Solved Paper

Neurology

1. Cerebral palsy

Subject: Neurology

This is a cornerstone case in the MD Pediatrics practical exam. You aren't just diagnosing "Cerebral Palsy" (CP); you are defining a **functional status, a topographical distribution, and a physiological type**, while identifying the **timing of the insult** and **associated comorbidities**.

HISTORY

Chief Complaint

- "Delay in attaining motor milestones" (most common)
- "Stiffness of limbs" or "abnormal posturing"
- "Involuntary movements"
- "Seizures or difficulty in feeding"
- Duration: Since birth or early infancy.

History of Present Illness

Ask these questions naturally to build the "Non-progressive" nature of CP:

- **Motor Milestones:** "When did he first hold his head steady? Did he ever lose a skill he had already gained?" (If yes, think neurodegenerative, not CP).
- **Hand Preference:** "Does he use one hand more than the other?" (Early handedness before 12-18 months suggests hemiplegic CP).
- **Tone/Posturing:** "Do you find it difficult to put on his diaper because his legs cross like scissors?" or "Does he feel 'floppy' like a rag doll?"
- **Feeding:** "Does he choke, cough, or have milk come out of his nose while feeding?" (Bulbar/pseudobulbar involvement).
- **Seizures:** "Has he had any fits? Describe them." (Common in post-asphyxial or malformation-related CP).
- **Vision/Hearing:** "Does he track toys? Does he turn to your voice?"

Relevant Background History

- **Antenatal:** Maternal infections (TORCH), PIH, or antepartum hemorrhage.
- **Birth History [CRITICAL]:** "Did he cry immediately? Was he in the NICU? Did he have jaundice requiring exchange transfusion (Kernicterus/Dyskinetic CP)?"
- **Developmental:** Detailed 4-domain history. Look for **"Dissociation"** (Motor significantly delayed, Social/Language relatively preserved).

- **Nutritional:** Caloric intake (often deficient due to feeding oromotor dysfunction).
- **Socioeconomic:** Can the family afford long-term physiotherapy and orthotics?

EXAMINATION

General Survey

- **Observation:** Observe the child on the mother's lap first. Is there **scissoring** of legs? Is the child **obligatorily** looking to one side (ATNR)?
- **Activity:** Is there spontaneous movement? In hemiplegia, one side is "quiet."
- **Facies:** Look for "Open mouth" (drooling), "High arched palate," or "Dental enamel hypoplasia" (common in athetoid CP).
- **Microcephaly:** Measure Head Circumference. It's the single best surrogate for brain growth.

Vital Signs and Anthropometry

- **Weight/Height:** Plot on WHO/IAP charts. Use **Upper Arm Length** or **Tibia Length** if contractures prevent standing height.
- **Clinical Significance:** Failure to thrive is a marker of the severity of oromotor dysfunction.

Peripheral Signs

- **Spine:** Look for scoliosis (common in non-ambulatory CP).
- **Skin:** Neurocutaneous markers (Sturge-Weber can present as hemiplegic CP).

Systemic Examination — Primary System (Neurology)

1. Tone Assessment [THE CORE OF THE CASE]

- **Passive Tone:**
    - **Upper Limb:** Flexion/extension at elbow, wrist rotation.
    - **Lower Limb:**
        - **Adductor Angle:** With child supine, abduct hips. Wide angle = Hypotonia; Narrow (<70°) = Spasticity.
        - **Popliteal Angle:** Flex hip to 90°, then extend knee. [SEVERITY MARKER] >50° suggests hamstring spasticity.
        - **Heel-to-Ear Maneuver:** In infants, to check for truncal hypotonia.
- **Dynamic Tone:**
    - **Scarf Sign:** Pull arm across chest.
    - **Ankle Clonus:** Rapidly dorsiflex the foot. >5 beats is pathological.

2. Power and Bulk

- In CP, "Power" is hard to assess in young children. Look for **Functional Power**: Can they reach for a toy? Can they pull to stand?
- **Bulk:** Disuse atrophy is common in spastic limbs.

3. Reflexes

- **DTRs:** Brisk (3+) or exaggerated (4+) with radiation.
- **Plantars:** Extensor (Babinski positive).
- **Primitive Reflexes [EXAMINER FAVORITE]:** Check if **Moro, ATNR, or STNR** are persistent beyond 6 months. Persistence prevents the development of voluntary motor skills.

4. Postural Reactions (The "Righting" Reactions)

- **Parachute Reflex:** Hold child mid-air and tilt forward. They should extend arms. Absence after 9 months is a major red flag for CP.

5. Gait (If Ambulatory)

- **Crouch Gait:** Flexion at hips/knees (Spastic diplegia).
- **Scissoring Gait:** Adductor spasticity.
- **Hemiplegic Gait:** Circumduction of one leg, "fencing" posture of the arm.

Systemic Examination — Secondary Systems

- **Vision:** Check for Strabismus (very common).
- **Hearing:** Distraction test at bedside.
- **Oromotor:** Check for drooling, tongue thrust, and gag reflex.

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **Persistence of ATNR:** If you turn the head and the "fencing" posture is obligatory and cannot be broken, it's a definitive sign of cortical pathology.
- **The "Commando Crawl":** Child pulls with arms but drags legs (Classic for Spastic Diplegia).
- **Early Handedness:** A child using one hand exclusively before age 1 is almost always Hemiplegic CP.
- **Gower's Sign (Pseudo):** Children with CP may use hands to get up, but due to spasticity/balance, not proximal weakness (Differentiate from DMD).

Severity Assessment

- **GMFCS (Gross Motor Function Classification System):** You **must** stage the child from Level I (walks without limitations) to Level V (transported in a manual wheelchair).

DIAGNOSIS

Diagnostic Criteria

- **Definition:** A group of **permanent** disorders of the development of movement and posture, causing activity limitation, attributed to **non-progressive** disturbances that occurred in the **developing fetal or infant brain**.

Differentials

1. **Neurodegenerative Disorders:** (Metabolic/Genetic) - History of **regression** of milestones.
2. **Slowly Progressive Spinal Cord Tumor:** Look for a sensory level and bladder/bowel involvement.
3. **Hereditary Spastic Paraplegia:** Strong family history, purely motor, slowly progressive.

Investigations

- **Tier 1:** Vision and Hearing screening (mandatory for all).
- **Tier 2: MRI Brain** (Gold Standard). Look for Periventricular Leukomalacia (PVL) in diplegia or Multicystic Encephalomalacia in quadriplegia.
- **Tier 3:** EEG (if seizures), Metabolic screen (if no clear perinatal insult).

Management Outline

- **Multidisciplinary Team:** Pediatrician, Physiatrist, PT/OT, Speech therapist, Orthopedic surgeon.
- **Medical:** Baclofen or Diazepam for spasticity; Glycopyrrolate for drooling.
- **Focal Spasticity:** Botulinum Toxin (Botox) injections.
- **Surgical:** Selective Dorsal Rhizotomy (SDR) or tendon lengthening.

EXAMINER'S VIVA

1. **Q: Why is CP called a "Static Encephalopathy" if the clinical picture changes?**
    - *A:* The *insult* to the brain is static and non-progressive, but the *clinical expression* changes as the child grows and the nervous system matures.
2. **Q: What is the most common MRI finding in a preterm child with Spastic Diplegia?**
    - *A:* Periventricular Leukomalacia (PVL).
3. **Q: How do you differentiate Spasticity from Rigidity at the bedside?**
    - *A:* Spasticity is "Clasp-knife" (velocity-dependent); Rigidity is "Lead-pipe" or "Cogwheel" (not velocity-dependent).
4. **Q: What is the "Hand-Regard" sign?**

- *A:* Normally disappears by 4-5 months. Persistence suggests developmental delay or visual impairment.

5. **Q: Technique: How do you elicit the ATNR?**
    - *A:* Child supine, turn head to one side for 10 seconds. Positive if ipsilateral limbs extend and contralateral limbs flex. It is "obligatory" if the child cannot break the posture.

LONG CASE PRESENTATION TIPS

- **The Opening:** "Master X, a 3-year-old male, born at 32 weeks gestation with a history of NICU stay, presents with a non-progressive delay in motor milestones and increased stiffness of all four limbs, suggestive of Spastic Diplegic Cerebral Palsy, GMFCS Level III."
- **Mistake to Avoid:** Don't just say "Developmental Delay." Use the term **"Global Developmental Delay"** only if ≥2 domains are affected.
- **Watch for:** Examiners will watch how you handle the child. Always warm your hands and perform the "least intrusive" parts of the exam (observation, head circumference) before "intrusive" parts (checking tone or reflexes).

---

2. Global developmental delay

Subject: Neurology

This is a cornerstone of pediatric neurology. In a Global Developmental Delay (GDD) case, the examiner isn't just looking for a list of milestones; they are looking for your ability to **localize the insult** (pre, peri, or postnatal) and **determine the trajectory** (static vs. progressive).

HISTORY

Chief Complaint

- "My child is not doing what other children his age are doing" (delayed milestones).
- "He is not talking yet" or "He is not walking yet."
- Associated complaints: Seizures, abnormal movements, or behavioral issues.

History of Present Illness

Phasing is key here. Don't just list milestones; tell a story of the child's development.

- **The "When" and "How":** "When did you first notice he was different from his siblings/peers?" "Was he always slow from birth, or did he gain skills and then lose them?" (Crucial for Static vs. Neurodegenerative).
- **Domain-Specific Characterization:**
    - **Gross Motor:** "When did he get neck control? Does he drag one side of his body while crawling?" (Points to CP/Hemiplegia).

    - **Fine Motor/Adaptive:** "Can he transfer an object from one hand to another? Can he feed himself with a spoon?"
    - **Language (often the most delayed):** "Does he turn to his name? Does he use gestures (pointing) to show you what he wants?" (Lack of pointing is a red flag for Autism).
    - **Social/Cognitive:** "Does he make eye contact? Does he play with other children or prefer to be alone?"
- **The "Why" (Etiology hunting):**
    - "Were there any flickers or stiffening episodes?" (Seizures/Infantile spasms).
    - "Does he have a strange odor to his urine?" (Inborn Errors of Metabolism - IEM).
    - "How is his vision and hearing?" (Sensory impairment can mimic GDD).

Relevant Background History

- **Antenatal:** Ask about maternal infections (TORCH), radiation, or drug intake. "Did you feel the baby move normally during pregnancy?" (Decreased movements suggest early fetal insult or neuromuscular disorder).
- **Birth History [CRITICAL]:** "Did the baby cry immediately? Did he need oxygen or a NICU stay? Was there jaundice requiring exchange transfusion?" (Kernicterus).
- **Developmental:** You must calculate the **Developmental Quotient (DQ)** for each domain:
- **Family History:** Consanguinity (AR disorders/IEMs). "Are there any other children in the family with similar slowness or early deaths?"
- **Socioeconomic:** "What toys does he have? Who spends time talking to him?" (Environmental deprivation).

EXAMINATION

General Survey

- **Observation:** Observe the child on the floor/mother's lap before touching them. Is there "scissoring" of legs? Is the child "floppy" like a ragdoll? Is there "hand-wringing" (Rett Syndrome)?
- **Facies [EXAMINER FAVORITE]:** Look for dysmorphism.
    - *Down Syndrome:* Up-slanting palpebral fissures, flat nasal bridge, Brushfield spots.
    - *Fragile X:* Long face, prominent ears.
    - *Williams:* Elfin facies.
    - *Fetal Alcohol:* Smooth philtrum, thin upper lip.
- **Neurocutaneous Markers:** Examine the skin in bright light for Ash-leaf spots (Tuberous Sclerosis) or Café-au-lait spots (Neurofibromatosis).

Vital Signs and Anthropometry

- **Head Circumference (OFC):** [CRITICAL] Measure the maximum circumference from the occipital protuberance to the supraorbital ridges.
    - *Microcephaly:* Suggests intrauterine insult or genetic syndrome.
    - *Macrocephaly:* Suggests hydrocephalus, storage disorders, or Canavan disease.
- **Height/Weight:** Plot on WHO/IAP charts. Stunting may suggest a chronic systemic or endocrine cause (Hypothyroidism).

Peripheral Signs

- **Eyes:** Check for cataracts (Galactosemia/TORCH), cherry-red spot on fundoscopy (Tay-Sachs), or optic atrophy.
- **Ears:** Assess startle response to sound.
- **Spine:** Look for tufts of hair, dimples, or lipomas (Spinal dysraphism).

Systemic Examination — Primary System (Neurology)

- **Tone:**
    - *Technique:* Move joints (elbow, knee, wrist) through passive range of motion at varying speeds.
    - *Significance:* Spasticity (clasp-knife) suggests UMN/Pyramidal lesion. Hypotonia (ragdoll) suggests LMN, cerebellar, or early "evolving" CP.
- **Power:** In a non-cooperative child, observe Gower's sign (climbing up oneself) or how they reach for a toy.
- **Reflexes:**
    - *Technique:* Use a reflex hammer, not your fingers.
    - *Significance:* Brisk reflexes + extensor plantar = UMN lesion (CP). Absent reflexes = LMN (SMA).
- **Primitive Reflexes:** In an older infant, check if the Moro, ATNR, or Palmar grasp have persisted. [SEVERITY MARKER] Persistence of primitive reflexes beyond 6 months is a strong predictor of CP.
- **Cerebellar Signs:** Watch the child reach for a biscuit (intention tremor) or their gait (ataxia).

Systemic Examination — Secondary Systems

- **Abdomen:** Organomegaly (Hepatosplenomegaly) points toward Lysosomal Storage Disorders (LSDs) or Mucopolysaccharidosis (MPS).
- **CVS:** Murmurs (Williams syndrome or TORCH).

SIGNS THAT CLINCH THE DIAGNOSIS [EXAMINER FAVORITES]

- **The "Scissoring" Posture:** When picked up by the axillae, the child crosses their legs. Indicates spastic diplegic CP.
- **Vertical Suspension Test:** If the child slips through your hands, it indicates shoulder girdle hypotonia.
- **Hand-Regard:** Persistence of looking at one's own hands beyond 20 weeks is a sign of cognitive delay.
- **The "Social Smile":** If absent by 2 months, it's the earliest indicator of GDD or visual impairment.

DIAGNOSIS

Diagnostic Criteria

- **GDD Definition:** Significant delay (>2 SD below the mean) in **two or more** developmental domains (Gross/fine motor, speech/language, cognition, social/personal, and activities of daily living). Usually reserved for children < 5 years.

Differentials

1. **Cerebral Palsy:** Non-progressive motor delay with history of birth asphyxia.
2. **Global Developmental Delay (Syndromic):** Associated with dysmorphism (e.g., Down Syndrome).
3. **Neurodegenerative/Metabolic:** Loss of milestones (regression) + organomegaly.
4. **Environmental Deprivation:** Poor stimulation but normal head circumference and no focal neuro signs.

Investigations

- **Tier 1:** Thyroid profile (TSH/T4), BERA (Hearing), Vision assessment, Metabolic screen (TMS/GCMS).
- **Tier 2:** Neuroimaging (MRI Brain is preferred over CT to see myelination patterns).
- **Tier 3:** Genetic testing (Karyotyping, Chromosomal Microarray, or Exome Sequencing).

MANAGEMENT OUTLINE

- **Multidisciplinary Approach:** The "Medical Home" concept.
- **Early Intervention:** Physiotherapy (Neurodevelopmental Therapy - NDT), Occupational therapy, and Speech therapy.
- **Pharmacotherapy:** For comorbidities (e.g., Levetiracetam for seizures, Baclofen for spasticity).
- **Counseling:** Recurrence risk for parents (Genetic counseling).

EXAMINER'S VIVA

1. **Q: What is the difference between GDD and Intellectual Disability (ID)?**
    - *A:* GDD is used for children < 5 years when clinical severity cannot be reliably assessed. ID is used for children > 5 years when IQ testing is more reliable.
2. **Q: How do you differentiate between a "Static" and "Progressive" insult?**
    - *A:* By the "Trajectory of Milestones." If the child is gaining skills (even slowly), it's likely static. If the child is losing previously attained skills (regression), it's progressive/degenerative.
3. **Q: What is the significance of the "Handedness" before 1 year of age?**
    - *A:* Early handedness (e.g., using only the right hand at 6 months) is pathological and suggests a motor deficit (Hemiplegia) in the contralateral limb.
4. **Q: Technique: How do you elicit the Landau Reflex?**
    - *A:* Suspend the child prone in mid-air. The child should extend the head and legs (the "superman" pose). If you flex the head, the legs should flex. Absence suggests motor delay.

LONG CASE PRESENTATION TIPS

- **The Opening:** "I am presenting [Name], a [Age] old boy, born of a [consanguineous/non-consanguineous] marriage, with a history of delay in [list domains] since early infancy, with a static/progressive course, currently functioning at a developmental age of [X] months."
- **Mistake to Avoid:** Don't just say "milestones are delayed." You must specify the **Developmental Age** for each of the four domains separately.
- **Watch for:** Examiners love to see if you checked the **Head Circumference** correctly. Use a non-stretchable tape and measure three times, taking the largest value.

---

3. Seizure disorder

Subject: Neurology

This is a high-stakes case. In a Neurology long case, the diagnosis is often 80% history. The examiner isn't just looking for "seizures"; they want you to classify the seizure type, the epilepsy syndrome, and the etiology.

HISTORY

Chief Complaint

- "Episodes of [describe movement/behavior]" rather than using the word "seizure" immediately.

- Duration of the disorder (months/years) and the time since the last event.

History of Present Illness

You must reconstruct the event chronologically. Ask the parent:

- **The Lead-up (Pre-ictal):** "Was the child acting differently before it started? Did they complain of a funny smell, a stomach sensation, or fear?" (Aura suggests focal onset). "What was the child doing exactly when it started?" (Triggers: flickering lights, sleep deprivation, fever).
- **The Event (Ictal):** "Did it start in one limb or the face and then spread, or did it involve the whole body from the start?" (Focal vs. Generalized). "Were the eyes open? Which way were they looking?" (Version). "Was there stiffening (tonic), jerking (clonic), or limpness (atonic)?" "Did the child lose consciousness—could they respond if you called their name?"
- **The Finish (Post-ictal):** "How long did the jerking last?" (Use a watch/timer analogy). "Was the child sleepy, confused, or aggressive afterward? Did they have weakness in one arm or leg?" (Todd's Palsy—confirms focal onset).
- **Inter-ictal state:** "Between episodes, is the child's behavior, school performance, or motor skills normal?" (Assessing for epileptic encephalopathy or underlying neurodegenerative process).

Symptom Clusters:

- **Absence:** Brief staring spells, no post-ictal confusion, provoked by hyperventilation.
- **Infantile Spasms:** Sudden clusters of "jack-knife" flexions in an infant, often on awakening.
- **GTCS:** Sudden loss of consciousness, tonic-clonic movements, tongue biting, incontinence.

Relevant Background History

- **Antenatal/Birth:** "Was there any bleeding or infection during pregnancy? Was the baby born at term? Did the baby cry immediately or need the NICU/oxygen?" (HIE is a leading cause of symptomatic epilepsy).
- **Developmental:** "Did the child reach milestones on time? Has there been any loss of previously gained skills?" (Regression suggests a neurodegenerative or metabolic cause).
- **Family History:** "Does anyone else have seizures? Any history of sudden unexplained deaths or consanguinity?"
- **Past History:** History of meningitis, encephalitis, or head trauma.

EXAMINATION

General Survey

- **Observation:** Watch the child's spontaneous activity. Is there a hemiparetic gait? Is the child hyperactive (common in certain syndromes or as a side effect of Phenobarbitone/Levetiracetam)?
- **Neurocutaneous Markers [CRITICAL]:** Use a torch or Wood's lamp. Look for:

    - *Ash-leaf spots/Shagreen patch:* Tuberous Sclerosis.
    - *Café-au-lait spots:* NF1.
    - *Port-wine stain (Trigeminal distribution):* Sturge-Weber Syndrome.
- **Dysmorphism:** Look for features of chromosomal anomalies (e.g., Angelman or Down syndrome).

Vital Signs and Anthropometry

- **Head Circumference:** Measure precisely. Microcephaly (congenital infections/HIE) or Macrocephaly (Leukodystrophies/Hydrocephalus).
- **Blood Pressure:** Essential to rule out hypertensive encephalopathy as a cause of seizures.

Peripheral Signs

- **Hands:** Look for "fanning" or tremors (side effects of Sodium Valproate). Check for single palmar crease.
- **Eyes:** Fundoscopy is mandatory. Look for chorioretinitis (TORCH), optic atrophy, or papilledema (raised ICP).
- **Gums:** Gingival hyperplasia (Phenytoin side effect).

Systemic Examination — Primary System (Neurology)

- **Higher Mental Functions:** Assess age-appropriate cognition. In older children, check orientation and simple memory.
- **Cranial Nerves:** Focus on extraocular movements (nystagmus) and facial symmetry.
- **Motor System:**
    - **Bulk/Tone:** Look for focal wasting or spasticity.
    - **Power:** Grade 0-5. Subtle hemiparesis may be the only clue to a focal structural lesion.
    - **Reflexes:** Asymmetry in DTRs or an upgoing plantar (Babinski) suggests a contralateral cortical lesion.
- **Cerebellar Signs:** Ataxia or dysmetria (could be drug toxicity, e.g., Phenytoin, or a posterior fossa lesion).
- **Meningeal Signs:** If the history is acute, check for Kernig's/Brudzinski's.

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **Hyperventilation Provocation:** Ask the child to "blow out a candle" repeatedly for 3 minutes. If they develop a staring spell/absence seizure, it's diagnostic of Childhood Absence Epilepsy.
- **Todd's Paralysis:** Transient focal weakness following a seizure; proves the seizure had a focal cortical onset even if the parent described it as generalized.
- **The "Fencer's Posture":** If described or seen, indicates a seizure arising from the Supplementary Motor Area (Focal).

- **Phakomatoses:** Finding an ash-leaf spot in a child with infantile spasms clinches Tuberous Sclerosis.

Severity Assessment

- **Frequency:** Daily vs. monthly.
- **Status Epilepticus:** Any seizure >5 mins or serial seizures without recovery of consciousness. [SEVERITY MARKER]
- **Developmental Regression:** Indicates an epileptic encephalopathy (e.g., West Syndrome or Lennox-Gastaut). [SEVERITY MARKER]

DIAGNOSIS

Diagnostic Criteria

- **ILAE 2014 Definition:**
    1. At least two unprovoked seizures occurring >24h apart.
    2. One unprovoked seizure and a probability of further seizures (at least 60%) over the next 10 years (e.g., structural lesion on MRI + one seizure).
    3. Diagnosis of an epilepsy syndrome.

Differentials

1. **Breath-holding spells:** Occur only when crying/angry; child turns blue/pale then limp.
2. **Syncope:** Occurs on standing; preceded by lightheadedness/tunnel vision; rapid recovery.
3. **Psychogenic Non-Epileptic Seizures (PNES):** Long duration, pelvic thrusting, side-to-side head shaking, eyes tightly closed, no post-ictal phase.
4. **Night Terrors:** Occur in NREM sleep; child is inconsolable but has no memory of it.

Investigations

- **Tier 1:** Blood glucose, Serum Calcium/Magnesium (especially in infants), Electrolytes.
- **Tier 2: EEG** (Standard, sleep-deprived, or video-EEG). *Note: A normal EEG does not rule out epilepsy.* **Neuroimaging (MRI Brain):** Preferred over CT to look for cortical dysplasia, mesial temporal sclerosis, or small tumors.
- **Tier 3:** Metabolic screen (TMS/GCMS) if regression is present; Genetic testing (Epilepsy panel).

Management Outline

- **Acute:** ABCs, Midazolam (0.2mg/kg intranasal/buccal) if seizure >5 mins.
- **Maintenance:** Start a single Anti-Seizure Medication (ASM) based on seizure type.
    - *Focal:* Oxcarbazepine, Levetiracetam.
    - *Generalized:* Sodium Valproate (avoid in adolescent girls), Levetiracetam.

    - *Absence:* Ethosuximide or Valproate.
- **Non-Pharmacological:** Ketogenic diet (for refractory cases), Vagal Nerve Stimulation (VNS), or Epilepsy surgery.

EXAMINER'S VIVA

- **Q: How do you distinguish a seizure from a jittery movement in a neonate?**
    - A: Jitteriness is stimulus-evoked, stops with passive flexion/holding the limb, and lacks abnormal eye movements. Seizures do not stop with holding.
- **Q: What is the "Rule of Halves" in epilepsy?**
    - A: 50% of patients respond to the first drug, 15% to the second, and only ~1-5% to the third.
- **Q: When can you consider stopping ASMs?**
    - A: Usually after a 2-year seizure-free interval, provided the EEG is normal and there is no structural lesion.
- **Q: Show me how to perform the Schamroth window test.**
    - A: (Resident demonstrates placing dorsal surfaces of terminal phalanges together). *Why?* To check for clubbing, which might suggest cyanotic heart disease (source of brain abscess/embolic stroke leading to seizures).

LONG CASE PRESENTATION TIPS

- **The Opening:** "I am presenting the case of [Name], a [Age] year old male, born of [Consanguineous/Non-consanguineous] marriage, with normal/delayed development, who presents with recurrent episodes of [Type] movements for [Duration]..."
- **Common Mistake:** Forgetting to ask about the **first-ever** seizure. Often parents forget a febrile seizure that happened years ago, which is vital for the history.
- **The "Negative" History:** Always explicitly state the absence of fever, head injury, and poisoning to rule out "provoked" seizures.

---

4. Duchenne muscular dystrophy

Subject: Neurology

This is a classic "bread and butter" neurology long case. In Duchenne Muscular Dystrophy (DMD), the diagnosis is often made the moment the child walks into the room. Your job as a postgraduate is to demonstrate that you can systematically prove it is a primary muscle disease, assess the functional stage, and screen for life-threatening complications.

HISTORY

Chief Complaint

- "Difficulty in getting up from the floor" (onset usually 3–5 years).
- "Frequent falls" or "clumsiness."
- "Walking on toes" or "waddling like a duck."
- "Difficulty climbing stairs" (needs to hold the railing).

History of Present Illness

Ask these exact questions to map the progression:

- **Onset:** "When did you first notice he was different from other children?" (Look for a delay in walking—often >15 months).
- **Proximal Weakness (Lower Limbs):** "Does he use his hands to push off his thighs to stand up?" (Gowers' maneuver). "Does he struggle to get into a bus or climb stairs?"
- **Proximal Weakness (Upper Limbs):** "Can he comb his hair or reach for a toy on a high shelf?" (Usually occurs years after lower limb involvement).
- **Distal Function:** "Can he still hold a spoon or button his shirt?" (Preserved until late stages in DMD).
- **Progression:** "Is it getting worse?" (DMD is relentlessly progressive; a static course suggests a different myopathy).
- **Bulbar/Respiratory:** "Does he have difficulty swallowing or a weak cough?" "Does he wake up with headaches?" (Suggests nocturnal hypoventilation).

Differentiating Clusters:

- **DMD vs. BMD:** If the child is >12 years and still walking, think Becker Muscular Dystrophy (BMD).
- **DMD vs. SMA:** Ask about fasciculations (tongue/hands). SMA has early loss of deep tendon reflexes and no pseudohypertrophy.

Relevant Background History

- **Developmental:** Specifically ask about speech and language. 30% of DMD boys have cognitive impairment or global developmental delay due to lack of Dp140/Dp71 dystrophin isoforms in the brain.
- **Family History:** Construct a three-generation pedigree. Ask about maternal uncles or brothers with similar weakness or early death. (X-linked recessive).
- **Birth History:** Usually normal. High CK levels can sometimes cause neonatal jaundice, but this is rare.

EXAMINATION

General Survey

- **Observation:** Watch the child enter the room. Note the **Trendelenburg gait** (waddling) and **lumbar lordosis**.
- **Posture:** Standing with feet wide apart, belly thrust forward (compensatory lordosis to keep the center of gravity behind the hips).
- **Facies:** Usually normal. If there is ptosis or "hatchet facies," reconsider Myotonic Dystrophy or FSHD.

Vital Signs and Anthropometry

- **Pulse:** Check for tachycardia. [SEVERITY MARKER] Persistent tachycardia in DMD often signals early **Dilated Cardiomyopathy (DCM)**.
- **Weight/Height:** Plot on WHO/IAP charts. Steroid use often leads to obesity and stunted height.

Peripheral Signs

- **Calf Pseudohypertrophy:** Look for "rubbery" enlargement of the calves. It's not muscle; it's fat and fibrosis.
- **Other muscles:** Look for hypertrophy of the infraspinatus and deltoid (common in DMD).
- **Contractures:** Check the Achilles tendon (equinus deformity), hamstrings, and iliotibial bands.

Systemic Examination — Primary System (Neuromuscular)

1. Inspection:

- **Wasting:** Look for symmetric wasting of the pelvic girdle and shoulder girdle.
- **Pseudohypertrophy:** Calves [EXAMINER FAVORITE], occasionally tongue or vastus lateralis.

2. Palpation (Tone & Nutrition):

- **Tone:** Usually hypotonia or normal.
- **Muscle Consistency:** Calves feel firm/woody, not soft like normal muscle.

3. Motor Examination (The Core of the Case):

- **Power:** Use the MRC scale. Test proximal vs. distal.
    - *Lower limb:* Test hip extensors (Gluteus maximus) and abductors. These are the first to go.
    - *Upper limb:* Test serratus anterior (look for winging) and biceps.
- **Gowers' Sign [EXAMINER FAVORITE]:** Ask the child to sit on the floor and stand up.
    - *Technique:* Ensure the floor isn't slippery.
    - *What to look for:* The child turns prone, moves to a "four-point" position, extends knees, and "walks" his hands up his legs to upright the torso.
    - *Significance:* Indicates severe pelvic girdle and paraspinal muscle weakness.

4. Reflexes:

- **Deep Tendon Reflexes (DTRs):** Knee jerks are lost early (quadriceps wasting). **Ankle jerks are preserved** until very late stages. [EXAMINER FAVORITE - this dissociation is classic for DMD].
- **Plantar:** Always flexor.

5. Gait:

- **Waddling Gait:** Due to weak gluteus medius. The pelvis drops on the non-weight-bearing side.
- **Toe Walking:** To compensate for weak hip extensors and due to Achilles contracture.

Systemic Examination — Secondary Systems

- **CVS:** Displaced apex beat or S3 (Cardiomyopathy).
- **Respiratory:** Chest expansion (decreased in late stages), use of accessory muscles.
- **Spine:** Check for scoliosis (common after the child becomes wheelchair-bound).

SIGNS THAT CLINCH THE DIAGNOSIS [EXAMINER FAVORITES]

1. **Gowers' Sign:** The classic "climbing up oneself" maneuver.
2. **Calf Pseudohypertrophy:** Firm, enlarged calves in a weak child.
3. **Valley Sign:** A hollow behind the axilla caused by wasting of the posterior axillary fold (latissimus dorsi) against a hypertrophied infraspinatus.
4. **Reflex Dissociation:** Absent knee jerks with preserved ankle jerks.
5. **Meryon's Sign:** When you try to lift the child by the axillae, he "slips through" your hands due to shoulder girdle weakness.

DIAGNOSIS

Diagnostic Criteria

- **Clinical:** Progressive symmetric proximal weakness, Gowers' sign, calf hypertrophy, X-linked inheritance.
- **Biochemical:** Serum Creatine Kinase (CK) >10–50x normal.
- **Genetic:** Gold Standard. Multiplex PCR or MLPA (Multiplex Ligation-dependent Probe Amplification) to detect deletions/duplications in the *DMD* gene.

Differentials

1. **Becker Muscular Dystrophy (BMD):** Later onset (>5 years), remains ambulatory beyond 15–16 years.
2. **Spinal Muscular Atrophy (SMA) Type 3 (Kugelberg-Welander):** Neurogenic. Look for fasciculations, absent ankle jerks, and no pseudohypertrophy.

3. **Limb-Girdle Muscular Dystrophy (LGMD):** Autosomal (affects girls too), no cognitive involvement, CK not as high as DMD.

Investigations

- **Tier 1:** Serum CK (always elevated in DMD, often >10,000 IU/L), AST/ALT (often elevated—don't mistake for liver disease!).
- **Tier 2: MLPA** (detects 70% of mutations). If MLPA is negative, do **Gene Sequencing** (for point mutations).
- **Tier 3:** Muscle Biopsy (only if genetics are inconclusive). Shows "opaque fibers," fat infiltration, and **absent Dystrophin staining**.
- **Complications:** Baseline ECG/Echo (at diagnosis or age 6), Pulmonary Function Tests (PFTs).

Management Outline

- **Multidisciplinary Team (MDT):** Neuro, Physio, Cardio, Pulmo, Genetics.
- **Steroids [UPDATED]:** Prednisolone (0.75 mg/kg/day) or Deflazacort (0.9 mg/kg/day). Start in the "plateau phase" (usually age 4–6). *Goal: Prolong ambulation and protect cardiac/respiratory function*.
- **Physical Therapy:** Stretching to prevent contractures; night splints.
- **Genetic Counseling:** Mandatory for the mother (carrier testing).
- **Newer Therapies:** Exon skipping (e.g., Eteplirsen for Exon 51) or Gene therapy (recently FDA approved for specific ages).

EXAMINER'S VIVA

Q1: Why do these children walk on their toes? *A:* It is a compensatory mechanism. Weakness of the hip extensors (gluteus maximus) causes the center of gravity to shift forward; toe walking and lumbar lordosis help keep the center of gravity behind the hip joint to maintain balance. Later, it is fixed due to Achilles tendon contracture.

Q2: Why is the CK so high? *A:* Dystrophin acts as a "shock absorber" linking the cytoskeleton to the extracellular matrix. Without it, the sarcolemma tears during muscle contraction, leaking CK into the blood.

Q3: How do you distinguish DMD from SMA Type 3 at the bedside? *A:* SMA 3 will have fine tremors (minipolymyoclonus) of the hands, tongue fasciculations, and early loss of all DTRs (including ankles). DMD has calf hypertrophy and preserved ankle jerks.

Q4: What is the most common cause of death in DMD? *A:* Respiratory failure (due to weak diaphragm and intercostals) or Dilated Cardiomyopathy.

Q5: Show me how you measure the "Liver Span" in this child. *(Technique check: Percuss upper border in mid-clavicular line, palpate lower border, measure distance. Important because DMD patients often have high ALT/AST; you must prove the liver isn't actually enlarged to show the enzymes are from muscle).*

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting the case of a [Age] year old male, born of a non-consanguineous marriage, with a history of progressive difficulty in standing from a sitting position and frequent falls since age 4, currently having a waddling gait..."
- **Mistake to avoid:** Don't forget to check the **Upper Limb Proximal Muscles**. Residents often focus only on the legs. Check the Serratus Anterior for winging.
- **Mistake to avoid:** Don't say "The child has a positive Gowers' sign" without describing how he did it. The examiner wants to see your observation skills.
- **The "Liver Trap":** If you mention elevated ALT/AST in your labs, immediately clarify that these are found in muscle and the child has no clinical signs of liver disease (no icterus, no hepatomegaly).

---

5. Hydrocephalus

Subject: Neurology

This is a classic "bread and butter" neurology case. In the exam, the examiner isn't just looking for the diagnosis—they are watching your **handling of the infant** and your ability to differentiate between **communicating and non-communicating** types based on clinical signs.

HISTORY

Chief Complaint

- **Infants:** Rapidly increasing head size, "sunset eyes," irritability, vomiting, or failure to achieve milestones.
- **Older Children:** Headache (early morning), vomiting, blurring of vision, or declining school performance.

History of Present Illness

Ask these questions naturally:

- **"When did you first notice the head looking larger?"** (Onset: Congenital vs. Acquired).
- **"Is the child excessively irritable or crying in a high-pitched tone?"** (Signs of raised ICP).
- **"Does the child vomit, especially right after waking up, and is it forceful?"** (Projectile morning vomiting suggests obstructive hydrocephalus).
- **"Have you noticed the eyes looking downwards, even when the child is trying to look at you?"** (Setting-sun sign).
- **"Has the child lost any previously gained milestones, like sitting or neck holding?"** (Regression vs. Global Delay).

- **"Does the child have any weakness in the legs or difficulty walking?"** (Stretching of paracentral fibers around the ventricles).
- **"Any history of seizures or fever with neck stiffness in the past?"** (Post-meningitic hydrocephalus is the most common cause in developing countries).

Relevant Background History

- **Antenatal:** Ask about maternal infections (TORCH) or antenatal ultrasounds showing "water in the brain."
- **Birth History:** Prematurity (Germinal Matrix Hemorrhage) or birth trauma/asphyxia.
- **Developmental:** Detailed charting of motor milestones. Hydrocephalus often causes "gross motor delay" due to the weight of the head and stretching of pyramidal tracts.
- **Past History:** History of neonatal meningitis or TB meningitis is crucial.

EXAMINATION

General Survey

- **Observation:** Observe the child's head-to-body proportion. Is the head "heavy" and tilted to one side? Look for the **"Setting-Sun Sign"** (downward deviation of eyes with sclera visible above the iris).
- **Activity:** Is the child lethargic or hyper-irritable?
- **Skin:** Look for a **lumbosacral myelomeningocele** (turn the child over!). Look for neurocutaneous markers (café-au-lait spots for NF-1, which causes aqueductal stenosis).

Vital Signs and Anthropometry

- **Head Circumference (HC): [CRITICAL STEP]** Use a non-stretchable tape. Measure from the glabella to the most prominent part of the occiput (widest diameter). Plot on WHO/IAP growth charts. A crossing of percentiles is more significant than a single high reading.
- **Heart Rate:** Look for **Cushing's Triad** (Bradycardia, Hypertension, Irregular respirations) — this is a late sign of impending herniation.

Peripheral Signs

- **Hands/Feet:** Look for spasticity. In hydrocephalus, the lower limbs are often more affected than the upper limbs (due to the stretching of periventricular white matter fibers from the motor cortex to the legs).
- **Lymph Nodes:** Check for BCG scar and cervical nodes if TB is suspected.

Systemic Examination — Primary System (Neurology)

- **Inspection of Cranium:**
    - **Shape:** Frontal bossing (prominent forehead).

    - **Scalp Veins:** Look for dilated, non-pulsatile scalp veins. *Technique:* If you compress them, do they fill from the center outwards? (Sign of increased venous pressure).
    - **Sutures:** Palpate the sagittal and coronal sutures. Are they "separated" (diastasis)?
- **Palpation of Fontanelle:**
    - **Technique:** Child must be calm and held upright. Use the flat of your fingers.
    - **Findings:** A bulging, tense, non-pulsatile anterior fontanelle is the hallmark of raised ICP.
- **Percussion:**
    - **Macewen's Sign (Cracked Pot Sign):** [EXAMINER FAVORITE] Percuss at the junction of the frontal, temporal, and parietal bones. A "cracked pot" sound indicates separated sutures.
- **Transillumination:**
    - **Technique:** In a dark room, place a high-intensity torch against the skull.
    - **Significance:** A glow >2cm beyond the torch suggests thinned out cortex (Hydrocephalus or Hydranencephaly).
- **Cranial Nerves:**
    - **CN II:** Fundoscopy (look for papilledema in older children; rare in infants with open fontanelles). Check for optic atrophy.
    - **CN III, IV, VI:** Look for 6th nerve palsy (false localizing sign) and the sunset sign.
- **Motor System:**
    - Check for **Spastic Diplegia.** Test the "Adductor Angle" and "Popliteal Angle." Increased tone and brisk Deep Tendon Reflexes (DTRs) in the legs are common.

Systemic Examination — Secondary Systems

- **Spine:** Check for any midline defects, tufts of hair, or sinus tracts.
- **Abdomen:** Palpate for a palpable VP shunt chamber or any pseudocyst if the child already has a shunt.

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

1. **Macewen's Sign:** Percussion of the skull sounding like a cracked pot.
2. **Setting-Sun Phenomenon:** Downward gaze due to pressure on the midbrain tectum.
3. **Tense, Bulging Fontanelle:** In an upright, non-crying infant.
4. **Transillumination:** To differentiate from Megalencephaly (where transillumination is negative).

Severity Assessment [SEVERITY MARKER]

- **Rapidly increasing HC:** >2cm per month in the first trimester.

- **Altered Sensorium:** Suggests acute decompensation.
- **Optic Atrophy:** Suggests chronic, untreated high pressure leading to permanent blindness.

DIAGNOSIS

Diagnostic Criteria

- Clinical diagnosis based on **HC > +2SD for age/sex** plus signs of raised ICP.
- Confirmed by Neuroimaging (USG Cranium for infants, MRI for older children).

Differentials

1. **Megalencephaly:** Large brain, but no signs of raised ICP, normal fontanelle, negative transillumination.
2. **Chronic Subdural Hematoma:** History of trauma, seizures, and a more "box-shaped" head.
3. **Rickets:** Can cause frontal bossing and large-looking head, but sutures are not separated and HC is usually within normal limits.

Investigations

- **Tier 1:** USG Cranium (if fontanelle is open). It's bedside, no radiation. Shows ventricular size and Evan's Index.
- **Tier 2: MRI Brain (Gold Standard).** Look for "periventricular lucency" (suggests transependymal seepage of CSF).
- **Tier 3:** CSF analysis (if post-meningitic) and TORCH screen.

Management Outline

- **Medical:** Acetazolamide or Furosemide (only temporary/temporizing measures).
- **Surgical (Definitive):**
    - **Ventriculoperitoneal (VP) Shunt:** Most common.
    - **Endoscopic Third Ventriculostomy (ETV):** [UPDATED] Preferred for obstructive hydrocephalus (e.g., Aqueductal stenosis) as it avoids foreign body insertion.

EXAMINER'S VIVA

1. **Q: How do you measure Head Circumference in a child with a VP shunt?** *A: Always measure the maximum diameter, even if it goes over the shunt reservoir, but note the presence of the shunt in your report.*
2. **Q: What is Evan's Index?** *A: It is the ratio of the maximum width of the frontal horns of the lateral ventricles to the maximum internal diameter of the skull. A value >0.3 indicates ventriculomegaly.*

3. **Q: Why do we see "Sunset Sign"?** *A: Pressure on the periaqueductal gray matter and the superior colliculi (Parinaud's Syndrome).*
4. **Q: What are the signs of Shunt Malfunction?** *A: Recurrence of bulging fontanelle, vomiting, irritability, and redness along the shunt tract.*
5. **Q: Can you have hydrocephalus with a normal head circumference?** *A: Yes, in older children whose sutures have fused (usually >18 months to 2 years).*

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting [Name], a [Age] old male, born of a [Consanguineous/Non-consanguineous] marriage, presenting with a progressive increase in head size since [Duration] and delay in motor milestones..."
- **Common Mistake:** Forgetting to check the spine for a myelomeningocele. If you miss a scar or a sac on the back, you fail the neurology case.
- **Technique:** When percussing for Macewen's sign, ensure the child is not crying, as crying naturally tenses the fontanelle and changes the resonance.

---

6. Microcephaly

Subject: Neurology

LONG CASE FRAMEWORK: MICROCEPHALY

HISTORY

Chief Complaint

- "The head looks small compared to the body" (Parental observation)
- "Not reaching milestones on time" (Developmental delay)
- "Recurrent fits/shaking of limbs" (Seizures)
- "Stiffness of limbs or difficulty in feeding"

History of Present Illness

Focus on differentiating Primary (Genetic/Developmental) from Secondary (Insurative/Acquired) Microcephaly.

- **Onset and Progression:** "Was the head small at birth, or did it fail to grow later?" (Small at birth suggests genetic or intrauterine insult; postnatal slowing suggests metabolic or progressive encephalopathy).
- **Developmental Trajectory:** "Tell me exactly what the child can do now." (Map all 4 domains). "Was there a period of normal development followed by a loss of skills?" (Regression suggests neurodegenerative/metabolic causes like Rett syndrome or Krabbe disease).

- **Seizure History:** "Describe the episodes. Are they sudden jerks (infantile spasms) or stiffening?" (Infantile spasms + microcephaly often point to Aicardi or severe encephalopathy).
- **Antenatal Insults (TORCH):** "Did the mother have fever with rash, joint pains, or eye redness during pregnancy?" (Zika, Rubella, CMV). "Any history of radiation or drug intake (Antiepileptics/Alcohol)?"
- **Perinatal Events:** "Did the baby cry immediately? Was there a need for NICU/Ventilator?" (HIE is a major cause of secondary microcephaly).

Relevant Background History

- **Past History:** History of meningitis or head trauma (Secondary causes).
- **Antenatal/Birth:** Detailed birth weight and head circumference at birth (if available).
- **Developmental:** This is the core. You must quantify the Developmental Quotient (DQ) for each domain.
- **Family History:** "Are the parents related (Consanguinity)?" (Autosomal Recessive Microcephaly - MCPH). "Are any siblings or the parents themselves 'small-headed' but otherwise normal?" (Benign Familial Microcephaly).
- **Socioeconomic:** Impact on the family, access to early intervention centers.

EXAMINATION

General Survey

- **The "First Look":** Observe the child's interaction. Is there "Bird-like" facies (MCPH)? Is the child hyperactive or self-mutilating?
- **Nutritional Status:** Check for wasting. Children with severe neuro-disability often have oromotor dysfunction leading to PEM.
- **Skin (Crucial):**
    - *Hypopigmented macules (Ash-leaf):* Tuberous Sclerosis.
    - *Port-wine stain:* Sturge-Weber.
    - *Jaundice/Petechiae at birth:* TORCH.
- **Facies:** Look for sloping forehead (MCPH), flat philtrum/thin upper lip (Fetal Alcohol Syndrome), or cataracts (Rubella).

Vital Signs and Anthropometry

- **Head Circumference (OFC): [EXAMINER FAVORITE]**
    - *Technique:* Use a non-stretchable tape. Measure from the glabella (front) to the most prominent point of the occiput (back). Take the maximum reading.
    - *Interpretation:* Plot on WHO/IAP charts. Microcephaly = OFC < -3 SD for age and sex.

- **Parental OFC:** Always measure the parents' heads. If the child is -2.5 SD and parents are also -2 SD, it may be familial.
- **Chest Circumference:** In infants, OFC is usually > CC. If CC > OFC before 6-9 months, it's a red flag.

Peripheral Signs

- **Eyes:** Check for chorioretinitis (CMV/Toxo), optic atrophy, or cherry-red spot (Metabolic).
- **Ears:** Low-set ears or preauricular tags (Syndromic).
- **Hands:** Simian crease, clinodactyly, or overlapping fingers (Trisomies).

Systemic Examination — Primary System (Neurology)

- **Inspection:**
    - *Skull Shape:* Look for a sloping forehead and a prominent occiput or flat occiput.
    - *Sutures/Fontanelle:* Feel the anterior fontanelle. Is it closed prematurely? (Craniosynostosis). Is it bulging? (Post-meningitic hydrocephalus/atrophy).
- **Tone:** Assess for spasticity (HIE/Cerebral Palsy) or hypotonia (Genetic/Metabolic).
- **Power:** Usually difficult to grade in infants; look for spontaneous movements and antigravity reach.
- **Reflexes:** Brisk DTRs and extensor plantars are common in secondary microcephaly (spastic quadriparesis).
- **Cranial Nerves:** Focus on Vision (Fixing/Following) and Hearing (Startle/Turning to sound).

Systemic Examination — Secondary Systems

- **Abdomen:** Hepatosplenomegaly (Storage disorders or TORCH).
- **CVS:** Murmurs (Congenital Rubella Syndrome - PDA/PS).

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **Sloping Forehead:** Classic for Primary Microcephaly (MCPH).
- **Closed Anterior Fontanelle:** If closed before 3 months, suggests either Craniosynostosis or primary lack of brain growth.
- **Transillumination of Skull:** Perform in a dark room. If the skull "lights up," it suggests hydranencephaly or severe porencephaly (Secondary).
- **Ridge along Sutures:** Suggests Craniosynostosis (The head is small because the bones fused, not because the brain didn't grow).

Severity Assessment

- **Mild:** -2 to -3 SD.
- **Severe:** < -3 SD.

- **Functional Severity:** [SEVERITY MARKER] Presence of refractory seizures, inability to swallow (bulbar signs), or total lack of social milestone attainment.

DIAGNOSIS

Diagnostic Criteria

- **Microcephaly:** OFC < -3 SD below the mean for age, sex, and gestation.
- **Microcephaly Vera:** Specifically refers to genetic primary microcephaly with a sloping forehead and simplified gyral pattern.

Differentials

1. **Primary Microcephaly (MCPH):** Autosomal recessive, consanguinity, sloping forehead, moderate ID, no other malformations.
2. **Secondary (Insurative) Microcephaly:** History of HIE, meningitis, or TORCH; associated with spasticity and seizures.
3. **Craniosynostosis:** Primary fusion of sutures; palpable ridges, abnormal head shape (Scaphocephaly/Brachycephaly).
4. **Syndromic:** Down syndrome, Edward syndrome, Cri-du-chat (look for dysmorphism).

Investigations

- **Tier 1:** Detailed Anthropometry (OFC plotting), TORCH screen (IgM/PCR), Serum Ammonia/Lactate (if regression).
- **Tier 2: Neuroimaging (MRI Brain)** - Look for simplified gyral pattern (Primary), calcifications (CMV/Toxo), or periventricular leukomalacia (HIE). **Karyotyping/CMA** if syndromic.
- **Tier 3:** Targeted Gene Panels (e.g., ASPM gene for MCPH).

Management Outline

- **Multidisciplinary Approach:** No "cure" for the small head; focus on the brain's function.
- **Seizure Control:** Anti-epileptic drugs (Levetiracetam/Valproate).
- **Early Intervention:** Physiotherapy for spasticity, occupational therapy, speech therapy.
- **Nutrition:** High-calorie diet, Ryle's tube feeding if bulbar palsy.
- **Genetic Counseling:** Recurrence risk (25% for AR conditions like MCPH).

EXAMINER'S VIVA

1. **Q: How do you differentiate Craniosynostosis from Microcephaly on examination?**
    - *A:* In craniosynostosis, there are palpable bony ridges over sutures and an abnormal skull shape (e.g., boat-shaped). In microcephaly, the shape is usually symmetrical (though small) and sutures aren't necessarily ridged.

2. **Q: What is the "Catch-up" growth of the head?**
    - *A:* Premature infants may have a small head at birth, but it should follow the curve and catch up by 2 years. If it stays below -2 SD, it's true microcephaly.
3. **Q: What is the significance of the "Sloping Forehead"?**
    - *A:* It indicates a failure of development of the frontal lobes, typically seen in genetic primary microcephaly.
4. **Q: At what age does the Anterior Fontanelle normally close?**
    - *A:* Usually between 9 to 18 months. Closure before 3 months is pathological.
5. **Q: Technique: Show me how you measure the OFC in a crying child.**
    - *A:* (Demonstrate) Ensure the tape is above the eyebrows and ears, over the most prominent part of the occiput. If the child is struggling, have the mother hold the head. Take the average of three readings or the maximum reading.

LONG CASE PRESENTATION TIPS

- **The Opening:** "I am presenting [Name], a [Age] old male/female, born of [Consanguineous/Non-consanguineous] marriage, presenting with a small head since birth and global developmental delay, currently functioning at a DQ of [X]%."
- **Mistake to Avoid:** Don't just say "the head is small." You must state the exact measurement and how many SDs it is below the mean.
- **The "Why":** Examiners want to see if you can distinguish between a "static" insult (HIE) and a "progressive" one (Metabolic). Emphasize the presence or absence of developmental regression.

Cardiovascular

7. Congestive cardiac failure

Subject: Cardiovascular

This is a high-stakes long case. In Pediatrics, Congestive Cardiac Failure (CCF) is a **clinical diagnosis**. You must demonstrate to the examiner that you can distinguish between "left-sided" (pulmonary congestion) and "right-sided" (systemic venous congestion) features, while simultaneously hunting for the underlying etiology (Congenital vs. Acquired).

HISTORY

Chief Complaint

- **Infants:** Poor feeding, "suck-rest-suck" cycle, excessive forehead sweating, rapid breathing, and poor weight gain.

- **Older Children:** Breathlessness on exertion, easy fatigability, orthopnea, paroxysmal nocturnal dyspnea (PND), or swelling of feet/abdomen.

History of Present Illness

Ask these exact questions to characterize the severity and type:

- **"Does the baby stop midway through breastfeeding to catch their breath?"** (Poor feeding/Feeding fatigue is the infant equivalent of NYHA Class III/IV dyspnea).
- **"Do you notice beads of sweat on the forehead specifically during feeding?"** (Sympathetic overactivity).
- **"How many pillows does your child use at night, or do they prefer sleeping propped up?"** (Orthopnea).
- **"Does the child develop a cough or wheeze when lying down?"** (Cardiac asthma/Pulmonary congestion).
- **"Is the puffiness worse in the morning (renal) or does the swelling increase towards the evening (cardiac)?"**
- **"Has there been a recent history of sore throat or joint pains?"** (Points toward Rheumatic Heart Disease).
- **"Is there a history of bluish discoloration of lips or nails during crying?"** (Cyanotic heart disease with failure, e.g., TAPVC or TGA).

