HY Pediatrics Shelf Review

Some MS4
Introduction

-The Peds shelf is somewhat challenging. It is essentially a medicine shelf for little people. You need to know a ton of stuff related to all body systems like you would for a medicine shelf.

-One challenging part of the peds exam is the issue with many non-descript questions. The NBME question writers for Peds are quite stingy with providing enough clues that should help with making a dx.

-However, they almost always introduce 1 keyword that can nudge you towards one correct answer on the exam.

-The goal of this presentation is to use cases and questions (most of the set) to review most of the HY pertinent peds material/keywords with time spent during the session on “aftermath whiteboard teaching”. Knowing the information in this slide set thoroughly should fetch you a large number of correct answers on your shelf.
A 4.5 yo male is brought to a well child visit by his concerned parents. For the past 4 months, he has woken up agitated and in tears, usually before midnight. He goes to sleep around 9.30 PM. He often appears drowsy when these episodes occur and goes right back to sleep after his mom caresses him for a few minutes. He has no memory of the event when he wakes up around 8AM the next morning. The patient is in the 60th percentile for height, weight, and head circumference. A physical exam conducted in the pediatrician’s office is normal. What is the next best step in the management of this patient?

a. 2 month prescription for diphenhydramine.
b. Recommendation of an exercise and weight loss program.
c. Polysomnography studies for obstructive sleep apnea.
d. Contact Child Protective Services.
e. Reassurance.
- The best answer here is E, **reassurance**.

- This child has **sleep terror disorder** (usually occurs in Stage N3 of sleep) which is characterized by **zero recollection** of the event in question.

- Contrast with **nightmare disorder** where the child has **vivid recollections** of his dreams. This occurs during **REM sleep (beta waves on EEG)**.

- If you see a presentation of a child physically walking from his bed to some other location in the process of sleep, **sleepwalking disorder** is your diagnosis. The next step on your exam may involve having to do something that would **physically protect** the child from harming himself during these episodes.

- Reassurance should be your go to answer for the most part as kids often outgrow these **parasomnias**.
An 18 mo male is brought to the ED with shortness of breath. A CXR on admission is notable for a well defined consolidation in the left upper lung lobe. This is the child’s 6th episode of pneumonia in the past 9 mo. He was recently placed on a course of high dose amoxicillin and clavulanate for a severe ear infection. His vital signs include a temperature of 102.9, HR 170 bpm, RR 30 bpm. There is no cervical lymphadenopathy or evidence of reactive hyperplasia in the back of his throat. His mom’s brother died last year from a severe Strep Pneumo infection. What is the most likely cause of this patient’s presentation?

a. Failed development of the 3rd and 4th pharyngeal pouches.
b. Failed differentiation of CD19 and 20+ cells.
c. Deficiency of a dimeric immunoglobulin.
d. Deficiency of an enzyme that deaminates adenosine.
e. X linked mutation in the WASP gene.
- The best answer here is B. This child has **Bruton’s agammaglobulinemia**.

- Consider this diagnosis if you get a history of a **boy** (it should 100% not be a girl on the NBME) with **recurrent sinopulmonary infections**.

- Since the disorder is **X linked**, you should have a family history of men having this disorder.

- Diagnosis is with a simple CBC. Consider giving regular **IVIG injections**.

- Most of these problems crop up at **> 6 mo age** since **maternal IgG** disappears from the infant’s serum around that time.

- Pay attention to the “**bug pattern**” on your exam. **Encapsulated bacterial** infections = **B cell problem**. **Viruses/fungi** = **T cell problem**. The combo of a **T cell deficiency (no thymus)**, **hypocalcemia (no parathyroids, seizures)**, **truncus arteriosus**, **low set ears** = **DiGeorge Syndrome**.

- The HY immunodeficiency syndromes are detailed on the next slide.
The Immunodeficiency Syndromes (memorize cold!)

- **Bruton’s**
  - X-linked (boys)
  - B cell problems (BTK mutation)
  - Respiratory/ GI bacterial infections
  - All Ig’s
  - Lymphoid hypoplasia tonsils
  - Give IVIG

- **IgA deficiency**
  - Much older
  - Respiratory and GI infections
  - Giardia
  - Transfusion anaphylaxis
  - Ig A 214

- **Wiskott Aldrich**
  - X-linked
  - Eczema
  - Low platelets
  - WASP mutation

- **Ataxia Telangiectasia**
  - Ataxia before telangiectasia
  - Risk of lymphoma

- **SCID**
  - B/T cell problems
  - IL2R mutation
  - ADA deficiency
  - Viruses, fungi, PCP
  - < 6mo

- **Leukocyte Adhesion Deficiency**
  - Delayed umbilical cord separation
  - Non-purulent abscesses
  - X Integrin (8)
- Kids with SCID often present with FTT, recurrent opportunistic infections (e.g. Candida), and a chronic diarrhea. You should not find a thymic shadow on a CXR (just like DiGeorge Syndrome). Avoid live attenuated vaccines with these patients.

- Patients with ataxia telangiectasia have IgA and T Cell deficiencies.

- Consider CGD as the dx (NADPH oxidase deficiency) in a kid with recurrent infections with catalase +ve bugs (especially Aspergillus/S. Aureus). Remember the Nitroblue Tetrazolium/Dihydrorhodamine tests which are used for dx. IFN-Gamma may help.

- In a kid with partial albinism, recurrent respiratory infections, and neuro problems (+ exam Q telling you of giant granules in the cytosol), think of Chediak Higashi dz as your dx (LYST gene mutation).
An 18 mo infant is brought to a pediatrician by his mom after she noticed a bulge in his abdomen when lifting him up from a crib. The presence of a left, infraumbilical mass is confirmed on a physical exam. Vital signs are notable for a BP of 138/97. All other vitals and a spot urinalysis are within normal limits. The infant has no relevant PMH and had a congenital hypospadias corrected 6 weeks after delivery. An abdominal CT scan is notable for a unilateral, non-calcified mass. The most likely pathophysiology of this child’s presentation is?

a. A tumor derived from cells of neural crest origin.
b. A tumor derived from embryonal renal cells.
c. Proliferation of catecholamine producing cells.
d. Metastases of proliferating cells found in the posterior mediastinum.
-The best answer here is B. This child most likely has a **Wilms Tumor**. Remember the association with the **WAGR complex** (*Wilms, Aniridia, GU anomalies, Mental Retardation*). The mutation is often ascribed to **Chromosome 11 on the WT1 gene** (or on some exams the **PAX6 gene**).

-There is a “**classic triad**” of HTN, hematuria, and a flank mass. This classic triad is super rare though (but common on exams).

-Dx is often with an **US first, followed by a CT scan** (mass is not calcified and is often unilateral).

-Do not confuse WT with a neuroblastoma which has a similar presentation (abdominal mass in a young kid). **Neuroblastomas** are not derived from the kidney parenchyma. They are derived from **neural crest cells** and often produce **catecholamines**. These masses are **CALCIFIED/often cross the midline** on a CT Scan.
Q3 Key

- It is also HY to know that neuroblastomas present as posterior mediastinal masses (+ may have an opsoclonus/myoclonus “sign”). Contrast with the anterior mediastinal presentation of teratomas (although most are sacrococcygeal) and thymomas.

- The breakdown products of catecholamines (VMA and HVA) may be elevated in the serum and urine of patients with a neuroblastoma.

- An unusual exam scenario may be to recognize a tumor that lights up with MIBG scanning. As an aside, remember the Horseshoe kidney association with Turner’s.

- You should also be able to recognize the presentation cluster for Beckwith-Wiedemann Syndrome (hemihypertrophy, hepatoblastoma, Wilms Tumor, macroglossia, and neonatal hypoglycemia from islet cell hypertrophy which may present as seizures). Neonatal hypoglycemia may also present as seizures in an IODM. Neonatal hypocalcemia in DiGeorge Syndrome may also present as seizures.
Q4

Given the following information cluster, what is the most likely diagnosis?

- Head circumference < 2nd percentile.
- Symmetric intrauterine growth restriction.
- Thin upper lip and smooth philtrum.
- Childhood behavioral problems.
Q4 Key

Given the following information cluster, what is the most likely diagnosis?

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This is fetal ROH syndrome. Other findings include clinodactyly and hypoplasia of the maxilla (contrast with mandibular hypoplasia in the Pierre Robin Sequence). The combo of epicanthal folds, flat midface, a single transverse palmar crease, hirschsprung’s disease, duodenal atresia, endocardial cushion defects, ALL = Down’s Syndrome. For trisomy 18, think of rocker bottom feet, a large occiput, clenched fists, overlapping toes, etc. For T13, think more of cleft lip/palate, and polydactyly.
Q4 Key contd.

- You should also note the association of Down’s syndrome with increased nuchal translucency on a prenatal US.

- On the quad screen, the B-HCG and Inhibin A will be elevated. AFP and Estriol will be low.

- Down’s syndrome commonly arises from maternal nondisjunction in the process of meiosis and occasionally from an unbalanced Robertsonian translocation.
Some Other HY Associations

- Remember the association of Fetal ROH Syndrome with VSDs, ADHD, and symmetric IUGR.

- In symmetric IUGR, the baby is small (including the head). This is IUGR that often arises from problems early in development like FAS or exposure to a TORCH infection.

- Asymmetric IUGR presents as a kid that is SGA with a normal sized head. This is less severe than symmetric IUGR, has a better prognosis, and often reflects an insult later in development (e.g. maternal malnutrition or any cause of uteroplacental insufficiency).
Q5

A 2 yo F is brought to the ED by her mom. PE is notable for bilateral, nonexudative conjunctival injection. A HEENT exam reveals cracked, fissured lips, tongue erythema, and left anterior cervical lymphadenopathy. There is no lymphadenopathy on the right. VS are notable for a temperature of 105 F. What is the next best step in the management of this patient?

a. Prednisolone and IVIG therapy.
b. Administration of high dose acetaminophen.
c. Supportive care.
d. Administration of IVIG and an anti-inflammatory dose of aspirin.
e. Prophylactic tPA administration to prevent myocardial infarction.
- The best answer here is D. This child (usually Asian) has **Kawasaki’s disease**. One HY symptom not mentioned in the Q stem is the presence of a **rash on the palms and soles and in some cases edema/desquamation of the hands/fingers**.

- If you get a question detailing RUQ pain in a patient with Kawasaki’s, your diagnosis is **gallbladder hydrops**.

- Remember the association of ASA with **Reye’s syndrome**

- The treatment of Kawasaki’s dz is **IVIG and high dose ASA**. Monitor for the development of coronary dz with an **echocardiogram**.

