 1, 2, 5

WHY THIS IS RIGHT

The presentation of sudden morning jerks causing the child to drop objects is characteristic of Juvenile Myoclonic Epilepsy (JME).

- Typical onset: adolescence (around 12-18 years), not infancy.
- Seizure type: myoclonic jerks, usually soon after waking.
- EEG finding: generalized 4-6 Hz polyspike-and-slow-wave discharges.
- Triggers: commonly sleep deprivation, stress, or alcohol.
- Genetics: mutations reported in genes such as CACNB4, CLCN2, and EJM2.
- KCNQ2 and SLC2A1 mutations -> False (associated with other epilepsies)

QUESTION

A 14-year-old girl is brought to the clinic by her mother for a yearly physical examination. The patient feels well but is worried that she has not yet started puberty. Temperature is 36.7 C (98 F), blood pressure is 152/91 mm Hg, pulse is 75/min, and respirations are 18/ min. Physical examination is significant for a lack of secondary sexual characteristics; a blind vagina is noted on pelvic examination. Laboratory studies reveal hypokalemia and low testosterone and estradiol levels. Cytogenetic analysis shows a 46, XY karyotype. This patient most likely has deficiency of which of the following enzymes?

A 5 alpha-reductase

B 11 beta-hydroxylase

C 17 alpha-hydroxylase

CORRECT

D 21 alpha-hydroxylase

RIGHT ANSWER

OK

C. 17 alpha-hydroxylase

WHY THIS IS RIGHT

17 α -hydroxylase deficiency is a rare form of congenital adrenal hyperplasia characterized by impaired synthesis of cortisol and sex steroids (androgens and estrogens).

Key features include:

- 46, XY individual with undervirilization -> female or ambiguous external genitalia and lack of secondary sexual characteristics
- Blind vagina due to absence of androgen-driven male genital development
- Low testosterone and estradiol levels
- Hypertension and hypokalemia due to excess mineralocorticoid activity ($\uparrow$ deoxycorticosterone)

QUESTION

A 12-year-old girl is brought to the emergency department by her parents for assessment of confusion and rapid breathing. Three days ago the patient developed rhinorrhea, cough, and fever, which have since resolved. The parents say that over the past 48 hours the patient has had increased urination, abdominal pain, and fatigue progressing to somnolence. On physical examination, she is ill-appearing with tachypnea, subcostal retractions, and dry mucous membranes. There is diffuse abdominal tenderness without rebound. Laboratory results are as follows:

Hematocrit-42%

Leukocytes-13,000/mm³

Serum Sodium-129 mEq/L

Potassium-4.8 mEq/L

Chloride-98 mEq/L

Bicarbonate-9 mEq/L

Blood urea nitrogen-24 mg/dL

Creatinine-1.2 mg/dL

Glucose-450 mg/dL

Which of the following is most likely decreased in this patient?

- A Blood renin activity
- B Circulating free fatty acids
- C Hypothalamic vasopressin production

D Total body potassium

CORRECT

RIGHT ANSWER

D. Total body potassium

WHY THIS IS RIGHT

This patient has diabetic ketoacidosis (DKA), suggested by:

- Hyperglycemia (glucose 450 mg/dL)
- Metabolic acidosis (bicarbonate 9 mEq/L)
- Polyuria, dehydration, abdominal pain, and Kussmaul breathing

In DKA, insulin deficiency leads to:

- Increased lipolysis → elevated free fatty acids
- Increased ketone production → metabolic acidosis
- Osmotic diuresis from hyperglycemia, causing loss of water and electrolytes in urine

Although serum potassium may appear normal or elevated, there is a significant depletion of total body potassium due to:

- Urinary potassium loss
- Shift of potassium from intracellular to extracellular space during acidosis

QUESTION

| All the following PFT findings in a child are suggestive of asthma except:

- A** Increase in FEV₁ > 12% after salbutamol inhalation
- B** FEV₁/FVC < 80%
- C** Day-night variation of FEV₁ > 15% CORRECT
- D** Decrease in FEV₁ > 15% after exercise

RIGHT ANSWER

- OK** **C. Day-night variation of FEV₁ > 15%**

WHY THIS IS RIGHT

Pulmonary function tests (PFTs) in asthma typically show reversible airway obstruction and bronchial hyperresponsiveness.

Typical PFT findings suggestive of asthma include:

- Reversibility with bronchodilator: Increase in FEV₁ ≥12% after salbutamol inhalation.
- Obstructive pattern: Reduced FEV₁/FVC ratio (<80%).
- Exercise-induced bronchoconstriction: Decrease in FEV₁ ≥10-15% after exercise.

Although asthma symptoms often show diurnal variation, the diagnostic criterion commonly used is diurnal variation in peak expiratory flow (PEF) >10-20%, not FEV₁ variation.

QUESTION

A 5-month-old girl is brought to the physician for a weight check. The patient has been evaluated several times for poor weight gain. Current weight is <5th percentile; length and head circumference have been tracking along the 25th percentile. The infant appears thin, but the remainder of the physical examination is unremarkable. Newborn screening results were normal. Laboratory results are as follows:

Serum Chemistry:

- Sodium: 140 mEq/L
- Potassium: 3 mEq/L
- Chloride: 121 mEq/L
- Blood urea nitrogen: 10 mg/dL
- Creatinine: 0.5 mg/dL
- Calcium: 9 mg/dL
- Glucose: 98 mg/dL

Arterial Blood Gases:

- pH: 7.21
- PaCO₂: 31 mm Hg
- Bicarbonate: 14 mEq/L

Urinalysis:

- pH: 7.9
- Potassium: Normal
- Sodium: Normal

Which of the following is the most likely cause of this patient's failure to thrive?

- A** Cystic fibrosis
- B** Gastroesophageal reflux
- C** Insufficient caloric intake

- D** Fanconi disease

CORRECT

RIGHT ANSWER

- OK** D. Fanconi disease

WHY THIS IS RIGHT

Failure to thrive with metabolic acidosis and electrolyte abnormalities in an infant suggests a renal tubular disorder.

- Metabolic acidosis: pH 7.21, HCO₃⁻ 14 mEq/L
- Hyperchloremia (Cl⁻ 121 mEq/L) -> indicates normal anion gap (hyperchloremic) metabolic acidosis
- Hypokalemia (K⁺ 3 mEq/L)
- Alkaline urine (pH 7.9) despite systemic acidosis

These findings indicate renal tubular dysfunction, where the kidney fails to properly reabsorb bicarbonate and other solutes.

Fanconi syndrome is a proximal renal tubular disorder characterized by impaired reabsorption of:

- Bicarbonate
- Glucose
- Phosphate
- Amino acids
- Uric acid

Loss of these substances leads to metabolic acidosis, electrolyte abnormalities, and poor growth (failure to thrive).

QUESTION

| Which of these is incorrect about Wilms' tumor?

- A** Can invade into the IVC
- B** Presents in children < 5 years of age
- C** Calcification is uncommon

D Lung metastasis is rare

CORRECT

RIGHT ANSWER

OK **D. Lung metastasis is rare**

WHY THIS IS RIGHT

Wilms tumor (nephroblastoma) is the most common renal malignancy in children, typically presenting between 2-5 years of age with an asymptomatic abdominal mass.

Important features include:

- It may invade the renal vein and extend into the inferior vena cava (IVC).
- Calcification is uncommon on imaging, which helps differentiate it from neuroblastoma, where calcification is common.
- Lungs are the most common site of metastasis, followed by the liver.

QUESTION

A 5-year-old child is recovering from *Haemophilus influenzae* type b meningitis and is clinically stable for discharge. Which of the following investigations should be performed before discharge to detect the most important long-term complication?

A EEG

B Hearing test

CORRECT

C CT scan of the head

D Developmental screening

RIGHT ANSWER

OK

B. Hearing test

WHY THIS IS RIGHT

In children recovering from *Haemophilus influenzae* type b meningitis, the most important long-term complication is sensorineural hearing loss.

- Inflammation of the meninges can involve the cochlea and auditory nerve, leading to permanent hearing impairment.
- This complication may not be clinically obvious immediately after recovery.

Therefore, hearing assessment (audiological testing) should be performed before discharge or soon after recovery to allow early detection and intervention.

QUESTION

For a newborn with a cephalohematoma that is clinically uncomplicated, what is the next step in management?

- A** Start phototherapy
- B** Observe the child for neonatal jaundice
- C** Monitor vitals and occipital-frontal circumference
- D** Immediate incision and drainage

CORRECT

RIGHT ANSWER

- OK** **B. Observe the child for neonatal jaundice**

WHY THIS IS RIGHT

Cephalohematoma is a subperiosteal collection of blood that occurs due to birth trauma. It is confined by suture lines and usually appears hours after birth.

- Most cases are benign and self-resolving over weeks.
- As the blood within the hematoma breaks down, bilirubin levels can rise, increasing the risk of neonatal jaundice.
- Therefore, the appropriate management for an uncomplicated cephalohematoma is observation with monitoring for hyperbilirubinemia.

Why not the other options?

- Phototherapy -> only if significant jaundice develops.
- Monitoring OFC and vitals -> more relevant for subgaleal hemorrhage, which can cause significant blood loss.
- Incision and drainage -> contraindicated due to risk of infection and because the condition resolves spontaneously.

QUESTION

A newborn infant develops nephrotic syndrome within the first two weeks of life. Which of the following is the most likely cause of this patient's nephrotic syndrome?

- A Congenital toxoplasmosis
- B Congenital syphilis
- C Abnormality in the nephrin gene**
- D Abnormality in the polycystin gene type

CORRECT

RIGHT ANSWER

- OK **C. Abnormality in the nephrin gene**

WHY THIS IS RIGHT

Nephrotic syndrome presenting within the first 3 months (especially first few weeks) strongly suggests Congenital Nephrotic Syndrome (CNS).

The most classic form is Congenital nephrotic syndrome of the Finnish type, caused by mutation in the NPHS1 (nephrin) gene

This produces:

- massive proteinuria
- hypoalbuminemia
- edema
- progression to renal failure

QUESTION

| All the following are causes of neonatal hypocalcemia except:

- A** Prematurity
- B** DiGeorge syndrome
- C** Infant of diabetic mother

D Maternal hypoparathyroidism in pregnancy

CORRECT

RIGHT ANSWER

OK D. Maternal hypoparathyroidism in pregnancy

WHY THIS IS RIGHT

Neonatal hypocalcemia occurs due to immature parathyroid hormone (PTH) response, decreased calcium stores, or increased phosphate load in the newborn.

Common causes include:

- Prematurity -> decreased transplacental calcium transfer in the third trimester.
- Infant of diabetic mother (IDM) -> functional hypoparathyroidism and increased calcitonin levels lead to hypocalcemia.
- DiGeorge syndrome -> congenital absence or hypoplasia of parathyroid glands causing low PTH and hypocalcemia.

Maternal hypoparathyroidism, however, usually leads to maternal hypocalcemia, which stimulates the fetal parathyroid glands and results in normal or even increased fetal calcium levels, making neonatal hypocalcemia unlikely.

QUESTION

A 5-year-old presents with persistent respiratory difficulty. Breath sounds reveal a localized, fixed, monophonic wheeze that does not vary with bronchodilator therapy. There is no history of recurrent cough or sputum. Which is the most likely underlying condition?

A Atopic reactive airway disease

B Intraluminal airway obstruction

CORRECT

C Chronic suppurative airway disease

D Cardiogenic fluid overload

RIGHT ANSWER

OK B. Intraluminal airway obstruction

WHY THIS IS RIGHT

A localized, fixed, monophonic wheeze that does not improve with bronchodilators suggests focal airway obstruction, rather than diffuse airway disease like asthma.

Key reasoning:

- Monophonic wheeze -> single airway segment involved
- Localized and persistent -> structural or obstructive lesion
- No response to bronchodilator -> not bronchospasm

Common causes:

- Intraluminal foreign body
- Endobronchial tumor or mucus plug
- Airway stenosis

QUESTION

A newborn presents with vomiting, dehydration, hyponatremia, hyperkalemia, and hypotension at 10 days of life. Physical exam shows 46,XY genitalia with female-appearing external genitalia. Serum 17-hydroxyprogesterone, testosterone, and cortisol are all markedly low. Which diagnosis is most likely?

A 21-hydroxylase deficiency

B 11 β -hydroxylase deficiency

C Lipoid adrenal hyperplasia

CORRECT

D 17 α -hydroxylase deficiency

RIGHT ANSWER

OK

C. Lipoid adrenal hyperplasia

WHY THIS IS RIGHT

Lipoid congenital adrenal hyperplasia (lipoid CAH) is caused by a defect in the StAR (steroidogenic acute regulatory) protein, which is required for transporting cholesterol into mitochondria for steroid hormone synthesis.

Key consequences:

- Failure of synthesis of all adrenal and gonadal steroids
- ↓ Cortisol, ↓ aldosterone, ↓ androgens/testosterone
- Leads to salt-wasting crisis in the neonatal period (vomiting, dehydration, hyponatremia, hyperkalemia, hypotension)

QUESTION

A 14-year-old boy presents with delayed puberty, decreased facial hair, and gynecomastia. Examination reveals small, firm testes. Which hormonal profile is expected?

A ↓ FSH, ↓ LH, ↓ testosterone

B ↑ FSH, ↑ LH, ↓ testosterone

CORRECT

C ↓ FSH, ↑ LH, ↑ testosterone

D ↑ FSH, ↑ LH, ↑ testosterone

RIGHT ANSWER

OK

B. ↑ FSH, ↑ LH, ↓ testosterone

WHY THIS IS RIGHT

A teenage boy with delayed puberty, gynecomastia, and small firm testes is suggestive of primary testicular failure, classically seen in Klinefelter syndrome (47,XXY).

Because the testes fail to produce adequate testosterone, there is loss of negative feedback on the hypothalamic-pituitary axis, leading to increased gonadotropins.

Hormonal profile:

- ↑ FSH
- ↑ LH
- ↓ Testosterone

QUESTION

A 2-year-old child with unilateral Wilms tumor is found to have aniridia on exam. Which gene mutation is most commonly associated with this condition?

- A** WT1 gene on chromosome 11p13
- B** WT2 gene on chromosome 11p15
- C** TP53 gene on chromosome 17p
- D** RET proto-oncogene on chromosome 10q

CORRECT

RIGHT ANSWER

- OK** A. WT1 gene on chromosome 11p13

WHY THIS IS RIGHT

The association of Wilms tumor with Aniridia suggests WAGR syndrome, which is caused by a deletion or mutation of the WT1 gene, located on chromosome 11p13.

WAGR syndrome features:

- W - Wilms tumor
- A - Aniridia (absence of iris)
- G - Genitourinary abnormalities
- R - Intellectual disability (previously termed mental retardation)

The WT1 gene is a tumor suppressor gene involved in kidney and gonadal development, and its mutation predisposes to Wilms tumor in early childhood.

Why not other options?

- WT2 gene (11p15): Associated with Beckwith-Wiedeman syndrome, which presents with macroglossia, organomegaly, hemihypertrophy, and increased Wilms tumor risk but not aniridia.
- TP53 gene (17p): Mutation seen in Li-Fraumeni syndrome, associated with multiple early-onset cancers.
- RET proto-oncogene (10q): Mutation seen in Multiple endocrine neoplasia 1, associated with medullary thyroid carcinoma and pheochromocytoma.

QUESTION

A neonate born at term undergoes routine newborn metabolic screening. At which age is screening for congenital hypothyroidism ideally performed?

A Within the first 6 hours of life

B Between 48-72 hours after birth

CORRECT

C On day 7-10 of life

D At 2 weeks of age

RIGHT ANSWER

OK B. Between 48-72 hours after birth

WHY THIS IS RIGHT

Screening for Congenital hypothyroidism should ideally be performed between 48-72 hours after birth using a heel-prick blood sample.

- Immediately after birth, there is a physiologic neonatal TSH surge that occurs within the first few hours of life.
- Testing too early (eg, within the first 6-24 hours) may give false-positive elevated TSH levels.
- Waiting 48-72 hours allow the transient TSH surge to settle, providing more accurate detection of persistent hypothyroidism.

Early detection through newborn screening allows prompt treatment with Levothyroxine, which prevents irreversible intellectual disability and growth failure associated with untreated congenital hypothyroidism.

QUESTION

A 9 months old infant is brought for a routine check-up. The infant is able to sit without support and transfer objects. Which of the following developmental milestones is most likely to be present at this age?

