CHILD ABUSE MIMICS

Chan Lowe, MD, FAAP, SFHM
Division Chief, Pediatric Hospital Medicine
Associate Professor – Department of Pediatrics
The University of Arizona
Banner-Diamond Children’s Medical Center
Disclosures

- I have served as a paid expert witness in child abuse cases.
- I am an examiner at the Southern Arizona Children’s Advocacy Center
- I will not be discussing any unapproved or off-label treatments.
- Images are from the American Academy of Pediatrics Visual Diagnosis of Child Abuse for Medical Providers, 4th addition, unless otherwise specified.
Case 1

- 9 month old boy presents with bucket handle fracture of the left distal humerus. He is also found to have evidence of acute and healing posterior rib fractures. Bones appear otherwise normal.

- Head CT shows no bleeds, Ophtho eval shows no retinal hemorrhages.

- Labs show a low vitamin D level of 12 ng/ml (low). PTH is normal, ALP is slightly high. Serum calcium on the low side of normal.

- Diagnosis?
Rickets or just a rickety excuse?

- Vitamin D deficiency is extremely common.
  - *Estimates range between 1 and 78%*

- No definite consensus on levels for peds
  - *Generally accepted < 10 ng/ml = overt deficiency*
  - 11-20 ng/ml = deficiency
  - 21-30 ng/ml = insufficiency

- Manifests as low calcium, low phosphorus, high PTH, low vitamin D, elevated ALP
  - As *Rickets develops, serum calcium often returns to normal range due to increased PTH action.*
## Biochemical findings in rickets

<table>
<thead>
<tr>
<th>Increased</th>
<th>Decreased</th>
</tr>
</thead>
<tbody>
<tr>
<td>Parathyroid hormone</td>
<td>Calcium</td>
</tr>
<tr>
<td>Alkaline Phosphatase</td>
<td>Phosphate</td>
</tr>
<tr>
<td>Urinary phosphate levels</td>
<td>Calcidiol</td>
</tr>
<tr>
<td></td>
<td>Calcitriol</td>
</tr>
<tr>
<td></td>
<td>Urinary calcium</td>
</tr>
</tbody>
</table>
Body manifestations of rickets

Images: Open source, Wikipedia
Imaging findings of rickets
Imaging findings of rickets
Vitamin D deficiency ≠ Rickets

- Biochemical evidence of vitamin D deficiency does not mean ricketic changes are occurring.
- Biochemical evidence of rickets without evidence on x-ray does not lead to increased risk of fractures.
Our patient

- Diagnosis = Child abuse
- Laboratory findings support vitamin D deficiency but would not predispose to developing these fractures.
Case 2

- 3 month old boy comes to ER with parents due to worsening cough and congestion. CXR reveals several old rib fractures and a left sided humerus fracture.

- His parents indicate he had a leg fracture when he was born due to him being a “hard delivery and having to be pulled out.”
Osteogenesis imperfecta

- Bone fragility disorder primarily caused by mutations in COL1A1 and COL1A2
  - Encodes type 1 procollagen
  - Types 1-4
- 17 specific forms now identified (likely there are more)
- Over 800 genetic mutations identified thus far
- Forlino and Marini described an alternative classification system in 2015
  - Group A: Primary defects in collagen structure or function (COL1A1, COL1A2, BMP1)
  - Group B: Collagen modification defects (CRTAP, LEPRE1, PPIB, TMEM38B)
  - Group C: Collagen folding and cross-linking defects (SERPINH1, FKB10, PLOD2)
  - Group D: Ossification or mineralization defects (IFITM5, SERPINF1)
  - Group E: Osteoblast development defects with collagen insufficiency (WNT1, CREB3L1, SP7)
Osteogenesis Imperfecta

- Incidence: estimated at 1 in 15,000 – may be an underestimate as milder forms may not be diagnosed
- 1:1 male:female
- No racial differences reported
Type 1 OI – mild, nondeforming

- No long bone deformity
- May have dentinogenesis imperfect
- Sclera can be blue or white
- Height can be normal but may be less than non-affected family members
- About 50% end up with hearing loss
- Fractures can occur at any age
Type 2 OI – Perinatal lethal

- Patients usually die at birth or shortly after due to extreme rib fragility, lung hypoplasia and CNS malformations or hemorrhages.
- Blue sclera may be present
- All patients have in utero fractures – can involved long bones, skull, vertebrae
- Long bones are severely deformed
- Typically a new mutation as patients do not survive to pass on the mutation.
Type 3 OI – Severe, progressively deforming

- Often have fractures in utero
- Sclera have variable coloring and can be bluish
- Typically have a triangular face with frontal and temporal bossing
- Limb deformities can be severe, compromising function and mobility
- Vertigo is a common issue
- Kyphoscoliosis can be progressive as well, leading to compromised respiratory status
- Renal stones from hypercalciuria can occur (usually as adults)
Type 4 OI – Variable presentation