- Another HY vasculitide to consider for your test is **Henoch Schonlein purpura**. This should be your dx in a kid with a **recent URI that has abdominal pain, palpable purpura on the buttocks and lower extremities, joint pain, and hematuria** (nephritic syndrome ->IgA nephropathy). Tx is supportive. Watch out for **intussusception** as a potential sequel of the dz.
A 7 yo M is brought to the ED by his mom after having low grade fevers for the past 2 weeks. Physical exam is notable for tenderness to palpation of the large joints of the upper and lower extremities. The patient recently went on a class trip to Hartford, CT. In addition to confirmatory lab testing, what is the next best step in the management of this patient?

a. Administer a treatment course of Amoxicillin.
b. Administer a treatment course of Ceftriaxone.
c. Administer a treatment course of doxycycline.
d. Administer a treatment course of Chloramphenicol.
e. Administer a treatment course of Ciprofloxacin.
The best answer is A. This child has Lyme dz (carried by the Ixodes tick, bug is a spirochete->Borrelia Burgdorferi). The regular tx of LDZ is Doxycycline with the notable exception of children that are < 8 yo. Amoxicillin is the preferred treatment in this age group to prevent permanent tooth discoloration.

In the setting of lyme cardiac/neurological dz, consider using Ceftriaxone as your preferred agent.

Where possible, avoid administering chloramphenicol to little kids to prevent grey baby syndrome. A FQ like Cipro may cause cartilage damage.
Given the following description, what is the most likely diagnosis?

- 2 mo old with his head twisted to the left and his chin twisted to the right.

- PE is notable for a mass palpated along the sternocleidomastoid muscle.
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-PE is notable for a mass palpated along the sternocleidomastoid muscle.

**Congenital Torticollis.** Arise from difficulties or malpositioning of the baby in the process of labor/delivery leading to SCM injury that heals with scar formation. Scar may tighten and shorten the muscle which leads to the clinical presentation. Consider passive neck stretching as first line before seeking other treatment.
Given the following description, what is the most likely diagnosis?

- 2 yo F heading to daycare with a babysitter is throwing tantrums by “throwing herself” on the ground. She is trying to pull away from the babysitter.

- A pop sound is heard.

- The 2 yo begins to complain of pain at the elbow. Forearm is held in a prone position.
Given the following description, what is the most likely diagnosis?

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This is a nursemaid elbow. It is essentially a subluxation of the radial head. Consider hypersupinating/hyperpronating the affected extremity. Common exam scenario.
Q9

Given the following injury scenario related to osteomyelitis, what is the most likely bug?

Most common overall cause of osteomyelitis

Osteomyelitis in a 16 d old neonate

2 yo M w/left ankle osteomyelitis. Recent history of stepping on a “sharp substance”

5 yo M w/right knee osteomyelitis. Spleen is not visible on abdominal imaging.
Q9 Key

Given the following injury scenario related to osteomyelitis, what is the most likely bug?

Most common overall cause of osteomyelitis-\textbf{S. Aureus}.

Osteomyelitis in a 16 d old neonate-\textbf{S. Agalactiae (GBS)}.

2 yo M w/left ankle osteomyelitis. Recent history of stepping on a “sharp substance”-\textbf{P. Aeruginosa (remember association with footwear)}.

5 yo M w/right knee osteomyelitis. Spleen is not visible on abdominal imaging-\textbf{Salmonella (remember the association with sickle cell disease)}.

On the exam, always get an arthrocentesis if you see signs of inflammation at a joint. If an XR is -ve and you suspect osteomyelitis, proceed to an MRI. Get cultures as well.
Q10

Given the following information cluster, what is the most likely diagnosis?

- 7 yo M with fevers for the past 6 weeks.
- He has 2 febrile episodes/day.
- PE is notable for diffuse cervical lymphadenopathy and hepatosplenomegaly.
- Mom reports that he occasionally has a “salmon colored” skin rash that seems to appear with these febrile episodes. They disappear not too long after his fevers subside.
Given the following information cluster, what is the most likely diagnosis?

- 7 yo M with fevers for the past 6 weeks.

- He has 2 febrile episodes/day.

- PE is notable for diffuse cervical lymphadenopathy and hepatosplenomegaly.

- Mom reports that he occasionally has a “salmon colored” skin rash that seems to appear with these febrile episodes. They disappear not too long after his fevers subside.

This is systemic arthritis (Still’s disease). It is one of 3 HY types of JIA that you need to know for your test. There is an association with the “salmon colored rash” and “picket fence fevers”. Treatment typically involves NSAIDs for most kinds of JIA. When this tx fails, proceed to DMARDs like Methotrexate.
- **Oligoarticular JIA** affects only 1-4 joints. The HY association for your test is anterior uveitis. A slit lamp exam may be the correct next best step in management on an NBME. This is the JIA subtype that has the strongest association with **ANA positivity**.

- **Polyarticular JIA** affects 5 or more joints. It is important to know that this one is further subdivided into the **RF+ and RF- subtype**. The RF+ subtype is very destructive and bears a strong semblance to adult RA.

- **Dactylitis** on your test in a young kid may **not always equal Sickle Cell Disease**. In the presence of **“skin findings”** (which for the purposes of the NBME are NOT are a feature of SCD) and other **arthritic symptoms**, consider choosing **Psoriatic Arthritis** as your answer.

- The NBME is very good at presenting “adult” diseases in the peds population to throw you off (especially on Step 2) so watch out.
Given the following clinical presentations, what is the most likely diagnosis?

7 yo M recently had an URI. He presents with a painful limp that began yesterday. WBC is wnl. ESR is marginally elevated. There is no evidence of joint erythema or effusion on PE.

4 yo F is brought to the ED by concerned parents. She is completely fine during the day but cries to sleep at night from bilateral calf pain. Her PE is completely normal. She runs around and plays with her friends during the day.
Q11 Key

Given the following clinical presentations, what is the most likely diagnosis?

7 yo M recently had an URI. He presents with a painful limp that began yesterday. WBC is wnl. ESR is marginally elevated. There is no evidence of joint erythema or effusion on PE—**This is Transient Hip Synovitis. Give NSAIDs. If there is any mention of “joint findings” on the exam, get an arthrocentesis to r/o bad stuff.**

4 yo F is brought to the ED by concerned parents. She is completely fine during the day but cries to sleep at night from bilateral calf pain. Her PE is completely normal. She runs around and plays with her friends during the day—**Growing pains.** The pain classically **shows up at night** and disappears during the day. Consider an **Osteoid Osteoma** as the dx if your exam describes a kid with **localized bone pain that is worse at night and relieved with analgesics.** The PE is typically +ve for a bone mass.
Hemophilia A (F8 deficiency) and B (F9 deficiency) can also present as knee pain from bleeding into the joint (hemarthrosis).

Consider calling CPS if you see fractures in “crawling kids” (< 1 yr) or imaging revealing fractures in different stages of healing (or spiral fractures).

An obese male kid (usually teenager) with hip pain who is also undergoing a big growth spurt should trigger thoughts of SCFE in your mind. XRs are used for dx. **Surgical pinning** is the NBSIM with regards to tx. SCFE is a Type 1 Salter Harris fracture.

Consider **DDH (flat acetabulum)** as the dx in a Q stem detailing an infant with a +ve **Barlow** (bring the hips towards the midline, apply pressures to the knees which may cause a posterior dislocation) or **Ortolani maneuver** (clunk with gentle abduction of the hips and knees held in flexion). Tx w/placement in a Pavlik harness.
-Think of **Legg Calve Perthes disease (LCPD, 6 yo)** as the dx in a kid that has a gradual onset of severe hip pain that creates a trendelenburg pattern of ambulation. One buzzword you’re looking for on an exam is the mention of a teenager with an “antalgic gait”. It is super HY to know that this is an **avascular necrosis of the femoral head**.

-With mention of a physically active child who has pain over the anterior tibial tubercle or anterior leg pain worsened by jumping, consider **Osgood Schlatter dz** as your underlying dx. Pathophysiology involves an inflammatory response to repeated traction of the patellar tendon on the tibial tubercle. A HY buzzword to recognize on your test is the pathophys as a “traction apophysitis”.

-In kids with **clubfoot** (talipes equinovarus), consider **serial manipulation and casting** as the tx of choice.

-Do not forget (**Osteosarcomas/Rb gene mutation**). **Ewings** (11/22, onion skinning).
Given the following information cluster, what is the most likely diagnosis?

- Neonate failed to pass meconium (FTPM) at birth.
- Rectal prolapse as a teenager along with failure to thrive.
- Infertility. Recurrent respiratory infections.
- Diabetes mellitus. Elevated PTH.
Q12 Key (CFTR/Cl- channel/Delta 508 mutation/Cr 7)

Given the following information cluster, what is the most likely diagnosis?

- Neonate failed to pass meconium (FTPM) at birth.
- Rectal prolapse as a teenager along with failure to thrive.
- Infertility. Recurrent respiratory infections.
- Diabetes mellitus. Elevated PTH.

This child has cystic fibrosis. The elevated PTH arises from hypocalcemia secondary to Vit D deficiency since fat soluble vitamins are not reabsorbed (no pancreatic lipase). Remember the association with S. Aureus respiratory infections before 20 and P. Aeruginosa/Burkholderia > 18. Tx involves pancreatic enzyme supplementation, pulmonary toilet, Dornase Alpha, tobramycin for bug coverage, bronchodilators, etc. There’s also a sweat chloride test with diagnostic utility. Super HY to know of the association with nasal polyps/digital clubbing.
A word on respiratory infections Part 1

- Pay attention to the words “wheeze vs stridor” in picking out answers on your shelf.

- The term “wheeze” should get you thinking about a lower respiratory tract infection (LRTI). For a kid < 2 yo, consider RSV as the offending organism (bronchiolitis, usually there’s cough/runny nose some days before). To go with PNA, you need other evidence in the Q like an XR revealing consolidation, or mention of terms like hyperresonance to percussion and increased tactile fremitus.

- The term “stridor” should get you thinking about an URTI like epiglottitis (usually the Q will mention something about super high fevers and acute onset respiratory distress/drooling/difficulty swallowing) OR laryngotracheobronchitis (croup).

- For RSV bronchiolitis, supportive care/humidified O2 is the TOC. Ribavirin use is controversial. For high risk kids (e.g. pulmonary problems), consider palivizumab as prophylaxis. Bronchodilators DO NOT help.
A word on respiratory infections Part 2

- **Croup** is typically caused by the *parainfluenza virus* and is a subglottic obstruction. Consider this dx in an infant with **fever/cough/runny nose** who then develops **inspiratory stridor and a barky/seal like cough**. The Q may be nice and mention the sxs being **worse at night**. Tx is with **racemic epinephrine**. A CXR will be consistent with the “**steeple sign**”.

- **Epiglottitis** arises from supraglottic obstruction. Associate this with the **thumbprint sign** on a CXR. Do not perturb the child. Prepare for emergent **intubation** (call your friendly anesthesiologist).

- The Hemophilus Influenza B vaccine has strongly reduced the incidence of epiglottitis.

- For a kid that was perfectly normal who develops **sudden respiratory distress**, think of **foreign body ingestion** (use a flexible laryngo/bronchoscope).
A word on respiratory infections Part 3

-For a kid that is blue (cyanotic) at rest but pink (acyanotic) with crying, think of choanal atresia as the underlying dx. Crying opens up another O2 route which relieves the cyanosis.