A Mature pincer grasp

B Creeps well on hands and knees

CORRECT

C Can hop on one foot

D Runs steadily

RIGHT ANSWER

OK

B. Creeps well on hands and knees

WHY THIS IS RIGHT

At 9 months of age, infants typically achieve gross motor milestones such as sitting without support and beginning to creep or crawl on hands and knees. Fine motor development at this stage includes transferring objects from one hand to another and developing an immature pincer grasp.

- Gross motor: sits without support, creeps/crawls
- Fine motor: hand-to-hand transfer of objects, immature pincer grasp
- Advanced milestones like mature pincer grasp (12 months), running (18-24 months), and hopping (4 years) occur later

QUESTION

A child who can make a cross, tell stories make tower of 6 cubes and goes to toilet alone, what is the age of the child:

A 1 year

B 2 years

C 3 years

D 4 years

CORRECT

RIGHT ANSWER

OK

D. 4 years

WHY THIS IS RIGHT

A 4-year-old child demonstrates advanced fine motor, language, and social independence milestones.

- Fine motor: able to copy a cross and build a tower of about 6 cubes
- Language: can tell simple stories and use well-formed sentences
- Social/adaptive: goes to the toilet independently

Age correlation:

- 1 year: basic motor skills like standing, few words
- 2 years: 2-word phrases, stacks 4-6 cubes
- 3 years: copies a circle, partial toilet training
- 4 years: copies a cross, storytelling, complete toilet training

QUESTION

A 9-month-old infant is able to roll over, babble ‘mama’ and ‘dada,’ and exhibits stranger anxiety but is unable to sit without support. Which of the following is the most appropriate next step in management?

A Reassurance

CORRECT

B Get development test

C Give training to sit

D Get an MRI

RIGHT ANSWER

OK

A. Reassurance

WHY THIS IS RIGHT

Developmental milestones can show normal variation, and a single mildly delayed milestone in the presence of otherwise appropriate development does not necessarily indicate pathology.

- At 9 months, expected milestones include babbling (“mama/dada”), stranger anxiety, and rolling over, all of which are present in this child
- Sitting without support typically develops around 8-9 months, but slight delay can still fall within normal variation
- Absence of red flag signs (loss of milestones, global delay, abnormal tone) supports reassurance

Clinical approach:

- If development is normal in other domains (social, language, fine motor), isolated borderline delay can be observed
- Regular follow-up is important to ensure progression

QUESTION

Which of these milestones is correctly matched with the corresponding age?

- A Stacking two cubes in a tower 2 years
- B Mature pincer grasp at 9 months
- C Transferring the object from one hand to another hand by 6 months**
- D Reaching out to the object with one hand by the age of 8 months

CORRECT

RIGHT ANSWER

OK

C. Transferring the object from one hand to another hand by 6 months

WHY THIS IS RIGHT

Object transfer from one hand to another is a key fine motor developmental milestone achieved at ~6 months of age.
 • It reflects improving hand coordination and bilateral integration.

Why other options are incorrect:

- A. Stacking two cubes -> occurs at ~15 months, not 2 years
- B. Mature pincer grasp -> develops at ~12 months, not 9 months
- D. Reaching with one hand -> seen earlier (~5 months), not 8 months

Age	Social Milestone
2m	Social smile
3m	Recognizes mother
4m	Stranger anxiety, inhibits to no
6m	Waves bye-bye, repeats activity when appreciated
9m	Comes when called, plays simple ball game
12m	Jargon, points to objects of interest
18m	Copies parents in task
2y	Asks for food, drink, toilet
3y	Shares toys, knows full name age gender
4y	Plays cooperatively in group, goes to toilet alone
5y	Helps in household tasks

Age	Language milestone
1m	Alerts to sound
3m	Coos
4m	Laughs loud
6m	Monosyllables
9m	Bisyllables
12m	1-2 words with meaning
18m	8-10-word vocabulary
2y	2-3 word sentences, uses pronouns
3y	Asks question
4y	Sings song, tells stories
5y	Asks meaning of words

Age	Gross motor Milestone
3m	Neck holding
4m	Rolls over
6m	Sits in tripod position
9m	Sits without support, crawling
10m	Stand with support, creeps
12m	Stands without support
15m	Walks alone
18m	Runs
2y	Walks up and downstairs, 2 feet step
3y	Rides tricycle, alternate feet going upstairs
4y	Hops on one foot, alternate feet going downstairs

Age	Fine motor milestone
4m	Bidextrous approach
6m	Unidextrous approach
9m	Immature pincer grasp
12m	Mature pincer grasp
15m	Imitates scribbling, tower of 2 blocks, drinks from cup
18m	Scribbles, tower of 3 blocks
2y	Tower of 6 blocks, vertical and circular strokes, undresses, feeds with spoon
3y	Tower of 9 blocks, copies circle, dresses
4y	Copies cross, bridge with blocks
5y	Copies triangle, gate with blocks

QUESTION

Identify the correct statements:

1. Social smile - 2months
2. Walk without support -12months
3. Babbling gibberish-15months
4. Walk upstairs with alternate foot - 4yrs
5. Copy a circle-2yrs

A 1, 2, 3, 4

B 1, 3, 4, 5

C 1, 3

CORRECT

D 1, 3, 5

RIGHT ANSWER

OK C. 1, 3

WHY THIS IS RIGHT

Developmental milestones occur at predictable ages and are used to assess normal child development.

Evaluation of the statements:

1. Social smile - 2 months -> Correct (appears around 6-8 weeks).
2. Walk without support - 12 months -> Incorrect in strict milestone charts; independent walking is typically around 12-15 months, not fixed at exactly 12 months.
3. Babbling gibberish - ~15 months -> Correct (jargon speech with varied sounds appears around this age).
4. Walk upstairs with alternate feet - 4 years -> Incorrect; alternating feet usually occurs around 3 years.
5. Copy a circle - 2 years -> Incorrect; copying a circle occurs around 3 years (2 years: scribbling).

QUESTION

While you are evaluating a baby, you show him a bright pink teddy bear that he reaches out to with both hands. What is the earliest age by which this milestone is typically achieved?

A 4 months

CORRECT

B 5 months

C 6 months

D 7 months

RIGHT ANSWER

OK

A. 4 months

WHY THIS IS RIGHT

Reaching out for an object with both hands is an early gross motor and visual-motor coordination milestone that typically appears by 4 months of age.

At this age:

- Infants develop improved head control and upper limb coordination.
- They begin to visually track objects and voluntarily reach for them with both hands.
- This marks the transition from reflexive grasping to purposeful reaching.

QUESTION

| All of the following milestones are delayed in a child with Down syndrome except:

A Personal and social development

CORRECT

B Gross motor

C Fine motor

D Language

RIGHT ANSWER

OK

A. Personal and social development

WHY THIS IS RIGHT

Children with Down syndrome typically show global developmental delay, particularly affecting:

- Gross motor skills (due to hypotonia and ligamentous laxity)
- Fine motor skills
- Language development (often the most significantly delayed)

However, personal and social development is relatively preserved and may even be better compared to other developmental domains.

QUESTION

A 15-month-old infant is not yet walking independently. Examination and history are normal. Appropriate next step:

A Reassure and observe until 18 months

CORRECT

B Immediate brain MRI

C Start physiotherapy for cerebral palsy

D Genetic testing for SMA

RIGHT ANSWER

OK **A. Reassure and observe until 18 months**

WHY THIS IS RIGHT

Independent walking normally develops between 9-18 months of age. Therefore, a 15-month-old child who is not yet walking but has otherwise normal development and examination can still fall within the normal range of motor milestone acquisition.

In such cases, the appropriate approach is reassurance and observation, with follow-up until 18 months, when delayed walking would warrant further evaluation.

Why not other options?

- B. Immediate brain MRI: Not indicated because the child has normal history and neurological exam. Imaging is reserved if neurologic abnormalities or regression are present.
- C. Start physiotherapy for cerebral palsy: CP would show abnormal tone, delayed milestones, or abnormal reflexes, which are not present here.
- D. Genetic testing for SMA: SMA presents with hypotonia, weakness, and delayed motor milestones, usually with abnormal exam findings.

QUESTION

A 2½-year-old boy is brought for evaluation of delayed speech. His parents report that he speaks only a few single words and does not combine words into phrases. He follows simple commands, shows appropriate eye contact, points to objects of interest, and engages in pretend play. His motor milestones were appropriate for age: he sat without support at 7 months, stood at 12 months, and walked independently by 14 months. There are no feeding issues or seizures. Hearing screening is normal.

This clinical presentation most likely represents:

A Developmental dissociation

CORRECT

B Developmental delay

C Developmental regression

D Developmental deviation

RIGHT ANSWER

OK A. Developmental dissociation

WHY THIS IS RIGHT

Developmental dissociation refers to uneven development in different developmental domains, where one area (e.g., language) is delayed while others such as social interaction, cognition, and motor development remain normal. In this child Speech delay is present with Normal social interaction (eye contact, pointing, pretend play) and Normal motor milestones. This pattern indicates isolated language delay with otherwise normal development, which is characteristic of developmental dissociation.

Key distinctions:

- Developmental delay: Delay in multiple developmental domains
- Developmental regression: Loss of previously acquired skills
- Developmental deviation: Abnormal pattern of development (e.g., autism spectrum behaviors)

QUESTION

A 2-year-old infant presents with high conjugated (direct) bilirubin, dark coloured urine and clay-coloured stools. On examination, the infant is visibly jaundiced but is feeding normally. What is the most likely diagnosis?

A Biliary atresia

CORRECT

B Gilbert syndrome

C Crigler Najjar Syndrome

D Physiological jaundice

RIGHT ANSWER

OK

A. Biliary atresia

WHY THIS IS RIGHT

A young child with conjugated (direct) hyperbilirubinemia, dark urine, and pale (clay-colored) stools is suggestive of obstructive cholestasis, most commonly due to biliary atresia.

- Conjugated hyperbilirubinemia always indicates pathological jaundice
- Pale stools occur due to absence of bile pigments in the intestine
- Dark urine is due to excretion of conjugated bilirubin
- Biliary atresia presents in early infancy with persistent jaundice and requires early surgical intervention (Kasai procedure)
- Conditions like Gilbert syndrome, Crigler-Najjar syndrome, and physiological jaundice cause unconjugated hyperbilirubinemia

QUESTION

A 6-month-old infant presents with sudden, brief spasms involving flexion of the neck, trunk, and limbs occurring in clusters. EEG reveals a hypsarrhythmia pattern. What is the first-line treatment for this condition?

A ACTH

CORRECT

B Valproate

C Ethosuximide

D Carbamazepine

RIGHT ANSWER

OK

A. ACTH

WHY THIS IS RIGHT

A 6-month-old infant with clustered flexor spasms and a hypsarrhythmia pattern on EEG is characteristic of infantile spasms (West syndrome). This is an age-specific epileptic encephalopathy requiring urgent treatment to prevent neurodevelopmental deterioration.

- Classic triad:
 - Infantile spasms (sudden flexion/extension movements in clusters)
 - Hypsarrhythmia on EEG (chaotic, high-amplitude disorganized pattern)
 - Developmental delay or regression
- Peak onset: 3-8 months of age
- First-line treatment: ACTH (or oral steroids); vigabatrin is preferred in tuberous sclerosis
- Drugs like valproate, ethosuximide, and carbamazepine are not effective for this condition

QUESTION

A newborn born to a diabetic mother, presents after 24 hours of life with a blood glucose level of 35 mg/dL. The baby is symptomatic. What is the next best step in management?

A Administer IV glucose and recheck blood glucose after 20 minutes

CORRECT

B Administer IV bolus of Hartmann solution

C Recheck blood glucose after 20 minutes

D Start oral feeds and observe

RIGHT ANSWER

OK

A. Administer IV glucose and recheck blood glucose after 20 minutes

WHY THIS IS RIGHT

Neonatal hypoglycemia is a medical emergency, especially in infants of diabetic mothers, and requires immediate correction to prevent neurological injury.

- Infants of diabetic mothers are at risk due to hyperinsulinemia
- Symptomatic hypoglycemia (e.g., lethargy, seizures, poor feeding) requires urgent treatment
- Initial management: IV bolus of glucose (10% dextrose, 2 ml/kg) followed by infusion

Clinical approach:

- Confirm hypoglycemia (<40-45 mg/dL depending on age)
- Give IV glucose immediately
- Recheck blood glucose after 15-20 minutes

Definition: Blood <40mg/dL/Plasma <45mg/dL

Asymptomatic: >20mg/dL - oral [BF] - >40mg/dL

└ <20mg/dL → 40mg/dL → Glycaemic infusion

Symptomatic: iv dextrose bolus 2mL/kg - infusion
[jitteriness; seizure; coma]

QUESTION

A previously healthy child presented with acute-onset dyspnea. A chest X-ray shows unilateral hyperinflation of the lungs. What is true for this patient?

CORRECT

A Focal area of decreased air entry will be suggestive of foreign body

B Flexible bronchoscopy used for removal

C In complete obstruction, ball and valve mechanism causes hyperinflation

D The child has developed acute laryngotracheobronchitis

RIGHT ANSWER

OK

A. Focal area of decreased air entry will be suggestive of foreign body

WHY THIS IS RIGHT

Foreign body aspiration should be suspected in a previously healthy child with sudden onset respiratory distress and unilateral hyperinflation on chest X-ray.

- Partial bronchial obstruction creates a “ball-valve” effect -> air enters but cannot exit -> unilateral hyperinflation
- Clinical sign: focal decreased air entry on the affected side is a key clue
- Most common in young children due to aspiration of small objects (e.g., peanuts)

Management:

- Rigid bronchoscopy is the procedure of choice for both diagnosis and removal
- Flexible bronchoscopy is mainly diagnostic, not preferred for removal

Why other options are incorrect:

- Ball-valve mechanism occurs in partial, not complete obstruction
- Laryngotracheobronchitis (croup) causes bilateral airway involvement, not unilateral hyperinflation

QUESTION

Which of the following children are considered at-risk babies?

1. Baby with a birth weight of 2.5 kg
2. Baby on artificial feeds
3. Baby of working mother/single parent
4. Baby with weight <85% of expected weight
5. Birth order of 3 or more

A 2, 3

CORRECT

B 1, 2, 3, 4

C 4, 5

D 1, 4

RIGHT ANSWER

OK

A. 2, 3

WHY THIS IS RIGHT

At-risk babies” are infants who have higher chances of morbidity and mortality due to biological or social risk factors, requiring closer monitoring.

- Social and feeding-related risks are important.
- Artificial feeding increases risk of infections and malnutrition
- Baby of a working mother or single parent may have inadequate care or feeding issues

Why other options are not included:

- Birth weight of 2.5 kg is normal (low birth weight is <2.5 kg)
- Weight <85% expected suggests malnutrition but applies to older infants/children, not specifically “at-risk baby” category
- Birth order alone is not a direct criterion unless associated with poor care

QUESTION

A child presented with a history of loose stools with an increase in frequency for 4 days. On examination, he is drowsy, unable to feed, and skin on pinching goes back very slowly. According to the integrated management of neonatal and childhood illness (IMNCI), this child will be classified as having:

A Mild dehydration

B Some dehydration

C Severe dehydration

CORRECT

D Moderate dehydration

RIGHT ANSWER

OK C. Severe dehydration

WHY THIS IS RIGHT

IMNCI classifies dehydration in children based on clinical signs into no dehydration, some dehydration, and severe dehydration to guide urgent management.

Parameters	No Dehydration	Some Dehydration
Appearance	Well, alert	Restless, irritable
Eyes	Normal	Sunken
Thirst	Drinks normally, not thirsty	Thirsty, drinks eagerly
Skin pinch	Goes back quickly (<1 second)	Goes back slowly (1 second)

Key concepts for severe dehydration:

- Lethargic or unconscious (drowsy child)
- Unable to drink or drinks poorly
- Skin pinch goes back very slowly

Clinical importance:

- Requires immediate IV fluid therapy (Plan C)
- Rapid recognition prevents shock and death

QUESTION

A 2-month-old infant born to an HIV-positive mother presents with recurrent diarrhea. What is the next best step?

A Test stool for giardia and give antibiotics

B Dried spot sample for HIV DNA PCR

CORRECT

C Antibody tests for HIV

D Aerobic culture

RIGHT ANSWER

OK

B. Dried spot sample for HIV DNA PCR

WHY THIS IS RIGHT

In infants born to HIV-positive mothers, diagnosis of HIV infection is made using virological tests (HIV DNA PCR), not antibody tests.

