

# High Yield Pediatrics

Shelf Exam Review

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# The Newborn

# APGAR

- Pulse of 130, acrocyanotic, grimaces to stimulation, moving all extremities and crying.
- Score? 8.

2pts for pulse, 1 for color, 1 for irritability, 2 for tone and 2 for respiration

- What does the APGAR tell you?  
General info about how the newborn tolerated labor (1min) and the newborn's response to resuscitation (5min)
- What does the APGAR *not* tell you  
What to do next (does not guide therapy)  
How the baby will turn out (does NOT predict neurologic outcome)

# And on physical exam you find...

- When assessing Moro on an LGA newborn, the right arm remains extended and medially rotated.
- When palpating the clavicles on a LGA newborn, you feel crepitus and discontinuity on the left.

**Erb-Duchenne C5-C6.**  
(Klumpke is C7-C8 + T1)  
Refer if not better by 3-6mo for neuroplasty

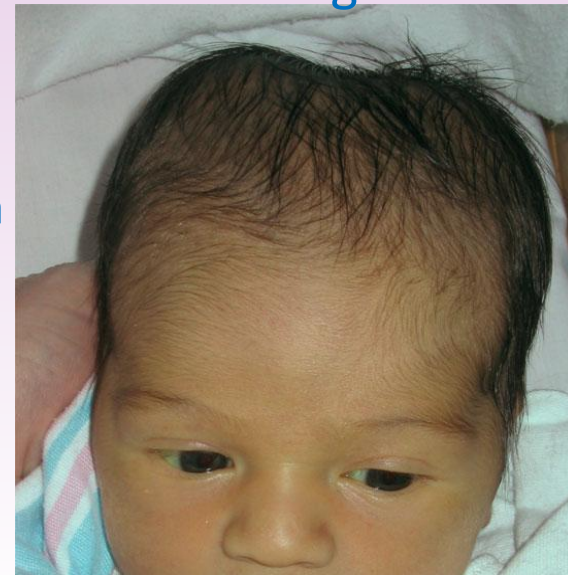
## **Clavicular Fracture.**

Will form a callus in 1wk. No tx needed. Can use figure of 8 splint.



## **Caput succedaneum**

“Edema. Crosses suture lines.”



## **Cephalo-hematoma**

“Fluctuance. Doesn't cross suture lines.”



Mongolian Spots



Nevus Simplex  
(Salmon Patch)



Milia



Erythema toxicum



Strawberry  
Hemangioma



Neonatal Acne





## Nevus Sebaceous

Described as “an area of alopecia with orange colored nodular skin”.

What to do?

Remove before adolescence b/c it can undergo malignant degeneration.



## Seborrheic Dermatitis

Described as “thick, yellow/white oily scale on an inflammatory base”.

What to do?

Gently clean w/ mild shampoo

# Neonatal Screen

- Two disorders screened for in every state because they are disastrous if not caught early (and happen to be a contraindication to breast feeding...)

## Phenylketonuria.

- Deficient Phe hydroxylase.
- Sxs = MR, vomiting, athetosis, seizures, developmental delay over 1<sup>st</sup> few mos
- Signs = fair hair, eyes, skin, musty smell.
- Low Phe diet.

## Galactosemia.

- Deficient G1p-uridyl-transferase. G1p accum to damage kidney, liver, brain.
- Sxs = MR direct hyperbili & jaundice, ↓glc, cataracts, seizures.
- Predisposed to E. coli sepsis.
- No lactose por vida.

# A Yellow Baby

- 3 days old, bili @ 10, direct is 0.5. Eating & pooping well.
- 7 days old, bili @ 12, direct is 0.5. dry mucous membranes, not gaining weight.
- 14 days old, bili @ 12, direct is 0.5. Baby regained birth weight, otherwise healthy.
- 1 day old, bili @ 14, direct is 0.5. Are you worried?

- Next best test? **Coombs**

- If positive? **Means Rh or ABO incompatibility**

- If negative? **Means twin/twin or mom/fetus transfusion, IDM, spherocytosis, G6p-DH deficiency, etc.**

**Physiologic Jaundice.** Gone by 5<sup>th</sup> DOL.

Liver conjugation not yet mature.

**Breast feeding Jaundice.** ↓feeding = dehydration = retain meconium & re-absorb deconjugated bili.

**Breast milk Jaundice.** Breast milk has glucuronidase and de-conj bili.

**Pathologic Jaundice** = on 1<sup>st</sup> DOL, bili >12, d-bili >2, rate of rise >5/day.



- 7 days old. Dark urine, pale stool. Bili @ 12, dbili is 8. LFTs also elevated. **Biliary atresia.** Bile ducts cannot drain bile. Causes liver failure. Need surgery.
- Other causes of direct hyperbilirubinemia? **Always r/o sepsis!** Galactosemia, hypothyroid, choledochal cyst, CF
- Random inherited causes of indirect hyperbili? (2) **Gilbert.** ↓ glucoronyl transferase level **Crigler-Najjar.** (type1) total deficiency
- Random inherited causes of direct hyperbili (2) **Dubin Johnson.** black liver. **Rotor.** No black liver.
- Why do we care about hyperbilirubinemia? **Indirect bili** can cross BBB, deposit in BG and brainstem nuclei and cause kernicterus. (esp if bili is >20)
- What is the treatment? **Phototherapy** → ionizes the unconj bili so it can be excreted. **Double volume exchange transfusion** if that doesn't work.

# Respiratory Disorders



Baby is born w/ respiratory distress, scaphoid abdomen & this CXR.

Diaphragmatic hernia

- Biggest concern? Pulmonary hypoplasia
- Best treatment? If dx prenatally, plan delivery at @ place w/ ECMO. Let lungs mature 3-4 days then do surg

Baby is born w/ respiratory distress w/ excess drooling. TE- Fistula

- Best diagnostic test? Place feeding tube, take xray, see it coiled in thorax
- What else do you look for? VACTER associated anomalies- vertebral, anal atresia, cardiac, radial and renal.

1 week old baby becomes cyanotic when feeding but pinks up when crying. Choanal Atresia

- What else do you look for? CHARGE associated anomalies- coloboma, heart defects, retarded growth, GU anomalies, Ear anomalies and deafness

32 wk premie has dyspnea, RDS  
RR of 80 w/ nasal flaring.

\*Prenatal dx?  $L/S < 2$ , give antenatal betamethasone

\*Pathophys? Surfactant def, can't keep alveoli open.

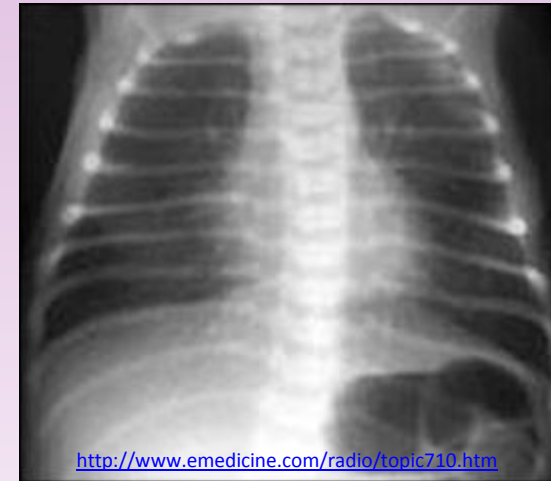
\*Tx? O<sub>2</sub> therapy with nasal CPAP to keep alveoli open



38 wk LGA infant born by C/S to an A2GDM has dyspnea/grunting TTN

\*Pathophys? Lung fluid not squeezed out, retained

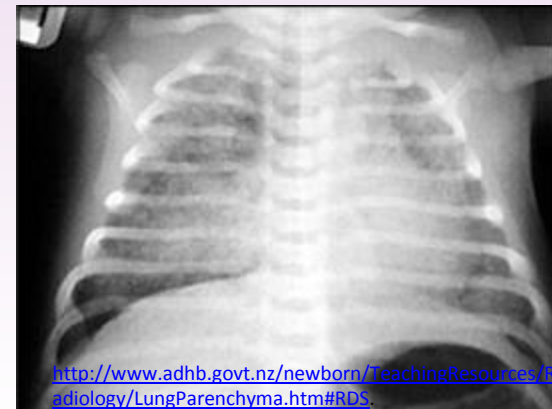
\*Prognosis? Usually minimal O<sub>2</sub> needed. Self-resolves in hours to days.