-For a neonate requiring extensive amounts of oxygenation/vent support for prolonged periods after birth, think of bronchopulmonary dysplasia as a potential sequel.
Q13

A 5 mo is brought to the ED by his mom. The infant’s parents have observed what appears to “jerky” movements of the head, arms, and legs for the past 2 weeks. These episodes occur multiple times a day over short time periods. His medical history is notable for gross and fine motor developmental delay. PE reveals hypopigmented macules that are more prominent on the trunk. The rest of the PE is wnl. What is the next best step in the management of this patient?

a. Supportive care.
b. IV Diphenhydramine therapy.
c. Daily oral ethosuximide therapy.
d. Neurosurgical hyperstimulation of the subthalamic nucleus.
e. Symptom control with a Proopiomelanocortin derivative.
Q13 Key

-The best answer is E. This child has **Tuberous Sclerosis and the accompanying infantile spasms** (also known as **West Syndrome**).

-Associate this dx with the classic **hypsarrhythmia** on an EEG. **ACTH** (or vigabatrin) are first line tx options.

-In a 2-6 yo with a wide variety of seizure types, consider **Lennox Gastaut Syndrome** as your underlying dx. Associate this with a 2.5 Hz slow and spike wave activity on an EEG.

-For a **teenager** (note the age difference) that wakes up in the **morning with generalized tonic, clonic seizures**, think of **Juvenile Myoclonic Epilepsy** as your dx.

-You can almost guarantee a question that involves a kid with tons of “**staring episodes**”. These are absence seizures (3 Hz spike and wave). Give ethosuximide.
Q13 Key contd.

- The NBME also loves febrile seizures (MCC seizures in the peds population).

- For a “typical” febrile seizure, administer an NSAID. Resist the temptation to administer antiepileptics or perform any sort of dx testing.

- For “atypical febrile seizures”, a neuro work up may be necessary (focal seizures, lasting > 15 mins, neuro deficits, > 1 in a 24 hr period). Know the criteria.

- For kids with cerebral palsy (of which the spastic type is the most common), consider administering a muscle relaxant like baclofen/dantrolene (or injecting botox) to prevent contractures.
Q14

Given the following information cluster, what is the most likely dx?

-Mutation in chromosome 17.

-PE is notable for iris hamartomas, freckles under the axilla, and multiple “growths” on the body. There are hyperpigmented macules on the skin.

-The infant has a history of a tumor causing an afferent pupillary defect.
Q14 Key

Given the following information cluster, what is the most likely dx?

-Mutation in chromosome 17 (Von Recklinghausen Dz).

-PE is notable for iris hamartomas (Lisch nodules), freckles under the axilla, and multiple “growths” on the body (neurofibromas). There are hyperpigmented macules on the skin (cafe au lait spots).

-The infant has a history of a tumor causing an afferent pupillary defect (optic nerve gliomas).

This child has NF1 (autosomal dominant inheritance like NF2). NF2 is characterized by bilateral sensorineural hearing loss (bilateral vestibular schwannomas). Many patients with NF have an increased predisposition to forming tumors in the brain (like meningiomas), amongst other malignancies (like Pheochromocytomas).
Given the following clinical descriptors, what is the most likely cause of HTN?

15 yo F who recently became sexually active.

16 yo M with an abdominal mass + episodic headache. Urinary metanephrines are elevated.

16 yo F with a normal plasma aldosterone/renin ratio. Plasma renin and aldosterone are elevated.

16 yo M with an elevated plasma aldosterone/renin ratio. Plasma renin is extremely low.

11 yo F with elevated BP in her arms. Dorsalis pedis pulses are not palpable bilaterally.

5 yo F with a thoracic CT revealing a posterior mediastinal mass.

12 yo F with Atrial fibrillation.

8 mo F with ambiguous genitalia.
Q15 Key

Given the following clinical descriptors, what is the most likely cause of HTN?

15 yo F who recently became sexually active—**OCPs.**

16 yo M with an abdominal mass/episodic HA. Urinary metanephrines are elevated—**This is a pheochromocytoma. Remember the association with the MEN syndromes. Block alpha receptors first before blocking betas to prevent unopposed HTN.**

16 yo F with a normal plasma aldosterone/renin ratio. Plasma renin and aldosterone are elevated—**Fibromuscular dysplasia.**

16 yo M with an elevated plasma aldosterone/renin ratio. Plasma renin is extremely low—**Conn’s Syndrome (adrenal adenoma secreting aldosterone, give Spironolactone).**

11 yo F with elevated BP in her arms. Dorsalis pedis pulses are not palpable bilaterally—**Coarctation of the aorta.**

5 yo F with a thoracic CT revealing a posterior mediastinal mass—**Neuroblastoma.**

12 yo F with Atrial fibrillation—**hyperthyroidism.**

8 mo F with ambiguous genitalia—**Congenital Adrenal Hyperplasia (11-beta hydroxylase deficiency, remember that 21-hydroxylase deficiency is the most common cause).**
Some Renal Considerations 1

-In a child presenting with peripheral edema, hematuria, and HTN a few days after an upper respiratory (or abdominal infection), consider IgA nephropathy as the most common dx (especially if the proteinuria is < 3.5g/day).

-A similar presentation 2-6 weeks after an URI should clue you into PSGN (or PIGN). Keywords to associate with this include subepithelial humps, “tea colored urine”, and appropriate testing like ASO and anti-Dnase B antibodies. C3 and 4 are also low with PSGN (since the antibody that constitutes the Ag-Ab complex activates the complement cascade). Note the tetrad of HTN + hematuria + edema + recent infection.

-IgA nephropathy + abdominal pain + joint pain + palpable purpura on the buttocks, thighs, and calf = Henoch Schonlein purpura (systemic vasculitis associated with IgA nephropathy).

-As an addendum, think of Alport Syndrome in a Q stem describing a kid with hearing loss (also in multiple family members), eye problems (cataracts), and kidney problems. Inheritance is in an X-linked dominant pattern secondary to a COL4A5 mutation.
Some Renal Considerations 2

- In a Q stem describing hypoalbuminemia, hyperlipidemia, generalized edema, and heavy proteinuria (with no other antecedents or a relatively “normal looking” renal biopsy result/electron microscopy revealing podocyte foot process effacement), consider Minimal Change Disease as the underlying dx.

- Remember the association of MCD with malignancies like lymphomas.

- In a peds patient with a history of nephrotic syndrome presenting with acute onset flank pain, think of renal vein thrombosis. Nephrotic syndrome is a hypercoagulable state since AT3 is lost in the nephron.

- It is also certainly HY to remember the triad of hemolytic anemia (could be presented as a normocytic anemia with schistocytes/helmet cells on a test, coombs -ve), thrombocytopenia, and acute renal failure (presented as a rising creatinine) as your cue into hemolytic uremic syndrome. There is often bloody diarrhea (E. Coli O157:H7). Avoid antibiotics.
Some Renal Considerations 3

-The pathophysiology of HUS involves infection with E. Coli O157:H7 (bloody diarrhea usually preceded by severe abdominal pain and watery diarrhea) which creates a toxin that injures endothelial cells.

-Injury to endothelial cells causes them to express factors (like VWF) that promote the clotting cascade (which leads to continuous platelet activation).

-The formation of platelet thrombi in blood vessels leads to the “shearing” of RBCs as they traverse these vessels (which explains the hemolytic anemia).

-DO NOT administer antibiotics to these patients since the death of more bacteria serves to increase the toxin pool which will in turn inflict more damage.
Some Renal Considerations 4

- Your friends at the NBME occasionally prefer to throw in some questions relating to benign renal pathology like **transient proteinuria** (after a febrile episode/serious workout regimen) OR **orthostatic proteinuria** (where urine protein goes up during the course of the day when an individual is physically active but goes down when at rest).

- Remember the association of ARPKD with bilateral renal masses in a newborn that could also have liver disease and elements of the POTTER sequence (pulmonary problems, oligohydramnios, skin/face anomalies, extremities defects).
Q16

Given the following descriptors, what is the most likely dx?

A 2 day old is brought to the pediatric surgery suite prior to a TEF repair. He was born at 30 weeks gestation. 5 mins after entry into the suite, the neonate’s skin appears mottled. The room temp is 45.

A 5 day old AA neonate is brought for his 1 week postnatal appointment. PE is notable for grey macules over the intergluteal cleft. The med student alerts child protective services. The rest of the PE are wnl.

A 2 mo old infant is brought to the pediatrician by her concerned mom. PE is notable for a raised red lesion just above her left eyelid. What is the next best step in the management of this patient?
Given the following descriptors, what is the most likely dx?

A G1P1 female is concerned about a rash she noticed on her newborn’s leg and ankle. PE is notable for erythematous yellow white papules/pustules on the involved extremities.
Q16 Key

Given the following descriptors, what is the most likely dx?

A 2 day old is brought to the pediatric surgery suite prior to a TEF repair. He was born at 30 weeks gestation. 5 mins after entry into the suite, the neonate’s skin appears mottled. The room temp is 45-**Cutis Marmorata (vasomotor instability in preemies)**.

A 5 day old AA neonate is brought for his 1 week postnatal appointment. PE is notable for grey macules over the intergluteal cleft. The med student alerts child protective services. The rest of the PE are wnl-**Mongolian Spots (don’t call CPS, common in AAs)**.

A 2 mo old infant is brought to the pediatrician by her concerned mom. PE is notable for a raised red lesion just above her left eyelid. What is the next best step in the management of this patient?-**Capillary hemangioma. Reassure the parent. Most involute in a few years and require no tx. A hemangioma in a dangerous spot, e.g. airway, should be resected.**
Q16 Key contd.

Given the following descriptors, what is the most likely dx?

A G1P1 female is concerned about a rash she noticed on her newborn’s leg and ankle. PE is notable for erythematous yellow white papules/pustules on the involved extremities—**Erythema Toxicum Neonatorum.** Reassure the mom. It is super HY to know that these lesions contain eosinophils. They typically resolve within 2ish weeks.
A 2hr old neonate born at 31 weeks gestation is transferred to the NICU for increasing oxygen requirements. A CXR obtained a few minutes before transfer is notable for lung hypoinflation with obscured left and right sided heart borders. In addition to empiric therapy with a cholesterol derivative, positive pressure ventilation is started. What is the most likely etiology of this neonate’s condition?

a. Chemical pneumononitis from meconium aspiration.
b. Decreased production of surfactant by Type 1 pneumocytes.
c. Increased lung compliance secondary to surfactant deficiency.
d. Delayed resorption of fetal lung fluid.
e. Atelectasis secondary to increased alveolar surface tension.
-The best answer is E, **atelectasis secondary to increased alveolar surface tension**.

-This neonate has **respiratory distress syndrome (RDS)** secondary to a **surfactant deficiency**. The decreased surfactant levels triggers an **increase in lung surface tension**, a **decrease in lung compliance**, an **increased work of breathing**, **atelectasis (lung collapse)**, and a **reduction in SA available for gas exchange**.

-The decreased SA for gas exchange explains the ensuing hypoxia and respiratory distress.

-Tx involves **+ve pressure ventilation using lung protective strategies (low tidal volumes with adequate PEEP)**. Surfactant can also be **delivered directly** to the airway.

-Note the association with **decreased lung volumes (hypoinflation)**, **air bronchograms**, and a “white out lung” on CXR. Do not choose RDS in non-preemies on your exam!
- In contrast, transient tachypnea of the newborn is caused by delayed resorption of fetal lung fluid. RFs (which are usually given on the exam) include maternal DM and cesarean delivery (or super short labor times).