- Maternal IgG antibodies cross the placenta -> antibody tests remain positive up to 18 months
- Therefore, antibody-based tests are unreliable in infants
- Early infant diagnosis is done using HIV DNA PCR (often via dried blood spot)

Clinical importance:

- Allows early detection and timely initiation of antiretroviral therapy
- Improves survival and reduces disease progression

QUESTION

An 8-day-old newborn was found to have thyroid-stimulating hormone level of more than 100 mIU/L. Which of the following will be the next best investigation?

- A Urine iodine excretion
- B Serum thyroid receptor antibody

C Radiotracer uptake with technetium

CORRECT

- D Perchlorate secretion test

RIGHT ANSWER

C. Radiotracer uptake with technetium

WHY THIS IS RIGHT

A markedly elevated TSH (>100 mIU/L) in a neonate strongly suggests congenital hypothyroidism and requires prompt evaluation of thyroid gland anatomy and function.

- Most common causes: thyroid dysgenesis (agenesis, ectopia) or dyshormonogenesis
- Next step after diagnosis is imaging to determine etiology
- Radionuclide scan (technetium or radioiodine uptake) helps identify:
 - Absent gland (agenesis)
 - Ectopic thyroid
 - Normal gland with impaired hormone synthesis

Clinical importance:

- Treatment with levothyroxine should be started immediately without waiting for investigations
- Early treatment prevents irreversible neurodevelopmental delay

QUESTION

A 1-day-old neonate has not passed urine since birth. What is the next step in management?

A Continue breastfeeding and observe

CORRECT

B Admit to NICU

C Start artificial feeding

D Start intravenous fluids

RIGHT ANSWER

OK

A. Continue breastfeeding and observe

WHY THIS IS RIGHT

In healthy term neonates, delayed passage of urine in the first 24 hours can be a normal finding and does not always indicate pathology.

- Most neonates void within the first 24 hours, but some may pass urine slightly later
- If the baby is otherwise well (feeding, active, normal exam), observation is appropriate
- Continue breastfeeding to ensure adequate hydration

When to investigate:

- No urine after 48 hours
- Associated signs: poor feeding, abdominal distension, vomiting, or suspected urinary obstruction

QUESTION

An infant is brought to OPD with hepatosplenomegaly and thrombocytopenia. Neuroimaging with CT reveals periventricular calcifications. What is the most likely diagnosis?

- A** Congenital rubella syndrome
- B** Congenital herpes simplex virus infection
- C** Congenital toxoplasmosis
- D** Congenital cytomegalovirus infection

CORRECT

RIGHT ANSWER

OK

D. Congenital cytomegalovirus infection

WHY THIS IS RIGHT

Congenital cytomegalovirus (CMV) infection is the most common congenital infection and is characterized by periventricular intracranial calcifications along with systemic features.

Classic features:

- Periventricular calcifications (hallmark)
- Hepatosplenomegaly
- Thrombocytopenia (-> petechiae, "blueberry muffin" rash)
- Sensorineural hearing loss

Differentiation:

- Toxoplasmosis: diffuse intracranial calcifications, hydrocephalus, chorioretinitis
- Rubella: cataracts, PDA, deafness
- HSV: encephalitis with temporal lobe involvement



QUESTION

A 3-month-old baby is brought to OPD with jaundice and clay-colored stools. Lab work up reveals that the baby has conjugated hyperbilirubinemia. A liver biopsy was done and shows periductal proliferation. What is the most likely diagnosis?

A Crigler-Najjar syndrome

B Rotor syndrome

C Dubin-Johnson syndrome

D Biliary atresia

CORRECT

RIGHT ANSWER

OK

D. Biliary atresia

WHY THIS IS RIGHT

Biliary atresia is a cause of neonatal cholestasis characterized by progressive obliteration of the extrahepatic bile ducts, leading to conjugated hyperbilirubinemia.

- Presents in early infancy with persistent jaundice, clay-colored (acholic) stools, and dark urine
- Lab findings: conjugated hyperbilirubinemia
- Liver biopsy shows bile duct proliferation, portal fibrosis, and bile plugs

Clinical importance:

- Requires early diagnosis and intervention
- Kasai portoenterostomy should be performed ideally before 6-8 weeks for best outcomes

Differentiation:

- Crigler-Najjar: unconjugated hyperbilirubinemia
- Dubin-Johnson & Rotor: conjugated hyperbilirubinemia but no obstructive features like pale stools

QUESTION

A 3-month-old baby comes with complaints of deafness, cataract, and patent ductus arteriosus. Which of the following is the most likely diagnosis?

A Congenital herpes simplex virus infection

B Congenital toxoplasmosis

C Congenital cytomegalovirus infection

D Congenital rubella syndrome

CORRECT

RIGHT ANSWER

OK

D. Congenital rubella syndrome

WHY THIS IS RIGHT

Congenital rubella syndrome (CRS) is characterized by a classic triad of sensorineural deafness, cataracts, and congenital heart defects (most commonly patent ductus arteriosus).

- Caused by transplacental infection with rubella virus, especially during the first trimester

Classic triad:

- Sensorineural hearing loss (most common)
- Eye defects (cataracts, retinopathy)
- Cardiac defects (PDA, pulmonary artery stenosis)

Other features:

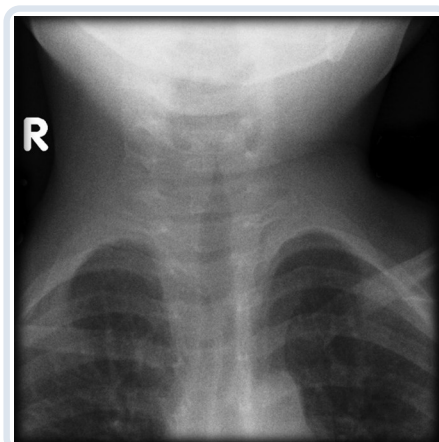
- Blueberry muffin rash
- Microcephaly, developmental delay
- Hepatosplenomegaly

Differentiation:

- CMV: periventricular calcifications, hearing loss
- Toxoplasmosis: hydrocephalus, chorioretinitis, diffuse calcifications
- HSV: skin vesicles, encephalitis

QUESTION

A child is brought to the hospital with features of respiratory distress and biphasic stridor. There is a history of upper respiratory infection. The radiograph is shown below. Mark the correct diagnosis?



A Acute epiglottitis

B Acute laryngotracheobronchitis

CORRECT

C Foreign body in glottis

D Laryngomalacia

RIGHT ANSWER

OK

B. Acute laryngotracheobronchitis

WHY THIS IS RIGHT

Acute laryngotracheobronchitis (croup) is a common cause of upper airway obstruction in children, typically following a viral upper respiratory infection.

- Presents with barking cough, hoarseness, and inspiratory or biphasic stridor
- X-ray (AP neck) shows “steeple sign” due to subglottic narrowing
- Most commonly caused by parainfluenza virus

Management:

- Mild: humidified air
- Moderate to severe: nebulized adrenaline and corticosteroids

Differentiation:

- Epiglottitis: thumb sign on lateral X-ray, toxic child, drooling
- Foreign body: sudden onset without preceding URI
- Laryngomalacia: chronic stridor since infancy

QUESTION

A woman came to OPD with a newborn who has complaints of chest retractions, dyspnea and lethargy. The paediatrician diagnosed the baby with respiratory distress syndrome. This occurs due to the deficiency of:

- A** Dipalmitoyl inositol
- B** Lecithin
- C** Sphingomyelin
- D** Dipalmitoyl phosphatidylethanolamine

CORRECT

RIGHT ANSWER

OK B. Lecithin

WHY THIS IS RIGHT

Neonatal respiratory distress syndrome (hyaline membrane disease) is caused by deficiency of pulmonary surfactant, particularly dipalmitoyl phosphatidylcholine (lecithin), produced by type II pneumocytes.

- Lecithin is the major component of surfactant responsible for reducing alveolar surface tension
- Deficiency leads to alveolar collapse (atelectasis), decreased lung compliance, and respiratory distress
- Common in preterm infants due to immature lungs

Clinical relevance:

- L/S ratio (lecithin:sphingomyelin) is used to assess fetal lung maturity
- Antenatal corticosteroids increase surfactant production

QUESTION

A mother delivers a 2.5 kg baby with normal vaginal delivery in a rural area. Which of the following is an incorrect statement?

A Exclusive breastfeeding

B Bath the baby with warm water immediately

CORRECT

C Clean the eyes

D Skin to skin contact

RIGHT ANSWER

OK B. Bath the baby with warm water immediately

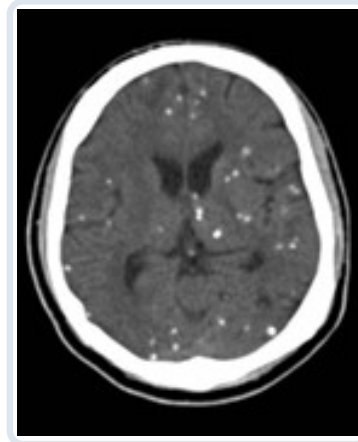
WHY THIS IS RIGHT

Essential newborn care focuses on thermal protection, early breastfeeding, and infection prevention.

- Immediate bathing should be avoided because it can cause hypothermia; bathing is delayed for at least 24 hours
- Early skin-to-skin contact (kangaroo care) helps maintain temperature and promotes bonding
- Exclusive breastfeeding should be initiated within the first hour of life
- Eye care helps prevent neonatal infections

QUESTION

A 3-month-old baby with intracranial diffuse calcifications, chorioretinitis, and hydrocephalus. What is the most likely diagnosis?



A Toxoplasmosis

CORRECT

B CMV

C Zika virus

D Rubella

RIGHT ANSWER

OK

A. Toxoplasmosis

WHY THIS IS RIGHT

Congenital toxoplasmosis classically presents with the triad of chorioretinitis, hydrocephalus, and intracranial calcifications.

- Caused by transplacental infection with *Toxoplasma gondii*
- Intracranial calcifications are typically diffuse (not periventricular)
- Associated features include seizures, developmental delay, and hepatosplenomegaly

Differentiation:

- CMV: periventricular calcifications
- Zika virus: microcephaly with calcifications
- Rubella: cataracts, cardiac defects, deafness

QUESTION

An HIV-positive mother with a viral load of 1200 copies/mL delivers a newborn baby. What is the most appropriate antiretroviral prophylaxis for the newborn?

A Nevirapine for 6 weeks

B Nevirapine + Zidovudine for 12 weeks

CORRECT

C Nevirapine for 12 weeks

D Zidovudine for 4 weeks

RIGHT ANSWER

OK B. Nevirapine + Zidovudine for 12 weeks

WHY THIS IS RIGHT

In infants born to HIV-positive mothers, the choice and duration of antiretroviral prophylaxis depend on the risk of transmission, primarily determined by maternal viral load.

- High-risk exposure (maternal viral load >1000 copies/mL or unknown) requires combination prophylaxis
- Recommended regimen: dual therapy with Nevirapine + Zidovudine for extended duration (e.g., 12 weeks)
- Low-risk exposure (viral load <1000 copies/mL, good ART adherence) may receive single-drug prophylaxis

Clinical importance:

- Early initiation (within hours of birth) significantly reduces vertical transmission
- Adherence to prophylaxis and follow-up testing is essential

QUESTION

A 6-month-old infant presents with recurrent infections and failure to thrive. Laboratory investigations reveal a deficiency of adenosine deaminase (ADA). Which of the following immunodeficiency disorders is most likely associated with this condition?

A Severe Combined Immunodeficiency (SCID)

CORRECT

B Hypogammaglobulinemia

C DiGeorge Syndrome

D Wiskott-Aldrich Syndrome

RIGHT ANSWER

OK A. Severe Combined Immunodeficiency (SCID)

WHY THIS IS RIGHT

Adenosine deaminase (ADA) deficiency is a cause of severe combined immunodeficiency (SCID), a life-threatening primary immunodeficiency characterized by defective T-cell and B-cell function.

- ADA deficiency leads to accumulation of toxic metabolites (deoxyadenosine) -> lymphocyte destruction
- Results in profound deficiency of both cellular (T-cell) and humoral (B-cell) immunity
- Presents in infancy with recurrent severe infections, failure to thrive, and opportunistic infections
- Requires urgent management such as hematopoietic stem cell transplantation
- Enzyme replacement therapy and gene therapy are also options

Contrast:

- Hypogammaglobulinemia: primarily B-cell defect
- DiGeorge syndrome: thymic aplasia -> T-cell deficiency
- Wiskott-Aldrich syndrome: triad of eczema, thrombocytopenia, and recurrent infections

QUESTION

At 6 weeks of life, a child developed a fever after DPT vaccination. And now child is 10 weeks due for vaccination. What should be done next?

- A** Give DPT Vaccination
- B** Give DT
- C** Don't give DPT
- D** Don't give any vaccination

CORRECT

RIGHT ANSWER

- OK** **A. Give DPT Vaccination**

WHY THIS IS RIGHT

Mild adverse events following immunization, such as low-grade fever after DPT vaccination, are common and not a contraindication to subsequent doses.

- DPT vaccine can cause minor reactions like fever, irritability, and local pain
- These reactions do not warrant withholding or changing future doses
- True contraindications include severe allergic reaction (anaphylaxis) or encephalopathy after a previous dose

Clinical application:

- The child should continue the routine immunization schedule
- Subsequent doses of DPT should be given as planned

QUESTION

INSURE technique is used in which condition?

- A** Meconium aspiration syndrome
- B** Transient tachypnea of newborn
- C** **Hyaline membrane disease**
- D** Neonatal jaundice

CORRECT

RIGHT ANSWER

- OK** **C. Hyaline membrane disease**

WHY THIS IS RIGHT

The INSURE technique (Intubation-Surfactant-Extubation) is used in preterm neonates with hyaline membrane disease (neonatal respiratory distress syndrome) to deliver surfactant while minimizing duration of mechanical ventilation.

- Indicated in surfactant deficiency seen in preterm infants
- Procedure: brief intubation -> administer exogenous surfactant -> early extubation to CPAP
- Reduces complications of prolonged ventilation such as bronchopulmonary dysplasia

Clinical significance:

- Improves oxygenation and lung compliance
- Decreases need for prolonged mechanical ventilation

QUESTION

| All of the following are true about neonatal cholestasis, except?

- A** Biliary atresia is the most common cause of neonatal cholestasis
- B** Kasai procedure should be done within 90 days
- C** HIDA scan is the best investigation for diagnosis
- D** Can be differentiated from other differential diagnoses by biopsy

CORRECT

RIGHT ANSWER

OK

B. Kasai procedure should be done within 90 days

WHY THIS IS RIGHT

Neonatal cholestasis is defined as conjugated hyperbilirubinemia in early infancy and requires prompt evaluation, with biliary atresia being the most important cause to rule out.

- Early diagnosis is critical because Kasai portoenterostomy is most effective when performed before 6-8 weeks of age (60 days is the usual cut-off)
- HIDA scan helps assess bile flow but is not definitive
- Liver biopsy is a key investigation to differentiate biliary atresia from other causes (e.g., neonatal hepatitis)

QUESTION

Which among the following is false about breast milk jaundice:

- a. Breast feeding should be completely stopped
- b. Unconjugated hyperbilirubinemia.
- c. Phototherapy is required to treat most cases
- d. Bilirubin levels are usually more than 10 mg/dL at 3-4 weeks

A a, c and d

B a and c

CORRECT

C a, b and d

D a and d

RIGHT ANSWER

OK B. a and c

WHY THIS IS RIGHT

Breast milk jaundice is a benign, late-onset unconjugated hyperbilirubinemia seen in otherwise healthy, thriving breastfed infants and usually does not require interruption of breastfeeding or aggressive treatment.

- Occurs after the first week of life and may persist up to 3-12 weeks
- Caused by increased enterohepatic circulation due to beta-glucuronidase in breast milk
- Infant is clinically well with normal feeding and weight gain
- Bilirubin levels may remain mildly elevated (often >10 mg/dL) during 3-4 weeks

Why statements a and c are false:

- Breastfeeding should not be completely stopped; it is usually continued
- Phototherapy is not required in most cases unless bilirubin reaches treatment thresholds

QUESTION

| Which of the following statements regarding breastfeeding technique is/are correct?