41 wk AGA infant was born Meconium aspiration syndrome  
after ROM yielded greenish-brown fluid.

\*Next best step? Intubate & suction before stimulation

\*Complications? Pulmonary artery HTN, pneumonitis



# GI disorders

- Defect lateral (usually R) of the midline, no sac. *\*will see high maternal AFP*
  - Assoc w/ other disorders? *Not usually.*
  - Complications? *May be atretic or necrotic req removal. Short gut syndrome*
- Defect in the midline. Covered by sac. *Assoc w/ Edwards & Patau Beckwith Wiedemann Syndrome = big baby w/ big tongue, ↓glc, ear pits*
  - Assoc w/ other disorders?
- Defect in the midline. No bowel present.
  - Assoc w/ other disorders? *Assoc w/ congenital hypothyroidism. (also big tongue)*
  - Treatment? *Repair not needed unless persists past age 2 or 3.*

## Gastroschisis



## Omphalocele



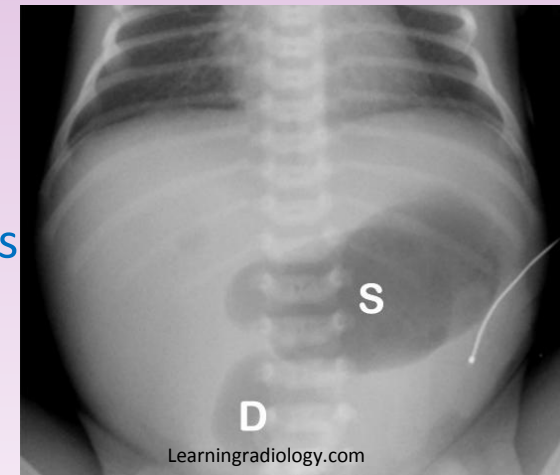
## Umbilical Hernia



# A vomiting baby

- 4wk old infant w/ non-bileous vomiting and palpable “olive”  
**Pyloric Stenosis**
  - Metabolic complications? **Hypochloremic, metabolic alkalosis**
  - Tx? **Immediate surg referral for myotomy**

- 2wk old infant w/ bileous vomiting. The pregnancy was complicated by polyhydramnios.  
**Intestinal Atresia**  
**Or Annular Pancreas**
  - Assoc w/? **Down Syndrome (esp duodenal)**



- 1 wk old baby w/ bileous vomiting, draws up his legs, has abd distension.  
**Malrotation and volvulus**
  - \*Ladd's bands can kink the duodenum
  - Pathophys? **Doesn't rotate 270 ccw around SMA**

# Pooping Problems

- A 3 day old newborn has still not passed meconium.

- DDX? (name 2)

**Meconium ileus**- consider CF if +FH

\*gastrograffin enema is dx & tx

**Hirschsprung's**- DRE → exposure of poo. bx showing no ganglia is gold standard

- A 5 day old former 33 weeker develops bloody diarrhea

**Necrotizing Enterocolitis**

- What do you see on xray?

**Pneumocystis intestinalis** (air in the wall)

- Treatment? **NPO, TPN** (if nec), antibiotics and resection of necrotic bowel

- Risk factors? **Premature gut**, introduction of feeds, formula.

- A 2mo old baby has colicky abd pain and current jelly stool w/ a sausage shaped mass in the RUQ.

**Intussusception**

\*Barium enema is dx and tx



# GU disorders

- Newborn male with no palpable testes.  
**Cryptorchidism**  
Assoc w/ prune belly syndrome
  - Where are they usually? **Inguinal canal**
  - Next best test? **Ultrasound if not palpable**
  - When to do surgery? **If not descended by 1yr to avoid sterility/cancer**
- Newborn male with urethral opening on the ventral surface.  
**Hypospadias**
  - What do you NOT do? **Circumcise! Foreskin is used in eventual repair.**
- Newborn child with ambiguous genitalia. One month later has vomiting & ↓Na ↑K and acidosis.  
**Congenital Adrenal Hyperplasia**
  - MC Cause? **21 Hydroxylase deficiency. (autosomal recessive)**
  - Definitive test? **17-OH progesterone before and after ACTH bolus**
  - Tx? **Hydrocortisone and fludrocortisone (↑ doses in times of stress)**

# Infants of Diabetic Mothers

- Mothers with pre-existing diabetes (esp type 1)
  - Control glc in the 1<sup>st</sup> trimester & take 4mg folate/day
  - Placental insufficiency/IUGR, Congenital heart dz, NTD, Caudal regression syndrome, Small left colon syndrome
- Mothers with gestational diabetes
  - **LGA.** Complications? ↑risk of birth trauma (clavicle, Erbs), C/S & TTN
  - **Hypoglycemia.** Why? Maternal hyperglycemia → fetal hyperinsulinemia
    - Complications? Neonatal seizure (always check glc!)
    - Treatment? Feed frequently if <40. IV dextrose if <20
  - **Hypocalcemia.** Complications? Neonatal seizure (always check Ca!)
  - **Polycythemia.** Why? Big baby needs more O<sub>2</sub>, hypoxia → ↑EPO
    - Complications? Renal or splenic vein thromboses
  - **Jaundice.** Why? More RBCs to break down. Risk for kernicterus
  - **RDS.** Why? ↑insulin interferes w/ cortisol surge prior to birth that normally stimulates lung maturation. Check L/S ratio >2

# Neonatal Fever Work up

- If a baby < 28 days has a fever >100.4 = sepsis until proven otherwise.
- Sxs might include *irritability, poor feeding*.
- What tests do you order?

CBC w/ diff, CXR, blood cultures, urine cultures (use catheter), LP

- Risk factors for neonatal sepsis?

Prematurity, chorioamnionitis, intrapartum fever, maternal leukocytosis, prolonged rupture of membranes (>18hrs), GBS+ mom.

- Most common bugs?

Group B Strep, E. Coli, Lysteria monocytogenes.

- Empiric treatment? Amp + gent until 48hr cx are negative.  
Cefotaxime + Amp if meningitis suspected

# TORCH infections

- Maculopapular rash on palms and soles, snuffles, periostitis. Syphilis. Tx w/ PCN
- Hydrocephalus, intracranial calcifications and chorioretinitis. Toxoplasmosis. Tx w/ sulfadiazine + leucovorin.
- Cataracts, deafness and heart defects (esp PDA, VSD), extramedullary hematopoiesis. Rubella. No tx.
- Microcephaly, periventricular calcifications, deafness, thrombocytopenia and petechiae. CMV. Tx w/ ganciclovir, but won't prevent MR
- Limb hypoplasia, cutaneous scars, cataracts, chorioretinitis, cortical atrophy. Congenital Varicella if mom infected 1<sup>st</sup> or 2<sup>nd</sup> trimester. If mom is exposed 5 days before – 2 days after delivery, baby gets VZIG.

# Neonatal conjunctivitis

- DOL 1-3, red conjunctiva and tearing.

**Chemical conjunctivitis** caused by silver nitrate drops. Not common anymore b/c we use erythromycin.

- DOL 3-5, bilateral purulent conjunctivitis can cause corneal ulceration.