Relevant Background History

- **Antenatal/Birth:** Maternal diabetes (Hypertrophic Cardiomyopathy), Rubella (PDA/PS), or SLE (Congenital Heart Block).
- **Developmental:** Gross motor delay is common in chronic CCF due to malnutrition and fatigue.
- **Nutritional:** Detailed caloric intake. CCF is a hypermetabolic state; "cardiac cachexia" is a real entity.
- **Past History:** Recurrent pneumonia (Increased pulmonary blood flow - L to R shunts).

EXAMINATION

General Survey

- **Activity:** Observe for "Air Hunger." Is the child tachypneic at rest?
- **Posture:** Is the child sitting up and leaning forward (Tripod position)?
- **Nutrition:** Look for visible wasting of temporalis muscle and loss of gluteal fat. [SEVERITY MARKER]
- **Facies:**
    - Down Syndrome (AVSD)

- Williams Syndrome (Elfin facies - Supravalvular AS)
- DiGeorge (Conotruncal anomalies)

Vital Signs and Anthropometry

- **Heart Rate:** Must count for a full minute. Look for **Tachycardia out of proportion to fever**.
- **Respiratory Rate:** Count while the child is calm. Look for subcostal/intercostal retractions.
- **Blood Pressure:** [EXAMINER FAVORITE] **Must measure in all four limbs.** A lower BP in the lower limbs compared to upper limbs indicates Coarctation of the Aorta.
- **Weight:** Plot on WHO charts. Sudden weight gain may indicate occult edema.

Peripheral Signs

- **Pulse:**
  - *Volume:* Low volume (Myocarditis/AS), Bounding (PDA/AR).
  - *Radio-femoral delay:* Feel simultaneously to rule out Coarctation.
- **Capillary Refill Time (CRT):** Press over the sternum for 5 seconds. >3 seconds indicates poor peripheral perfusion (Cold Heart Failure).
- **Clubbing:** Use Schamroth's window. Significant in Cyanotic CCF or infective endocarditis.
- **JVP:** Difficult in infants due to short necks. In older children, look for the highest point of pulsation of the internal jugular vein. **Hepatojugular Reflux:** Press the liver for 10-30 seconds; a persistent rise in JVP >3cm is positive.

Systemic Examination — Primary System (CVS)

- **Inspection:**
  - **Precordial Bulge:** Indicates long-standing cardiomegaly in a compliant young chest wall.
  - **Apical Impulse:** Locate it. Is it shifted down and out?
- **Palpation:**
  - **Apex Beat:** Characterize it. *Heaving* (Pressure overload - AS/HTN), *Hyperdynamic* (Volume overload - VSD/PDA/AR).
  - **Parasternal Heave:** Place the heel of your hand on the left parasternal area. Lift indicates Right Ventricular Enlargement.
  - **Thrills:** Palpate with the MCP joints. A thrill makes a murmur Grade 4/6.
- **Percussion:** Only to define the right heart border if you suspect dextrocardia or massive pericardial effusion (dullness in the 2nd LICS).
- **Auscultation:**
  - **S1/S2:** Is S2 loud (Pulmonary Hypertension)?

- **S3 (Gallop Rhythm):** [EXAMINER FAVORITE] Best heard at the apex with the bell. It is the hallmark of ventricular failure in children.
- **Murmurs:** Describe site, timing, character, and radiation. (e.g., Pansystolic murmur of VSD or MR).

Systemic Examination — Secondary Systems

- **Respiratory:** Fine crepitations at the bases (Pulmonary edema). Note: In infants, CCF often presents with generalized wheezing.
- **Abdomen: Tender Hepatomegaly.** Measure the liver span. A firm, smooth, tender liver edge is the most reliable sign of Right Heart Failure in infants. [EXAMINER FAVORITE]
- **CNS:** Assess for embolic strokes if you suspect Infective Endocarditis or R-to-L shunts.

DIAGNOSIS

Diagnostic Criteria

- **Ross Classification (for Infants):**
  - Class I: Asymptomatic.
  - Class II: Tachypnea or sweating with feeds; mild growth retardation.
  - Class III: Marked tachypnea/sweating; prolonged feeding time; marked growth retardation.
  - Class IV: Symptoms at rest (retractions, grunting).
- **Modified NYHA (for older children).**

Differentials

1. **Severe Pneumonia:** Will have fever, bronchial breath sounds, but no hepatomegaly or gallop.
2. **Severe Anemia:** Can cause "High Output Failure." Look for extreme pallor and hemic murmurs.
3. **Acute Nephritic Syndrome:** Edema and hypertension, but the liver is usually not tender, and the heart size may be normal.

Investigations

- **Tier 1:** Chest X-ray (Cardiomegaly - CTR >60% in infants, >50% in children; Pulmonary plethora). ECG (Chamber hypertrophy, arrhythmias).
- **Tier 2: Echocardiography (Gold Standard).** To define anatomy and calculate Ejection Fraction/Fractional Shortening.
- **Tier 3:** NT-proBNP (Biomarker for heart strain); Cardiac Catheterization (if surgery is planned).

Management Outline

1. **Positioning:** Propped up (30-45 degrees).

2. **Oxygen:** If saturations are low.
3. **Diuretics:** IV Furosemide (1-2 mg/kg) to reduce preload.
4. **Inotropes:** If in cardiogenic shock (Dopamine/Dobutamine).
5. **Afterload Reduction:** ACE Inhibitors (Enalapril/Captopril) — [UPDATED] Standard of care for chronic failure.
6. **Nutrition:** High-calorie feeds (MCT oil, frequent small feeds).

EXAMINER'S VIVA

1. **Q: Why is the liver tender in CCF?**
    - A: Rapid stretching of the Glisson's capsule due to acute venous congestion.
2. **Q: How do you differentiate a cardiac murmur from a hemic murmur of anemia?**
    - A: Hemic murmurs are always systolic, soft (Grade 1-2), heard best at the base, and disappear once the anemia is corrected.
3. **Q: What is "Preload" and "Afterload" in simple terms?**
    - A: Preload is the volume of blood returning to the heart (stretch). Afterload is the resistance the heart must pump against (squeeze).
4. **Q: How do you measure Liver Span in a 2-year-old?**
    - A: Percuss the upper border (usually 5th ICS) and palpate the lower border. Measure the distance in cm.
5. **Q: What is the significance of a "Gallop Rhythm"?**
    - A: It represents rapid ventricular filling into a non-compliant, failing ventricle. It is a sign of significant myocardial dysfunction.

LONG CASE PRESENTATION TIPS

- **The Opening:** "I am presenting 8-month-old [Name], who presents with a 2-week history of feeding transition fatigue, forehead sweating, and excessive weight gain, currently in Ross Class III congestive cardiac failure, likely secondary to a high-flow left-to-right shunt."
- **Common Mistake:** Forgetting to check the BP in the legs. If you don't do this, the examiner will assume you missed a Coarctation.
- **What Examiners Watch For:** They watch how you handle the infant. If the baby is crying, your HR and RR measurements are invalid. Always do the "quiet" parts of the exam (auscultation, observation) first.

---

8. Ventricular septal defect (VSD)

Subject: Cardiovascular

This is a classic "Left-to-Right Shunt" case. In the exam, you aren't just diagnosing a VSD; you are quantifying the shunt, assessing pulmonary pressures, and looking for heart failure.

HISTORY

Chief Complaint

- **Infants:** Breathlessness during feeds, poor weight gain, excessive sweating (forehead), and recurrent "chest congestion" (LRTI).
- **Older Children:** Exercise intolerance, palpitations, or often an incidental murmur detection.

History of Present Illness

Ask these naturally to build the hemodynamic picture:

- **Feeding History (The "Stress Test"):** "Does he take breaks while breastfeeding? Does he sweat profusely on the forehead while sucking? How long does a single feed take?" (Interrupted feeds/suck-rest-suck cycle >30 mins suggests heart failure).
- **Growth:** "Compared to his siblings or peers, how is his weight? Have you had to change clothing sizes recently?"
- **Respiratory:** "How many times has he been hospitalized for pneumonia? Does he have a persistent cough or wheeze?"
- **Cyanosis/Squatting:** "Do his lips turn blue while crying? Does he sit down abruptly after running?" (Negative history helps exclude Tetralogy of Fallot or Eisenmenger).
- **Hyperdynamic state:** "Can you feel his heart beating fast against his chest wall even at rest?"

Relevant Background History

- **Antenatal:** Maternal diabetes, SLE, or intake of anti-epileptics (associated with CHD).
- **Birth:** Birth weight is crucial to differentiate "failure to thrive" from "small for gestational age."
- **Development:** Motor delays are common in large shunts due to poor caloric reserve and frequent illnesses.
- **Socioeconomic:** Can they afford the surgery? This is a standard PG question regarding "Modified Kuppuswamy Scale."

EXAMINATION

General Survey

- **Activity:** Is the infant tachypneic at rest? Look for **subcostal/intercostal retractions** [SEVERITY MARKER].

- **Nutritional Status:** Look for "cardiac cachexia." Assess weight-for-length and mid-upper arm circumference.
- **Facies:** Look for Down Syndrome (AVSD is more common, but VSD occurs), Turner, or Williams syndrome.
- **Precordial Bulge:** Look for a bulge to the left of the sternum. This indicates long-standing cardiomegaly in a compliant young ribcage.

Vital Signs and Anthropometry

- **Respiratory Rate:** Count for a full minute while the child is asleep or calm. Tachypnea is the earliest sign of failure.
- **Pulses:** Palpate all four limb pulses.
    - **Technique:** Simultaneously palpate right radial and right femoral to rule out Coarctation (often associated with VSD).
    - **Character:** In a large VSD, the pulse is often **bounding** (hyperdynamic circulation).
- **Blood Pressure:** Must be measured in at least one upper and one lower limb using the correct cuff size (bladder width = 40% of arm circumference).

Peripheral Signs

- **Clubbing/Cyanosis:** Check the tongue and nail beds. If present in a VSD case, it suggests **Eisenmenger Syndrome** (Reversal of shunt).
- **Edema:** In infants, look for **eyelid swelling** rather than pedal edema.
- **JVP:** Difficult in infants due to short necks. In older children, look for a prominent 'v' wave if tricuspid regurgitation (secondary to PH) has set in.

Systemic Examination — Cardiovascular

1. Inspection:

- **Apical Impulse:** Look for the location. If shifted down and out, it suggests LV volume overload.
- **Precordial Activity:** Look for a "hyperdynamic" precordium (visible pulsations).

2. Palpation:

- **Apex Beat:** Use the palm first, then fingertips. In VSD, it is **hyperdynamic** (forceful but ill-sustained) and displaced.
- **Thrills:** [EXAMINER FAVORITE] Palpate with the ulnar border of your hand at the left lower sternal border (LLSB). A systolic thrill here is classic for a small-to-moderate VSD (Maladie de Roger).
- **Parasternal Heave:** Use the heel of your hand at the left sternal border. A lift suggests Right Ventricular Hypertrophy (RVH) due to Pulmonary Hypertension (PH).
- **P2 Palpation:** Palpate the 2nd left intercostal space. If P2 is palpable, it signifies **Pulmonary Hypertension**.

3. Percussion:

- Not usually done in children, but you may percuss the right cardiac border to check for RA enlargement.

4. Auscultation:

- **S1:** Usually normal.
- **S2:** Listen at the base.
    - Wide, variable split: Due to prolonged LV ejection.
    - **Loud P2:** [SEVERITY MARKER] Indicates Pulmonary Hypertension.
- **S3:** A low-pitched sound at the apex indicates a large shunt and incipient heart failure.
- **Murmur of VSD:**
    - **Character:** Holosystolic (pansystolic).
    - **Location:** Left 3rd/4th intercostal space.
    - **Radiation:** Like a wheel-spoke pattern, but primarily to the right sternal border.
- **Mitral Diastolic Flow Rumble:** [EXAMINER FAVORITE] Listen at the apex with the bell. This indicates a **large shunt** (Qp:Qs > 2:1) because of increased blood returning from lungs through the mitral valve.

Secondary Systems

- **Respiratory:** Fine crepitations at the bases (Pulmonary edema/CCF).
- **Abdomen:** Palpate the liver. A firm, enlarged liver (>2cm) in an infant is a reliable sign of Right Heart Failure.

SIGNS THAT CLINCH THE DIAGNOSIS [EXAMINER FAVORITES]

1. **Pansystolic Murmur at LLSB:** The hallmark of VSD.
2. **Systolic Thrill:** Paradoxically, a loud murmur and thrill often mean a *smaller* (restrictive) VSD.
3. **Mid-Diastolic Rumble at Apex:** Indicates a large L-to-R shunt (High flow across the mitral valve).
4. **Palpable P2:** Indicates the development of Pulmonary Arterial Hypertension (PAH).
5. **Reverse Murmur Intensity:** If the murmur becomes shorter and softer while P2 becomes louder, the patient is developing Eisenmenger Syndrome.

DIAGNOSIS

Differentials

1. **Tricuspid Regurgitation:** Murmur increases with inspiration (Carvallo's sign); associated with pulsatile liver.
2. **Mitral Regurgitation:** Murmur radiates to the axilla; apex is usually more displaced.
3. **Atrioventricular Septal Defect (AVSD):** Common in Down Syndrome; ECG shows "Superior Axis" (Left axis deviation).

Investigations

- **ECG:**
    - Small VSD: Normal.
    - Large VSD: LVH (tall R in V5-V6), LAH (broad P wave).
    - PAH: RVH (Tall R in V1), **Katz-Wachtel Phenomenon** (Large biphasic QRS complexes in V2-V4).
- **Chest X-ray:** Cardiomegaly (LV/LA type), increased pulmonary vascular markings, and a prominent pulmonary artery segment.
- **Echocardiography (Gold Standard):** Defines location (perimembranous, muscular, subarterial), size, and pressure gradient.

Management Outline

1. **Medical:**
    - Anti-failure: Diuretics (Furosemide, Spironolactone) and ACE inhibitors (Enalapril) to reduce afterload.
    - Nutrition: High-calorie feeds (MCT oil, formula fortification).
2. **Surgical:**
    - Indications: Failure to thrive, refractory CCF, or Qp:Qs > 1.5:1.
    - Procedure: Primary patch closure (Dacron/Pericardium).
    - Timing: Usually 3–6 months for large VSDs; earlier if intractable.

EXAMINER'S VIVA

- **Q: Why does a small VSD have a louder murmur than a large one?**
    - A: A small VSD is "restrictive," creating a high-pressure gradient between the LV and RV, leading to high-velocity, turbulent flow.
- **Q: What is the Katz-Wachtel phenomenon?**
    - A: It is the presence of large biphasic QRS complexes in mid-precordial leads (V2-V4), indicating biventricular hypertrophy.
- **Q: How do you differentiate a VSD murmur from an ASD murmur?**

    - A: VSD is pansystolic at the LLSB. ASD is an ejection systolic murmur at the ULSB (due to increased flow across the pulmonary valve, not the defect itself) with a fixed split S2.
- **Q: What is Eisenmenger Syndrome?**
    - A: It is the reversal of a long-standing L-to-R shunt to a R-to-L shunt due to irreversible pulmonary vascular obstructive disease. The murmur disappears, and the patient becomes cyanotic.
- **Q: Technique: How do you differentiate a thrill from a heave?**
    - A: A thrill is a palpable murmur (vibration/tactile), felt best with the ulnar border or MCP joints. A heave is a palpable impulse (lifting the hand), indicating chamber hypertrophy.

LONG CASE PRESENTATION TIPS

- **The Opening:** "I am presenting [Name], a [Age] old male, with a history of recurrent lower respiratory tract infections, feeding difficulties, and poor weight gain, currently presenting with a loud pansystolic murmur and features of pulmonary hypertension..."
- **Common Mistake:** Forgetting to check the back for a thoracotomy scar (previous PA banding).
- **Watch out:** If you say "P2 is loud," the examiner will immediately ask you to palpate it. Never say it's loud if you haven't felt for it in the 2nd ICS.

---

9. Atrial septal defect (ASD)

Subject: Cardiovascular

This is a classic "stable" long case. The challenge isn't finding the diagnosis—it’s the precision of your physical exam and your ability to explain the hemodynamics. In an ASD, the findings are subtle. If you miss the fixed splitting of S2 or the grade II ejection systolic murmur, you’ve missed the case.

HISTORY

Chief Complaint

- Often asymptomatic (detected on routine screening/school physical).
- Frequent lower respiratory tract infections (LRTIs).
- Poor weight gain or "tires easily" during play (older child) or feeding (infant).
- Palpitations or exercise intolerance (usually adolescent/older child).

History of Present Illness

Ask these exact questions to gauge the shunt size and impact:

- **"Does the child get breathless while running compared to peers of the same age?"** (Assesses NYHA/Ross class).

- **"How many times in the last year has the child had a cough/fever lasting >5 days requiring antibiotics?"** (LRTIs suggest increased pulmonary blood flow).
- **"Does the child complain of their heart 'thumping' or 'skipping a beat'?"** (Atrial arrhythmias, more common in older patients).
- **"Has there ever been any bluish discoloration of lips or nails during crying or exertion?"** (Crucial to rule out Eisenmenger syndrome or associated cyanotic heart disease).
- **"Has the child ever fainted or complained of chest pain?"** (Suggests pulmonary hypertension or associated lesions).

Relevant Background History

- **Antenatal:** Ask about maternal diabetes or SLE (though more common in VSD/CHB). Ask about first-trimester febrile illness (Rubella).
- **Birth History:** Prematurity (increased incidence of Secundum ASD/PDA).
- **Developmental:** Gross motor delays often seen in large shunts due to poor weight gain.
- **Nutritional:** Detailed calorie count. Large left-to-right shunts cause a hypermetabolic state.
- **Family History:** ASD has a high recurrence risk in siblings. Ask about "sudden deaths" or "early strokes" (Paradoxical embolism).

EXAMINATION

General Survey

- **Activity:** Is the child active in the bed or tachypneic at rest?
- **Facies:** Look for **Holt-Oram Syndrome** (triphalangeal thumbs/hypoplastic radius) or **Down Syndrome** (Primum ASD/AVSD).
- **Nutritional Status:** Check for "Precordial Bulge"—a sign of long-standing right ventricular enlargement in a growing skeleton.

Vital Signs and Anthropometry

- **Respiratory Rate:** Count for a full minute. Look for subtle subcostal retractions.
- **Pulse:** Assess for **Atrial Fibrillation** (irregularly irregular) in older children.
- **Blood Pressure:** Compare upper and lower limbs (always rule out associated Coarctation).
- **Growth:** Plot on WHO/IAP charts. ASDs typically affect weight more than height.

Peripheral Signs

- **Clubbing/Cyanosis:** Should be absent. If present, think Eisenmenger or associated PS.
- **JVP:** Look for a prominent **'a' wave** (if RV compliance is low) or **'v' wave** (if tricuspid regurgitation has developed).
- **Liver:** Palpate for hepatomegaly and feel for **systolic pulsations** (indicates significant TR).

Systemic Examination — Cardiovascular

Inspection

- **Precordial Bulge:** Look from the foot of the bed. In ASD, the bulge is typically over the left parasternal area (RV enlargement).
- **Apical Impulse:** Usually shifted laterally but not necessarily downwards (Volume overload of RV, not LV).

Palpation

- **Apex Beat:** Use the ulnar border of your hand. In ASD, the apex is formed by the **Right Ventricle**. It feels "tapping" or "diffuse."
- **Left Parasternal Heave:** [SEVERITY MARKER] Use the heel of your hand at the left 3rd/4th intercostal space. Grade it (1-3). This confirms RV volume/pressure overload.
- **Pulsation in 2nd Left IS:** Palpable P2 (suggests Pulmonary Hypertension).
- **Thrills:** Usually **absent** in isolated ASD. If a thrill is present, look for associated Pulmonic Stenosis or VSD.

Percussion

- Not routinely done, but you may find the right heart border shifting beyond the right sternal edge.

Auscultation

This is where the exam is won or lost. Use the diaphragm for high-pitched sounds.

1. **S1:** Usually normal or loud (due to forceful closure of Tricuspid valve).
2. **S2: [EXAMINER FAVORITE]** Listen at the 2nd left Intercostal Space.
    - **Wide and Fixed Splitting:** The hallmark. Explain why: The split is wide because of RV volume overload (delayed P2). It is fixed because the respiratory variations in venous return are equalized between the two atria through the defect.
3. **Ejection Systolic Murmur (ESM):** Grade II-III/VI at the **Pulmonary Area**.
    - *Crucial Point:* This murmur is NOT due to flow across the ASD (the pressure gradient is too low). It is due to **increased stroke volume across the pulmonary valve** (Relative Pulmonic Stenosis).
4. **Mid-Diastolic Murmur (MDM):** Listen at the **Lower Left Sternal Border** (Tricuspid area) with the bell.
    - [SEVERITY MARKER] This indicates a large shunt (Qp:Qs > 2:1) causing relative Tricuspid Stenosis.

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **Wide, Fixed Split S2:** Pathognomonic for ASD.
- **ESM at Pulmonary Area without a thrill:** Distinguishes it from organic Pulmonic Stenosis.
- **Mid-diastolic flow murmur at Tricuspid area:** Indicates a large L-to-R shunt.
- **Right Ventricular Heave:** Confirms the hemodynamic impact is on the right side of the heart.

Severity Assessment

- **Mild:** Asymptomatic, normal growth, no MDM, no heave.
- **Moderate:** Frequent LRTIs, mild growth lag, RV heave present.
- **Severe:** Failure to thrive, MDM present, palpable P2, signs of heart failure (hepatomegaly).

DIAGNOSIS

Diagnostic Criteria

- Clinical diagnosis based on S2 characteristics and murmur.
- Confirmed via **Transthoracic Echocardiography (TTE)**.

Differentials

1. **Partial Anomalous Pulmonary Venous Connection (PAPVC):** Clinically identical to ASD; requires Echo/CT to differentiate.
2. **Pulmonic Stenosis (PS):** S2 split is wide but **not fixed** (moves with respiration). Murmur is louder, harsher, and usually accompanied by a **thrill** and an **ejection click**.
3. **Ventricular Septal Defect (VSD):** Murmur is pansystolic, loudest at the 4th IS, and the apex is LV type.

Investigations

- **ECG:** Look for **Right Axis Deviation** and **rSR' pattern** in V1 (Incomplete RBBB). *Primum ASD will show Left Axis Deviation*.
- **Chest X-ray:** Cardiomegaly (RV type), prominent pulmonary artery segment, and **increased pulmonary vascular markings** (Plethora).
- **Echocardiography (Gold Standard):** Defines type (Secundum is most common), size, direction of shunt, and pulmonary pressures.

Management Outline

- **Medical:** Treat CCF if present (Diuretics like Furosemide). Treat LRTIs.
- **Surgical/Interventional:**
    - **Device Closure:** Treatment of choice for Secundum ASD if margins are adequate (>5mm).
    - **Surgical Patch:** For Primum, Sinus Venosus types, or Secundum with poor margins.

- **Timing:** Usually elective at **3–5 years of age**, or earlier if the child is symptomatic or has significant growth failure.

EXAMINER'S VIVA

Q1: Why is the murmur in ASD an Ejection Systolic Murmur? *A: It is a flow murmur caused by the increased volume of blood being ejected through a normal-sized pulmonary valve. The flow across the actual ASD is low-velocity and silent.*

Q2: Why is the S2 split "fixed"? *A: Normally, inspiration increases venous return to the RV, delaying P2. In ASD, during inspiration, the decrease in left-to-right shunting compensates for the increase in systemic venous return, keeping the total RV volume and the timing of P2 constant throughout the respiratory cycle.*

Q3: How do you differentiate a Secundum ASD from a Primum ASD on an ECG? *A: Secundum ASD shows Right Axis Deviation. Primum ASD shows **Left Axis Deviation** (due to early activation of the left ventricle and conduction system abnormalities).*

Q4: What is Lutembacher Syndrome? *A: The combination of an acquired Mitral Stenosis with a congenital ASD.*

Q5: Can an ASD cause cyanosis? *A: Only if: 1. Pulmonary hypertension develops leading to a reversal of shunt (Eisenmenger Syndrome), or 2. In the neonatal period if RV compliance is very low, or 3. If there is an associated lesion like Ebstein's anomaly.*

LONG CASE PRESENTATION TIPS

- **The Opening:** "I am presenting [Name], a [Age] year old male/female, who is currently asymptomatic but was referred for an incidental murmur, with a history suggestive of increased pulmonary blood flow in the form of recurrent LRTIs..."
- **Common Mistake:** Don't say "S2 is split." In a PG exam, you must specify: "S2 is wide and fixedly split."
- **What Examiners Watch For:** They will watch your stethoscope placement. If you don't spend significant time at the **pulmonary area** and then move to the **tricuspid area** to look for the flow murmur, they will know you aren't looking for the right signs.

---

10. Patent ductus arteriosus (PDA)

Subject: Cardiovascular

HISTORY

Chief Complaint

- **Infants:** Fast breathing, difficulty in feeding (suck-rest-suck cycle), excessive sweating over the forehead while feeding, and poor weight gain.

- **Older Children:** Often asymptomatic (detected on routine screening), easy fatigability, or recurrent lower respiratory tract infections (LRTIs).
- **Duration:** Usually noted since the first few weeks of life in significant shunts.

History of Present Illness

- **Feeding History (The "Stress Test" of Infancy):** Ask: "Does the baby take a full feed at one go, or does he stop to breathe heavily?" "Does he sweat profusely on the forehead during feeds?" (Indicates sympathetic overactivity and heart failure).
- **Respiratory Symptoms:** "How many times has he been hospitalized for pneumonia?" (PDA increases pulmonary blood flow, making lungs "wet" and prone to infection).
- **Growth:** "Has he outgrown his clothes at the same rate as his peers?" (Failure to thrive is common in large PDAs).
- **Exercise Tolerance:** In older kids, ask: "Can he keep up with friends during play, or does he need to sit down sooner?"
- **Cyanosis/Squatting:** "Have you noticed any blue discoloration of the lips or toes?" (Differential cyanosis is the hallmark of reversed PDA/Eisenmenger).
- **Neurological:** "Has there been any sudden weakness or seizures?" (Rule out infective endocarditis with embolic phenomena).

Relevant Background History

- **Antenatal/Birth:** Ask specifically about **Maternal Rubella** (Gregg's triad: PDA, Cataract, Deafness) and **Prematurity** (the most common cause of functional PDA).
- **Developmental:** Gross motor delay is common due to cardiac failure and poor weight gain.
- **Socioeconomic:** Crowding increases risk of recurrent LRTIs.

EXAMINATION

General Survey

- **Activity:** Is the infant tachypneic but alert, or lethargic (low output)?
- **Nutritional Status:** Look for **Precordial Bulge** (indicates long-standing cardiomegaly in a growing skeleton). Assess for "Wasting" (loss of subcutaneous fat in the axilla and groin).
- **Facies:** Look for "Salt and Pepper" retinopathy or cataracts (Rubella). Look for dysmorphism (Down Syndrome can have PDA, though VSD/AVSD are more common).

Vital Signs and Anthropometry

- **Respiratory Rate:** Count for a full minute while the child is quiet. [SEVERITY MARKER: Tachypnea at rest].

- **Blood Pressure: CRITICAL.** Measure in both upper and lower limbs. Look for **Wide Pulse Pressure** (e.g., 90/30 mmHg). The diastolic pressure drops because of the "runoff" from the aorta into the pulmonary artery.
- **Growth:** Plot Weight-for-Age and Length-for-Age. PDA often causes "Type 2" growth failure (weight affected more than height).

Peripheral Signs

- **Pulse:** Feel the radial and femoral pulses simultaneously. Look for **Water-hammer/Bounding pulses** (Corrigan's pulse). This is due to a large stroke volume and rapid diastolic runoff.
- **Capillary Refill:** Check on the sternum; >3 seconds suggests low systemic output.
- **Differential Cyanosis:** [EXAMINER FAVORITE] Compare the color of the fingers (pink) with the toes (blue/dusky). This indicates Eisenmenger syndrome where desaturated blood from the PA enters the descending aorta via the ductus.
- **Clubbing:** Check toes specifically if differential cyanosis is suspected.

Systemic Examination — Cardiovascular

Inspection

- **Precordial Activity:** Look for a hyperdynamic impulse.
- **Apex Beat:** Usually shifted downwards and outwards (Left Ventricular Volume Overload).

Palpation

- **Apex Beat:** Characterize as **Hyperdynamic** (brisk, forceful, but ill-sustained).
- **Thrills:** Palpate the **Left Second Intercostal Space** (Infraclavicular area). A continuous thrill is pathognomonic for PDA.
- **P2:** Palpate the pulmonary area for a palpable P2 (suggests Pulmonary Hypertension).

Percussion

- Usually not performed in infants, but in older children, you may find an increased area of dullness to the left of the sternum.

Auscultation

- **The Murmur:** [EXAMINER FAVORITE] **Gibson's Murmur.**
    - *Character:* Continuous, "machinery-like" (crescendo-decrescendo in systole, continuing through S2 into diastole).
    - *Location:* Left infraclavicular area and 2nd left intercostal space.
    - *Radiation:* To the back or left clavicle.
- **S2:** Often obscured by the murmur. If heard, P2 may be loud (PAH).

- **Mid-Diastolic Rumble:** Listen at the apex with the bell. This is a "flow murmur" across the mitral valve due to increased pulmonary venous return. [SEVERITY MARKER: Indicates a large Shunt (Qp:Qs > 2:1)].

Secondary Systems

- **Respiratory:** Fine crepitations at the bases (Pulmonary edema/Heart failure).
- **Abdomen:** Hepatomegaly (congestive heart failure). Feel for liver span.

SIGNS THAT CLINCH THE DIAGNOSIS [EXAMINER FAVORITES]

1. **Continuous Machinery Murmur:** Best heard in the left infraclavicular area.
2. **Bounding Peripheral Pulses:** With wide pulse pressure (diastolic "runoff").
3. **Differential Cyanosis/Clubbing:** Pink hands, blue toes (indicates reversed PDA).
4. **Hyperdynamic Apex:** Shifted down and out, indicating LV volume overload.

DIAGNOSIS

Differentials

1. **Aortopulmonary (AP) Window:** Murmur is similar but usually louder and lower down the sternal border; pulses are also bounding.
2. **Ruptured Sinus of Valsalva (RSOV):** Sudden onset, murmur is loudest at the lower sternal border (not infraclavicular).
3. **Venous Hum:** Disappears when the child lies flat or when the jugular vein is compressed (PDA murmur does not).
4. **Truncus Arteriosus:** Continuous murmur may be present due to truncal insufficiency or flow, but the child is usually cyanotic from birth.

Investigations

- **Tier 1: CXR** (Cardiomegaly, increased pulmonary vascular markings, prominent aortic knuckle). **ECG** (LV hypertrophy; look for deep Q waves and tall R waves in V5-V6).
- **Tier 2: Echocardiography (Gold Standard).** Confirms the ductus, measures the smallest diameter, determines shunt direction, and assesses LV/LA dilation.
- **Tier 3: Cardiac Catheterization.** Only if Eisenmenger syndrome is suspected to check for vasoreactivity of pulmonary vessels.

Management Outline

- **Medical:**
    - Infants: Diuretics (Furosemide) and ACE inhibitors for heart failure.

    - Preterm neonates: **Indomethacin or Ibuprofen/Paracetamol** [UPDATED] to close the ductus.

- **Surgical/Interventional:**
    - **Device Closure:** Treatment of choice for most PDAs (Amplatzer Duct Occluder).
    - **Surgical Ligation:** Indicated for very large PDAs or very small infants where device closure is technically impossible.

EXAMINER'S VIVA

1. **Q: Why is the murmur continuous?**
    - A: Because the pressure in the aorta is higher than the pressure in the pulmonary artery during both systole and diastole, maintaining a constant flow across the ductus.
2. **Q: What is "Silent PDA"?**
    - A: A PDA so small it has no audible murmur, detected only on Echo. Usually requires no treatment.
3. **Q: Why does differential cyanosis occur in PDA?**
    - A: When pulmonary hypertension develops (Eisenmenger), the shunt reverses (Right-to-Left). The ductus enters the aorta *after* the origin of the head and neck vessels, so deoxygenated blood goes only to the lower body.
4. **Q: How do you differentiate a PDA murmur from a Venous Hum at the bedside?**
    - A: [TECHNIQUE] Apply gentle pressure over the neck veins or turn the child's head; a venous hum will change or disappear, whereas a PDA murmur remains constant.
5. **Q: What is the significance of a mid-diastolic murmur at the apex in PDA?**
    - A: It signifies a large shunt (high flow across the mitral valve). It is a marker of severity.

LONG CASE PRESENTATION TIPS

- **The Opening:** "I am presenting the case of a [Age] old male, born [Term/Preterm], presenting with features of increased pulmonary blood flow and congestive cardiac failure, currently having a hyperdynamic circulation and a continuous machinery murmur..."
- **Common Mistake:** Forgetting to check the pulses in the lower limbs or failing to compare the color of the fingers and toes.
- **Watch out:** Examiners will watch how you position your stethoscope. For PDA, you *must* auscultate the back (interscapular area) and the infraclavicular area specifically.

---

11. Tetralogy of Fallot

Subject: Cardiovascular

This is a classic "Cyanotic Congenital Heart Disease with Decreased Pulmonary Blood Flow" case. In the exam, the examiner is looking for your ability to differentiate "cyanotic spells" from seizures, your precision in localizing the murmur, and your assessment of the degree of right ventricular outflow tract (RVOT) obstruction.

HISTORY

Chief Complaint

- Bluish discoloration of skin/nails (Cyanosis) since early infancy (usually 2–6 months).
- Paroxysmal episodes of increased bluishness, rapid breathing, and irritability (Cyanotic Spells).
- Difficulty in breathing on exertion.
- Squatting episodes (in older, ambulatory children).

History of Present Illness

- **Cyanosis:** "When did you first notice the blue color? Was it at birth or later? Does it increase during crying or feeding?" (TOF cyanosis is usually not immediate at birth but appears as the ductus closes or the RVOT obstruction progresses).
- **Cyanotic Spells (Hypercyanotic Spells):** "Does the child ever suddenly become very blue, breathe very fast, and then become limp or sleepy? What do you do to stop it?" (Look for the classic sequence: Crying → Hyperpnea → Deep Cyanosis → Syncope/Sleep).
- **Squatting:** "Does your child stop to sit on their haunches while playing?" (Squatting increases systemic vascular resistance, forcing more blood through the lungs).
- **Exercise Tolerance:** "How far can he walk compared to peers? Does he get tired during feeds (suck-rest-suck cycle)?"
- **Complications:** Ask about focal neurological deficits or seizures (Brain abscess/Thrombosis) and fever (Infective Endocarditis).

Relevant Background History

- **Antenatal:** Maternal diabetes or intake of retinoic acid/anti-epileptics (increased risk of Conotruncal defects).
- **Birth:** Birth weight (TOF babies usually have preserved birth weight compared to TGA).
- **Development:** Gross motor delay is common due to hypoxia and exercise intolerance.
- **Family History:** Recurrence risk is ~3%. Ask about DiGeorge syndrome features in family (cleft palate, infections).

EXAMINATION

General Survey

- **Observation:** Note the degree of cyanosis at rest. Is the child comfortable or tachypneic?
- **Posture:** Is the child naturally assuming a knee-chest position on the examination table?
- **Nutritional Status:** Assess for "cardiac cachexia." TOF children are often underweight but less prone to severe congestive heart failure (CHF) than those with VSD/PDA.
- **Facies:** Look for DiGeorge features (hypertelorism, low-set ears, micrognathia).

Vital Signs and Anthropometry

- **Pulse:** Usually normal volume. If bounding, suspect an associated PDA or MAPCAs.
- **Respiratory Rate:** Count for a full minute. [SEVERITY MARKER]: Persistent tachypnea at rest suggests severe hypoxia or an impending spell.
- **Saturation (SpO2):** Measure in all four limbs (to rule out associated arch anomalies).
- **Blood Pressure:** Measure in upper and lower limbs to rule out associated Coarctation (though rare in TOF).

Peripheral Signs

- **Clubbing:** [EXAMINER FAVORITE] Use Schamroth's window test. In TOF, clubbing usually appears after 6 months of age. Grade it (Grade 1: Softening of nail bed; Grade 2: Loss of Lovibond angle; Grade 3: Curvature/Drumsticking; Grade 4: Hypertrophic osteoarthropathy).
- **Cyanosis:** Check tongue, buccal mucosa, and conjunctiva. Central cyanosis in TOF is due to right-to-left shunting across the VSD.
- **JVP:** Usually not elevated in uncomplicated TOF. A prominent 'a' wave suggests a restrictive VSD or severe RV hypertrophy (uncommon as VSD is typically large/non-restrictive).

Systemic Examination — Cardiovascular

Inspection:

- **Precordial Bulge:** May be present over the left parasternal area due to RV hypertrophy.
- **Apical Impulse:** Usually normal position; not displaced (since there is no volume overload of the LV).

Palpation:

- **Apex Beat:** Usually in the 4th/5th intercostal space, mid-clavicular line. Character is "tapping" (suggests RV dominance).
- **Left Parasternal Heave:** [EXAMINER FAVORITE] Use the heel of your hand. Grade it. This confirms Right Ventricular Hypertrophy (RVH).
- **Systolic Thrill:** Palpate at the left 2nd and 3rd intercostal spaces. A thrill indicates the murmur is at least Grade 4/6. Note: In very severe RVOT obstruction (near atresia), the thrill disappears.

Percussion:

- Usually not required in Peds CV exam unless looking for dextrocardia or dullness of a large pericardial effusion.

Auscultation:

- **S1:** Normal.
- **S2:** [EXAMINER FAVORITE] Usually **single and loud**. Why? The pulmonary component (P2) is soft or inaudible due to low pulmonary pressure and the aortic component (A2) is loud because the aorta is anteriorly displaced (overriding).
- **S3/S4:** Usually absent.
- **Murmur:** Ejection Systolic Murmur (ESM) at the left mid-to-upper sternal border.
    - *Origin:* It is NOT from the VSD (the VSD in TOF is large and non-restrictive, so it's silent). The murmur is from the **RVOT obstruction (Pulmonary Stenosis)**.
    - *Inverse Relationship:* [SEVERITY MARKER] The louder and longer the murmur, the milder the obstruction. A short, soft murmur indicates severe obstruction (less blood crossing the narrowed valve). During a "spell," the murmur may disappear entirely.

Secondary Systems

- **Respiratory:** Lungs are usually clear (no signs of congestion/crackles, as pulmonary blood flow is decreased).
- **Abdomen:** Check for hepatomegaly. If present, think of something else (TOF does not typically cause CHF unless there is severe anemia or infective endocarditis).
- **CNS:** Brief screen for focal deficits (rule out paradoxical embolism/abscess).

DIAGNOSIS

Diagnostic Criteria (The Tetrad)

1. Ventricular Septal Defect (Large, subaortic).
2. Right Ventricular Outflow Tract Obstruction (Infundibular and/or Valvular).
3. Overriding of the Aorta (<50%).
4. Right Ventricular Hypertrophy.

Differentials

1. **Transposition of Great Arteries (TGA) with VSD and PS:** Presents similarly; differentiated by Echo.
2. **Tricuspid Atresia:** ECG shows Left Axis Deviation (TOF has Right Axis Deviation).
3. **Single Ventricle with PS:** Differentiated by Echo.

Investigations

- **CXR:** "Boot-shaped heart" (Coeur-en-sabot) due to upturned apex (RVH) and concave pulmonary bay. Lung fields are oligemic (dark).
- **ECG:** Right Axis Deviation (RAD) and RVH (Tall R waves in V1).

- **Echocardiography:** Gold standard. Confirms the four components and measures the "Z-score" of pulmonary arteries.
- **CBC:** Polycythemia (high Hb/Hct) as a compensatory mechanism for chronic hypoxia.

Management Outline

1. **Medical Management of Spells:** Knee-chest position, Oxygen, Morphine (0.1mg/kg), IV fluids, Sodium Bicarbonate (for acidosis), Beta-blockers (Propranolol).
2. **Surgical:**
    - *Palliative:* Modified Blalock-Taussig (mBT) shunt (subclavian artery to pulmonary artery) if the child is too small or anatomy is unfavorable for repair.
    - *Definitive:* Total Correction (VSD closure + RVOT resection/patch) usually performed between 6–12 months of age.

EXAMINER'S VIVA

Q1: Why is the VSD silent in TOF? A: Because the VSD is large and non-restrictive. There is no pressure gradient between the two ventricles; they function as a common pumping chamber.

Q2: Why does the murmur shorten during a cyanotic spell? A: During a spell, RVOT obstruction increases or systemic resistance drops, causing almost all blood to shunt R-to-L through the VSD. Since less blood passes through the pulmonary valve, the ESM becomes softer or disappears.

Q3: How do you differentiate a TOF spell from a breath-holding spell? A: Breath-holding spells are usually preceded by a clear provocative trigger (anger/frustration), occur during expiration, and the child recovers very quickly once breathing resumes. TOF spells are characterized by hyperpnea and prolonged exhaustion/sleep afterward.

Q4: What is the significance of a continuous murmur in a known TOF patient? A: It suggests the presence of MAPCAs (Major Aortopulmonary Collateral Arteries) or a surgical shunt (like a BT shunt).

Q5: Show me how you assess for a Parasternal Heave. A: (Technique) Place the heel of your hand over the left parasternal area (3rd-5th ICS). Keep your fingers lifted. If your hand is lifted with each heartbeat, a heave is present.

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting the case of a [Age] male, born of a non-consanguineous marriage, presenting with central cyanosis since 4 months of age, history of squatting, and recurrent hypercyanotic spells, currently having compensated polycythemia and no signs of congestive failure."
- **Mistake to avoid:** Never say the murmur is a "VSD murmur." Always specify it is an "ESM of RVOT obstruction."
- **Observation:** Examiners watch how you handle the child. If the child cries, use it to your advantage to observe the increase in cyanosis and the single S2.

12. Rheumatic heart disease

Subject: Cardiovascular

This is a classic "bread and butter" long case for the MD Pediatrics practical exam. In RHD, the examiner is not just looking for the diagnosis; they are looking for your ability to assess **hemodynamic significance, activity of the disease, and the presence of congestive cardiac failure (CCF).**

HISTORY

Chief Complaint

- Typically a school-aged child (5–15 years) presenting with:
- Breathlessness (duration, progression)
- Palpitations
- Joint pains/swelling (past or present)
- Precordial bulge or chest pain
- Decreased exercise tolerance

History of Present Illness

Ask these questions naturally to build the "hemodynamic story":

- **Breathlessness:** "Does he get tired before his friends when playing?" (NYHA Class I/II). "Does he struggle to breathe while walking to the bathroom or dressing?" (Class III). "Does he have trouble breathing while lying flat? Does he need extra pillows?" (Orthopnea). "Does he wake up gasping at night?" (PND).
- **Palpitations:** "Does he feel his heart racing or skipping beats?" (Suggests arrhythmia like AF or severe AR/MR).
- **Joint Symptoms:** "Did he have painful, swollen joints that moved from one to another (migratory)?" "Did the pain respond dramatically to aspirin?"
- **Chorea:** "Have you noticed any clumsy movements, change in handwriting, or emotional outbursts?"
- **Fever:** "Is there a low-grade fever?" (Suggests Reactivation or Infective Endocarditis).
- **Negative clusters:** Ask about cyanosis and cyanotic spells to rule out Congenital Heart Disease (CHD). Ask about facial puffiness and decreased urine output to rule out Chronic Kidney Disease with secondary cardiac involvement.

Relevant Background History

- **Past History:** [CRITICAL] Ask about recurrent sore throats (treated or untreated). Ask if the child is already on "the painful injection" (Benzathine Penicillin prophylaxis) and the date of the last dose.

- **Developmental:** Usually normal in RHD (unlike CHD), but chronic hypoxia/failure can cause secondary growth failure.
- **Socioeconomic:** Overcrowding, sleeping arrangements (number of people per room) – RHD is a disease of poverty.

EXAMINATION

General Survey

- **Position:** Is the child propped up? (Orthopnea).
- **Distress:** Look for alae nasi flaring or accessory muscle use.
- **Nutritional Status:** Assess for "Cardiac Cachexia." Measure weight/height; chronic RHD often leads to stunting and wasting.
- **Facies:** Look for "Mitral Facies" (malar flush) – rare in children but seen in severe MS.

Vital Signs and Anthropometry

- **Pulse:**
    - *Technique:* Feel the radial and femoral simultaneously (rule out Coarctation).
    - *Findings:* Tachycardia out of proportion to fever (suggests Carditis). Irregularly irregular (Atrial Fibrillation). Water-hammer pulse (AR).
- **Blood Pressure:**
    - *Technique:* Use the correct cuff size (bladder width 40% of arm circumference).
    - *Findings:* Wide pulse pressure in AR.
- **Respiratory Rate:** Count for a full minute. Look for tachypnea [SEVERITY MARKER].

Peripheral Signs

- **Clubbing:** [EXAMINER FAVORITE] Check Schamroth's window. If present in RHD, think **Infective Endocarditis**.
- **Pallor:** Check palmar creases. Anemia worsens cardiac failure.
- **JVP:**
    - *Technique:* Child at 45 degrees, head turned slightly left. Look for the double flicker of internal jugular pulsation.
    - *Significance:* Elevated JVP indicates right heart failure or severe MS.
- **Rheumatic Stigmata:** Look for Subcutaneous nodules (over bony prominences) and Erythema Marginatum (rare in dark skin).

Systemic Examination — Cardiovascular (The Core)

Inspection

- **Precordial Bulge:** Indicates long-standing cardiomegaly before the rib cage calcified.
- **Apical Impulse:** Locate it. Is it shifted down and out? (Volume overload/LV enlargement).
- **Visible Pulsations:** Look for epigastric pulsations (RV enlargement) or suprasternal pulsations (AR).

Palpation

- **Apex Beat:** Use the ulnar border of your hand first, then fingertips.
    - *Character:* "Heaving" (Pressure overload, e.g., AS) vs. "Hyperdynamic/Ill-sustained" (Volume overload, e.g., MR/AR) vs. "Tapping" (Palpable S1 in MS).
- **Parasternal Heave:** Use the heel of your hand at the left lower sternal border. Indicates RV enlargement.
- **Thrills:** Palpate with the flat of your hand at the apex (systolic thrill = MR) and base.
- **P2:** Palpate the 2nd left intercostal space. If palpable, it signifies Pulmonary Hypertension [SEVERITY MARKER].

Percussion

- Not routinely done, but you may percuss the right heart border to check for dextrocardia or massive cardiomegaly.

Auscultation

- **Mitral Area (Apex):**
    - *Pansystolic murmur:* High pitched, blowing, radiates to axilla (MR).
    - *Mid-diastolic murmur:* Use the **bell**, patient in left lateral position. Listen for the "rumble" of MS.
- **Aortic Area:**
    - *Early Diastolic Murmur (EDM):* Patient sitting up, leaning forward, breath held in expiration. Listen at the 3rd/4th left intercostal space (Erb's point) for AR.
- **Dynamic Auscultation:** If the murmur is faint, ask the child to do 10 squats (if stable) to increase afterload and accentuate MR/AR.

Systemic Examination — Secondary Systems

- **Respiratory:** Fine crepitations at the bases (Pulmonary edema).
- **Abdomen:** Tender hepatomegaly (Right heart failure). Check for splenomegaly (Infective Endocarditis).

SIGNS THAT CLINCH THE DIAGNOSIS [EXAMINER FAVORITES]

- **Carey Coombs Murmur:** A short, mid-diastolic murmur at the apex during acute carditis (due to mitral valve leaflet edema), disappearing as the episode resolves.

- **Tapping Apex Beat:** This is actually a palpable First Heart Sound (S1), pathognomonic for MS.
- **Radiation to Axilla:** Distinguishes the pansystolic murmur of MR from VSD (which radiates all over the precordium/tricuspid area).
- **Gallop Rhythm (S3):** Heard at the apex; indicates heart failure or a very dilated LV.

SEVERITY ASSESSMENT [SEVERITY MARKER]

- **Severe MR:** Presence of S3, mid-diastolic flow rumble, displaced apex, and pulmonary hypertension (loud P2).
- **Severe MS:** Narrowing of the S2-OS (Opening Snap) interval. The closer the OS is to S2, the more severe the MS.
- **CCF Signs:** Gallop, hepatomegaly, basal creps, orthopnea.

DIAGNOSIS

Diagnostic Criteria

- **Revised Jones Criteria (2015):** Differentiates between Low-risk and Moderate/High-risk populations.
    - *Major:* Carditis, Polyarthritis (or Monoarthritis/Polyarthralgia in high-risk), Chorea, Erythema Marginatum, Subcutaneous nodules.
    - *Minor:* Fever, Polyarthralgia, ESR ≥60, Prolonged PR interval.
    - *Evidence of preceding Strep infection:* Rising ASO titer or positive throat culture.

Differentials

1. **Congenital Mitral Valve Prolapse:** Mid-systolic click, non-pansystolic murmur.
2. **VSD:** Harsh pansystolic murmur, maximal at the left lower sternal border, not axilla.
3. **Infective Endocarditis:** Fever, new murmur, splenomegaly, petechiae.

Investigations

- **Tier 1:** ECG (Look for PR interval - 1st degree block; P-mitrale); CXR (Cardiomegaly, "Straightening of the left heart border" in MS).
- **Tier 2: Echocardiography (Gold Standard):** Assess valve morphology (thickening, subvalvular fusion), chamber dimensions, and EF%.
- **Tier 3:** ASO Titer, ESR/CRP (markers of activity).

Management Outline

- **Acute Episode:** Bed rest, Aspirin (high dose for arthritis), Steroids (for severe carditis/CCF).
- **CCF:** Diuretics (Furosemide), ACE inhibitors (for MR/AR), Digoxin.

- **Secondary Prophylaxis:** Inj. Benzathine Penicillin G (1.2 million units) every 3–4 weeks. [UPDATED: 3-weekly is preferred in high-risk areas].
- **Surgical:** Mitral Valve Repair or Replacement; Balloon Mitral Valvotomy (BMV) for MS.

EXAMINER'S VIVA

1. **Q: How do you differentiate the murmur of MR from TR at the bedside?**
    - *A:* Carvallo's Sign: The murmur of TR increases in intensity during inspiration; MR does not.
2. **Q: Why do we use steroids in RHD?**
    - *A:* Only for severe carditis with cardiomegaly or CCF. It doesn't prevent valve damage but reduces acute inflammation and life-threatening failure.
3. **Q: What is the duration of secondary prophylaxis?**
    - *A:* RHD with persistent valvular disease: Until age 40 or lifelong. RHD with resolved carditis: 10 years or until age 21 (whichever is longer).
4. **Q: How do you measure the Liver Span?**
    - *A:* [Technique] Percuss the upper border (usually 5th ICS) and palpate the lower border. Measure the distance in cm. Essential to distinguish "downward displacement" from true "hepatomegaly."
5. **Q: What is the significance of a "silent" chest in a known RHD patient?**
    - *A:* Could indicate a massive pericardial effusion (associated with acute carditis) or very low cardiac output.

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting the case of [Name], a [Age] year old male, resident of [Area], who presented with symptoms suggestive of NYHA Class III cardiac failure, with a past history suggestive of Acute Rheumatic Fever..."
- **Mistake:** Don't forget to check the throat and skin. Examiners love to see you looking for the "source" or "stigmata."
- **Observation:** If you see a scar on the chest, look for it *before* you start palpating. A midline sternotomy vs. a lateral thoracotomy tells you if they've had a valve replacement or a closed mitral valvotomy.

---

13. Infective endocarditis

Subject: Cardiovascular

LONG CASE FRAMEWORK: INFECTIVE ENDOCARDITIS (IE)

HISTORY

Chief Complaint

- Fever (prolonged/unexplained), malaise, weight loss, joint pains, or new-onset neurological deficits.
- In children with known Congenital Heart Disease (CHD): "Worsening of baseline symptoms" (increased cyanosis, decreased exercise tolerance).

History of Present Illness

- **Fever:** Ask, "Is the fever continuous or intermittent? Does it have a specific pattern?" (Usually low-grade and remittent in subacute IE; high-grade/hectic in acute IE).
- **Constitutional Symptoms:** "Has the child lost interest in play? Is there a noticeable loss of weight or appetite?" (Suggests chronicity).
- **Embolic Phenomena:**
    - *CNS:* "Have you noticed any sudden weakness of limbs, facial drooping, or seizures?" (Mycotic aneurysm rupture or embolic stroke).
    - *Renal:* "Is the urine red or smoky in color?" (Glomerulonephritis or renal infarct).
    - *Pulmonary:* "Is there sudden chest pain or coughing up blood?" (Septic pulmonary emboli, common in Right-Sided IE/VSD).
- **Cardiac Failure:** "Is the child breathing faster than usual? Is there difficulty feeding or sweating over the forehead?" (Suggests new valvular regurgitation).
- **Musculoskeletal:** "Are there pains in the small joints or lower back?" (Immunological phenomena).
- **Predisposing Events:** "In the last 2 months, did the child have a dental extraction, professional teeth cleaning, or any surgery/endoscopy?"