- A** The infant's lower lip should be inverted
- B** The infant's cheeks should not appear full during feeding
- C** Upper areola should be covered more than lower
- D** The infant's chin should touch the breast

CORRECT

RIGHT ANSWER

- OK** **D. The infant's chin should touch the breast**

WHY THIS IS RIGHT

Correct breastfeeding technique (proper latch) is essential for effective milk transfer and prevention of maternal nipple problems.

Key features of proper latch:

- Infant's chin should touch the breast
- Mouth wide open with lips everted (not inverted)
- More of the lower areola is inside the mouth than the upper
- Cheeks appear full (not sunken) during feeding

Why others are incorrect:

- Lower lip should be everted, not inverted
- Cheeks should be full, not sunken
- More lower areola (not upper) should be covered

QUESTION

A 3-month-old infant presents with prolonged jaundice, dark urine, and pale stools. The child is exclusively breastfed and otherwise well. HIDA scan is non-excretory. What is the next best step in management?

- A** Rule out galactosemia using enzyme assay.
- B** The child has breast milk jaundice and start formula feeding after 3 weeks.
- C** Immediate liver biopsy to confirm diagnosis. CORRECT
- D** Perform a hepatic ultrasound to look for causes like biliary atresia.

RIGHT ANSWER

- OK** **C. Immediate liver biopsy to confirm diagnosis.**

WHY THIS IS RIGHT

Biliary atresia should be strongly suspected in an infant with prolonged jaundice, pale (acholic) stools, dark urine, and a non-excretory HIDA scan. The next best step is liver biopsy to confirm the diagnosis and differentiate it from other causes of neonatal cholestasis.

- Conjugated hyperbilirubinemia presenting after 2 weeks of life is always pathological
- Pale stools and dark urine indicate obstructive cholestasis
- Non-excretion on HIDA scan suggests biliary obstruction but is not definitive
- Liver biopsy shows bile duct proliferation, portal fibrosis, and bile plugs, helping confirm biliary atresia

Why other options are incorrect:

- Galactosemia presents with systemic illness (vomiting, hypoglycemia, sepsis), not isolated cholestasis
- Breast milk jaundice causes unconjugated hyperbilirubinemia with normal stool and urine color
- Ultrasound is useful but not the next best step after a non-excretory HIDA; biopsy provides definitive diagnosis

QUESTION

A term neonate delivered by cesarean section develops tachypnea and mild respiratory distress shortly after birth. Chest x-ray shows prominent vascular markings with fluid in the interlobar fissure and a normal cardiac shadow. The symptoms dissolve completely within 48 hours. What is the likely diagnosis?

- A** RDS
- B** Meconium aspiration syndrome
- C** Pneumonia
- D** Transient tachypnea of newborn

CORRECT

RIGHT ANSWER

- OK** D. Transient tachypnea of newborn

WHY THIS IS RIGHT

The source quiz contains the placeholder “\$19” here and does not include a written explanation for this item.

QUESTION

A 12-year-old girl is brought by her mother, who asks about the normal sequence of puberty. The girl is noted to have Tanner stage 2 breast development and Tanner stage 2 pubic hair. What is the next physiological event expected in her pubertal progression?

A Axillary hair

B Peak growth in height

CORRECT

C Menarche

D Start of growth spurt

RIGHT ANSWER

OK

B. Peak growth in height

WHY THIS IS RIGHT

In normal female puberty, breast development (thelarche, Tanner stage 2) is the first sign, followed by a sequence of predictable events culminating in menarche.

- Thelarche (breast budding, Tanner stage 2)
- Pubarche (pubic hair)
- Peak height velocity (maximum growth spurt)
- Menarche (typically occurs about 2-2.5 years after thelarche)

Application:

- At Tanner stage 2 for breast and pubic hair, the next expected event is peak growth in height

QUESTION

A 12-year-old child presents with short stature. On evaluation, bone age is found to be 10 years, and the upper segment to lower segment (US:LS) ratio is 1.1:1. There are no dysmorphic features, and the child is otherwise healthy. What is the most likely diagnosis?

A Achondroplasia

B Familial short stature

C Constitutional delay of growth and puberty

CORRECT

D Chondrodysplasia

RIGHT ANSWER

OK C. Constitutional delay of growth and puberty

WHY THIS IS RIGHT

Constitutional delay of growth and puberty (CDGP) is a common cause of short stature characterized by delayed bone age with otherwise normal body proportions and health.

- Bone age is delayed compared to chronological age
- Normal upper segment to lower segment ratio indicates proportionate short stature
- No dysmorphic features or systemic illness
- Growth velocity is usually normal, but puberty is delayed

Differentiation:

- Familial short stature: bone age matches chronological age
- Achondroplasia/chondrodysplasia: disproportionate short stature with abnormal body ratios

QUESTION

Which of the following is the correct sign indicating adequate growth in an infant with a birth weight of 3 kg?

- A** Increase in length of 25 cm in the first year
- B** Weight gain of 300 grams per month till 1 year
- C** Anterior fontanelle closure by 6 months of age
- D** Weight under 75th percentile and height under 25th percentile

CORRECT

RIGHT ANSWER

- OK** A. Increase in length of 25 cm in the first year

WHY THIS IS RIGHT

Normal infant growth follows predictable patterns, and linear growth (length gain) is a reliable indicator of adequate growth.

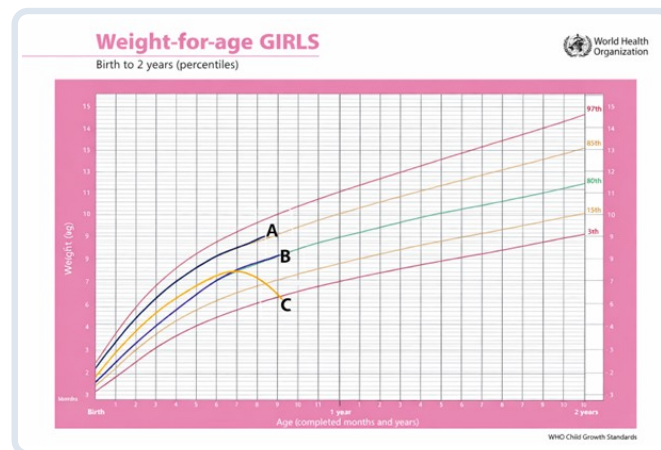
- Length increases by about 25 cm in the first year (≈50 cm at birth → ~75 cm at 1 year)
- Weight typically triples by 1 year, but monthly gain is not constant throughout the year
- Anterior fontanelle normally closes between 9-18 months (not by 6 months)
- Growth assessment depends on tracking along percentiles, not absolute percentile positions

Why option A is correct:

- Increase of ~25 cm in length in the first year is a standard normal growth parameter

QUESTION

Identify the child whose growth is compromised:



A Line A

B Line B

C Line C

CORRECT

D Both B and C

RIGHT ANSWER

OK C. Line C

WHY THIS IS RIGHT

Growth monitoring using WHO growth charts is essential to identify growth faltering, with particular attention to trends rather than single measurements.

- A child's growth should follow a consistent percentile channel
- Crossing down of percentiles or a falling growth curve indicates growth faltering
- Line C shows a downward trend crossing percentiles -> indicates compromised growth
- Lines A and B follow expected growth trajectories without significant deviation

Clinical significance:

- Growth faltering suggests underlying issues such as malnutrition, chronic illness, or malabsorption
- Requires further evaluation and intervention

QUESTION

Maximum brain growth occurs at which year of life?

A 2 years

CORRECT

B 6 years

C 10 years

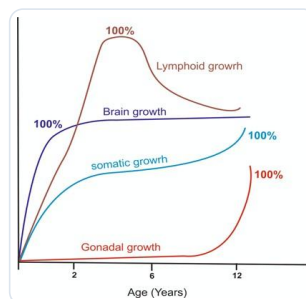
D 12 years

RIGHT ANSWER

OK **A. 2 years**

WHY THIS IS RIGHT

The brain undergoes rapid growth in the first 2 years of life, reaching about 80% of adult brain size. This period is critical for synaptogenesis and myelination, making it highly sensitive to nutrition and environmental factors. Hence, maximum brain growth occurs by 2 years of age.



QUESTION

Calculate the Downe score of a newborn who has cyanosis which improves on giving oxygen, mild to moderate retractions, grunting heard on naked ear, air entry markedly decreased, RR 40 per minute?

A 4

B 5

C 6

CORRECT

D 7

RIGHT ANSWER

OK C. 6

WHY THIS IS RIGHT

The Downe score is used to assess the severity of respiratory distress in newborns.

1 (cyanosis) + 1 (retractions) + 2 (grunting) + 2 (air entry) + 0 (RR) = 6

A score of 6 in this case indicates moderate respiratory distress, characterized by cyanosis that improves with oxygen, mild to moderate retractions, grunting heard on naked ear, markedly decreased air entry, and a normal respiratory rate.

Parameter	0	1	2
Respiratory Rate	< 60/min	60–80/min	> 80/min
Cyanosis	None	In room air	Even with 40% O ₂
Air Entry	Normal	Decreased	Barely audible
Grunting	None	Audible with stethoscope	Audible without stethoscope
Retractions	None	Mild	Moderate–severe

QUESTION

| Which of the following does not play a role in fetal growth?

CORRECT

- A Growth hormone
- B Insulin-like growth factors
- C Thyroxine
- D All of the above

RIGHT ANSWER

OK A. Growth hormone

WHY THIS IS RIGHT

Fetal growth is primarily regulated by insulin-like growth factors (IGF-1, IGF-2) and maternal-placental factors, not by growth hormone.

- IGFs are the main drivers of fetal growth and are influenced by fetal insulin and nutrient supply
- Thyroxine plays a supportive role in overall growth and development
- Growth hormone has minimal to no role in intrauterine growth and becomes important only after birth

QUESTION

A 4-year-old child with weight for height less than -1 SD & height for age is -2 SD.
What is the best possible assessment?

- A Acute malnutrition
- B Chronic malnutrition**
- C Acute on chronic malnutrition
- D None of the above

CORRECT

RIGHT ANSWER

- B. Chronic malnutrition**

WHY THIS IS RIGHT

Different anthropometric indices help distinguish between acute and chronic malnutrition in children.

- Height-for-age reflects long-term nutritional status (stunting) -> indicator of chronic malnutrition
- Weight-for-height reflects current nutritional status (wasting) -> indicator of acute malnutrition

Application to the question:

- Height-for-age = -2 SD -> indicates stunting -> chronic malnutrition
- Weight-for-height = -1 SD -> within normal/mild range, does not indicate acute malnutrition

QUESTION

| What is the rate of increment of head circumference in the first three months of life?

A 3 cm per month

B 2 cm per month

CORRECT

C 1 cm per month

D 0.5 cm per month

RIGHT ANSWER

OK B. 2 cm per month

WHY THIS IS RIGHT

Head circumference is a key indicator of brain growth in early infancy, with the most rapid increase occurring in the first few months of life.

Key growth pattern:

- 0-3 months: increases by about 2 cm per month
- 3-6 months: increases by about 1 cm per month
- 6-12 months: increases by about 0.5 cm per month

Clinical importance:

- Rapid head growth reflects normal brain development
- Deviations may indicate conditions like microcephaly or hydrocephalus

QUESTION

Treatment non-responsive Severe Acute Malnutrition (SAM) includes all except:

- A** Appetite not increased by day 4
- B** No edema reduction by day 4
- C** **No Complete absence of edema by day 20**
- D** Weight gain less than 5 gm/kg/day for 3 consecutive days

CORRECT

RIGHT ANSWER

OK

C. No Complete absence of edema by day 20

WHY THIS IS RIGHT

Non-responsive severe acute malnutrition (SAM) is identified when a child fails to show expected clinical or nutritional improvement despite appropriate treatment, prompting evaluation for underlying causes or complications.

Criteria suggesting non-response:

- Poor appetite not improving by day 3-4
- Failure of edema to start reducing by day 4
- Inadequate weight gain (<5 g/kg/day) over consecutive days

Why option C is incorrect:

- Complete resolution of edema is expected by around 2-3 weeks, but not necessarily by exactly day 20
- Lack of complete edema disappearance by day 20 alone is not used as a standard criterion for non-response

QUESTION

Which among the following is not included in the severe acute malnutrition definition?

A Weight for height

B Height for age

CORRECT

C MUAC <11.5

D Pedal oedema

RIGHT ANSWER

OK

B. Height for age

WHY THIS IS RIGHT

Severe acute malnutrition (SAM) is defined using indicators of acute nutritional status, not chronic growth failure.

Diagnostic criteria for SAM:

- Weight-for-height/length < -3 SD
- Mid-upper arm circumference (MUAC) < 11.5 cm (6-59 months)
- Presence of bilateral pitting edema

Why height-for-age is not included:

- Height-for-age reflects chronic malnutrition (stunting), not acute malnutrition
- SAM focuses on wasting and edema, which indicate recent or severe nutritional deficiency

QUESTION

Which of the following statements is true regarding Marasmus in children?

- A Seen due to calories deficiency
- B Protein loss is responsible for edema in this patient
- C Subcutaneous fat is preserved
- D Voracious appetite is seen

CORRECT

RIGHT ANSWER

- OK
- D. Voracious appetite is seen

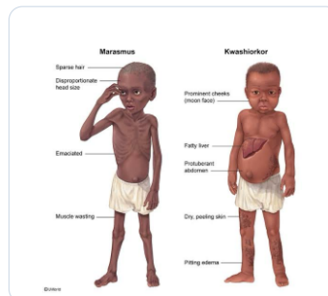
WHY THIS IS RIGHT

Marasmus is a form of severe acute malnutrition caused by deficiency of total caloric intake (both protein and energy), leading to severe wasting without edema.

- Marked loss of subcutaneous fat and muscle mass -> “skin and bones” appearance
- No edema (distinguishes it from kwashiorkor)
- Child is typically alert with a voracious appetite
- Growth failure with significant weight loss

Why others are incorrect:

- A is incomplete: marasmus involves deficiency of both calories and protein
- B describes kwashiorkor (protein deficiency -> edema)
- C is incorrect: subcutaneous fat is markedly reduced



QUESTION

Which of the following condition best explains the development of hypophosphatemia and hypomagnesemia in a malnourished child following the initiation of nutritional rehabilitation?

A Mechanism of refeeding syndrome

CORRECT

B Insulin mediated uptake of electrolytes

C Urinary loss of phosphates

D None of the above

RIGHT ANSWER

OK

A. Mechanism of refeeding syndrome

WHY THIS IS RIGHT

Refeeding syndrome occurs when nutrition is reintroduced in a severely malnourished child, leading to rapid metabolic and electrolyte shifts driven by a sudden increase in insulin secretion.

- Starvation state: low insulin, high glucagon -> depletion of intracellular electrolytes (phosphate, magnesium, potassium)
- On refeeding (especially carbohydrates): insulin release increases -> drives glucose and electrolytes (phosphate, magnesium, potassium) into cells
- This results in hypophosphatemia, hypomagnesemia, and hypokalemia in the blood
- Hypophosphatemia is the hallmark and can lead to muscle weakness, respiratory failure, cardiac dysfunction, and hemolysis
- Hypomagnesemia can cause arrhythmias and neuromuscular irritability

Why others are incomplete:

- Insulin-mediated uptake is part of the mechanism but does not fully define the syndrome
- Urinary loss is not the primary cause

QUESTION

A three-year-old child presents with cough, wheeze, and recurrent infection for which he has been taking oral medications. He now presents with excessive weight gain, a round face, hirsutism, and hair growth in the forehead. Developmental milestones were normal, child's BMI is more than +2 standard deviation, but height for age is below -2 standard deviation. What is the next best step in management?

- A** Checking for congenital hypothyroidism with T3, T4, TSH.
- B** Checking for early morning cortisol and history of exogenous cortisol administration. CORRECT
- C** Advise exercise and dietary changes for the child.
- D** Evaluate for genetic causes of obesity.

RIGHT ANSWER

- OK** **B. Checking for early morning cortisol and history of exogenous cortisol administration.**

WHY THIS IS RIGHT

Exogenous Cushing syndrome should be suspected in a child with excessive weight gain, growth failure (short stature), and cushingoid features such as moon face, hirsutism, and facial fullness, especially with a history of recurrent respiratory illness suggesting possible prolonged corticosteroid use.