**Gonococcal conjunctivitis** tx w/ topical erythromycin and IV 3<sup>rd</sup> gen ceph.



- DOL 7-14, red conjunctiva w/ mucoid discharge & lid swelling

**Chlamydia conjunctivitis** tx w/ oral erythromycin. Complication is chlamydial pneumonia → cough, nasal drainage, scattered crackles + bilat infiltrates on CXR

# Genetic Diseases & Syndromes



A newborn baby has decreased tone, oblique palpebral fissures, a simian crease, big tongue, white spots on his iris

### Down's Syndrome

- What can you tell his mother about his expected IQ?  
He will likely have moderate MR. Speech, gross and fine motor skill delay
- Common medical complications?
  - Heart? VSD, endocardial cushion defects
  - GI? Hirschsprung's, intestinal atresia, imperforate anus, annular pancreas
  - Endocrine? Hypothyroidism
  - Msk? Atlanto-axial instability
  - Neuro? Incr risk of Alzheimer's by 30-35. (APP is on Chr21)
  - Cancer? 10x increased risk of ALL

- Omphalocele, rocker-bottom feet/hammer toe, microcephaly and clenched hand, multiple others.
- Holoprosencephaly, severe mental retardation and microcephaly, cleft lip/palate, multiple others.
- 14 year old girl with no breast development, short stature and high FSH.

Edward's syndrome  
(Trisomy 18)

Patau's syndrome  
(Trisomy 13)

Turner's syndrome.  
XO. MC genotype of  
aborted fetuses

- Assoc anomalies? Horseshoe kidney, coarctation of aorta, bicuspid aortic valve
  - Tx? Estrogen replacement for secondary sex char, and avoid osteoporosis
- 18 year old tall, lanky boy with mild MR has gynecomastia and hypogonadism. \*increased risk for gonadal malignancy\*

Klinefelter's  
syndrome

- Café-au-lait spots, seizures large head. Autosomal dominant
- Mandibular hypoplasia, glossoptosis, cleft soft palate. W/ FAS or Edwards.
- Broad, square face, short stature, self-injurious behavior. Deletion on Chr17
- Hypotonia, hypogonadism, hyperphagia, skin picking, aggression. Deletion on paternal Chr15.
- Seizures, strabismus, sociable w/ episodic laughter. Deletion on maternal Chr15.
- Elfin-appearance, friendly, increased empathy and verbal reasoning ability. Deletion on Chr7.

Neurofibromatosis

Pierre Robin Sequence

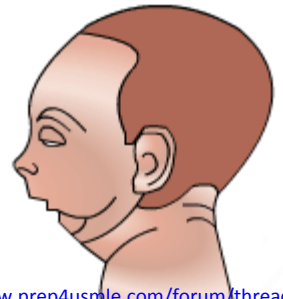
Smith Magenis

Prader-Willi

Angelman

Williams

Bird Facies



[www.prep4usmile.com/forum/thread/938](http://www.prep4usmile.com/forum/thread/938)



[beehive.thisishull.co.uk/default.asp?WCI=Disp](http://beehive.thisishull.co.uk/default.asp?WCI=Disp)

- IUGR, hypertonia, distinctive facies, limb malformation, self-injurious behavior, hyperactive.
- Microcephaly, smooth philtrum, thin upper lip, ADHD-like behavior. Most common cause of mental retardation.
- Most common type of MR in boys, CGG repeats on the X-chr w/ anticipation. Macrocephaly, macroorchidism, large ears.
- Autosomal dominant, or assoc w/ advanced paternal age. Short palpebral fissures, white forelock and deafness.

Cornelia de Lange



Fetal Alcohol Syndrome

Fragile X Syndrome

Waardenburg Syndrome

# Immune Deficiency

- 2 y/o M w/ multiple ear infxns, diarrheal episodes & pneumonias. No tonsils seen on exam.
  - Labs? **Absence of B cells on flow cytometry, low levels of all Igs**

Bruton agammaglobulinemia  
-x-linked  
-infx start @ 6-9mo (why?)
- 17 y/o F with decreased levels of IgG, IgM, IgE, and IgA but normal numbers of B cells.
  - Complication? **Increased lymphoid tissue → increased risk for lymphoma**

Combined variable immune deficiency. (acquired)
- Most common B-cell defect. **Selective IgA deficiency**  
Recurrent URIs, diarrhea.
  - Complication? **Anaphylaxis reaction if given blood containing IgA**
- 3wk old M with seizure, truncus arteriosus, micrognathia.
  - Genetic defect? **Microdeletion on Chr22**
  - What types of infxns in childhood? **Candida, viruses, PCP pneumonia**

DiGeorge Syndrome

- Infant w/ severe infxns, no thymus or tonsils. Severe lymphopenia.
  - Inheritance? SCID. See infxns w/ bacterial, viral and opportunistic bugs.
  - Tx? MC is XLR. AR is an ADA deficiency  
Pediatric emergency! Need bone marrow transplant by age 1 or death.
- 3 y/o M child w/ recurrent swollen, infected lymph nodes in groin and staph aureus skin abscesses.
  - How to diagnose? Chronic granulomatous disease XLR. PMNs can ingest but not kill catalase + bugs.  
Nitroterazolium blue (yellow means they have the dz). New test is Flow cytometry w/ DHR-123
- 18mo M baby w/ severe eczema, petechiae, and recurrent ear infxns.
  - Ig make up? Wiskott-Aldrich Syndrome. Often present w/ prolonged bleeding after circumcision.  
Low IgM, high IgA and IgE, slightly low IgG.



# Growth and Development

# Growth & Nutrition

- Newborns lose 10% of birth weight in 1<sup>st</sup> week. Why? Diuresis of extravascular fluid
- Should regain birth weight by? 2 weeks
- Should double weight by? 6 months
- Should triple weight by? 1 year
- Increased 50% of length by? 1 year
- Double length by? 5 years
- Breast milk is best for babies. True. Duh.  
True or false?
- Contraindications to breast-feeding. Galactosemia, PKU, HIV, HSV on the breast, chemo, Li, Iodide, alcohol.
- Breast milk vs. Formula- Breast milk is whey dominant, more lactose, more LCFA, less Fe but its better absorbed.

# Abnormal Growth

- 14 y/o boy, always been below 5% in height. Parents are tall & were “late bloomers”.
- Same story, but father is 5’2” and mom is 4’10”.
- 14 y/o boy, 50% in height, 97% for weight.
- Other causes of same bone age findings?
- 14 y/o boy, starts out in 50% for height, in the past 2 years is now between the 5%-10%.

## **Constitutional Growth Delay**

Bone age < Real age.

Child is likely to have normal final adult height.

## **Familial Short Stature**

Bone age = Real age.

## **Obesity**

Bone age > Real age.

Precocious puberty, CAH,  
Hyperthyroidism

## **Pathologic Short Stature**

Consider craniopharyngioma (vision problems, chest CT),  
Hypothyroidism (check TFTs),  
Hypopituitarism (check IgF1),  
Turners (check karyotype).

# Primitive Reflexes

- When head is extended, arms and legs both flex.
- When you place your finger in palm, flexes hand.
- Rub cheek, head turns to that side.
- When stimulate dorsum of foot, steps up.
- When neck is turned to one side, opposite arm flexes and ipsilateral arm extends
- When a fall is simulated, arms are extended.
- CNS origin of these reflexes?

Moro.

From birth – 4/6mo

Grasp.

From birth – 4/6mo

Rooting.

From birth – 4/6mo

Placing.

From birth – 4/6mo

Tonic neck.

From birth – 4/6mo

Parachute.

**From 6-8mo – por vida**

Brainstem and vestibular nuclei



# Developmental Milestones

- Roll over? **6mo.** Also, sits w/ support, creep/crawl, stranger anxiety.
- Skips & copies a triangle? **60mo.** Also draws a person w/ 8-10 parts.
- Walk alone? **15mo.** Also, builds 3 cube tower and scribbles w/ crayon.
- Walk upstairs w/ alternating feet? **30mo.** Also, stands on 1 foot, knows name, refers to self as “I”.
- Copy cross and square? **48mo.** Also, hops on 1 foot, throws ball overhead, group play and goes to toilet alone.
- Sit unsupported + Pincer grasp? **9mo.** Also, walks w/ hand held, object permanence, peak-a-boo & bye-bye
- Walks downstairs, copies a circle and can jump with both feet. **36mo.** Also, knows age and sex. Understands taking turns. Counts to 3.
- ½ of speech is comprehensible & 2-3word sentences? **24mo.** Also, runs well, builds 7 cube tower, holds spoon, helps undress.
- Social smile, start to coo? **2mo.** Also, sustains head in plane of body, follows an object 180deg, some vowel sounds

# Potty Training

- Urinary continence should be attained by: 5 years
- Primary if continence never achieved, Secondary if after a 6mo period of dryness.
- Medical causes to r/o? UTI (do a UA), constipation (disimpact) or Diabetes (check sugar)
- Tx of Enuresis? 1<sup>st</sup>- behavioral- reward system, pee before bed, bell-alarm pad.  
2<sup>nd</sup>- pharmacological- DDAVP or imipramine
- Fecal continence should be attained by: 4 years
- Most common cause? Constipation, fecal retention.
- Treatment? Disimpact, stool softeners, high fiber diet
- Behavioral modification? Post-prandial toilet sitting.