Relevant Background History

- **Past History:** Document the underlying cardiac lesion. "Was the child born with a hole in the heart or a blue baby? Has there been a previous cardiac surgery or prosthetic valve placement?"
- **Antenatal/Birth:** Relevant if the CHD was diagnosed in utero or at birth.
- **Immunization:** Specifically ask about Pneumococcal and Hib vaccines (asplenic patients or post-cardiac surgery).
- **Socioeconomic:** Overcrowding and poor dental hygiene are significant risk factors.

EXAMINATION

General Survey

- **Appearance:** Look for the "Cafe-au-lait" complexion (a sallow, brownish-yellow tint seen in chronic IE due to anemia and low-grade hemolysis).
- **Activity:** Is the child toxic/septic (Acute IE - *S. aureus*) or just chronically ill (Subacute IE - *Viridans strep*)?
- **Nutritional Status:** Check for significant wasting (chronic infection).

Vital Signs and Anthropometry

- **Temperature:** Must be recorded. Look for "fever of unknown origin" pattern.
- **Heart Rate:** Tachycardia out of proportion to fever suggests myocarditis or heart failure.
- **Blood Pressure:** Look for wide pulse pressure (if IE causes acute Aortic Regurgitation).
- **Respiratory Rate:** [SEVERITY MARKER] Tachypnea indicates congestive heart failure (CHF).

Peripheral Signs [CRITICAL FOR IE]

- **Hands & Feet:**
    - **Splinter Hemorrhages:** Linear, dark-red streaks under the distal third of the nail. *Technique:* View the nail bed in tangential light.
    - **Osler Nodes:** Small, tender, pea-sized nodules on the pads of fingers/toes. [EXAMINER FAVORITE] *Significance:* Immunological phenomenon.
    - **Janeway Lesions:** Non-tender, erythematous macules on palms/soles. *Significance:* Embolic phenomenon.
    - **Clubbing:** Look for early sponginess of the nail bed. Common in chronic IE or cyanotic CHD.
- **Eyes:**
    - **Roth Spots:** Pale-centered retinal hemorrhages. *Technique:* Fundoscopy is mandatory in a suspected IE case.
    - **Conjunctival Hemorrhages:** Look for small petechiae in the palpebral conjunctiva (flip the lid).
- **Splenomegaly:** Palpate the left hypochondrium. A soft, tender spleen suggests acute IE; a firm spleen suggests chronic.

Systemic Examination — Cardiovascular (Primary)

- **Inspection:** Look for a hyperdynamic precordium or scars from previous cardiac surgeries (thoracotomy/sternotomy).
- **Palpation:**
    - **Apex Beat:** Note shifts (suggesting ventricular dilation from acute valvular insufficiency).
    - **Thrills:** May change or disappear if a vegetation interferes with flow.

- **Auscultation:**
    - **The "Changing Murmur":** [EXAMINER FAVORITE] This is the hallmark. Listen for the appearance of a *new* regurgitant murmur (MR or AR) or a change in the intensity/character of a pre-existing murmur.
    - **Gallop Rhythm (S3):** Indicates heart failure.
    - **Pericardial Rub:** Suggests extension of infection (ring abscess) into the pericardium.

Systemic Examination — Secondary Systems

- **Abdomen:** Check for hepatomegaly (CHF) and splenic tenderness (infarction).
- **CNS:** Detailed focal neurological exam to rule out embolic strokes.
- **Respiratory:** Fine crepitations at bases (Left-sided heart failure) or localized diminished breath sounds (Pulmonary infarct in Right-sided IE).

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **New Valvular Regurgitation:** Especially AR or MR in a child previously known to have only a VSD or ASD.
- **Janeway Lesions vs. Osler Nodes:** Janeway = Embolic/Non-tender; Osler = Immune/Tender.
- **Splenomegaly + Unexplained Fever + CHD:** The classic clinical triad.
- **Microscopic Hematuria:** Use a dipstick at the bedside; suggests immune-complex glomerulonephritis.

DIAGNOSIS

Diagnostic Criteria

- **Modified Duke Criteria (2023 Update):**
    - **Major:** Positive blood cultures (typical organism from 2 sets), Echocardiographic evidence (vegetation, abscess, new valvular regurgitation), or positive PCR/Q-fever serology.
    - **Minor:** Predisposition (heart condition/IV drug use), Fever ≥38.0°C, Vascular phenomena (Janeway, emboli), Immunological phenomena (Osler, Roth spots, RF+), Microbiological evidence not meeting major criteria.
    - **Definite IE:** 2 Major OR 1 Major + 3 Minor OR 5 Minor.

Differentials

1. **Acute Rheumatic Fever:** Migratory polyarthritis, high ESR, but blood cultures are negative and murmurs don't change as rapidly.
2. **Systemic Juvenile Idiopathic Arthritis (sJIA):** Evanescent rash, high spiking fevers, but no valvular vegetations.

3. **Typhoid/Tuberculosis:** Common causes of prolonged fever in tropics; excluded by culture and echo.

Investigations

- **Tier 1:** CBC (leukocytosis, normocytic anemia), ESR/CRP (markedly elevated), Urinalysis (microscopic hematuria).
- **Tier 2: Blood Cultures.** [TECHNIQUE] 3 sets from different peripheral sites; first two within 24 hours. *Do not wait for fever spikes.*
- **Tier 3: Echocardiography.** Transthoracic (TTE) first in children; Transesophageal (TEE) if TTE is inconclusive or if the child has a prosthetic valve.

MANAGEMENT

- **Stabilization:** Treat CHF with diuretics (Furosemide) and oxygen.
- **Antibiotics:**
    - **Empiric:** Vancomycin + Gentamicin (covers Staph, Strep, and Enterococci).
    - **Targeted:** Based on MIC of the organism. Duration: 4–6 weeks of IV therapy.
- **Surgery Indications:** Refractory heart failure, fungal IE, persistent bacteremia >7 days despite antibiotics, large mobile vegetations (>10mm) with embolic risk.
- **Prophylaxis:** [UPDATED] Only for high-risk patients (prosthetic valves, previous IE, unrepaired cyanotic CHD) undergoing dental procedures involving gingival manipulation.

EXAMINER'S VIVA

1. **Q: Why is the "changing murmur" so significant?**
    - A: It indicates acute structural damage—like chordae tendineae rupture or a vegetation obstructing a valve orifice—which is pathognomonic for endocardial involvement.
2. **Q: How do you take blood cultures in a suspected IE case?**
    - A: Take 3 sets (aerobic and anaerobic) from different venipuncture sites. The volume is key (1-3ml in infants, 5-10ml in older children). Strict aseptic technique is mandatory to avoid skin contaminants like *S. epidermidis*.
3. **Q: What is a "Culture-Negative Endocarditis"?**
    - A: IE where standard cultures are negative after 5 days. Causes: Prior antibiotic use (most common), fastidious organisms (HACEK group), or intracellular organisms (*Coxiella, Bartonella*).
4. **Q: Explain the pathophysiology of Osler nodes.**
    - A: They are caused by the deposition of immune complexes in the dermal vessels (Type III Hypersensitivity), leading to localized inflammation and pain.

5. **Q: Which CHD is most prone to IE?**
    - A: VSD, TOF, and PDA. Note: Secundum ASD is rarely associated with IE due to low-pressure gradient.

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting the case of a [Age] year old [Gender], a known case of [Specific CHD], who now presents with a prolonged febrile illness of [Duration] associated with new-onset respiratory distress and peripheral embolic markers..."
- **Mistake:** Forgetting to check the urine for hematuria. It is a "bedside" test that counts as a Duke's minor criterion.
- **Watch-out:** Examiners will watch your hand-washing and aseptic technique if you are asked to demonstrate how to draw a blood culture. They also watch if you check for splinter hemorrhages specifically under the nails.

Respiratory

14. Bronchial asthma

Subject: Respiratory

This is a classic long case. In the exam, the child is often in a stable phase or a mild exacerbation. Your goal is to prove three things: **Reversibility, Recurrence, and Atopy.**

HISTORY

Chief Complaint

- "Difficulty in breathing," "Cough," or "Whistling sound from chest."
- **Note the duration:** Is it an acute-on-chronic presentation?

History of Present Illness

Ask these exact questions to build the "Asthma Phenotype":

- **The Cough:** "Does the cough get worse at night or in the early morning hours?" (Diurnal variation is key). "Is it triggered by running, laughing, or crying?" (Exercise-induced bronchospasm).
- **The Wheeze:** "Do you hear a whistling sound when the child breathes out?" (Confirm it is expiratory).
- **Triggers:** "Does a change in weather, dust, strong smells, or a common cold trigger these episodes?"

- **Interval Symptoms:** "Between these major attacks, does the child have a persistent cough or get tired easily during play?" (Indicates poor control).
- **Severity/ED visits:** "How many times in the last year did you have to rush to the ER for a nebulization?" "Has the child ever been admitted to the ICU or needed a 'machine' (ventilator) to breathe?" [SEVERITY MARKER]

Relevant Background History

- **Past History:** Look for the "Allergic March." Ask about recurrent itchy skin rashes in infancy (Atopic Dermatitis) or frequent sneezing/runny nose (Allergic Rhinitis).
- **Birth History:** Was the child premature? (Differential: Bronchopulmonary Dysplasia).
- **Family History:** "Does anyone in the immediate family use an inhaler or have 'skin allergies'?" (Strong genetic link).
- **Socioeconomic/Environmental:** "Is there a smoker in the house?" "Do you use incense sticks (agarbatti) or mosquito coils?" "Are there pets or carpets in the bedroom?"
- **Drug History:** Ask specifically about the *technique* of inhaler use. "Show me how the child takes the medicine." (Most common cause of 'refractory' asthma).

EXAMINATION

General Survey

- **Position:** Is the child sitting up and leaning forward (tripod position)? This indicates significant respiratory distress.
- **Speech:** Can the child speak in full sentences, or only short phrases/words? [SEVERITY MARKER]
- **Mental Status:** Is the child agitated (hypoxia) or drowsy (hypercapnia)?
- **Skin:** Look at the flexural surfaces (elbows, knees) for lichenification or eczema.

Vital Signs and Anthropometry

- **Respiratory Rate:** Count for a full minute. Look for tachypnea relative to age.
- **Heart Rate:** Tachycardia may be due to distress or side effects of Beta-2 agonists (Salbutamol).
- **Pulsus Paradoxus:** [EXAMINER FAVORITE] Feel the radial pulse. Does it weaken significantly during inspiration? (A drop in SBP >10-20 mmHg indicates severe airway obstruction).
- **Growth:** Plot height/weight. Chronic steroid use or poorly controlled chronic hypoxia can lead to growth stunting.

Peripheral Signs

- **Clubbing:** Usually *absent* in asthma. If present, think Cystic Fibrosis, Bronchiectasis, or Congenital Heart Disease.

- **Nasal Signs:** Look for a "transverse nasal crease" (from the 'allergic salute') and pale, boggy nasal turbinates.
- **Chest Deformity:** In chronic asthma, look for a "Barrel Chest" (increased AP diameter) or "Harrison's Sulcus" (a groove at the insertion of the diaphragm due to chronic tugging).

Systemic Examination — Respiratory System

Inspection

- **Shape:** Check for increased AP diameter.
- **Work of Breathing:** Look for suprasternal, intercostal, and subcostal retractions.
- **Symmetry:** Asthma is a generalized disease; asymmetric chest expansion suggests a complication like pneumothorax or collapse.

Palpation

- **Trachea:** Usually midline. Displacement suggests a complication.
- **Chest Expansion:** Often reduced bilaterally in an acute attack.
- **Vocal Fremitus:** Usually normal or decreased (due to hyperinflation).

Percussion

- **Note:** Hyper-resonant note throughout both lung fields.
- **Liver Dullness:** The upper border of liver dullness may be pushed down (below the 6th intercostal space) due to hyperinflated lungs.
- **Cardiac Dullness:** May be obliterated.

Auscultation

- **Breath Sounds:** Vesicular with **prolonged expiration**.
- **Adventitious Sounds:**
    - **Wheeze:** High-pitched, polyphonic, musical sounds. Note if they are only end-expiratory (mild) or throughout expiration (moderate).
    - **Silent Chest:** [SEVERITY MARKER] If you see severe retractions but hear no wheeze, the airway is so tight there is no air movement. This is a medical emergency.
- **Vocal Resonance:** Decreased bilaterally.

Secondary Systems

- **CVS:** Look for signs of Cor Pulmonale (loud P2, raised JVP) – rare in asthma, suggests alternative diagnosis or very chronic severe disease.

SIGNS THAT CLINCH THE DIAGNOSIS [EXAMINER FAVORITES]

1. **Polyphonic Expiratory Wheeze:** Indicates widespread narrowing of different-sized airways.

2. **Reversibility:** Documented improvement in wheeze or Peak Expiratory Flow Rate (PEFR) after bronchodilator inhalation.
3. **Hyper-resonant percussion note:** Confirms air trapping/hyperinflation.
4. **Allergic Shiners/Crease:** Periorbital darkening and nasal crease pointing to an atopic phenotype.

DIAGNOSIS

Diagnostic Criteria

- **GINA Guidelines (Global Initiative for Asthma):** Based on the pattern of respiratory symptoms (wheeze, shortness of breath, chest tightness, cough) that vary over time and in intensity, together with documented expiratory airflow limitation.

Differentials

1. **Foreign Body Aspiration:** Sudden onset, unilateral wheeze/diminished air entry.
2. **Tropical Pulmonary Eosinophilia (TPE):** Persistent cough, high absolute eosinophil count, weight loss.
3. **Cystic Fibrosis:** Failure to thrive, malabsorptive stools, digital clubbing.
4. **Vascular Ring:** Stridor/wheeze since early infancy, often positional.

Investigations

- **Tier 1:** Chest X-ray (to rule out pneumonia/pneumothorax; shows hyperinflation in asthma), Pulse Oximetry.
- **Tier 2: Spirometry (Gold Standard for >5-6 years):** Shows obstructive pattern (FEV1/FVC < 0.8) with >12% reversibility after Salbutamol.
- **Tier 3:** Skin Prick Testing (for triggers), Absolute Eosinophil Count (AEC), Serum IgE.

Management Outline

1. **Acute Attack:** Oxygen (target 94-98%), SABA (Salbutamol) via MDI+Spacer or Nebulizer, systemic corticosteroids (Prednisolone 1-2 mg/kg).
2. **Maintenance:** Stepwise approach (GINA).
    - **Step 1:** As needed SABA or low dose ICS-formoterol.
    - **Step 2:** Daily low-dose Inhaled Corticosteroids (ICS).
3. **Education:** Inhaler technique check, Asthma Action Plan, trigger avoidance.

EXAMINER'S VIVA

- **Q: Why use a spacer with an MDI?**

    - A: It reduces the need for hand-breath coordination, decreases oropharyngeal deposition (reducing thrush), and increases lung delivery of the drug.

- **Q: How do you differentiate Asthma from Wheeze Associated Control Infection (WACI) in a toddler?**
    - A: Use the **Modified Asthma Predictive Index (mAPI)**. Positive if frequent wheezing + 1 major criteria (parental asthma/eczema) or 2 minor criteria (allergic rhinitis/eosinophilia/wheezing unrelated to colds).
- **Q: What is the 'Silent Chest' and why is it ominous?**
    - A: It occurs when airflow is insufficient to generate a wheeze. It indicates impending respiratory failure.
- **Q: How do you perform a PEFR measurement?**
    - A: [Technique] Child standing, slide marker to zero, deep inspiration, wrap lips tight around mouthpiece, blow out as hard and fast as possible ("like blowing out birthday candles"). Take best of three.

LONG CASE PRESENTATION TIPS

- **The Opening:** "I am presenting the case of [Name], a [Age] year old male, a known case of episodic breathlessness and wheezing for 2 years, now presenting with an acute exacerbation triggered by a viral prodrome."
- **Mistake to Avoid:** Don't just say "Wheeze." Specify that it is **expiratory and polyphonic**.
- **The "Social" Angle:** Examiners love to ask about school absenteeism. Always mention how many school days were missed in the last month to justify the "Severity/Control" classification.

---

15. Bronchiectasis

Subject: Respiratory

LONG CASE FRAMEWORK: BRONCHIECTASIS

HISTORY

Chief Complaint

- **Chronic Cough:** Usually >4 weeks, productive of large volumes of sputum (often worse in the morning or on changing posture).
- **Recurrent Pneumonias:** Multiple episodes of fever, cough, and respiratory distress requiring antibiotics.
- **Hemoptysis:** Ranging from blood-streaked sputum to massive bleed.
- **Shortness of Breath:** Initially on exertion, later at rest.

History of Present Illness

- **Characterizing the Cough:** "When your child coughs, is it dry or wet? Does it sound 'rattly'?" "How much sputum is produced in a day? (Estimate in teaspoons/cups)." "What color is it? Does it ever smell foul?" (Foul smell suggests anaerobes).
- **Postural Variation:** "Does the cough worsen when the child lies down or turns to one side?" (Suggests drainage from a specific bronchiectatic lobe).
- **Hemoptysis:** "Have you seen streaks of blood or frank clots?" (Differentiate from hematemesis: bright red, frothy, alkaline).
- **Exacerbations:** "How often does the child need antibiotics? How many hospitalizations in the last year?"
- **Associated Sinus Symptoms:** "Does the child have a persistent runny nose or headache?" (Points toward Primary Ciliary Dyskinesia - PCD or Cystic Fibrosis - CF).
- **Systemic Features:** "Is there significant weight loss or night sweats?" (Think TB). "Are there bulky, oily, foul-smelling stools?" (Points toward CF).

Relevant Background History

- **Past History:** Ask specifically about a severe episode of measles, pertussis, or adenovirus in early childhood (Post-infectious bronchiectasis). Ask about foreign body inhalation (localized bronchiectasis).
- **Antenatal/Birth:** Meconium ileus or delayed passage of meconium (>48 hours) is a huge red flag for CF.
- **Immunization:** Specifically check for Measles and Pertussis (DTP) coverage.
- **Family History:** Consanguinity (Autosomal Recessive conditions like CF or PCD). Sibling deaths with similar symptoms.
- **Socioeconomic:** Overcrowding and exposure to biomass fuel (risk for TB).

EXAMINATION

General Survey

- **Observation:** Is the child thin, stunted, or barrel-chested? Look for the "suppurative facies" (pale, puffy, chronically ill).
- **Nutritional Status:** [SEVERITY MARKER] Assess for visible wasting of gluteal and axillary folds. Bronchiectasis is a "consuming" disease; poor weight gain indicates high inflammatory load or malabsorption (CF).
- **Skin:** Look for BCG scar. If absent, ask why.

Vital Signs and Anthropometry

- **Respiratory Rate:** Count for a full minute. Look for tachypnea at rest.

- **Oxygen Saturation:** Check on room air. [SEVERITY MARKER] SpO2 <94% indicates significant V/Q mismatch.
- **Growth:** Plot Height-for-Age and Weight-for-Age. Chronic suppurative lung disease often leads to **Stunting** (chronic malnutrition).

Peripheral Signs

- **Clubbing:** [EXAMINER FAVORITE]
    - **Technique:** Look for loss of the Schamroth window. Measure the phalangeal depth ratio (DPD/IPD >1).
    - **Significance:** Grade it (1-4). Grade 3 (Drumsticking) or 4 (Hypertrophic Osteoarthropathy with wrist pain) suggests long-standing suppuration or hypoxia.
- **Pallor:** Check palmar creases. Chronic inflammation leads to "Anemia of Chronic Disease."
- **Lymph Nodes:** Check cervical and axillary stations. Significant lymphadenopathy suggests TB or Sarcoidosis.
- **Nasal Polyps:** Use a torch to look for glistening, grape-like masses in the nostrils (Common in CF).

Systemic Examination — Respiratory System

Inspection:

- **Shape of Chest:** Look for **Pectus Carinatum** or increased AP diameter (Barrel chest) suggesting air trapping.
- **Symmetry:** Look for unilateral flattening or crowding of ribs (suggests collapse/fibrosis associated with bronchiectasis).
- **Movements:** Check for intercostal and subcostal retractions.

Palpation:

- **Trachea:** [CRITICAL] Feel for tracheal shift. In localized bronchiectasis with collapse/fibrosis, the trachea shifts *toward* the lesion.
- **Chest Expansion:** Measure with a tape at the nipple level. Reduced expansion is common.
- **Vocal Fremitus:** Increased over areas of bronchiectasis (due to surrounding consolidation) but may be decreased if the bronchus is plugged with mucus.

Percussion:

- **Technique:** Percuss symmetrically.
- **Findings:** Usually **Dull** over the affected area (due to secretions/fibrosis). **Hyper-resonant** if there is compensatory emphysema in other lobes.
- **Traube’s Space:** Percuss to ensure no splenomegaly (Situs inversus in Kartagener's).

Auscultation:

- **Breath Sounds:** Often Bronchial or Broncho-vesicular in the affected area.
- **Added Sounds:** [EXAMINER FAVORITE] **Coarse, leathery crepitations** that are **mid-inspiratory and expiratory**.
- **Technique:** Ask the child to cough. If the crepitations change or disappear after a vigorous cough, it confirms they are due to secretions in the dilated bronchi.
- **Wheeze:** May be present due to airway hyper-reactivity (common in CF).

Systemic Examination — Secondary Systems

- **Abdomen:** Palpate for the Liver on the *left* and Spleen on the *right* (Situs Inversus). Palpate for hepatomegaly (Cor Pulmonale).
- **CVS:** Look for a loud P2 and left parasternal heave (Signs of Pulmonary Hypertension).

SIGNS THAT CLINCH THE DIAGNOSIS [EXAMINER FAVORITES]

1. **Persistent Coarse Crepitations:** That persist across different examinations and change with coughing/postural drainage.
2. **Grade 3 Clubbing:** In a child with chronic wet cough, this is the hallmark of suppurative lung disease.
3. **Dextrocardia/Situs Inversus:** Heart sounds loudest on the right, liver on the left. Clinches **Kartagener Syndrome** (part of PCD).
4. **Halitosis:** Foul-smelling breath specifically during coughing episodes.

DIAGNOSIS

Diagnostic Criteria

- **Clinical:** Chronic wet cough >4 weeks + suggestive Chest X-ray/CT.
- **Radiological:** HRCT is the Gold Standard.

Differentials

1. **Cystic Fibrosis:** Look for failure to thrive, oily stools, and pansinusitis.
2. **Primary Ciliary Dyskinesia:** Look for situs inversus, chronic otitis media, and neonatal respiratory distress.
3. **Post-Tubercular Bronchiectasis:** History of contact, positive Mantoux, localized to upper lobes.
4. **Foreign Body Aspiration:** Sudden onset, localized monophonic wheeze, unilateral signs.

Investigations

- **Tier 1:** Chest X-ray (Look for "Tram-track" shadows, "Ring" shadows, or "Gloved-finger" appearance). Sputum culture and sensitivity. Mantoux test.

- **Tier 2: HRCT Chest** (Signet ring sign: internal diameter of bronchus > adjacent pulmonary artery). Sweat Chloride test (>60 mmol/L is positive).
- **Tier 3:** Nasal Nitric Oxide (low in PCD), Genetic testing for CFTR mutations, Immunoglobulin levels (IgG, IgA, IgM) to rule out hypogammaglobulinemia.

Management Outline

1. **Airway Clearance:** The cornerstone. Chest physiotherapy (percussion, vibration) + Active Cycle of Breathing Techniques (ACBT).
2. **Antibiotics:** For exacerbations (based on sputum culture). Long-term Azithromycin (anti-inflammatory/immunomodulatory) is often used.
3. **Nutrition:** High-calorie, high-protein diet. Fat-soluble vitamin supplementation (A, D, E, K) if CF.
4. **Surgery:** Lobectomy only if disease is strictly localized and medical management fails.

EXAMINER'S VIVA

1. **Q: What is the "Signet Ring Sign" on CT?**
    - A: It is seen when the internal diameter of the bronchus is larger than its accompanying pulmonary artery. Normally, the artery is slightly larger.
2. **Q: Why do we ask the patient to cough during auscultation?**
    - A: To see if the crepitations are "fixed" or "mobile." In bronchiectasis, coughing moves the secretions, which may temporarily clear or change the character of the crepitations.
3. **Q: What are the components of Kartagener Syndrome?**
    - A: Bronchiectasis, Situs Inversus, and Chronic Sinusitis.
4. **Q: How do you define a "Massive Hemoptysis" in a child?**
    - A: Usually >240 ml in 24 hours or >8 ml/kg/day. It is a life-threatening emergency.
5. **Q: What is the role of inhaled hypertonic saline?**
    - A: It acts as a mucolytic by hydrating the airway surface liquid, making it easier to expectorate sputum.

LONG CASE PRESENTATION TIPS

- **Opening Statement:** "I am presenting the case of a [Age] year old male/female, with a history of chronic productive cough and recurrent respiratory infections for [Duration], currently having clinical evidence of localized/generalized bronchiectasis with/without features of Cor Pulmonale."
- **Common Mistake:** Forgetting to check for Situs Inversus. If you miss Dextrocardia in a bronchiectasis case, it’s a major fail.

- **What Examiners Watch For:** They watch your **percussion technique** (ensure your pleximeter finger is firm and you percuss from the wrist) and how you **position the child** for auscultation of the lung bases (where bronchiectasis is most common).

---

16. Pulmonary tuberculosis

Subject: Respiratory

This is a classic long case. In the exam, the challenge with Pediatric Pulmonary TB (PTB) is that the signs are often subtle compared to adults. You aren't just looking for a cavity; you are looking for a story of chronic infection, nutritional failure, and focal lung pathology.

HISTORY

Chief Complaint

- **Cough:** Usually >2 weeks, non-remitting.
- **Fever:** Low-grade, evening rise, >2 weeks.
- **Weight loss:** Or failure to gain weight (check the growth card).
- **Decreased activity:** "Not playing as much as before."

History of Present Illness

Ask these questions naturally:

- **The Cough:** "Is the cough getting better with the usual syrups, or is it staying the same/getting worse?" (TB cough is unremitting). "Does he cough up blood?" (Hemoptysis is rare in children but suggests cavitary disease).
- **The Fever:** "Does the fever come only in the evening? Does he wake up drenched in sweat even when the fan is on?" (Classic night sweats).
- **Weight:** "Has he outgrown his clothes in the last 3 months? When was the last time he gained weight?" [SEVERITY MARKER]
- **Contact History:** "Is there anyone in the house, or a frequent visitor/neighbor, who has a chronic cough, is taking 'red capsules' (Rifampicin), or died of a lung problem recently?" (Crucial for the "Source Case").
- **Focal Signs:** "Any swellings in the neck? Any headaches or vomiting?" (To rule out Disseminated/Miliary/Meningitis).

Relevant Background History

- **Past History:** History of measles or pertussis in the last 6 months? (These are potent immunosuppressants that "wake up" latent TB).
- **Birth/Antenatal:** Only if neonatal TB is suspected (maternal history).

- **Immunization:** "Was the BCG vaccine given? Can I see the scar?" (BCG protects against severe forms like TB Meningitis/Miliary, but not necessarily PTB).
- **Socioeconomic:** Overcrowding (number of people per room) and ventilation. This is a social disease.

EXAMINATION

General Survey

- **Observation:** Does the child look "toxic" (acute) or "chronically ill/wasted" (TB)? Look for the "TB Facies"—sunken eyes, prominent cheekbones, and a bright-eyed but exhausted look.
- **Nutritional Status:** Check for visible wasting of gluteal muscles and loss of buccal fat pads. TB is a catabolic state.
- **Skin:** Look for **Erythema Nodosum** (tender, reddish nodules on shins)—a hypersensitivity reaction to TB. Look for **Phlyctenular conjunctivitis** in the eyes.

Vital Signs and Anthropometry

- **Temperature:** Record it yourself; don't rely on history.
- **Respiratory Rate:** Count for a full minute. Tachypnea without significant distress is common in miliary TB.
- **Growth Parameters:** [CRITICAL] Plot Weight-for-Age and Height-for-Age. A "flat" growth curve for 3 months in an endemic area is TB until proven otherwise.

Peripheral Signs

- **BCG Scar:** Check the left deltoid. If absent, note it. If present, it doesn't rule out TB.
- **Clubbing:** Look for early clubbing (loss of Schamroth's window). Significant clubbing in TB suggests chronic lung destruction or bronchiectasis.
- **Lymph Nodes:** [EXAMINER FAVORITE] Examine the cervical, axillary, and epitrochlear nodes. Look for **matted, non-tender nodes**. If you find a discharging sinus, it's a "Scrofuloderma."
- **Anemia:** Common due to the "anemia of chronic disease."

Systemic Examination — Respiratory System

Position: Sitting up for back examination, supine for front.

1. Inspection:

- **Chest Deformity:** Look for flattening of the chest wall on the affected side (suggests volume loss/fibrosis).
- **Movement:** Watch from the foot end. Is one side lagging?
- **Apical Impulse:** Is it shifted? (Suggests collapse/fibrosis pulling it, or effusion pushing it).

2. Palpation:

- **Trachea:** Use your index finger to feel the gap between the trachea and the sternocleidomastoid on both sides. Shifted trachea = significant volume change.
- **Chest Expansion:** Measure with a tape at the nipple level. In older children, use your hands to feel the "bucket-handle" movement.
- **Vocal Fremitus:** Use the ulnar border of your hand. Increased fremitus = Consolidation; Decreased = Effusion/Thickened pleura.

3. Percussion:

- **Technique:** Middle finger of the left hand firmly on the intercostal space; strike with the right middle finger.
- **Findings:**
    - **Dullness:** Over a lobe (consolidation/collapse) or "stony dull" (effusion).
    - **Traube's Space:** Percuss the 6th rib in the left mid-axillary line. If dull, check for splenomegaly or left-sided pleural effusion.

4. Auscultation:

- **Breath Sounds:**
    - **Bronchial Breathing:** High-pitched, tubular. If heard in the periphery, it suggests a cavity or consolidation with a patent airway.
    - **Diminished Breath Sounds:** Common in effusion or collapse.
- **Adventitious Sounds:**
    - **Post-tussive Crepitations:** [EXAMINER FAVORITE] Ask the child to cough, then immediately inhale. Crepitations that appear or become louder after a cough are classic for TB.

Secondary Systems

- **Abdomen:** Palpate for hepatosplenomegaly (miliary TB) or "doughy abdomen" (TB peritonitis).
- **CNS:** Check for neck stiffness (Meningitis).
- **Spine:** Run your fingers down the vertebrae looking for a "gibbus" or tenderness (Pott's spine).

SIGNS THAT CLINCH THE DIAGNOSIS [EXAMINER FAVORITES]

1. **Matted Lymphadenopathy:** Non-tender, cervical nodes that feel stuck together.
2. **Post-tussive Crepitations:** Indicates the presence of secretions in small airways/cavities that clear or shift with coughing.
3. **Phlyctenular Conjunctivitis:** Small grey-yellow nodules at the limbus; a hypersensitivity sign.
4. **Stony Dullness on Percussion:** Pathognomonic for pleural effusion, often tubercular in endemic areas.

DIAGNOSIS

Diagnostic Criteria

- **IAP/NTEP Algorithm:** Based on persistent cough (>2 weeks), fever (>2 weeks), weight loss, and contact with a TB case.
- **UpToDate/WHO:** Focuses on "Bacteriological confirmation" vs. "Clinical Diagnosis."

Differentials

1. **Persistent Post-Infectious Pneumonia:** Usually follows an acute episode; fails to resolve but child isn't "wasting."
2. **Foreign Body Aspiration:** Sudden onset, localized wheeze (monophonic), no prodrome of fever/weight loss.
3. **HIV-associated Lung Disease (LIP):** Similar presentation; check for generalized lymphadenopathy and parotid swelling.

Investigations

- **Tier 1:**
    - **Chest X-ray (PA view):** Look for hilar lymphadenopathy (the hallmark of pediatric TB), Ghon complex, or miliary mottling.
    - **Mantoux Test:** 0.1ml of 5TU PPD intradermally. Read at 48–72 hours. **>10mm is positive** (>5mm if HIV+ or severely malnourished).
- **Tier 2:**
    - **CBNAAT (GeneXpert):** [UPDATED] This is now the first-line test. Use Gastric Aspirate (in young children who swallow sputum) or Induced Sputum. It detects *M. tuberculosis* and Rifampicin resistance.
- **Tier 3:**
    - **CT Chest:** If X-ray is inconclusive but suspicion is high (shows "tree-in-bud" appearance or necrotic nodes).

MANAGEMENT OUTLINE

- **Notification:** Mandatory reporting to the national portal (Nikshay).
- **Intensive Phase (2 months):** HRZE (Isoniazid, Rifampicin, Pyrazinamide, Ethambutol).
- **Continuation Phase (4 months):** HRE.
- **Nutrition:** High protein, high calorie.
- **Pyridoxine (Vit B6):** To prevent Isoniazid-induced peripheral neuropathy.

EXAMINER'S VIVA

1. **Q: How do you collect a Gastric Aspirate?**
    - *A:* Done early morning after 6-8 hours of fasting. Pass an NG tube, aspirate contents. If <5ml, instill 30ml sterile saline and re-aspirate. Neutralize with Sodium Bicarbonate if transport is delayed.
2. **Q: What is a Ghon Complex?**
    - *A:* A combination of a subpleural parenchymal lesion (Ghon focus), lymphangitis, and the draining hilar lymph node.
3. **Q: Why is Ethambutol now used in all pediatric cases?**
    - *A:* [UPDATED] Previously avoided due to optic neuritis risk, but now included in the 4-drug regimen (NTEP) because the risk is low at 15-25mg/kg and it prevents resistance.
4. **Q: How do you differentiate a Mantoux reaction from a BCG scar effect?**
    - *A:* Reactions >15mm or those with blistering/necrosis are almost always due to TB infection, not the BCG vaccine.

LONG CASE PRESENTATION TIPS

- **The Opening:** "I am presenting 8-year-old [Name], who presents with a triad of unremitting cough, evening rise of fever, and significant weight loss over 3 weeks, with a positive contact history from his father."
- **Common Mistake:** Forgetting to check the spine or the abdomen. TB is a systemic disease.
- **Technique:** When performing Mantoux, ensure you mention you are measuring **induration** (the hard part), not the erythema (the red part). Use a ruler across the forearm.

---

17. Pleural effusion

Subject: Respiratory

LONG CASE FRAMEWORK: PLEURAL EFFUSION

HISTORY

Chief Complaint

- **Cough:** Duration and nature (usually dry/hacking in early stages).
- **Chest Pain:** "Does it hurt more when you take a deep breath or cough?" (Pleuritic pain).
- **Breathlessness:** "Is he breathing faster than usual? Does he struggle to speak in full sentences?"
- **Fever:** Duration and grade (High grade/chills point toward Empyema).

History of Present Illness

- **Onset of Dyspnea:** "Did the breathing difficulty start suddenly or gradually?" (Sudden onset with pain suggests pneumothorax; gradual suggests effusion).
- **Positioning:** "Does the child prefer lying on one side?" (Children with large effusions often lie on the **affected side** to allow the healthy lung maximum expansion—*Trepopnea*).
- **Character of Pain:** "Where exactly is the pain? Does it radiate to the shoulder or abdomen?" (Diaphragmatic pleurisy can radiate to the shoulder via phrenic nerve).
- **Constitutional Symptoms:** "Has there been weight loss, night sweats, or contact with a chronic cougher?" (Points toward TB).
- **Symptom Clusters:**
    - **Pneumonic:** High fever, productive cough, followed by worsening dyspnea (Parapneumonic effusion/Empyema).
    - **Tubercular:** Low-grade evening pyrexia, weight loss, anorexia, often adolescent age group.
    - **Congestive/Systemic:** Swelling of feet, puffiness of face, decreased urine output (Suggests transudate: Heart failure, Nephrotic syndrome, or Malnutrition).

Relevant Background History

- **Past History:** Previous episodes of pneumonia, asthma, or known cardiac/renal disease.
- **Contact History:** [CRITICAL] Ask specifically about household contacts with TB (adults with chronic cough).
- **Immunization:** BCG scar presence; Hib and Pneumococcal vaccination status (reduces likelihood of bacterial empyema).
- **Socioeconomic:** Overcrowding and poor ventilation (TB risk).

EXAMINATION

General Survey

- **Position:** Note if the child is propped up or lying on the affected side.
- **Respiratory Distress:** Look for nasal flaring, grunting, and use of accessory muscles (sternocleidomastoid, scalene).
- **Nutrition:** Assess for visible wasting (TB/Malignancy) or generalized edema (Nephrotic/CCF).

Vital Signs and Anthropometry

- **Respiratory Rate:** Count for a full minute. [SEVERITY MARKER]: Tachypnea out of proportion to fever suggests significant lung/pleural involvement.
- **SpO2:** Measure on room air. <94% indicates significant compromise.
- **Temperature:** Persistent high-grade fever despite 48h of antibiotics suggests Empyema.

Peripheral Signs

- **Clubbing:** [EXAMINER FAVORITE] Look for loss of Schamroth's window. Grade 2+ clubbing in a respiratory case suggests **Chronic Suppurative Lung Disease** (Bronchiectasis, Lung Abscess, or Chronic Empyema).
- **Tracheal Position:** [CRITICAL] Use the index and ring finger on the sternoclavicular joints and the middle finger to palpate the trachea. **Shift to the opposite side** is the hallmark of a large effusion.
- **Lymph Nodes:** Check the **Supraclavicular (Virchow's)** and Axillary nodes. Matted nodes suggest TB; stony hard nodes suggest malignancy.

Systemic Examination — Respiratory System

Inspection

- **Shape of Chest:** Look for fullness or bulging of intercostal spaces on the affected side.
- **Movements:** "Watch the chest from the foot of the bed." You will see **diminished respiratory excursions** on the affected side.
- **Apical Impulse:** Look for the pulsation. It may be shifted laterally (away from the effusion).

Palpation

- **Confirm Inspection:** Confirm the diminished movement by placing your hands on the chest wall (Palpatory expansion).
- **Trachea:** Confirm shift. If the trachea is central but there is dullness, think of collapse or a small effusion.
- **Vocal Fremitus:** Use the ulnar border of your hand. Ask the child to say "99" or "Ek Do Teen." **Significantly decreased or absent** over the effusion.
- **Apex Beat:** Palpate the exact site. Shifted away from the side of dullness confirms a space-occupying lesion (Effusion/Tension Pneumothorax).

Percussion

- **Technique:** Percuss the intercostal spaces, not the ribs. Compare side to side.
- **Note: Stony Dullness** [EXAMINER FAVORITE]. This is the most characteristic sign of pleural effusion. It feels like percussing a stone or a wall.
- **Upper Border:** In a moderate effusion, the upper limit of dullness is highest in the axilla (Ellis S-shaped curve).
- **Shifted Dullness:** Not usually performed in the chest unless you suspect a hydropneumothorax (fluid + air).

Auscultation

- **Breath Sounds:** Characteristically **diminished or absent** over the area of dullness.

- **Upper Border:** Just above the fluid level, you may hear **Bronchial Breath Sounds** (due to compression of the lung) and **Egophony** (nasal 'bleating' quality when the child says "Eeee").
- **Vocal Resonance:** Decreased over the fluid.
- **Friction Rub:** May be heard in the early stage (pleurisy) or during the resolution phase. It sounds like creaking leather.

Secondary Systems

- **CVS:** Look for signs of heart failure (S3, gallop, hepatomegaly).
- **Abdomen:** Check for **Hepatomegaly** (displaced downward by a right-sided effusion or due to CCF) and **Ascites** (Polyserositis).

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

1. **Stony Dull Percussion Note:** The most reliable sign distinguishing effusion from consolidation (which is 'dull' but not 'stony').
2. **Tracheal and Mediastinal Shift:** Confirms the presence of a "plus" lesion (pushing the structures away).
3. **Absent Vocal Fremitus:** Distinguishes effusion from consolidation (where fremitus is increased).
4. **Auscultatory Silence:** The "Silent Chest" over the dull area.

Severity Assessment [SEVERITY MARKER]

- **Severe:** Tracheal shift to the opposite side, respiratory rate >60 (infant) or >40 (older child), SpO2 <92%, paradoxical breathing, or cyanosis.
- **Tension Pleural Effusion:** Rapidly progressive dyspnea with mediastinal shift causing obstructive shock (tachycardia, poor perfusion).

DIAGNOSIS

Diagnostic Criteria

- Clinical diagnosis based on **Stony Dullness + Shifted Mediastinum + Absent Breath Sounds.**
- Confirmed by Imaging (X-ray/Ultrasound).

Differentials

1. **Consolidation:** Dull note (not stony), **Increased** vocal fremitus/resonance, Bronchial breath sounds, No mediastinal shift.
2. **Collapse:** Dull note, **Shift of trachea TOWARD** the affected side, depressed intercostal spaces.
3. **Thickened Pleura:** Dull note, but no mediastinal shift; usually a history of treated empyema.

4. **Diaphragmatic Hernia:** (In infants) Scaphoid abdomen, bowel sounds heard in the chest.

Investigations

- **Tier 1:** Chest X-ray (PA view: Obliteration of costophrenic angle; Lateral decubitus: for small effusions). Ultrasound Chest (to detect loculations and guide aspiration).
- **Tier 2: Diagnostic Pleural Tap (Thoracocentesis):**
    - *Biochemistry:* Protein, LDH (Light's Criteria), Glucose, pH.
    - *Cytology:* Total/Differential count (Lymphocytic = TB/Malignancy; Neutrophilic = Parapneumonic).
    - *Microbiology:* Gram stain, Culture, AFB stain, GeneXpert (on fluid).
- **Tier 3:** Pleural biopsy (if TB suspected but fluid negative), CT Chest (for underlying lung pathology).

Management Outline

1. **Stabilization:** Oxygen, IV fluids (if in shock), positioning.
2. **Thoracocentesis:** Diagnostic and therapeutic (if respiratory distress is present).
3. **Specific Rx:**
    - **Parapneumonic/Empyema:** IV Antibiotics (Ceftriaxone + Vancomycin/Clindamycin) + **Intercostal Drainage (ICD) Tube**.
    - **Tubercular:** Antitubercular Therapy (ATT) as per national guidelines.
4. **Fibrinolytics:** [UPDATED] Intrapleural Urokinase or Streptokinase for loculated empyema to avoid surgery.
5. **Surgery:** VATS (Video-Assisted Thoracoscopic Surgery) for organized empyema.

EXAMINER'S VIVA

Q1: How do you differentiate between Pleural Effusion and Consolidation on examination? *A: In effusion, the trachea is shifted away, percussion is stony dull, and vocal fremitus/breath sounds are absent. In consolidation, the trachea is central, percussion is dull, and vocal fremitus/breath sounds (bronchial) are increased.*

Q2: What is Light's Criteria? *A: It distinguishes Exudate from Transudate. Fluid is an **Exudate** if: 1. Fluid protein/Serum protein >0.5; 2. Fluid LDH/Serum LDH >0.6; 3. Fluid LDH > 2/3rd the upper limit of normal serum LDH.*

Q3: Where do you insert an ICD tube? *A: Usually in the **"Triangle of Safety"**: Bordered by the anterior border of the latissimus dorsi, lateral border of pectoralis major, and a line superior to the 5th intercostal space, in the mid-axillary line.*

Q4: Why do we percuss the upper border of the rib when doing a pleural tap? *A: To avoid the neurovascular bundle (intercostal artery, vein, and nerve) which runs along the **lower border** of each rib.*

Q5: What is a "Massive Effusion" on X-ray? *A: Opacification of the entire hemithorax with mediastinal shift to the opposite side.*

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting the case of a [Age] year old male, previously healthy, who presented with acute onset high-grade fever, cough, and progressive breathlessness of 1-week duration..."
- **Mistake to avoid:** Don't forget to check the **BCG scar**; examiners look for this specifically in any respiratory case.
- **Mistake to avoid:** Don't say "Breath sounds are absent" without checking the **Axilla**. Small effusions are often missed because the resident only auscultates the back.
- **Observation:** If the child has an ICD tube in situ, describe the "swing" (oscillation) of the fluid column in the tube with respiration—this shows the system is patent.

---

18. Empyema

Subject: Respiratory

LONG CASE FRAMEWORK: PEDIATRIC EMPYEMA THORACIS

HISTORY

Chief Complaint

- **Fever:** Duration (usually >7-10 days), grade, and pattern.
- **Cough:** Nature (dry vs. productive), duration.
- **Chest Pain:** Pleuritic (sharp, worsens with deep breath/cough).
- **Difficulty Breathing:** Onset and progression.
- **Decreased Activity/Appetite:** Often more pronounced than in simple pneumonia.

History of Present Illness

- **The "Stalled Recovery":** Ask: "Was the child initially diagnosed with pneumonia, started on antibiotics, showed slight improvement, but then developed high-grade fever and worsening respiratory distress?" (Classic history of parapneumonic effusion progressing to empyema).
- **Character of Pain:** "Does the child cry or winced when coughing or moving? Does he prefer to lie on one specific side?" (Children often lie on the affected side to splint the chest).

- **Cough:** "Is the cough productive of foul-smelling sputum?" (Suggests anaerobic infection or bronchopleural fistula).
- **Systemic Symptoms:** "Has there been significant weight loss or night sweats?" (To differentiate from Tubercular etiology).
- **Negative Predictors:** Ask about choking episodes (foreign body), contact with TB, or history of recurrent skin boils (Staphylococcal etiology).

Relevant Background History

- **Past History:** Previous episodes of pneumonia, asthma, or hospitalizations. History of measles or varicella in the preceding month (predisposes to severe bacterial empyema).
- **Immunization:** Specifically ask about **PCV (Pneumococcal Conjugate Vaccine)** and **Hib**. A fully vaccinated child with empyema raises suspicion of non-vaccine serotypes or *Staph. aureus*.
- **Nutritional History:** Detailed caloric and protein intake. Malnutrition is both a risk factor and a consequence of chronic empyema.
- **Socioeconomic:** Overcrowding and smoking in the house.

EXAMINATION

General Survey

- **Observation:** Is the child "toxic-looking"? Empyema patients often look sicker than those with simple pleural effusion.
- **Posture:** [EXAMINER FAVORITE] Look for **scoliosis**—the child may lean *towards* the side of the lesion to reduce tension on the pleura.
- **Respiratory Effort:** Note nasal flaring, grunting, and accessory muscle use.
- **Nutritional Status:** Assess for visible wasting of the shoulder girdle and ribs (common in chronic cases).

Vital Signs and Anthropometry

- **Temperature:** Often high-grade (102-104°F) or "hectic" in the fibrinopurulent stage.
- **Respiratory Rate:** Count for a full minute. Tachypnea out of proportion to fever suggests significant lung compression or sepsis.
- **Heart Rate:** Tachycardia; if excessive, consider pericardial extension or tension pyothorax.
- **Growth:** Plot weight-for-age. Acute weight loss is common due to the catabolic state.

Peripheral Signs

- **Clubbing:** [SEVERITY MARKER] Grade 1-2 clubbing can develop rapidly (within 2-4 weeks) in empyema. If present, it suggests a chronic process or underlying bronchiectasis/cystic fibrosis.

- **Pallor:** Significant anemia of inflammation is common.
- **Lymph Nodes:** Check for supraclavicular or axillary nodes (TB or malignancy differentials).
- **Edema:** Check for pedal edema (hypoproteinemia due to protein loss in the pleural pus).

Systemic Examination — Respiratory System

Inspection

- **Shape of Chest:** Look for fullness or bulging of intercostal spaces on the affected side.
- **Movement:** Diminished chest excursion on the affected side.
- **Tracheal Position:** Inspect the suprasternal notch. Tracheal shift to the opposite side indicates a large collection.
- **Apex Beat:** May be shifted laterally/medially depending on the side of the empyema.

Palpation

- **Confirm Tracheal Shift:** Use your index finger in the suprasternal notch.
- **Confirm Apex Beat:** Locate the point of maximal impulse.
- **Chest Expansion:** Measure using a tape measure at the nipple level (in older children).
- **Vocal Fremitus:** [TECHNIQUE] Use the ulnar border of your hand. It will be **markedly diminished or absent** over the empyema.
- **Tenderness:** [EXAMINER FAVORITE] Gently palpate the intercostal spaces. **Intercostal tenderness** is a hallmark of empyema (pus under tension).

Percussion

- **Note: Stony Dull** note over the affected area.
- **Technique:** Percuss from top to bottom, comparing sides.
- **Upper Border:** In empyema, the upper limit of dullness is often highest in the axilla (Ellis S-shaped curve), though this is harder to elicit in a crying child.
- **Shift:** Shifting dullness is usually *not* elicited in empyema because the fluid is thick or loculated.

Auscultation

- **Breath Sounds:** Absent or markedly diminished over the area of dullness.
- **Bronchial Breathing:** [EXAMINER FAVORITE] Listen at the **upper border** of the effusion. You may hear tubular bronchial breathing due to the compressed lung just above the fluid level.
- **Vocal Resonance:** Diminished or absent. Look for **egophony** (E-to-A change) at the upper limit.
- **Crackles:** May be heard in the rest of the lung if there is associated pneumonia.

Systemic Examination — Secondary Systems

- **Abdomen:** Check for hepatomegaly (congestive heart failure or downward displacement by a right-sided empyema).
- **CVS:** Listen for muffled heart sounds (pericardial effusion).

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

1. **Stony Dull Percussion Note:** The most reliable sign of fluid in the pleural space.
2. **Intercostal Tenderness:** Highly suggestive of empyema (infected fluid) rather than simple transudative effusion.
3. **Scoliosis towards the affected side:** Indicates pleuritic pain and splinting.
4. **Bulging of Intercostal Spaces:** Indicates a large volume of fluid under pressure.
5. **Empyema Necessitans:** [RARE BUT CLASSIC] Look for a soft tissue swelling on the chest wall where the pus is tracking out through the pleura.

Severity Assessment [SEVERITY MARKER]

- **Mediastinal Shift:** Significant displacement of trachea/apex indicates tension and risk of cardiovascular collapse.
- **Respiratory Failure:** SpO2 <90%, severe retractions, inability to speak.
- **Sepsis:** Poor perfusion, altered sensorium, hypotension.
- **Bronchopleural Fistula (BPF):** Sudden onset of massive expectoration of pus; shifting air-fluid level on X-ray.

DIAGNOSIS

Diagnostic Criteria

- **Clinical:** Fever, respiratory distress, stony dullness, diminished breath sounds.
- **Radiological:** Opacification of the hemithorax with mediastinal shift.
- **Biochemical (Pleural Fluid):** Frank pus on aspiration OR Pleural fluid with:
    - pH < 7.2
    - Glucose < 40 mg/dL
    - LDH > 1000 IU/L
    - Presence of bacteria on Gram stain/culture.

Differentials

1. **Pleural Effusion (Serous):** Child looks less toxic, no intercostal tenderness, fluid is straw-colored.

2. **Chylothorax:** History of cardiothoracic surgery, milky fluid on aspiration.
3. **Diaphragmatic Hernia:** Scaphoid abdomen, bowel sounds heard in the chest (usually left side).
4. **Massive Consolidation:** Trachea is central, vocal fremitus is increased (not decreased), no stony dullness (usually dull but not "stony").

Investigations

- **Tier 1:** CBC (high TLC with left shift), CRP/ESR (elevated), Chest X-ray (PA and Lateral - look for meniscus sign).
- **Tier 2: Ultrasound Chest** (Best for identifying loculations and guiding aspiration). **Pleural fluid analysis** (Cell count, Biochemistry, Gram stain, Culture/Sensitivity).
- **Tier 3: CT Chest (Contrast):** To look for pleural thickening (peel), lung abscess, or BPF. **GeneXpert** on pleural fluid if TB is suspected.

Management Outline

1. **Stabilization:** Oxygen, IV fluids, antipyretics.
2. **Antibiotics:** Start IV Ceftriaxone + Vancomycin/Clindamycin (to cover *Staph* and *Strep*). Adjust based on culture. Duration: 3-4 weeks.
3. **Drainage:**
    - **Stage 1 (Exudative):** Thoracocentesis or ICD (Intercostal Drainage).
    - **Stage 2 (Fibrinopurulent):** ICD + Fibrinolytics (Urokinase/Streptokinase) OR **VATS (Video-Assisted Thoracoscopic Surgery)** [UPDATED: VATS is now preferred early in many centers].
    - **Stage 3 (Organizing):** Decortication (open surgery).
4. **Nutrition & Physio:** High-protein diet and chest physiotherapy (incentive spirometry).