- Look for the following buzzwords on your exam—tachypnea with no hypoxia, perihilar streaking on a CXR with "fluid in the fissures", etc. Resolution typically occurs in a few hours.

- Meconium aspiration syndrome (MAS) is another HY peds respiratory pathology. The HY RF here is post term delivery (> 42 weeks).

- MAS is associated with patchy atelectasis and lung hyperinflation on CXR. Note the difference in lung size with RDS vs MAS. Also note the difference in being preterm with RDS vs being post term with MAS.
Q18

A 5 week old male is brought to the ED by his mom who reports continuous emesis for the past 4 days. BP is 60/40, HR 190 bpm, T 99, RR 25 bpm. PE is notable for a sunken fontanelle and a succussion splash on auscultation of the epigastrium. Further lab testing would most likely reveal?

a. Hypochloremic hypokalemic metabolic acidosis.
b. Hyperchloremic hypokalemic metabolic alkalosis.
c. Hypochloremic hyperkalemic metabolic acidosis.
d. Hypochloremic hypokalemic metabolic alkalosis.
e. Non anion gap hyperkalemic metabolic acidosis.
This is a classic peds NBME question. The best answer is D.

This child has **pyloric stenosis (dx w/US)**. You should have some very cogent reasons for not making this an answer choice in a kid that is usually weeks old with **projectile, nonbilious vomiting** (contrast with the bilious vomiting in duodenal atresia).

The loss of HCl in the process of emesis induces a **hypochloremic metabolic alkalosis**. The concomitant volume depletion upregulates the activity of the RAAS system which coincides with the **hypokalemia**.

PE is often notable (on exams) for a palpable **“olive like” mass in the epigastrium**. The term **“succussion splash”** is code word on NBME exams for a **gastric outlet obstruction**.

HY RFs include being a **firstborn male and exposure to erythromycin** in the first few weeks of life. **Replenish elytes and volume before proceeding to a pyloromyotomy**.
Other HY GI Pathologies

- Consider **intussusception** (telescoping of 1 part of bowel into another) as the dx in a young kid with **episodic abdominal pain that is completely fine b/w episodes**. The NBME may be nice and include the classic “**currant jelly stools**”.

- HY RFs to be aware of include a hx of **Meckel’s diverticulum or some severe GI infection**. As an aside, **DO NOT administer the Rotavirus vaccine to kids with a history of intussusception**.

- Dx is typically with a **contrast (barium) or air enema**. Both methods are **diagnostic and therapeutic**. Dx can also be made with an **US which reveals a “target sign”**. PE may be notable for the palpation of a “**sausage like mass**”.

- In utero (around week 10), the bowel leaves the abdominal cavity, hangs out for a bit, and then returns to its original spot (with 2 rotations) with the small intestine in the center and the large intestine draping over the sides and top of the small intestine.
Other HY GI Pathologies contd.

- Problems with this process can lead to the large and small bowels being in weird spots (e.g. small intestine on the right of the abdomen, large intestine on the left).

- On NBME exams, they often mention imaging revealing the appendix/cecum being in the RUQ. The weird positioning of stuff from incomplete rotation is known as malrotation.

- These “malrotated” organs are ok for the most part. Unfortunately the poor positioning of stuff may make the small intestine twist on itself with the “twisting” ultimately tying off a blood vessel and causing an infarction. This “intestinal twisting on itself” is known as volvulus.

- Dx is with an Upper GI Series (introduce contrast through an NGT and then take pretty XR pictures). Tx is with surgery.
Given the following clinical presentation, what is the most likely dx?

- 4 day old newborn has not pooped since birth.
- PE is notable for abdominal distension.
- An in utero US was notable for endocardial cushion defects.
Q19 Key

Given the following clinical presentation, what is the most likely dx?

-4 day old newborn has not pooped since birth.

-PE is notable for abdominal distension.

-An in utero US was notable for endocardial cushion defects.

This is Hirschsprung’s disease (stool will not be palpated in the rectal vault vs regular constipation with palpable stool) which arises secondary to problems with neural crest cell migration to the GI tract (no Meissner’s/Auerbach’s plexus). Peristaltic waves are absent (often in the terminal GI tract). As an aside, this child has Down’s Syndrome (Tri 21). You need to know certain T21 HY associations (duodenal atresia, hirschsprung’s dz, annular pancreas, celiac dz, endocardial cushion defects, atlantoaxial instability->perform imaging before allowing sports, early ALZ, epicanthal folds).

Meconium ileus can also present in kids with CF (thick secretions).
Given the following clinical presentation, what is the most likely dx?

- Recent history of a febrile adenovirus infection.
- Home aspirin was used to treat the fevers.
- The infant presents today with copious vomiting. He ultimately becomes comatose.
- Blood glucose is 45 g/dl.
Q20 Key

Given the following clinical presentation, what is the most likely dx?

- Recent history of a febrile adenovirus infection.
- Home aspirin was used to treat the fevers.
- The infant presents today with copious vomiting. He ultimately becomes comatose.
- Blood glucose is 45 g/dl.

This is Reye Syndrome. Avoid aspirin as tx with febrile viral infections in kids. The liver and brain stop working (hypoglycemia, confusion/coma). Remember the exception of aspirin with Kawasaki’s disease.
Given the following clinical presentation, what is the most likely dx?

- Neonate born at 30 weeks presents with bilious vomiting and diarrhea. She had her first course of breast milk 5 days ago.

- PE is notable for abdominal distension. BP is 60/40.

- Blood pH is 7.1, Na is 123.

- Abdominal XRs show free air under the diaphragm.
Q21 Key

Given the following clinical presentation, what is the most likely dx?

- Neonate born at 30 weeks presents with bilious vomiting and diarrhea. She had her first course of breast milk 5 days ago.

- PE is notable for abdominal distension. BP is 60/40.

- Blood pH is 7.1, Na is 123.

- Abdominal XRs show free air under the diaphragm.

This is necrotizing enterocolitis. A classic finding on imaging is pneumatosis intestinalis (air in the wall of the bowel). The HY RF to be aware of here is enteral feeding in a preemie. For abdominal perforation (free air under the diaphragm), proceed to surgery. These kids often get antibiotics as well.
Given the following clinical scenario, what is the most likely dx?

- 3 day old newborn is brought to the ED by his mom because he started turning yellow 28 hrs ago. He was born w/o complication.

- PE is completely normal and notable only for scleral icterus and a yellow hue on his face.

- Vital signs are wnl. Hb is 19 (normal). Total bilirubin is 9. Direct bilirubin is 0.7. WBC and other serum markers are wnl. Coombs test is -ve.
Given the following clinical scenario, what is the most likely dx?

-3 day old newborn is brought to the ED by his mom because he started turning yellow 28 hrs ago. He was born w/o complication.

-PE is completely normal and notable only for scleral icterus and a yellow hue on his face.

-Vital signs are wnl. Hb is 19 (normal). Total bilirubin is 9. Direct bilirubin is 0.7. WBC and other serum markers are wnl. Coombs test is -ve.

This is physiologic jaundice. Physiologic jaundice is usually unconjugated and occurs secondary to a transient UGT deficiency in newborns. The T. Bili levels are often mildly elevated (low teens and mostly single digits). Jaundice in the first 24 hrs of life or conjugated hyperbilirubinemia in the newborn is always pathologic. Your NBSIM should revolve around getting bilirubin levels, checking Hb (to look for hemolysis), and getting a Coombs test (to delineate immune mediated causes). Tx is usually supportive (or phototx).
Physiologic jaundice;

- Is always an unconjugated hyperbilirubinemia.
- Does not arise in the first 24 hrs of life.
- For NBME purposes is usually < 12.9
- Does not rise very quickly (rises in 4s/5s)
- Resolves by about 2 weeks of life.
- Can be treated supportively or with phototherapy (if on the higher end of bad).
- Arises from the transient UDPGT deficiency present in newborns.
Some More HY Info Related to Peds Jaundice 1

- With **conjugated hyperbilirubinemia**, the one dx to consider as you prep for your exam is **biliary atresia** which is fixed with the **Kasai procedure** (although most of the these kids end up requiring liver transplants). A lower yield dx to consider is a **choledochal cyst**. As was mentioned earlier, **direct hyperbilirubinemia (DH)** is always pathologic. **DO NOT** pick an answer that encourages the use of phototherapy for DH (does not work!).

- Consider **Rh/ABO incompatibility** as the cause of **indirect hyperbilirubinemia (IHB)** if a Q stem mentions incongruent blood groups in the setting of a +ve coombs test (since the destruction is immune mediated).

- Consider **structural deficiencies** like hereditary spherocytosis (AD inheritance, dx with the osmotic fragility test/eosin-5-maleimide test, spectrin/ankyrin defects, RBCs with no central pallor, elevated MCHC) and h.elliptocytosis or enzyme defects like G6PD deficiency (Heinz bodies, bite cells, fava beans, oxidative drugs, mediterranean populations) and Pyruvate Kinase deficiency if the Coombs test is -ve, Hb is low, and the retic count is elevated.
Some More HY Info Related to Peds Jaundice 2

-An unusual mind bender on an exam would be to give you a question involving a -ve Coombs with an elevated hematocrit. Don’t freak out. Consider causes that give the baby “extra blood” like maternal-fetal transfusion, and the twin-twin transfusion syndrome. Fetal hyperinsulinemia (from maternal DM) may also cause this.

-For indirect hyperbilirubinemia, phototherapy is the TOC. For bilirubin levels > 25 or very close to, proceed to a partial exchange transfusion. Delaying PET can lead to Kernicterus from indirect bilirubin deposition in the basal ganglia and other regions of the brain.

-Consider a Hb problem (like SCD/thalassemia) if problems begin to crop up after 6 mo when HbF has receded.
Pediatric Jaundice—Quick and Dirty Summary

**Jaundice**

- All conjugated / Some unconjugated hyperbilirubinemia
- 1st 24 hr of life

**Physiologic**

- a) Unconjugated hyperbilirubinemia
  - b) ~ 3-4 g/L
  - c) Disappears by ~2 wks
  - d) 1st wk → Breastfeeding Jaundice
  - e) Dehydration / Exaggerated phys
  - f) 1st wk → Breastmilk Jaundice

**Pathologic**

- Unconjugated
  - a) Autosomal Dominant
  - b) Fava beans, Malaria, Meds, Greek, Heinz bodies = G6PD deficiency
  - c) Thalassemias
  - d) ABO/Rh incomp.

- Conjugated
  - a) Biliary Atresia
  - b) #1 dx in newborn unconjugated hyperbilirubinemia

Q23

Given the following info cluster, what is the most likely dx?

-Newborn has a “blueberry muffin rash” and a RUQ mass. A bilateral white reflex is observed on a fundoscopic exam.

-A continuous machine like murmur is heard on auscultation of the chest.

-Otoacoustic emission testing is abnormal 3 days after delivery.

-Mom had poor prenatal care and a febrile illness at 13 weeks with a rash.