# Immunizations

- Due at birth? HepB (remember to give HepBIV if mom is HbsAg +)
- Due at 2mo, 4mo and 6mo? HepB, Rota, Dtap, HiB, PCV and IPV
- Starting at 6mo and then yearly? Influenza
  - Contraindications to flu vac? Egg allergy, also CI for yellow fever vac
- Due at 12mo? MMR, varicella, HepA (live vaccines not for kiddos < 12mo)
  - Contraindications to MMR? Neomycin or streptomycin allergy
- Due before age 2? Dtap and 2<sup>nd</sup> HepA (6mo after the 1<sup>st</sup> one)
- Due before kindergarden? Last IPV, Dtap, MMR and varicella
- Due at age 12? Tdap booster, meningococcal vaccine, and HPV (girls)

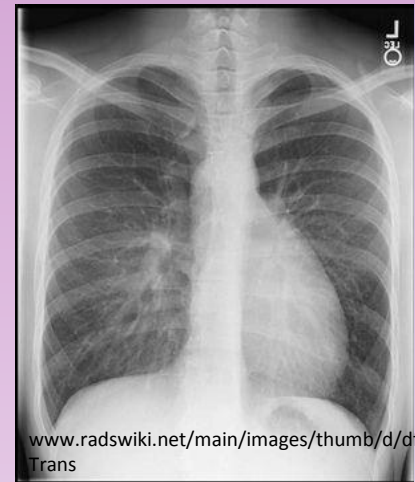
# Heart Disease

# Benign Murmurs

- Not all murmurs are a cause for alarm, >30% of kiddos have them.
- Good characteristics =
  - **Stills murmur**- SYSTOLIC, <II/VI, soft, vibratory and musical, heard best @ lower mid-sternum
  - **Venous hum**- best heard in anterior neck, disappears when jugular vein is compressed.
- Never normal =
  - Anything DIASTOLIC.
  - Anything >II/VI
  - Get an echo

## Transposition of the Great Arteries

- Newborn is cyanotic @ birth, O2 does not improve.
  - Most common in? **Infants of diabetic mothers**
  - Associated murmur? **NONE!** (unless PDA or VSD)
  - Immediate tx? **PGE1 to keep PDA patent**



- 2y/o child who gets cyanotic and hypernea while playing, squats down.
  - Associated murmur? **Harsh SEM + single S2**
  - Treatment? **O2 and knee-chest position, surgical correction.**

## Tetralogy of Fallot

VSD + RA hypertrophy + overriding aorta, pulmonary stenosis



- Bipolar woman gives birth to a child w/ holosystolic murmur worse on inspiration.

## Ebstein Anomaly-

Tricuspid insufficiency 2/2 TV displacement into RV.

- Associated arrhythmia? **Wolff-Parkinson-White**

- Cyanosis @ birth with holosystolic murmur, depends on VSD or ASD for life. EKG shows LVH.

- Heart defect associated with DiGeorge syndrome. CXR shows ↑pulm blood flow and bi-ventricular hypertrophy.

- #1 congenital heart lesion. Harsh holosystolic murmur over LL sternal border, loud P2.

- If II/VI in a 2mo old? If no sx's, continue to monitor. Most close by 1-2yr
- Gold standard dx test? Echo
- When is surgery indicated? FTT, 6-12mo w/ pHTN, >2yrs w/ Qp/Qs >2:1
- Is louder better or worse? Better. It means the defect is small. Most often membranous. More likely to spontaneously close.

### Tricuspid atresia.

Give PGE1 until surgery

### Truncus arteriosus.

Eisenmenger develops early. Do surg in 1<sup>st</sup> few weeks of life

### Ventriculoseptal defect.

- Loud S1 w/ fixed and split S2. Older child w/ exercise intolerance.

## ASD

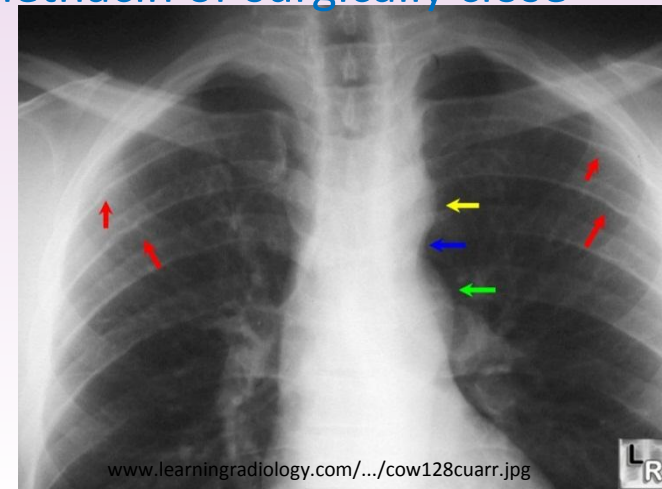
- Most common defect in Down Syndrome baby. Fixed & split S2 + SEM w/ diastolic rumble.

## Endocardial Cushion Defect

- Tx? @ risk for early Eisenmengers. Surgery before pHTN @ 6-12mo.
- Continuous machine-like murmur w/ bounding pulses and wide pulse pressure.
  - Associations? Prematurity, congenital rubella syndrome
  - Treatment? If not closed by 1wk, give indomethacin or surgically close
- Most common defect in Turner's baby. Decreased femoral pulses, "reverse 3 sign", "notching" @ inf rib border 2/2 incr collateral. May see asymmetry in arm BPs

## PDA

## Coarctation of the Aorta



# Other cardiac diseases:

- 15 year old athlete complains of occasional palpitations, angina and dizziness. Last week he fainted during the 1<sup>st</sup> inning of his baseball game. **HOCM**
  - Murmur? **SEM, better w/ ↑ preload (squat, handgrip) louder w/ valsalva, standing, exercise (↓preload)**
  - Treatment of this child? **Beta blockers or CCB (no diuretics or dig- why?)  
Alcohol ablation or surgical myotomy**
  - Restrictions? **No sports or heavy exercise!**
- 7 year old girl presents with vague chest pain, pain in several different joints over the past few days, and a rash. Her ESR is elevated, and her EKG shows prolongation of the PR interval. **Acute Rheumatic Fever**
  - Treatment? **Oral PCN (erythromycin) for 10 days, then prophylactic till 20**
  - Complications? **Mitral stenosis, (then aortic or tricuspid involvement)**

# Respiratory Disease



# Cystic Fibrosis

- Signs at birth? Meconium ileus = dilated loops, “ground glass”, dx/tx with gastrograffin enema  
Can also see rectal prolapse from chronic diarrhea.
- In early childhood, suspect it when: failure to thrive (<5<sup>th</sup> % weight & height), foul-smelling, bulky, floating stools, recurrent respiratory infections and nasal polyps.
- Genetic Defect & Inheritance? AR, mutation on Chr7, CFTR protein.
- Diagnosis? Sweat test → >60mEq/L chloride is diagnostic
- Treatment?
  - For thick resp. secretions? DNase (mucolytic), albuterol/saline nebs
  - For pneumonia? Most often pseudomonas or colonized w/ b. cepacia  
Tx w/ piperacillin + tobramycin or ceftazidime
  - For pancreatic insuff? Enzy replacement w/ meals + ADEK supplement
  - For electrolyte loss through skin? Adequate fluid replacement when exercising or when hot.