EXAMINER'S VIVA

Q1: Why is the percussion note "stony" dull in empyema? *A: Because the fluid/pus completely replaces the air-containing lung and dampens all vibrations, unlike consolidation where some air-filled bronchi remain.*

Q2: How do you differentiate between a lung abscess and empyema with BPF on X-ray? *A: In empyema with BPF, the air-fluid level is wide and extends to the chest wall, and the height of the fluid level is unequal on PA and Lateral views. In a lung abscess, the air-fluid level is usually equal in both views.*

Q3: What are the indications for an ICD in a child? *A: Frank pus, pH < 7.2, positive Gram stain/culture, or a large effusion causing respiratory distress.*

Q4: Where do you insert an ICD? *A: Usually in the 4th or 5th intercostal space in the mid-axillary line (Safe Triangle).*

Q5: [Technique] Show me how you percuss the axilla. *A: (Demonstrate) Ask the child to place their hand on their head to expose the axillary area. Percuss in the mid-axillary line. This is crucial as fluid often accumulates highest here (Ellis S-curve).*

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting the case of [Name], a [Age] old male, who presented with high-grade fever and cough for 10 days, and progressive respiratory distress for 3 days, with physical findings consistent with a right-sided massive empyema thoracis, currently in the fibrinopurulent stage."
- **Mistake:** Forgetting to check for tracheal shift. Examiners watch your finger placement in the suprasternal notch very closely.
- **Mistake:** Not mentioning the PCV vaccination status. In the post-PCV era, the microbiology of empyema has shifted.
- **Observation:** If you see an ICD in situ, describe the column movement (respiratory swing) and the nature of the drainage (serous, purulent, or bloody).

---

19. Interstitial lung disease

Subject: Respiratory

This is a challenging long case. In Pediatric Interstitial Lung Disease (chILD), the diagnosis is often "hidden" behind a label of "recurrent pneumonia" or "difficult asthma." As a PG resident, your job is to demonstrate that you can distinguish parenchymal restrictive disease from airway disease through meticulous physical signs.

HISTORY

Chief Complaint

- **Progressive breathlessness:** Usually insidious. Ask for duration (months vs. weeks).
- **Persistent cough:** Typically dry, non-productive, and "hacking."
- **Failure to thrive:** Inability to gain weight despite adequate intake.
- **Exercise intolerance:** In older children, "Can you keep up with peers during play?"

History of Present Illness

- **Breathlessness:** "When did you first notice the fast breathing? Is it only during feeds (infants) or while walking (older children)?" Characterize using the NYHA/Ross classification.
- **Cough:** "Is there any phlegm? Does it change with posture or time of day?" (ILD cough is usually dry and constant).

- **Feeding History (Crucial for infants):** "Does the baby take breaks during breastfeeding? Does he sweat profusely while feeding? How long does a single feed take?" (Suck-rest-suck cycle suggests respiratory distress).
- **Cyanosis:** "Have you noticed bluish discoloration of lips or tongue? Does it happen only during crying or even at rest?"
- **Negative Predictors:**
    - "Is there any wheezing?" (Absence of wheeze points away from asthma/bronchiolitis).
    - "Any history of choking or sudden onset coughing?" (Excludes foreign body).
    - "Any history of recurrent foul-smelling stools?" (Excludes Cystic Fibrosis).

Relevant Background History

- **Past History:** Recurrent "pneumonias" that never fully resolve on X-ray.
- **Antenatal/Birth:** Prematurity, prolonged oxygen requirement, or NICU stay (points toward BPD or surfactant protein deficiencies).
- **Developmental:** Gross motor delay due to dyspnea and hypoxia.
- **Environmental/Exposure:** "Do you keep pigeons or birds? Is there dampness/mold on the walls? Any family members with TB or chronic lung disease?" (Hypersensitivity pneumonitis).
- **Family History:** Consanguinity (Autosomal recessive surfactant mutations) or unexplained sibling deaths in infancy.

EXAMINATION

General Survey

- **The "Look":** Observe the child's respiratory effort. In ILD, you see **"Quiet Tachypnea"**—rapid breathing without the loud grunting or wheezing of airway disease.
- **Nutritional Status:** Look for "Spindly extremities" (muscle wasting) and loss of buccal fat pads. ILD is a high-caloric state.
- **Skin:** Look for heliotrope rash or Gottron papules (Juvenile Dermatomyositis associated ILD).

Vital Signs and Anthropometry

- **Respiratory Rate:** Count for a full minute. Expect tachypnea out of proportion to the clinical "illness."
- **Pulse Oximetry [CRITICAL]:** Check SpO2 at rest AND after a 6-minute walk test (or after feeding in infants). **Exertional desaturation** is a hallmark of ILD.
- **Growth:** Plot Weight-for-Age and Height-for-Age. You will likely see a "stalled" growth curve.

Peripheral Signs

- **Clubbing [EXAMINER FAVORITE]:**

    - *Technique:* Look for obliteration of the diamond-shaped Schamroth's window.
    - *Significance:* Suggests chronic hypoxia and advanced architectural distortion (e.g., NEHI usually does *not* have clubbing; surfactant protein C deficiency often *does*).
- **Cyanosis:** Check the tongue and sublingual mucosa.
- **BCG Scar:** Always check; TB can mimic any lung pathology.

Systemic Examination — Respiratory (Primary)

- **Inspection:**
    - **Shape of Chest:** May be normal or "pigeon chest" (pectus carinatum) due to chronic increased work of breathing.
    - **Retractions:** Subcostal and intercostal retractions are common.
- **Palpation:**
    - **Trachea:** Usually midline (unlike the shift seen in collapse/effusion).
    - **Chest Expansion:** Will be **symmetrically reduced**. This is a restrictive disease.
    - **Vocal Fremitus:** May be increased over areas of fibrosis.
- **Percussion:**
    - **Note:** Usually resonant, but may be "woody" or dull if there is extensive fibrosis.
    - **Liver Dullness:** Check the upper border; in ILD, the lungs are small (restrictive), so the liver is not displaced downward (unlike asthma/emphysema).
- **Auscultation:**
    - **Breath Sounds:** Bronchial or harsh vesicular.
    - **Adventitious Sounds [EXAMINER FAVORITE]:** Look for **"Velcro Crepitations."**
    - *Technique:* Listen at the lung bases posteriorly. These are fine, end-expiratory, non-shifting, "dry" crackles.
    - *Significance:* They represent the sudden opening of small airways/alveoli stiffened by fibrosis.
    - **Wheeze:** Usually absent. If present, consider "Overlap" or Neuroendocrine Hyperplasia of Infancy (NEHI).

Systemic Examination — Secondary Systems

- **Cardiovascular:** Look for a **loud P2** and a left parasternal heave (Signs of Pulmonary Hypertension/Cor Pulmonale).
- **Abdomen:** Hepatomegaly (Right heart failure).

SIGNS THAT CLINCH THE DIAGNOSIS [EXAMINER FAVORITES]

1. **Exertional Desaturation:** SpO2 drops significantly with minimal activity.
2. **Velcro Crepitations:** Fine, dry, end-expiratory crackles that do not clear with coughing.
3. **Quiet Tachypnea:** High RR with minimal accessory muscle use compared to the degree of hypoxia.
4. **Clubbing in a non-cyanotic heart disease/non-CF patient:** Strongly points to ILD or Bronchiectasis.

SEVERITY ASSESSMENT [SEVERITY MARKER]

- **Mild:** Tachypnea at rest, normal SpO2, normal growth.
- **Moderate:** Exertional desaturation, growth failure, significant retractions.
- **Severe:** Resting cyanosis, signs of Pulmonary Hypertension (loud P2), Cor Pulmonale.

DIAGNOSIS

Diagnostic Criteria

- No single "Jones-like" criteria. Diagnosis is based on the **"Triple Trigger"**:
    1. Clinical (Tachypnea, crackles, growth failure).
    2. Radiologic (Diffuse infiltrates on HRCT).
    3. Physiological (Restrictive pattern on PFT or hypoxia).

Differentials

1. **Cystic Fibrosis:** Distinguished by productive cough, malabsorptive stools, and sweat chloride >60 mmol/L.
2. **Recurrent Aspiration:** History of choking, neurological impairment, or GERD.
3. **Hypersensitivity Pneumonitis:** History of bird/mold exposure; improves when removed from environment.
4. **Post-Infectious Bronchiolitis Obliterans (PIBO):** Follows a severe viral (Adenovirus) pneumonia; HRCT shows "Mosaic attenuation."

Investigations

- **Tier 1:** CXR (reticulonodular opacities, "ground glass"), Pulse oximetry, HIV/Immunoglobulins (to rule out PJP/Infection).
- **Tier 2: HRCT Chest (Gold Standard):** Look for ground-glass opacities, honeycombing, or "crazy paving."
- **Tier 3:** Genetic testing (SFTPB, SFTPC, ABCA3 genes), Bronchoalveolar Lavage (BAL) to rule out infection/hemosiderosis, and **Lung Biopsy** (the definitive histological diagnosis).

Management Outline

- **Supportive:** High-calorie nutrition (150% RDA), Oxygen therapy to maintain SpO2 >92%.
- **Specific:**
    - **Corticosteroids:** Pulse Methylprednisolone (30mg/kg/day for 3 days monthly).
    - **Steroid-sparing agents:** Azathioprine, Hydroxychloroquine (especially for surfactant protein C mutations).
- **Monitoring:** Monthly 6-minute walk tests and serial PFTs (Spirometry/DLCO).

EXAMINER'S VIVA

1. **Q: Why do we call them "Velcro" crackles?**
    - A: Because they sound like the pulling apart of the hook-and-loop fastener. They are fine, high-pitched, and occur at the very end of inspiration.
2. **Q: What is the significance of a "Mosaic pattern" on HRCT?**
    - A: It indicates areas of different aeration. In ILD, it suggests air trapping or patchy fibrosis.
3. **Q: How do you perform a 6-minute walk test in a 4-year-old?**
    - A: It is difficult. We use the "Timed Floor to Stand" test or simply observe SpO2 while the child plays or climbs stairs for a set period.
4. **Q: What is NEHI?**
    - A: Neuroendocrine Hyperplasia of Infancy. A specific chILD syndrome presenting with tachypnea and crackles, but with a characteristic "geographic" ground-glass pattern on HRCT involving the right middle lung and lingula. It has a good prognosis.
5. **Q: Why is the liver not palpable in ILD compared to Asthma?**
    - A: In asthma, hyperinflation pushes the diaphragm down (pseudo-hepatomegaly). In ILD, the lungs are stiff and small (low lung volumes), so the diaphragm remains high.

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting [Name], a [Age] old male, with a history of progressive breathlessness and dry cough for 6 months, associated with significant weight loss, currently having quiet tachypnea and fine end-expiratory crackles on examination."
- **Mistake:** Don't call every crackle "crepitations." Specify if they are fine/coarse and if they change with posture.
- **Observation:** Examiners watch how you handle the pulse oximeter. Always check the waveform (pleth) before reading the SpO2 to ensure it's accurate.

Gastrointestinal and Liver

20. Chronic liver disease

Subject: Gastrointestinal and Liver

HISTORY: CHRONIC LIVER DISEASE (CLD)

Chief Complaint

- Abdominal distension (insidious onset)
- Yellowish discoloration of eyes/skin (recurrent or persistent)
- Hematemesis or melena (sudden onset, indicates portal hypertension)
- Altered sensorium or sleep-wake reversal (encephalopathy)
- Failure to gain weight or height

History of Present Illness

Ask these questions naturally to build the "Life History of the Liver":

- **The Onset:** "When did you first notice the tummy getting bigger? Was it sudden or did it creep up over months?" (Sudden suggests acute-on-chronic or Budd-Chiari; insidious suggests cirrhosis).
- **Jaundice:** "Is the yellow color always there, or does it come and go? Is the urine staining the clothes deep yellow?" (Persistent conjugated jaundice vs. intermittent hemolysis).
- **Pruritus:** "Does the child scratch more at night? Do you see scratch marks?" (Suggests cholestatic etiologies like Alagille or PFIC).
- **GI Bleed:** "Has he ever vomited blood or passed black, tarry, sticky stools?" (Distinguish melena from iron-supplement stools).
- **Neuropsychiatric:** "Has his school performance dropped? Is he irritable during the day and awake all night?" (Early Hepatic Encephalopathy).
- **The "Negative" Screen for Etiology:**
    - "Any history of umbilical catheterization in the newborn period?" (Portal vein thrombosis).
    - "Any recurrent fractures or tremors?" (Wilson Disease).
    - "Any chronic cough or greasy stools?" (Cystic Fibrosis).
    - "Any history of blood transfusions?" (Hepatitis B/C or Hemochromatosis/Thalassemia).

Relevant Background History

- **Antenatal/Birth:** Ask about neonatal jaundice. "Did it start in the first 24 hours? Did it last more than 2 weeks?" (Biliary atresia/Neonatal hepatitis).

- **Developmental:** "Has he lost any milestones he previously gained?" (Wilson Disease, Galactosemia).
- **Nutritional:** Detailed 24-hour recall. CLD patients are often in a state of "accelerated starvation."
- **Family:** Consanguinity (Autosomal Recessive conditions like PFIC, Wilson). "Any sibling deaths due to liver disease?"
- **Socioeconomic:** Ability to afford long-term transplant or immunosuppression.

EXAMINATION

General Survey

- **The "CLD Look":** Observe for a "pot-bellied" child with thin extremities (muscle wasting) and a sallow complexion.
- **Activity:** Is the child "flapping"? Look for asterixis even while they play.
- **Nutritional Status:** Look for temporal wasting and loss of gluteal fat. [SEVERITY MARKER]
- **Skin:** Look for **Spider Naevi** (usually in SVC distribution - neck, upper chest). Press the center with a glass slide; it should blanch and refill from the center.
- **Facies:**
    - **Alagille Syndrome:** Broad forehead, deep-set eyes, pointed chin (triangular face).
    - **Wilson Disease:** Mask-like facies or "risus sardonicus."

Vital Signs and Anthropometry

- **Pulse:** Look for a **bounding pulse** (hyperdynamic circulation in portal hypertension).
- **BP:** Often low-normal in cirrhosis due to systemic vasodilation.
- **Growth:** Height and Weight are mandatory. Plot on WHO/IAP charts. **Stunting** indicates the chronicity of the liver insult.

Peripheral Signs

- **Hands:**
    - **Leukonychia:** (White nails) due to hypoalbuminemia.
    - **Clubbing:** Suggests Hepatopulmonary Syndrome or biliary cirrhosis.
    - **Palmar Erythema:** Redness over thenar/hypothenar eminences (hyperestrogenism).
- **Eyes:**
    - **Icterus:** Best seen in the superior bulbar conjunctiva in natural light.

    - **Kayser-Fleischer (KF) Ring:** [EXAMINER FAVORITE] Use a torch from the side. Look for a greenish-brown copper deposit at the periphery of the cornea (Descemet's membrane). If suspected, must request Slit-lamp exam.

- **Lymph Nodes:** Check for Virchow's node (left supraclavicular) – rare in kids, suggests malignancy.
- **Edema:** Check over the medial malleolus. In CLD, edema usually follows ascites (unlike nephrotic syndrome).

Systemic Examination — Abdomen (Primary System)

Inspection:

- **Shape:** Protuberant, flanks full.
- **Umbilicus:** Everted or transverse (due to pressure).
- **Veins: Caput Medusae** (veins radiating from umbilicus) or lateral thoracic veins. Determine the direction of flow: [TECHNIQUE] Empty the vein with two fingers, release one. If it fills from bottom to top above the umbilicus, it's normal/portal HTN. If it fills from bottom to top *below* the umbilicus, suspect IVC obstruction.

Palpation:

- **Liver:**
    - Technique: Start in the Right Iliac Fossa. Move up with respiration.
    - Note: Firm/Hard consistency suggests Cirrhosis. Irregular surface suggests Macronodular cirrhosis or Hepatoma.
    - **Liver Span:** Measure in the mid-clavicular line. A small, shrunken liver is a hallmark of advanced cirrhosis.
- **Spleen:**
    - Technique: Start from RIF towards the Left Costal Margin.
    - Significance: Splenomegaly is the most reliable clinical sign of **Portal Hypertension**.
- **Gallbladder:** If palpable in a jaundiced child, think of a choledochal cyst.

Percussion:

- **Shifting Dullness:** [TECHNIQUE] Percuss from umbilicus to the flank until dull. Keep finger there, turn child 45 degrees towards you. Wait 30 seconds. If it becomes resonant, it's positive. (Requires ~100ml fluid).
- **Fluid Thrill:** [TECHNIQUE] Need an assistant's hand on the midline to stop skin-wave transmission. Tap one flank, feel on the other. (Indicates massive ascites).

Auscultation:

- **Cruveilhier-Baumgarten Murmur:** A venous hum over the epigastrium due to collateral circulation in the falciform ligament.

- **Bruit:** Over the liver suggests Hepatoma or Hemangioma.

Secondary Systems

- **Respiratory:** Listen for basal dullness (Pleural effusion/Hepatic hydrothorax) or crepitations.
- **CNS:** Assess for **Asterixis** (Flapping tremor). Ask the child to "stop traffic" with their hands. Look for jerky, irregular flexion-extension at the MCP and wrist joints.

SIGNS THAT CLINCH THE DIAGNOSIS [EXAMINER FAVORITES]

- **Splenomegaly in the absence of significant lymphadenopathy:** Points strongly toward Portal Hypertension.
- **KF Ring:** Pathognomonic for Wilson Disease (if >5 years old).
- **Direction of flow in abdominal veins:** Distinguishes Portal HTN from IVC obstruction.
- **Palpable Left Lobe of Liver in the epigastrium:** Often enlarged in cirrhosis while the right lobe shrinks.
- **Spider Naevi:** Highly specific for significant parenchymal liver disease.

SEVERITY ASSESSMENT

- **Child-Pugh Score:** (Bilirubin, Albumin, INR, Ascites, Encephalopathy).
- **PELD/MELD Score:** Used for transplant prioritization.
- **Clinical "Red Flags":** Deep jaundice, gross ascites, muscle wasting, and any degree of altered sensorium.

DIAGNOSIS

Diagnostic Criteria

- **Cirrhosis:** A pathological diagnosis, but clinically diagnosed by the triad of: **Liver dysfunction + Portal Hypertension + Shrunken/Firm Liver.**

Differentials

1. **Wilson Disease:** Presents with liver failure, tremors, or school failure; KF ring positive.
2. **Extra-Hepatic Portal Vein Obstruction (EHPVO):** Massive spleen, *normal* liver functions, no stigmata of CLD (like spider naevi).
3. **Chronic Budd-Chiari Syndrome:** Massive ascites, hepatomegaly, and prominent veins on the back.
4. **Autoimmune Hepatitis:** Often adolescent girls, associated with other autoimmune features (joint pains, rash).

Investigations

- **Tier 1:** CBC (look for thrombocytopenia = hypersplenism), LFT (Albumin/Globulin ratio reversal), PT/INR (best marker of synthetic function).
- **Tier 2:** Ultrasound Abdomen with Doppler (portal vein diameter, flow direction), Serum Ceruloplasmin/24hr Urinary Copper.
- **Tier 3:** Upper GI Endoscopy (to screen for varices), Liver Biopsy (Gold Standard for etiology/fibrosis), Fibroscan.

Management Outline

1. **Nutrition:** High calorie (1.5x RDA), high protein (unless encephalopathy), MCT-rich oil.
2. **Portal HTN:** Propranolol (primary prophylaxis) or Endoscopic Variceal Ligation (EVL).
3. **Ascites:** Salt restriction + Spironolactone (first line) +/- Furosemide.
4. **Definitive:** Liver Transplantation.

EXAMINER'S VIVA

1. **Q: Why is the spleen enlarged in CLD?** *A: Congestive splenomegaly due to increased resistance to portal blood flow.*
2. **Q: How do you differentiate a liver mass from a kidney mass?** *A: Liver moves with respiration, is not bimanually palpable/ballottable, and has no resonance (bowel) anterior to it.*
3. **Q: What is the first sign of Hepatic Encephalopathy in a child?** *A: Sleep-wake reversal and irritability.*
4. **Q: Why do we measure Liver Span?** *A: To detect a shrunken liver; a liver may not be palpable but can still be diseased. Normal span is 6-12 cm depending on age.*
5. **Q: [Technique] How do you elicit 'Dipping' or 'Ballottement' of the liver?** *A: In massive ascites, push your fingers sharply toward the liver; you will feel the liver 'sink' and 'float' back to hit your fingertips.*

LONG CASE PRESENTATION TIPS

- **The Opening:** "I am presenting [Name], a [Age] year old male, with a history of progressive abdominal distension and jaundice for 6 months, now presenting with an episode of hematemesis, suggesting Chronic Liver Disease with Portal Hypertension."
- **Mistake:** Forgetting to check the back for dilated veins or the chest for spider naevi.
- **Mistake:** Calling it "Cirrhosis" without evidence of both synthetic failure and portal hypertension. Stick to "Chronic Liver Disease" until you have the full triad.

---

21. Portal hypertension

Subject: Gastrointestinal and Liver

This is a classic "bread and butter" long case for the MD exam. In Portal Hypertension (PHT), the examiner is testing your ability to differentiate between **Pre-hepatic** (Extrahepatic Portal Venous Obstruction - EHPVO), **Hepatic** (Cirrhosis), and **Post-hepatic** causes, and your skill in performing a meticulous abdominal examination.

HISTORY

Chief Complaint

- Abdominal distension (duration)
- Hematemesis or Melena (the "frightening" event)
- Yellowish discoloration of eyes/skin
- Mass in the left upper abdomen (Spleen)

History of Present Illness

Focus on the "Site of Block" and "Complications".

- **Characterizing the Bleed:** "Was the blood bright red or coffee ground? Was it preceded by retching (Mallory-Weiss) or did it come up effortlessly (Variceal)? How many episodes? Did the child require a transfusion?"
- **Characterizing the Mass:** "When did you first notice the lump under the left ribs? Is it growing? Does it cause 'fullness' after eating only a small meal (Early satiety)?"
- **Differentiating the Cause:**
    - **Pre-hepatic (EHPVO):** "Was there any history of umbilical sepsis, exchange transfusion, or severe dehydration in the neonatal period?" (Crucial for EHPVO).
    - **Hepatic (Cirrhosis):** "Is there jaundice? Has the urine been dark? Is there any history of itching (Pruritus)?"
    - **Post-hepatic:** "Is there any history of tender, rapid abdominal swelling with weight gain (Budd-Chiari)?"
- **Negative Predictors:** "Is there any history of altered sleep-wake cycle, irritability, or flapping tremors (Encephalopathy)?" *Note: Encephalopathy is rare in EHPVO but common in Cirrhosis*.
- **Growth/Nutrition:** "Has the child's growth slowed down? Any history of easy bruising or bone pain?"

Relevant Background History

- **Antenatal/Birth:** Neonatal umbilical catheterization or sepsis (Risk for EHPVO).
- **Past History:** Previous sclerotherapy/banding (EVL) sessions. History of recurrent fractures or infections (Hypersplenism/Malnutrition).
- **Family History:** Consanguinity (Wilson’s, Galactosemia). Sibling with liver disease.
- **Socioeconomic:** Ability to afford long-term follow-up and beta-blockers.

EXAMINATION

General Survey

- **Observation:** Look for the "Wasted but Distended" appearance. In EHPVO, the child often looks surprisingly well despite a massive spleen. In Cirrhosis, the child looks "sick" (Chronic Liver Disease facies).
- **Nutritional Status:** Check for temporal wasting, loss of gluteal fat, and mid-upper arm circumference (MUAC).
- **Skin:** Look for **Spider Naevi** (usually in SVC distribution), **Palmar Erythema**, and **Caput Medusae** (dilated veins around the umbilicus).

Vital Signs and Anthropometry

- **Pulse:** Tachycardia may indicate compensated shock (recent bleed) or hyperdynamic circulation.
- **BP:** Check for orthostatic hypotension if there is a history of recent hematemesis.
- **Growth:** Plot Height and Weight. Stunting is common in Cirrhosis; EHPVO children are often better preserved.

Peripheral Signs

- **Icterus:** Best seen in the upper bulbar conjunctiva. [SEVERITY MARKER]
- **Clubbing:** Suggests Hepatopulmonary Syndrome or biliary cirrhosis.
- **Edema:** Check the shins and sacrum. Pitting edema suggests hypoalbuminemia (Cirrhosis).
- **Lymphadenopathy:** Look for Virchow's node (unlikely in kids, but rule out malignancy).

Systemic Examination — Abdomen (The Primary System)

1. Inspection:

- **Shape:** Protuberant. Look for fullness in the flanks (Ascites).
- **Umbilicus:** Everted? Displaced downwards?
- **Veins:** Look for prominent veins. **[EXAMINER FAVORITE]** Determine the direction of flow. If flow is *away* from the umbilicus, it is Portal Hypertension. If flow is *upward* from the pubis, it is IVC obstruction.

2. Palpation:

- **Spleen:** Start in the Right Iliac Fossa. Use the "Classical" or "Middleton's" maneuver.
    - *Characterize:* Size in cm from the costal margin, notch (medial border), moves with respiration, cannot get above it.
    - *Significance:* Massive splenomegaly with no/minimal jaundice strongly suggests EHPVO.

- **Liver:** Palpate for size, consistency (firm/hard), and surface (nodular in cirrhosis).
    - **Liver Span:** Measure in the mid-clavicular line. A shrunken liver suggests advanced Cirrhosis.
- **Fluid:** Perform **Shifting Dullness** (if moderate ascites) or **Fluid Thrill** (if massive ascites).

3. Percussion:

- **Traube's Space:** Percuss in the 6th-9th ICS in the left mid-axillary line. Dullness indicates splenomegaly even before it is palpable.

4. Auscultation:

- **Cruveilhier-Baumgarten Murmur:** A venous hum over the epigastrium/umbilicus due to collateral flow in the falciform ligament.

Secondary Systems

- **Respiratory:** Check for dullness at the right base (Hepatic Hydrothorax) or tachypnea (Hepatopulmonary Syndrome).
- **CNS:** Check for **Asterixis (Flapping Tremor)**: Ask the child to extend arms and dorsiflex wrists. Look for jerky, irregular flexion-extension. [SEVERITY MARKER]

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

1. **Splenomegaly with Notch:** Confirms the mass is the spleen and indicates portal congestion.
2. **Direction of Flow in Abdominal Veins:** Centrifugal flow (away from umbilicus) confirms PHT.
3. **Liver Span:** A small, firm liver with a massive spleen = Cirrhosis. A normal liver with a massive spleen = EHPVO.
4. **Shifting Dullness:** Confirms ascites, suggesting either Cirrhosis or Budd-Chiari (rare in EHPVO unless there is severe malnutrition).

Severity Assessment

- **Child-Pugh Score:** (Bilirubin, Albumin, INR, Ascites, Encephalopathy).
- **PELD/MELD Score:** Used for transplant prioritization.
- **Signs of Decompensation:** Ascites, Jaundice, Encephalopathy, GI Bleed.

DIAGNOSIS

Diagnostic Criteria

- Clinical diagnosis based on Splenomegaly + Ascites/Variceal bleed.
- Confirmed by Ultrasound Doppler (Portal vein diameter > age-specific norms, reduced/reversed flow).

Differentials

1. **EHPVO:** Massive spleen, normal liver functions, history of neonatal umbilical sepsis.
2. **Cirrhosis (Post-Hepatitic/Wilson's):** Jaundice, firm/shrunken liver, stunting, stigmata of CLD.
3. **Budd-Chiari Syndrome:** Rapid onset ascites, tender hepatomegaly, flow in abdominal veins is *upwards*.
4. **Hematological Malignancy:** Fever, lymphadenopathy, bone pain (but PHT signs like caput medusae will be absent).

Investigations

- **Tier 1:** CBC (look for **Pancytopenia** suggesting hypersplenism), LFT (Albumin/Bilirubin), PT/INR (Synthetic function).
- **Tier 2: USG Doppler of Abdomen** (Look for "Cavernoma" transformation in EHPVO). **Upper GI Endoscopy** (to grade varices).
- **Tier 3:** Slit lamp exam (KF rings), Ceruloplasmin (Wilson's), Liver Biopsy (if etiology unknown and safe).

Management Outline

1. **Acute Bleed:** NPO, IV fluids, Octreotide infusion, IV Antibiotics (Ceftriaxone), Endoscopic Band Ligation (EVL) within 12-24 hours.
2. **Prophylaxis:** Propranolol (non-selective beta-blocker) to reduce portal pressure.
3. **Definitive (EHPVO):** Meso-Rex bypass (restores flow) or Splenorenal shunt.
4. **Definitive (Cirrhosis):** Liver Transplantation.

EXAMINER'S VIVA

Q: Why is the spleen enlarged in PHT? A: It is primarily due to passive venous congestion, but also due to hyperplasia of the reticuloendothelial cells and increased splenic blood flow.

Q: How do you differentiate a spleen from a kidney clinically? A: Spleen has a notch, is not bimanually palpable (usually), has no resonance over it (kidneys have colonic resonance), and moves diagonally with respiration.

Q: What is "Hypersplenism"? A: A triad of: 1. Splenomegaly, 2. Cytopenia in one or more cell lines (Anemia, Leukopenia, Thrombocytopenia), and 3. Normal/Hypercellular bone marrow.

Q: Why do we avoid Aspirin/NSAIDs in these children? A: Risk of precipitating a variceal bleed due to gastric mucosal irritation and anti-platelet effects.

Q: What is the "Cavernoma" seen on USG? A: In EHPVO, the portal vein is thrombosed. The body forms multiple small collateral vessels around the block to bypass it; this "tangle" of vessels is the cavernoma.

LONG CASE PRESENTATION TIPS

- **Opening Statement:** "I am presenting [Name], a [Age] year old male, who presented with a massive left-sided abdominal lump and two episodes of hematemesis, with no prior history of jaundice, suggesting a pre-hepatic portal hypertension, likely EHPVO."
- **Common Mistake:** Forgetting to check the "Direction of Flow" in abdominal veins. Use two fingers to empty a segment of the vein and see which way it refills.
- **Watch out:** Examiners will watch your hand placement during spleen palpation. Always start from the Right Iliac Fossa; if you start too high, you will miss a massive spleen.

---

22. Severe acute malnutrition

Subject: Gastrointestinal and Liver

HISTORY

Chief Complaint

- "Failure to gain weight" or "Weight loss" (Duration is key: acute vs. chronic)
- "Looseness of clothes" or "Thinning of limbs"
- "Swelling of feet and face" (Suggests Kwashiorkor/Edematous malnutrition)
- "Irritability or lethargy"
- "Skin changes or hair loss"

History of Present Illness

- **Weight Loss:** "When did you first notice the child becoming thin? Was it after a specific illness like diarrhea or measles?" (Helps identify a precipitating event).
- **Edema:** "Where did the swelling start? Did it start from the feet and move up, or was it first seen around the eyes?" (Nutritional edema is usually dependent and bilateral).
- **Appetite:** "How is the child's 'hunger'? Does he reach for food, or do you have to force-feed him?" (Loss of appetite is a [SEVERITY MARKER] in SAM).
- **Diarrhea:** "How many stools per day? Is there blood or mucus? Is it watery?" (Persistent diarrhea is a common co-morbidity).
- **Fever/Cough:** "Has there been a persistent cough for more than 2 weeks or contact with a TB patient?" (SAM is a TB mimic and vice versa).
- **Activity Level:** "Is the child playing as usual, or does he just sit in one place and cry?" (Apathy is classic for Kwashiorkor).

Relevant Background History

- **Dietary History (Crucial):** Perform a 24-hour recall. Calculate the **Calorie and Protein gap**.
    - "Was the child exclusively breastfed for 6 months?"

    - "When were solids started? What was the consistency (watery dal vs. thick mash)?"
    - "How many times a day is the child fed?"
- **Birth History:** "Was the child born small (IUGR/LBW)?" (Distinguishes stunted growth from acute wasting).
- **Immunization:** Specifically ask about **Measles** (a common precipitant of SAM) and **BCG** (TB risk).
- **Developmental:** "Has the child lost any previously gained milestones?" (Gross motor delay is common due to muscle wasting).
- **Socioeconomic:** "Who earns in the family? How much is spent on food? What is the source of drinking water?" (SAM is a social disease).

EXAMINATION

General Survey

- **Initial Impression:** Does the child look like "skin and bones" (Marasmus) or "miserable and swollen" (Kwashiorkor)?
- **Activity:** Observe for **Apathy** (child doesn't smile or interact) or **Irritability**.
- **Wasting:** Look for the **"Old Man Facies"** (loss of buccal fat pads—the last fat to go) and **"Baggy Pant" appearance** (excess skin folds in the gluteal region).
- **Skin:** Look for **"Flaky Paint Dermatosis"** (hyperpigmented patches that peel off to reveal pale skin underneath—pathognomonic for Kwashiorkor).

Vital Signs and Anthropometry

- **Temperature:** Check for **Hypothermia** (<35°C/95°F). SAM children don't always mount a fever even with severe infection.
- **Heart Rate/RR:** Look for signs of heart failure or pneumonia.
- **Weight:** Use a digital scale (tared) for infants.
- **Height/Length:** Use an infantometer (<2yrs) or stadiometer (>2yrs).
- **MUAC:** Use the Shaker tape. Measure at the midpoint between the acromion and olecranon processes. [SEVERITY MARKER: <11.5 cm in 6–59 months].
- **Weight-for-Height (W/H):** Plot on WHO growth charts. [SEVERITY MARKER: < -3SD].

Peripheral Signs

- **Hands/Nails:** Look for **Koilonychia** (Iron deficiency) or **Beau's lines**. Check capillary refill (dehydration is hard to assess in SAM).
- **Eyes:**

    - **Bitot's spots:** Foamy triangular patches on the bulbar conjunctiva (Vitamin A deficiency).
    - **Conjunctival Xerosis:** Dryness.
    - **Keratomalacia:** [EMERGENCY] Cloudy/soft cornea.
- **Hair: "Flag Sign"** (alternating bands of light and dark hair reflecting periods of poor vs. good nutrition), easy pluckability, sparse/thin hair.
- **Mouth:** Angular stomatitis (B2 deficiency), bleeding gums (Vitamin C), or **Cancrum Oris** (Noma—severe infection).
- **Edema:** [EXAMINER FAVORITE] Press firmly with your thumb over the dorsum of the foot for **3 full seconds**. If a pit remains, it is positive. It must be **bilateral** to diagnose SAM.

Systemic Examination — Primary System (Abdomen)

- **Inspection:** Scaphoid or distended? Look for visible peristalsis.
- **Palpation:**
    - **Liver:** Feel for hepatomegaly. In Kwashiorkor, the liver is often enlarged and soft due to **fatty infiltration**.
    - **Skin Turgor:** In Marasmus, skin turgor is poor due to loss of subcutaneous fat, but this is a **false positive** for dehydration. Use the "Abdominal Pinch" cautiously.
- **Percussion:** Check for shifting dullness if ascites is suspected (rare, usually due to severe hypoalbuminemia).

Systemic Examination — Secondary Systems

- **Chest:** Listen for fine crepitations (Pneumonia often presents without cough/fever in SAM).
- **CVS:** Listen for a hemic murmur (Anemia) or signs of heart failure (S3 gallop) during refeeding.
- **CNS:** Assess tone (usually hypotonic) and mental status.

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **Visible Bony Prominences/Loss of Gluteal Fat:** Indicates Marasmus.
- **Bilateral Pitting Edema:** The defining feature of Kwashiorkor.
- **MUAC < 11.5 cm:** Rapid screening tool for SAM in children 6–59 months.
- **Flaky Paint Dermatosis:** Specific to edematous malnutrition.
- **Appetite Test:** If the child refuses RUTF (Ready-to-Use Therapeutic Food), it indicates a metabolic crisis/infection.

DIAGNOSIS

Diagnostic Criteria (WHO/IAP)

For a child aged 6 months to 5 years, any one of:

1. **W/H Z-score < -3 SD**
2. **MUAC < 11.5 cm**
3. **Bilateral Pitting Edema** (Nutritional)

Differentials

1. **Celiac Disease:** Chronic diarrhea, wasting, but usually no edema; look for abdominal distension.
2. **Disseminated Tuberculosis:** Fever, lymphadenopathy, contact history.
3. **HIV/AIDS:** Recurrent infections, oral thrush, persistent lymphadenopathy.
4. **Congestive Heart Failure:** Edema and hepatomegaly, but will have raised JVP and cardiomegaly.

Investigations

- **Tier 1 (Bedside):** Random Blood Sugar (Hypoglycemia is a silent killer), Hemoglobin, Urine routine (check for UTI), Stool for parasites.
- **Tier 2 (Confirmatory/Baseline):** Serum electrolytes (look for **Hypokalemia**, Hypomagnesemia), Serum Albumin (low in Kwashiorkor), Chest X-ray (rule out TB/Pneumonia).
- **Tier 3 (Complications):** Blood culture (if hypothermic/lethargic), HIV screening, Mantoux test (though often false negative in SAM).

Management Outline

1. **Stabilization Phase (Days 1–7):**
    - Treat/Prevent **Hypoglycemia** (10% Dextrose).
    - Treat/Prevent **Hypothermia** (Kangaroo Mother Care).
    - Treat **Dehydration** (Use **ReSoMal**—low sodium, high potassium).
    - Correct **Electrolytes** (No extra Sodium; give Potassium and Magnesium).
    - Treat **Infection** (Broad-spectrum antibiotics are mandatory for all SAM).
    - Correct **Micronutrients** (Vitamin A on Day 1; **No Iron** in the first week).
    - Start **F-75** diet (low calorie, low protein to prevent refeeding syndrome).
2. **Rehabilitation Phase (Weeks 2–6):**
    - Transition to **F-100** or RUTF.
    - Start **Iron** supplementation once the child is eating well.
    - Sensory stimulation and emotional support.

EXAMINER'S VIVA

- **Q: Why is Iron contraindicated in the stabilization phase?**
    - A: Free iron can promote bacterial growth and act as a pro-oxidant, worsening the child's fragile metabolic state.
- **Q: How do you distinguish dehydration from wasting in a Marasmic child?**
    - A: Skin turgor is unreliable. Look for **moist mucous membranes** and the **child's sensorium**. If the child is alert and the mouth is moist, the "tenting" of skin is likely due to loss of fat, not dehydration.
- **Q: What is the "Refeeding Syndrome"?**
    - A: A metabolic complication occurring when high-calorie feeding is started too quickly, leading to shifts in Phosphate, Potassium, and Magnesium, resulting in heart failure and arrhythmias.
- **Q: Why do we use ReSoMal instead of standard ORS?**
    - A: Standard ORS has too much Sodium (75 mmol/L) and too little Potassium for a SAM child, which can lead to over-hydration and heart failure.
- **Q: Demonstrate how to measure MUAC.**
    - *Technique:* Find the tip of the shoulder (acromion) and the tip of the elbow (olecranon). Use a string or tape to find the midpoint. Wrap the MUAC tape around this midpoint, neither too tight nor too loose.

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting [Name], a [Age] old male, who presents with a history of significant weight loss over [Duration] and bilateral pedal edema, currently meeting the WHO criteria for Severe Acute Malnutrition."
- **Common Mistake:** Forgetting to mention the **Appetite Test**. In the exam, always state whether the child accepted the feed you offered.
- **Examiner Watch-out:** They will watch how you handle the child. SAM children are often irritable; handle them gently and keep them warm. If you leave the child uncovered after examination, you will lose marks for ignoring the risk of hypothermia.

---

23. Chronic diarrhea

Subject: Gastrointestinal and Liver

This is a classic long case that tests your ability to differentiate between malabsorption, osmotic diarrhea, and secretory diarrhea, while simultaneously assessing your skill in nutritional rehabilitation.

HISTORY

Chief Complaint

- "Loose stools for >4 weeks" (Standard definition of chronic diarrhea).
- Age-specific: In infants, look for "failure to thrive"; in older children, look for "stunted growth" or "abdominal distension."

History of Present Illness

Ask these questions naturally to categorize the diarrhea:

- **Characterize the stool:** "How many times a day? Is the volume large (small bowel) or small (large bowel)? Is it oily, greasy, and difficult to flush (steatorrhea/malabsorption) or watery (osmotic/secretory)?"
- **Diurnal variation:** "Does the diarrhea stop when the child sleeps or is fasted?" (If yes, points to **Osmotic**; if no, points to **Secretory**).
- **Relation to feeds:** "Did this start exactly when you introduced cow's milk (CMA) or wheat/cereal (Celiac)?"
- **Blood/Mucus:** "Is there visible blood?" (Suggests IBD, Polyps, or Chronic Amebiasis).
- **Associated GI symptoms:** "Is there significant bloating or excessive flatus?" (Carbohydrate malabsorption). "Does the child have recurrent vomiting?"
- **Systemic features:** "Is there a persistent cough?" (Think Cystic Fibrosis). "Are there skin rashes or mouth sores?" (Think Acrodermatitis Enteropathica or IBD). "Is there joint pain?" (IBD).

Relevant Background History

- **Antenatal/Birth:** Delayed passage of meconium (>48 hours)? (Think Hirschsprung's with enterocolitis or CF).
- **Dietary History [CRITICAL]:**
    - Detailed 24-hour recall to calculate calorie and protein deficit.
    - Exact timing of weaning and introduction of gluten.
    - History of "juice" or "sugar-sweetened beverage" intake (Toddler's diarrhea).
- **Past History:** Recurrent pneumonia (CF/Immunodeficiency); Previous abdominal surgery (Short bowel syndrome).
- **Family History:** Celiac disease, IBD, or early sibling deaths (Primary Immunodeficiency).
- **Socioeconomic:** Water source and sanitation (Giardiasis/Parasites).

EXAMINATION

General Survey

- **The "First Look":** Is the child "Pot-bellied" with wasted buttocks? (Classic malabsorption/Celiac). Is the child irritable (Celiac) or apathetic (Kwashiorkor)?
- **Nutritional Assessment:** Look for "Baggy pants" appearance of the skin around the gluteal region (loss of subcutaneous fat).
- **Skin:**
    - Perianal/Periorificial dermatitis (Zinc deficiency/Acrodermatitis Enteropathica).
    - Hyperpigmentation over knuckles (Vitamin B12 deficiency).
    - Easy bruisability (Vitamin K deficiency).
- **Facies:** "Old man facies" (Marasmus). Check for pale conjunctiva and Bitot's spots (Vitamin A).

Vital Signs and Anthropometry

- **Vitals:** Check for orthostatic hypotension (chronic dehydration).
- **Anthropometry [EXAM CORE]:**
    - Measure Weight, Length/Height, Head Circumference, and MUAC.
    - **Plot on WHO Growth Charts.**
    - Calculate: Weight-for-Height (Wasting - acute malnutrition) and Height-for-Age (Stunting - chronic malnutrition). Chronic diarrhea almost always leads to stunting.

Peripheral Signs

- **Hands:**
    - **Clubbing:** [EXAMINER FAVORITE] Look for loss of Schamroth's window. In GI cases, this suggests IBD, Celiac, or Liver Cirrhosis.
    - **Koilonychia/Spoon nails:** Iron deficiency due to malabsorption.
- **Eyes:** Pallor (Iron/B12/Folate malabsorption); Xerophthalmia (Vitamin A).
- **Edema:** Check pedal edema. If present, think **Protein Losing Enteropathy (PLE)** or severe malnutrition.
- **Mouth:** Aphthous ulcers (IBD/Celiac); Angular stomatitis (B-complex deficiency); Glossitis (Beefy red tongue in B12 deficiency).

Systemic Examination — Primary System (Abdomen)

- **Inspection:**
    - **Distension:** Is it generalized? Note if the flanks are full.
    - **Everted Umbilicus:** Suggests ascites.
    - **Visible Peristalsis:** From left to right in the upper abdomen (not typical for diarrhea, but check for obstruction).

    - **Perianal Exam:** [DO NOT OMIT] Look for skin tags, fistulae, or fissures (Crohn's disease).

- **Palpation:**
    - **Tenderness:** Localized (IBD) or generalized.
    - **Organomegaly:** Check for hepatomegaly (could be fatty liver due to malnutrition or part of a systemic disease like Gaucher's).
    - **Masses:** Feel for "doughy" abdomen (Abdominal TB).
- **Percussion:**
    - **Tympany:** Over distended loops (gas).
    - **Shifting Dullness:** If you suspect ascites (Hypoproteinemia).
- **Auscultation:**
    - Hyperactive bowel sounds (Borborygmi) are common in malabsorption.

Systemic Examination — Secondary Systems

- **Chest:** Auscultate for crepitations (CF/Immunodeficiency).
- **Joints:** Swelling or restricted range of motion (Extra-intestinal manifestations of IBD).

SIGNS THAT CLINCH THE DIAGNOSIS [EXAMINER FAVORITES]

- **Gluteal Wasting:** Indicates severe, chronic calorie/protein malabsorption.
- **Dermatitis Herpetiformis:** Intensely pruritic vesicles on elbows/knees; pathognomonic for Celiac Disease.
- **Perianal Fistulae/Tags:** Highly suggestive of Crohn's Disease.
- **Positive Hair Signs:** "Flag sign" (alternating bands of light and dark hair) indicating intermittent periods of malnutrition.
- **Abdominal Doughiness:** Suggests Tuberculous Peritonitis.

SEVERITY ASSESSMENT [SEVERITY MARKER]

- **Clinical Dehydration:** Sunken eyes, slow skin pinch, lethargy.
- **Electrolyte Imbalance:** Muscle weakness/paralytic ileus (Hypokalemia); Trousseau/Chvostek signs (Hypocalcemia/Hypomagnesemia).
- **Growth Failure:** Height-for-age < -3 Z-scores.

DIAGNOSIS

Diagnostic Criteria

- **Chronic Diarrhea:** >14 days (WHO) or >4 weeks (most GI societies).
- **Celiac Disease:** ESPGHAN 2020 Criteria (Symptoms + TTG IgA >10x ULN + Positive EMA; Biopsy can be skipped if criteria met).

Differentials

1. **Celiac Disease:** Growth failure, irritability, distension, gluten link.
2. **Post-Infectious Malabsorption (Tropical Sprue):** Follows an acute episode, secondary lactose intolerance.
3. **Cystic Fibrosis:** Steatorrhea + Recurrent pneumonia + failure to thrive.
4. **Giardiasis:** Bloating, greasy stools, foul-smelling flatus.
5. **Inflammatory Bowel Disease (IBD):** Blood in stool, fever, joint pain, perianal tags.

Investigations

- **Tier 1 (Screening):** Stool Routine (pH <5.5 and Reducing substances >0.5% suggest Carbohydrate malabsorption); Stool fat (Sudan III stain); CBC (Anemia); Serum Albumin.
- **Tier 2 (Specific):** Anti-tissue Transglutaminase (tTG) IgA; Sweat Chloride test; Stool for Giardia antigen.
- **Tier 3 (Advanced):** Upper GI Endoscopy + D2 biopsy (Marsh grading for Celiac); Colonoscopy (for IBD); Fecal Calprotectin (marker of gut inflammation).

Management Outline

1. **Stabilization:** Correct dehydration (ORS/IVF) and electrolytes (K+, Mg++, Ca++).
2. **Nutritional Rehabilitation:**
    - High calorie, high protein diet.
    - Vitamin/Mineral supplementation (Zinc, Vitamin A, Iron, Folic acid).
3. **Specific Therapy:**
    - Gluten-free diet (Celiac).
    - Pancreatic enzyme replacement (CF).
    - Antibiotics/Antiparasitics (Metronidazole for Giardia).
4. **Monitoring:** Weight gain (the best indicator of recovery).

EXAMINER'S VIVA

1. **How do you differentiate Osmotic from Secretory diarrhea at the bedside?**

- *Answer:* Ask if the diarrhea persists during fasting or at night. Osmotic diarrhea (like lactose intolerance) stops when the offending agent is removed; Secretory diarrhea (like Cholera or certain tumors) continues regardless of oral intake.

2. **What is the "Stool Osmotic Gap" and how is it used?**
    - *Answer:* Gap = 290 - 2([Na] + [K]). A gap >100 mOsm/kg suggests Osmotic; <50 mOsm/kg suggests Secretory.
3. **Why do children with Celiac disease have a "pot-belly"?**
    - *Answer:* It is a combination of generalized muscle wasting (hypotonia of abdominal wall) and distension of bowel loops due to fermentation of unabsorbed carbohydrates.
4. **How do you perform a skin pinch test correctly?**
    - *Answer:* Pinch the skin of the abdomen halfway between the umbilicus and the flank for 1 second. Do not use fingertips; use the pads of the thumb and index finger. Note if it returns immediately, slowly (<2s), or very slowly (>2s).
5. **What is the significance of "Reducing Substances" in stool?**
    - *Answer:* It indicates unabsorbed sugars (lactose, glucose, fructose) which are being fermented by colonic bacteria.

LONG CASE PRESENTATION TIPS

- **The Opening:** "I am presenting [Name], a [Age] old male, with a 2-month history of large-volume, greasy stools associated with significant weight loss and irritability, starting 3 months after the introduction of wheat-based porridges." (This immediately tells the examiner you are thinking Celiac).
- **Common Mistake:** Forgetting to perform a perianal examination. In a chronic diarrhea case, this is a "fail" point for many examiners.
- **Focus:** Spend less time on the "diarrhea" and more time on the "nutritional consequences" (the deficiencies and growth failure). That is what we treat in the ward.

---

24. Malabsorption

Subject: Gastrointestinal and Liver

This is a classic "bread and butter" long case for the MD Pediatrics practical exam. It tests your ability to integrate nutrition, growth, and systemic examination. When you see a child with chronic diarrhea and failure to thrive, your mind must immediately differentiate between **Intraluminal Maldigestion** (e.g., Pancreatic Insufficiency/CF), **Mucosal Malabsorption** (e.g., Celiac Disease), and **Post-mucosal Obstruction** (e.g., Lymphangiectasia).

HISTORY

Chief Complaint

- "Looseness of stools" or "increased frequency" for >4 weeks.
- "Failure to gain weight" or "weight loss" despite adequate intake.
- "Abdominal distension" or "bloating."

History of Present Illness

Ask these questions naturally to differentiate the site of malabsorption:

- **Stool Character:** "Does the stool look oily, greasy, or difficult to flush?" (Steatorrhea points to pancreatic/biliary issues). "Is it watery, frothy, and causes a skin rash on the buttocks?" (Osmotic diarrhea/Carbohydrate malabsorption).
- **Timing:** "Did this start exactly after introducing wheat/semolina (suji)?" (Celiac disease usually presents 6–24 months of age).
- **Appetite:** "Is the child always hungry (voracious) or refuses to eat?" (Celiac often has anorexia; CF/Pancreatic insufficiency often has a voracious appetite).
- **Abdominal Symptoms:** "Does the belly swell up like a drum after meals?" (Fermentation in the gut).
- **Systemic Clues:** "Does the child have a chronic cough or recurrent pneumonia?" (Cystic Fibrosis). "Does the child have skin rashes or hair loss?" (Acrodermatitis enteropathica/Zinc deficiency).

Relevant Background History

- **Dietary History (Crucial):** Perform a 24-hour recall. Calculate the calorie and protein gap. Ask specifically about the timing of gluten introduction.
- **Birth History:** Was there delayed passage of meconium? (Cystic Fibrosis/Meconium ileus).
- **Family History:** Any siblings with similar chronic diarrhea or early childhood deaths? (Celiac, CF, Primary Immunodeficiency).
- **Socioeconomic:** To rule out "Environmental Enteropathy" or simple "Secondary Malabsorption" due to repeated infections and poverty.

EXAMINATION

General Survey

- **The "Celiac Facies":** Look for a sad, fretful expression with a prominent forehead and wasted cheeks.
- **Attitude:** Is the child irritable? (Common in Celiac).
- **Wasting:** Look for the "baggy pants" appearance of the gluteal region. [SEVERITY MARKER]
- **Skin:** Look for hyperpigmented dermatitis (Niacin/B3), easy bruising (Vitamin K), or dry/scaly skin (Essential Fatty Acids).

Vital Signs and Anthropometry

- **Technique:** Use an infantometer for <2 years, stadiometer for >2 years. Measure Mid-Upper Arm Circumference (MUAC).
- **Interpretation:** Plot on WHO Growth Charts. In malabsorption, **Weight-for-Height** (Wasting) is affected first, followed by **Height-for-Age** (Stunting). If both are severely low, the process is chronic.