-The baby is small for gestational age (SGA).
Q23 Key

Given the following info cluster, what is the most likely dx?

- Newborn has a “blueberry muffin rash” (also present in CMV) and a RUQ mass. A bilateral white reflex is observed on a fundoscopic exam (bilateral cataracts).

- A continuous machine like murmur is heard on auscultation of the chest (PDA).

- Otoacoustic emission testing is abnormal 3 days after delivery (sensorineural HL).

- Mom had poor prenatal care and a febrile illness at 13 weeks with a rash.

- The baby is small for gestational age (SGA).

This is congenital rubella syndrome. It would be a good idea to commit this slide to memory.
Q24

Given the following info cluster, what is the most likely dx?

- Newborn with the triad of chorioretinitis, hydrocephalus, and intracranial calcifications. The baby is SGA.

- Mom routinely cleaned out the cat litter box while she was pregnant.
Q24 Key
Given the following info cluster, what is the most likely dx?

-Newborn with the triad of *chorioretinitis, hydrocephalus, and intracranial calcifications* (vs *periventricular calcifications in CMV*). The baby is SGA.

-Mom routinely cleaned out the cat litter box while she was pregnant.

This is congenital toxoplasmosis. Treatment (of the neonate) is with pyrimethamine and sulfadiazine. Given the administration of folate blocking drugs, consider adding Leucovorin (regenerates folinic acid) to the medication regimen. Spiramycin can be used to treat the infection in the first trimester of pregnancy.

As an aside, consider getting “TORCH Titers” as your dx test whenever you suspect any of these infections. For CMV, consider this as your dx in a Q stem that mentions a kid with periventricular (has a V like CMV, calcifications, microcephaly, and congenital deafness).
Q25

Given the following info cluster, what is the most likely dx?

-Newborn with “snuffles” -> copious amount of nasal secretions.

-Saddlenose deformity, anterior bowing of the lower limbs, upward indentation of the teeth, frontal bossing of the skull.
Q25 Key

Given the following info cluster, what is the most likely dx?

- Newborn with “snuffles” --> copious amount of nasal secretions (on the NBME, snuffles in a newborn is syphilis until proven otherwise!).

- Saddlenose deformity (depression in the bridge of the nose, this is also associated with granulomatosis with polyangiitis/Wegener’s), anterior bowing of the lower limbs (saber shins), upward indentation of the teeth (hutchinson’s teeth), frontal bossing of the skull.

This is congenital syphilis. Testing requires screening with RPR/VDRL and confirmatory testing with MHA-TP and FTA-ABS. Tx mom with PCN. If mom is allergic to PCN and is pregnant (and has syphilis), desensitize her, and still give PCN. Avoid the temptation to give any other drug for syphilis in pregnancy.
Other HY Congenital Infections

- HSV is treated with acyclovir. A common presentation is one revolving around encephalitis/meningitis/sepsis in a neonate with involvement of the temporal lobes in the setting of CSF containing a large number of RBCs.

- If mom has a hx of HSV and has NO active lesions at the time of labor, deliver vaginally. The presence of active vaginal lesions at the time of labor is an indication for cesarean delivery.

- For non-descript questions describing any infectious process/sepsis in a neonate < 28 days, choose GBS as the offending organism. It is the MCC of meningitis, PNA, sepsis, etc in neonates. Most neonatal infections deserve tx with Ampicillin (to cover Listeria) and Cefotaxime.

- A stillborn child with abscesses in multiple body organs/cavities has granulomatosis infantiseptica which is associated with in utero listeria bacteremia.
A 5 yo is brought to the pediatrician for a routine well child visit. Height, weight, and head circumference have tracked consistently in the 30th percentile. BP 98/60, HR 71 bpm, RR 13. A BMP/CBC with differential is wnl with the exception of a Na of 134. Physical exam is unremarkable although the patient complains of constant itching around his scalp. An examination of the scalp reveals patchy hair loss with no specific distribution. A KOH hair scraping is positive for organisms with long tubular structures. What is the next best step in the management of this patient?

a. Oral griseofulvin therapy.
b. Reassurance.
c. Topical administration of selenium sulfide shampoo.
d. Regular hair washing and preventive care.
e. Topical application of ketoconazole to the scalp lesion.
The best answer is A, **oral griseofulvin therapy**. This child has **tinea capitis**.

Tinea capitis is a fungal infection of the scalp caused primarily by **Trichophyton**, **Epidermophyton**, and **Microsporum** species (all dermatophytes, **microsporum fluoresces under Woods Lamp**).

Tinea capitis/unguium require treatment with systemic antifungal therapy (not topical unlike tinea corporis/tinea versicolor).

Consider **trichotillomania** as the dx in a child with a hx of **OCD** who presents with alopecia. A child with **alopecia** who comes in with **abdominal pain/distension** has a **bezoar**.

Well circumscribed hair loss on the NBME is a prescription for **alopecia areata**. Consider **traction alopecia** in situations where a child has “tightly knit” hair.
Q27

Given the following clinical descriptions, what is the most likely diagnosis?

- 4 year old F presents with fever and pain with urination. This is her 2nd UTI in the past 6 mo. PE is notable for CVA tenderness and a palpable LUQ mass. Urine cx obtained through a suprapubic cath (or midstream clean catch) grows 120k colonies of gram -ve lactose fermenting rods.

Newborn with fullness on suprapubic palpation. Gestational hx is notable for oligohydramnios.
Q27 Key
Given the following clinical descriptions, what is the most likely diagnosis?

- A 4-year-old female presents with fever and pain with urination. This is her 2nd UTI in the past 6 months. Physical examination is notable for CVA tenderness and a palpable LUQ mass. Urine culture obtained through a suprapubic cath (or midstream clean catch) grows 120k colonies of gram-negative lactose-fermenting rods—this is pyelonephritis more than likely from vesicoureteral reflux. For a young kid (F) with this many UTIs (commonly E. Coli), consider getting a voiding cystourethrogram to rule out anatomic causes of recurrent urinary infections. Treatment with Ceftriaxone or TMP-SMX. On your exam, certainly get a VCUG if you get a question about any young male infant/female < 5 yo with a UTI (this should never happen). To reiterate, a urine culture is the gold standard for UTI dx.

Newborn with fullness on suprapubic palpation. Gestational history is notable for oligohydramnios—Posterior Urethral Valves which can lead to VUR. Your next step is to get a VCUG. If this is not an option, place a suprapubic catheter.
Some NBME Urinary Oddities

- As you encounter 3rd year, think long and hard about Q stems that encourage you to **admit a patient to the hospital**. This is usually the right answer.

- A kid with cystitis can be sent home on outpatient abx with good f/u. A kid with **pyelonephritis should be admitted** to the hospital.

- Another common pitfall to watch out for is when to administer IV abx. A kid that is dehydrated (low BP, skin tenting, depressed fontanelle), vomiting, or super young (aka neonate) should receive **IV NOT ORAL antibiotics**. They will include both options in the answer choices.
A Few Words On Cerebral Palsy

-CP is a motor disease that is NON-PROGRESSIVE.

-The pathophysiology of CP is not exactly known. However, most experts believe that it all happens from an insult to the child’s brain in development.

-Consider this as your diagnosis in an exam question detailing a child that is otherwise normal and fails to attain motor milestones.

-The exam often describes a child that is spastic.

-Treatment options for the spasticity include baclofen intrathecal pumps and botulinum toxin injections.
Q28

Given the following clinical descriptor, what is the most likely diagnosis?

- 6 mo M comes for his routine Peds visit. His weight at birth was 5 Lbs. His current weight is 9 Lbs. His first bowel movement was at 47 hrs after birth.

- He has experienced 3 bouts of PNA since birth and has foul smelling, greasy stools.

- He was recently treated for rectal prolapse. His mom has noticed that he always tastes salty when the heater is turned on or when he is rocked in the sun.

- PE is notable for digital clubbing and recurrent nasal polyps which have not been well controlled with intranasal steroids.
Q28 Key

Given the following clinical descriptor, what is the most likely diagnosis?

- 6 mo M comes for his routine Peds visit. His weight at birth was 5 Lbs. His current weight is 9 Lbs. His **first bowel movement was at 47 hrs** after birth.

- He has experienced 3 bouts of PNA since birth and has **foul smelling, greasy stools**.

- He was recently treated for **rectal prolapse**. His mom has noticed that he always tastes salty when the heater is turned on or when he is rocked in the sun.

- PE is notable for **digital clubbing and recurrent nasal polyps** which have not been well controlled with intranasal steroids.

This patient has CF. **Sweat chloride testing** is diagnostic. Note that **serum trypsinogen will be decreased** at birth (since the pancreatic duct is clogged).
- CF (autosomal recessive) arises secondary to mutations in the CFTR gene on chromosome 7. Remember the association with S. Aureus infections before 20, P. Aeruginosa infections after 20, and Burkholderia infections before death.

- Tx includes dornase alpha/N-Acetylcysteine (break secretions), inhaled tobramycin (Pseudomonas coverage), pancreatic enzyme replacement, fat soluble vitamin replenishment (ADEK), and chest physiotherapy.

- Greasy stools in a peds patient may also be a presentation of celiac disease (anti-TTG/endomysial/gliadin antibodies). Remember the association with dermatitis herpetiformis and the recommendation to stop consuming gluten.

- Respiratory problems in a kid with a heart displaced to the right (situs inversus) on a CXR should guide you towards Kartagener syndrome (dynein defect).
Given the following clinical scenarios, what is the most likely diagnosis?

7 yo F is brought to the pediatrician by her mom with complaints of sore throat, high fevers, abdominal pain, and vomiting that began suddenly yesterday. PE is notable for tender anterior cervical LND, redness of the hard and soft palate, and a “sandpaper rash” along her UE.

3 yo M is brought to the pediatrician by his mom with complaints of sore throat that started over 3 days ago. He has had a persistent cough and rhinorrhea for the past week.

5 yo F is brought to the pediatrician by her mom with complaints of sore throat and “watery eyes” that started over 3 days ago. PE is notable for severe conjunctival erythema and hyperemia.
Given the following clinical scenarios, what is the most likely diagnosis?

6 yo M is brought to a pediatrician by his mom with complaints of sore throat and high fevers. PE is notable for erythematous lesions with central clearing on the palms. HEENT exam is notable for ulcerative lesions in the mouth.

14 yo F is brought to a pediatrician by her mom with complaints of sore throat and mild fevers for the past 2 weeks. She has been extremely tired and unable to participate in competitive lacrosse over the last 3 weeks. PE is notable for generalized cervical LND. There is fullness on palpation of the LUQ of the abdomen. Initial tx with amoxicillin triggered a severe dermatologic skin reaction which led to the drug being discontinued.

2 yo M is brought to a pediatrician by his “enlightened parents” with complaints of a sore throat. They live in Palo Alto, CA. A HEENT exam is notable for the presence of a grayish “pseudomembrane” in the posterior pharynx with what looks like a “bull's neck”.
Q29A Key

Given the following clinical scenarios, what is the most likely diagnosis?