# Asthma

- If pt has sxs twice a week and PFTs are normal? *Albuterol only*
- If pt has sxs 4x a week, night cough 2x a month and PFTs are normal? *Albuterol + inhaled CS*
- If pt has sxs daily, night cough 2x a week and FEV1 is 60-80%? *Albuterol + inhaled CS + long-acting beta-ag (salmeterol)*
- If pt has sxs daily, night cough 4x a week and FEV1 is <60%? *Albuterol + inhaled CS + salmeterol + montelukast and oral steroids*
- Exacerbation → tx w/ inhaled albuterol and PO/IV steroids. *Watch peak flow rates and blood gas. PCO2 should be low. Normalizing PCO2 means impending respiratory failure → INTUBATE.*
- Complications → *Allergic Brochopulmonary Aspergillus*

# Endocrine

# Diabetes

A 12 y/o girl presents with a 2 day history of vomiting. For the last 4 weeks, she noticed weight loss, polyphagia, polydipsia and polyuria.

Na = 130, Cl = 90, HCO<sub>3</sub> = 15, glucose = 436.

- Next best step? Start insulin drip + IVF. Monitor BGL and anion gap.  
Start K. Bridge w/ glargine once tolerating PO.
- Pathophysys? T-cell mediated destruction of islet cells, insulin autoAb, glutamic acid decarboxylase autoAb
- Long term treatment? Will need insulin tx.
- Diagnostic criteria for diabetes? Fasting glc >125 (twice)  
2hr OGTT (75g) > 200  
Any glc > 200 + symptoms

# Renal Disease

# A kiddo is peeing blood...

- Best 1<sup>st</sup> test? Urinalysis
- Dysmorphic RBCs or RBC casts? Glomerular source
- Definition of nephritic syndrome? Proteinuria (but <2g/24hrs), hematuria, edema and azotemia
- 1-2 days after runny nose, sore throat & cough? Berger's Dz (IgA nephropathy). MC cause.
- 1-2 weeks after sore throat or skin infxn? Post-strep GN- smoky/cola urine, best 1<sup>st</sup> test is ASO titer. Subepithelial IgG humps
- Hematuria + Hemoptysis? Goodpasture's Syndrome. Abs to collagen IV
- Hematuria + Deafness? Alport Syndrome. XLR mutation in collagen IV

# Kidney Stones

- Flank pain radiating to groin + hematuria.
- Best test? CT.
- Types-
  - Most common type? Calcium Oxalate. Tx w/ HCTZ
  - Kid w/ family hx of stones? Cysteine. Can't resorb certain AA.
  - Chronic indwelling foley and alkaline pee? Mg/Al/PO4 = struvite. proteus, staph, pseudomonas, klebsiella
  - If leukemia being treated w/ chemo? \*Uric Acid  
Tx by alkalinizing the urine + hydration
  - If s/p bowel resection for volvulus? Pure oxylate stone. Ca not reabsorbed by gut (pooped out)
- Treatment
  - Stones <5mm Will pass spontaneously. Just hydrate
  - Stones >2cm Open or endoscopic surgical removal
  - Stones 5mm-2cm Extracorporeal shock wave lithotripsy

# A kiddo is peeing protein...

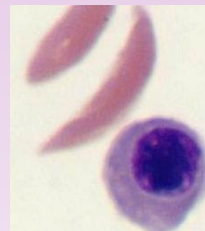
- Best 1<sup>st</sup> test? Repeat test in 2 weeks, then quantify w/ 24hr urine
- Definition of nephrotic syndrome? >3.5g protein/24hrs, hypoalbuminemia, edema, hyperlipidemia (fatty/waxy casts)
- MC in kiddos?
  - Minimal change dz- fusion of foot processes
  - Treat with prednisone for 4-6wks.
  - Most common complication is infection-
    - Make sure immunized against pneumococcus and varicella.
- If nephrotic patient suddenly develops flank pain? Suspect renal vein thrombosis! 2/2 peeing out ATIII, protein C and S. Do CT or U/S stat!
- Other random causes? Orthostatic = MC in school aged kids. Normal while supine, increased when standing.



Heme-Onc

# An African American (or Mediterranean) kid w/ sickle cell disease comes in...

- Swollen, painful hands and feet. **Dactylitis. 2/2 necrosis of small bones**
- Excruciating pain in the extremities, ulcers, hip pain. **Pain crises. Ischemic damage 2/2 sickling.**
- Point tenderness on femur, fever, and malaise. **Osteomyelitis. MC bug is salmonella**
- Things seen on blood smear?  
**↑retics, nl MCV sickles, targets, HJ bodies.**
- Acute drop in HCT with ↓↓retics? **Aplastic crisis. Parvo B19**
- Recurrent RUQ pain after meals. **Pigment gallstones. Do Chole.**
- Respiratory distress & emergent tonsilectomy? **Waldyer Ring hyperplasia.**
- Proteinuria and increased creatinine + recurrent UTIs? **Kidney infarcts due to sickled RBCs**



<http://emedicine.medscape.com/article/954598-media>

# More Sickle Cell Pearls

- Most common cause of sepsis? **Strep Pneumo**
- Presents w/ fever, cough, chest pain, chills, and SOB? **Acute Chest syndrome. Pulmonary infarction. MC cause of death.**
  - Tx? **O2, abx and exchange transfusion.**
- Acute confusion and focal neurologic deficits? **Stroke**
  - Tx? **Exchange transfusion (NOT tPA!)**
  - Assessing risk? **Transcranial doppler (v <200cm/sec), keep HbS <30%**
- Vaccination and prophylaxis?  
**23-valent pneumococcal vaccine @ age 2 + H. flu and N. meningitides.**  
**PCN prophylaxis from age 2mo until age 6yrs.**
- If a patient presents w/ fatigue and megaloblastic anemia? **Most likely folate deficiency.**  
**Has higher need 2/2 ↑retics**
- Treatment? **Hydroxyurea incr production of HbF**  
**Tx infx aggressively and manage pain.**  
**Bone marrow transplant cures, but has 10% post op mortality**

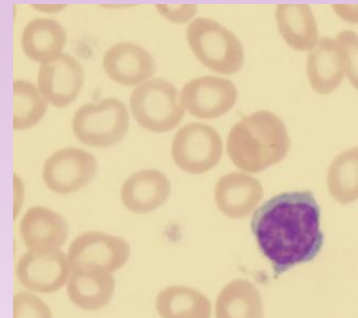
# Kids with Anemia

- When is anemia not a big deal?

Physiologic drop in H&H for 1<sup>st</sup> 2-3mo. Transient Erythroblastopenia occurs later (3mo-6yrs) = immune suppression after viral infxn (not B19)

- 18 mo kiddo, picky eater, drinks lots of cow's milk.

↓H&H, MCV 75, ↓ferritin, ↑TIBC

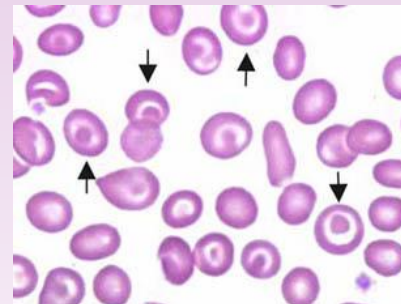


**Fe-deficiency.**

Tx w/ oral ferrous salts.

- 18 mo kiddo, eats a varied diet. Mom is Italian.

↓H&H, MCV 60, ↓RDW

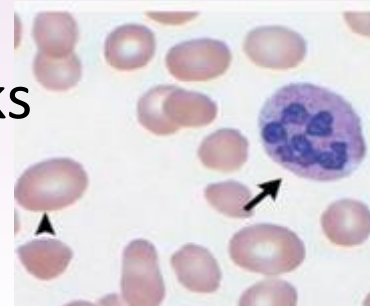


**Thalassemia.**

Varying degrees. Tx w/ transfusion & deferoxamine.