Peripheral Signs

- **Hands:**
    - **Clubbing:** [EXAMINER FAVORITE] Use Schamroth's window. If present, think Cystic Fibrosis, Liver Cirrhosis, or IBD.
    - **Koilonychia:** Spoon-shaped nails indicating chronic Iron deficiency.
- **Eyes:** Bitot's spots (Vitamin A), Pallor of palpebral conjunctiva (Iron/B12/Folate).
- **Mouth:** Angular stomatitis (B2), Glossitis (B12/Folate), Aphthous ulcers (Crohn's/Celiac), Dental enamel hypoplasia (Celiac).
- **Edema:** Check pedal edema. If present, it indicates **Protein Losing Enteropathy** or severe malnutrition (Kwashiorkor-like state).

Systemic Examination — Primary System (Abdomen)

- **Inspection:**
    - **Shape:** Look for "Pot-belly" (distended abdomen) contrasting with wasted extremities.
    - **Visible Peristalsis:** May be seen in thin abdominal walls.
    - **Umbilicus:** Everted due to high intra-abdominal pressure from gas/fluid.
- **Palpation:**
    - **Consistency:** The abdomen often feels "doughy" or "full of gas."
    - **Liver/Spleen:** Usually not enlarged in pure Celiac, but hepatomegaly may be seen in CF (cirrhosis) or Gaucher's.
- **Percussion:**
    - **Tympany:** Generalized hyper-resonance due to dilated, gas-filled loops.
    - **Shifting Dullness:** Perform this carefully. If positive, it suggests Ascites (Hypoproteinemia).
- **Auscultation:**
    - Listen for increased bowel sounds (borborygmi).

Systemic Examination — Secondary Systems

- **Chest:** Auscultate for crepitations/wheeze (Cystic Fibrosis).

- **CNS:** Check deep tendon reflexes. Absent DTRs + Ataxia = Vitamin E deficiency.

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **Gluteal Wasting:** The most sensitive sign of muscle protein catabolism in malabsorption.
- **Doughy Abdomen:** Suggests dilated, fluid-and-gas-filled loops of bowel typical of malabsorption.
- **Dermatitis Herpetiformis:** Intensely pruritic vesicles on elbows/knees (Pathognomonic for Celiac).
- **Rectal Prolapse:** Can occur in CF or severe malnutrition with chronic diarrhea.

DIAGNOSIS

Diagnostic Criteria

- **Celiac Disease:** ESPGHAN 2020 Criteria (Symptoms + Anti-tTG IgA >10x ULN + Positive EMA = Diagnosis without biopsy in some cases).
- **Cystic Fibrosis:** Clinical features + Sweat Chloride >60 mmol/L.

Differentials

1. **Celiac Disease:** Gluten-related, irritability, severe wasting.
2. **Cystic Fibrosis:** Respiratory symptoms + Steatorrhea.
3. **Giardiasis:** Chronic diarrhea, bloating, but usually less severe growth failure.
4. **Cow's Milk Protein Allergy (CMPA):** Usually in infants, blood in stools, eczema.
5. **Tropical Enteropathy:** History of poor sanitation, multiple nutrient deficiencies.

Investigations

- **Tier 1 (Bedside/Basic):**
    - Stool for pH (<5.5 = CHO malabsorption), Reducing substances (Positive = Lactose intolerance), Stool fat (Sudan III stain).
    - CBC (Anemia), Peripheral Smear (Dimorphic anemia).
- **Tier 2 (Confirmatory):**
    - Serology: Anti-tTG IgA (Check total IgA first to avoid false negatives).
    - Sweat Chloride test.
    - Endoscopy + D2 Biopsy (Marsh Grading).
- **Tier 3 (Complications):**
    - Serum Albumin, Calcium, Magnesium, Vitamin D levels, INR (Vitamin K).
    - Bone age (X-ray wrist).

Management Outline

1. **Stabilization:** Correct dehydration and electrolyte imbalances (K+, Mg++).
2. **Nutritional Rehabilitation:**
    - High calorie (150% of RDA), High protein.
    - **Celiac:** Strict Gluten-Free Diet (GFD) for life.
    - **CF:** Pancreatic Enzyme Replacement Therapy (PERT) + Fat-soluble vitamins (ADEK).
3. **Micronutrient Supplementation:** Iron, Folic acid, B12, Zinc, and Vitamin A.

EXAMINER'S VIVA

- **Q: Why is the abdomen distended in malabsorption?**
    - A: Due to a combination of hypotonic abdominal muscles (protein deficiency) and fermentation of undigested carbohydrates by colonic bacteria producing gas.
- **Q: How do you perform the "Sudan III" stool test?**
    - A: Mix a drop of stool with Sudan III stain on a slide. Large orange-red droplets indicate neutral fat (maldigestion).
- **Q: What is the "Gold Standard" for Celiac diagnosis?**
    - A: Histopathology of the duodenum (Marsh classification), though ESPGHAN now allows serological diagnosis if tTG is >10x normal.
- **Q: How do you differentiate between Osmotic and Secretory diarrhea?**
    - A: Osmotic diarrhea stops with fasting and has a high stool osmotic gap (>100 mOsm/kg). Secretory diarrhea continues during fasting.
- **Q: Why do we check Total IgA before ordering Anti-tTG IgA?**
    - A: IgA deficiency is more common in Celiac patients. If the child is IgA deficient, the Anti-tTG IgA will be falsely low/negative. In such cases, IgG-based tests (Anti-DGP IgG) are used.

LONG CASE PRESENTATION TIPS

- **The Opening:** "I am presenting [Name], a [Age] old male, with a history of chronic large-volume diarrhea for 6 months, significant weight loss, and abdominal distension, currently having a weight-for-height Z-score of <-3..."
- **Common Mistake:** Forgetting to ask about the "consistency" of the stool. Don't just say "loose"; describe if it's oily or frothy.
- **The "Trap":** If you say the child has Celiac, the examiner will ask if you checked the "Dermatitis Herpetiformis" sites. Always look at the elbows and knees!

Nephrology

25. Nephrotic syndrome

Subject: Nephrology

HISTORY

Chief Complaint

- **Swelling (Edema):** Usually starts in the periorbital region ("puffiness of eyes"), most prominent in the morning, progressing to involve the abdomen (distension), scrotum/labia, and lower limbs.
- **Decreased Urine Output (Oliguria):** Ask about frequency and volume.
- **Frothy/Foamy Urine:** Indicative of significant proteinuria.
- **Duration:** Typically days to weeks.

History of Present Illness

- **Characterizing the Edema:**
    - "When did you first notice the swelling?" (Onset)
    - "Does it disappear by evening and reappear in the morning?" (Diurnal variation – classic for renal edema).
    - "Is it painful or red?" (To rule out cellulitis).
- **Assessing for Complications:**
    - **Infection:** "Has there been any fever, abdominal pain, or vomiting?" (Spontaneous Bacterial Peritonitis is common).
    - **Hypovolemia:** "Is the child excessively thirsty, dizzy, or having cold extremities?"
    - **Thrombosis:** "Any sudden leg swelling, pain, or respiratory distress?"
- **Assessing for Etiology (Primary vs. Secondary):**
    - **Systemic Lupus Erythematosus (SLE):** "Any skin rashes (malar), joint pains, or prolonged fever?"
    - **Post-Streptococcal:** "Was there a sore throat or skin sores 2-3 weeks ago?" (Though NS is rarely post-infectious, Nephritic-Nephrotic overlap exists).
    - **HSP:** "Any rashes on the buttocks or legs?"
- **Negative Predictors:** "Is the urine red or cola-colored?" (Hematuria points toward Nephritic syndrome or MPGN). "Has the blood pressure been high?"

Relevant Background History

- **Past History:**

    - **Crucial:** "Is this the first episode or a relapse?"
    - If relapse: "How many relapses in the last 6 months/year?" (To define Infrequent vs. Frequent Relapse/Steroid Dependency).
    - "What medications was the child on? Dose? Duration?" (Check for steroid toxicity).
- **Birth/Neonatal:** "Was there swelling at birth or in the first 3 months?" (Congenital/Infantile Nephrotic Syndrome – usually genetic).
- **Immunization:** "Was any vaccine given recently?" (Relapses can be triggered by vaccines). "Has the child had Varicella?" (Crucial for steroid safety).
- **Socioeconomic:** "Can the family afford long-term therapy and home urine protein monitoring?"

EXAMINATION

General Survey

- **"The Look":** Observe for the "Puffy Face" and "Slit-like eyes." Is the child irritable (uremia/electrolyte imbalance) or lethargic (hypovolemia)?
- **Nutritional Status:** Look past the edema. Check for **Muehrcke's lines** (transverse white bands on nails) indicating chronic hypoalbuminemia. Check mid-upper arm circumference (MUAC) as weight is unreliable due to fluid.
- **Skin:** Look for striae (steroid use), thinning of skin, or fungal infections (immunosuppression). Check for needle prick marks (frequent hospitalizations).

Vital Signs and Anthropometry

- **Blood Pressure:** [SEVERITY MARKER] Use the correct cuff size (bladder width 40% of arm circumference). Compare with age/sex/height-specific centiles. Hypertension may indicate a non-minimal change pathology.
- **Weight:** Measure daily on the same scale. This is the best indicator of fluid balance.
- **Height:** Plot on growth charts. Chronic steroid use or steroid resistance often leads to growth failure.

Peripheral Signs

- **Eyes:** Check for periorbital edema. Check for **cataracts** (steroid side effect) using a torch.
- **Pulse:** Assess volume. A low-volume, thready pulse with cold peripheries indicates **intravascular volume depletion** (despite total body fluid overload).
- **Lymph Nodes:** Generalized lymphadenopathy might suggest SLE or lymphoma as a secondary cause.
- **Edema:** [TECHNIQUE] Press over the dorsum of the foot, medial malleolus, and the pretibial surface for at least 10 seconds. In children, also check the sacrum if bedridden.

Systemic Examination — Primary System (Abdomen)

- **Inspection:**
    - Distended abdomen, umbilicus may be everted or transverse ("smile" sign).
    - Look for prominent veins (distinguish between IVC obstruction vs. portal hypertension).
- **Palpation:**
    - The abdomen will be "doughy."
    - **Liver/Spleen:** May be difficult to feel if ascites is tense. Use the "dipping" (ballottement) technique.
- **Percussion:**
    - **Shifting Dullness:** [TECHNIQUE] Percuss from umbilicus to the flank. When dullness is reached, have the child turn 45 degrees toward you. Wait 30 seconds for fluid to shift. Percuss again.
    - **Fluid Thrill:** [TECHNIQUE] Only for massive ascites. Need an assistant's hand on the midline to block skin-fold transmission.
- **Auscultation:** Listen for bowel sounds (absent in peritonitis).

Systemic Examination — Secondary Systems

- **Respiratory:** Check for dullness at the bases (pleural effusion) and tachypnea (pulmonary edema or compensation for metabolic acidosis).
- **Cardiovascular:** Check for loud S2 (hypertension) or gallop rhythm (heart failure).

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **Pitting Edema:** Demonstrates the transudative nature of the fluid.
- **Ascites with no signs of Portal Hypertension:** (No caput medusae, no splenomegaly) points toward hypoproteinemia.
- **Leukonychia/Muehrcke's lines:** Confirms chronic low albumin.
- **Steroid Toxicity Signs:** Cushingoid facies, buffalo hump, hirsutism, and striae (indicates a child with frequent relapses or steroid dependence).

Severity Assessment [SEVERITY MARKER]

- **Hypovolemic Shock:** Tachycardia, delayed capillary refill (>3 sec), cold extremities, low BP (late sign).
- **Tense Ascites:** Causing respiratory distress or umbilical hernia.
- **Peritonitis:** Guarding, rigidity, and rebound tenderness.

DIAGNOSIS

Diagnostic Criteria (ISKDC / IPNA)

1. **Nephrotic Range Proteinuria:** >40 mg/m^2/hr or Spot Protein/Creatinine ratio >2 mg/mg.
2. **Hypoalbuminemia:** Serum albumin <2.5 g/dL.
3. **Edema.**
4. **Hyperlipidemia:** Serum cholesterol >200 mg/dL (supportive, not mandatory).

Differentials

1. **Acute Glomerulonephritis (AGN):** Distinguished by gross hematuria, significant hypertension, and less severe edema.
2. **Protein Losing Enteropathy:** Distinguished by history of diarrhea and absence of proteinuria.
3. **Hepatic Cirrhosis:** Distinguished by icterus, splenomegaly, and stigmata of chronic liver disease.
4. **Congestive Heart Failure:** Distinguished by raised JVP, hepatomegaly (tender), and cardiomegaly.

Investigations

- **Tier 1 (Bedside):** Urine dipstick (3+ or 4+ protein). Urine microscopy (look for fatty casts; RBCs >5/hpf suggest non-minimal change).
- **Tier 2 (Confirmatory):** 24-hour urine protein (gold standard but difficult) or spot UPr/UCr. Serum Albumin. Lipid profile.
- **Tier 3 (Etiology/Complications):** C3, C4 levels (low in SLE/MPGN), ANA (if SLE suspected), USG Abdomen (confirm ascites, renal size).

Management Outline

1. **Induction:** Prednisolone 2 mg/kg/day (max 60 mg) for 6 weeks, followed by 1.5 mg/kg alternate days for 6 weeks. [UPDATED: Some protocols now suggest shorter durations].
2. **Supportive:** Salt restriction (no added salt), adequate protein (RDA for age), fluid restriction only if hyponatremic or oliguric.
3. **Diuretics:** Use only if edema is tense or causing distress. Caution: risk of hypovolemia.
4. **Vaccination:** Pneumococcal and Varicella vaccines are mandatory (during remission).

EXAMINER'S VIVA

- **Q: How do you define a "Frequent Relapser"?** A: Two or more relapses in the first six months of initial response, or four or more relapses in any 12-month period.
- **Q: Why are these children prone to infection?** A: Loss of IgG and Complement Factor B in urine, and impaired T-cell function.

- **Q: What is the "Dipping Technique"?** A: In tense ascites, you push your fingers quickly and firmly into the abdomen; the fluid is displaced, and you feel the organ "tap" your fingertips.
- **Q: When would you perform a Renal Biopsy in a child with NS?** A: Age <1 year or >12 years, persistent hypertension, gross hematuria, low C3, or steroid resistance.
- **Q: How do you differentiate between a Steroid Responder and Steroid Resistant?** A: Resistance is defined as failure to achieve remission after 4 weeks of daily prednisolone at 2 mg/kg/day.

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting the case of [Name], a [Age] year old male, known case of [First episode/Relapse] Nephrotic Syndrome, currently presenting with generalized edema and oliguria of [Duration]..."
- **Common Mistake:** Forgetting to check the blood pressure or using a cuff that is too small (gives a falsely high reading).
- **What Examiners Watch For:** Your technique for shifting dullness and how you palpate for the liver in an ascitic child. They also look to see if you checked for steroid side effects (striae, cataracts).

---

26. Chronic kidney disease

Subject: Nephrology

This is a classic long case that tests your ability to integrate multisystem findings. In Chronic Kidney Disease (CKD), the diagnosis is often sitting right there in the growth chart and the skin color before you even touch the patient.

HISTORY

Chief Complaint

- "Swelling of the body" (Edema) – duration and progression.
- "Decreased urine output" or "Increased frequency of urination at night" (Polyuria/Nocturia).
- "Paleness of skin" (Anemia).
- "Poor weight gain or short stature" (Growth failure).
- "Bone deformities" (Rickets/Osteodystrophy).

History of Present Illness

- **Edema:** Ask: "Where did the swelling start? Was it around the eyes in the morning (renal) or feet by evening (cardiac)?" In CKD, edema is often less dramatic than in Nephrotic Syndrome unless there is end-stage heart failure or significant proteinuria.

- **Urinary Symptoms:** Ask: "Does he wake up multiple times at night to pass urine?" (Loss of concentrating ability). Ask about the stream: "Is there straining or a poor stream?" (Points to Posterior Urethral Valves as the etiology).
- **Anemia/Fatigue:** "Does he get tired faster than his peers? Has he started looking pale?"
- **Uremic Symptoms:** Ask: "Is there persistent nausea or early morning vomiting? Any itching (uremic pruritus)? Any unusual sleepiness or twitching?"
- **Bone Pain/Deformity:** "Have you noticed bowing of the legs or pain when walking?"
- **Cardiovascular:** "Any breathlessness or palpitations?" (Anemia or Hypertension).

Relevant Background History

- **Antenatal/Birth:** Crucial. Ask: "Was the liquor low (Oligohydramnios) during pregnancy?" (Suggests renal dysplasia/PUV). "Was any abnormality detected on fetal ultrasound?"
- **Developmental:** CKD causes global developmental delay, especially in motor and cognitive domains.
- **Nutritional:** Detailed 24-hour recall. CKD children are often anorexic. You must calculate the caloric deficit.
- **Family History:** Ask about dialysis, early deaths, or "kidney stones" in the family (Alport syndrome, Cystinosis, Polycystic Kidney Disease).
- **Socioeconomic:** Essential for management planning. "Can the family afford long-term dialysis or transplant?"

EXAMINATION

General Survey

- **The "CKD Look":** Observe for a combination of **Pallor + Sallow Skin Pigmentation** (due to urochrome deposition) + **Growth Retardation**.
- **Activity:** Is the child listless or encephalopathic?
- **Facies:** Look for "Potter facies" (low set ears, flattened nose, prominent epicanthal folds) suggesting chronic oligohydramnios.

Vital Signs and Anthropometry

- **Blood Pressure:** [CRITICAL] Use the right cuff size (bladder width 40% of arm circumference). Measure in both upper and lower limbs to rule out coarctation if HTN is found. Compare against age/sex/height centiles.
- **Respiration:** Look for Kussmaul breathing (deep, sighing respirations) indicating metabolic acidosis.
- **Anthropometry:** Plot Height-for-Age. CKD is a premier cause of **Pathological Short Stature**. Measure Mid-Upper Arm Circumference (MUAC) for wasting.

Peripheral Signs

- **Hands:** Look for **Pallor** of palmar creases. Check for **Mees' lines** or **Muehrcke's lines** (hypoalbuminemia).
- **Pulse:** Check for "Water-hammer" character (Anemia/Fluid overload).
- **Eyes:** Check for **Conjunctival Pallor**. Look for **Band Keratopathy** (calcium-phosphate deposition in the cornea).
- **Lymph Nodes:** Usually not enlarged unless there is a chronic infection or SLE.
- **Edema:** Check the dorsum of the feet and the pretibial area. In infants, check the **sacral area**.

Systemic Examination — Primary System (Renal/Abdomen)

- **Inspection:** Look for fullness in the flanks. Look for scars (previous surgeries for PUV or reflux). Look for a visible suprapubic bulge (distended bladder in PUV).
- **Palpation:**
    - **Bimanual Palpation of Kidneys:** Use one hand posteriorly in the loin and one anteriorly. A palpable kidney in CKD suggests PCKD, Hydronephrosis, or Multicystic Dysplastic Kidney.
    - **Bladder:** Palpate for a thick-walled, palpable bladder (PUV).
- **Percussion:**
    - **Shifting Dullness:** To detect ascites.
    - **Bladder Percussion:** To confirm a full bladder.
- **Auscultation:** Listen for **Renal Bruits** (Renal Artery Stenosis) 2cm above and lateral to the umbilicus.

Systemic Examination — Secondary Systems

- **CVS:** Look for the **Apex Beat** (displaced in LVH due to HTN). Listen for a **Loud S2** (HTN) or a **Hemic Murmur** (Anemia). Listen for a **Pericardial Rub** [SEVERITY MARKER - Uremic Pericarditis].
- **RS:** Listen for crepitations at the bases (Pulmonary edema).
- **Skeletal:** Look for **Rachitic Rosary**, **Harrison's Sulcus**, and **Bowing of legs** (Renal Osteodystrophy).

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **Sallow Skin/Urochrome Pigmentation:** Earthy-brown tint combined with pallor; indicates chronic retention of urinary pigments.
- **Renal Rickets:** Bowing of legs in a child with stunted growth and high BP; distinguishes renal from nutritional rickets.
- **Palpable, Thick-walled Bladder:** In a male child with CKD, this almost certainly points to PUV.

- **Uremic Frost:** (Rare now) White powdery urea crystals on the skin in severe uremia.

Severity Assessment [SEVERITY MARKER]

- **Fluid Overload:** Basal creps, gallop rhythm, hepatomegaly.
- **Uremic Encephalopathy:** Altered sensorium, asterixis (flapping tremors), seizures.
- **Acidosis:** Kussmaul breathing.
- **Hyperkalemia:** Weakness, peaked T-waves on ECG (this is a clinical emergency).

DIAGNOSIS

Diagnostic Criteria

- **KDIGO Criteria:** Kidney damage or GFR < 60 mL/min/1.73m^2 for **> 3 months**.
- Staging is based on GFR (Stage 1 to 5).

Differentials

1. **Chronic Glomerulonephritis:** History of hematuria, significant proteinuria, and HTN.
2. **Obstructive Uropathy (e.g., PUV):** Male child, poor stream, palpable bladder/kidneys.
3. **Juvenile Nephronophthisis:** Polyuria, polydipsia, normal-sized kidneys, no HTN until late.
4. **Reflux Nephropathy:** History of recurrent UTIs and febrile illnesses.

Investigations

- **Tier 1:** CBC (Normocytic normochromic anemia), Urine R/M (isosthenuria – fixed Sp. Gravity 1.010), Serum Electrolytes (High K, Low Na, Low Ca, High PO4).
- **Tier 2:** BUN/Creatinine (calculate GFR using **Schwartz Formula**:). Ultrasound (Small echogenic kidneys with loss of CMD).
- **Tier 3:** PTH levels (High in secondary hyperparathyroidism), Vitamin D levels, Echocardiography (LVH), Bone X-rays (Subperiosteal resorption).

Management Outline

1. **Diet:** High calorie (100% RDA), protein restriction (only if not on dialysis), low potassium/phosphate.
2. **Anemia:** Erythropoietin (EPO) injections + Oral/IV Iron.
3. **Bone Health:** Phosphate binders (Calcium carbonate) + Active Vitamin D (Calcitriol).
4. **Acidosis:** Oral Sodium Bicarbonate.
5. **Renal Replacement Therapy (RRT):** Peritoneal Dialysis, Hemodialysis, or the gold standard: **Renal Transplant**.

EXAMINER'S VIVA

- **Q: How do you calculate GFR in a child at the bedside?**
    - *A:* Use the Modified Schwartz Formula: .
- **Q: Why is there anemia in CKD?**
    - *A:* Primarily Erythropoietin deficiency; secondarily iron deficiency, chronic inflammation, and shortened RBC lifespan in uremic milieu.
- **Q: What is the first sign of Renal Osteodystrophy on X-ray?**
    - *A:* Subperiosteal resorption of the phalanges (usually the radial side of the middle phalanx).
- **Q: How do you differentiate Renal Rickets from Nutritional Rickets?**
    - *A:* Renal rickets has high phosphate (nutritional has low/normal), presence of HTN, and signs of CKD like sallow skin.
- **Q: What is the significance of "Isosthenuria"?**
    - *A:* It means the kidney has lost the power to concentrate or dilute urine; the urine osmolality is fixed at that of plasma (Sp. Gravity ~1.010).

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting the case of [Name], a [Age] year old male, known/suspected case of CKD, currently presenting with [Chief Complaint], with evidence of growth failure and renal osteodystrophy."
- **Mistake:** Forgetting to check the BP or using the wrong cuff size.
- **Mistake:** Not plotting the height on a growth chart. In CKD, the height is your "biochemical marker" of long-term control.
- **Watch out:** Examiners will watch how you perform bimanual palpation of the kidney. Ensure your posterior hand is lifting the loin while the anterior hand feels for the organ.

---

27. Acute glomerulonephritis

Subject: Nephrology

This is a classic long case. In Acute Glomerulonephritis (AGN), the diagnosis is often made at the bedside through a meticulous history and a focused cardiovascular and renal examination. Pay close attention to the complications—hypertensive emergency and acute kidney injury (AKI)—as these are what the examiners will grill you on.

HISTORY

Chief Complaint

- "Swelling of the face/eyes for 3 days"
- "Passing cola-colored or reddish urine for 2 days"
- "Decreased urine output for 2 days"
- "Headache, vomiting, or seizures" (Indicating hypertensive encephalopathy)

History of Present Illness

- **Edema:** "Where did the swelling start?" (Typically periorbital, morning-heavy in AGN). "Does it involve the feet or abdomen?" (Usually less than Nephrotic Syndrome).
- **Hematuria:** "What is the color of the urine?" (Cola, smoky, or tea-colored suggests glomerular origin; bright red with clots suggests lower urinary tract).
- **Oliguria:** "How many times is he passing urine, and what is the approximate volume?" (Define oliguria: <0.5 ml/kg/hr or <400 ml/m^2/day).
- **Hypertension/Encephalopathy:** "Has he had any severe headaches, blurring of vision, or vomiting?" "Did he have any fits or altered consciousness?"
- **Preceding Infection:** "Did he have a sore throat or fever 1–3 weeks ago?" (Pharyngitis-associated) or "Did he have any skin sores, pyoderma, or 'scabies' 3–6 weeks ago?" (Pyoderma-associated). **Note the latent period; it is crucial.**
- **Systemic Review (To exclude secondary GN):** "Any joint pains or rashes?" (SLE/HSP). "Any prolonged fever or weight loss?" (Endocarditis/Chronic infection). "Any history of hearing loss?" (Alport Syndrome).

Relevant Background History

- **Past History:** Similar episodes in the past? (Recurrent hematuria suggests IgA Nephropathy or Alport's).
- **Family History:** Any family members on dialysis or with early-onset deafness? (Alport's).
- **Socioeconomic:** Overcrowding and poor hygiene (Predisposes to streptococcal pyoderma).

EXAMINATION

General Survey

- **The "Look":** Observe for **"Nephritic Facies"**—periorbital puffiness, a slightly pale, "muddy" complexion. Is the child tachypneic or orthopneic? (Suggests circulatory congestion).
- **Nutritional Status:** Unlike Nephrotic Syndrome, these children are usually well-nourished.
- **Skin:** [EXAMINER FAVORITE] Look specifically for healing crusts of pyoderma, scars of old skin infections, or active scabies lesions on the webs of fingers and feet. Look for the purpuric rash of HSP on the extensor surfaces.

Vital Signs and Anthropometry

- **Blood Pressure:** [CRITICAL] Use the correct cuff size (bladder width 40% of mid-arm circumference). Measure in the supine position. Compare with age, sex, and height-based centiles.
- **Pulse:** Look for high volume and bounding character (fluid overload) or bradycardia (reflex response to acute hypertension).
- **Respiration:** Look for tachypnea and use of accessory muscles (Pulmonary edema).

Peripheral Signs

- **Edema:** Check the pretibial area and sacrum. In AGN, edema is usually **pitting but firm**, and less massive than the "doughy" edema of Nephrotic Syndrome.
- **Pallor:** Often present due to hemodilution (dilutional anemia).
- **JVP:** [EXAMINER FAVORITE] In an older child, check the JVP. It is typically **raised** in AGN due to hypervolemia, unlike Nephrotic Syndrome where it is normal or low.

Systemic Examination — Primary System (Renal/CVS)

- **Inspection:** Check for abdominal distension (ascites is rare but possible).
- **Palpation:**
    - **Liver:** Palpate for tender hepatomegaly (sign of congestive heart failure/fluid overload).
    - **Kidneys:** Usually not palpable. If palpable, consider other diagnoses (e.g., HUS, vein thrombosis).
- **Percussion:** Check for shifting dullness if ascites is suspected.
- **Auscultation (CVS/Lungs):**
    - **Heart:** Listen for an **S3 gallop** at the apex (volume overload).
    - **Lungs:** [SEVERITY MARKER] Listen for **fine crepitations** at the bases, progressing upwards. This indicates pulmonary edema—a medical emergency.

Systemic Examination — Secondary Systems

- **CNS:** Check sensorium and fundoscopy. Look for **papilledema** or retinal hemorrhages if BP is severely high.

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **Periorbital Edema + Hypertension:** The classic "Nephritic" duo.
- **Healing Pyoderma Scars:** Points specifically to Post-Streptococcal GN (PSGN).
- **Raised JVP + S3 Gallop:** Confirms the state of circulatory congestion.
- **Cola-colored urine in a test tube:** Ask the nurse for a sample to show the examiner.

Severity Assessment [SEVERITY MARKER]

- **Hypertensive Emergency:** BP >95th centile + 30mmHg OR symptomatic (headache, seizures).

- **Pulmonary Edema:** Tachypnea, basal creps, orthopnea.
- **Acute Kidney Injury:** Anuria or severe oliguria with rising creatinine.

DIAGNOSIS

Diagnostic Criteria

- **Clinical:** Acute onset of hematuria, edema, and hypertension.
- **Laboratory:** Evidence of preceding Strep infection + Low C3 complement levels.

Differentials

1. **IgA Nephropathy:** Short latent period (1–2 days after URTI), recurrent episodes, normal C3.
2. **HSP Nephritis:** Presence of characteristic purpuric rash, joint pain, and abdominal pain.
3. **Lupus Nephritis:** Multi-system involvement (malar rash, arthritis), low C3 AND low C4.
4. **MPGN:** Persistent low C3 beyond 8 weeks, more severe proteinuria.

Investigations

- **Tier 1 (Bedside):** Urine dipstick (Proteinuria <2+, Hematuria), Urine Microscopy (Dysmorphic RBCs, **RBC casts**—pathognomonic of GN).
- **Tier 2 (Confirmatory):** ASO titer (raised), Anti-DNase B (best for skin infections), **Serum C3 (low in 90% of PSGN)**.
- **Tier 3 (Complications):** Sr. Creatinine, Electrolytes (look for hyperkalemia), Chest X-ray (cardiomegaly, pulmonary congestion).

Management Outline

1. **Fluid Management:** Fluid restriction (Insensible loss + urine output).
2. **Hypertension:** First line: Loop diuretics (Furosemide) to handle volume. Second line: Calcium channel blockers (Nifedipine/Amlodipine).
3. **Antibiotics:** 10-day course of Penicillin/Amoxicillin to eradicate nephritogenic strain (does not change course of GN but prevents spread).
4. **Monitoring:** Daily weight (best indicator of fluid status), BP monitoring, intake-output charting.

EXAMINER'S VIVA

- **Q: Why is there anemia in AGN?**
    - A: It is primarily **dilutional anemia** due to ECF expansion, though a mild hemolytic component can exist.
- **Q: How do you differentiate glomerular from non-glomerular hematuria?**

    - A: Glomerular: Smoky/Cola color, RBC casts, >20% dysmorphic RBCs, no clots. Non-glomerular: Bright red, clots present, isomorphic RBCs.

- **Q: What is the natural history of C3 levels in PSGN?**
    - A: It drops early and **must return to normal by 6–8 weeks**. If it remains low, consider MPGN or Lupus.
- **Q: When would you consider a Renal Biopsy in AGN?**
    - A: [EXAMINER FAVORITE] 1. Normal C3 at onset. 2. Persistent low C3 >8 weeks. 3. Rapidly rising creatinine (RPGN). 4. Massive proteinuria (Nephrotic range). 5. Absence of Strep infection evidence.
- **Q: Technique: How do you measure the liver span in a child?**
    - A: (Demonstrate) Percuss upper border (usually 5th ICS), palpate lower border, measure distance in mid-clavicular line. Essential to differentiate "palpable" liver from "enlarged" liver.

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting [Name], a [Age] year old male, who presented with acute onset of periorbital edema and hematuria following a skin infection 3 weeks ago, currently complicated by hypertension but without signs of heart failure."
- **Common Mistake:** Forgetting to check the BP in the lower limbs if hypertension is found (to rule out Coarctation).
- **Watch for:** The examiner will watch how you handle the stethoscope—always warm the diaphragm before touching the child's chest.

---

28. Posterior urethral valves

Subject: Nephrology

This is a classic "Surgical-Medical" crossover case in Pediatric Nephrology. In the exam, you are often given a male infant or toddler with a palpable bladder and poor growth. You must demonstrate that you can distinguish between a simple UTI and a structural obstructive uropathy.

HISTORY

Chief Complaint

- **Infant/Toddler:** Poor weight gain, crying during micturition, weak urinary stream, or unexplained fever (UTI).
- **Older Child:** Daytime/nighttime wetting (secondary enuresis), frequency, urgency, or a palpable abdominal mass.

History of Present Illness

Ask these questions naturally to the mother:

- **The Stream:** "When he passes urine, does it come out in a strong arc, or does it just dribble down his legs?" (Dribbling suggests obstruction).
- **The Effort:** "Does he have to strain or push with his tummy muscles to start peeing?"
- **The Bladder:** "After he finishes peeing, does his lower belly still feel hard or full?"
- **The UTI Cluster:** "Has he had high-grade fevers with shivering? Was the urine foul-smelling or cloudy?"
- **Renal Failure Symptoms:** "Is he unusually tired? Is he breathing fast even when resting? Does he look more pale than his siblings?" (Suggests CKD/Metabolic acidosis).
- **Polyuria:** "Does he drink excessive amounts of water and soak through more diapers than usual?" (Loss of urinary concentrating ability in obstructive uropathy).

Relevant Background History

- **Antenatal (Crucial):** "Was the 'water' (liquor) around the baby low during pregnancy? Were any scans done? Did they show 'swollen kidneys' (hydronephrosis) or a 'thick bladder'?"
- **Birth:** Was he born preterm? (PUV is often associated with prematurity due to early induction for oligohydramnios).
- **Developmental:** Gross motor delay is common if the child has CKD/Renal Osteodystrophy.
- **Nutritional:** Detailed calorie count. These children often have "Renal Rickets" and failure to thrive.

EXAMINATION

General Survey

- **Activity:** Is the child irritable (uremia) or tachypneic (acidosis)?
- **Nutritional Status:** Look for "Potter facies" features (flattened nose, recessed chin) if severe/neonatal. Check for visible wasting of gluteal muscles.
- **Skin:** Look for sallow complexion (earthy hue of CKD) and scratch marks (uremic pruritus).

Vital Signs and Anthropometry

- **Blood Pressure:** [CRITICAL] Use the correct cuff size (covering 2/3rd of the upper arm). Measure in all four limbs if hypertensive. PUV is a common cause of pediatric hypertension.
- **Respiration:** Look for deep, acidotic breathing (Kussmaul).
- **Growth:** Plot Weight, Height, and Head Circumference. Height is usually more affected than weight in chronic PUV (Renal Stunting).

Peripheral Signs

- **Hands:** Look for widening of wrists (Renal Rickets).

- **Pallor:** Check palmar creases and conjunctiva. Anemia is common due to EPO deficiency in CKD.
- **Edema:** Usually absent unless the child has progressed to end-stage renal disease or has severe hypoalbuminemia from chronic infection.
- **Rachitic Rosary:** Feel the costochondral junctions for beads.

Systemic Examination — Primary System (Abdomen)

Position: Supine, knees slightly flexed, bladder should NOT be emptied just before the exam.

- **Inspection:**
    - Look for fullness in the suprapubic area.
    - Look for visible peristalsis or "flank fullness" (bilateral hydronephrosis).
    - Check the urinary meatus for stenosis (to rule out other causes of poor stream).
- **Palpation:**
    - **The Bladder:** Use the ulnar border of your hand, starting from the umbilicus and moving downwards. A PUV bladder is typically **thick-walled, firm, and does not disappear after voiding** (expressed as "palpable post-void").
    - **The Kidneys:** Bimanual palpation (ballottement). If kidneys are palpable, note if they are smooth (hydronephrosis) or cystic.
- **Percussion:**
    - Percuss the bladder to define the upper limit. A dull note confirms it is a solid/fluid-filled structure and not gas-filled bowel.
- **Auscultation:**
    - Listen for renal bruits (though rare in PUV, it's a PG-level skill for any hypertensive child).

Systemic Examination — Secondary Systems

- **Respiratory:** Auscultate for crepitations (fluid overload) or features of pulmonary hypoplasia in neonates.
- **CVS:** Loud A2 (hypertension), flow murmurs (anemia).
- **Skeletal:** Check for bowing of legs or wrist widening (Renal Osteodystrophy).

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **Palpable, Thick-walled Bladder:** In a male child, this is PUV until proven otherwise.
- **Poor Urinary Stream:** Observe the child voiding if possible. A "dribbling" stream is pathognomonic.
- **Bilateral Ballottable Kidneys:** Suggests bilateral hydronephrosis secondary to high-pressure bladder.

- **Indwelling Catheter Check:** If the child is already catheterized, ask to see the urine. Is it clear? Is the bladder still palpable? (If yes, suggests a thick-walled, non-compliant bladder).

Severity Assessment [SEVERITY MARKER]

- **Serum Creatinine:** Elevated for age indicates renal parenchymal damage.
- **Failure to Thrive:** Indicates chronic disease.
- **Rickets:** Indicates long-standing CKD (Mineral Bone Disorder).
- **Oliguria/Anuria:** Emergency sign of acute-on-chronic renal failure.

DIAGNOSIS

Diagnostic Criteria

- **Gold Standard:** Voiding Cystourethrogram (VCUG).
- **Findings:** Dilated and elongated posterior urethra, "spinning top" appearance of the bladder neck, bladder trabeculations, and often Vesicoureteral Reflux (VUR).

Differentials

1. **Neurogenic Bladder:** Look for spinal dysmorphism (sacral dimple, tuft of hair).
2. **Prune Belly Syndrome:** Triad of absent abdominal muscles, undescended testes, and urinary tract dilation.
3. **Severe Phimosis:** Obvious on local examination of the penis.

Investigations

- **Tier 1:** Urine R/M (pus cells), S. Creatinine, Electrolytes (look for hyperkalemia/acidosis), Ultrasound (KUB) showing "Keyhole Sign" (dilated posterior urethra).
- **Tier 2: VCUG** (after stabilizing and clearing infection). This is the definitive diagnostic step.
- **Tier 3:** DMSA scan (to look for renal scarring/functional cortical mass).

Management Outline

1. **Stabilization:** Bladder drainage with a small-gauge (5F or 6F) feeding tube (not a Foley, as the balloon can irritate the trigone).
2. **Medical:** Correct dehydration, treat UTI with IV antibiotics, and manage hyperkalemia.
3. **Surgical: Primary Endoscopic Valve Ablation** (Treatment of choice).
4. **Alternative:** If the child is too small for an endoscope, a temporary **Vesicostomy** is performed.
5. **Long-term:** Monitoring for "Bladder Valve Syndrome" (non-compliant bladder) and CKD progression.

EXAMINER'S VIVA

- **Q: Why don't we use a Foley catheter for initial drainage?**
    - *A:* The balloon of the Foley can rest on the trigone, causing bladder spasms and further obstructing the ureters, potentially worsening the hydronephrosis.
- **Q: What is the "Keyhole Sign" on Ultrasound?**
    - *A:* It is the appearance of the dilated posterior urethra (the keyhole) situated below the thick-walled bladder.
- **Q: What is the most common cause of CKD in male children?**
    - *A:* Posterior Urethral Valves.
- **Q: How do you measure BP in an infant?**
    - *A:* Use an oscillometric device (like Dinamap) or Doppler. Ensure the child is quiet; the cuff bladder should encircle 80-100% of the arm circumference.
- **Q: Even after valve ablation, why does the creatinine sometimes stay high?**
    - *A:* Due to "Renal Dysplasia" (irreversible damage during fetal development) or "Valve Bladder Syndrome" (permanent loss of bladder elasticity).

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting the case of [Name], a [Age] old male child, who presented with a history of poor urinary stream and failure to thrive, found to have a palpable, thick-walled bladder on examination..."
- **Mistake:** Forgetting to examine the spine. If you miss a meningomyelocele, you miss the diagnosis of neurogenic bladder.
- **Mistake:** Not checking the blood pressure. In a renal case, not checking BP is an automatic "fail" in many centers.
- **Observation:** Examiners watch how you palpate the bladder. Use a light touch first; a full bladder is tender. If you poke it aggressively, the child will cry, and you won't be able to feel the kidneys.

Hematology

29. Thalassemia major

Subject: Hematology

This is a classic "bread and butter" long case for the MD Pediatrics practical exam. While the diagnosis is often obvious from the foot of the bed, the examiner is looking for your ability to assess the **complications of the disease** and the **complications of the treatment** (iron overload and transfusion-transmitted infections).

HISTORY

Chief Complaint

- "Pallor noticed since [Age - usually 6 months to 2 years]"
- "Requirement of repeated blood transfusions every [Number] weeks"
- "Increasing abdominal girth or yellowish discoloration of eyes"

History of Present Illness

Focus on the "Thalassemia Phenotype" and "Transfusion Burden":

- **Onset of Pallor:** "At what age did you first notice the child looking pale?" (If <6 months, think Diamond-Blackfan; if >2 years, think Thalassemia Intermedia).
- **Transfusion History:** "How many transfusions has the child received in total? How frequent are they now?" (Calculate the transfusion index: ml/kg/year of packed RBCs. >200 ml/kg/year suggests hypersplenism).
- **Growth and Development:** "Have you noticed the child is shorter than peers? Has there been a delay in puberty?" (Suggests iron overload in pituitary/gonads).
- **Iron Chelation:** "Which medicine are you giving to remove iron? How many days a week? Any side effects like blurring of vision or hearing loss?" (Crucial for assessing compliance).
- **Pressure Symptoms:** "Is the abdomen getting so large it interferes with breathing or eating?" (Massive splenomegaly).
- **Bone Changes:** "Have you noticed any change in facial features or forehead shape?" (Extramedullary hematopoiesis).
- **Infections:** "Any history of frequent fever?" (Post-splenectomy sepsis or Yersinia infection from Desferrioxamine).

Relevant Background History

- **Antenatal:** Was any screening (HPLC/CVS) done? (Important for "preventable disease" discussion).
- **Family History:** Consanguinity? History of siblings with similar illness or unexplained early deaths?
- **Socioeconomic:** Can the family afford chelation? (This dictates the prognosis more than the biology).

EXAMINATION

General Survey

- **"Thalassemic Facies":** Stand at the foot of the bed. Look for frontal bossing, prominent malar bones, depressed nasal bridge, and maxillary hypertrophy with protrusion of upper teeth (chipmunk facies).

- **Nutritional Status:** Look for stunting (chronic anemia/endocrinopathy) and wasting.
- **Skin:** Look for a "muddy" or "bronze" complexion (combination of icterus, pallor, and iron deposition/melanosis).

Vital Signs and Anthropometry

- **Pulse:** Tachycardia and a hyperdynamic state (bounding pulse) suggest severe anemia.
- **Growth:** [SEVERITY MARKER] Plot Height and Weight. Thalassemics often fall off the curve by age 8–10 due to iron overload in the GH-IGF1 axis.

Peripheral Signs

- **Pallor:** Check palmar creases and conjunctiva.
- **Icterus:** Check the sclera in natural light (indicates ineffective erythropoiesis/hemolysis).
- **Lymphadenopathy:** Usually absent. If present, think of transfusion-transmitted infections (HIV/CMV).
- **Nails:** Look for koilonychia (though Thalassemia is a hyperferremic state, some may have concomitant nutritional IDA).

Systemic Examination — Primary System (Abdomen)

Position: Supine, knees flexed, bladder empty.

- **Inspection:** Protuberant abdomen, everted umbilicus. Look for a midline scar (splenectomy) or scars in the flanks (cholecystectomy).
- **Palpation:**
    - **Spleen:** Use the "Hooking method" for massive spleens. Measure in centimeters from the left costal margin to the tip along the splenic axis. Feel for the splenic notch. [SEVERITY MARKER] A massive spleen (>6cm) suggests hypersplenism.
    - **Liver:** Measure the liver span. A firm, non-tender hepatomegaly is common due to extramedullary hematopoiesis and hemosiderosis.
- **Percussion:** Confirm the upper border of the liver (usually 5th ICS). Percuss Traube’s space (will be dull).
- **Auscultation:** Listen for a splenic rub (infarction).

Systemic Examination — Secondary Systems

- **CVS:** Look for a hyperdynamic apex beat. Listen for a **Hemic Murmur** (soft, systolic, ejection type at the pulmonary area). Check for signs of Heart Failure (S3, basal creps) due to siderotic cardiomyopathy.
- **Respiratory:** Basal crepitations if in heart failure.
- **Endocrine:** Assess Tanner staging (delayed puberty is the most common endocrine complication).

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **Frontal Bossing/Malar Prominence:** Indicates marrow expansion due to ineffective erythropoiesis.
- **Massive Splenomegaly with Notch:** Differentiates from other causes of abdominal lumps.
- **Muddy Complexion:** The "triad" of pallor, icterus, and hyperpigmentation.
- **Transfusion Siderosis Signs:** Darkening of skin folds and scars.

DIAGNOSIS

Diagnostic Criteria

- **Clinical:** Anemia, hepatosplenomegaly, facies.
- **Laboratory:** Hb electrophoresis or HPLC showing Absence/Reduction of HbA, increased HbF (>90% in non-transfused TM).

Differentials

1. **Thalassemia Intermedia:** Later onset (>2 years), maintains Hb 7–10 g/dL without regular transfusions.
2. **Sickle-Thalassemia:** Bone pains/vaso-occlusive crises, splenomegaly (unlike pure Sickle Cell).
3. **Hereditary Spherocytosis:** Family history (autosomal dominant), splenomegaly, but usually no thalassemic facies.

Investigations

- **Tier 1:** CBC (Low Hb, Low MCV/MCH, High RDW), Peripheral Smear (Target cells, nucleated RBCs, anisopoikilocytosis).
- **Tier 2: HPLC (Gold Standard)**. Serum Ferritin (aim to keep <1000 ng/ml).
- **Tier 3:** T2* MRI of Heart and Liver (to quantify iron overload), Liver Function Tests (ALT for hepatitis), Viral markers (HBsAg, Anti-HCV, HIV).

Management Outline

1. **Transfusion:** Leucodepleted PRBCs. Target pre-transfusion Hb 9.5–10.5 g/dL.
2. **Chelation:** Start when Ferritin >1000 or after 10–20 transfusions.
    - Oral: Deferasirox (20–40 mg/kg/d) or Deferiprone.
    - SC: Desferrioxamine.
3. **Splenectomy:** If transfusion requirement >200 ml/kg/year. (Must give Pneumococcal/Meningococcal/H.influenzae vaccines 2 weeks prior).
4. **Cure:** Hematopoietic Stem Cell Transplant (HSCT) - best results if done early (Lucarelli Class I).

EXAMINER'S VIVA

- **Q: Why do thalassemics have a depressed nasal bridge?**
    - A: It is due to the expansion of the erythroid marrow within the maxillary and frontal bones, leading to cortical thinning and overgrowth of the facial bones.
- **Q: What is "Ineffective Erythropoiesis"?**
    - A: It is the destruction of erythroid precursors within the bone marrow itself due to the precipitation of unpaired alpha-globin chains, which cause membrane damage.
- **Q: How do you monitor for Desferrioxamine toxicity?**
    - A: Annual audiometry (high-frequency sensorineural hearing loss) and ophthalmological exam (retinopathy).
- **Q: What is the "Mentzer Index" and is it useful here?**
    - A: MCV/RBC count. <13 suggests Thalassemia trait; >13 suggests IDA. In Thalassemia Major, it is not used as the diagnosis is usually overt.
- **Q: Why do we keep pre-transfusion Hb at 9.5 g/dL?**
    - A: To suppress the child's own ineffective erythropoiesis, thereby preventing bone deformities and excessive iron absorption from the gut.

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting [Name], a [Age] year old male, a known case of Thalassemia Major on hypertransfusion therapy, currently presenting with [Chief Complaint]..."
- **Mistake:** Forgetting to calculate the "Transfusion Index." Examiners love numbers.
- **Mistake:** Not checking the vaccination status (especially Hepatitis B and post-splenectomy vaccines).
- **Watch for:** The examiner will watch how you palpate the spleen. Ensure you start from the Right Iliac Fossa and move diagonally toward the Left Hypochondrium. **Never** start palpating in the middle of the abdomen.

---

30. Sickle cell disease

Subject: Hematology

HISTORY: SICKLE CELL DISEASE (SCD)

Chief Complaint

- **Pain:** "Bone pain" or "stomach pain" (Vaso-occlusive crisis - VOC). Note the site, duration, and frequency.
- **Pallor:** Noticed by parents (Chronic anemia or Sequestration).

- **Jaundice:** Recurrent yellowish discoloration of eyes.
- **Abdominal Swelling:** (Splenomegaly in early childhood, Hepatomegaly later).
- **Fever:** (Increased susceptibility to encapsulated organisms).

History of Present Illness

Focus on the "Phenotype" of the disease—is this a "Pain" phenotype or a "Hemolysis" phenotype?

- **Characterizing the Pain (VOC):**
    - "Where exactly is the pain? Is it in the small bones of hands/feet (Dactylitis - common in infants) or long bones/girdles?"
    - "How many times in the last year has he needed hospital admission for pain?" (Defines severity: >3/year is severe).
    - "What triggered it? Cold weather, dehydration, infection, or strenuous exercise?"
- **Anemia & Hemolysis:**
    - "When was the first time you noticed he was pale?" (SCD usually presents after 4–6 months as HbF drops).
    - "Does the jaundice fluctuate? Is the urine dark (coca-cola colored) during crises?"
- **Neurological (Silent or Overt Stroke):**
    - "Has he ever had a sudden weakness of one side, a seizure, or a change in his school performance/speech?" [SEVERITY MARKER]
- **Respiratory (Acute Chest Syndrome - ACS):**
    - "Has he ever had chest pain with fever and difficulty breathing?" (ACS is a leading cause of mortality).
- **Splenic Sequestration:**
    - "Has there been a sudden increase in pallor accompanied by rapid abdominal swelling and weakness?" (A pediatric emergency).

Relevant Background History

- **Past History:** Number of blood transfusions (assess iron overload risk). History of surgeries (cholecystectomy for gallstones).
- **Birth History:** Was he screened at birth? (NBS - Newborn Screening).
- **Developmental:** Look for "Sickle Cell Dystrophy"—delayed puberty and physical growth are hallmark features.
- **Immunization: CRITICAL.** "Has he received Pneumococcal (PCV13 and PPSV23), Meningococcal, and H. influenzae vaccines? Is he on daily Penicillin prophylaxis?"
- **Family History:** Consanguinity? History of "anemia" or "early sudden deaths" in siblings/cousins. Construct a 3-generation pedigree.

- **Socioeconomic:** Cost of hydroxyurea, distance to a center with blood transfusion facilities.

EXAMINATION

General Survey

- **Sickle Cell Facies:** Look for frontal bossing, prominent maxilla (due to extramedullary hematopoiesis—though less common than in Thalassemia, it occurs in severe SCD).
- **Habitus:** Typically thin, underweight, with long spindly extremities (Asthenic build).
- **Puberty:** Assess Tanner staging; delay is very common.

Vital Signs and Anthropometry

- **Heart Rate:** Tachycardia is common due to chronic anemia.
- **Temperature:** Fever must be taken seriously (Sepsis risk due to autosplenectomy).
- **Growth:** Plot Weight and Height. You will often see the "Sickle lag"—falling below the 5th centile.

Peripheral Signs

- **Hands:**
    - **Dactylitis:** Look for residual thickening of metacarpals/metatarsals.
    - **Clubbing:** Can occur in chronic ACS/pulmonary hypertension.
    - **Pallor:** Check palmar creases.
- **Eyes:**
    - **Icterus:** Best seen in the upper bulbar conjunctiva.
    - **"Comma-shaped" vessels:** [EXAMINER FAVORITE] Use a slit lamp or ophthalmoscope to look at the bulbar conjunctiva for isolated, comma-shaped vascular segments (due to sickling in small vessels).
- **Lymph Nodes:** Usually not enlarged in SCD; if present, consider infection or other pathology.
- **Leg Ulcers:** [EXAMINER FAVORITE] Look around the medial/lateral malleoli in adolescents. These are chronic, punched-out, and slow to heal.

Systemic Examination — Primary System (Abdomen)

- **Inspection:** Scaphoid or distended? Look for surgical scars (cholecystectomy).
- **Palpation:**
    - **Spleen:** In infants/toddlers, the spleen is often enlarged. If you find a massive spleen in an older child (>6–8 years), think of **HbSC disease** or **S-β Thalassemia**, because "pure" SS disease usually results in a shrunken, fibrotic **Autosplenectomy** by age 5.
    - **Liver:** Often enlarged (2–4 cm) due to extramedullary hematopoiesis or congestion.

- **Percussion:** Confirm liver span. Check for shifting dullness if you suspect heart failure/cirrhosis (rare).

Systemic Examination — Secondary Systems

- **Cardiovascular:**
    - **Hyperdynamic Precordium:** Visible apical impulse.
    - **Murmurs:** Hemic murmur (systolic, grade 2/6 at the left upper sternal border).
    - **P2:** Loud P2 suggests Pulmonary Hypertension [SEVERITY MARKER].
- **Respiratory:** Look for signs of chronic lung disease (crepitations, tachypnea).
- **CNS:** Full neurological exam to rule out focal deficits from a previous stroke.