- Weight gain with decreased height velocity is a red flag for endocrine pathology (not simple obesity)
- Features like moon face, hirsutism, and truncal obesity point toward hypercortisolism
- Most common cause in children is exogenous steroid use (e.g., repeated oral steroids for wheeze/asthma)
- Initial step is to take a detailed drug history and check early morning cortisol (suppressed in exogenous cause)

Contrast:

- Simple obesity: weight gain with normal or increased height
- Hypothyroidism: weight gain is mild with lethargy, constipation, and delayed milestones
- Genetic obesity: usually early onset with normal or increased height initially

QUESTION

A 7-year-old child presents with short stature (height is -5 SD). The child is eating well and gaining weight. On examination, upper segment to lower segment ratio is 1.4:1 and systemic examinations are within normal. What is the most likely diagnosis?

A Spondyloepiphyseal dysplasia

B Achondroplasia

CORRECT

C Growth hormone deficiency

D Severe malnutrition

RIGHT ANSWER

OK B. Achondroplasia

WHY THIS IS RIGHT

Achondroplasia is a skeletal dysplasia characterized by disproportionate short stature with relatively normal trunk length and shortened limbs (rhizomelic shortening). A key clinical clue is an increased upper segment to lower segment ratio, reflecting short limbs compared to the trunk.

- Disproportionate short stature with normal weight gain and nutrition
- Increased upper segment to lower segment ratio
- Normal intelligence and systemic examination
- Due to mutation in FGFR3 causing impaired endochondral ossification

Contrast:

- Growth hormone deficiency: proportionate short stature with delayed growth and often poor weight gain
- Severe malnutrition: both height and weight are reduced
- Spondyloepiphyseal dysplasia: primarily affect

QUESTION

A teenage boy, who was previously raised as a girl, presents with primary amenorrhea and absence of secondary sexual characteristics. On examination, he is tall, has small testes, and gynecomastia. Serum testosterone levels are within the normal range. Which of the following is the most likely diagnosis?

A 45, X

B 46, XX

C 47, XXY

CORRECT

D 47, XYY

RIGHT ANSWER

OK

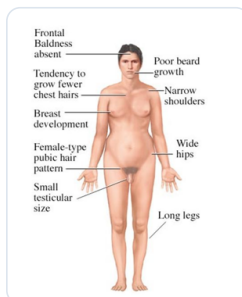
C. 47, XXY

WHY THIS IS RIGHT

Tall stature, small testes, gynecomastia, and primary amenorrhea-like presentation with normal-range testosterone point to Klinefelter syndrome (47, XXY).

It causes primary testicular failure with low fertility and often learning/behavioral issues.

Labs typically show $\uparrow$ FSH/ $\uparrow$ LH with relatively low-normal testosterone.



QUESTION

A young boy is brought to the clinic for developmental assessment. On examination, he has a prominent epicanthal fold, a single transverse palmar crease, hypotonia and intellectual disability. Facial features appear flat with upslanting palpebral fissures. What is the most likely diagnosis?

A Down Syndrome

CORRECT

B Patau Syndrome

C Edward Syndrome

D Fragile X Syndrome

RIGHT ANSWER

OK

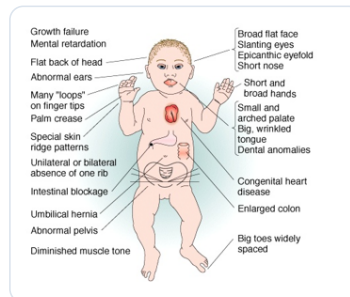
A. Down Syndrome

WHY THIS IS RIGHT

The findings-flat facial profile, upslanting palpebral fissures, epicanthal folds, hypotonia, intellectual disability, and single transverse palmar crease-are classic for Down syndrome (Trisomy 21).

Common associated issues include AV septal defect, duodenal atresia, and hypothyroidism.

Early screening for cardiac, hearing, vision, and thyroid problems is essential.



QUESTION

Which genetic syndrome results from a deletion of the short arm of chromosome 5 (5p deletion)?

A Edward syndrome

B Patau syndrome

C Cri du chat syndrome

CORRECT

D Turner Syndrome

RIGHT ANSWER

OK C. Cri du chat syndrome

WHY THIS IS RIGHT

Cri du chat syndrome is caused by deletion of the short arm of chromosome 5 (5p deletion) and is characterized by a distinctive high-pitched “cat-like” cry in infancy.

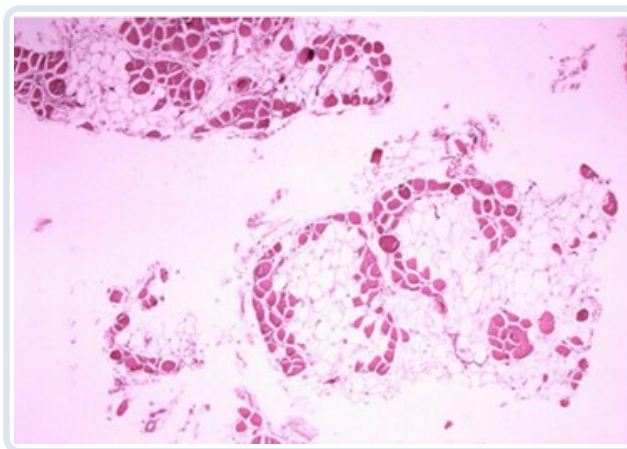
- Genetic defect: deletion of 5p
- Clinical features:
 - High-pitched cry
 - Microcephaly
 - Intellectual disability
 - Hypertelorism and facial dysmorphism

Differentiation:

- Edward syndrome: trisomy 18
- Patau syndrome: trisomy 13
- Turner syndrome: monosomy X (45,X)

QUESTION

An 8-year-old child has difficulty walking and getting up from a squatting position. A muscle biopsy was done and is as shown in the image. Which of the following is true about this condition?



A Death occurs in 3rd decade

CORRECT

B Previous history of viral prodrome

C It is a mitochondrial storage disorder

D Early treatment has excellent prognosis

RIGHT ANSWER

OK

A. Death occurs in 3rd decade

WHY THIS IS RIGHT

Duchenne muscular dystrophy (DMD) is an X-linked recessive disorder caused by mutation in the dystrophin gene, leading to progressive muscle degeneration.

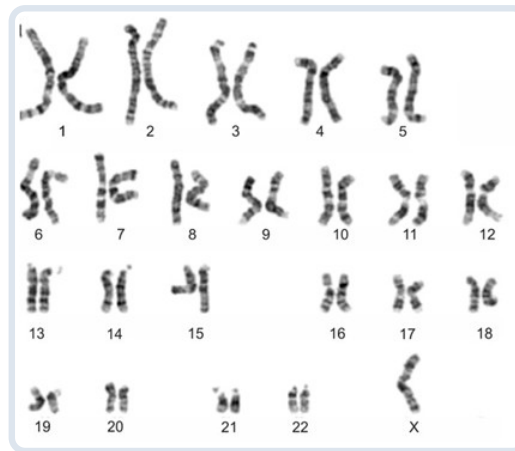
- Presents in early childhood with proximal muscle weakness and Gowers' sign (difficulty getting up from squatting)
- Muscle biopsy shows muscle fiber degeneration with replacement by fat and connective tissue
- Serum creatine kinase (CK) is markedly elevated
- Progressive weakness leading to loss of ambulation by adolescence
- Death usually occurs in the second to third decade due to respiratory or cardiac failure

Differentiation:

- Not associated with viral prodrome (rules out myositis)
- Not a mitochondrial disorder

QUESTION

A 15-year-old girl with wide-spaced nipples, short height, and the following karyotype.



A Turner syndrome

CORRECT

B Klinefelter syndrome

C Down syndrome

D Partial mole

RIGHT ANSWER

OK

A. Turner syndrome

WHY THIS IS RIGHT

Turner syndrome is a chromosomal disorder characterized by monosomy X (45,X), leading to gonadal dysgenesis and characteristic physical features.

- Clinical features: short stature, webbed neck, shield chest with widely spaced nipples, primary amenorrhea
- Karyotype: 45,X (absence of one X chromosome)
- Streak gonads → infertility

Associated conditions:

- Congenital heart defects (e.g., coarctation of aorta)
- Renal anomalies
- Hypothyroidism

Differentiation:

- Klinefelter syndrome: 47,XXY males
- Down syndrome: trisomy 21
- Partial mole: triploid karyotype

QUESTION

An abnormally tall patient presents with lens subluxation, long limbs, and arachnodactyly. A mutation in which of the following genes is most commonly associated with this condition?

A Fibrillin-1 gene (FBN1)

CORRECT

B Collagen Type I gene

C Elastin gene

D Dystrophin gene

RIGHT ANSWER

OK

A. Fibrillin-1 gene (FBN1)

WHY THIS IS RIGHT

Marfan syndrome is an autosomal dominant connective tissue disorder caused by mutation in the FBN1 gene encoding fibrillin-1, leading to abnormal elastic fiber formation and increased TGF- β activity

Clinical features: tall stature, long limbs (dolichostenomelia), arachnodactyly, lens subluxation (typically upward), and aortic root dilation

- Risk of aortic aneurysm and dissection (most serious complication)
- Requires regular cardiovascular monitoring

Contrast:

- Collagen type I defect -> osteogenesis imperfecta
- Elastin gene defect -> supravalvular aortic stenosis (e.g., Williams syndrome)
- Dystrophin defect -> Duchenne muscular dystrophy

QUESTION

| Which of the following is not seen in down syndrome?

A Hypothyroidism

B Hearing loss

C Short stature

D Caudal regression syndrome

CORRECT

RIGHT ANSWER

OK D. Caudal regression syndrome

WHY THIS IS RIGHT

Down syndrome (Trisomy 21) is associated with multiple systemic abnormalities affecting endocrine, auditory, and growth parameters.

- Common associations include hypothyroidism, hearing loss, and short stature
- Other features: intellectual disability, characteristic facies, congenital heart defects (e.g., AV septal defect)

Why caudal regression syndrome is not seen:

- Caudal regression syndrome is a separate congenital anomaly associated with maternal diabetes, not Down syndrome

QUESTION

A tall 10-year-old boy presents with Sexual Maturity Rating 5 and hyperpigmentation. His BP is 150/100 mm Hg. What is the likely diagnosis?

A 11 β -Hydroxylase deficiency

CORRECT

B 21 β -Hydroxylase deficiency

C 17 β -Hydroxylase deficiency

D SCC deficiency

RIGHT ANSWER

OK

A. 11 β -Hydroxylase deficiency

WHY THIS IS RIGHT

11 β -Hydroxylase deficiency in congenital adrenal hyperplasia causes hypertension due to salt retention and precocious puberty due to excess androgen production.

| Enzyme Deficiency | Mineralocorticoids | Glucocorticoids | Androgens | Symptoms |

| ----- | ----- | ----- | ----- | ----- |

| 11 β -hydroxylase | $\uparrow^*$ | $\downarrow$ | $\uparrow$ | • Hypertension, $\downarrow$ K $\square$

• Ambiguous genitalia in girls |

| 17 α -hydroxylase | $\uparrow$ | $\uparrow^{**}$ | $\downarrow$ | • Hypertension, $\downarrow$ K $\square$

• Ambiguous genitalia in boys •

Absent puberty |

| 21-hydroxylase | $\downarrow$ | $\downarrow$ | $\uparrow$ | • Salt wasting (vomiting, hypotension, $\downarrow$ Na $\square$, $\uparrow$ K $\square$)

• Ambiguous genitalia in girls |

QUESTION

| Which of the following is not an example of an aneuploidy disorder?

CORRECT

A Bloom syndrome

B Trisomy 21

C Trisomy 13

D Klinefelter syndrome

RIGHT ANSWER

OK A. Bloom syndrome

WHY THIS IS RIGHT

Aneuploidy refers to an abnormal number of chromosomes due to gain or loss of whole chromosomes, typically caused by nondisjunction.

- Trisomy 21, Trisomy 13, and Klinefelter syndrome (47,XXY) are classic examples of aneuploidy
- These conditions involve numerical chromosomal abnormalities

Why Bloom syndrome is not aneuploidy:

- Bloom syndrome is a chromosomal instability disorder caused by mutation in the BLM gene
- It involves defective DNA repair and increased sister chromatid exchanges, not a change in chromosome number

QUESTION

Which of the following is not seen in Turner syndrome?

A Widely spaced nipples

B Short stature

C Prominent occiput

CORRECT

D Barr body

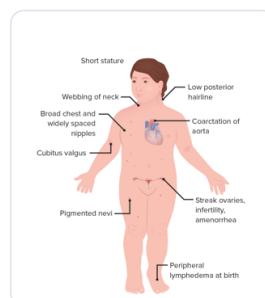
RIGHT ANSWER

OK C. Prominent occiput

WHY THIS IS RIGHT

Turner syndrome is a chromosomal disorder (45,XO) characterized by gonadal dysgenesis and distinctive somatic features, with absence of Barr body due to monosomy X.

- Common features include short stature, webbed neck, shield chest with widely spaced nipples, and streak gonads
- Other findings: coarctation of aorta, lymphedema at birth, primary amenorrhea
- Barr body is absent because there is only one X chromosome
- Prominent occiput is not a feature of Turner syndrome



QUESTION

| Which of the following has autosomal recessive pattern of inheritance?

- A** Achondroplasia
- B** Huntington's disease
- C** Cystic fibrosis
- D** Familial Hypercholesterolemia

CORRECT

RIGHT ANSWER

- OK** C. Cystic fibrosis

WHY THIS IS RIGHT

Cystic fibrosis is an autosomal recessive disorder caused by mutations in the CFTR gene, which encodes a chloride channel involved in fluid and electrolyte transport across epithelial cells.

Defective CFTR leads to thick, viscous secretions in multiple organs, resulting in:

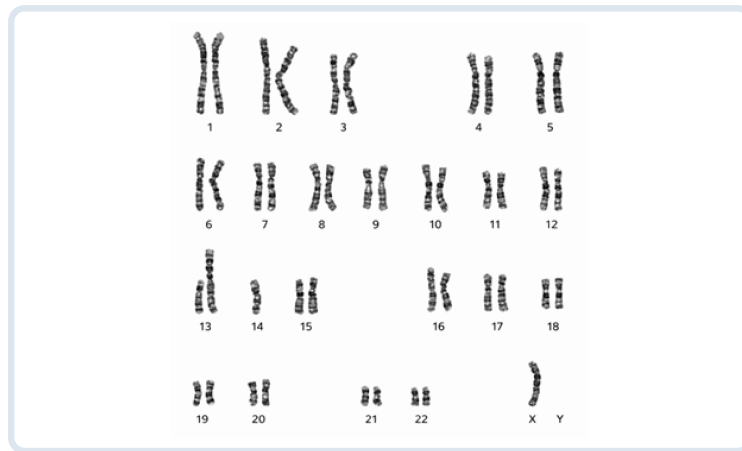
- Chronic lung infections
- Pancreatic insufficiency and malabsorption
- Meconium ileus in newborns

Why other options are incorrect:

- Achondroplasia: Autosomal dominant (FGFR3 mutation).
- Huntington's disease: Autosomal dominant (trinucleotide repeat disorder).
- Familial hypercholesterolemia: Typically autosomal dominant (LDL receptor defect).

QUESTION

A female patient with infertility presented to the clinic and underwent karyotyping. What is the likely diagnosis in the karyotype shown?



A Down syndrome

B Turner syndrome

CORRECT

C Klinefelter syndrome

D Edward syndrome

RIGHT ANSWER

OK

B. Turner syndrome

WHY THIS IS RIGHT

The karyotype shows monosomy X (45,X), which is diagnostic of Turner syndrome, a common chromosomal cause of female infertility.

- Absence of one X chromosome -> 45, X karyotype
- No Barr body present

Clinical features:

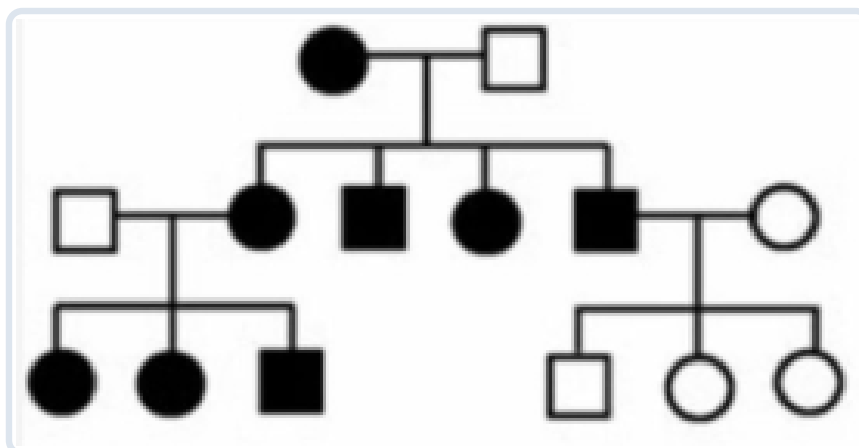
- Primary amenorrhea & infertility
- Streak ovaries (gonadal dysgenesis)
- Short stature
- Webbed neck
- Shield chest with widely spaced nipples

Why other options are incorrect:

- Down syndrome: Trisomy 21
- Klinefelter syndrome: 47,XXY (male infertility)
- Edward syndrome: Trisomy 18

QUESTION

Which of the following diseases is shown by the pedigree chart?