7 yo F is brought to the pediatrician by her mom with complaints of sore throat, high fevers, abdominal pain, and vomiting that began suddenly yesterday. PE is notable for tender anterior cervical LND, redness of the hard and soft palate, and a “sandpaper rash” along her UE. **This is Strep pharyngitis caused by GAS.** Note the association with anterior cervical LND, palatal erythema, the sandpaper rash, and presence of “extra sxss” like abdominal pain/vomiting. Tx is with PCN/Ampicillin/Amoxicillin. If a child is PCN allergic, consider erythromycin/some other macrolide. A HY clinical decision rule (the centor criteria) is attached on the next slide. Abx are given to prevent rheumatic fever. However, abx do not prevent glomerulonephritis. In general, you need a very good reason to pick strep throat in a kid < 1-2 yo on your NBME. It is very rare in this age group. As an aside, ALWAYS LOOK AT THE AGE WITH PEDS NBME Q’s!
Clinical Decision Rule for Management of Sore Throat

Patient with sore throat
Apply streptococcal score

Criteria
- Absence of cough
- Swollen and tender anterior cervical nodes
- Temperature > 100.4°F (38°C)
- Tonsillar exudates or swelling
- Age
  - 3 to 14 years
  - 15 to 44 years
  - 45 years and older

Points
- 1
- 1
- 1
- 1
- 1
- 0
- 1

Cumulative score: ______

Score ≤ 0
- Risk of GABHS pharyngitis 1 to 2.5%
  - No further testing or antibiotics indicated

Score = 1
- Risk of GABHS pharyngitis 5 to 10%
  - Option
    - Perform throat culture or RADT

Score = 2
- Risk of GABHS pharyngitis 11 to 17%
  - Option
    - Consider empiric treatment with antibiotics

Score = 3
- Risk of GABHS pharyngitis 28 to 35%

Score ≥ 4
- Risk of GABHS pharyngitis 51 to 53%
  - Consider empiric treatment with antibiotics

Negative
- No antibiotics indicated

Positive
- Treat with antibiotics
Given the following clinical scenarios, what is the most likely diagnosis?

3 yo M is brought to the pediatrician by his mom with complaints of sore throat that started over 3 days ago. He has had a persistent cough and rhinorrhea for the past week—**Viral pharyngitis. Note the more gradual onset (vs the more abrupt onset in GABHS pharyngitis), and the presence of “URI” sx before the sore throat begins.**

5 yo F is brought to the pediatrician by her mom with complaints of sore throat and “watery eyes” that started over 3 days ago. PE is notable for severe conjunctival erythema and hyperemia—**This is Adenoviral conjunctivitis. DO NOT get this wrong on an exam—Conjunctivitis + Pharyngitis +/- Abdominal pain = Adenovirus. GAS does not cause conjunctivitis on the NBME.**
Given the following clinical scenarios, what is the most likely diagnosis?

6 yo M is brought to a pediatrician by his mom with complaints of sore throat and high fevers. PE is notable for erythematous lesions with central clearing on the palms. HEENT exam is notable for ulcerative lesions (herpangina) in the mouth—Coxsackie A pharyngitis. **Remember this as one cause of a rash on the palms and soles (HFM dz, also myocarditis).**

14 yo F is brought to a pediatrician by her mom with complaints of sore throat and mild fevers for the past 2 weeks. She has been extremely tired and unable to participate in competitive lacrosse over the last 3 weeks. PE is notable for generalized cervical LND. There is fullness of palpation of the LUQ of the abdomen. Initial tx with amoxicillin triggered a severe dermatologic skin reaction which led to the drug being discontinued—**This is infectious mononucleosis caused by EBV. Tx involves supportive care and avoidance of contact sports. Remember the increased risk of Burkitt’s lymphoma and nasopharyngeal carcinoma with EBV infection. Note the age range. An infant should not have EBV on tests.**
Q29B Key contd.

Given the following clinical scenarios, what is the most likely diagnosis?

2 yo M is brought to a pediatrician by his “enlightened parents” with complaints of a sore throat. They live in Palo Alto, CA. A HEENT exam is notable for the presence of a grayish “pseudomembrane” in the posterior pharynx with what looks like a “bull's neck”- **This is Corynebacterium Diphtheriae. Entirely vaccine preventable. Consider as the dx in immigrants and kids of “enlightened parents”**.

-It is really hard to distinguish between a peritonsillar and retropharyngeal abscess but the clues I use for PTA are the deviation of the uvula, and the “hot potato voice”. RPA is usually associated with a more toxic presentation/drooling where there may be airway compromise (sort of similar to epiglottitis). Retropharyngeal abscesses are usually in kids < 5 on the NBME.
Some HY Vaccine Associations

- A child with a **severe, febrile illness should not be vaccinated**. Vaccines are ok with mild infections.

- A **prior anaphylactic reaction to a given vaccine/component of the vaccine** (e.g. egg allergy in the influenza vaccine) is certainly a contraindication.

- Avoid **live, attenuated vaccines in pregnant women/the immunocompromised** (intranasal influenza, MMR, varicella).

- Kids with **SCD (no spleen) deserve vaccination against encapsulated bugs** (S. Pneumo, H. Influenza, N. Meningitidis). They also deserve regular **PCN prophylaxis**.

- A history of **intussusception/Meckel’s diverticulum** are HY contraindications to the **Rotavirus vaccine**.
Some HY Feeding Associations

- As an aside, **exclusively breastfed kids require Vitamin D supplementation. Avoid honey** till the child is > 1 yo.

- Milk confers some immunity from its **IgA content**. Moms taking **chemo or those with HIV/active herpes on the breast** should not breastfeed. Kids with **essential galactosemia (remember the HY association with death from E. Coli sepsis)** should also NOT breastfeed. Cow’s milk should be avoided at < 12 mo.

- **Milk caries** could occur in kids that are put to bed with a feeding bottle left in their mouths.

- You should still be able to distinguish b/w **UC (continuous, does not touch the terminal ileum)** and Crohn’s (nukes terminal ileum, can cause a B12 deficiency, affects anywhere along the GIT, skip lesions, non-caseating granulomas) in the pediatric population.
Other HY Associations

- Breast milk/most other formula products have an energy value of 20 Kcal/ounce.

- HY RFs for SIDS (unexplained childhood death < 1 yr) include sleeping prone, having parents that smoke, sleeping on a soft surface, being the kid of a super young mom (<20), and overheated rooms.

- Thrush in an infant should be treated with oral nystatin.
Given the following clinical description, what is the most likely diagnosis?

- Child with nasty coughing episodes. The child vomits after particularly severe fits. PE is notable for a unilateral subconjunctival hemorrhage.

- Treatment is with a macrolide antibiotic.

- Bacterial infection associated with a massive lymphocytosis.

- Can be isolated/grown on Regan Lowe/Bordet Gengou media.
Given the following clinical description, what is the most likely diagnosis?

- Child with nasty coughing episodes. The child vomits after particularly severe fits. PE is notable for a unilateral subconjunctival hemorrhage.

- Treatment is with a macrolide antibiotic.

- Bacterial infection associated with a massive lymphocytosis.

- Can be isolated/grown on Regan Lowe/Bordet Gengou media.

This is whooping cough caused by Bordetella Pertussis. The cough is typically followed by an inspiratory whoop. There’s a prodromal URI phase, a catarrhal phase with intense cough, and a convalescent phase where the child recovers. Close contacts should receive macrolide ppx. Spread is via respiratory droplets. Vaccine preventable.
Q31

Given the following clinical description, what is the most likely diagnosis?

- 2 yo presents with bilateral swelling above the jaw.

- His sx started 2 days ago as neck pain, fever, and a severe headache.

- Vaccine preventable.

- HY sequelae include inflammation of the testes (causing infertility) and pancreatitis.
Q31 Key

Given the following clinical description, what is the most likely diagnosis?

- 2 yo presents with bilateral swelling above the jaw.
- His sx started 2 days ago as neck pain, fever, and a severe headache.  
- Vaccine preventable. 
- HY sequelae include inflammation of the testes (causing infertility) and pancreatitis.

This is mumps. Classic presentation is as orchitis/parotitis. Prevent with the MMR vaccine (avoid in pregnant women and kids < 1 yo).
Q32

Given the following clinical description, what is the most likely diagnosis?

-Present in 2% of the population. Is 2 ft from the ileocecal valve. Contains 2 types of tissue.

-Kid presents with painless, episodic rectal bleeding.

-Diagnosis is with a technetium scan.

-Can serve as a lead point for intussusception.
Q32 Key

Given the following clinical description, what is the most likely diagnosis?

- Present in 2% of the population. Is 2 ft from the ileocecal valve. Contains 2 types of tissue.

- Kid presents with painless, episodic rectal bleeding.

- Diagnosis is with a technetium scan.

- Can serve as a lead point for intussusception.

This is Meckel’s diverticulum (a true diverticulum). After dx with the Technetium scan, proceed to surgical resection.

As mentioned earlier, associate intussusception with colicky abdominal pain, currant jelly stools, and a “sausage shaped mass” in the RUQ. Tx with an air/contrast enema.
Given the following clinical description, what is the most likely diagnosis?

-A 2 yo is angry with mom for not giving him breast milk.

-His mom notices the child suddenly passing out on the carpet.

-He regains consciousness within 15s.
Q33 Key

Given the following clinical description, what is the most likely diagnosis?

-A 2 yo is angry with mom for not giving him breast milk.

-His mom notices the child suddenly passing out on the carpet.

-He regains consciousness within 15s.

This is a breath holding spell (the cyanotic kind often associated with anger/fear). There’s also the “pallidic” type which is often associated with pain/element of surprise. Consider replenishing Fe in kids with BHS (if Fe deficiency exists).
Q34

Given the information cluster, what is the most likely dx?

- Female with short stature, webbing of the neck, and infertility.

- Coarctation of the aorta and horseshoe kidney.

- Common cause of primary amenorrhea.
Q34 Key

Given the information cluster, what is the most likely dx?

- Female with short stature, webbing of the neck, and infertility.

- Coaractation of the aorta and horseshoe kidney.

- Common cause of primary amenorrhea.

Another reminder for Turner Syndrome. In general, your dx revolves around getting a karyotype which will be 45 XO. Consider giving exogenous estrogen/progesterone for uterine protection in addition to growth hormone.
Q35

Given the information cluster, what is the most likely dx?

-Super small testicles.

-6ft tall male with gynecomastia and infertility.

-IQ is 80 but patient is otherwise normal.
Q35 Key

Given the information cluster, what is the most likely dx?

- Super small testicles.
- 6ft tall male with gynecomastia and infertility.
- IQ is 80 but patient is otherwise normal.

This is Klinefelter’s syndrome (47 XXY).

Think of the XYY karyotype in a tall guy with significant acne.