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **The "Disappearing Spleen":** A documented history of a palpable spleen in infancy that is no longer palpable in a school-aged child (Autosplenectomy).
- **Dactylitis (Hand-Foot Syndrome):** Symmetrical swelling of the dorsum of hands/feet in an infant.
- **Frontal Bossing + Icterus + Splenomegaly:** The classic "Hemolytic" triad.
- **Gnathopathy:** Prominent maxilla with malocclusion of teeth.

Severity Assessment

- **Frequent VOC:** >3 hospitalizations per year.
- **History of ACS or Stroke.**
- **Transfusion dependency.**
- **TCD (Transcranial Doppler) Velocity:** >200 cm/sec (High risk for stroke).

DIAGNOSIS

Diagnostic Criteria

- **Gold Standard:** Hb Electrophoresis or HPLC (High-Performance Liquid Chromatography).
- **Pattern:** HbS > 80%, HbF variable (higher is better), HbA2 < 3.5%, **HbA is absent** in SS disease.

Differentials

1. **Sickle-β^0 Thalassemia:** Very similar clinically; distinguished by low MCV and high HbA2 (>4.5%) on HPLC.
2. **HbSC Disease:** Milder anemia, significant splenomegaly persists into adulthood, more retinopathy.
3. **Hereditary Spherocytosis:** Positive family history (autosomal dominant), spherocytes on smear, negative sickling test.

Investigations

- **Tier 1:** CBC (Low Hb 6–9 g/dL, High Retic count), Peripheral Smear (Sickle cells, target cells, Howell-Jolly bodies—signifying autosplenectomy). Sickling test with Sodium Metabisulfite.
- **Tier 2:** HPLC or Hb Electrophoresis.
- **Tier 3:** TCD (Transcranial Doppler) for stroke screening (ages 2–16 years), Chest X-ray (for ACS), Abdominal USG (for gallstones).

Management Outline

1. **Maintenance:**
    - **Folic Acid:** 1–5 mg/day.
    - **Penicillin V Prophylaxis:** Until at least age 5.
    - **Hydroxyurea:** [UPDATED] Now recommended for all children with SCA starting at 9 months of age, regardless of clinical severity, to increase HbF.
2. **Crisis Management:**
    - **VOC:** Hydration (1.5x maintenance), aggressive analgesia (Step ladder: NSAIDs → Opioids).
    - **ACS:** Oxygen, antibiotics (Macrolide + Cephalosporin), exchange transfusion.
3. **Cure:** Hematopoietic Stem Cell Transplant (HSCT) or Gene Therapy (recent FDA approvals).

EXAMINER'S VIVA

1. **Q: Why do SCD symptoms only start after 4–6 months of age?**
    - A: Because of the presence of high levels of HbF (fetal hemoglobin) which inhibits the polymerization of HbS. Symptoms appear as HbF levels decline.
2. **Q: What is the significance of Howell-Jolly bodies on a smear?**
    - A: They indicate functional asplenia. The spleen is no longer filtering nuclear remnants from RBCs.
3. **Q: How do you perform a Sickling Test?**
    - A: Mix a drop of blood with 2% sodium metabisulfite (a reducing agent) on a slide, seal it with paraffin, and look for sickling under a microscope after 1–2 hours.
4. **Q: What is the "Sequestration Crisis" and how do you identify it at the bedside?**
    - A: Sudden pooling of blood in the spleen. Bedside signs: Rapidly enlarging spleen, dropping Hb (shock), and high reticulocyte count.
5. **Q: Why is Hydroxyurea used in SCD?**
    - A: It increases HbF production, which prevents HbS polymerization, reduces VOCs, ACS, and the need for transfusions.

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting [Name], a [Age] year old male, a known case of Sickle Cell Anemia, currently presenting with a vaso-occlusive crisis, with a background of [Number] previous admissions and [Number] transfusions..."
- **Mistake:** Don't forget to check the **immunization status** for special vaccines. Examiners hate it when you miss the preventive aspect of SCD.
- **Observation:** The examiner will watch how you palpate the spleen. Ensure you start from the right iliac fossa because a sequestration crisis can lead to a massive spleen.

---

31. Aplastic anemia

Subject: Hematology

LONG CASE FRAMEWORK: APLASTIC ANEMIA

HISTORY

Chief Complaint

- **Anemia symptoms:** Progressive pallor, lethargy, easy fatigability (Duration: weeks to months).
- **Thrombocytopenia symptoms:** Spontaneous skin bleeds (petechiae, ecchymosis), mucosal bleeds (epistaxis, gum bleeds), or menorrhagia in adolescent girls.
- **Leukopenia symptoms:** Recurrent or persistent fevers, oral ulcers, or slow-healing infections.
- *Note:* In Aplastic Anemia (AA), the onset is usually insidious. If it's hyperacute, think of Acute Leukemia.

History of Present Illness

- **Characterizing the Cytopenias:**
    - "When did you first notice the child looking pale? Was it sudden or gradual?" (Gradual suggests AA; sudden suggests hemolysis).
    - "Are there any red spots on the skin? Do they disappear when you press them?" (Distinguishes petechiae from rashes).
    - "Has there been bleeding from the nose or gums? Any blood in the stools or dark-colored urine?"
    - "Has the child had a fever? If yes, was it associated with chills, rigors, or a sore throat?"
- **Excluding Differentials (The "Negative" History):**
    - **No Bone Pain:** "Does the child cry when you touch their legs or back? Does he refuse to walk?" (Bone pain/limp strongly suggests Leukemia or Neuroblastoma).

    - **No B-symptoms:** "Has there been significant weight loss or drenching night sweats?" (Suggests Lymphoma/Malignancy).
    - **No Lumps:** "Have you noticed any swelling in the neck, armpits, or groin?"
- **Etiological Clues:**
    - **Post-Hepatitis:** "Did the child have jaundice 2–3 months before the onset of these symptoms?" (Hepatitis-associated AA).
    - **Drugs/Toxins:** "Any history of taking chloramphenicol, sulfonamides, or anti-epileptics? Any exposure to pesticides or benzene?"
    - **Congenital Clues:** "Was the child born with any birth defects like a small thumb or a small head?" (Fanconi Anemia).

Relevant Background History

- **Past History:** Previous blood transfusions (calculate the frequency; frequent transfusions suggest a chronic process).
- **Birth History:** Low birth weight or small for gestational age (SGA) is common in Fanconi Anemia (FA).
- **Developmental History:** Developmental delay is often seen in FA.
- **Family History:** Consanguinity? Any sibling with similar "blood failure" or early deaths? (Suggests inherited bone marrow failure syndromes).

EXAMINATION

General Survey

- **The "Sick" Look:** The child usually looks pale but "otherwise well" unless they are currently febrile or severely anemic. Unlike Leukemia, they don't usually look "wasted" or cachectic early on.
- **Nutritional Status:** Assess for weight and height. [SEVERITY MARKER] Short stature is a hallmark of Fanconi Anemia.
- **Skin:** Look for "Café-au-lait" spots or areas of hypopigmentation/hyperpigmentation (slate-grey skin). Check for petechiae (pinpoint, non-blanchable) and ecchymoses over bony prominences.

Vital Signs and Anthropometry

- **Pulse:** Tachycardia and a "bounding" pulse (Hyperdynamic circulation due to severe anemia).
- **Temperature:** Document fever accurately. Any fever in a neutropenic child is a medical emergency.
- **Growth:** Plot Height/Weight on WHO/IAP charts. Microcephaly (small head circumference) is a key pointer to FA.

Peripheral Signs

- **Hands:** [EXAMINER FAVORITE] **The Thumb Examination.** Check for absent, hypoplastic, bifid, or triphalangeal thumbs. Check for a narrow first web space. This is the most common skeletal abnormality in FA.
- **Nails:** Look for dystrophic nails (Dyskeratosis Congenita).
- **Eyes:**
    - **Pallor:** Examine the palpebral conjunctiva. In AA, pallor is usually "paper-white."
    - **Icterus:** Usually *absent* in AA. If present, think of Hepatitis-associated AA or PNH.
    - **Retinal Hemorrhages:** Use an ophthalmoscope if the child is cooperative; indicates severe thrombocytopenia.
- **Lymph Nodes:** Systematically palpate cervical, axillary, and inguinal stations. **Crucial Finding:** Significant lymphadenopathy is *absent* in AA. Its presence points toward Leukemia or Lymphoma.

Systemic Examination — Primary System (Hematopoietic/Reticuloendothelial)

- **Inspection:** Look for any sternal deformity or bruising.
- **Palpation:**
    - **Liver and Spleen:** [EXAMINER FAVORITE] Use the "edge of hand" technique or "hooking" method.
    - **The Diagnostic Clincher:** In idiopathic Aplastic Anemia, there is **NO hepatosplenomegaly**.
    - *Clinical Significance:* If you feel a significant spleen, you must reconsider the diagnosis (think Leukemia, Hypersplenism, or Myelofibrosis). A palpable liver edge (1-2cm) in an infant may be normal, but significant enlargement is a "red flag" against AA.
- **Percussion:** Check for sternal tenderness. Apply firm pressure with your thumb over the manubrium and body of the sternum. [SEVERITY MARKER] Sternal tenderness is common in Leukemia but rare in AA.

Systemic Examination — Secondary Systems

- **Cardiovascular:** Auscultate for a "Hemic Murmur" (soft, systolic, ejection-type murmur at the base) due to anemia-induced turbulence.
- **Respiratory:** Check for basal crepitations (if in heart failure) or signs of pneumonia (opportunistic infection).
- **Oral Cavity:** Look for gingival hyperplasia (Leukemia), oral thrush, or necrotic ulcers (Neutropenia).

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

1. **Pancytopenia without Organomegaly:** The absence of a palpable spleen/lymph nodes in a pale child with petechiae is AA until proven otherwise.

2. **Thumb/Radial Anomalies:** Absent or hypoplastic thumbs point specifically to Fanconi Anemia.
3. **Café-au-lait spots:** Suggests FA.
4. **Mucosal Bleeding:** Indicates the severity of thrombocytopenia.

DIAGNOSIS

Diagnostic Criteria (Camitta Criteria)

Severe Aplastic Anemia (SAA) requires Bone Marrow cellularity <25% PLUS at least two of:

1. ANC (Absolute Neutrophil Count) < 500/mm^3
2. Platelet count < 20,000/mm^3
3. ARC (Absolute Reticulocyte Count) < 40,000/mm^3 (or <1% corrected) *Very Severe AA (VSAA):* Same as above but ANC < 200/mm^3.

Differentials

1. **Acute Leukemia (Aleukemic phase):** Distinguished by bone pain, lymphadenopathy, and hepatosplenomegaly.
2. **Hypocellular Myelodysplastic Syndrome (MDS):** Distinguished by dysplastic features on marrow aspirate and cytogenetics.
3. **Paroxysmal Nocturnal Hemoglobinuria (PNH):** Suggested by dark urine (hemoglobinuria) and positive flow cytometry (CD55/59 deficiency).

Investigations

- **Tier 1 (CBC & Peripheral Smear):** Pancytopenia with **normocytic normochromic** RBCs. Low reticulocyte count. No blasts.
- **Tier 2 (The Gold Standard):**
    - **Bone Marrow Aspirate (BMA):** "Dry tap" or "Empty particles."
    - **Bone Marrow Biopsy:** Shows replacement of hematopoietic tissue by fat cells.
- **Tier 3 (Etiology):**
    - **Chromosomal Breakage Analysis:** (Mitomycin C or Diepoxybutane test) - Positive in Fanconi Anemia.
    - **Liver Function Tests:** To check for post-hepatitis AA.

Management Outline

1. **Stabilization:** Leucocyte-depleted, irradiated PRBC and Platelet transfusions (keep Hb >7, Platelets >10k).
2. **Definitive Rx:**

- **Matched Sibling Donor (MSD) HSCT:** Treatment of choice for SAA.
- **Immunosuppressive Therapy (IST):** If no donor. Triple therapy: Antithymocyte Globulin (ATG) + Cyclosporine + Eltrombopag [UPDATED].

3. **Supportive:** Febrile neutropenia protocol (broad-spectrum antibiotics).

EXAMINER'S VIVA

1. **Q: Why do we use irradiated blood products in AA?** *A: To prevent Transfusion-Associated Graft-Versus-Host Disease (TA-GVHD), as these children are severely immunosuppressed and may undergo future transplant.*
2. **Q: How do you calculate the Absolute Neutrophil Count (ANC)?** *A: ANC = Total WBC count × (% Neutrophils + % Bands) / 100.*
3. **Q: What is the significance of a "Dry Tap" on BMA?** *A: It means no marrow could be aspirated. Common in AA (empty marrow), Leukemia (packed marrow), or Myelofibrosis.*
4. **Q: Why is Eltrombopag now used in AA?** *A: It is a TPO-receptor agonist that stimulates stem cells and has shown to improve response rates when added to ATG/Cyclosporine.*
5. **Q: Technique: How do you differentiate a petechiae from a hemangioma at the bedside?** *A: Diascopy (The Glass Slide Test). Press a transparent slide against the lesion. Petechiae do not blanch because the blood is extravasated; vascular lesions like hemangiomas will blanch.*

LONG CASE PRESENTATION TIPS

- **The Opening:** "I am presenting [Name], a [Age] year old male, who presented with a triad of progressive pallor, spontaneous skin bleeds, and recurrent febrile episodes, in the absence of bone pain or significant organomegaly."
- **Common Mistake:** Forgetting to mention the "Negative" findings. In AA, the *absence* of splenomegaly and lymphadenopathy is just as important as the *presence* of pallor.
- **What Examiners Watch For:** They will watch your hand placement during abdominal palpation. If you "dig" for a spleen and miss it, or if you fail to check the thumbs in a suspected AA case, it shows a lack of clinical maturity.

---

32. Leukemia (ALL)

Subject: Hematology

This is a high-stakes long case. In a Leukemia case, the examiner isn't just looking for the diagnosis—they are looking for your ability to assess **bone marrow failure**, **organ infiltration**, and **oncologic emergencies**.

HISTORY

Chief Complaint

- **Fever:** Duration and pattern (usually low-grade but persistent, or high-grade if secondary infection).
- **Pallor/Fatigue:** "When did you notice he stopped playing or started wanting to be carried more?"
- **Bleeding:** Epistaxis, gum bleeds, or "unexplained bruises" (purpura/petechiae).
- **Bone Pain:** "Does he wake up at night crying with leg pain?" or "Has he stopped walking entirely?" (Refusal to bear weight is a classic ALL presentation).
- **Lumps/Swellings:** Neck, axilla, or groin swellings; abdominal distension.

History of Present Illness

- **Bone Marrow Failure (The Triad):**
    - *Anemia:* "Is he breathless while eating or crying?" "Has he become pale suddenly or gradually?"
    - *Neutropenia:* "Any recurrent mouth ulcers, sore throat, or skin boils?" "How long did the last fever last?"
    - *Thrombocytopenia:* "Do you see tiny red spots on the skin?" "Does he bleed for a long time after a small scratch?"
- **Extramedullary Infiltration:**
    - *Reticuloendothelial:* "Have you noticed any fullness in the belly?" (Hepatosplenomegaly).
    - *CNS:* "Any morning headaches, vomiting, or squint?" (Suggests CNS leukemia/increased ICP).
    - *Mediastinal (T-cell ALL):* "Does he have a dry cough or wheeze when lying flat?" "Is there any facial puffiness?" (Superior Vena Cava Syndrome).
    - *Testicular:* "Any painless swelling or heaviness in the private parts?"
- **Constitutional:** "Has there been significant weight loss?" (Quantify if possible).

Relevant Background History

- **Past History:** Previous radiation exposure, prior chemotherapy, or history of Down Syndrome/Bloom Syndrome/Fanconi Anemia.
- **Family History:** History of early childhood cancers (Li-Fraumeni syndrome).
- **Socioeconomic:** Essential for ALL. "Can the family afford a 2.5-year treatment protocol?" "How far do they live from a tertiary center?" (Treatment abandonment is the leading cause of failure in developing settings).

EXAMINATION

General Survey

- **Initial Impression:** Does the child look "waxy pale" (Anemia) or "toxic" (Sepsis)? Note the activity level—is the child irritable or lethargic?
- **Nutritional Status:** Check for temporal wasting and loss of gluteal fat. Malnutrition at diagnosis is a poor prognostic marker.
- **Facies:** Look for Down syndrome features (20x increased risk of leukemia). Look for "chipmunk facies" (though more common in Thalassemia, extramedullary hematopoiesis can occur in chronic leukemias).

Vital Signs and Anthropometry

- **Temperature:** Document fever.
- **Heart Rate:** Tachycardia out of proportion to fever suggests severe anemia.
- **Respiratory Rate:** Tachypnea might indicate severe anemia, pneumonia, or a mediastinal mass.
- **Blood Pressure:** Check for hypertension (can occur with hyperuricemia/renal involvement or steroid therapy).

Peripheral Signs

- **Pallor:** [TECHNIQUE] Check palmar creases, palpebral conjunctiva, and buccal mucosa. In ALL, pallor is often "dead white."
- **Skin Markers:**
    - *Petechiae/Ecchymosis:* Look at pressure points and shins.
    - *Leukemia Cutis:* Multiple firm, non-tender, blue-gray/violaceous nodules (more common in AML/Neonatal leukemia).
- **Lymph Nodes:** [TECHNIQUE] Systematically palpate cervical, axillary, and inguinal chains.
    - *Note:* Size, consistency (leukemic nodes are usually "rubbery," non-tender, and discrete), and fixity.
    - *Specific:* Look for **Epitrochlear** and **Supraclavicular** nodes—these are almost always pathological.
- **Sternum:** [EXAMINER FAVORITE] **Sternal Tenderness.** Use your thumb to apply firm pressure over the mid-sternum. A wince or withdrawal indicates marrow expansion/infiltration.

Systemic Examination — Primary System (Abdomen)

- **Inspection:** Look for fullness in the flanks or epigastrium. Note any dilated veins (SVC or IVC obstruction).
- **Palpation:**
    - *Liver:* Measure liver span. Note the edge (usually firm, smooth, non-tender in ALL).

    - *Spleen:* [TECHNIQUE] Start in the right iliac fossa. Use the "hooking method" for massive spleens. Note the notch. Splenomegaly in ALL is due to infiltration.
- **Percussion:** Confirm organomegaly; check for shifting dullness (ascites is rare but can occur in advanced disease).

Systemic Examination — Secondary Systems

- **Respiratory:** Auscultate for decreased breath sounds or stony dullness (pleural effusion associated with mediastinal mass).
- **Cardiovascular:** Listen for a hemic murmur (systolic, flow murmur) at the apex/base due to anemia.
- **CNS:** Check for cranial nerve palsies (III, IV, VI, VII) and signs of meningismus.
- **Genitalia:** [MANDATORY] Palpate both testes. Leukemic infiltration causes **painless, stony-hard enlargement**, usually bilateral but can be unilateral.

Signs That Clinch the Diagnosis [EXAMINER FAVORITE]

- **The "Rubbery" Lymphadenopathy:** Generalized, non-matted, non-tender nodes.
- **Sternal Tenderness:** Highly suggestive of marrow infiltrative process.
- **Painless Testicular Enlargement:** In a pale child, this is leukemia until proven otherwise.
- **Petechiae + Pallor + Hepatosplenomegaly:** The classic "Leukemic Triad."

Severity/Emergency Markers [SEVERITY MARKER]

- **Superior Vena Cava (SVC) Syndrome:** Facial puffiness, orthopnea, distended neck veins.
- **Hyperleukocytosis:** Confusion, priapism, or respiratory distress (Leukostasis).
- **Tumor Lysis Syndrome (TLS):** Oliguria, tetany (hypocalcemia), or arrhythmias.

DIAGNOSIS

Diagnostic Criteria

- **Gold Standard:** Bone marrow aspiration/biopsy showing **>20% blasts** (WHO criteria).
- **Morphology:** FAB Classification (L1, L2, L3).
- **Immunophenotyping:** Flow cytometry to distinguish B-ALL (CD19, CD20, CD22) from T-ALL (CD3, CD5, CD7).

Differentials

1. **Aplastic Anemia:** Will have pancytopenia but **no** hepatosplenomegaly and **no** lymphadenopathy.

2. **Infectious Mononucleosis:** Fever, sore throat, lymphadenopathy, and splenomegaly. Distinguished by atypical lymphocytes (not blasts) and positive Monospot/EBV titers.
3. **Juvenile Idiopathic Arthritis (Systemic JIA):** Fever, joint pain, and rash. However, bone pain in leukemia is usually metaphyseal, not just in the joints.
4. **Neuroblastoma (Stage IV):** Can present with marrow infiltration and bone pain. Look for a fixed, irregular abdominal mass that crosses the midline.

Investigations

- **Tier 1:** CBC (WBC may be low, normal, or high; anemia and thrombocytopenia usually present), Peripheral Smear (look for blasts), Uric acid, LDH (marker of cell turnover).
- **Tier 2:** Bone Marrow Aspirate & Biopsy (Morphology, Cytochemistry like MPO negative in ALL), Flow Cytometry.
- **Tier 3:** Cytogenetics (t(12;21) is good prognosis; t(9;22) Philadelphia+ is poor), CSF Analysis (to rule out CNS involvement), Chest X-ray (look for mediastinal mass).

Management Outline

- **Stabilization:** Hydration (2x maintenance), Allopurinol (for TLS), transfusion of PRBC/Platelets if symptomatic.
- **Chemotherapy Phases:**
    1. Induction (4 weeks): Aim for complete remission (<5% blasts).
    2. Consolidation/Intensification.
    3. Maintenance (2-3 years).
- **CNS Prophylaxis:** Intrathecal Methotrexate (essential for all patients).

EXAMINER'S VIVA

1. **Q: Why is it important to check the testes in a child with ALL?**
    - *A:* The blood-testis barrier makes it a "sanctuary site" where systemic chemo doesn't reach well. It’s a common site for relapse.
2. **Q: How do you differentiate a blast from a mature lymphocyte on a smear?**
    - *A:* Blasts are larger, have a high N:C ratio, fine "open" chromatin, and visible nucleoli.
3. **Q: What is the "Great Imitator" in pediatric oncology?**
    - *A:* ALL, because it can mimic JIA, ITP, or even osteomyelitis.
4. **Q: A child with ALL develops a high fever during chemo. What is your first step?**
    - *A:* Assume **Febrile Neutropenia**. Start IV broad-spectrum antibiotics (e.g., Piperacillin-Tazobactam) immediately after taking cultures, without waiting for results.
5. **Q: [Technique] Show me how you palpate for axillary lymph nodes.**

- *A:* (Demonstrate: Support the child's arm to relax the pectoral muscles, use the pads of your fingers to palpate against the chest wall in all four quadrants—anterior, posterior, medial, lateral, and apex).

LONG CASE PRESENTATION TIPS

- **The Opening:** "I am presenting the case of a [Age] year old male, who presented with a short history of 3 weeks characterized by the triad of bone marrow failure—fever, progressive pallor, and skin bleeds—associated with significant bone pain and abdominal distension."
- **Mistake to Avoid:** Never forget to mention the **fundus examination** (for leukemic infiltrates or hemorrhages) and **testicular exam**.
- **The "Why":** If you find splenomegaly, always state if it is "massive" or "moderate" as this helps in risk stratification (Standard vs. High Risk).

---

33. Hemophilia

Subject: Hematology

LONG CASE FRAMEWORK: HEMOPHILIA

HISTORY

Chief Complaint

- **Joint swelling and pain:** (Hemarthrosis) Most common in toddlers and older children.
- **Prolonged bleeding:** Following trauma, tooth extraction, or circumcision.
- **Easy bruising:** Large, palpable ecchymoses (not petechiae).
- **Muscle pain/swelling:** (Muscle hematoma) Leading to limb weakness or paresthesia.

History of Present Illness

Ask these questions naturally to build the "bleeding phenotype":

- **"When did the swelling start, and was there a specific injury?"** (Spontaneous vs. traumatic; spontaneous bleeds suggest severe deficiency <1%).
- **"How long did it take for the swelling to reach its peak?"** (Hemophilic bleeds are usually delayed by a few hours, unlike immediate traumatic effusions).
- **"Is this the first time, or has this happened in other joints?"** (Identify "target joints" – 3 or more bleeds in a single joint in 6 months).
- **"Did the bleeding stop and then start again a few hours later?"** (Classic for coagulation defects; primary hemostasis/platelet plug is fine, but secondary hemostasis/fibrin mesh is weak).
- **"Does he have any difficulty moving the limb or numbness in the fingers/toes?"** (Checking for compartment syndrome or nerve compression by a hematoma).

- **"Have you noticed any dark urine or black stools?"** (Internal mucosal bleeds).
- **"Has he ever had a headache, vomiting, or altered sensorium after a minor fall?"** (Crucial: rule out Intracranial Hemorrhage).

Relevant Background History

- **Birth History:** "Was there a scalp swelling (cephalohematoma) after birth?" "Did he bleed excessively after the umbilical cord fell off or after Vitamin K injection?"
- **Developmental:** "When did he start crawling or walking?" (Bleeding often starts with increased mobility).
- **Family History:** [CRITICAL] Draw a 3-generation pedigree. "Are any maternal uncles, brothers, or the maternal grandfather affected?" (X-linked recessive pattern). Note: 30% are *de novo* mutations with no family history.
- **Treatment History:** "What products has he received? (FFP, Cryoprecipitate, or Factor concentrate?)" "How many times a year?" "Has the usual dose stopped working lately?" (Suggests **Inhibitor** formation).
- **Socioeconomic:** Cost of factor is prohibitive; assess if they are on a government-sponsored program (e.g., NHM in India).

EXAMINATION

General Survey

- **State:** Is the child in pain? (Acute hemarthrosis is excruciating).
- **Posture:** Look for "pseudoflexion" deformity of the knee or elbow. The child holds the joint in a position of maximum comfort (usually semi-flexed).
- **Gait:** Observe for limping or "antalgic gait."
- **Pallor:** Check for chronic anemia (from multiple bleeds) or acute anemia (large psoas/muscle bleed).

Vital Signs and Anthropometry

- **Pulse:** Tachycardia may indicate acute pain or significant internal blood loss.
- **BP:** Check for hypotension in large muscle bleeds (Psoas).
- **Weight/Height:** Chronic disease and restricted mobility can lead to growth failure or, conversely, obesity due to inactivity (which worsens joint stress).

Peripheral Signs

- **Skin:** Look for **Ecchymoses** (large, blue-purple bruises). [EXAMINER FAVORITE]: Note the *absence* of petechiae (petechiae suggest platelet disorders, not factor deficiency).
- **Joints (General):** Look for muscle wasting (Quadriceps wasting is prominent in chronic knee hemarthrosis).

Systemic Examination — Musculoskeletal (The Primary System)

Examine the affected joint and compare it with the contralateral normal joint.

1. Inspection:

- **Swelling:** Note if it is localized to the joint capsule.
- **Skin over joint:** Is it red or stretched?
- **Muscle wasting:** Measure mid-thigh circumference (for knee) or mid-arm (for elbow). Chronic bleeds lead to disuse atrophy.
- **Deformity:** Look for fixed flexion deformity (FFD) or valgus/varus angulations.

2. Palpation:

- **Temperature:** Use the back of your hand. Acute hemarthrosis is **warm**.
- **Tenderness:** Diffuse tenderness over the synovium.
- **Synovial thickening:** In chronic cases, the synovium feels "boggy" or "doughy" (Hemophilic Synovitis).
- **Bony expansion:** Chronic bleeds cause epiphyseal overgrowth (the joint looks disproportionately large).

3. Range of Motion (ROM):

- **Active vs. Passive:** Always check active first. [SEVERITY MARKER]: Severe restriction in all planes suggests acute bleed or end-stage arthropathy.
- **Patellar Tap:** If the knee is swollen, perform the tap to confirm intra-articular fluid.

4. Neurological (Secondary):

- Check for **Nerve Palsies**: Specifically the Femoral nerve (psoas bleed) or Median/Ulnar nerve (forearm hematoma).
- Check distal pulses to rule out **Compartment Syndrome**.

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **The "Target Joint":** A joint with repetitive bleeds, characterized by boggy synovial thickening and restricted ROM.
- **Quadriceps Wasting:** A sensitive sign of chronic knee involvement.
- **Pseudotumor:** A rare but pathognomonic finding; a firm, enlarging, non-tender mass (usually on the pelvis or long bones) caused by a subperiosteal hemorrhage.
- **Psoas Sign:** Child keeps the hip flexed and experiences pain on passive extension (indicates Iliopsoas bleed).

Severity Assessment [SEVERITY MARKER]

- **Mild:** Factor level 5–40%. Bleeding only after major trauma/surgery.
- **Moderate:** Factor level 1–5%. Bleeding after minor trauma.
- **Severe:** Factor level <1%. Frequent spontaneous bleeds (joints/muscles).

DIAGNOSIS

Diagnostic Criteria

- **Clinical:** Male child, X-linked family history, musculoskeletal bleeds.
- **Laboratory:** Prolonged aPTT, normal PT, normal Bleeding Time/Platelet count, and **low Factor VIII (Hemophilia A) or Factor IX (Hemophilia B) activity levels.**

Differentials

1. **von Willebrand Disease (vWD):** Bleeding is usually mucosal (epistaxis, gum bleeds). aPTT may be prolonged, but BT is also prolonged.
2. **Vitamin K Deficiency:** Usually in neonates or those with malabsorption. Both PT and aPTT are prolonged.
3. **Immune Thrombocytopenic Purpura (ITP):** Characterized by petechiae and low platelet count; joints are never involved.

Investigations

- **Tier 1 (Screening):** CBC (normal platelets), PT (normal), aPTT (prolonged). [EXAMINER FAVORITE]: **Mixing Study.** Mix patient plasma 1:1 with normal plasma. If aPTT corrects, it's a deficiency. If it doesn't, an **Inhibitor** is present.
- **Tier 2 (Confirmatory):** Factor VIII and IX assays.
- **Tier 3 (Complications):**
    - **Bethesda Assay:** To quantify inhibitors (if aPTT doesn't correct).
    - **X-ray/MRI of joints:** Arnold-Hilgartner staging for arthropathy.
    - **Screening:** HIV, Hep B, Hep C (for those who received older blood products).

Management Outline

1. **Acute Bleed: RICE** (Rest, Ice, Compression, Elevation) + Immediate Factor Replacement.
2. **Factor Replacement:**
    - Hemophilia A: 1 unit/kg raises FVIII by 2%. Target 40-50% for joint bleeds, 100% for life-threatening bleeds.
    - Hemophilia B: 1 unit/kg raises FIX by 1%.
3. **Prophylaxis [UPDATED]:** Primary prophylaxis (starting before the 2nd joint bleed) is now the gold standard to prevent arthropathy.

4. **Novel Therapies: Emicizumab** (bispecific antibody for Hemophilia A) – can be used even in patients with inhibitors.
5. **Adjuncts:** Tranexamic acid (for dental/mucosal bleeds); Desmopressin (for mild Hemophilia A).

EXAMINER'S VIVA

Q1: Why do we avoid intramuscular (IM) injections in these children? *A: It causes massive muscle hematomas. All vaccines should be given subcutaneously, and firm pressure applied for 5 minutes.*

Q2: How do you differentiate a Psoas bleed from Acute Appendicitis? *A: In a Psoas bleed, the hip is held in flexion, and pain occurs on **extension**. In appendicitis, pain is usually localized to McBurney's point and associated with fever and rebound tenderness. A Psoas bleed may also show a drop in hemoglobin.*

Q3: What is the "Mixing Study" and why is it important? *A: We mix the patient's plasma with normal pool plasma. If the prolonged aPTT corrects, the patient has a factor deficiency. If it fails to correct, it means the patient has developed **Inhibitors** (antibodies) against the factor.*

Q4: Show me how you measure for a fixed flexion deformity of the knee. *(Technique: Perform Thomas Test or simply attempt to flatten the popliteal space against the bed. Measure the angle of the lower leg relative to the bed using a goniometer.)*

Q5: What is the most common cause of death in Hemophilia today? *A: In developed areas, it is Intracranial Hemorrhage. In areas with poor screening in the past, complications of transfusion-transmitted infections (HIV/HCV) were common.*

LONG CASE PRESENTATION TIPS

- **Opening:** "Master X, a [Age]-year-old male, born of a non-consanguineous marriage, presenting with recurrent spontaneous painful joint swellings since he started walking, with a significant maternal family history, currently presenting with an acute right knee hemarthrosis..."
- **Mistake to avoid:** Never say "The child has Hemophilia" in the HPI. Say "The child has a life-long bleeding diathesis suggestive of a coagulation disorder."
- **Examiner Watch-point:** They will watch how you handle the joint. Be extremely gentle. If the child is in acute pain, do not force a full Range of Motion. Mention this to the examiner: "I am not performing full passive ROM to avoid causing further pain and bleeding."

Endocrine

34. Type 1 diabetes mellitus

Subject: Endocrine

HISTORY: Type 1 Diabetes Mellitus (T1DM)

Chief Complaint

- **Polyuria:** "Increased frequency and volume of urine" (Ask if the child is waking up at night or has started wetting the bed again—secondary enuresis).
- **Polydipsia:** "Excessive thirst" (Ask if they are drinking water even at night).
- **Polyphagia:** "Increased appetite" despite weight loss.
- **Weight loss:** Rapid, despite good intake.
- **Altered sensorium/Vomiting/Abdominal pain:** Suggests Diabetic Ketoacidosis (DKA).

History of Present Illness

- **The "Osmotic" Symptoms:** "When did you first notice he was going to the toilet more often? Is he waking up 3-4 times a night? Is he drinking more than 2-3 liters a day?"
- **Weight Loss:** "How much weight has he lost in the last month? Are his clothes fitting looser?" (Quantify the loss).
- **DKA Screen:** "Has there been any vomiting or abdominal pain? Is his breathing heavy or fast? Is he unusually drowsy or irritable?"
- **Infection Triggers:** "Did he have a fever, cough, or urinary burning just before these symptoms started?" (Infections often precipitate DKA).
- **Negative Screen for Type 2/Secondary DM:**
    - "Any darkening of the skin around the neck (Acanthosis)?"
    - "Any long-term steroid use for asthma or nephrotic syndrome?"
    - "Any history of recurrent pancreatitis or oily stools (Cystic Fibrosis related)?"

Relevant Background History

- **Past History:** Previous episodes of DKA or "heavy breathing" treated with IV fluids.
- **Developmental:** Usually normal, but chronic poor control can lead to growth failure and delayed puberty.
- **Immunization:** Check for flu and pneumococcal vaccines (recommended for T1DM).
- **Family History:** History of T1DM (only 5-10% have it) or other autoimmune diseases (Hypothyroidism, Celiac disease, Vitiligo).
- **Socioeconomic: Crucial.** "Who gives the injections? Can the family afford insulin and strips? Is there a refrigerator at home?" (This dictates your management plan).

EXAMINATION

General Survey

- **The "Doorway" Assessment:** Does the child look dehydrated? Is there a fruity odor (acetone) to the breath? Is the child tachypneic (Kussmaul breathing)?
- **Activity:** Is the child lethargic or playing?
- **Nutritional Status:** Look for visible wasting of the gluteal muscles and loss of buccal fat pads.
- **Skin:** Look for fungal infections (Candidal intertrigo, paronychia), necrobiosis lipoidica (rare, pretibial), or vitiligo (autoimmune association).

Vital Signs and Anthropometry

- **Respiration:** Count for a full minute. Look for **Kussmaul breathing** (deep, sighing, rapid respirations). This is a compensatory mechanism for metabolic acidosis.
- **Hydration:** Check capillary refill time (CRT), skin turgor (on the abdomen), and dryness of tongue.
- **Blood Pressure:** Use the correct cuff size (covering 2/3rd of the upper arm). Check for orthostatic hypotension if the child is old enough to stand.
- **Growth:** Plot Height, Weight, and BMI on WHO/IAP charts. **[SEVERITY MARKER]** Poor height velocity suggests long-standing poor glycemic control (Mauriac Syndrome).

Peripheral Signs

- **Hands:**
    - **Limited Joint Mobility (Cheiroarthropathy):** Ask the child to join palms in a "Prayer Sign." If they cannot appose the PIP and MCP joints, it suggests chronic hyperglycemia and glycosylation of collagen.
    - **Insulin Injection Sites:** Inspect the thighs, abdomen, and deltoids. Look for **Lipohypertrophy** (rubbery, fatty lumps). *Technique:* You must palpate, not just look.
- **Eyes:** Fundoscopy (if age permits) to look for background retinopathy (rare at diagnosis, but common in long-standing cases). Look for cataracts (snowflake cataracts).
- **Thyroid:** Palpate for goiter (Hashimoto’s thyroiditis is a common co-morbidity).

Systemic Examination — Primary System (Abdomen & Metabolic)

- **Inspection:** Look for abdominal distension (DKA can cause paralytic ileus).
- **Palpation:**
    - **Liver:** Check for hepatomegaly. In poorly controlled T1DM, glycogen deposition can cause a firm, large liver (Mauriac Syndrome).
    - **Tenderness:** DKA often presents with "surgical abdomen" (generalized tenderness/guarding).
- **Auscultation:** Bowel sounds may be sluggish in DKA.

Systemic Examination — Secondary Systems

- **Neurological:** Check sensorium (Glasgow Coma Scale). Look for signs of cerebral edema (bradycardia, hypertension, altered breathing) if the child is in DKA.
- **Pubertal Staging:** Use Tanner staging. T1DM can cause delayed puberty.

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **Kussmaul Respiration:** Deep, labored breathing without chest indrawing (unlike pneumonia). Indicates pH < 7.2.
- **Acetone Breath:** Sweet, fruity smell (like rotting apples or nail polish remover).
- **Lipohypertrophy:** Palpable lumps at injection sites. Indicates poor rotation of sites and leads to erratic insulin absorption.
- **Prayer Sign:** Inability to flatten palms against each other. A marker of long-term microvascular risk.

Severity Assessment

- **Mild/Moderate:** Polyuria, polydipsia, weight loss, but hemodynamically stable and conscious.
- **Severe (DKA):** Vomiting, abdominal pain, Kussmaul breathing, dehydration >5%, altered sensorium (GCS <15).

DIAGNOSIS

Diagnostic Criteria (ISPAD/ADA)

1. **Symptoms of DM** (Polyuria, polydipsia, weight loss) **PLUS** Random Plasma Glucose mg/dL.
2. **Fasting Plasma Glucose** mg/dL (Fasting = no caloric intake for 8 hrs).
3. **2-hour Post-load Glucose** mg/dL during OGTT.
4. **HbA1c** (Must be performed in a lab using NGSP certified method).

Differentials

- **Type 2 DM:** Usually adolescent, obese, Acanthosis nigricans, strong family history of T2DM.
- **Diabetes Insipidus:** Polyuria/polydipsia present, but urine is dilute (low specific gravity) and blood sugar is normal.
- **Renal Glycosuria:** Glucose in urine but normal blood glucose.
- **MODY (Maturity Onset Diabetes of the Young):** Autosomal dominant, usually presents <25 years, non-ketotic, often misdiagnosed as T1DM.

Investigations

- **Tier 1 (Bedside):** Urine dipstick (Glucose +++, Ketones positive), Fingerstick blood glucose.
- **Tier 2 (Confirmatory):** Venous blood glucose, HbA1c, C-peptide (low in T1DM), Beta-cell antibodies (GAD-65, IA-2, Zinc Transporter 8) to confirm autoimmune etiology.

- **Tier 3 (DKA/Complications):** Arterial/Venous Blood Gas (pH, HCO3), Serum Electrolytes (check for "pseudo-hyponatremia" and potassium shift), Renal function, TSH and Anti-TTG (screen for associated autoimmune diseases).

Management Outline

1. **DKA Stabilization:** IV fluids (0.9% NS), Potassium replacement (even if serum K is normal), and IV Insulin infusion (0.05–0.1 U/kg/hr) after 1 hour of fluids.
2. **Maintenance:** Subcutaneous Insulin (Basal-Bolus regimen is gold standard).
    - **Basal:** Glargine or Degludec (once daily).
    - **Bolus:** Aspart, Lispro, or Regular insulin before meals.
3. **Monitoring:** SMBG (Self-monitoring of blood glucose) at least 4 times/day; HbA1c every 3 months (Target < 7.0% per ISPAD).
4. **Education:** Injection technique, site rotation, hypoglycemia recognition/management (Rule of 15), and "Sick Day Rules."

EXAMINER'S VIVA

1. **Q: Why do we wait 1 hour before starting insulin in DKA?**
    - A: To allow for initial volume expansion and reduce the risk of a sudden drop in osmolality, which can precipitate cerebral edema.
2. **Q: How do you differentiate T1DM from T2DM clinically?**
    - A: T1DM: Lean, younger, ketosis-prone, no acanthosis. T2DM: Obese, pubertal, acanthosis present, strong family history.
3. **Q: What is the "Honeymoon Phase"?**
    - A: A period shortly after starting insulin where the remaining beta cells recover temporarily, leading to very low insulin requirements (<0.5 U/kg/day) and stable sugars.
4. **Q: Explain the technique for checking Lipohypertrophy.**
    - A: Use a "sweeping" motion with fingertips and then a "pinch" technique over injection sites. It feels like a firm, rubbery nodule under the skin.
5. **Q: What are "Sick Day Rules"?**
    - A: Never stop insulin during illness (even if not eating); check blood glucose and ketones more frequently (every 2-4 hours); maintain hydration; and give extra rapid-acting insulin if sugars are high.

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting [Name], a [Age] year old male, who presented with a 2-week history of osmotic symptoms and significant weight loss, currently admitted with/without features of metabolic acidosis."

- **Common Mistake:** Forgetting to check the injection sites or the thyroid. Examiners look for this.
- **Common Mistake:** Not quantifying the weight loss or the amount of fluid intake.
- **Examiner Focus:** They will watch how you plot the growth chart. A child "falling off the curve" is a major indicator of chronic poor control.

---

35. Hypothyroidism

Subject: Endocrine

HISTORY

Chief Complaint

- **Infants:** Prolonged jaundice, poor feeding, constipation, "quiet baby" (excessive sleepiness), or failure to thrive.
- **Older Children:** Short stature (most common), weight gain despite poor appetite, declining school performance, or delayed puberty.
- **Duration:** Crucial to establish when the growth velocity slowed down or when developmental milestones lagged.

History of Present Illness

- **Growth:** "When did you notice he stopped outgrowing his clothes or shoes?" (Differentiates congenital from acquired). "Is he the shortest in his class?"
- **Activity/Neurodevelopment:** "Is she as active as her peers, or does she prefer sitting in one place?" "Has there been a recent change in her grades or teacher's feedback?" (Hypothyroidism causes "pseudo-dementia" or sluggishness, not true intellectual disability if acquired late).
- **Gastrointestinal:** "How many times does he pass stool per week?" "Is the stool hard or like pellets?" (Constipation is a hallmark).
- **Energy/Sleep:** "Does she sleep more than usual?" "Is it difficult to wake her up in the morning?"
- **Skin/Hair:** "Have you noticed her skin becoming dry or rough like sandpaper?" "Is there increased hair fall or thinning of the eyebrows?"
- **Cold Intolerance:** "Does he demand a sweater when others are comfortable?" "Does he avoid fans or AC?"
- **Puberty:** "Has there been any breast development or menses?" (Look for **Van Wyk-Grumbach Syndrome**: precocious puberty with hypothyroidism).

Relevant Background History

- **Antenatal/Birth:** Ask about maternal thyroid status, intake of antithyroid drugs, or goitrogens. Ask about birth weight (often high/macrosomic in congenital hypothyroidism) and prolonged neonatal jaundice.
- **Newborn Screening:** "Was a heel-prick test done after birth?" (Crucial for congenital cases).
- **Developmental:** Detailed milestones. In congenital cases, delay is global; in acquired cases, motor and social skills are preserved but "slowed down."
- **Nutritional:** Distinguish thyroid-related weight gain (myxedema/water retention) from caloric obesity. Thyroid patients have poor appetite but still gain weight.
- **Family History:** Autoimmune thyroiditis in the mother or siblings; history of consanguinity (points to dyshormonogenesis).

EXAMINATION

General Survey

- **Initial Impression:** Look for a "placid" or "dull" expression. Is the child interactive or disinterested?
- **Facies:** Look for periorbital puffiness, a broad flat nasal bridge, and a protruding tongue (macroglossia).
- **Skin:** [EXAMINER FAVORITE] Feel the skin with the back of your hand. It should be cold, dry, and coarse (xeroderma). Look for a yellowish tinge (carotenemia—check palms/soles, but sclera will be white).
- **Hair:** Brittle, thinning of the lateral 1/3rd of eyebrows (Hertoghe's sign).

Vital Signs and Anthropometry

- **Heart Rate:** Measure for a full minute. **Bradycardia** is a key finding.
- **Blood Pressure:** Check for narrow pulse pressure.
- **Height/Length:** [CRITICAL] Must use a stadiometer for older children or an infantometer for <2 years. Plot on WHO/IAP charts.
- **Upper Segment:Lower Segment (US:LS) Ratio:** [EXAMINER FAVORITE]
    - *Technique:* Measure total height and lower segment (symphysis pubis to floor). US = Total height - LS.
    - *Significance:* Hypothyroidism causes **disproportionate short stature** (infantile proportions preserved). The US:LS ratio will be high for the age.
- **Weight:** Often increased (BMI high), but height is more severely affected.

Peripheral Signs

- **Hands:** Feel for coldness. Check for "puffy" dorsum of hands.
- **Pulse:** Low volume, slow rate.

- **Eyes:** Periorbital edema (non-pitting).
- **Neck/Thyroid:** [TECHNIQUE]
    - Stand behind the child. Use both hands. Ask the child to swallow (give a sip of water).
    - Note size, consistency (rubbery in Hashimoto's), nodules, and presence of a bruit.
    - If no thyroid is felt, look for an ectopic thyroid at the base of the tongue (lingual thyroid).

Systemic Examination — Primary System (Neuromuscular & Endocrine)

- **Deep Tendon Reflexes:** [EXAMINER FAVORITE] Focus on the **Ankle Jerk**.
    - *Technique:* Ensure the patient is relaxed.
    - *Finding:* **Delayed relaxation phase** (Woltman sign). This is due to slowed calcium reuptake by the sarcoplasmic reticulum.
- **Muscle Tone:** Often hypotonic in infants ("floppy baby"). In older children, look for "pseudohypertrophy" of muscles (Kocher-Debre-Semelaigne syndrome).
- **Abdomen:**
    - *Inspection:* Look for a protuberant abdomen and **umbilical hernia** (common in congenital cases).
    - *Palpation:* Check for fecal masses (constipation).

Systemic Examination — Secondary Systems

- **CVS:** Listen for muffled heart sounds (pericardial effusion).
- **Respiratory:** Noisy breathing or snoring (macroglossia).

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

1. **Disproportionate Short Stature:** High US:LS ratio for age indicating skeletal immaturity.
2. **Delayed Relaxation of Reflexes:** Pathognomonic for the metabolic state of hypothyroidism.
3. **Xeroderma & Carotenemia:** Cold, dry, yellowish skin with white sclera.
4. **Macroglossia & Umbilical Hernia:** In an infant, these two together strongly suggest congenital hypothyroidism.
5. **Goiter with Hypothyroid Features:** Points specifically to Hashimoto's or dyshormonogenesis.

Severity Assessment

- **Severe [SEVERITY MARKER]:** Myxedema coma (rare in kids), pericardial effusion (muffled sounds), or profound developmental delay in an infant.
- **Moderate:** Significant growth failure (Height < -3 SD), delayed bone age.

DIAGNOSIS

Diagnostic Criteria

- **Biochemical:** Elevated TSH with low Free T4.
- **Subclinical:** Elevated TSH with normal Free T4.

Differentials

1. **Down Syndrome:** Also has macroglossia and hypotonia, but has distinct epicanthal folds and Simian crease. (Note: Down syndrome often co-exists with hypothyroidism).
2. **Growth Hormone Deficiency:** Proportionate short stature, "doll-like" facies, normal skin texture, and normal reflexes.
3. **Mucopolysaccharidosis (MPS):** Coarse facies and organomegaly, but usually has corneal clouding and skeletal deformities (dysostosis multiplex).

Investigations

- **Tier 1:** Serum TSH (Screening) and Free T4 (Confirmatory).
- **Tier 2:**
    - **Bone Age:** X-ray of left hand/wrist (or knee/foot in neonates). Look for **epiphyseal dysgenesis** (stippled epiphysis).
    - **Thyroid Ultrasound/Scintigraphy (Technetium-99m):** To differentiate agenesis, ectopy, or dyshormonogenesis.
- **Tier 3:** Anti-TPO and Anti-thyroglobulin antibodies (if Hashimoto's suspected).

Management Outline

- **Drug of Choice:** Levothyroxine (L-T4).
- **Dosing:** Age-dependent. Neonates require higher doses (10–15 mcg/kg/day) to prevent brain damage.
- **Administration:** Give on an empty stomach, 30–60 mins before breakfast. Do not mix with soy, iron, or calcium.
- **Monitoring:** Check TSH and FT4 every 1–2 months in the first year of life; every 3–6 months thereafter.

EXAMINER'S VIVA

- **Q: Why is the US:LS ratio high in hypothyroidism?**
    - A: Thyroid hormone is essential for epiphyseal maturation and linear growth of long bones. Deficiency leads to failed limb elongation while the trunk continues to grow, preserving infantile proportions.
- **Q: How do you differentiate carotenemia from jaundice at the bedside?**

    - A: In carotenemia, the yellow pigment deposits in the stratum corneum (palms, soles, nasolabial folds), but the **sclera remains white**. In jaundice, the sclera is yellow.
- **Q: What is the most common cause of preventable intellectual disability worldwide?**
    - A: Congenital Hypothyroidism.
- **Q: Describe the technique for measuring the thyroid gland.**
    - A: (Demonstrate standing behind the patient, using the pads of the fingers, palpating while the patient swallows).
- **Q: What is "Bone Age" and what do you expect in this child?**
    - A: Bone age is the degree of maturation of a child's bones. In hypothyroidism, bone age is significantly delayed and usually lags behind even the height age.

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting the case of [Name], a [Age] year old male, who presented with chief complaints of failure to gain height for 2 years and sluggishness..."
- **Mistake:** Don't just say "the child is short." You must state if the shortness is **proportionate or disproportionate**.
- **Focus:** For PGs, the examiner wants to see your **growth chart plotting** and your **US:LS ratio technique**. If you get these wrong, the rest of the case fails.
- **Nuance:** Mention "School Performance" specifically. A drop in grades is a sensitive indicator of acquired hypothyroidism in an older child.

---

36. Short stature

Subject: Endocrine

LONG CASE FRAMEWORK: SHORT STATURE

HISTORY

Chief Complaint

- "My child is the shortest in the class" or "Not outgrowing clothes for the last 2 years."
- Age-specific: In toddlers, it’s often "failure to thrive"; in adolescents, it’s "delayed puberty/not catching up to peers."

History of Present Illness

Ask these questions to differentiate between Constitutional, Familial, Endocrine, and Systemic causes:

- **Birth Weight and Gestation:** "Was the baby born small for gestational age (SGA)?" (SGA children who don't catch up by age 2 need GH consideration).

- **Growth Velocity:** "Has he stayed the same size for a year, or is he slowly growing but always behind?" (Flat curve = Pathological; Parallel to curve = Constitutional/Familial).
- **Nutritional Intake:** "Tell me exactly what he ate yesterday from morning to night." (Rule out malnutrition/caloric deficit).
- **Systemic Review (The "Hidden" Chronic Illnesses):**
    - "Any chronic cough or steatorrhea?" (Cystic Fibrosis/Celiac).
    - "Does he have frequent unexplained fevers or joint pains?" (Juvenile Idiopathic Arthritis/SLE).
    - "How is his appetite and bowel habit?" (Constipation + poor appetite = Hypothyroidism).
    - "Does he have polyuria/polydipsia?" (Renal Tubular Acidosis/Diabetes Insipidus).
- **Neurological/Midline Defects:** "Any history of headaches, visual changes, or was he born with a cleft lip?" (Pituitary tumors or Septo-optic dysplasia).

Relevant Background History

- **Antenatal/Birth:** Look for breech delivery, birth asphyxia, or neonatal hypoglycemia/prolonged jaundice (clues to Growth Hormone Deficiency).
- **Developmental:** Global delay suggests chromosomal issues or hypothyroidism; normal development with short stature suggests isolated GH deficiency or Turner syndrome.
- **Family History [CRITICAL]:**
    - **Mid-Parental Height (MPH):** Get exact heights of both parents.
    - **Pubertal Timing:** "When did the mother have her first period? Did the father continue growing after high school?" (Late bloomers = Constitutional Delay of Growth and Puberty - CDGP).
- **Socioeconomic:** Psychosocial dwarfism (emotional deprivation) is a real differential in neglected children.