Can see expanded medullary space

- 8 mo kiddo is irritable, has glossitis & FTT. Picky eater, drinks lots of goat's milk.



**Folate-deficiency.**

See low serum (RBC) folate. Tx w/ daily folate.

- 4mo pale baby, normal plts, WBCs but hemoglobin is 4. Incr RBC ADA and low retics. Triphalangeal thumbs.

Blackfan-Diamond  
Anemia

- Tx? Corticosteroids, transfusions, stem cell transplant.

- 18mo baby presents w/ low plts, low WBCs and profound anemia. He has café-au-lait spots, microcephaly, and absent thumbs.

Fanconi Anemia

- Dx? Bone marrow shows hypoplasia, Cytogenetic studies for chr breaks

- Tx? Corticosteroids, androgens, bone marrow transplant.

- Complications? Incr risk for AML and other cancers.

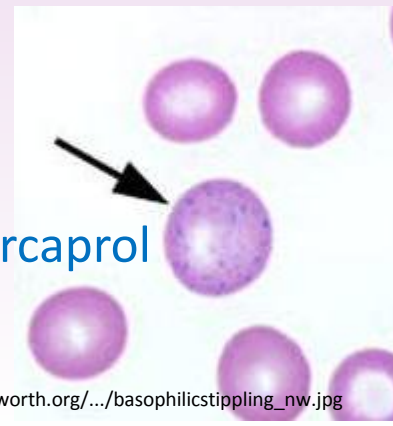
- 2 y/o baby presents w/ hyperactivity, impaired growth, abdominal pain and constipation.

Lead Poisoning

- Dx? Venous blood sample, check lead level

- Tx? >45- tx w/ succimer. >70- admit and tx w/ EDTA + dimercaprol

- Screening? Test blood lead levels btwn 12-24 mo if low SES, live in old house (<1960).



# A kiddo walks in with thrombocytopenia

- 15 y/o F recurrent epistaxis, heavy menses & petechiae. ↓ plts only. **ITP.** Tx w/ IVIG for 1-2 days, then prednisone, then splenectomy. NO plts!!
- 15 y/o F recurrent epistaxis, heavy menses, petechiae, normal plts, ↑ bleeding time and PTT. **VWD.** DDAVP for bleeding or pre-op. Replace factor VIII (contains vWF) if bleeding continues.
- 7 y/o M recurrent bruising, hematuria, & hemarthroses, ↑ PTT that corrected w/ mixing studies\*. **Hemophilia.** If mild, tx w/ DDAVP, otherwise, replace factors.
- 1wk old newborn, born at home, comes in with bleeding from the umbilical stump & bleeding diathesis **VitK def.** ↓ II, VII, IX and X. Same in CF kid with malabsorption  
Tx w/ FFP acutely + vitK shot
- 9 y/o F with Wilson's disease developed fulminant liver disease.
  - 1<sup>st</sup> factor depleted? **VII**, so PT increases 1st
  - 2 factors not depleted? **VIII and vWF** b/c they are made by endothelial cells.

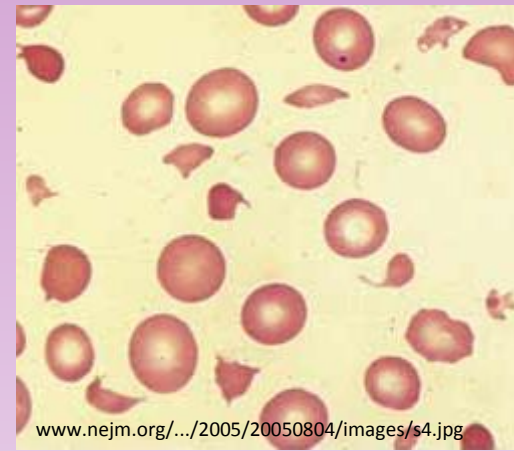
A 3 y/o child is brought in with petechiae, abdominal pain, vomiting and lethargy. He had bloody diarrhea 5 days ago after eating hamburgers at a family picnic.

Labs reveal thrombocytopenia and ↑creatinine

### Hemolytic Uremic Syndrome

Most common cause? \*E. Coli O157H7\*, Shigella, Salmonella, Campylobacter

Treatment? NO platelets! Tx w/ aggressive nutrition (TPN) and early peritoneal dialysis. Don't give abx for bloody diarrhea. Can ↑ risk of HUS



A 5 y/o child is brought in with purpura on his legs and buttocks, abdominal pain, joint pain, current jelly stool. His smear appears normal, as are his coagulation studies and electrolytes. IgA and C3 are deposited in the skin.

### Henoch Schonlein Purpura

Most common cause? Usually follows a URI

Treatment? Symptomatic treatment. Can use steroids for GI or renal dz.



- New onset seizure, ataxia and HA worse in the AM with vomiting for a month. Brain tumor. Most likely infratentorial
  - Most common- Pilocytic astrocytoma of cerebellum. Resect. ~90% survive.
  - 2<sup>nd</sup> most common, worse prognosis- Medulloblastoma. Vermis, obstruct 4<sup>th</sup> V
- Adolescent with height in 5<sup>th</sup> %, w/ bitemporal hemianopsia. See calcifications in sella turcica. Craniopharyngioma. Suprasellar  
A remnant of Rathke's pouch.
- 2 year old hypertensive child with asymptomatic abdominal mass. Wilm's tumor
  - Associations? Aniridia, GU anoms, Hemihypertrophy, Beckwith-Weidemann
  - Best test? Abdominal CT. Do CXR to check lung involvement
  - Treatment? Surgery, chemo, rads
- 4 year old with jerking movements of eyes and legs, bluish skin nodules and an tender abdominal mass. Neuroblastoma
  - Diagnostic tests? ↑urine homovanillic or vanillylmandelic acid.

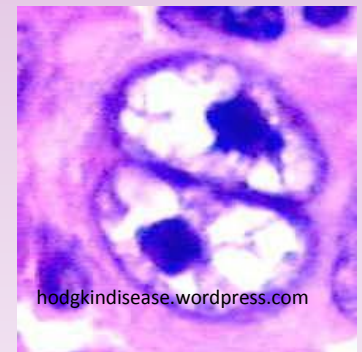


- 3 year old girl w/ a limp & left leg pain, T = 99.9, HSM, petechiae, & pallor. Cells are shown. Cells are CALLA and TdT +.
  - Best test? Bone marrow biopsy → >30% lymphoblasts
  - Treatment? VDP + CNS tx w/ intrathecal methotrexate
  - Poor prognostic factors? <1 or >10, >100K WBC
- 14 y/o boy w/ enlarged, painless, rubbery nodes, drenching fevers, and 10% weight loss.
  - Best test? Excisional biopsy.
  - And then? Staging CT or laparoscopy. (determines tx)
  - Treatment? Chemo + Rads. 90% cure if stage I or II
- 7 year old girl with non-productive cough and large anterior mediastinal mass on CXR.
  - Best test? Biopsy of mass, bone marrow bx for staging
  - Treatment? Surgical excision if abdominal tumor. Can use anti-CD20 if B-cell tumor.  
Rads for some.

ALL



Hodgkin Lymphoma

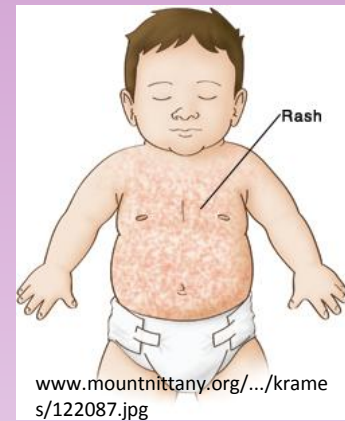


Non-Hodgkin Lymphoma

# Infectious Disease

- 2y/o w/ a fever to 105, 3 days later gets a pink, mac-pap rash on trunk arms and legs.