Think of the XXX karyotype in a female with “menstrual problems” but the specific association of a normal height with an abnormally small head circumference (classically 80% for height but 25% for head circumference)
Q36-Genetic/Toxin/Miscellaneous Triggers

ID, low BW, microcephaly, high pitched cat like cry
Obesity, infantile hypotonia, ID, small extremities, Cr 15 deletion
Happy mood, ID, inappropriate laughter, ataxia, Cr 15 deletion (mom)
“Elfin facies”, short upward turned nose, long philtrum
X-linked, CGG repeat, anticipation, long face, large ears, ID, macroorchidism
Short palpebral fissures, thin upper lip, smooth philtrum, microcephaly, VSD
Tricuspid valve displaced downward, RV hypoplasia
Lateral neck mass that does not move with swallowing, derived from ectoderm
Midline neck mass, moves with swallowing, may have ectopic thyroid tissue
Hypopigmented macules identified with Wood’s lamp, shagreen patches, adenoma sebaceum (face), infantile spasms, subependymal nodules in brain, cardiac rhabdomyoma, renal angiomyolipoma
Severe fasting hypoglycemia, lactic acidosis, hepatomegaly, gout, may have kidney sxs (since this also does gluconeogenesis), no muscle sxs, give cornstarch
GSD associated with heart failure and death at < 2 yo
Mild hypoglycemia, hepatomegaly, muscle sxs, no lactic acidosis
No lactic acidosis, muscle weakness, no hepatomegaly
Q36 Key-Genetic/Toxin/Miscellaneous Triggers (Sorry!)

ID, low BW, microcephaly, high pitched cat like cry-Cri du Chat syndrome
Obesity, infantile hypotonia, ID, small extremities, Cr 15 deletion-Prader Willi (dad deletion)
Happy mood, ID, inappropriate laughter, ataxia, Cr 15 deletion (mom)-Angelman
“Elfin facies”, short upward turned nose, long philtrum-Williams Syndrome (also cardiac problems)
X-linked, CGG repeat, anticipation, long face, large ears, ID, macroorchidism-Fragile X Syndrome
Short palpebral fissures, thin upper lip, smooth philtrum, microcephaly, VSD-Fetal ROH syndrome
Tricuspid valve displaced downward, RV hypoplasia-Li toxicity (Ebstein’s anomaly)
Lateral neck mass that does not move with swallowing, derived from ectoderm-branchial cleft cyst
Midline neck mass, moves with swallowing, may have ectopic thyroid tissue-Thyroglossal duct cyst
Hypopigmented macules identified with Wood’s lamp, shagreen patches, adenoma sebaceum (face),
infantile spasms, subependymal nodules in brain, cardiac rhabdomyosarcoma, renal
angiomyolipoma-Tuberous Sclerosis
Severe fasting hypoglycemia, lactic acidosis, hepatomegaly, gout, may have kidney sxs (since this also
does gluconeogenesis), no muscle sxs, give cornstarch-Von Gierke (GSD 1, G-6-P’ase gone)
GSD associated with heart failure and death at < 2 yo-Pompe’s dz (GSD 2, lysosomal acid maltase gone)
Mild hypoglycemia, hepatomegaly, muscle sxs, no lactic acidosis-Cori Dz (GSD 3, debranching enzyme
gone)
No lactic acidosis, muscle weakness, no hepatomegaly-McArdle’s dz (GSD 5, muscle glycogen
phosphorylase gone). Also remember I-Cell dz with the mannose-6-P problems.
Given the following clinical description, what is the most likely diagnosis?

- On December 1, a 4 yo immigrant has a T of 102, cough, bilateral ocular erythema, and runny nose.

- PE in the office reveals whitish spots on the buccal mucosa and palate. There is a red maculopapular rash on the face that spreads down to the trunk.

- 5 days after supportive care, the rash resolves in the same distribution.
Given the following clinical description, what is the most likely diagnosis?

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- 5 days after supportive care, the rash resolves in the same distribution.

This is measles (rubeola). Remember the 3C's (cough, coryza, and conjunctivitis). Note the progression in symptoms (URI prodrome->Koplik spots->rash->rash resolution). Tx is supportive but it is very HY to know that Vitamin A is helpful. Consider Subacute Sclerosing Panencephalitis (SSPE) as the dx if the Q stem describes a child with a distant measles infection now presenting with neurologic problems (e.g. seizures).
Q38

Given the following clinical description, what is the most likely diagnosis?

- On December 1st, a 2 yo has low grade fevers and does not “feel well”.

- By December 5th, posterior auricular and suboccipital lymphadenopathy can be appreciated.

- Rash starts on the head and moves downward.
Given the following clinical description, what is the most likely diagnosis?

- On December 1st, a 2 yo has low grade fevers and does not “feel well”.

- By December 5th, **posterior auricular and suboccipital lymphadenopathy** can be appreciated.

- Rash starts on the head and moves downward.

**This is German measles (Rubella)**

As an aside, do not forget your Coxsackievirus (A) association with hand, foot, mouth dz.

Think of Roseola/HHV 6 (exanthem subitum) as the dx in a “playful” kid with high fevers that precede a “sudden appearing” rash.

In a 2 yo with recurrent, well timed monthly fevers, aphthous ulcers, sore throat, and painful LND, consider PFAPA as the dx (Periodic Fever with Aphthous ulcer, Pharyngitis, Adenitis).
Q39

Given the following clinical descriptors, what is the most likely dx/NBSIM?

A medical student rotating in the nursery notices severe scalp swelling in a newborn male. The edema from the lesion crosses suture lines.

A medical student rotating in the nursery notices severe scalp swelling in a newborn female. The edema from the lesion does not cross suture lines.

A medical student rotating in the nursery notices streaks of blood emanating from the vagina of a 3 day old newborn female. What is the NBSIM?
Q39 Key
Given the following clinical descriptors, what is the most likely dx/NBSIM?

A medical student rotating in the nursery notices severe scalp swelling in a newborn male. The edema from the lesion **crosses suture lines—Caput Succedaneum.** Is simply a swelling of the scalp secondary to “barotrauma” from going through the birth canal. Completely benign and resolves within days.

A medical student rotating in the nursery notices severe scalp swelling in a newborn female. The edema from the lesion **does not cross suture lines—Cephalohematoma.** Is a subperiosteal bleed. Resolves in weeks-months. Increased risk for anemia/jaundice as the blood resorbs.

A medical student rotating in the nursery notices streaks of blood emanating from the vagina of a 3 day old newborn female. What is the NBSIM? **Normal/reassure the parents.** Arises secondary to withdrawal of maternal hormones (uterus shedding).
The APGAR Score (no correlation w/outcomes)

<table>
<thead>
<tr>
<th>Activity (muscle tone)</th>
<th>0 Points</th>
<th>1 Point</th>
<th>2 Points</th>
<th>Points totaled</th>
</tr>
</thead>
<tbody>
<tr>
<td>Absent</td>
<td>Arms and legs flexed</td>
<td>Active movement</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Pulse</td>
<td>Absent</td>
<td>Below 100 bpm</td>
<td>Over 100 bpm</td>
<td></td>
</tr>
<tr>
<td>Grimace (reflex irritability)</td>
<td>Flaccid</td>
<td>Some flexion of Extremities</td>
<td>Active motion (sneeze, cough, pull away)</td>
<td></td>
</tr>
<tr>
<td>Appearance (skin color)</td>
<td>Blue, pale</td>
<td>Body pink, Extremities blue</td>
<td>Completely pink</td>
<td></td>
</tr>
<tr>
<td>Respiration</td>
<td>Absent</td>
<td>Slow, irregular</td>
<td>Vigorous cry</td>
<td></td>
</tr>
</tbody>
</table>

| Severe depressed       | 0-3      |
| Moderately depressed   | 4-6      |
| Excellent condition    | 7-10     |
Tetralogy of Fallot Pathophysiology

- Normal
- Aorta-pulmonary Septum (APS)
- Aorta (A)
- Pulmonary Trunk (PT)
- RV
- LV
- Muscular Interventricular Septum (MIVS)

**Note**: Equal division

**A**: Smaller PT = Pulmonic Stenosis

**B**: Bigger A = Overriding Aorta

**C**: RV Hypertrophy

- RV Pump against the "stenosed" PT
- VSD due to APS mal-positioning

**Note**: Unequal division

**APS (displaced to right)**

**PT**

**LV**

**RV**

**MIVS**
Q40

Given the following clinical description, what is the most likely dx?

- 15 yo M presents to his pediatrician with a 3 mo history of fevers and pain just below the femoral head.

- The pain was initially well controlled with acetaminophen but has progressively gotten worse (and unbearable) over the past 4 weeks.

- Labs are notable for an increased ESR and a mild leukocytosis.

- Imaging of the femur is consistent with a moth eaten appearance and a “concentric whorling” periosteal bone pattern. Biopsy of the lesion reveals “small blue cells”.

Q40 Key

Given the following clinical description, what is the most likely dx?

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- The pain was initially well controlled with acetaminophen but has progressively gotten worse (and unbearable) over the past 4 weeks.

- Labs are notable for an increased ESR and a mild leukocytosis.

- Imaging of the femur is consistent with a moth eaten appearance and a “concentric whorling” periosteal bone pattern. Biopsy of the lesion reveals “small blue cells”.

Ewing’s sarcoma. Note the association with “onion skinning” on imaging. Remember your 11:22 translocation (EWS-FLI) and potential treatment with dactinomycin.
Q41 A

Given the following clinical descriptors, what is the most likely pediatric cardiac anomaly?

Newborn presents with cyanosis 30 mins after delivery. Pulmonary vascular markings are normal. An emergent CXR reveals a heart that is shaped like an “egg on a string”

A med19 student rotating in the nursery calls the peds attending to report a pertinent finding. Auscultation of a 3 day old newborn with APGAR scores of 8 and 9 at 1 and 5 mins reveals a systolic ejection murmur at the LUSB and a wide, fixed, splitting of the S2 heart sound. A CXR is notable for right heart enlargement and increased pulmonary vascular markings
Q41 A Key
Given the following clinical descriptors, what is the most likely pediatric cardiac anomaly?

Newborn presents with cyanosis 30 mins after delivery. Pulmonary vascular markings are normal. An emergent CXR reveals a heart that is shaped like an “egg on a string” - This is a transposition of the great vessels (no spiral of the aorticopulmonary septum which is derived from neural crest cells). Some communication must exist b/w the left and right heart in utero for the child to survive. Consider administering a PGE1 analog as the NBSIM with future plans to reverse the arterial orientations.

A med19 student rotating in the nursery calls the peds attending to report a pertinent finding. Auscultation of a 3 day old newborn with APGAR scores of 8 and 9 at 1 and 5 mins reveals a systolic ejection murmur at the LUSB and a wide, fixed, splitting of the S2 heart sound. A CXR is notable for right heart enlargement and increased pulmonary vascular markings - This is an ASD. For all cardiac pathologies, consider getting an echocardiogram as a part of your initial dx plan. Remember the HY sequelae of Eisenmenger’s syndrome that is associated with an ASD. Note that an Ostium Primum ASD is more common in T21 (Down’s Syndrome). Remember kids with ASD having stereotypical/fixed behaviors just like the fixed split in ASD.
Q41 B

Given the following clinical descriptors, what is the most likely pediatric cardiac anomaly?

A 16 yo F is brought to her pediatrician with a 4 year history of severe, recurrent headaches. She is 4 ft tall and has a low posterior hairline in addition to chronic lymphedema on both sides of her neck. She had surgery 5 years ago for some “kidney problem”. PE is notable for a loud murmur around the left scapula. Her LE posterior tibial and dorsalis pedis pulses are barely palpable. A CXR is notable for what appears to be rib indentations and a “3” sign just after the aortic arch.
Q41 B Key

Given the following clinical descriptors, what is the most likely pediatric cardiac anomaly?