EXAMINATION

General Survey

- **Proportion:** Look at the child from a distance. Does the trunk look too long for the legs (Achondroplasia/Skeletal dysplasias) or is the shortening symmetrical?
- **Facies:**
    - **"Doll-like" facies:** Chubby cheeks, high forehead, depressed nasal bridge (GH Deficiency).
    - **Coarse facies:** Large tongue, thick skin (Hypothyroidism).
    - **Elfin facies:** (Williams Syndrome).

    - **Webbed neck/Low hairline:** (Turner Syndrome - check every short girl for this).
- **Nutritional Status:** Assess for "Chubby but short" (Endocrine: GH deficiency/Cushing's) vs. "Thin and short" (Systemic illness/Malnutrition).

Vital Signs and Anthropometry

- **Height/Length:** Use a **Stadiometer** for >2 years. Ensure 5-point contact (Heels, calves, buttocks, scapulae, occiput). Head in **Frankfort Plane**. Measure 3 times and average.
- **Weight:** Measure accurately; plot Weight-for-Height.
- **Arm Span:** Should equal height. If Arm Span < Height by >5cm, suspect skeletal dysplasia.
- **Upper Segment (US) to Lower Segment (LS) Ratio:**
    - *Technique:* Measure LS from symphysis pubis to floor. US = Total Height minus LS.
    - *Significance:* At birth 1.7:1; at 7 years 1:1; Adults 0.9:1. Increased ratio = Hypothyroidism or Skeletal Dysplasia.
- **Growth Velocity:** If you have two points in time, calculate cm/year. <4cm/year in a school-age child is pathological.

Peripheral Signs

- **Hands:**
    - **Trident hand:** (Achondroplasia).
    - **Short 4th metacarpal:** (Turner syndrome - Archibald's sign).
    - **Nail dysplasia:** (Turner syndrome).
- **Eyes:** Check for papilledema (increased ICP from craniopharyngioma) and visual fields by confrontation.
- **Thyroid:** Palpate for goiter (Hashimoto's is a common cause of growth failure).
- **Midline defects:** Check for a single central incisor [EXAMINER FAVORITE] (Sign of Holoprosencephaly/GH deficiency).

Systemic Examination — Primary System (Endocrine/Auxology)

- **Tanner Staging (Puberty):** You **must** stage the child.
    - Short stature + Delayed puberty = CDGP or Hypopituitarism.
    - Short stature + Precocious puberty = Early growth plate closure (e.g., CAH).
- **Abdomen:** Palpate for organomegaly (Storage diseases) or masses.
- **Skin:** Check for striae, buffalo hump, or central obesity (Cushing's). Check for dry, cold skin (Hypothyroidism).

Systemic Examination — Secondary Systems

- **CNS:** Fundoscopy and visual fields are mandatory if you suspect a pituitary tumor.

- **CVS:** Murmur of bicuspid aortic valve or Coarctation of Aorta (Turner Syndrome).

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **Micropenis/Hypoglycemic seizures:** Points strongly to Congenital Hypopituitarism.
- **Cubitus Valgus:** Increased carrying angle in girls (Turner Syndrome).
- **Frontal Bossing/Midface Hypoplasia:** Classic for Achondroplasia or GH deficiency.
- **Goiter + Delayed Bone Age:** Hypothyroidism.

Severity Assessment

- **Height SDS (Standard Deviation Score):**
    - Mild: -2 to -2.5 SD.
    - Severe: < -3 SD (Requires urgent endocrine workup).
- **[SEVERITY MARKER]:** Crossing two major centiles downwards on the growth chart.

DIAGNOSIS

Diagnostic Criteria

- **Short Stature:** Height < 3rd centile or < -2 SD for age and sex on appropriate population charts.

Differentials

1. **Constitutional Delay of Growth and Puberty (CDGP):** "Late bloomers," family history of late puberty, bone age = height age.
2. **Familial Short Stature (FSS):** Short parents, normal growth velocity, bone age = chronological age.
3. **Growth Hormone Deficiency (GHD):** Decreased growth velocity, doll-like facies, truncal obesity, delayed bone age.
4. **Turner Syndrome:** Girls, webbed neck, increased US:LS ratio, primary amenorrhea.
5. **Celiac Disease:** May present with *only* short stature without overt GI symptoms ("Silent Celiac").

Investigations

- **Tier 1 (Screening):** CBC, ESR (Chronic inflammation), Creatinine/Electrolytes (RCD), Tissue Transglutaminase (tTG) IgA (Celiac), TSH/Free T4.
- **Tier 2 (Bone Age):** X-ray of Left Hand and Wrist (Greulich-Pyle Atlas).
    - Delayed in GHD, Hypothyroidism, CDGP.
    - Normal in FSS.
- **Tier 3 (Confirmatory):**

- **GH Stimulation Tests:** (Clonidine, Insulin, or Glucagon). Peak GH <10 ng/mL is diagnostic of deficiency.
- **Karyotype:** Mandatory for any girl with unexplained short stature.
- **MRI Brain:** To visualize the pituitary/hypothalamus if GH deficiency is proven.

Management Outline

- **Nutritional Rehabilitation:** If systemic/nutritional.
- **Thyroxine:** If hypothyroid (Do not give GH first as it can worsen the metabolic state).
- **Recombinant Human Growth Hormone (rhGH):**
  - Indications: GHD, Turner syndrome, SGA (no catch-up), Chronic Renal Failure, Prader-Willi.
  - Route: Daily subcutaneous injection at night.
- **Monitoring:** Height every 3–6 months, monitor for side effects (Slipped Capital Femoral Epiphysis, Pseudotumor Cerebri).

EXAMINER'S VIVA

1. **Q: How do you calculate Mid-Parental Height (MPH)?**
   - A: Boys: [Father's height + Mother's height + 13cm] / 2. Girls: [Father's height + Mother's height - 13cm] / 2. Target range is ± 5cm.
2. **Q: What is the difference between "Height Age" and "Bone Age"?**
   - A: Height age is the age at which the child's current height would be the 50th centile. Bone age is the biological maturation of the skeleton.
3. **Q: Why is a single GH level useless?**
   - A: GH is secreted in pulsatile bursts. A random low level could just be the "trough" between pulses. Stimulation tests are required.
4. **Q: [Technique] Show me how to measure the Upper Segment:Lower Segment ratio.**
   - A: (Demonstrate measuring from the top of the pubic symphysis to the floor for LS, then subtracting from total height for US).
5. **Q: When do you stop GH therapy?**
   - A: When growth velocity falls below 2cm/year or when bone age reaches 14 (girls) or 16 (boys).

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting [Name], a [Age] year old male, whose height is [X] cm, which is well below the 3rd centile, with a growth velocity of [Y] cm/year, suggesting pathological short stature."
- **Common Mistake:** Forgetting to check the child's birth weight. If they were SGA, the entire diagnostic pathway changes.
- **What Examiners Watch For:** They watch your stadiometer technique. If you don't remove the child's shoes or don't check the Frankfort plane, you will lose marks immediately.

---

37. Disorders of puberty

Subject: Endocrine

This is a high-stakes long case. In puberty cases, the examiner is not just looking for a diagnosis; they are watching your **sensitivity, privacy maintenance, and precision in staging.** You must demonstrate that you can distinguish between "normal variation," "early/late but normal," and "pathological."

HISTORY

Chief Complaint

- **Precocious Puberty:** "Appearance of [secondary sexual characteristic] before age 8 in girls or age 9 in boys."
- **Delayed Puberty:** "No signs of puberty by age 13 in girls (no breast bud) or age 14 in boys (testicular volume <4ml)."

History of Present Illness

- **The "First Sign":** Ask the mother, "What was the very first change you noticed? Was it breast development, pubic hair, body odor, or a sudden increase in height?" (Helps distinguish *Thelarche* vs. *Adrenarche*).
- **Tempo of Progression:** "How quickly have these changes progressed? Has she had her first period (menarche) yet?" (Rapid progression suggests central pathology).
- **Growth Velocity:** "Have you noticed the child outgrowing shoes or clothes faster than peers? Has he/she suddenly become the tallest in class?" [SEVERITY MARKER: Rapid height gain suggests bone age advancement].
- **Neurological Red Flags:** "Does the child have headaches, morning vomiting, or any changes in vision/peripheral sight?" (Crucial for hypothalamic/pituitary tumors).
- **Behavioral Changes:** "Any mood swings, increased aggression, or 'adult-like' interests?"
- **Exogenous Exposure:** "Are there any hormonal creams, hair products, or medications (OCPs) in the house the child might have touched or ingested?"

Relevant Background History

- **Antenatal/Birth:** Ask about birth weight and length. (SGA infants are at higher risk for premature adrenarche and PCOS). Ask about neonatal "mini-puberty" (breast buds/vaginal bleed in infancy).
- **Developmental:** "Was there any delay in milestones?" (Associated with certain syndromes like Prader-Willi or Septo-optic dysplasia).
- **Family History [CRITICAL]:** "At what age did the mother have her first period? At what age did the father have his 'growth spurt' or start shaving?" (To identify **Constitutional Delay of Growth and Puberty - CDGP**).
- **CNS Insults:** History of meningitis, encephalitis, head trauma, or cranial irradiation.

EXAMINATION

General Survey

- **The "First Impression":** Does the child look older than their stated age? Look at the face—is there acne, comedones, or a "mature" facial contour?
- **Stature:** Measure Standing Height and Sitting Height (to calculate Upper Segment: Lower Segment ratio).
    - *Significance:* Hypothyroidism (delayed puberty) causes an increased US:LS ratio (immature proportions).
- **Skin:**
    - **Café-au-lait spots:** Look for jagged "Coast of Maine" borders (McCune-Albright Syndrome).
    - **Acanthosis Nigricans:** Check neck and axilla (Insulin resistance/PCOS).
    - **Axillary hair/Sweat odor:** Markers of adrenarche.

Vital Signs and Anthropometry

- **Blood Pressure:** Use the correct cuff size. (Elevated in certain CAH types like 11-beta hydroxylase deficiency).
- **Growth Charting [MANDATORY]:** Plot Height, Weight, and BMI on age-appropriate charts (WHO or IAP).
    - *Finding:* In precocious puberty, the height curve will cross percentiles upwards (acceleration). In delayed puberty/hypothyroidism, the curve flattens.

Peripheral Signs

- **Eyes:** Visual field testing by confrontation (looking for bitemporal hemianopia from a pituitary mass). Fundoscopy for papilledema.
- **Thyroid:** Palpate for goiter (Hypothyroidism is a common cause of delayed puberty).
- **Hands:** Check for short 4th/5th metacarpals (Turner Syndrome).

- **Midline Defects:** Look for cleft lip/palate or a single central incisor (associated with GnRH deficiency/Kallmann syndrome).

Systemic Examination — Primary System (Genitalia & Pubertal Staging)

This is the core of the case. You must ask for a chaperone and ensure privacy.

1. Tanner Staging (Sexual Maturity Rating - SMR)

- **Girls (Breast - B1 to B5):**
    - *Technique:* Palpate the breast tissue, don't just look. Distinguish between true glandular tissue (firm, subareolar) and "lipomastia" (soft fat in obese girls).
    - *B2 (Thelarche):* Breast bud—elevation of breast and papilla as a small mound, enlargement of areolar diameter.
- **Boys (Genitalia - G1 to G5):**
    - *Technique:* Use an **Orchidometer** [EXAMINER FAVORITE].
    - *Significance:* Testicular volume **≥ 4 ml** (or length > 2.5 cm) defines the onset of true puberty (G2).
    - *Discordance:* If a boy has pubic hair (P2/3) but pre-pubertal testes (<4ml), the source of androgens is peripheral (Adrenal/Tumor), not the brain.
- **Pubic Hair (P1 to P5):** Describe distribution and texture.

2. Specific Genital Exam

- **Girls:** Look for estrogenization of the vaginal mucosa (pink/moist vs. red/shiny) and any discharge.
- **Boys:** Check for hypospadias or hyperpigmentation of the scrotum (suggests high ACTH/CAH).

Systemic Examination — Secondary Systems

- **CNS:** Detailed cranial nerve exam (I, II, III, IV, VI). Smell test (Coffee/Soap) to rule out **Kallmann Syndrome** (Anosmia + Hypogonadotropic Hypogonadism).
- **Abdomen:** Palpate for masses (Ovarian or Adrenal tumors).

SIGNS THAT CLINCH THE DIAGNOSIS [EXAMINER FAVORITES]

- **Testicular Volume (Orchidometer):**
    - *Bilateral Enlargement (≥4ml):* Central Precocious Puberty (CPP).
    - *Pre-pubertal Testes with Pubic Hair:* Peripheral Precocious Puberty (PPP) or Premature Adrenarche.
    - *Unilateral Enlargement:* Testicular tumor.
- **The "Coast of Maine" Café-au-lait spot:** Large, irregular, unilateral spots + Precocious puberty = **McCune-Albright Syndrome**.

- **Anosmia:** Inability to smell + delayed puberty = **Kallmann Syndrome**.
- **Stigmata of Turner Syndrome:** Webbed neck, low posterior hairline, wide-spaced nipples (Shield chest), cubitus valgus.

DIAGNOSIS

Diagnostic Criteria

- **Precocious:** Secondary sexual characteristics <8y (F) or <9y (M).
- **Delayed:** No breast buds by 13y (F); Testicular vol <4ml by 14y (M).

Differentials

1. **Central Precocious Puberty (GnRH Dependent):** Idiopathic (most common in girls) or CNS lesion (Hamartoma).
2. **Peripheral Precocious Puberty (GnRH Independent):** CAH, McCune-Albright, Ovarian/Testicular tumors.
3. **Normal Variations:** Isolated Premature Thelarche or Adrenarche.
4. **Delayed Puberty:** CDGP (most common), Hypergonadotropic Hypogonadism (Turner/Klinefelter), Hypogonadotropic Hypogonadism (Kallmann, Chronic illness).

Investigations

- **Tier 1: Bone Age X-ray (Left Hand/Wrist).** If Bone Age > Chronological Age by >1 year, it confirms a significant hormonal effect.
- **Tier 2:**
    - **Basal LH/FSH:** High LH (>0.3 IU/L) suggests Central Puberty.
    - **GnRH Stimulation Test:** Gold standard. Peak LH >5 IU/L confirms Central Precocious Puberty.
    - **Pelvic USG (Girls):** Look for uterine length >3.4 cm and ovarian volume >2 ml.
- **Tier 3: MRI Brain (Sella/Hypothalamus)** to rule out tumors in all boys with CPP and girls <6 years with CPP.

MANAGEMENT OUTLINE

- **Central Precocious Puberty: GnRH Analogs** (e.g., Leuprolide depot) to "pause" puberty and preserve adult height.
- **Peripheral:** Treat the cause (e.g., Surgery for tumors, Steroids for CAH).
- **Delayed (CDGP):** Reassurance or "jump-start" with low-dose Testosterone (boys) or Estrogen (girls) for 3-6 months.

EXAMINER'S VIVA

1. **Q: How do you distinguish Premature Thelarche from Central Precocious Puberty?**
    - *A:* Thelarche is isolated breast development without increased growth velocity, without advanced bone age, and with pre-pubertal LH levels.
2. **Q: What is the significance of "discordant" puberty in a boy?**
    - *A:* If he has pubic hair and acne but small testes, it indicates a peripheral source of androgens (like CAH or adrenal tumor), as the testes are not being stimulated by pituitary LH.
3. **Q: Why do children with precocious puberty end up as short adults?**
    - *A:* Sex steroids cause rapid bone maturation and early fusion of the epiphyseal plates.
4. **Q: Show me how to use an orchidometer.**
    - *A:* (Demonstrate) Hold the testis with one hand, find the matching ellipsoid with the other. 4ml is the cutoff for puberty.

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting [Name], a [Age] year old [Male/Female], who presented with [Breast development/Testicular enlargement] noted since the age of [X], which is [Precocious/Delayed] for their age."
- **Mistake:** Forgetting to mention the mother's age of menarche. This is the single most common omission in a puberty case.
- **Ethics:** Always mention that the genital exam was done in the presence of a guardian and a chaperone.

Growth and Development

38. Failure to thrive

Subject: Growth and Development

This is a classic "bread and butter" long case that tests your ability to be a clinical detective. In Failure to Thrive (FTT), the diagnosis isn't just a label; it’s a process of identifying the **mechanism** (inadequate intake, malabsorption, or increased demand).

HISTORY

Chief Complaint

- "Not gaining weight" or "looking small/thin" compared to peers.
- "Poor feeding" or "refusal to eat."

- Associated symptoms: Diarrhea, cough, or developmental delay.
- *Note:* Duration is critical. Is it a recent drop or has the child always been below the 3rd centile?

History of Present Illness

You must categorize the HPI into the "Three Buckets of FTT":

1. Inadequate Caloric Intake (The most common)

- "Tell me exactly what the child ate yesterday from the moment they woke up." (24-hour dietary recall).
- "How is the formula prepared? Show me the scoop and the bottle." (Look for over-dilution).
- "Does the child choke, gag, or tire during feeds?" (Suggests oromotor dysfunction or CHD).
- "How long does a feed take?" (>30 mins suggests struggle; <5 mins suggests neglect or poor supply).

2. Excessive Caloric Loss (Malabsorption/Loss)

- "Describe the stools. Are they oily, foul-smelling, and hard to flush?" (Steatorrhea/Cystic Fibrosis/Celiac).
- "Is there vomiting? Is it projectile or effortless spit-up?" (GERD/Pyloric stenosis).
- "Are there worms in the stool or perianal itching?"

3. Increased Caloric Demand (Chronic Disease)

- "Does the child breathe fast even while sleeping?" (CHD/Chronic Lung Disease).
- "Are there frequent fevers or recurrent pneumonias?" (Immunodeficiency/TB/HIV).

Relevant Background History

- **Antenatal/Birth:** "Was the child small at birth (SGA)?" SGA children have different catch-up trajectories than AGA children who fall off the curve.
- **Developmental:** "When did he start walking?" FTT can cause global delay, but if delay preceded the growth failure, think of organic brain syndromes or metabolic disorders.
- **Social/Environmental:** "Who feeds the child? Is there enough food in the house? Are the parents stressed?" [CRITICAL] You must screen for neglect without being accusatory.
- **Immunization:** Focus on Measles (can trigger malnutrition) and BCG (TB risk).

EXAMINATION

General Survey

- **Observation:** Is the child "happy but small" (constitutional/genetic) or "miserable and wasted" (malnutrition/organic disease)?
- **Activity:** Does the child interact with the environment or sit listlessly?

- **Wasting:** Look for the "Old Man Facies" (loss of buccal fat pads—this is the *last* fat store to go, indicating severe chronicity).
- **Skin:** Look for "flaky paint" dermatosis (Kwashiorkor) or excessive wrinkling of the skin on the buttocks ("baggy pants" appearance).

Vital Signs and Anthropometry

- **Heart Rate:** Tachycardia might suggest CCF or hyperthyroidism; Bradycardia suggests severe marasmus or hypothyroidism.
- **Anthropometry [THE CORE OF THE CASE]:**
    - **Weight-for-age:** Reflects acute-on-chronic status.
    - **Weight-for-height:** Reflects **Wasting** (Acute malnutrition).
    - **Height-for-age:** Reflects **Stunting** (Chronic malnutrition).
    - **Head Circumference:** If this is also low, think of TORCH infections, genetic syndromes, or very early severe malnutrition.
    - **Mid-Upper Arm Circumference (MUAC):** Use the Shakir tape. <11.5 cm in 6–59 months indicates SAM.

Peripheral Signs

- **Hands/Nails:** Look for koilonychia (Iron deficiency) or clubbing (Cystic Fibrosis, Cyanotic CHD, or Celiac).
- **Eyes:** Bitot's spots or xerosis (Vitamin A deficiency is common in FTT).
- **Edema:** Check the dorsum of the feet. If bilateral pitting edema is present, it is **Kwashiorkor** regardless of anthropometry.
- **Hair:** Flag sign (alternating bands of light and dark hair) indicating intermittent protein intake.

Systemic Examination — Primary Focus

- **Abdomen:**
    - **Inspection:** Scaphoid (severe wasting) or Distended (Celiac, Malabsorption, or Giardiasis).
    - **Palpation:** Check for hepatomegaly (Fatty liver in Kwashiorkor or Congestive hepatomegaly in CHD).
- **Respiratory:** Listen for crackles (Bronchiectasis/CF) or signs of respiratory distress.
- **Cardiovascular:** Listen for murmurs. A quiet precordium with a gallop may suggest nutritional cardiomyopathy.

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **Loss of Buccal Fat Pads:** Indicates the most severe stage of calorie deprivation.

- **Baggy Pants Appearance:** Loss of gluteal fat; indicates significant wasting.
- **Pot Belly with Wasted Extremities:** Classic for protein-energy malnutrition or Celiac disease.
- **Visible Peristalsis:** In a thin child, may suggest a partial obstruction or simply extreme thinness.

Severity Assessment [SEVERITY MARKER]

- **Red Flags:** Lethargy, hypothermia, hypoglycemia, or visible severe wasting (MUAC <11.5cm).
- **The "Shift":** If the child's growth curve crosses two major centile lines (e.g., from 50th to 5th), it is clinically significant FTT.

DIAGNOSIS

Diagnostic Criteria

- **Gomez Classification:** Based on Weight-for-Age.
- **Waterlow Classification:** Distinguishes Wasting (Wt/Ht) from Stunting (Ht/Age).
- **WHO Criteria for SAM:** Wt/Ht < -3SD, MUAC <11.5cm, or nutritional edema.

Differentials

1. **Non-Organic FTT (Psychosocial):** Most common. History of poverty or poor feeding technique.
2. **Celiac Disease:** Distended abdomen, wasted buttocks, irritability.
3. **Congenital Heart Disease:** Tachypnea, murmur, diaphoresis during feeds.
4. **Cystic Fibrosis:** Recurrent pneumonia + steatorrhea.

Investigations

- **Tier 1:** CBC (anemia), Urine R/M (UTI is a silent cause of FTT), Stool for ova/parasites/reducing substances.
- **Tier 2:** Electrolytes, Albumin, Liver Function Tests, Sweat Chloride test (if respiratory symptoms), Celiac Serology (Anti-tTG IgA).
- **Tier 3:** Bone age X-ray (to assess growth potential), Echocardiogram.

Management Outline

1. **Stabilization:** Treat hypoglycemia, hypothermia, and infection.
2. **Nutritional Rehabilitation:**
    - **Initial phase:** F-75 diet (low protein/calorie to prevent Refeeding Syndrome).
    - **Rehabilitation phase:** F-100 or Ready-to-Use Therapeutic Food (RUTF).
3. **Catch-up Growth:** Aim for 10-15g/kg/day of weight gain.
4. **Social Intervention:** Parental counseling and feeding education.

EXAMINER'S VIVA

Q: How do you distinguish between organic and non-organic FTT on history? A: Organic FTT children are often "difficult" to feed (vomiting, diarrhea, cough), whereas non-organic FTT children are often "not offered" enough food or have poor interaction with the caregiver.

Q: What is the "Rule of Thumb" for expected weight gain in an infant? A: Double birth weight by 5 months, triple by 1 year, quadruple by 2 years.

Q: [TECHNIQUE] How do you measure height vs. length? A: Use a stadiometer for children >2 years (standing). Use an infantometer for children <2 years (supine). Never use a measuring tape on a bed; it is inaccurate due to surface contour.

Q: What is Refeeding Syndrome? A: A life-threatening shift in electrolytes (primarily hypophosphatemia, hypokalemia, and hypomagnesemia) that occurs when starting high-calorie feeds too quickly in a starved child.

LONG CASE PRESENTATION TIPS

- **The Opening:** "I am presenting [Name], a [Age] old male, who presents with a primary concern of failure to gain weight over the last [Duration], currently plotting below the 3rd centile for weight and height."
- **Common Mistake:** Forgetting to calculate the **Caloric Deficit**. You must be able to tell the examiner exactly how many calories the child is currently getting vs. what they *should* be getting for their "Ideal Weight for Height."
- **What Examiners Watch For:** They watch how you handle the child. A child with FTT is often irritable. If you can soothe the child while performing a meticulous anthropometric measurement, you demonstrate senior-level clinical competence.

---

39. Rickets

Subject: Growth and Development

HISTORY

Chief Complaint

- **Infants (6–18 months):** Delayed motor milestones (not sitting/walking), "softness" of the head, or abnormal skull shape.
- **Toddlers/Older Children:** Bowing of legs, waddling gait, or "pigeon chest" deformity.
- **Acute presentation:** Seizures (hypocalcemic), tetany, or frequent respiratory infections.

History of Present Illness

- **Motor Milestones:** Ask: "When did your child start holding their head, sitting without support, and standing?" (Rickets causes hypotonia and delayed gross motor skills).
- **Bony Deformities:** Ask: "When did you first notice the bowing of the legs? Is it getting worse? Does the child complain of leg pain when walking?"
- **Dentition:** Ask: "When did the first tooth erupt? Are the teeth decaying easily?" (Delayed dentition and enamel hypoplasia are common).
- **Hypocalcemic Symptoms:** Ask: "Has the child ever had a fit, or sudden stiffening of the body, or a strange 'crowing' sound while breathing (laryngospasm)?"
- **Respiratory:** Ask: "Does the child get frequent coughs and colds?" (Rachitic children have poor chest expansion and are prone to pneumonia).
- **Dietary History (Crucial):**
    - "Is the child exclusively breastfed? For how long?" (Breast milk is low in Vitamin D).
    - "When were solids started? What exactly does the child eat?" (Look for lack of fortified cereals/milk).
    - "Does the child drink cow's milk? How much?" (High phosphate in cow's milk can worsen calcium balance).
- **Sunlight Exposure:** "How many hours a day does the child play outside? Is the child kept covered or indoors most of the day?" (Assess for Vitamin D synthesis).

Relevant Background History

- **Antenatal/Birth:** Ask about maternal Vitamin D intake and sun exposure. Ask if the child was premature (Preterm rickets is a distinct entity due to low mineral stores).
- **Past History:** Ask about chronic diarrhea or oily stools (Malabsorption/Celiac disease) and history of jaundice (Biliary atresia/Chronic liver disease).
- **Family History:** "Are there other family members with short stature or leg deformities?" (Points toward X-linked Hypophosphatemic Rickets).
- **Socioeconomic:** Overcrowding and lack of open spaces (limited sun exposure).

EXAMINATION

General Survey

- **Observation:** Look for a "miserable" child (chronic bone pain). Observe the gait—look for a **Waddling Gait** (due to pelvic girdle weakness and coxa vara).
- **Activity:** Note if the child is hypotonic (the "floppy" rachitic child).
- **Nutritional Status:** Check for concomitant Protein Energy Malnutrition (PEM). Rickets often co-exists with Vitamin A and Iron deficiency.

Vital Signs and Anthropometry

- **Height/Length:** [SEVERITY MARKER] Plot on WHO charts. Stunting is common in chronic rickets.
- **Head Circumference:** May be increased (Frontal bossing gives a false impression of hydrocephalus).
- **Vitals:** Check for tachypnea (if the child has a "soft" chest or pneumonia).

Peripheral Signs

- **Hands:** Look for **Wrist Widening** (at the distal radius/ulna).
- **Nails/Eyes:** Check for pallor (Iron deficiency is a common co-morbidity).
- **Lymph Nodes:** Generally not enlarged unless there is an active infection.

Systemic Examination — Primary System (Skeletal)

- **Head:**
    - **Craniotabes:** [EXAMINER FAVORITE] *Technique:* Press firmly with your thumb over the occipital or posterior parietal bones (avoid sutures). *Finding:* A "ping-pong ball" sensation (collapsing and recoiling). *Significance:* Only significant after 3 months of age (normal in neonates).
    - **Frontal Bossing:** Inspect from the side for a prominent forehead.
    - **Large Anterior Fontanelle:** Feel for delayed closure (normally closes by 18 months).
- **Chest:**
    - **Rachitic Rosary:** [EXAMINER FAVORITE] *Technique:* Palpate the costochondral junctions lateral to the sternum using your middle three fingers. *Finding:* Bead-like enlargements. *Significance:* These are non-tender (unlike the "scorbutic rosary" of Scurvy, which is sharp and tender).
    - **Harrison's Sulcus:** Look for a horizontal groove along the lower border of the thorax at the insertion of the diaphragm.
    - **Pigeon Chest (Pectus Carinatum):** Anterior projection of the sternum.
- **Extremities:**
    - **Wrist Widening:** Palpate the distal ends of the radius and ulna.
    - **Genu Varum (Bowing):** Measure the **Inter-condylar distance** with the medial malleoli touching.
    - **Genu Valgum (Knock-knees):** Measure the **Inter-malleolar distance** with the knees touching.
    - **Double Malleoli (Marfan's Sign):** Look for a second prominence above the medial malleolus due to widening of the distal tibia.
- **Spine:** Look for **Rachitic Kyphosis** (sitting cat deformity)—a smooth curve that disappears when the child is placed prone (unlike the fixed gibbus of Pott's disease).

Systemic Examination — Secondary Systems

- **Abdomen:** Look for **Pot-belly** (due to hypotonia of abdominal muscles) and palpate for hepatosplenomegaly (associated with anemia/infections).
- **Neuromuscular:**
    - **Chvostek Sign:** Tap over the facial nerve in front of the ear. Positive if the upper lip twitches (Latent tetany).
    - **Trousseau Sign:** Inflate BP cuff above systolic pressure for 3 mins. Positive if carpal spasm occurs.

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **Non-tender Rachitic Rosary:** Indicates expansion of the osteoid at the costochondral junction.
- **Wrist Widening:** The most reliable peripheral sign of active rickets in a walking child.
- **Craniotabes:** The earliest sign of rickets in an infant.
- **Waddling Gait:** Indicates proximal muscle weakness and pelvic deformities.

Severity Assessment

- **Mild:** Only biochemical changes or subtle wrist widening.
- **Moderate:** Visible bony deformities (bowing, rosary) but child is ambulatory.
- **Severe:** [SEVERITY MARKER] Inability to walk, "cat-back" kyphosis, fractures (pseudofractures), or symptomatic hypocalcemia (seizures/stridor).

DIAGNOSIS

Diagnostic Criteria

- Diagnosis is clinical (skeletal signs) + Biochemical (High ALP, Low/Normal Ca, Low PO4) + Radiological (Cupping/Fraying).

Differentials

1. **Scurvy:** Tender, sharp rosary; subperiosteal bleeds; "froglike" position; no wrist widening.
2. **Achondroplasia:** Short-limb dwarfism, trident hand, normal biochemistry.
3. **Physiological Bowing:** Common in children <2 years; no other rachitic signs; normal biochemistry.
4. **Blount's Disease:** Idiopathic tibia vara; localized to the medial tibial epiphysis.

Investigations

- **Tier 1 (Biochemical):** Serum Calcium (low/normal), Phosphorus (low), **Alkaline Phosphatase (elevated - best marker for activity)**.

- **Tier 2 (Radiological):** X-ray of the wrist (AP view). Look for:
    - **Cupping:** Concave distal metaphysis.
    - **Fraying:** Shaggy, irregular metaphyseal margins.
    - **Splaying:** Widening of the metaphysis.
- **Tier 3 (Specialized):** 25-OH Vitamin D levels (low in nutritional), PTH (elevated), Urine Calcium:Creatinine ratio (to exclude renal causes).

Management Outline

- **Stoss Therapy:** Single oral dose of 300,000 to 600,000 IU Vitamin D3 (if compliance is an issue).
- **Daily Therapy:** 2,000–6,000 IU/day of Vitamin D3 for 2–3 months, followed by 400–600 IU/day.
- **Calcium Supplementation:** 50–75 mg/kg/day of elemental calcium is mandatory to avoid "Hungry Bone Syndrome."
- **Monitoring:** Repeat ALP and X-ray at 4 weeks. The first sign of healing is the **line of calcification** across the zone of provisional calcification.

EXAMINER'S VIVA

1. **Q: Why is the rosary in rickets not tender, while in Scurvy it is?**
    - *A:* In rickets, the enlargement is due to excess uncalcified osteoid (soft tissue expansion). In Scurvy, it is due to micro-fractures and subperiosteal hemorrhage, which is highly painful.
2. **Q: What is the first radiological sign of healing?**
    - *A:* Appearance of a dense transverse line (line of provisional calcification) across the metaphyseal end.
3. **Q: How do you differentiate between Vitamin D dependent and Vitamin D resistant rickets?**
    - *A:* Resistant rickets (Hypophosphatemic) usually presents later, has a family history, and has normal Serum Calcium and PTH, but very low Serum Phosphorus.
4. **Q: What is "Hungry Bone Syndrome"?**
    - *A:* When Vitamin D treatment starts, calcium rapidly moves from the blood into the "hungry" demineralized bones, potentially causing acute hypocalcemic tetany.
5. **Q: Technique: How do you measure the inter-condylar distance?**
    - *A:* With the child supine, bring the medial malleoli together. Use a tape measure or calipers to measure the distance between the medial femoral condyles. >5cm is usually pathological.

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting [Name], a [Age] old male, born of a non-consanguineous marriage, presenting with delayed walking and progressive bowing of legs since the age of 1 year..."
- **Mistake:** Forgetting to check the fontanelle or teeth in an older child.
- **Mistake:** Diagnosing rickets based solely on bowing without checking for wrist widening or rosary (bowing can be physiological).
- **Examiner Watch-point:** They will watch how you palpate the wrist and rosary. Use the finger pads, not the tips, and be gentle.

---

40. Down syndrome

Subject: Growth and Development

This is a classic "spot diagnosis" case, but the examiner isn't testing your ability to recognize the facies—they are testing your ability to systematically screen for the multi-system complications of Trisomy 21.

HISTORY

Chief Complaint

- Usually brought for "delayed milestones," "floppiness," "protruding tongue," or "not growing well."
- Duration: Since birth.

History of Present Illness

- **Developmental Delay:** Ask specifically: "When did he first hold his head up? When did he sit without support?" (Expect global delay, but social/language often relatively better than motor).
- **Feeding Difficulties:** "Does he tire easily during feeds? Does he turn blue or sweat profusely over the forehead while sucking?" (Points toward Congenital Heart Disease - CHD).
- **Recurrent Infections:** "How many times has he had pneumonia or ear discharge?" (Due to hypotonia, poor clearance, and immune deficiency).
- **Vision/Hearing:** "Does he follow light? Does he turn his head to loud sounds?" (High incidence of cataracts, refractive errors, and conductive hearing loss).
- **Thyroid/Growth:** "Is there excessive constipation or skin dryness?" (Hypothyroidism).
- **Neurological:** "Have you noticed any sudden jerking of limbs or neck?" (Infantile spasms/Seizures). "Any change in gait or neck pain?" (Atlanto-axial instability).

Relevant Background History

- **Antenatal:** "What was the mother's age at conception?" (**Critical:** Risk increases significantly >35 years). "Were any dual/triple markers or USG (nuchal translucency) done?"
- **Birth History:** Often born preterm or small for gestational age.

- **Family History:** "Are there any other children with similar features?" (Relevant for Robertsonian Translocation).
- **Socioeconomic:** Essential for planning rehabilitation and special schooling.

EXAMINATION

General Survey

- **Initial Observation:** Note the "Happy-go-lucky" personality. Observe the posture—often "frog-leg" position due to generalized hypotonia.
- **Nutritional Status:** Assess for stunting. Use **Down Syndrome-specific growth charts** (standard WHO charts will show them as "failing to thrive" incorrectly).
- **Facies [EXAMINER FAVORITE]:**
    - **Brachycephaly:** Flat occiput.
    - **Eyes:** Upslanting palpebral fissures, epicanthic folds, and **Brushfield spots** (speckled white spots in the iris—easier to see in light-colored eyes).
    - **Nose:** Flat nasal bridge.
    - **Mouth:** Small oral cavity leading to a "seemingly" large, protruding tongue (macroglossia) with fissuring (scrotal tongue).
    - **Ears:** Small, low-set, overfolded helix.

Vital Signs and Anthropometry

- **Pulse:** Check for tachycardia (Heart failure) or bradycardia (Hypothyroidism).
- **BP:** Measure in all four limbs if you suspect Coarctation (though less common in Down than Turner).
- **Head Circumference:** Usually microcephalic.

Peripheral Signs

- **Hands:**
    - **Simian Crease:** Single transverse palmar crease (present in 50%, but not pathognomonic).
    - **Clinodactyly:** Incurving of the 5th finger due to hypoplastic middle phalanx.
    - **Dermatoglyphics:** Increased ulnar loops.
- **Feet: Sandal Gap** (wide space between 1st and 2nd toes) and a plantar crease extending from the gap.
- **Skin:** Xerosis (dryness), cutis marmorata.
- **Joints:** Hyperflexibility. Check the "Thumb-to-forearm" apposition.

Systemic Examination — Primary System (Cardiovascular)

- *Why? 40-50% have CHD (AVSD is most common).*
- **Inspection:** Precordial bulge, increased apical impulse.
- **Palpation:** Feel for a hyperdynamic apex or a thrill at the left lower sternal border.
- **Auscultation:**
    - **Loud S2:** Suggests Pulmonary Hypertension [SEVERITY MARKER].
    - **Pansystolic murmur:** At the left lower sternal border (VSD or AVSD).
    - **Mid-diastolic rumble:** At the apex (increased flow across mitral valve in large left-to-right shunts).

Systemic Examination — Secondary Systems

- **Abdomen:** Check for **rectus diastasis** and umbilical hernia (common due to hypotonia). Palpate for hepatomegaly (Congestive Heart Failure).
- **CNS:**
    - **Tone:** Perform "Pull-to-sit" (look for head lag) and "Ventral suspension" (inverted U-shape).
    - **Reflexes:** Usually diminished (hyporeflexia).
- **Musculoskeletal:** Screen for **Atlanto-axial instability**. Ask the child to walk; look for broad-based gait or signs of spasticity (which would be a *change* from their baseline hypotonia).

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

1. **Upslanting palpebral fissures with epicanthic folds.**
2. **Generalized Hypotonia:** The "floppy" feel when lifting the child.
3. **Sandal Gap and Simian Crease.**
4. **Brushfield Spots:** Small white/gray spots on the periphery of the iris.
5. **Excess nuchal skin:** Redundant skin folds at the back of the neck.

DIAGNOSIS

Diagnostic Criteria

- Clinical diagnosis is based on the **Hall's Criteria** (10 signs; if 6 or more are present, the clinical diagnosis is highly likely).

Differentials

1. **Congenital Hypothyroidism:** Also has macroglossia, hypotonia, and developmental delay, but lacks the specific dysmorphic facies (upslanting eyes) and has a large posterior fontanelle.

2. **Zellweger Syndrome:** Severe hypotonia and high forehead, but usually associated with seizures and early liver dysfunction.
3. **Fragile X Syndrome:** (In older children) Large ears and macro-orchidism, but eyes are not upslanting.

Investigations

- **Tier 1 (Bedside/Basic):**
    - **Karyotyping:** [GOLD STANDARD] To differentiate Trisomy 21 (95%), Translocation (4%), or Mosaicism (1%).
    - **CBC:** Look for leukemoid reaction in neonates or increased risk of AML/ALL later.
- **Tier 2 (Confirmatory/Screening):**
    - **Echocardiography:** Mandatory for all Down syndrome patients at birth, regardless of murmur.
    - **Thyroid Function Test (TFT):** Annual screening for hypothyroidism.
- **Tier 3 (Complications):**
    - **Cervical Spine X-ray (Lateral):** Neutral, flexion, and extension views (after age 3) to check for Atlanto-axial subluxation.
    - **Bera/Audiometry:** Every 6–12 months.

MANAGEMENT

- **Multidisciplinary Approach:**
    1. **Medical:** Treat CHD (diuretics/surgery), Thyroid replacement.
    2. **Surgical:** Correction of AVSD (usually by 6 months), Duodenal atresia if present.
    3. **Rehabilitation:** Physiotherapy for hypotonia, Speech therapy, Occupational therapy.
    4. **Education:** Early intervention programs and special schooling.
    5. **Counseling:** Recurrence risk (1% for Trisomy 21; up to 10-15% if mother is a carrier of 14;21 translocation).

EXAMINER'S VIVA

1. **Q: How do you assess hypotonia in a neonate?**
    - A: Check for head lag on pull-to-sit, ventral suspension (inverted U), and the "scarf sign" (elbow crossing midline easily).
2. **Q: What is the most common cardiac lesion in Down Syndrome?**
    - A: Atrioventricular Septal Defect (AVSD), followed by VSD and ASD.

3. **Q: Why is it important to identify a translocation?**
    - A: Because if a parent is a balanced translocation carrier, the recurrence risk is much higher than the 1% seen in non-disjunction.
4. **Q: At what age do you screen for Atlanto-axial instability?**
    - A: Usually after age 3, before the child starts active sports or if they require surgery (for intubation safety).
5. **Q: What is "Mosaicism" in Down syndrome?**
    - A: A condition where some cells have 46 chromosomes and some have 47. These children often have milder clinical features and higher IQ.

LONG CASE PRESENTATION TIPS

- **The Opening:** "I am presenting [Name], a [Age] old male, born to a [Age] old mother, who presents with global developmental delay and dysmorphic features suggestive of a chromosomal anomaly, likely Down Syndrome."
- **The "Trap":** Don't just say "The child has a Simian crease." Say "There is a single transverse palmar crease, which is a recognized feature of Down syndrome but can be found in 5-10% of the normal population."
- **The "Must-Do":** Always mention that you checked the **red reflex** (to rule out congenital cataracts) and the **anal patency** (to rule out imperforate anus/Hirschsprung's). Examiners love multi-system screening.

---

41. Autism spectrum disorder

Subject: Growth and Development

This is a challenging long case because the "examination" is largely observational and behavioral. In a PG exam, the examiner isn't just looking for the diagnosis; they are watching how you interact with a child who has social communication deficits.

HISTORY

Chief Complaint

- "Not speaking yet" or "Speech is not clear" (Language delay is the most common presenting complaint).
- "Doesn't listen when called" (often mistaken for hearing loss).
- "Stays in his own world" or "Doesn't play with other children."
- Unusual behaviors (hand flapping, spinning).

History of Present Illness

Focus on the two core pillars of DSM-5: Social Communication and Restricted/Repetitive Patterns.

1. Social Communication & Interaction:

- **Joint Attention:** "If you point at a bird in the sky, does he look where you are pointing, or does he just look at your finger?" [CRITICAL DIFFERENTIATOR]
- **Social Reciprocity:** "Does he bring toys to show you just for fun, or only when he needs help?" "Does he smile back when you smile at him?"
- **Response to Name:** "When you call his name from another room, does he look up? How many times out of ten does he ignore you?"
- **Non-verbal communication:** "Does he use gestures like waving 'bye-bye' or nodding 'yes'?" "Does he take your hand and lead you to the fridge like a 'tool' rather than pointing?"

2. Restricted, Repetitive Patterns of Behavior (RRBs):

- **Stereotypies:** "Does he flap his hands when excited, rock his body, or walk on his toes?"
- **Insistence on Sameness:** "Does he get a meltdown if you take a different route to the park or change the furniture?"
- **Sensory Issues:** "Does he cover his ears at the sound of a pressure cooker?" "Does he sniff objects or stare at spinning fans/wheels for a long time?"

3. Associated Features:

- **Regression:** "Did he have words or social smiles earlier that he later lost?" (Occurs in ~30%, usually between 15-24 months).
- **Comorbidities:** Ask about sleep disturbances, pica, hyperactivity, and seizures.

Relevant Background History

- **Antenatal:** Maternal metabolic syndrome, valproate exposure, advanced parental age (both mother and father).
- **Developmental:** Detailed milestones. In ASD, motor milestones are often normal, but social/language milestones are delayed. Look for "Splinter Skills" (e.g., can read alphabet but can't ask for water).
- **Family History:** Ask about siblings with ASD, ADHD, or learning disabilities (High heritability).
- **Screening History:** "Has he been screened with M-CHAT-R/F at the 18 or 24-month visit?"

EXAMINATION

General Survey

- **The "Waiting Room" Observation:** Your exam starts before you touch the child. Observe the child in the mother's lap or playing on the floor.
- **Eye Contact:** Is it absent, fleeting, or "piercing" but socially inappropriate?
- **Activity Level:** Is he darting around the room (hyperactive) or sitting in a corner lining up blocks?

- **Attachment:** Does he use the mother as a "secure base," or is he indifferent to her presence/absence?

Vital Signs and Anthropometry

- **Head Circumference:** [EXAMINER FAVORITE] Measure accurately. Macrocephaly (HC >95th centile) is seen in 15-20% of ASD cases due to accelerated brain growth in early childhood.
- **Growth:** Plot on WHO charts. Look for failure to thrive (common in children with severe sensory-based food aversions).

Peripheral Signs

- **Skin:** Examine under a Wood's lamp if possible. Look for Ash-leaf spots or Shagreen patches. *Why?* Tuberous Sclerosis is a major genetic association with ASD.
- **Dysmorphism:** Look for large ears and a long face (Fragile X Syndrome).
- **Neurological Soft Signs:** Check for poor coordination, toe-walking, or unusual gait.

Systemic Examination — Primary System (Developmental & Behavioral)

This is not a traditional "percussion/auscultation" exam. It is a structured observation.

1. Social Interaction Assessment:

- **Technique:** Try to engage the child with a toy. Offer a "social smile."
- **What to look for:** Does the child ignore you? If you withhold a toy, does he make eye contact to request it, or does he just scream?

2. Language and Communication:

- **Technique:** Observe spontaneous vocalizations.
- **Findings:** Look for **Echolalia** (repeating your questions back), **Pronoun Reversal** (referring to himself as "He" or "You"), and lack of "Proto-declarative pointing" (pointing to show interest).

3. Play Assessment:

- **Technique:** Give the child a doll and a spoon, or a toy car.
- **Functional Play:** Does he push the car?
- **Symbolic/Pretend Play:** Does he "feed" the doll? (Usually absent or delayed in ASD).
- **Atypical Play:** Does he just spin the wheels of the car or line the blocks up in a perfect row?

4. Sensory Profile:

- **Technique:** Observe reaction to your stethoscope (cold) or the lights in the room.
- **Significance:** Hyper- or hypo-reactivity to sensory input is now a core diagnostic criterion.

Systemic Examination — Secondary Systems

- **Neurology:** Focus on hearing (distinguish from "tuning out") and vision. Check for focal deficits (uncommon in primary ASD).

- **Abdomen:** Check for hepatosplenomegaly (metabolic storage disorders can mimic ASD).

SIGNS THAT CLINCH THE DIAGNOSIS [EXAMINER FAVORITES]

- **Lack of Joint Attention:** The failure to coordinate attention between a person and an object. This is the "hallmark" of ASD.
- **Absence of Proto-declarative Pointing:** Pointing to *share interest* (e.g., "Look at that dog!") is usually absent, while proto-imperative pointing (pointing to *get* something) may be preserved.
- **Hand-Flapping/Stereotypies:** Rhythmic, repetitive movements triggered by excitement or boredom.
- **The "Tool Use" Sign:** Taking the examiner's hand and placing it on a jar to open it, without making eye contact with the examiner.

DIAGNOSIS

Diagnostic Criteria

- **DSM-5 Criteria:** Must meet all 3 criteria in Category A (Social Communication) and at least 2 of 4 in Category B (Restricted/Repetitive Behaviors). Symptoms must be present in early developmental period.

Differentials

1. **Social Communication Disorder:** Deficits in social use of language but *without* the restricted/repetitive behaviors.
2. **Global Developmental Delay (GDD):** Social skills are delayed but commensurate with the overall mental age (in ASD, social skills are lower than the mental age).
3. **Hearing Impairment:** Always rule this out with BERA/Audiometry.
4. **Landau-Kleffner Syndrome:** Acquired epileptic aphasia (regression in language with abnormal EEG).

Investigations

- **Tier 1:** Audiometry/BERA (Mandatory). M-CHAT-R/F (Screening tool).
- **Tier 2: ISAA (Indian Scale for Assessment of Autism)** or **CARS-2** (Childhood Autism Rating Scale) to confirm and grade severity.
- **Tier 3:** Genetic testing (Chromosomal Microarray, Fragile X testing), EEG (if seizures or regression suspected), MRI Brain (only if focal neuro signs or microcephaly).

Management Outline

- **Early Intervention:** The gold standard.
- **Applied Behavior Analysis (ABA):** Intensive behavioral therapy.

- **Occupational Therapy:** For sensory integration and fine motor skills.
- **Speech Therapy:** Focus on functional communication (PECS - Picture Exchange Communication System).
- **Pharmacotherapy:** Risperidone or Aripiprazole (only for irritability/aggression, NOT for core social symptoms). [UPDATED: FDA approved for ages 5+ and 6+ respectively].

EXAMINER'S VIVA

Q1: How do you differentiate a "late talker" from a child with ASD? *A: A late talker usually compensates with gestures (pointing, nodding, miming). A child with ASD has a "global" communication deficit, including lack of gestures and poor eye contact.*

Q2: What is "Theory of Mind" in ASD? *A: It is the ability to understand that others have beliefs, desires, and intentions different from one's own. Children with ASD struggle with this (Social Cognition deficit).*

Q3: Why is it important to check for a "pincer grasp" in a child with ASD? *A: To assess if the developmental delay is global or specific. If they have a perfect pincer grasp but no words, it suggests a "dissociation" favoring ASD over Global Developmental Delay.*

Q4: What is the M-CHAT and at what age is it used? *A: Modified Checklist for Autism in Toddlers. Validated for children 16 to 30 months of age.*

Q5: [Technique Question] Show me how you would test for "Joint Attention" right now. *A: (Action) Get the child's attention, then suddenly look and point toward a distant object (like a clock) and say, "Oh look!" Observe if the child follows your gaze and then looks back at you to share the experience.*

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting [Name], a [Age] old male, who presented with a primary concern of language delay and social withdrawal, with a developmental profile suggestive of Autism Spectrum Disorder."
- **Mistake to Avoid:** Don't say "The child is uncooperative" in your exam. In an ASD case, the "uncooperativeness" *is* the clinical finding. Describe it as "The child was difficult to engage in reciprocal play."
- **What Examiners Watch For:** They watch if you try to force physical contact (like palpating the abdomen) too early. With ASD children, you must build rapport and perform the "hands-off" developmental exam first.

Infectious Diseases

42. HIV in children

Subject: Infectious Diseases

HISTORY

Chief Complaint

- **Infants/Young Children:** Recurrent "colds" or pneumonia, persistent diarrhea, failure to gain weight, or persistent oral thrush.
- **Older Children/Adolescents:** Chronic cough, skin rashes, unexplained fever, or lymph node swelling.
- **Duration:** Usually chronic (>1 month) or recurrent in nature.

History of Present Illness

- **Growth Failure:** "Has the child outgrown their clothes or shoes in the last 6 months?" "How does his weight compare to his siblings at this age?" (Points to HIV-associated wasting).
- **Recurrent Infections:** "How many episodes of pneumonia has he had?" "Were any life-threatening or required ICU stay?" (PCP/PJP often presents early and severe).
- **Chronic Diarrhea:** "Has the stool been loose for more than 14 days?" "Is there blood or mucus?" (Cryptosporidium or Microsporidia).
- **Central Nervous System:** "Has he lost any milestones he previously achieved?" "Is he struggling more in school lately?" (HIV Encephalopathy).
- **Skin:** "Does he have itchy bumps that don't go away?" (Pruritic Papular Eruption - PPE).
- **Fever/Cough:** "Is there a night sweat or contact with an adult who has a chronic cough?" (TB co-infection is the most common opportunistic infection).

Relevant Background History

- **Antenatal/Birth (CRITICAL):** "Did the mother receive any 'red and white' syrups or tablets during pregnancy?" "Was the delivery vaginal or C-section?" "Did the baby receive any prophylaxis (Nevirapine/Zidovudine) after birth?"
- **Feeding History:** "Was the child exclusively breastfed, formula-fed, or mixed-fed?" (Mixed feeding in the first 6 months carries the highest transmission risk).
- **Developmental:** Look for "Static Encephalopathy" (failure to attain milestones) vs. "Progressive Encephalopathy" (loss of milestones).
- **Immunization:** Specifically ask about BCG (look for the scar or BCG adenitis) and Measles (HIV children are high risk for severe measles).
- **Family History:** "Are the parents alive and healthy?" "Have any siblings died in infancy?" (The "Death of a sibling" is a major red flag for vertical transmission).

- **Socioeconomic:** "Who is the primary caregiver?" (Essential for assessing future ART adherence).

EXAMINATION

General Survey

- **The "HIV Look":** Observe for the "Old Man Facies" (loss of temporal and buccal fat), generalized lymphadenopathy, and a distended abdomen (organomegaly).
- **Activity:** Is the child playful or irritable/apathetic? (Apathy is common in HIV encephalopathy).
- **Skin:** Look for **Pruritic Papular Eruption (PPE)**—small, intensely itchy papules on extremities. Look for **Molluscum Contagiosum** (umbilicated papules) especially if giant or facial. Check for **Seborrheic Dermatitis** in the scalp and nasolabial folds.