- A Prader-Willi Syndrome
- B Achondroplasia
- C Kearns-Sayre Syndrome**
- D Williams syndrome

CORRECT

RIGHT ANSWER

- OK C. Kearns-Sayre Syndrome**

WHY THIS IS RIGHT

The pedigree shows maternal transmission, where an affected mother passes the disease to all offspring, while affected fathers do not transmit it.

This inheritance pattern is characteristic of mitochondrial disorders.

Kearns-Sayre syndrome is a classic mitochondrial disease caused by mtDNA deletions.

Classic Features (Triad)

- Progressive external ophthalmoplegia
- Pigmentary retinopathy
- Cardiac conduction defects

Pattern	Key Feature	Example
-----	-----	-----
Mitochondrial	Maternal only	Kearns-Sayre
Autosomal dominant	Every generation	Achondroplasia
Imprinting	Parent-specific	Prader-Willi
Microdeletion	Sporadic	Williams

QUESTION

Match the following:

Column A	Column B
A. Mitochondrial inheritance	1. Spinobulbar dystrophia
B. XLR	2. Cystic fibrosis
C. AR	3. Leber's disease
D. Trinucleotide repeat	4. Duchene's muscular dystrophy

A A-3, B-4, C-2, D-1

CORRECT

B A-1, B-2, C-4, D-3

C A-2, B-3, C-1, D-4

D A-4, B-1, C-3, D-2

RIGHT ANSWER

OK A. A-3, B-4, C-2, D-1

WHY THIS IS RIGHT

Mitochondrial inheritance is classically seen in Leber's hereditary optic neuropathy due to mtDNA mutations. Inherited only from mother.

X-linked recessive disorders include Duchenne muscular dystrophy caused by dystrophin gene deletion. Features:

- Gowers' sign
- Calf pseudohypertrophy

Autosomal recessive inheritance is typical of cystic fibrosis due to CFTR mutation. Features:

- Thick mucus
- Lung infections
- Pancreatic insufficiency

Spinobulbar muscular atrophy is caused by trinucleotide (CAG) repeat expansion. Also called: Kennedy disease

Similar concept seen in:

- Huntington disease
- Fragile X

QUESTION

| All of the following are characteristic features of Turner syndrome, except:

A Prominent occiput

CORRECT

B Short 4th metacarpal

C Webbed neck

D Wide spaced nipples

RIGHT ANSWER

OK

A. Prominent occiput

WHY THIS IS RIGHT

Turner syndrome (45,X) shows classic somatic features like webbed neck, shield chest with wide-spaced nipples, and short 4th metacarpal due to SHOX gene haploinsufficiency.

Classic Phenotype

- Short stature
- Webbed neck
- Shield chest
- Wide spaced nipples
- Streak ovaries -> primary amenorrhea

Skeletal abnormalities and lymphatic dysgenesis are characteristic.

Prominent occiput is not a recognized feature of Turner syndrome and is more associated with certain craniofacial anomalies in other syndromes.

A. Prominent occiput- Incorrect statement

- Not a feature of Turner syndrome
- Seen more in:
- Trisomy 18 (Edwards syndrome)
- Some neural tube defects

B. Short 4th metacarpal

- Gives knuckle dimpling when making a fist
- Also seen in:
- Pseudohypoparathyroidism

C. Webbed neck

- Due to cystic hygroma in fetal life
- Leaves behind lateral neck folds

QUESTION

A 5-year-old child presents with exertional dyspnoea and a history of cyanotic spell. On examination, there is clubbing and a harsh systolic murmur heard at left upper sternal border. Chest X-ray is shown below. What is the most likely diagnosis?



A Tetralogy of Fallot

CORRECT

B TAPVC

C TGA

D COA

RIGHT ANSWER

OK **A. Tetralogy of Fallot**

WHY THIS IS RIGHT

Tetralogy of Fallot is the most common cyanotic congenital heart disease beyond infancy, presenting with exertional dyspnea, cyanotic spells, clubbing, and a systolic murmur.

- Components (PROV):
- Pulmonary stenosis
- Right ventricular hypertrophy
- Overriding aorta
- Ventricular septal defect

Clinical features:

- Cyanotic (tet) spells due to increased right-to-left shunting
- Harsh systolic murmur at left upper sternal border
- Squatting relieves symptoms

Chest X-ray finding: "Boot-shaped heart" due to right ventricular hypertrophy

Differentiation:

- TAPVC: snowman sign
- TGA: egg-on-string appearance
- Coarctation: rib notching

QUESTION

| Which of the following is not a component of Tetralogy of Fallot (TOF)?

A ASD, left ventricular hypertrophy

CORRECT

B Pulmonary stenosis

C Overriding of aorta

D Right ventricular hypertrophy

RIGHT ANSWER

OK

A. ASD, left ventricular hypertrophy

WHY THIS IS RIGHT

Tetralogy of Fallot is a cyanotic congenital heart disease characterized by four structural abnormalities resulting in right-to-left shunting. Right ventricular outflow obstruction leads to right-to-left shunt and cyanosis

Severity of pulmonary stenosis determines clinical presentation

Components (remember: PROV):

- Pulmonary stenosis
- Right ventricular hypertrophy
- Overriding aorta
- Ventricular septal defect (VSD)

Why option A is incorrect:

- ASD is not a component of TOF
- Left ventricular hypertrophy is not seen; instead, right ventricular hypertrophy occurs

QUESTION

A one-month-old child presents with recurrent pneumonia and no cyanosis. On examination, a continuous murmur is heard below the left clavicle and a bounding pulse is present. What is the most likely diagnosis?

A ASD (Atrial Septal Defect)

B VSD (Ventricular Septal Defect)

C PDA (Patent Ductus Arteriosus)

CORRECT

D Coarctation of Aorta

RIGHT ANSWER

OK

C. PDA (Patent Ductus Arteriosus)

WHY THIS IS RIGHT

Patent ductus arteriosus is a congenital left-to-right shunt in which the ductus arteriosus fails to close after birth, leading to continuous blood flow from the aorta to the pulmonary artery.

- Continuous “machinery” murmur best heard below the left clavicle
- Bounding pulses and wide pulse pressure due to runoff of blood into pulmonary circulation
- Recurrent respiratory infections and signs of heart failure due to increased pulmonary blood flow
- Usually no cyanosis initially (acyanotic congenital heart disease)

Pathophysiology:

- Persistent connection between aorta and pulmonary artery -> left-to-right shunt
- Increased pulmonary blood flow -> pulmonary congestion -> recurrent pneumonia

Contrast:

- ASD: fixed split S2, no continuous murmur
- VSD: pansystolic murmur at left lower sternal border
- Coarctation of aorta: radio-femoral delay, upper limb hypertension, weak lower limb pulses

QUESTION

A child presents with recurrent sinusitis and repeated lung infection. Genetic testing shows a mutation in CFTR gene. Which test is considered gold standard to confirm the diagnosis?

A Sweat Chloride Test

CORRECT

B Lung Biopsy

C Chest X-ray

D Nasal Swab Culture

RIGHT ANSWER

OK

A. Sweat Chloride Test

WHY THIS IS RIGHT

A child with recurrent sinopulmonary infections and a CFTR gene mutation is suspected to have cystic fibrosis. The gold standard test for confirming the diagnosis is the sweat chloride test, which demonstrates elevated chloride levels due to defective chloride transport.

- Cystic fibrosis is caused by mutation in the CFTR gene leading to thick, viscous secretions
- Common features include recurrent sinusitis, chronic lung infections, malabsorption, and failure to thrive
- Sweat chloride test:
 - ≥ 60 mmol/L is diagnostic
 - 30-59 mmol/L is intermediate
- Newborn screening and genetic testing support diagnosis but confirmation is by sweat test

QUESTION

A 4-month-old infant is brought to the emergency department with a respiratory rate of 66 breaths per minute, along with chest indrawing and nasal flaring. There are no other significant findings. What is the most likely diagnosis?

A Pneumonia

CORRECT

B Severe Pneumonia

C Severe Asthma

D Common Cold

RIGHT ANSWER

OK

A. Pneumonia

WHY THIS IS RIGHT

According to IMNCI guidelines, a 2-12 month old infant with fast breathing (respiratory rate $\geq 50/\text{min}$) is classified as pneumonia. In this case, a respiratory rate of 66/min indicates fast breathing for age.

- Age-specific fast breathing:
 - <2 months: $\geq 60/\text{min}$
 - 2-12 months: $\geq 50/\text{min}$
 - 1-5 years: $\geq 40/\text{min}$
- Chest indrawing in infants may be present due to compliant chest wall and does not always indicate severe disease
- Severe pneumonia is diagnosed when there are danger signs such as inability to feed, lethargy, convulsions, or central cyanosis

QUESTION

| In which disease among the following, Chloride level in sweat is evaluated?

A

Phenylketonuria

B

Cystic fibrosis

CORRECT

C

Gaucher's disease

D

Osteogenesis imperfecta

RIGHT ANSWER

OK

B. Cystic fibrosis

WHY THIS IS RIGHT

Sweat chloride test is the gold standard diagnostic test for cystic fibrosis, reflecting defective chloride transport due to CFTR mutation.

- CFTR dysfunction -> impaired chloride reabsorption in sweat glands -> increased chloride concentration in sweat
- Sweat chloride level >60 mmol/L is diagnostic (in appropriate clinical setting)

Clinical relevance:

- Used in children with recurrent respiratory infections, failure to thrive, or steatorrhea
- Helps confirm diagnosis early and guide management

QUESTION

| Choose the true statement regarding CF (Cystic Fibrosis) :

A Chloride secretion is decreased in sweat and increased in pancreatic fluid

B Chloride secretion is increased in sweat and decreased in pancreatic fluid

CORRECT

C Chloride secretion is decreased in both sweat and pancreatic fluid

D Chloride secretion is increased in both sweat and pancreatic fluid

RIGHT ANSWER

OK

B. Chloride secretion is increased in sweat and decreased in pancreatic fluid

WHY THIS IS RIGHT

Cystic fibrosis is caused by CFTR channel dysfunction leading to abnormal chloride transport in different tissues, producing opposite effects in sweat glands and exocrine organs.

- In sweat glands: defective reabsorption of chloride -> increased chloride in sweat (basis of sweat chloride test)
- In pancreas and other exocrine glands: decreased chloride secretion -> thick, viscous secretions causing ductal obstruction

Clinical correlation:

- High sweat chloride is diagnostic
- Pancreatic insufficiency leads to malabsorption and failure to thrive

QUESTION

A 2-year-old child is brought to the emergency department with fever, hoarse voice, and a barking cough for 2 days. On examination, the child has inspiratory stridor that worsens at night. There is no drooling, and the child is able to swallow.

Which of the following is the most likely causative organism?

- A Haemophilus influenzae
- B Streptococcus
- C Parainfluenza virus
- D Corynebacterium diphtheriae

CORRECT

RIGHT ANSWER

OK C. Parainfluenza virus

WHY THIS IS RIGHT

- Barking cough + stridor + hoarseness -> classic triad
- Age: 6 months - 3 years
- No drooling -> rules out epiglottitis
- Most common cause of croup = Parainfluenza virus (Type 1)
- X-ray: Steeple sign
- Treatment: Steroids (dexamethasone) ± nebulized adrenaline

QUESTION

| All are seen in Cystic fibrosis except:

A Biliary atresia

CORRECT

B Meconium ileus

C Nasal polyp

D Bleeding diathesis

RIGHT ANSWER

OK

A. Biliary atresia

WHY THIS IS RIGHT

Cystic fibrosis is an autosomal recessive disorder caused by CFTR gene mutation leading to thick, viscous secretions affecting multiple organs.

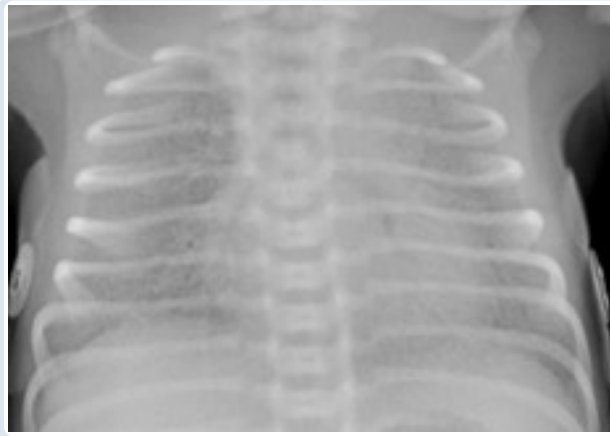
- Gastrointestinal: meconium ileus in neonates, pancreatic insufficiency causing malabsorption
- Respiratory: recurrent infections, bronchiectasis, nasal polyps
- Hepatobiliary: can cause biliary cirrhosis but not biliary atresia
- Hematological: fat-soluble vitamin (vitamin K) deficiency -> bleeding tendency

Why biliary atresia is not seen:

- Biliary atresia is a separate obstructive condition of the extrahepatic bile ducts, not related to CFTR dysfunction (bile duct obstruction is seen in CF, not atresia)

QUESTION

A 34-week newborn presents with breathing difficulty that improves with FiO_2 at 30%. What is the most likely diagnosis?



A Hyaline membrane disease

CORRECT

B Transient tachypnea of newborn

C Meconium aspiration syndrome

D None of the above

RIGHT ANSWER

OK

A. Hyaline membrane disease

WHY THIS IS RIGHT

Hyaline membrane disease (neonatal respiratory distress syndrome) occurs due to surfactant deficiency (produced by type II pneumocytes), most commonly in preterm infants, leading to alveolar collapse and impaired gas exchange.

- Seen in preterm or late preterm infants (like 34 weeks)
- Presents with respiratory distress soon after birth that improves with supplemental oxygen
- Chest X-ray shows diffuse reticulogranular (ground-glass) appearance with air bronchograms and reduced lung volumes
- Managed with oxygen, CPAP, and exogenous surfactant if needed
- Antenatal steroids reduce risk in preterm delivery

Differentiation:

- Transient tachypnea of newborn: hyperinflation, fluid in fissures, milder disease
- Meconium aspiration syndrome: patchy infiltrates, hyperinflation, term/post-term

QUESTION

A 10-year-old child presents with diarrhea and weight loss. On examination, the height and weight are lesser than expected. Laboratory investigations were positive for class II HLA-DQ2. Which of the following will you advise the child?

- A Fat-free diet
- B Lactose-free diet
- C Low carbohydrate diet

D Gluten-free diet

CORRECT

RIGHT ANSWER

D. Gluten-free diet

WHY THIS IS RIGHT

Celiac disease is an autoimmune disorder triggered by gluten in genetically predisposed individuals, commonly associated with HLA-DQ2 (or DQ8), leading to malabsorption and growth failure.

- Presents with chronic diarrhea, weight loss, and short stature
- Associated with HLA-DQ2/DQ8 positivity
- Pathology: villous atrophy in small intestine

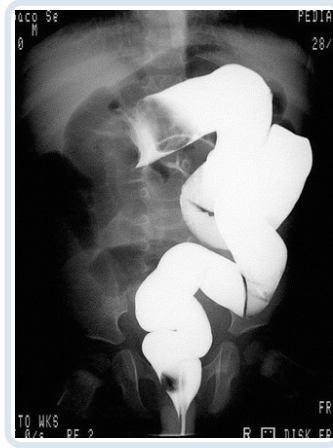
Management:

- Lifelong strict gluten-free diet (avoid wheat, barley, rye)
- Leads to symptom resolution and catch-up growth

Clinical importance: Early dietary intervention prevents complications like malnutrition and delayed growth

QUESTION

A baby presented with abdominal pain. On examination, a mass is palpated in the right lumbar region. A barium enema is done, and the image is given below. What is the likely diagnosis?