A 16 yo F is brought to her pediatrician with a 4 year history of severe, recurrent headaches. She is 4 ft tall and has a low posterior hairline in addition to chronic lymphedema on both sides of her neck. She had surgery 5 years ago for some “kidney problem”. PE is notable for a loud murmur around the left scapula. Her LE posterior tibial and dorsalis pedis pulses are barely palpable. A CXR is notable for what appears to be rib indentations and a “3” sign just after the aortic arch.

This is a coarctation of the aorta which is common in kids with Turner Syndrome (preductal). If the child presents with severe cyanosis at birth, proceed to PGE1 analog tx (alprostadil).
Given the following clinical descriptors, what is the most likely pediatric cardiac anomaly?

A newborn presents to a pediatrician for his 1 week postnatal appointment. Cardiac auscultation is notable for a holosystolic murmur best heard at the LLSB, a loud pulmonic second heart sound, and an apical diastolic rumble. A CXR is consistent with increased pulmonary vascular markings.

A newborn with multiple facial defects and hypocalcemic seizures is transported to the Med19 NICU for persistent cyanosis. Cardiac auscultation is notable for a single S2 heart sound.
Q41 C Key

Given the following clinical descriptors, what is the most likely pediatric cardiac anomaly?

A newborn presents to a pediatrician for his 1 week postnatal appointment. Cardiac auscultation is notable for a holosystolic murmur best heard at the LLSB, a loud pulmonic second heart sound, and an apical diastolic rumble. A CXR is consistent with increased pulmonary vascular markings—This is a VSD (membranous kinds are more common). Smaller VSDs sound louder.

A newborn with multiple facial defects and hypocalcemic seizures is transported to the Med19 NICU for persistent cyanosis. Cardiac auscultation is notable for a single S2 heart sound—This is Truncus Arteriosus. Arises from a failure of the aorticopulmonary septum to separate the truncus arteriosus into the pulmonary artery and aorta. Very common in DiGeorge Syndrome. Pulmonary vascular markings will be increased.
Q41 D

Given the following clinical descriptors, what is the most likely pediatric cardiac anomaly?

A 3 hr old newborn is seen by a med student in the NICU for persistent cyanosis. Cardiac auscultation is consistent with a wide, fixed, split of the second heart sound, a harsh, holosystolic murmur at the LLSB, and a “diminished” S1. A CXR is consistent with decreased pulmonary vascular markings. An EKG is notable for left axis deviation
Given the following clinical descriptors, what is the most likely pediatric cardiac anomaly?

A 3 hr old newborn is seen by a med student in the NICU for persistent cyanosis. Cardiac auscultation is consistent with a wide, fixed, split of the second heart sound, a harsh, holosystolic murmur at the LLSB, and a “diminished” S1. A CXR is consistent with decreased pulmonary vascular markings (same as TOF). An EKG is notable for left axis deviation—This is tricuspid atresia. This is the only congenital cyanotic defect with LAD. I’ll discuss mechanisms behind the findings in the review. Consider initial tx with our PGE1 analog. There’s a single S2 in some types (also in TGA and truncus arteriosus).

As an aside, remember the association of congenital rubella with a PDA (continuous machine like murmur since there is always flow through the defect in all phases of the cardiac cycle). Give indomethacin/close surgically. Also note the “maneuvers” that will make tet spells in tetralogy of fallot better.
Q42

A 6 ft 7 in tall 12 yo M is rushed to the ED by ambulance with chest pain. A screening EKG on presentation reveals ST elevations in V5-6, 1, and avL. A 325 mg acetylsalicylic acid dose is administered enroute to the cardiac catheterization lab. There is 85% stenosis of the left anterior descending and complete occlusion of the left circumflex artery detected on coronary angiography. The child has a history of severe mental retardation and lives at home with his parents. The pathophysiology of this patient’s presentation would most likely be observed in an individual with?

a. A normal IQ, gigantism, and a history of mitral valve prolapse.
b. Elevated serum titers of an autoantibody against parietal cells.
c. Low serum levels of a transketolase cofactor in the HMP shunt.
d. A normal IQ, gigantism, and a history of neural growth on the lips and buccal mucosa.
e. A smooth philtrum, thin vermilion border, and microcephaly.
- The best answer here is B. This child has **hyperhomocysteinemia (AR inheritance)**.

- Hyperhomocysteinemia is associated with deficiencies in **CBS and MTHFR** which leads to homocysteine (HCS) accumulation and accelerated atherosclerosis. There is no good tx, but serum HCS levels can be lowered with Vitamin B6/9/12.

- You should be able to **distinguish hyperhomocysteinemia** (marfanoid habitus, MR, lens displaced downwards) from **marfan syndrome** (marfanoid habitus, no MR, lens displaced upwards, MVP, aortic regurgitation, pectus issues, fibrillin-1 mutation) and **MEN2B** (marfanoid habitus, mucosal neuromas, medullary thyroid cancer, and pheochromocytoma).

- Other HY metabolic disorders to be aware of include-> **Galactosemia** (GALT deficiency) which presents with **hepatosplenomegaly (HSM), diarrhea, and cataracts in a newborn**. Sxs begin with administration of milk. Foods containing **lactose and galactose** should be avoided. A common cause of death is **E. Coli sepsis**.
Q42 Key contd.
-
**Fructose intolerance (aldolase B deficiency, no cataracts)** often presents with HSM, diarrhea, and other severe problems after “fruit juice” and other fructose containing foods have been introduced into the diet. In general, this presentation will begin at > 6 mo age on NBME exams, not the newborn period as observed for galactosemia.

-A self injurious boy (XLR inheritance) with gout and MR should get you thinking about **Lesch Nyhan Syndrome (HGPRT deficiency)**. Give allopurinol.

-A kid with a mousy/musty odor who presents with MR and “skin anomalies” should get you thinking of PKU (phenylalanine hydroxylase or tetrahydrobiopterin reductase/BH4 deficiency, AR inheritance). Avoid aspartame/phenylalanine containing foods. **Tyrosine** becomes an essential AA in these patients.

-As a pro tip, disorders involving **specific odors** on your exam correlate more with **amino acid metabolism** problems.
- A newborn with secretions (sweat, urine, etc) that smell like maple syrup and elevated levels of branched chain AAs in the serum (“LIV”-> leucine, isoleucine, and valine) should get you thinking about MSUD (AR inheritance, branched chain ketoacid dehydrogenase deficiency). Remember the “TLCFN” association from Step 1.

- A kid that presents with skin/neuro problems (or other signs of B3/Niacin deficiency-> the 4Ds) in the setting of increased levels of neutral AAs in the urine raise concerns for Hartnup dz. Note that a tryptophan deficiency explains most of the pathophysiology.

- Consider OTC (Ornithine transcarbamylase) deficiency as the dx in a kid presenting with neuro problems from elevated ammonia levels (with a concomitant orotic acidemia).
Finally;
A kid < 1 yo with a weird startle response, a cherry red spot on the macula, and no HSM = Tay Sachs disease (hexosaminidase deficiency).
Similar presentation with HSM = Niemann Pick dz (sphingomyelinase deficiency).
Neuro deficits + lactic acidosis + ragged red muscle fibers on biopsy = Some kind of mitochondrial disorder (e.g. LHON, MELAS, etc).
A hemangioma that sequesters platelets leading to thrombocytopenia = Kasabach Merritt Syndrome.
Low plts in the setting of skeletal deficits (like an absent radius) = TAR syndrome.
Corneal clouding = Hurler dz, no corneal clouding w/coarse facial features/X-linked = Hunter's.
Think of Gaucher dz (glucocerebrosidase deficiency) in a Q stem describing pancytopenia and the presence of cells with a “tissue paper” like cytoplasmic morphology on bone marrow biopsy.
Hypoketotic hypoglycemia is the buzzword for a fatty acid oxidation disorder. If you get exercise intolerance (muscle problems), think more of an MCAD deficiency vs a carnitine deficiency.
Anything relating to the accumulation of VLCFAs on your test should get you thinking of a peroxisomal disorder (like Zellweger syndrome)
Given the following clinical descriptors, what is the most likely dx?

A newborn presents with drooling after initial breastfeeding. The NICU nurse cannot pass a catheter all the way through to his stomach.

A newborn presents with leukocoria. This newborn is at increased risk of what malignancy in the future?

A 3 mo presents with circumoral pallor and cyanosis. CBC is notable for severe anemia. The child has a hx of thumb and craniofacial anomalies.
Q43 Key

Given the following clinical descriptors, what is the most likely dx?

A newborn presents with drooling after initial breastfeeding. The NICU nurse cannot pass a catheter all the way through to his stomach—**Esophageal atresia with TEF. Note the association with polyhydramnios and the VACTERL association.**

A newborn presents with leukocoria. This newborn is at increased risk of what malignancy in the future?—**This neonate has retinoblastoma. Associated with an increased risk of osteosarcoma in the future.**

A 3 mo presents with circumoral pallor and cyanosis. CBC is notable for severe anemia. The child has a hx of thumb and craniofacial anomalies—**This is Diamond Blackfan anemia. Note the age (< 1 yo) and associated anomalies. Contrast with Fanconi anemia (chromosomal breakage) where other “blood” cell lines could be down with the addition of cafe au lait spots.**
Final Pathologies

- Remember the association of terminal (C5-9) complement deficiencies with Neisseria infections. These kids deserve vaccines against “SHiN” organisms. An unusual exam Q would revolve around similar pathologies in a patient on Eculizumab for paroxysmal nocturnal hemoglobinuria (GPI anchor problems).

- An exam Q detailing a kid with progressive flaccid paralysis that is < 1 yo with muscle weakness, tongue fasciculations, and ultimately death from respiratory insufficiency should get you thinking of Werdnig Hoffman disease (also known as Spinal Muscular Atrophy) which is inherited in an AR fashion secondary to a mutation in the SMN1 gene.

- Displacement of cerebellar tonsils downward through the foramen magnum which ultimately presents as hydrocephalus should get you thinking about a Chiari 1 malformation (super HY to know its association with syringomyelia).
Final Pathologies contd.

- Displacement of the cerebellar tonsils and vermis towards the foramen magnum presenting as hydrocephalus should get you thinking about a Chiari 2 malformation (super HY to know the association with a lumbosacral myelomeningocele).

- A kid with dwarfism who has normal intelligence and bowed legs should get you thinking about achondroplasia which arises secondary to an AD mutation in FGFR3 (on Chromosome 4, endochondral bone formation problems).

- A child with blue sclera, hearing difficulty, and multiple fractures should get you thinking about an AD Type 1 collagen production defect (Osteogenesis Imperfecta).

- Consider Duchenne MD (X linked dystrophin mutation) in a teenager presenting with calf pseudohypertrophy, Gower’s sign, and elevated creatine kinase. Becker’s MD presents similarly but progresses slowly (there’s some dystrophin vs DMD with zero dystrophin).
All The Best!

KEEP CALM AND CRUSH THE EXAM