Vital Signs and Anthropometry

- **Temperature:** Check for low-grade persistent fever.
- **Respiratory Rate:** Count for a full minute. Persistent tachypnea without distress may suggest **LIP (Lymphoid Interstitial Pneumonitis)**.
- **Growth [SEVERITY MARKER]:** You **must** plot Weight-for-Age, Height-for-Age, and Weight-for-Height. Wasting (acute) and Stunting (chronic) are hallmarks of advanced pediatric HIV.

Peripheral Signs

- **Oral Cavity [EXAMINER FAVORITE]:** Use a tongue depressor. Look for **Oral Candidiasis** (curdy white plaques that bleed when scraped). Check the side of the tongue for **Oral Hairy Leukoplakia** (EBV-related). Look for **Gingival Erythema** or **Noma** (cancrum oris) in severe cases.
- **Lymph Nodes:** Examine all stations (Cervical, Axillary, Epitrochlear, Inguinal). **Persistent Generalized Lymphadenopathy (PGL)** is defined as nodes >1cm in two or more extra-inguinal sites for >3 months. Note consistency (rubbery in HIV, matted in TB).
- **Clubbing:** Look for Grade 1-4 clubbing. In an HIV-positive child, clubbing is strongly suggestive of **LIP** or **Bronchiectasis**.
- **Parotid Swelling:** Look for bilateral, painless parotid enlargement. This is a classic sign of the LIP spectrum in children.
- **BCG Scar:** Check the left deltoid. Absence of a scar in a vaccinated child or a "suppurative" BCG scar suggests poor T-cell response.

Systemic Examination — Primary Systems

- **Respiratory:**
    - *Inspection:* Look for "barrel chest" (chronic airway disease) or intercostal retractions.
    - *Auscultation:* Listen for "fine, velcro-like crepitations" at the bases (LIP) or localized bronchial breathing (TB/Pneumonia).

- **Abdomen:**
    - *Palpation:* Feel for **Hepatosplenomegaly**. In HIV, this is often due to chronic infections (CMV, EBV, TB) or HIV-associated lymphoproliferative disease.
- **Central Nervous System:**
    - Assess muscle tone. **HIV Encephalopathy** often presents with "Diplegic" features—increased tone in lower limbs, brisk DTRs, and an upward plantar response.
    - Check head circumference (Microcephaly due to brain growth failure).

SIGNS THAT CLINCH THE DIAGNOSIS [EXAMINER FAVORITES]

1. **Oral Candidiasis beyond the neonatal period:** Highly suggestive of immunosuppression.
2. **Bilateral Painless Parotid Swelling:** In a child with respiratory symptoms, this is almost pathognomonic for LIP.
3. **Pruritic Papular Eruption (PPE):** Symmetrical, hyperpigmented papules on the shins and forearms.
4. **Persistent Generalized Lymphadenopathy (PGL):** Rubbery, non-tender nodes in multiple chains.
5. **Loss of Developmental Milestones + Hyperreflexia:** Suggests HIV Encephalopathy.

SEVERITY ASSESSMENT

- **WHO Clinical Staging (1 to 4):**
    - *Stage 1:* Asymptomatic/PGL.
    - *Stage 2:* Minor skin infections, recurrent URIs, parotid swelling.
    - *Stage 3:* Moderate malnutrition, persistent thrush, LIP, TB (pulmonary).
    - *Stage 4 (AIDS):* Wasting syndrome, PJP, Extrapulmonary TB, HIV Encephalopathy, Kaposi Sarcoma.

DIAGNOSIS

Diagnostic Criteria

- **Age <18 months:** Virological testing required (**HIV DNA PCR**). Maternal antibodies (IgG) persist, so ELISA is not diagnostic.
- **Age >18 months: HIV Antibody testing (ELISA/Rapid)**. Three different kits/assays are required for a positive diagnosis.

Differentials

1. **Severe Acute Malnutrition (SAM):** Can cause secondary immunosuppression, but won't have PGL or LIP.
2. **Disseminated Tuberculosis:** Overlaps significantly; however, TB usually has a more acute/subacute downhill course.
3. **Primary Immunodeficiency (e.g., SCID):** Presents much earlier (first 6 months) and usually has absent lymphoid tissue (no tonsils/nodes).

Investigations

- **Tier 1:** CBC (look for lymphopenia, anemia, thrombocytopenia), Chest X-ray (look for reticulonodular patterns of LIP or focal infiltrates of TB).
- **Tier 2: CD4 Count/Percentage** (Age-specific: <25% is significant in young children). **Viral Load** (Baseline before ART).
- **Tier 3:** Sputum for GeneXpert (TB), CSF analysis (if encephalopathy), Liver Function Tests (before starting ART).

Management Outline

- **Stabilization:** Treat acute infections (Pneumonia/Diarrhea).
- **Prophylaxis: Cotrimoxazole Preventive Therapy (CPT)** for all HIV-exposed or infected children to prevent PJP.
- **ART [UPDATED]:** "Test and Treat" policy—start ART regardless of CD4 count or clinical stage.
    - *Preferred Regimen:* **Dolutegravir (DTG)-based regimens** are now first-line for children (e.g., ABC + 3TC + DTG) provided weight is >3kg.
- **Monitoring:** Viral load at 6 months, then annually. Monitor growth and development at every visit.

EXAMINER'S VIVA

1. **Q: Why don't we use ELISA in a 6-month-old?**
    - *A:* Because maternal IgG antibodies cross the placenta and can persist in the infant's blood for up to 18 months, leading to a false positive.
2. **Q: How do you differentiate LIP from TB on a Chest X-ray?**
    - *A:* LIP typically shows bilateral, diffuse, symmetrical reticulonodular shadows with hilar lymphadenopathy and stays stable for months. TB is usually asymmetrical, with focal consolidation or miliary shadows, and is progressive.
3. **Q: What is the significance of a "Flat" Weight Curve in HIV?**
    - *A:* It is often the first sign of treatment failure or an occult opportunistic infection like TB.
4. **Q: How do you perform a proper oral exam for thrush?**

    - *A:* (Technique) Use a tongue depressor to scrape the white patch. If it's milk curd, it wipes off easily leaving normal mucosa. If it's Candidiasis, it's adherent and leaves an erythematous, bleeding base.

5. **Q: What is the "Window Period"?**
    - *A:* The time between HIV infection and the production of detectable antibodies (usually 3-12 weeks).

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting [Name], a [Age] year old male, born to a mother with [Known/Unknown] HIV status, who presents with a chronic history of [Main symptoms], currently classified as WHO Clinical Stage [X]."
- **Mistake:** Forgetting to check the immunization status of the mother or the HIV status of siblings.
- **Observation:** Examiners watch how you handle the "Social History." Be sensitive. Do not use the word "AIDS" loudly in front of the family; use "Stage 4 disease" or "Advanced HIV."

---

43. Congenital infections

Subject: Infectious Diseases

This is a classic "stump the resident" case. Congenital infections (TORCH-S: Toxoplasmosis, Others [Syphilis, HIV, Parvovirus, Zika], Rubella, CMV, Herpes) often present with overlapping features like IUGR, jaundice, and hepatosplenomegaly. Your job is to find the "discriminators"—those specific physical findings that point to one specific virus.

HISTORY

Chief Complaint

- "Yellowish discoloration of skin since birth"
- "Small head size or failure to gain weight"
- "Abnormal skin rashes or blisters"
- "Failure to respond to sound or abnormal eye appearance"
- "Abdominal distension"

History of Present Illness

Focus on the timeline of the insult and the constellation of symptoms.

- **Antenatal Insult:** "Mother, did you have any fever with a rash during the first three months of pregnancy?" (Rubella/Parvovirus). "Did you have any painful genital sores or discharge?" (HSV/Syphilis). "Did you consume undercooked meat or handle cat litter?" (Toxo).
- **Growth:** "Was the baby born small for their month?" (Symmetric IUGR is common in TORCH).

- **Jaundice:** "When did the yellowness start?" (Onset in first 24 hours suggests hemolysis or hepatitis). "Is the urine staining the cloth yellow and are the stools pale?" (Suggests cholestasis/CMV/Biliary atresia).
- **Neurological:** "Has the baby had any fits or abnormal posturing?" (Intracranial calcifications/encephalitis). "Do you feel the baby is too quiet or doesn't startle to loud noises?" (SNHL).
- **Skin:** "Were there any 'water bubbles' (vesicles) or 'blueberry spots' on the skin at birth?"

Relevant Background History

- **Antenatal:** Ask about routine screening (VDRL/HBsAg/HIV). Ask about "soft markers" on mid-trimester USG (intracranial calcifications, echogenic bowel, hydrops).
- **Birth:** Mode of delivery (HSV risk is higher in vaginal delivery with active lesions).
- **Developmental:** This is crucial. If the child is older, look for global developmental delay, especially motor and sensory (hearing/vision).
- **Immunization:** Mother's Rubella vaccination status.

EXAMINATION

General Survey

- **Initial Observation:** Look for the "Small for Gestational Age" (SGA) phenotype. Is the baby microcephalic? Is there an "anxious" or "staring" look (suggests visual impairment)?
- **Nutritional Status:** Measure birth weight, current weight, and length. Plot on Fenton's or WHO charts. TORCH babies often have "Symmetric IUGR" (Weight, Length, and HC all <10th centile).
- **Skin:**
    - **Blueberry Muffin Rash:** Look for non-blanching, purplish-blue papulo-nodules (Extramedullary hematopoiesis - classic for Rubella/CMV).
    - **Vesicles:** Look for clusters on an erythematous base (HSV).
    - **Desquamation:** Look for peeling skin on palms and soles (Congenital Syphilis).
    - **Jaundice:** Assess depth (Kramer's scale) and look for a greenish hue (cholestasis).

Vital Signs and Anthropometry

- **Head Circumference (HC):** [CRITICAL] Measure the maximal occipito-frontal circumference. Use a non-stretchable tape. **Microcephaly** (Z-score < -2) is a hallmark of CMV and Zika. **Macrocephaly** (due to hydrocephalus) points toward Toxoplasmosis.
- **Temperature:** Check for hypothermia or fever (neonatal sepsis mimic).

Peripheral Signs

- **Eyes:**

    - **Cataract:** Use a torch; look for a white pupillary reflex (Leukocoria). [EXAMINER FAVORITE: Rubella cataracts are often pearly white and bilateral].
    - **Microphthalmia:** Compare the size of the globes.
    - **Chorioretinitis:** You must mention you would perform a dilated fundus exam (Salt and pepper retinopathy = Rubella; Focal necrotizing = Toxo).
- **Lymph Nodes:** Feel for generalized lymphadenopathy, especially epitrochlear nodes (highly suggestive of Syphilis).
- **Hands/Nails:** Look for "Paronychia" and "Snuffles" (persistent mucoid nasal discharge) in Syphilis.

Systemic Examination — Primary System (Abdomen & CNS)

Abdomen (Hepatobiliary):

- **Inspection:** Scaphoid or distended? Look for prominent veins.
- **Palpation:**
    - **Liver:** Use the "edge of hand" technique from the RIF upwards. Measure the liver span. In TORCH, the liver is usually firm and enlarged due to extramedullary hematopoiesis or hepatitis.
    - **Spleen:** Feel for the notch. Massive splenomegaly is more common in CMV and Syphilis.
- **Percussion:** Check for ascites (shifting dullness) if the abdomen is very distended.

Central Nervous System:

- **Tone:** Assess for axial hypotonia and peripheral hypertonia.
- **Reflexes:** Check neonatal reflexes (Moro, Sucking, Palmar grasp). They are often depressed or asymmetrical in CNS involvement.
- **Fontanelle:** Palpate the Anterior Fontanelle. Is it bulging (Hydrocephalus/Toxo) or depressed/small (Microcephaly/CMV)?

Systemic Examination — Secondary Systems

- **CVS:** Auscultate for a continuous "machinery" murmur at the left infraclavicular area (PDA) or a systolic murmur of Pulmonary Artery Stenosis—both classic for **Congenital Rubella Syndrome (CRS)**.
- **Skeletal:** Look for pseudoparalysis (Parrot's pseudoparalysis in Syphilis—the baby doesn't move a limb because of bone pain from osteochondritis).

SIGNS THAT CLINCH THE DIAGNOSIS [EXAMINER FAVORITES]

- **The "Gregg's Triad" (Rubella):** Cataract + SNHL (Sensorineural Hearing Loss) + PDA.

- **Chorioretinitis + Hydrocephalus + Intracranial Calcifications:** The classic triad for **Toxoplasmosis**.
- **Periventricular Calcifications:** Specific for **CMV** (Toxo calcifications are usually scattered throughout the parenchyma).
- **Hutchinson's Triad (Late Syphilis):** Interstitial keratitis + Hutchinson teeth (notched incisors) + 8th nerve deafness.
- **Snuffles and Palmar/Plantar Desquamation:** Pathognomonic for **Early Congenital Syphilis**.

SEVERITY ASSESSMENT [SEVERITY MARKER]

- **Neurological:** Refractory seizures, bulging fontanelle, or decerebrate posturing.
- **Hematological:** Petechiae/Purpura (suggests severe thrombocytopenia).
- **Hepatic:** Direct hyperbilirubinemia >2mg/dL or signs of liver failure (prolonged PT/INR).

DIAGNOSIS

Diagnostic Criteria

- **Congenital Rubella:** WHO criteria (Suspected/Probable/Confirmed based on clinical signs + IgM/PCR).
- **Congenital Syphilis:** CDC criteria (Confirmed if *T. pallidum* seen on darkfield; Probable if infant titer is 4x maternal titer).

Differentials

1. **Neonatal Sepsis:** Presents with lethargy and jaundice, but usually lacks the dysmorphism/microcephaly of TORCH.
2. **Biliary Atresia:** Presents with cholestatic jaundice and hepatomegaly, but usually no microcephaly or intracranial calcifications.
3. **Inborn Errors of Metabolism (IEM):** Galactosemia can present with cataracts and jaundice, but usually starts after feeding begins.

Investigations

- **Tier 1:** CBC (look for thrombocytopenia, anemia), LFT (conjugated jaundice), Skeletal X-rays (look for "celery stalking" in Rubella or periostitis in Syphilis).
- **Tier 2 (The "Gold Standards"):**
    - **CMV:** Urine or Saliva PCR within the first 3 weeks of life (after 3 weeks, it could be postnatal infection).
    - **Toxo/Rubella:** Serum IgM (infant).
    - **Syphilis:** RPR/VDRL on infant serum (not cord blood).

- **Tier 3:** Neuroimaging (USG Cranium or CT/MRI) to differentiate calcification patterns; BERA for hearing; Slit-lamp exam for eyes.

Management Outline

- **General:** Nutritional support (high calorie for IUGR), seizure control.
- **Specific:**
    - **CMV:** Oral Valganciclovir (6 months) to improve hearing/developmental outcomes.
    - **Toxo:** Pyrimethamine + Sulfadiazine + Folinic acid (1 year).
    - **Syphilis:** IV Penicillin G for 10 days.
    - **HSV:** IV Acyclovir (21 days for CNS/Disseminated).

EXAMINER'S VIVA

1. **Q: Why is urine PCR preferred over serum IgM for CMV?**
    - *A: CMV IgM has low sensitivity and can be false positive. PCR of urine/saliva is the gold standard; however, it must be done before 21 days of life to distinguish congenital from perinatally acquired infection.*
2. **Q: How do you differentiate Toxo and CMV calcifications on CT?**
    - *A: CMV calcifications are typically **periventricular** (subependymal), whereas Toxoplasmosis calcifications are **diffuse/scattered** throughout the cortex and basal ganglia.*
3. **Q: What is the significance of "Snuffles" in a neonate?**
    - *A: It is a highly contagious, syphilitic rhinitis. The discharge is loaded with spirochetes. It often precedes the skin rash.*
4. **Q: How do you perform a Schamroth's window test in an infant?**
    - *A: (Technique) Place the dorsal surfaces of the distal phalanges of corresponding fingers together. Look for the diamond-shaped window. Loss of this window indicates clubbing (rare in TORCH, but seen in associated chronic cyanotic heart disease).*
5. **Q: Can you give the Rubella vaccine to a pregnant woman?**
    - *A: No, it is a live attenuated vaccine and is contraindicated. If given inadvertently, it is generally not an indication for termination, but the mother must be monitored.*

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting a [Age]-old [Gender] neonate, born SGA at [Gestational Age], presenting with a constellation of jaundice, microcephaly, and hepatosplenomegaly, suggesting a congenital intrauterine infection."

- **Mistake:** Don't just say "TORCH." Try to commit to one (e.g., "Most likely Congenital CMV") based on your discriminators (e.g., "due to the presence of microcephaly and periventricular calcifications").
- **Watch out:** Examiners will watch how you handle the baby. Always warm your hands and stethoscope. When checking the fontanelle, ensure the baby is calm and upright.

---

44. Tuberculosis

Subject: Infectious Diseases

HISTORY

Chief Complaint

- **Fever:** Usually low-grade, evening rise of temperature, lasting >2 weeks.
- **Cough:** Persistent, non-remitting cough for >2 weeks.
- **Weight Loss:** Or failure to gain weight/crossing centiles downwards over the last 3 months.
- **Lump/Swelling:** Usually in the neck (cervical lymphadenopathy).
- **Decreased Activity:** "Not playing as much," "always tired," or "refusing to walk" (in bone/joint TB).

History of Present Illness

- **Fever Characterization:** "Does the fever come every day? Is it higher in the evening? Does he sweat profusely at night when the fever breaks?" (Classic evening rise and night sweats).
- **Cough Nuances:** "Is the cough getting better with the antibiotics the local GP gave, or is it the same? Is there blood in the spit (hemoptysis - rare in children, suggests cavitary disease)?"
- **Weight/Growth:** "When was the last time he fit into these clothes? Have you noticed his ribs becoming more prominent?"
- **Contact History (Crucial):** "Is there anyone in the house, or a frequent visitor, who has a chronic cough, is taking 'red tablets,' or was treated for a lung condition recently?" *Note: Ask about the 'grandfather's smoker's cough'—it's often undiagnosed TB.*
- **Systemic Review for Dissemination:**
    - **CNS:** "Has his behavior changed? Is he unusually irritable? Any vomiting or headaches?" (TBM).
    - **Abdominal:** "Is his belly getting bigger? Any change in bowel habits or persistent pain?" (Abdominal TB).
    - **Skeletal:** "Is there a limp? Does he cry when you pick him up? Any deformity in the back?" (Pott's spine).

Relevant Background History

- **Birth/Antenatal:** Only if neonatal TB is suspected (maternal history of TB or infertility).

- **Immunization:** "Was the BCG vaccine given at birth? Can you show me the scar on the left deltoid?" (Presence of scar doesn't rule out TB but its absence is noted).
- **Nutrition:** Detailed 24-hour recall. TB is a disease of "poverty and protein-energy malnutrition." Malnutrition is both a risk factor and a consequence.
- **Socioeconomic:** "How many people sleep in one room? Is there a window for sunlight and air?" (Overcrowding and poor ventilation are key drivers).
- **Past History:** History of recent Measles or Pertussis (these cause transient immunosuppression and can reactivate latent TB).

EXAMINATION

General Survey

- **Appearance:** Does the child look "chronically ill"? Look for the "TB Facies"—sunken eyes, prominent cheekbones, and a bright-eyed but exhausted look.
- **Activity:** Is the child interactive or listless? [SEVERITY MARKER: Irritability in a child with fever is TBM until proven otherwise].
- **Nutritional Status:** Look for visible wasting of gluteal fat and "baggy pants" appearance. Check for hair changes (flag sign) suggesting associated kwashiorkor.

Vital Signs and Anthropometry

- **Temperature:** Document the evening rise if possible.
- **Respiratory Rate:** Count for a full minute. Tachypnea without significant distress or wheeze in a stable child suggests interstitial involvement or effusion.
- **Weight-for-Age / BMI:** Plot on WHO charts. A "flat" growth curve over 3 months is a diagnostic criterion in the NTP.

Peripheral Signs

- **BCG Scar:** Check left deltoid. If absent, it indicates the child missed the vaccine or it "didn't take."
- **Lymph Nodes:** [EXAMINER FAVORITE]
    - Examine cervical, axillary, and inguinal groups.
    - **Technique:** Stand behind the child for cervical nodes. Use the pads of your fingers.
    - **What to look for:** Look for "matted" nodes (nodes that feel stuck together in a clump). Note consistency (firm vs. fluctuant). Look for a **scrofuloderma** (skin breakdown over a node) or a healed puckered scar.
- **Clubbing:** Look for early clubbing (loss of Schamroth’s window) which can occur in chronic cavitary TB or bronchiectasis secondary to TB.

- **Phlyctenular Conjunctivitis:** Small grayish-pink nodules at the limbus. This is a hypersensitivity reaction to TB antigen. [EXAMINER FAVORITE].
- **Skin:** Look for **Erythema Nodosum** (tender, reddish nodules on shins)—another hypersensitivity sign.

Systemic Examination — Respiratory (Primary System)

- **Inspection:** Look for asymmetrical chest expansion. Tracheal shift (though difficult in infants).
- **Palpation:** Confirm the expansion. Check for increased or decreased Vocal Fremitus.
- **Percussion:**
    - **Dullness:** Stony dullness suggests pleural effusion.
    - **Impaired Note:** Over the apex (Supraclavicular/Infraclavicular) suggests infiltration or collapse-consolidation.
- **Auscultation:**
    - **Breath Sounds:** Look for "Amphoric" breathing over a cavity (rare in kids). More commonly, you will find diminished breath sounds over an effusion or bronchial breathing over a consolidated segment.
    - **Adventitious sounds:** Persistent localized crepitations (crackles) that do not clear with coughing.

Systemic Examination — Secondary Systems

- **Abdomen:** Palpate for hepatosplenomegaly (disseminated/miliary TB). Feel for "doughy abdomen" (peritoneal TB) or palpable matted mesenteric lymph nodes (tabes mesenterica).
- **CNS:** Check for neck stiffness (Kernig's/Brudzinski's) and cranial nerve palsies (especially VI nerve).
- **Spine:** Run your fingers down the spinous processes looking for a "gibbus" (sharp angulation) or localized tenderness.

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **Matted Lymphadenopathy:** Highly suggestive of TB over other causes of lymphadenitis.
- **Phlyctenular Conjunctivitis/Erythema Nodosum:** These are "clues" that the body is reacting to TB proteins.
- **Stony Dull Percussion Note:** Points to pleural effusion, which in an endemic area is TB until proven otherwise.
- **Gibbus Deformity:** Pathognomonic for Pott's Spine.
- **BCG Scar + Positive TST:** In a symptomatic child, this triad is very strong evidence.

Severity Assessment

- **Mild/Moderate:** Isolated lymph node TB, localized pulmonary TB in an older child.

- **Severe [SEVERITY MARKER]:**
    - Miliary TB (look for "millet seed" spots on fundoscopy).
    - TB Meningitis (altered sensorium, seizures).
    - Disseminated TB (involvement of >2 organ systems).
    - Severe respiratory distress (suggesting massive effusion or pneumothorax).

DIAGNOSIS

Diagnostic Criteria

- **Index of Suspicion (NTEP/IAP):** Persistent fever/cough >2 weeks, weight loss, and history of contact.
- **Microbiological:** Sputum/Gastric Aspirate/Induced Sputum for CBNAAT (GeneXpert).

Differentials

1. **Refractory Pneumonia:** Doesn't respond to standard antibiotics; usually more acute onset.
2. **Lymphoma:** Nodes are usually rubbery, non-tender, and discrete (not matted); often associated with massive splenomegaly.
3. **HIV-associated Lung Disease (LIP):** Similar presentation; requires HIV testing.
4. **Foreign Body Aspiration:** Sudden onset, localized wheeze, no constitutional symptoms.

Investigations

- **Tier 1:**
    - **Chest X-ray:** Look for hilar lymphadenopathy, paratracheal widening, or miliary mottling.
    - **Mantoux Test (TST):** >10mm is positive (in endemic areas). Read at 48-72 hours.
- **Tier 2:**
    - **CBNAAT (GeneXpert):** [UPDATED] This is now the first-line test. Use Gastric Aspirate in children who cannot expectorate. It also detects Rifampicin resistance.
- **Tier 3:**
    - **Contrast-Enhanced CT (CECT) Chest:** If X-ray is inconclusive but suspicion is high (shows "ring enhancement" in nodes).
    - **Fine Needle Aspiration Cytology (FNAC):** From lymph nodes to look for "caseating granulomas."

Management Outline

- **Notification:** TB is a notifiable disease.
- **Nutritional Support:** High protein, high calorie diet.

- **Medical (NTEP Guidelines):**
    - **Intensive Phase (2 months):** HRZE (Isoniazid, Rifampicin, Pyrazinamide, Ethambutol).
    - **Continuation Phase (4 months):** HRE.
    - *Note: Use child-friendly Dispersible Tablets (FDCs) based on weight bands.*
- **Steroids:** Indicated in TBM and Pericardial TB to reduce inflammation.

EXAMINER'S VIVA

Q1: Why do we do Gastric Aspirate in children? *A: Children are "pauci-bacillary" and usually swallow their sputum. Gastric aspirate collected early morning (after 6 hours of fasting) traps the swallowed sputum in the stomach.*

Q2: What is the "Ghon Complex"? *A: It consists of three things: The primary parenchymal focus (Ghon focus), lymphangitis, and the regional (hilar) lymphadenitis.*

Q3: How do you perform a Mantoux test correctly? *A: Inject 0.1 ml of 5TU PPD intradermally on the volar aspect of the forearm using a 26G needle to create a 6-10mm wheal. Read the **induration** (not erythema) after 48-72 hours.*

Q4: If a child has a positive Mantoux but no symptoms and a normal X-ray, what is the diagnosis? *A: Latent TB Infection (LTBI). [UPDATED] Current guidelines recommend TPT (TB Preventive Treatment) for these children, especially if they are household contacts.*

Q5: What is the significance of a "negative" Mantoux in a very sick child? *A: It's called "Anergy." It occurs in severe malnutrition, miliary TB, or disseminated disease because the immune system is too weak to mount a response.*

LONG CASE PRESENTATION TIPS

- **The Opening:** "I am presenting [Name], a [Age] year old male, resident of [Location], belonging to [Socioeconomic Class], who presented with a 3-week history of low-grade evening rise fever, persistent cough, and significant weight loss, with a positive history of contact with his paternal grandfather who is on anti-TB treatment."
- **The "Contact" Trap:** Never just say "No history of contact." Say "I specifically searched for a history of contact in the household and neighborhood, and found..."
- **The Examination Flow:** Examiners watch how you handle the child. If you go straight for the throat with a tongue depressor, you lose marks. Start with the BCG scar and lymph nodes.
- **The "Matted" Node:** Be prepared to demonstrate how you feel the nodes are fixed to each other rather than being discrete.

Neonatology

45. Preterm with complications

Subject: Neonatology

This is a high-stakes case. In a Neonatal Long Case, the examiner isn't just looking for a diagnosis; they are watching your **handling** of the neonate. A preterm baby is fragile. Your movements must be purposeful, warm, and aseptic.

HISTORY

Chief Complaint

- "Baby born at [Gestational Age] via [Mode of Delivery] presented with [Respiratory Distress/Poor Feeding/Jaundice/Seizures] since [Age of Onset]."

History of Present Illness

- **Gestational Age Assessment:** "Was the mother sure of her dates? When was the first trimester ultrasound done?" (Crucial for dating).
- **The "Why" of Prematurity:** "Did the mother have leaking P/V, fever, or foul-smelling discharge?" (PPROM/Chorioamnionitis). "Was there high BP or protein in the urine?" (Pre-eclampsia).
- **Antenatal Steroids:** "Did the mother receive injections to mature the baby's lungs? How many doses and how long before delivery?" [CRITICAL: 2 doses of Betamethasone 24h apart, ideally >24h but <7 days before birth].
- **Resuscitation History:** "Did the baby cry immediately? Was any 'tube' put in the windpipe or 'bag and mask' used?" (Assessing Birth Asphyxia risk).
- **Respiratory Distress:** "Did the grunting start immediately (RDS) or after a few hours (Sepsis/Pneumonia)?"
- **Feeding & Gut:** "Has the baby tolerated feeds? Any abdominal distension or blood in stools?" (NEC screening).
- **Neurological:** "Any abnormal jerky movements or periods of duskiness/apnea?"

Relevant Background History

- **Antenatal:** Maternal blood group (Rh isoimmunization), TILT (Toxoplasmosis, etc.), and GDM (Preterm/Large for dates).
- **Birth Weight:** "What was the exact birth weight?" (To plot on Fenton's/Intergrowth-21st charts).
- **Socioeconomic:** "Can the family afford surfactant or long-term NICU care?" (Practical ethics).

EXAMINATION

General Survey

- **The "First Look":** Observe the baby in the incubator/warmer *before* touching. Note the posture (Preterms have "froglike" posture due to hypotonia; term babies are flexed).
- **Activity:** Is the baby "vibrant" or "lethargic"? Note spontaneous movements.
- **Color:** Look for central cyanosis (tongue), pallor (anemia/shock), or plethora (polycythemia).
- **Skin:** Look for vernix (scant in very preterm), peeling (post-term), or visible veins (preterm skin is translucent).

Vital Signs and Anthropometry

- **Temperature:** Use a digital thermometer in the axilla for 3 mins. [SEVERITY MARKER]: Cold stress (36–36.4°C) vs. Neonatal Cold Injury (<32°C).
- **Heart Rate:** Auscultate the apex for a full minute. (Normal: 120–160 bpm).
- **Respiratory Rate:** Count for 60 seconds (periodic breathing is common in preterms; look for true apnea >20 sec).
- **CRT:** Press over the sternum for 5 seconds. Normal is <3 seconds.
- **Anthropometry:**
    - **Weight:** Use a digital scale (subtract weight of tubes/diapers).
    - **Length:** Use an infantometer (requires two people).
    - **OFC:** Use a non-stretchable tape; measure the maximum circumference (occiput to supraorbital ridges).
    - **Interpretation:** Plot on **Fenton's Charts**. Is the baby SGA, AGA, or LGA?

Peripheral Signs

- **Sole Creases:** [EXAMINER FAVORITE] Examine the anterior 1/3, 2/3, or entire sole. Preterms <32 weeks have almost no creases.
- **Breast Bud:** Measure the diameter. (Absent in <34 weeks).
- **Ear Cartilage:** Fold the pinna. Does it recoil? (Preterms have slow/no recoil).
- **Genitalia:** Males (undescended testes, smooth scrotum); Females (Labia majora do not cover minora).
- **Perfusion:** Check peripheral pulses (brachial and femoral). Weak femorals? Think Coarctation.

Systemic Examination — Primary System (Respiratory/Neonatal)

- **Inspection:**
    - **Silverman-Anderson Score:** [CRITICAL] Assess 5 parameters: Upper chest retraction, Lower chest retraction, Xiphoid retraction, Nares dilation, and Expiratory grunt. Score >7 is severe distress.
- **Palpation:** Check for apex beat (shifted in pneumothorax or diaphragmatic hernia).

- **Auscultation:** Use a neonatal diaphragm. Listen for air entry (often reduced in RDS) and "fine creps" (pneumonia).
- **The Heart:** Listen for a continuous "machinery" murmur at the left infraclavicular area (PDA—very common in preterms).

Systemic Examination — Secondary Systems

- **Abdomen:** Palpate for liver (normal 1–2cm). Look for visible bowel loops or discoloration (NEC).
- **CNS:**
    - **Tone:** Scarf sign (in preterm, the elbow crosses the midline easily).
    - **Reflexes:** Sucking and Rooting (mature by 34 weeks). Moro reflex (exhaustible and incomplete in preterms).
- **Eyes:** Look for "white reflex" (ROP/Cataract).

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **New Ballard Score:** You must be able to demonstrate at least 2 physical (e.g., Skin, Plantar surface) and 2 neuromuscular (e.g., Square window, Popliteal angle) criteria to justify your gestational age assessment.
- **The Grunt:** Explain that it is expiration against a partially closed glottis to maintain Functional Residual Capacity (FRC).
- **Apnea vs. Periodic Breathing:** Periodic breathing has no bradycardia/cyanosis; Apnea >20 seconds does.
- **Patent Ductus Arteriosus (PDA) Signs:** Hyperdynamic precordium, bounding pulses, and a wide pulse pressure.

DIAGNOSIS

Diagnostic Criteria

- **Preterm:** Born before 37 completed weeks.
- **LBW:** <2500g; **VLBW:** <1500g; **ELBW:** <1000g.
- **RDS (Hyaline Membrane Disease):** Clinical distress + X-ray (Ground glass opacities + Air bronchograms).

Differentials

1. **Transient Tachypnea of Newborn (TTNB):** Usually near-term, C-section, improves within 24–48 hours.
2. **Neonatal Sepsis/Pneumonia:** History of maternal fever, foul liquor, or prolonged ROM.
3. **Meconium Aspiration Syndrome:** History of meconium-stained liquor (rare in very preterm).

Investigations

- **Tier 1:** Sepsis screen (CRP, ANC, I:T ratio), Blood sugar (Dextrostix), Chest X-ray.
- **Tier 2:** Arterial Blood Gas (ABG) for Type 1 vs Type 2 respiratory failure; Cranial Ultrasound (to rule out IVH).
- **Tier 3:** Echocardiography (for PDA/PPHN); Screening for ROP (at 4 weeks or 31 weeks PMA).

Management Outline

1. **Warmth:** Neutral Thermal Environment (Incubator/Warmer).
2. **Breathing:** CPAP (First line for RDS) or Surfactant (InsurE technique).
3. **Fluids/Nutrition:** TPN if <1500g; Early Trophic feeding with EBM (Expressed Breast Milk).
4. **Infection:** Aseptic precautions; antibiotics only if sepsis screen positive.
5. **Developmental Supportive Care:** Nesting, dimmed lights, minimal handling.

EXAMINER'S VIVA

- **Q: How do you differentiate a Growth Restricted Term baby from a Preterm baby?**
    - A: Use the New Ballard Score. A growth-restricted term baby will have mature physical features (creases, ear cartilage) despite low weight.
- **Q: What is the "Golden Hour" in neonatology?**
    - A: The first 60 minutes of life where stabilization, thermoregulation, and early respiratory support (CPAP) determine long-term outcomes.
- **Q: Why do preterms get IVH (Intraventricular Hemorrhage)?**
    - A: Due to the fragile "Germinal Matrix" and poor autoregulation of cerebral blood flow.
- **Q: Technique: How do you perform the "Square Window" test?**
    - A: Flex the wrist toward the forearm. Apply gentle pressure to get maximum flexion. In preterms, the angle is >90°; in term babies, it is 0°.

LONG CASE PRESENTATION TIPS

- **The Opening:** "I am presenting the case of a [GA] male/female neonate, AGA/SGA, born to a [Gravida] mother, currently [Post-natal age] days old, with a birth weight of [X] grams..."
- **Mistake to Avoid:** Never forget to mention **Vitamin K** administration and **Eye care** in the birth history.
- **The "Hand-Wash" Rule:** If you touch the baby without sanitizing your hands in front of the examiner, you may fail the station immediately. Always sanitize before and after touching the baby.

---

46. Hypoxic ischemic encephalopathy sequelae

Subject: Neonatology

This is a classic "Chronic Stable" case in Neonatology/Neurology. You aren't just examining a child with Cerebral Palsy; you are reconstructing a perinatal catastrophe and mapping its permanent neurological footprint.

HISTORY

Chief Complaint

- "Delay in reaching milestones" (usually the primary concern).
- "Stiffness of limbs" or "abnormal posturing."
- "Recurrent seizures" or "staring spells."
- "Difficulty in feeding or swallowing."

History of Present Illness

Focus on the evolution of the neurological deficit.

- **The "Static" Nature:** Ask, "Has the child lost any skills they previously gained, or are they just slow to acquire them?" (HIE sequelae are static; regression suggests neurodegenerative disorders).
- **Motor Delay:** "When did the child start holding their head? Is there a preference for one hand? Do the legs cross like scissors when you pick them up?"
- **Seizures:** "Describe the episodes. Are they sudden jerks (myoclonic), stiffening (tonic), or blank stares? How frequent are they despite medications?"
- **Feeding/Bulbar issues:** "Does the child cough or choke while feeding? Does milk come out of the nose? How long does a single feed take?" (Indicates pseudobulbar palsy).
- **Vision/Hearing:** "Does the child respond to your face? Do they turn to loud sounds?" (Cortical Visual Impairment is common in HIE).

Relevant Background History

- **Antenatal:** Ask about maternal PIH, GDM, or decreased fetal movements (fetal distress).
- **Birth History (The Core):**
    - "Was the liquor meconium-stained?"
    - "Did the baby cry immediately? If not, for how long was the baby blue or limp?"
    - "Was bag-and-mask ventilation or chest compressions required?" (Determines Sarnat staging retrospectively).
- **Neonatal Period:**
    - "Did the baby have seizures in the first 24–48 hours?"

    - "How many days was the baby on a ventilator?"
    - "When did the baby start direct breastfeeding?" (Delayed sucking is a strong predictor of poor outcome).
- **Developmental:** Detailed domain-wise delay. In HIE, motor is usually more affected than social.
- **Nutritional:** Caloric intake (often deficient due to feeding difficulties).

EXAMINATION

General Survey

- **Observation:** Observe the child on the mother's lap. Look for **scissoring of lower limbs**, **fisting of hands**, or **obligate cortical thumb**.
- **Activity:** Spontaneous movements? Are they purposeful or extrapyramidal (athetosis/chorea)?
- **Nutritional Status:** Assess for wasting. Use mid-upper arm circumference (MUAC) if the child has contractures making height/weight difficult.
- **Microcephaly:** [SEVERITY MARKER] Measure Head Circumference. Secondary microcephaly (due to lack of brain growth) is a hallmark of severe HIE sequelae.

Vital Signs and Anthropometry

- **Vitals:** Standard. Check for tachypnea (aspiration risk).
- **Growth:** Plot weight, height, and HC on WHO charts. Note the "falling off" of the HC curve.

Peripheral Signs

- **Hands:** Look for **Simian crease** (though more common in Trisomy 21, it can be seen in malformations mimicking HIE). Check for **contractures** at the wrists and small joints.
- **Eyes:** Check for **strabismus** (very common) and the **threat reflex** (to assess Cortical Visual Impairment).
- **Spine:** Look for scoliosis (common in non-ambulatory spastic quadriplegia).

Systemic Examination — Primary System (Central Nervous System)

1. Tone (The most critical part)

- **Technique:** Move joints (elbow, knee, ankle) at varying speeds.
- **Findings:** Look for **Spasticity** (clasp-knife, velocity-dependent).
- **Adductor Angle:** With the child supine, extend legs and abduct hips. An angle <90° suggests high adductor tone.
- **Popliteal Angle:** Flex hip to 90°, then extend the knee. Resistance/angle >120° suggests hamstring spasticity.

2. Power and Reflexes

- **Power:** Often difficult to grade 0–5 in a non-cooperative child; describe as "functional power" (e.g., "can reach for objects," "can push against resistance").
- **DTRs:** Expect **brisk reflexes** (3+ or 4+) with **clonus**.
- **Technique for Ankle Clonus:** Rapidly dorsiflex the foot and maintain pressure. Count the beats. >5 beats is pathological.

3. Primitive Reflexes [EXAMINER FAVORITE]

- At this age (usually >1 year), these should be gone. Their persistence confirms an upper motor neuron lesion.
- **Asymmetric Tonic Neck Reflex (ATNR):** Turn head to one side; "fencing posture" should not persist beyond 6 months.
- **Moro Reflex:** Should disappear by 4–6 months.
- **Parachute Reflex:** [TECHNIQUE] Hold child horizontally and tip forward. Failure to extend arms to "break the fall" after 8–9 months is abnormal.

4. Cranial Nerves

- Focus on **Bulbar function**: Check gag reflex and look for drooling (pseudobulbar palsy).
- **Fundoscopy:** Look for **Optic Atrophy** (pale disc).

Systemic Examination — Secondary Systems

- **Respiratory:** Auscultate for coarse crepitations (chronic aspiration/GERD).
- **Musculoskeletal:** Check for hip dislocation (Ortolani/Barlow if young, or limited abduction if older).

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **The "Scissor Gait" or Posture:** Due to overactive adductors.
- **Obligate Cortical Thumb:** Thumb tucked inside the fist beyond 4 months of age.
- **Persistence of Primitive Reflexes:** Specifically a strong ATNR that prevents the child from rolling over.
- **Evolutionary Change:** History of a "floppy" neonate who became a "stiff" infant (classic HIE progression).

Severity Assessment

- **GMFCS (Gross Motor Function Classification System):** You MUST classify the child from Level I (walks without limitations) to Level V (transported in a manual wheelchair).
- **Presence of Refractory Epilepsy:** Indicates more extensive cortical damage.

DIAGNOSIS

Diagnostic Criteria

- **Clinical Diagnosis:** Based on a non-progressive motor deficit resulting from an insult to the developing brain (Cerebral Palsy, Spastic Quadriplegic type, GMFCS Level IV/V, likely secondary to HIE).

Differentials

1. **Leukodystrophies:** Distinguished by **regression** of milestones and early loss of vision/hearing.
2. **Symmetric Spinal Muscular Atrophy (SMA):** Distinguished by **absent** DTRs, tongue fasciculations, and no cognitive impairment.
3. **Metabolic Disorders (e.g., Glutaric Aciduria):** Often have a "trigger" (fever) followed by acute dystonia.

Investigations

- **Tier 1:** Vision and Hearing screening (BERA/OAE).
- **Tier 2: MRI Brain** (The Gold Standard). Look for:
    - *Term HIE:* Parasagittal watershed infarcts or Basal Ganglia/Thalamus (putamen) lesions.
    - *Preterm HIE:* Periventricular Leukomalacia (PVL).
- **Tier 3:** EEG (if seizures present), Hip X-ray (to rule out subluxation).

Management Outline

- **Multidisciplinary Approach:**
    - **Medical:** Baclofen/Diazepam for spasticity; Antiepileptics (Levetiracetam/Valproate).
    - **Physical:** Stretching, bracing (AFOs), and positioning.
    - **Nutritional:** High-calorie feeds, thickened liquids, or G-tube if aspiration risk is high.
    - **Surgical:** Selective Dorsal Rhizotomy or tendon lengthening (if contractures are fixed).

EXAMINER'S VIVA

1. **Q: Why is the head small in this child?**
    - *A:* It is "secondary microcephaly." The brain didn't grow due to the initial hypoxic insult; since the skull grows in response to brain expansion, the HC remains small.
2. **Q: What is the difference between "Stiffness" and "Spasticity"?**
    - *A:* Spasticity is a velocity-dependent increase in muscle tone with exaggerated DTRs (UMN sign). Stiffness is a lay term; as a clinician, I define this as spasticity or rigidity.
3. **Q: How do you differentiate a UMN lesion from a LMN lesion at the bedside?**
    - *A:* UMN (HIE) shows spasticity, brisk DTRs, and extensor plantar. LMN shows wasting, fasciculations, hypotonia, and absent DTRs.

4. **Q: [Technique] Show me how to elicit the ATNR.**
    - *A:* (Resident should turn the child's head to one side while supine). "The arm and leg on the side the face is turned to should extend, while the opposite side flexes."
5. **Q: What is the "Hand Preference" significance before 1 year of age?**
    - *A:* It is always pathological. It suggests hemiplegia on the opposite side.

LONG CASE PRESENTATION TIPS

- **The Opening:** "I am presenting [Name], a [Age] old male, born out of a [Consanguineous/Non-consanguineous] marriage, with a significant history of birth asphyxia, now presenting with global developmental delay and static neurological deficits, currently classified as GMFCS Level..."
- **Common Mistake:** Forgetting to check the hearing and vision. Examiners hate when a resident focuses only on the legs and ignores the "Social" and "Sensory" aspects.
- **Watch for:** The examiner will watch how you handle the child. Be gentle, keep the child warm, and always involve the mother in the examination.

---

47. Neonatal cholestasis

Subject: Neonatology

This is a high-stakes long case. In neonatal cholestasis, the examiner is looking for your ability to differentiate **Extrahepatic Biliary Atresia (EHBA)** from **Neonatal Hepatitis Syndrome (NHS)** because the former is a surgical emergency.

HISTORY

Chief Complaint

- Jaundice persisting beyond 14 days of life (term) or 21 days (preterm).
- High-colored urine (staining the diaper).
- Pale or clay-colored stools (acholic).

History of Present Illness

- **Onset of Jaundice:** "When did you first notice the yellowing? Was it on the first day (hemolysis) or did it appear after the first week?" (EHBA usually presents at 2–4 weeks; NHS often earlier).
- **Urine Color:** "Does the urine leave a dark yellow stain on the diaper that is hard to wash off?" (Conjugated bilirubin is water-soluble and excreted in urine).
- **Stool Color:** "What is the exact color of the stool? Is it like mustard, or like white clay/putty?" [Show a stool color card if available].
- **Progression:** "Is the jaundice getting deeper or staying the same?"

- **Pruritus:** "Is the baby irritable, scratching the skin, or having disturbed sleep?" (Suggests chronic cholestasis/Alagille syndrome).
- **Bleeding Manifestations:** "Have you noticed any bleeding from the cord, nose, or needle prick sites?" (Vitamin K deficiency due to malabsorption).
- **Feeding and Activity:** "Is the baby feeding well and gaining weight?" (EHBA babies often look deceptively healthy initially; NHS babies often look "sick").

Relevant Background History

- **Antenatal:** Maternal infections (TORCH titers), gestational diabetes, or any history of "itching" in the mother (PFIC/ICP).
- **Birth History:** Birth weight (SGA is common in NHS/TORCH).
- **Newborn Screening:** Was a screen done for Galactosemia or Hypothyroidism?
- **Family History:** Consanguinity? Previous sibling deaths due to liver disease? (Suggests metabolic causes like Galactosemia, Tyrosinemia, or PFIC).
- **Medications/Nutrition:** History of TPN (TPN-induced cholestasis)?

EXAMINATION

General Survey

- **Appearance:** Does the baby look "greenish-yellow" (verdin jaundice - chronic) or "bright yellow"?
- **Activity:** Is the baby vigorous (EHBA) or lethargic/toxic (NHS/Sepsis)?
- **Nutritional Status:** Check for wasting of temples and gluteal folds. Look for Vitamin D deficiency (rickety rosary) and Vitamin A deficiency (Bitot spots - rare in neonates but look for corneal clouding).
- **Facies:** [EXAMINER FAVORITE] Look for Alagille Syndrome: Broad forehead, deep-set eyes, pointed chin (triangular facies).

Vital Signs and Anthropometry

- **Temperature:** Fever suggests sepsis or TORCH.
- **Growth:** Plot Weight, Length, and HC. EHBA usually has preserved growth initially; NHS/Metabolic causes often show early FTT.

Peripheral Signs

- **Skin:** Look for scratch marks (pruritus). Look for **Xanthomas** (yellowish papules on extensor surfaces/skin creases) indicating chronic high cholesterol.
- **Eyes:** Check for **Icterus** in natural light. Look for **Chorioretinitis** (TORCH) or **Posterior Embryotoxon** (Alagille).

- **Umbilicus:** Check for a prominent venous pattern (caput medusae) or umbilical hernia (hypothyroidism/ascites).

Systemic Examination — Abdomen (Primary System)

- **Inspection:**
    - Distension? (Ascites or organomegaly).
    - Everted umbilicus?
    - Visible veins (flow direction: away from umbilicus).
- **Palpation:**
    - **Liver:** Use the "edge of the hand" or "fingertips" starting from the RIF. In neonates, the liver is normally 1-2 cm below the costal margin.
    - **Character:** In EHBA, the liver is **firm to hard** with a sharp edge. In NHS, it is usually soft/firm.
    - **Spleen:** Palpate from RIF towards the left costal margin. Splenomegaly at presentation strongly suggests EHBA (portal hypertension develops early).
- **Percussion:**
    - **Liver Span:** Measure in the mid-clavicular line. Normal is 4–5 cm in a neonate.
    - **Ascites:** Check for shifting dullness. (Note: Fluid thrill is only for massive ascites).
- **Auscultation:** Bruit over liver (Hemangioma).

Systemic Examination — Secondary Systems

- **CVS:** Listen for murmurs. Peripheral pulmonary stenosis is associated with Alagille syndrome.
- **CNS:** Check tone and reflexes. Microcephaly suggests TORCH.

Signs That Clinch the Diagnosis [EXAMINER FAVORITES]

- **Firm/Hard Hepatomegaly:** Points toward EHBA and biliary cirrhosis.
- **Early Splenomegaly:** Suggests portal hypertension, common in EHBA.
- **Acholic Stools (Documented):** If you see white stools in the ward, it is the most significant sign of biliary obstruction.
- **Triangular Facies/Murmur:** Clinches Alagille Syndrome.
- **Cataracts:** Suggests Galactosemia or Rubella.

Severity Assessment [SEVERITY MARKER]

- **Encephalopathy:** Irritability, reversal of sleep-wake cycle, or coma (Late stage).
- **Ascites:** Indicates decompensated liver disease.
- **Vitamin K Deficiency:** Oozing from sites or intracranial hemorrhage.

DIAGNOSIS

Diagnostic Criteria

- **Definition:** Conjugated bilirubin > 1.0 mg/dL (if TSB < 5) or > 20% of TSB (if TSB > 5).

Differentials

1. **Extrahepatic Biliary Atresia:** Firm liver, early splenomegaly, persistently acholic stools, baby looks well.
2. **Neonatal Hepatitis Syndrome (Idiopathic/TORCH):** SGA baby, soft liver, intermittent stool color, looks "sick."
3. **Choledochal Cyst:** May have a palpable mass in the right hypochondrium.
4. **Metabolic (Galactosemia):** Jaundice starts after milk feeds, associated with vomiting, hypoglycemia, and cataracts.

Investigations

- **Tier 1 (Confirm Cholestasis):** Total/Fractionated Bilirubin, SGOT/SGPT, GGT (High in EHBA, low in PFIC-1/2), PT/INR (Check for coagulopathy).
- **Tier 2 (Etiology):**
    - **USG Abdomen:** Look for "Triangular Cord Sign" (fibrous cone at porta hepatis) and presence/absence of gallbladder. [EXAMINER FAVORITE]
    - **HIDA Scan:** If no excretion into the bowel after 24 hours (with priming by Phenobarbitone), EHBA is likely.
- **Tier 3 (Gold Standard):**
    - **Liver Biopsy:** Shows bile duct proliferation and plugs in EHBA.
    - **Intraoperative Cholangiogram (IOCG):** The definitive test to confirm EHBA.

Management Outline

- **Medical:** Medium Chain Triglycerides (MCT) oil, Fat-soluble vitamin supplementation (A, D, E, K), Ursodeoxycholic acid (UDCA) for bile flow.
- **Surgical: Kasai Portoenterostomy** (Best results if done < 60 days of life). [UPDATED]
- **Definitive:** Liver Transplantation for Kasai failures or late presentations.

EXAMINER'S VIVA

1. **Q: How do you differentiate EHBA from NHS on a biopsy?**
    - A: EHBA shows bile duct proliferation, portal fibrosis, and bile plugs. NHS shows lobular disarray and giant cell transformation.

2. **Q: What is the significance of GGT in neonatal cholestasis?**
    - A: High GGT suggests biliary obstruction (EHBA). Low GGT in a cholestatic baby suggests PFIC Type 1 or 2 or Bile Acid Synthesis defects.
3. **Q: Why do we give Phenobarbitone before a HIDA scan?**
    - A: To "prime" the liver enzymes and enhance biliary excretion, reducing false positives for EHBA.
4. **Q: What is the "Triangular Cord Sign"?**
    - A: It is a cone-shaped fibrotic mass seen on USG cephalad to the bifurcation of the portal vein; it is highly specific for EHBA.
5. **Q: Technique: How do you palpate the liver in a neonate?**
    - A: Start low in the right iliac fossa using the radial border of the index finger or fingertips. Palpate during inspiration. In neonates, always use light palpation as the liver is superficial.

LONG CASE PRESENTATION TIPS

- **Opening:** "I am presenting the case of a [Age] old [Male/Female] infant, born of [Consanguineous/Non-consanguineous] marriage, presenting with conjugated jaundice, acholic stools, and firm hepatomegaly, currently being evaluated for Extrahepatic Biliary Atresia."
- **Mistake:** Do not forget to mention the stool color. If you haven't seen the stool yourself, state: "Mother reports the stool is like clay."
- **Watch for:** Examiners will watch how you palpate the liver. If you poke vertically with your fingers, you will miss the soft edge. Use the flat of your hand.