- Can be difficult to detect.
- Symptoms may be very mild (difficult to detect) to moderately severe (not as bad as type 3).
- Height is quite variable. Some are near normal, some are significantly short.
- Fractures can occur in utero but not commonly.
- Often fractures will begin in infancy.
- Sclera are usually white to slightly grey.
- Femurs may be bowed at birth and tend to straighten with time.
Imaging findings
Diagnosis

- Diagnosis is primarily a clinical diagnosis based on history of fractures and physical exam.
- Routine labs are usually normal – can be useful in excluding other metabolic bone problems.
- Sequencing of genes – slowly becoming better and better but still difficult to estimate the sensitivity/specificity
- Skin biopsy for collagen synthesis analysis
  - False negative rate not completely clear but estimated at around 15%.
- Bone mineral density testing – generally low in patients with OI but can be very variable.
  - Normal density should at least prompt reconsideration of the diagnosis
Our patient

- Review of history reveals a CXR that was done at birth due to difficulty breathing that showed several new rib fractures. Imaging was also found that verified a femur fracture at birth.
- Patient sent to geneticist for further evaluation and diagnosed with OI type 3.

- Important note: OI patients still don’t get classical abuse fractures (classic metaphaseal lesions, etc.).
Case 3

- 9 month old girl develops a febrile viral illness. Several days later she is brought to the ER with acute dystonia. In the ED she is noted to have macrocephaly. A CT scan shows subdural hemorrhages bilaterally with marked enlargement of the sylvian fissure and apparent slight volume loss of the frontal lobe bilaterally. Ophthalmology examination reveals a couple scattered RH’s in each eye near the posterior poles.

- Acute neurological change + subdural hemorrhages + retinal hemorrhages = child abuse?
Glutaric aciduria type 1

- Autosomal recessive disorder
- Defective glutaryl-CoA dehydrogenase
  - Causes increased levels of glutaric acid and 3-hydroxyglutaric acid, both neurotoxic
- Estimated incidence 1:100,000-120,000
  - Old-Order Amish in Pennsylvania up to 1:300
  - Sweden 1:30,000
  - USA estimated at 1:50,000
GA1 clinical presentation

- Can be quite variable, even amongst siblings
- Most have macrocephaly at birth or develop it in the following weeks
- Most are asymptomatic beyond the macrocephaly
- Imaging will often show atrophy of the fronto-parietal area with widening of the sylvian fissures.
  - Micrencephalic macrocephaly
  - Causes stretched veins that are at risk of rupture even with minor trauma
GA1 clinical presentation

- Acute neurological deterioration can occur, usually between 6 and 18 months
  - *Usually triggered by a febrile illness with some degree of dehydration*
    - Usually a rapid deterioration
  - *Can still occur more insidiously*
- Usually follows 1-3 days of fever and vomiting
- Child becomes acutely hypotonic, loses head control and can have seizure-like movements (no seizure activity on EEG)
- After weeks there often is improvement in the dystonia but pt’s remains severely disabled.
- May live to adulthood but are wheelchair bound and require assistance.
Imaging findings in GA – 1

- **A** - Expanded anterior middle cranial fossa subarachnoid space due to frontotemporal atrophy

- **B** - “Batwing” appearance of the expanded Sylvian fissures

- **C** - Decrease in Attenuation of the putamen bilaterally

- **D** - Large bilateral convexity subdural hygromas.

GA1 clinical presentation

- Acute deterioration is rare after age 5.
- If patients do not have the acute deterioration they can live relatively normal lives, though MRI still shows brain substance loss.
Retinal hemorrhages are often thought to be “pathognomonic” for abusive head trauma.

In reality, the possible causes of RH's are extensive; however, almost all these causes only produce a few RH's near the posterior pole.

GA – 1 has been described to cause RH’s but they are different than are seen in AHT.

- *Typically only a few scattered hemorrhages near the posterior pole as opposed to TNC RH’s extending to the ora serrata involving multiple layers.*
GA-1 Diagnosis

- Urine organic acids will identify abnormal increases in 3-OH-glutaric acid.
- Newborn screening in Arizona now tests for this disorder
  - *GA1 produces carnitine deficiency and increased levels of C5 dicarboxylic (glutaryl) carnitine in urine and plasma.*
  - *C5-DC is used to screen.*
- Some affected infants will not have elevated levels of C5-DC so this is not 100% effective.
  - *Send urine organic acids if concerned*
- DNA sequencing also available
  - *Most helpful in screening for carriers if desired*
GA-1 Urine Organic Acid Profile

GA-1 Treatment

- No specific treatment has been established as gold standard yet.
- Low protein diet to minimize lysine supplemented with lysine free formula to help ensure other needs are met.
- Carnitine supplementation (100 mg/kg/day)
- Riboflavin has been suggested but current evidence suggests only normal dietary amounts are needed.

Crisis management:
- 200 mg/kg IV glucose infusion over a few minutes
- 10 – 20 ml/kg NS bolus to help correct hypovolemia
- Start on D10 1/2NS at maintenance+deficit correction rate.
- When no longer nauseated start lysine free amino acid formula enterally
- Carnitine 200 mg/kg/day IV supplementation
- May need IV insulin due to high glucose infusions
- Aggressive fever control
GA -1 Prognosis

■ If treatment is started before symptomatic there is good evidence children can live symptom free into adulthood.