Roseola-  
HHV6



- 2y/o w/ a low grade fever, lacy reticular rash on cheeks and upper body (spares the palms/soles)

5<sup>th</sup> Disease/Erythema  
Infectiosum-  
Parvovirus B19



- Who is this bad for? Preggos, sickle cell, thalessemia

- Fine, mac-pap desquamating rash begins on chest and spreads to neck, trunk, & extremities+ strawberry tongue. Sore throat 1-2wks prior.

Scarlett  
Fever  
(group A  
strep)



- Treatment PCN prevents rheumatic fever. (won't help reduce changes of APSGN)

- Cough, runny nose, fever → macular rash begins behind ears & spreads down. Gray spots on the buccal mucosa.
  - Tx? **Vitamin A + supportive care**
- Sore throat, joint pain fever → pinpoint rash on the face and spreads down. Rose spots on the palate.
  - Complications? **Congenital rubella syndrome**
- Baby with poor feeding. Vesicles in the mouth on palms and soles + rash on buttocks.
  - **Hand-Foot-and-Mouth Disease (Coxsackie virus A16)**
- 16 year old M with swollen parotid glands, fever & HA.
  - Complications? **Orchiditis and sterility**

**Measles**  
(paramyxovirus)



**Rubella**  
(paramyxovirus)



**Mumps**  
(paramyxovirus)

- 6y/o kid from central PA, went camping. Had fever. **Lyme Disease.**  
**Borrelia burgorferi**
  - Complications? **Arthritis, heart block, meningitis, Bells**
  - Treatment? **Amox for this kid. Doxy if >8.**



- 6y/o kid from coastal NC, went camping. Had fever, myalgias, abd pain.

**Rocky Mountain Spotted fever.**  
**Rickettsia rickettsii**

- Complications? **Vasculitis and gangrene**
  - Treatment? **Doxy no matter what age**
- 8y/o kid, multiple excoriations on arms. Itchy at night. **Scabies!**
  - Treatment? **5% permethrin for whole family!**



- Honey-colored crusted plaque on face. **Impetigo. MC bug is staph if bullus.**
  - Treatment? **Topical muciprocin if localized**
- Inflamed conjunctiva and multiple blisters. Nikolsky's +/- **Staph Scalded Skin Syndrome**  
**From exfoliative toxin**
  - Treatment? **Tx w/ IV ox or nafcillin**

# Meningitis

- Most Common bugs? Strep Pneumo, H. Influenza, N. meningitidis (tx w/ Ceftriaxone and Vanco)
- In young & immune suppressed? Add Lysteria. (tx w/ Ampicillin)
- In ppl w/ brain surg? Add Staph (tx w/ Vanco)
- Randoms? TB (RIPE + 'roids) and Lyme (IV ceftriazone)
- Best 1<sup>st</sup> step? Start empiric treatment (+ steroids if you think it is bacterial)
- Diagnostic test? Then, check CT if signs of increased ICP  
Then, do an LP:
- Roommate of the kid in the dorms who has bacterial meningitis and petechial rash? +Gram stain, >1000WBC is diagnostic.  
High protein and low glucose support bacterial  
Rifampin!!



# Ear Infections

- 2 y/o w/ fever to 102, tugging on his right ear. Patient's tympanic membrane is red and bulging. **Otitis Media**
  - Most sensitive dx test? **Limited mobility on insufflation or air-fluid level**
  - RF? **↓SES, Native Americans, formula fed, tobacco smoke, around kids**
  - Treatment? **Amox or azithromycin for 10days. If no improvement in 2-3 days, switch to amox-clav**
  - Complications? **Effusion-place tubes if bilat effusion >4mo or if bilateral hearing loss.**
- 12y/o in summer swim league has pain when adjusting his goggle straps behind his ear. Thick exudates coming from the ear and tender posterior auricular nodes. **Otitis Externa**
  - Treatment? **Topical ciprofloxacin**
  - Complications? **Malignant external otitis → can invade to temporal bone → facial paralysis, vertigo. Need CT and IV abx. May need surgery**

# Sore Throat

- 7y/o w/ exudative pharyngitis w/ tender cervical lymph nodes and fever to 102. Sounds like GABHS Pharyngitis
  - Best 1<sup>st</sup> test? Rapid strep antigen
  - If negative? If clinical suspicion ↑ (ie, no viral sxs) – do culture
  - Treatment? PCN or erythromycin. Why?\*
- A child presents w/ “muffled voice”, stridor and refuses to turn her head to the left. Retropharyngeal abscess
  - Treatment? I&D for C&S. GAS + anaerobes. 3<sup>rd</sup> gen ceph + amp or clinda
  - Complications? Retropharyngeal space communicates w/ mediastinum
- A child presents w/ “hot potato voice” and upon throat exam her uvula is deviated to the right 2/2 a bulge. Peritonsillar abscess
  - Treatment? Aspiration or I&D + abx, tonsillectomy if recurrent.
  - Indications for tonsillectomy? >5 episodes of strep/year for 2 years or >3 episodes/year for 3 years.



# Older kiddo with a sore throat...

- Other sxs = fever, fatigue, generalized adenopathy and splenomegaly (anterior and posterior cervical nodes).

Think Epstein-Barr virus

- What happens if you give them ampicillin or amoxicillin?

Maculopapular Rash (immune mediated vasculitic)

- Diagnosis? Blood smear shows lymphocytosis w/ atypical lymphs +Heterophile antibody (Monospot) test.

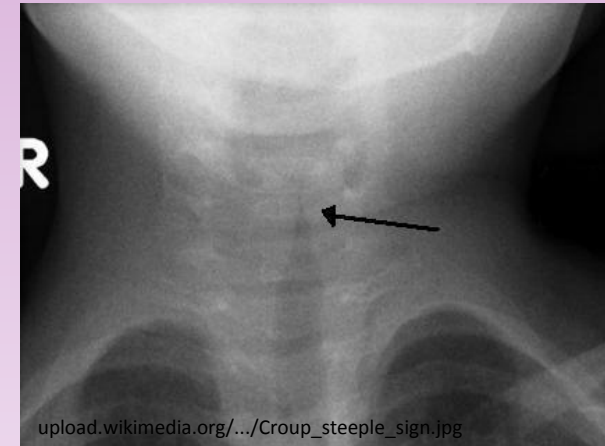
- Treatment? Rest and symptomatic therapy.

- Precautions? Splenic hemorrhage or rupture. (most in 2<sup>nd</sup> week)  
No contact sports until splenomegaly resolves.

# Respiratory Distress

- 1 y/o w/ fever to 100.5 & “barking” cough and loud noises on inspiration.
  - Most common bug? [Parainfluenza virus](#)
  - X-ray buzzword? [“steeple sign”](#)
  - Treatment? [Mist, epinephrine neb, steroids](#)
- 2 y/o w/ fever to 104 & drooling w/ intercostal retractions and tripod position.
  - Most common bug? [H. Flu B only in unimmunized](#)  
[Strep pyo, strep pneumo, staph](#)
  - X-ray buzzword? [“thumbprint sign”](#)
  - Next best step? [Go to OR and intubate](#)
  - Treatment? [Anti-staph abx + 3<sup>rd</sup> generation cephalosporin](#)

Croup



Epiglottitis



# Pneumonia

- Kid comes in w/ cough productive of yellow-green sputum, runny nose and T = 100.8. Lung exam only reveals some coarse rhonchi. **Acute Bronchitis**
  - Next best step? **Supportive tx w/ anti-pyretic, tussives, histamines.**
- Kid comes in w/ similar sxs but decrease breath sounds and crackles in the LLL and WBC = 16K. **Pneumonia**
  - Next best step? **CXR to confirm. Typical vs atypical.**
  - MC cause in neonates <28days? **GBS, E. Coli, Lysteria**
  - MC cause 1mo-3mo? **C. trachomatis, RSV, paraflu, strep pneumo**
  - Specific findings for chlamydia pneumonia? **Staccato cough, eosinophilia**
  - MC cause in kids 4mo- 5y/o? **VIRAL! (RSV) then s. pneumo**
  - MC cause in kids >5y/o? **Mycoplasma, s. pneumo**

# Kid with a cough

- 9mo infant w/ runny nose, wheezy cough, T = 101.5, and RR = 60. Retractions are visible and pulse ox is 91%.