A Intussusception

CORRECT

B Volvulus

C Duodenal atresia

D Intestinal obstruction

RIGHT ANSWER

OK **A. Intussusception**

WHY THIS IS RIGHT

Intussusception is the most common cause of intestinal obstruction in infants and young children, caused by telescoping of one segment of bowel into another.

- Presents with intermittent abdominal pain, vomiting, and a palpable abdominal mass (often right sided)
- May have "red currant jelly" stools (blood + mucus)
- Barium or air enema shows characteristic "coil spring" or "claw" sign

Clinical significance:

- Barium/air enema is both diagnostic and therapeutic
- Early diagnosis prevents complications like bowel ischemia and perforation

Differentiation:

- Volvulus: bilious vomiting, corkscrew appearance
- Duodenal atresia: double bubble sign in neonates

QUESTION

1-month-old baby came with complaints of projectile vomiting with an olive-shaped mass. X-ray abdomen was done which is shown. What is the diagnosis?



A Pyloric stenosis

CORRECT

B Intestinal obstruction

C Duodenal atresia

D Jejunal atresia

RIGHT ANSWER

OK **A. Pyloric stenosis**

WHY THIS IS RIGHT

Congenital hypertrophic pyloric stenosis is a common cause of non-bilious projectile vomiting in early infancy due to hypertrophy of the pyloric muscle causing gastric outlet obstruction.

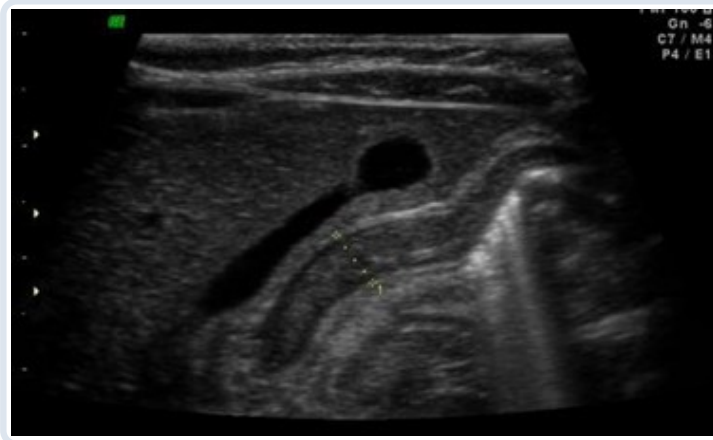
- Presents at 2-6 weeks with projectile non-bilious vomiting and an “olive-shaped” mass
- X-ray may show a distended stomach with paucity of distal bowel gas
- Ultrasound is the investigation of choice (elongated, thickened pylorus)

Clinical significance:

- Leads to hypochloremic, hypokalemic metabolic alkalosis
- Management requires fluid and electrolyte correction followed by pyloromyotomy

QUESTION

A 1-month-old baby presents with an olive-shaped mass and recurrent vomiting. An ultrasound (USG) was performed, as shown below. What is your diagnosis?



A CHPS (Congenital Hypertrophic Pyloric Stenosis)

CORRECT

B Intussusception

C Meckel's Diverticulum

D None of the above

RIGHT ANSWER

OK **A. CHPS (Congenital Hypertrophic Pyloric Stenosis)**

WHY THIS IS RIGHT

Congenital hypertrophic pyloric stenosis is a common cause of gastric outlet obstruction in early infancy, characterized by hypertrophy of the pyloric muscle.

- Presents at 2-6 weeks with non-bilious projectile vomiting and an "olive-shaped" palpable mass
- Infant is typically hungry after vomiting
- Ultrasound is the investigation of choice showing elongated and thickened pylorus (target or donut sign)
- Hypochloremic, hypokalemic metabolic alkalosis due to repeated vomiting

Management:

- Initial stabilization with fluids and electrolyte correction
- Definitive treatment is pyloromyotomy

QUESTION

A 24-hour-old neonate presents with severe respiratory distress and is admitted to the Intensive Care Unit (ICU). A chest X-ray of the neonate is provided. What is the most probable diagnosis?



A Congenital Pulmonary Airway Malformation (CPAM)

B Congenital Diaphragmatic Hernia (CDH)

CORRECT

C Congenital Lobar Emphysema

D Neonatal Pneumonia

RIGHT ANSWER

OK B. Congenital Diaphragmatic Hernia (CDH)

WHY THIS IS RIGHT

Congenital diaphragmatic hernia is a neonatal surgical emergency characterized by herniation of abdominal contents into the thoracic cavity, leading to lung compression and severe respiratory distress.

- Presents soon after birth with respiratory distress, scaphoid abdomen, and decreased breath sounds
- Chest X-ray shows bowel loops in the thorax with mediastinal shift
- Most commonly left-sided (Bochdalek hernia)

Clinical importance:

- Causes pulmonary hypoplasia and pulmonary hypertension
- Immediate management includes endotracheal intubation and gastric decompression (NG tube)
- Bag-mask ventilation is contraindicated

QUESTION

A 1-day old neonate presented with scaphoid abdomen, respiratory distress and mediastinal shift. All of the following can be done except?

- A O2 by mask
- B Positive pressure ventilation by ventilator
- C ET intubation
- D Bag and mask ventilation**

CORRECT

RIGHT ANSWER

- OK
- D. Bag and mask ventilation**

WHY THIS IS RIGHT

Congenital diaphragmatic hernia presents with respiratory distress, scaphoid abdomen, and mediastinal shift due to herniation of abdominal contents into the thoracic cavity causing lung compression.

- Immediate management focuses on preventing further gastric distension and lung compression
- Bag and mask ventilation is contraindicated because it forces air into the stomach, worsening respiratory compromise
- Endotracheal intubation with controlled ventilation is preferred
- Nasogastric tube should be inserted for gastric decompression

QUESTION

| What is the fluid of choice in pediatric patients?

- A RL
- B DNS**
- C N/2 saline
- D 5% dextrose

CORRECT

RIGHT ANSWER

OK **B. DNS**

WHY THIS IS RIGHT

In pediatric patients, maintenance fluid therapy should provide both water and glucose to prevent hypoglycemia and ketosis; hence dextrose-containing fluids are commonly used.

- DNS (dextrose normal saline) provides glucose along with sodium, making it suitable for many pediatric maintenance needs
- Children have limited glycogen stores and are prone to hypoglycemia during illness or fasting
- Pure crystalloids like normal saline or Ringer lactate do not provide glucose
- 5% dextrose alone lacks electrolytes and is not appropriate for routine use
- In emergency resuscitation (e.g., shock), isotonic fluids like normal saline are preferred initially

QUESTION

| Which of the following is the best indicator of some dehydration?

A Skin pinch

B Sunken eyeballs

C Urine output

D Irritable status

CORRECT

RIGHT ANSWER

OK

D. Irritable status

WHY THIS IS RIGHT

Some dehydration" is diagnosed when ≥ 2 of the following are present:

- Restless / irritable (early & sensitive sign)
- Sunken eyes
- Thirsty, drinks eagerly
- Skin pinch goes back slowly

Why Irritability is BEST indicator

- Reflects early CNS response to dehydration
- Appears before severe signs
- More reliable clinically than isolated physical signs

| Parameters | No Dehydration | Some Dehydration |

| ----- | ----- | ----- |

| Appearance | Well, alert | Restless, irritable |

| Eyes | Normal | Sunken |

| Thirst | Drinks normally,

not thirsty | Thirsty, drinks eagerly |

| Skin pinch | Goes back quickly

(<1 second) | Goes back slowly

(1 second) |

QUESTION

A 3-week-old infant presents with non-projectile, non-bilious vomiting and a normal abdominal examination. What is the most likely diagnosis?

A GERD

CORRECT

B CHPS

C Cow milk protein allergy

D Intestinal obstruction

RIGHT ANSWER

OK

A. GERD

WHY THIS IS RIGHT

Key Clinical Reasoning

- Age: 3 weeks (very early infancy)
- Vomiting:
- Non-projectile
- Non-bilious
- Abdominal exam: Normal

This pattern is classic for physiological reflux / GERD in infants

Why NOT others?

B. CHPS (Congenital Hypertrophic Pyloric Stenosis)

- Projectile vomiting (key feature)
- Usually presents at 3-6 weeks
- May have palpable olive, visible peristalsis

C. Cow milk protein allergy

- Associated with diarrhea, blood in stool, eczema
- Not isolated vomiting

D. Intestinal obstruction

- Bilious vomiting
- Abdominal distension

QUESTION

I All of the following statements regarding Hirschsprung disease are true except:

- A** Failure of migration of ganglion cells.
- B** Colonic segment proximal to the affected area is constricted. CORRECT
- C** Failure of passage of meconium in the postnatal period.
- D** Loss of coordinated peristalsis.

RIGHT ANSWER

- OK** **B. Colonic segment proximal to the affected area is constricted.**

WHY THIS IS RIGHT

Hirschsprung disease is caused by failure of neural crest cell migration, leading to absence of ganglion cells in the distal bowel (aganglionosis). This results in loss of coordinated peristalsis and functional obstruction.

Key concepts:

- The affected distal segment is spastic and constricted due to aganglionosis
- The proximal segment becomes dilated due to accumulation of intestinal contents (megacolon)
- Newborns typically present with failure to pass meconium within the first 48 hours
- There is absence of the rectoanal inhibitory reflex

QUESTION

A boy born to a normal father and mother had Hemophilia. Detailed history taking revealed that the mother's side had a family history of similar affected individuals. The children born to this boy are likely to have which of the following?

- A All boys and girl children equally affected
- B 50% boys affected and no girl children unaffected
- C No boy affected and 50% girl children are carrier
- D 25% boy affected and 25% girl children are carrier

CORRECT

RIGHT ANSWER

- OK
- C. No boy affected and 50% girl children are carrier

WHY THIS IS RIGHT

Hemophilia is an X-linked recessive disorder affecting males who inherit the mutant X chromosome. An affected male transmits his X chromosome to all daughters and Y chromosome to all sons. Hence, none of the sons are affected as they receive the father's Y chromosome.

All daughters receive the mutant X, making 100% daughters carriers; effectively, 50% of total children are carrier females.

Genotype setup:

- Affected male (boy): X^hY
- Assume wife is normal: X^HX^H
- Punnett Square:

Mother

$X^H X^H$

Father $X^h X^HX^h X^HX^h$

$Y X^HY X^HY$

- Interpretation:
- Daughters (X^HX^h) -> All are carriers (100%)
- Sons (X^HY) -> All are normal (0% affected)
- Final conclusion:
- No male child affected
- All female children are carriers

QUESTION

A 4-year-old child is brought to the clinic with a low grade fever, sore mouth and painful vesicular rash on the hands, feet and inside the mouth as shown in image below. The child attends daycare and similar cases have been reported among classmates. What is the most likely causative organism?



A Coxsackie A16

CORRECT

B EBV

C VZV

D Coxsackie B

RIGHT ANSWER

OK

A. Coxsackie A16

WHY THIS IS RIGHT

A child with low-grade fever, painful oral ulcers, and vesicular lesions on the hands and feet in a daycare setting is characteristic of hand, foot and mouth disease caused most commonly by Coxsackievirus A16.

- Spread is via feco-oral and respiratory routes, leading to outbreaks in children
- Typical features include oral ulcers, vesicles on palms and soles, and mild fever
- Illness is self-limiting and resolves within about a week

Differentials:

- Varicella: lesions in different stages, predominantly on trunk
- Epstein-Barr virus: pharyngitis with lymphadenopathy, no vesicular rash
- Coxsackie B -> Myocarditis, pleurodynia

QUESTION

A 8-year-old child presents with enuresis. The parents mention the child had a fall from bed once but has no other significant medical history. There are no signs of urinary tract infections or neurological deficits. What is the best next step in management?

A Behavioural modification

CORRECT

B Start anticholinergic drugs

C Prescribe Oxybutinin

D Start desmopressin therapy

RIGHT ANSWER

OK

A. Behavioural modification

WHY THIS IS RIGHT

Primary nocturnal enuresis in children is most often a benign, functional condition and is managed initially with non-pharmacological measures.

- Diagnosis is clinical after ruling out UTI, diabetes, and neurological causes
- Most children have normal bladder function and outgrow the condition

First-line management:

- Behavioral modification:
- Limit evening fluid intake
- Timed voiding before sleep
- Positive reinforcement (reward system)
- Enuresis alarm (most effective long-term)

Pharmacological therapy:

- Desmopressin or anticholinergics are reserved for refractory cases or specific indications

QUESTION

A 10-year-old presents with edema and anasarca. A diagnosis of minimal change disease is made. Which of the following is true about this condition?

A Light microscopy shows effacement of podocytes

B Good response to steroids

CORRECT

C Most common in adults

D Non-selective proteinuria

RIGHT ANSWER

OK **B. Good response to steroids**

WHY THIS IS RIGHT

Minimal change disease is the most common cause of nephrotic syndrome in children and is characterized by excellent response to corticosteroid therapy.

- Pathology:
- Light microscopy: appears normal
- Electron microscopy: effacement of podocyte foot processes
- Proteinuria is selective (mainly albumin)
- Most common in children, not adults
- Presents with edema, proteinuria, hypoalbuminemia
- Steroid-sensitive with good prognosis

Why other options are incorrect:

- Podocyte effacement is seen on electron microscopy, not light microscopy
- It is most common in children
- Proteinuria is selective, not non-selective

QUESTION

A male child presented with arthralgia and abdominal pain. On examination, there was palpable purpura over the lower limbs. There is a past history of upper respiratory tract infection prior to the onset of presenting symptoms. Which of the following is the treatment for this condition?

A Azathioprine

B Methotrexate

C Cyclosporine

D Glucocorticoids

CORRECT

RIGHT ANSWER

OK

D. Glucocorticoids

WHY THIS IS RIGHT

IgA vasculitis (Henoch-Schönlein purpura) is a small-vessel vasculitis seen in children, typically following an upper respiratory infection, presenting with palpable purpura, arthralgia, abdominal pain, and possible renal involvement.

- Classic tetrad:
- Palpable purpura (non-thrombocytopenic, mainly lower limbs)
- Arthralgia/arthritis
- Abdominal pain
- Renal involvement (hematuria/proteinuria)

Treatment: Usually self-limiting; supportive care is mainstay. Glucocorticoids are used in moderate to severe cases (significant abdominal pain, renal involvement)

Why others are incorrect:

- Azathioprine, methotrexate, cyclosporine are not first-line and used only in severe/refractory cases



QUESTION

Identify the condition:



A Bladder exstrophy

CORRECT

B Omphalocele

C Persistent vitellointestinal duct

D Gastroschisis

RIGHT ANSWER

OK A. Bladder exstrophy

WHY THIS IS RIGHT

Bladder exstrophy is a congenital defect with absence of the lower anterior abdominal wall, exposing the posterior bladder wall.

It is commonly associated with epispadias, widened pubic symphysis, and urinary incontinence.

Early surgical reconstruction is required to preserve renal function and continence.

QUESTION

A 10-month-old infant was brought with complaints of jerking movement of limbs towards the body. On examination, there is a regression of developmental milestones. Electroencephalogram shows hypsarrhythmia. Which of the following is the drug of choice in this condition?

A Phenytoin

B Adrenocorticotrophic hormone (ACTH)

CORRECT

C Levetiracetam

D Phenobarbitone

RIGHT ANSWER

OK B. Adrenocorticotrophic hormone (ACTH)

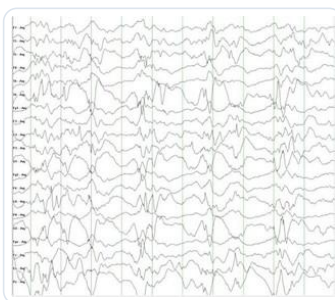
WHY THIS IS RIGHT

Infantile spasms (West syndrome) is an epileptic encephalopathy characterized by the triad of epileptic spasms, developmental regression, and hypsarrhythmia on EEG.

- Age: typically 3-12 months
- Seizures: brief flexor or extensor spasms ("jackknife seizures")
- EEG: chaotic, high-amplitude pattern called hypsarrhythmia

Treatment:

- First-line: ACTH (adrenocorticotrophic hormone)
- Alternative: oral steroids or vigabatrin (especially in tuberous sclerosis)
- Early treatment is crucial to improve neurodevelopmental outcomes
- Delay can lead to long-term cognitive impairment



QUESTION

A 2-year-old child with stable vitals and ECG showing PSVT. What is the appropriate management?