## Bronchiolitis

- Most common bug? RSV. Confirm w/ swab
- CXR findings? Hyperinflation w/ patchy atelectasis
- Treatment? Hospitalize if respiratory distress. Albuterol nebs. NO steroids
- Who needs vaccine? Palivizumab for premies, CHD, lung dz, immune dz
- 9mo infant with severe coughing spells with loud inspiratory whoops and vomiting afterwards. 2 weeks ago she had runny nose and dry cough.

## Whooping Cough

- Responsible bug? Bordetella pertussis
- Lab findings? CBC shows lymphocytosis
- Treatment? Erythromycin for 14 days
- \*Family members and kids in her daycare? Erythromycin for 14 days

# UTI

- In neonates- sxs are vague- fever, dehydration, fussy.
  - If fever is present → its pyelo. Cystitis has NO fever
- Before age 1, boys are more likely than girls to get UTI.
- Anatomic risk factor for UTI? Vesicoureteral reflux. Need abx prophylaxis
- Diagnosis of UTI? Clean catch or cath sample, UA and Culture (>10K CFU)
  - Need ultrasound if: Any febrile UTI for anatomy, abscess or hydronephrosis
- Treatment of UTI? PO trim-sulfa or nitrofurantoin
- Treatment of pyelo? 14 days of IV ceftriaxone or amp & gent
- Follow up? Test of cure to confirm sterility
- Who needs VCUG? All males, females <5, any pyelo, females >5 w/ 2<sup>nd</sup> UTI
- Role of Tc-labeled DMSA scan? It is most sensitive and accurate study of scarring and renal size, but is not first line.

# Bone and Joint Issues

# Kid with a limp

- Most common cause overall? **Trauma**
- 18mo F w/ asymmetric gluteal folds on exam. **Developmental hip dysplasia**
  - RF? **1<sup>st</sup> born F, +FH, breech position**
  - Dx and Tx? **clunk on Barlow. U/S of hip if unsure. Tx w/Pavilk harness, surg**
- 5 y/o M initially w/ painless limp now has pain in his thigh. **Legg-Calve'-Perthes Disease.** (avascular necrosis).
- 5 y/o M initially w/ a cold 1wk ago now presents w/ a limp & effusion in the hip. X-rays are normal and ESR is 35 (↑), T = 99.8, WBCs = 10K. **Transient Synovitis**
  - Next best step? **\*Bed rest for 1 wk + NSAIDS**
- 14 y/o lanky M w/ nagging knee pain and decreased ROM of the hip on exam. **SCFE.** Remember they're not always fat!
  - Tx **Surgically close and pin the epiphysis to avoid osteonecrosis.**
- 14 y/o basketball player has knee pain and swelling of the tibial tubercle **Osgood-Schlatter.** Overuse injury from jumping

- 12 y/o F w/ 2 wk history of daily fevers to 102 and a salmon colored evanescent rash on her trunk, thighs and shoulders. Her left knee and right knee are swollen. **JRA**
  - Good Prognostic factor? **+ANA**
  - Bad Prognostic factor? **+RF, also polyarticular and older age @onset**
  - Treatment? **1<sup>st</sup> line = NSAIDs, 2<sup>nd</sup> line = methotrexate, 3<sup>rd</sup> = steroids**
- 2y/o F w/ a 2 wk history of daily fevers to 102 and a desquamating rash on the perineum. She has swollen hands and feet, conjunctivitis and unilateral swollen cervical lymph node. **Kawasaki**
  - Other lab findings? **↑plts (wk2-3), ↑ urine WBC, ↑LFTs, ↑CSF protein**
  - Best 1<sup>st</sup> test? **2D echo and EKG. Repeat the Echo after 2-3wks of tx**
  - Treatment? **Acute = IVIG + high dose aspirin. Then aspirin + warfarin**
  - Most serious sequelae? **Coronary artery aneurysm or MI**



# Bone Pain due to Cancer

- If  $<10$ , more likely. M>F.  
More common if hx of **Ewing Sarcoma**  
retinoblastoma or previous radiation. “Onion skinning” on xray. (layers of periosteal development).
  - Treatment? **Rads and/or surgery**
- If  $>10$ , more likely. M>F. See **Osteogenic sarcoma**  
“sunburst” and “Codman’s triangle” on xray.
  - Treatment? **Chemo and/or surgery**
- More diffuse bone pain in a patient w/ petechiae, pallor and increased infections



Don't forget bone pain can be presenting sx for leukemia

# Neurology

# Hydrocephalus

- Anytime you see a meningocele or myelomeningocele...  
Do a head CT looking for hydrocephalus. (Arnold Chiari II)
- Anytime you see an infant with a head circumference  $>95^{\text{th}}$  %...  
Consider hydrocephalus. Also bulging fontanelle,  $\uparrow$ DTRs, HA, vomiting.
- Noncommunicating- Stenosis of CA, tumor/malformation near 4<sup>th</sup> ventr
- Communicating- SAH, pneumococcal/TB meningitis, leukemia
- Infant with increasing head size, prominent occiput, cerebellar ataxia and delayed motor development.
  - Dx? Dandy-Walker malformation
  - What will you see on CT or MRI? Cystic expansion of 4<sup>th</sup> ventricle. Can see Agenesis of cerebellar vermis.

# Seizures

- This morning, a 1 y/o develops a fever to 102.4. Four hours later, the parents bring her in after she has a 3-4 minute tonic-clonic seizure. **Febrile Seizure**
  - Next best step? **Give acetamenophen. NO ↑risk for epilepsy**
- An 8 year old boy gets in trouble in school because he is always “staring into space”. These episodes last only seconds, have lip smacking, and he goes right about his business after they are done. **Absence Seizure**
  - Common EEG finding? **3Hz spike and wave discharge**
  - Best Tx? **Ethosuxamide or valproic acid**
- A 6mo old is brought in for multiple symmetric contraction episodes of neck, trunk and extremities that occur in spells.
  - Dx? **Infantile Spasms**
  - Common EEG finding? **Hypsarrhythmia = asynchronous, chaotic, bilat**
  - Best Tx? **ACTH. Prednisone is 2<sup>nd</sup> line.**

# Neurodegenerative Disorders

- 8y/o w/ difficulty w/ balance while walking, no DTRs, bilateral Babinski and “explosive, dysarthric speech”. Friedrich Ataxia  
AR, trinuc repeat
  - Most common cause of death? HOCM → CHF.
- 2y/o w/ gait disturbance, loss of intellectual fxn, nystagmus and optic atrophy. Cresyl violet → metachromatic staining. Metachromatic leukodystrophy  
AR
  - Pathophys? Deficiency of arylsulfatase A → accum cerebroside sulfate
- 12y/o w/ decreased school performance, behavior changes, ataxia, spasticity, hyperpigmentation, ↑K, ↓Na, acidosis. Adrenoleukodystrophy  
XLR
  - Prognosis? Death w/in 10 years
- 9mo who had previously been reaching milestones starts to lag. Seizures, hypotonia, cherry red macula. Tay-Sachs  
XLR
  - Pathophys? Def of hexosaminidase A → accum GM2

# Neuromuscular Disorders

- 3mo infant lays in the “frog-leg” position, <5<sup>th</sup>% 2/2 feeding difficulties, hypotonic, fasciculations of the tongue and absent DTRs.
  - Dx? SMA 1- Werdnig Hoffman Disease
  - Prognosis? Most die before age 2
- 6y/o is brought in 2/2 “clumsiness” and frequent falls. The lower leg has decreased muscle bulk and appears “stork-like”. There are multiple small injuries on the hands and feet. You notice pes cavus and claw hand.
  - Dx? Marie-Charcot-Tooth Disease
  - Tests? Decreased motor/sensory nerve vel, sural nerve bx. \*CPK is normal
  - Treatment? Stabilize ankles w/ surgical fusion. Usually normal lifespan and most remain ambulatory.