- A** Cardioversion
- B** Adenosine 0.3 mg per kg
- C** Adenosine 0.2 mg/kg
- D** Adenosine 0.1 mg/kg

CORRECT

RIGHT ANSWER

- OK** **D. Adenosine 0.1 mg/kg**

WHY THIS IS RIGHT

Paroxysmal supraventricular tachycardia (PSVT) in a hemodynamically stable child is treated first with rapid-acting AV nodal blocking agents, with adenosine as the drug of choice.

- First-line dose of adenosine: 0.1 mg/kg rapid IV bolus (maximum 6 mg)
- If ineffective, second dose: 0.2 mg/kg (maximum 12 mg)
- Must be given rapidly followed by saline flush due to very short half-life

Clinical approach:

- Ensure child is hemodynamically stable
- Attempt vagal maneuvers first (if feasible)
- Use synchronized cardioversion only if unstable or drug therapy fails

QUESTION

A newborn baby is born with a congenital defect. Identify the defect shown in the image:



A Omphalocele

B Gastroschisis

C Bladder Exstrophy

CORRECT

D Meckel's Diverticulum

RIGHT ANSWER

OK C. Bladder Exstrophy

WHY THIS IS RIGHT

Bladder exstrophy is characterized by absence of the lower anterior abdominal wall, with exposed bladder mucosa. It is associated with epispadias, widened pubic symphysis, and urinary incontinence. This defect differs from omphalocele and gastroschisis, which involve bowel herniation rather than bladder exposure.

QUESTION

2-year-old was brought to the ED following choking on a peanut. The child was unresponsive. What is the next step for managing this patient?

- A** Give 5 back slaps
- B** Give abdominal thrusts
- C** Endotracheal intubation

D Chest compressions

CORRECT

RIGHT ANSWER

OK D. Chest compressions

WHY THIS IS RIGHT

- Unresponsive child with choking (foreign body airway obstruction)
- Treat as cardiac arrest -> start CPR immediately
- Begin with 30 chest compressions
- Then open airway -> look for visible foreign body
- Give 2 rescue breaths
- Repeat cycles
- Hemlich manoeuvre/ Back slaps / abdominal thrusts -> only if conscious child

QUESTION

A 3-year-old child is brought to the emergency department with a generalized seizure following a high-grade fever. Which of the following is the first-line drug of choice for seizure control in this acute febrile episode?

A Diazepam

CORRECT

B Valproate

C Fosphenytoin

D Doxycycline /Amoxicillin

RIGHT ANSWER

OK

A. Diazepam

WHY THIS IS RIGHT

Acute management of febrile seizures focuses on rapid termination of the seizure using benzodiazepines as first-line therapy.

- Diazepam (IV/rectal) or lorazepam (IV) are first-line drugs for ongoing seizures
- They act by enhancing GABA activity -> rapid seizure control
- Most febrile seizures are self-limiting, but if seizure persists >5 minutes, treatment is required

Clinical approach:

- Ensure airway, breathing, circulation first
- Give benzodiazepine if seizure is prolonged
- Long-term antiepileptic therapy is not indicated for simple febrile seizures

Why other options are incorrect:

- Valproate and fosphenytoin are second-line agents if seizures persist
- Antibiotics have no role unless infection is specifically indicated

QUESTION

| The following finding can be seen in:



A Molluscum contagiosum

B Parvovirus B19 infection

CORRECT

C HFMD

D Kawasaki disease

RIGHT ANSWER

OK B. Parvovirus B19 infection

WHY THIS IS RIGHT

Parvovirus B19 infection classically presents with erythema infectiosum showing a "slapped cheek" rash. It may be followed by a lacy rash on the trunk and limbs. The virus can also cause aplastic crisis in hemolytic anemias and hydrops fetalis in pregnancy, adding diagnostic value.

QUESTION

| Causes of non-immune hydrops fetalis are all except:

A ABO incompatibility

CORRECT

B Congestive cardiac failure

C Parvovirus infection

D Chromosomal anomalies

RIGHT ANSWER

OK

A. ABO incompatibility

WHY THIS IS RIGHT

Hydrops fetalis is defined as abnormal accumulation of fluid in two or more fetal compartments and is classified into immune and non-immune causes.

- Immune hydrops is caused by red cell alloimmunization (e.g., Rh incompatibility; ABO incompatibility is also immune-mediated, though usually milder)
- Non-immune hydrops (more common) includes:
 - Cardiac causes (congestive cardiac failure)
 - Infections (e.g., parvovirus B19 causing fetal anemia)
 - Chromosomal anomalies (e.g., Turner syndrome, trisomies)

Why option A is correct:

- ABO incompatibility is an immune cause, not a non-immune cause

QUESTION

A child came to OPD with fever for 6 days with strawberry tongue, conjunctival congestion, and peeling of skin. What should be the ideal treatment?

A IVIG

CORRECT

B Aspirin

C Plenty of fluids

D Cephalosporins

RIGHT ANSWER

OK
A. IVIG

WHY THIS IS RIGHT

Kawasaki disease is an acute vasculitis of childhood characterized by prolonged fever (>5 days), conjunctival congestion, strawberry tongue, mucocutaneous changes, and desquamation.

- Diagnosis is clinical: fever ≥ 5 days + mucocutaneous features
- Risk of coronary artery aneurysms is the most serious complication

Management:

- First-line treatment is intravenous immunoglobulin (IVIG) given early (within 10 days of illness) as it rapidly decreases inflammation and reduces risk of coronary artery aneurysms.
- High-dose aspirin is also used as adjunct therapy

Why others are incorrect:

- Aspirin alone is insufficient
- Fluids are supportive but not definitive
- Antibiotics (cephalosporins) have no role

QUESTION

Cannabinoids are approved in all of the following except:

- A** Dravet Syndrome
- B** Lennox-Gastaut Syndrome
- C** Tuberous Sclerosis

D Rett Syndrome

CORRECT

RIGHT ANSWER

OK **D. Rett Syndrome**

WHY THIS IS RIGHT

Cannabinoids (particularly cannabidiol) are approved for specific treatment-resistant epileptic syndromes in children, where they help reduce seizure frequency. Cannabidiol acts by modulating neuronal excitability and reducing seizure activity. Used as adjunct therapy in refractory epilepsy

Approved indications:

- Dravet syndrome
- Lennox-Gastaut syndrome
- Tuberous sclerosis complex-associated epilepsy

Why Rett syndrome is incorrect:

- Rett syndrome is a neurodevelopmental disorder with seizures, but cannabinoids are not an approved standard indication

QUESTION

| Which of the following statements is not true about febrile seizure?

- A Age 6 months - 5 years
- B Simple febrile seizure lasts for <15 minutes
- C No need for long term antiepileptics

D 54% recurrence

CORRECT

RIGHT ANSWER

OK D. 54% recurrence

WHY THIS IS RIGHT

Febrile seizures are benign, age-dependent seizures occurring in children between 6 months and 5 years in association with fever, without evidence of intracranial infection or metabolic cause.

- Simple febrile seizures are generalized, last less than 15 minutes, and do not recur within 24 hours
- Long-term antiepileptic therapy is not indicated due to the benign nature and low risk of epilepsy
- Recurrence risk is about 30-35%, not as high as 54%

QUESTION

All of the following are incorrect about resuscitation of a critically sick child, except?

- A** Preferred route of adrenaline intraosseous
- B** Compression to ventilation ratio should be 30:2 with 2 rescuers
- C** Chest compression depth should be one-third of A-P diameter of chest
- D** Injection Adenosine 0.01 ml/kg IV or IO (1:10,000)

CORRECT

RIGHT ANSWER

- OK** C. Chest compression depth should be one-third of A-P diameter of chest

WHY THIS IS RIGHT

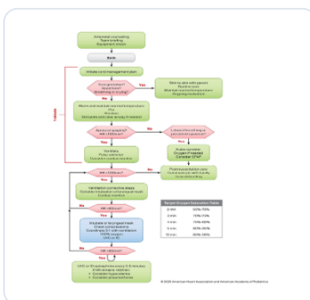
In pediatric resuscitation, effective chest compressions are critical and should be delivered to a depth of one-third of the anteroposterior diameter of the chest to ensure adequate cardiac output.

- Compression depth: one-third of chest A-P diameter (correct statement)
- Compression-ventilation ratio:
- Single rescuer: 30:2
- Two rescuers: 15:2 (not 30:2)
- Preferred route for emergency drugs is IV; intraosseous is used when IV access is not available (not preferred primarily)
- Adrenaline dose: 0.01 mg/kg (0.1 ml/kg of 1:10,000), not adenosine

Why other options are incorrect:

- IO is an alternative route, not the preferred first line if IV is available
- 30:2 ratio is for single rescuer, not two rescuers
- Adenosine dose and indication are incorrect in this context (used for SVT, not cardiac arrest)

NRP 2025:



QUESTION

A 2 yr old child presented with fever, thrombocytopenia, hematuria. The mother tells you that the baby also suffered from diarrhea a few days back. Which is the most common organism for likely cause?

A E Coli 0157:H7

CORRECT

B Salmonella

C Shigella

D Yersinia

RIGHT ANSWER

OK

A. E Coli 0157:H7

WHY THIS IS RIGHT

Hemolytic uremic syndrome (HUS) is a common cause of acute kidney injury in young children, classically occurring after a diarrheal illness caused by Shiga toxin-producing *Escherichia coli*, most commonly serotype O157:H7.

- Triad: microangiopathic hemolytic anemia, thrombocytopenia, and acute kidney injury (hematuria)
- Typically follows bloody diarrhea ("D+ HUS")
- Shiga toxin damages endothelial cells -> platelet activation and microthrombi formation
- Management is mainly supportive (fluids, dialysis if needed); antibiotics are generally avoided

Why other options are less likely:

- Shigella can also cause HUS but is less common
- Salmonella and Yersinia are not typical causes of HUS

QUESTION

A child is NS1 antigen positive and presents with fever and warning signs. Lab findings show hematocrit of 41% and platelet count of 50,000 per microliter. What is the most appropriate next step in the management?

- A Platelet transfusion.
- B Start IV fluids with crystalloids at 7 ml/kg/hour.**
- C Start IV fluids with crystalloids at 10-20 ml/kg/hour.
- D Start IV fluids with colloids at 7 ml/kg/hour.

CORRECT

RIGHT ANSWER

- B. Start IV fluids with crystalloids at 7 ml/kg/hour.**

WHY THIS IS RIGHT

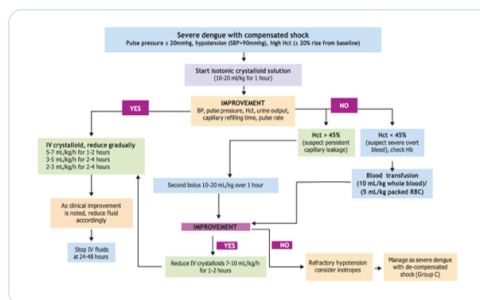
Dengue with warning signs requires careful fluid management using isotonic crystalloids to prevent progression to shock while avoiding fluid overload.

- Warning signs include abdominal pain, persistent vomiting, rising hematocrit, and thrombocytopenia
- Hematocrit of 41% with low platelets suggests plasma leakage phase
- First-line management is controlled IV fluid therapy with crystalloids (normal saline or Ringer lactate) at 5-7 ml/kg/hour. It helps maintain intravascular volume without causing fluid overload

Why others are incorrect:

- Platelet transfusion is not indicated unless there is significant bleeding
- 10-20 ml/kg/hour is used in dengue shock (more severe cases)
- Colloids are reserved for patients not responding to crystalloids or with severe shock

Screenshots from national Dengue management guidelines:



QUESTION

A 9 months old infant is brought for a routine check-up. The infant is able to sit without support and transfer objects. Which of the following developmental milestones is most likely to be present at this age?

A Mature pincer grasp

B Creeps well on hands and knees

CORRECT

C Can hop on one foot

D Runs steadily

RIGHT ANSWER

OK

B. Creeps well on hands and knees

WHY THIS IS RIGHT

At 9 months of age, infants typically achieve gross motor milestones such as sitting without support and beginning to creep or crawl on hands and knees. Fine motor development at this stage includes transferring objects from one hand to another and developing an immature pincer grasp.

- Gross motor: sits without support, creeps/crawls
- Fine motor: hand-to-hand transfer of objects, immature pincer grasp
- Advanced milestones like mature pincer grasp (12 months), running (18-24 months), and hopping (4 years) occur later

QUESTION

A child who can make a cross, tell stories make tower of 6 cubes and goes to toilet alone, what is the age of the child:

A 1 year

B 2 years

C 3 years

D 4 years

CORRECT

RIGHT ANSWER

OK

D. 4 years

WHY THIS IS RIGHT

A 4-year-old child demonstrates advanced fine motor, language, and social independence milestones.

- Fine motor: able to copy a cross and build a tower of about 6 cubes
- Language: can tell simple stories and use well-formed sentences
- Social/adaptive: goes to the toilet independently

Age correlation:

- 1 year: basic motor skills like standing, few words
- 2 years: 2-word phrases, stacks 4-6 cubes
- 3 years: copies a circle, partial toilet training
- 4 years: copies a cross, storytelling, complete toilet training

QUESTION

A 9-month-old infant is able to roll over, babble ‘mama’ and ‘dada,’ and exhibits stranger anxiety but is unable to sit without support. Which of the following is the most appropriate next step in management?

A Reassurance

CORRECT

B Get development test

C Give training to sit

D Get an MRI

RIGHT ANSWER

OK

A. Reassurance

WHY THIS IS RIGHT

Developmental milestones can show normal variation, and a single mildly delayed milestone in the presence of otherwise appropriate development does not necessarily indicate pathology.

- At 9 months, expected milestones include babbling (“mama/dada”), stranger anxiety, and rolling over, all of which are present in this child
- Sitting without support typically develops around 8-9 months, but slight delay can still fall within normal variation
- Absence of red flag signs (loss of milestones, global delay, abnormal tone) supports reassurance

Clinical approach:

- If development is normal in other domains (social, language, fine motor), isolated borderline delay can be observed
- Regular follow-up is important to ensure progression

QUESTION

Which of these milestones is correctly matched with the corresponding age?

- A Stacking two cubes in a tower 2 years
- B Mature pincer grasp at 9 months
- C Transferring the object from one hand to another hand by 6 months**
- D Reaching out to the object with one hand by the age of 8 months

CORRECT

RIGHT ANSWER

- OK C. Transferring the object from one hand to another hand by 6 months**

WHY THIS IS RIGHT

Object transfer from one hand to another is a key fine motor developmental milestone achieved at ~6 months of age.
• It reflects improving hand coordination and bilateral integration.

Why other options are incorrect:

- A. Stacking two cubes -> occurs at ~15 months, not 2 years
- B. Mature pincer grasp -> develops at ~12 months, not 9 months
- D. Reaching with one hand -> seen earlier (~5 months), not 8 months

Age	Social Milestone
2m	Social smile
3m	Recognizes mother
4m	Stranger anxiety, inhibits to no
6m	Waves bye-bye, repeats activity when appreciated
9m	Comes when called, plays simple ball game
12m	Jargon, points to objects of interest
18m	Copies parents in task
2y	Asks for food, drink, toilet
3y	Shares toys, knows full name age gender
4y	Plays cooperatively in group, goes to toilet alone
5y	Helps in household tasks

Age	Language milestone
1m	Alerts to sound
3m	Coos
4m	Laugh loud
6m	Monosyllables
9m	Bisyllables
12m	1-2 words with meaning
18m	8-10-word vocabulary
2y	2-3 word sentences, uses pronouns
3y	Asks question
4y	Sings song, tell stories
5y	Asks meaning of words

Age	Gross motor Milestone
3m	Neck holding
4m	Rolls over
6m	Sits in tripod position
9m	Sits without support, crawling
10m	Stand with support, creeps
12m	Stands without support
15m	Walks alone
18m	Runs
2y	Walks up and downstairs, 2 feet step
3y	Rides tricycle, alternate feet going upstairs
4y	Hops on one foot, alternate feet going downstairs

Age	Fine motor milestone
4m	Bidextrous approach
6m	Unidextrous approach
9m	Immature pincer grasp
12m	Mature pincer grasp
15m	Imitates scribbling, tower of 2 blocks, drinks from cup
18m	Scribbles, tower of 3 blocks
2y	Tower of 6 blocks, vertical and circular strokes, undresses, feeds with spoon
3y	Tower of 9 blocks, copies circle, dresses
4y	Copies cross, bridge with blocks
5y	Copies triangle, gate with blocks