■ Established treatment is new so it is unknown if they will have problems in adulthood.
Case 4

- 5 y/o girl presents with mild vulvar itching and some blood stains in her underwear. Mother looked and noticed what appeared to be abrasions and brought her to the clinic for evaluation as she was concerned that someone was abusing her.

- The patient does not report any inappropriate contact.
Lichen Sclerosus et Atrophica

- Chronic inflammatory dermatosis
- Presents as white plaques that slowly coalesce.
- Epidermal atrophy and scarring can follow.
- While not restricted to the genital area, this is a commonly affected site.
  - 5:1 genital:extragenital
- Frequency is not known
  - Male genital involvement seen almost exclusively in uncircumcised men
- No racial predilection has been identified
- 6:1 female to male
  - Most common presentation is vulvar area
- 15% of cases are in children
Classical appearance of a Figure-of-8 around the perineum and anus. Skin can be fissured and have some bleeding.
Treatment

- Ultra-high potency topical steroids (clobetasol, betamethasone)
  - *Must monitor for steroid induced skin atrophy*
  - *Treatment should not be abruptly discontinued-step down therapy indicated*
- Topical testosterone is widely used but newer data suggests it is no more effective than placebo, and can cause virulization.
- Topical calcineurin inhibitors (tacrolimus, pimecrolimus) have been shown to have some effect but generally are less effective than steroids. Possible use in maintenance phase?
- Anecdotal evidence that UVB/PUVA may have a beneficial effect
- Circumcision for boys can be considered (also treats increased risk of phimosis)
- Surgical excision not recommended for girls.
- Recurrence rate is 44-82%
- Unclear if early treatment in childhood can prevent disease from becoming chronic.
Prognosis

- Good for most acute genital cases
- Pediatric disease may resolve spontaneously or improve dramatically with puberty
- Exogenous and chronic atrophic genital cases generally have limited improvement
- Association with future malignancy (squamous cell carcinoma) is unclear for adults
Case 5

2 week old boy presents to ER with a “full sensation” to his anterior fontanel. ED obtains head imaging and small, bilateral SDH’s overlying the parietal areas. Ophtho happens to be in the ED and does a retinal exam, 6 to 7 RH’s along the posterior aspect of the eye bilaterally are seen, mostly flame hemorrhages with a few dot-blot type bleeds.

He is admitted for concern of child abuse.
Birth trauma

- More recent studies suggest 26-50% of asymptomatic term infants may have SDH’s (Holdon, Looney, Rooks)
- Abnormal labor significantly increases the risk
  - 1:664 delivered by forceps
  - 1:860 delivered by vacuum
  - 1:907 delivered by cesarean with a trial of labor
- Presence of SDH is not necessarily indicative of excessive birth trauma
- Method of assisted delivery may be more important than the urgency of delivery
Subdural hematomas

- Birth subdural can be supratentorial or infratentorial
- Generally located in the posterior cranium
  - Rarely seen along the frontal convexities but may occasionally be interhemispheric
  - Compared to AHT which generally are along the cerebral convexities (especially the frontal-occipital areas) and into the falx
- Rare to see SDH’s of varying ages or chronic subdurals.
  - Only 6 reported chronic subdurals at birth from 1977 to 2005
- Most resolve by a month of age.
  - 94-100% resolved by 1 month of age (Whitby, 2004)
  - Only 2 of 46 patients required follow-up imaging at 3 months, both showed complete resolution at 3 months (Rooks, 2008)
- Most with birth SDH’s have normal development at age 24 months.
Posterior fossa SDH in a neonate delivered via SVD

Neonate delivered via SVD with both supratentorial and infratentorial SDH.
Birth retinal hemorrhages

■ Most studies suggest 20-35% of neonates will have RH’s related to birth
  - *Incidence highly dependent on the time of exam.*
  - *Up to 45% can have hemorrhages seen in the first 36 hours*
  - 11-20% by 72 hours
  - Suggests that these hemorrhages resolve rapidly

■ More common after vacuum assisted delivery (46-78%)

■ Data is equivocal for forceps deliveries (some studies show increased risk, others show decreased risk)

■ Cesarean section seems to be protective
Retinal hemorrhages

- Typical RH’s from birth are located near the posterior pole of the eye
- Most birth RH’s are intraretinal (flame or dot-blot in nature), Most AHT RH’s are multilayered
- Flame type hemorrhages typically resolve by 2 weeks.
- Dot and blot hemorrhages resolve by 6-8 weeks.
- RH’s cannot be reliably dated
- Studies suggest birth hemorrhages may actually resolve faster than this.
- RH’s outside 2 months of age cannot be attributed to birth
- Traumatic retinoschisis has never been described from birth
Right eye of a child with Abusive Head trauma: White arrow-preretinal hemorrhage, black arrows-intraretinal dot-blot hemorrhages, white arrowhead – ora serrata

Birth Retinal Hemorrhages

Case 6

- 6 y/o presents to the PCP for well check. Exam reveals these markings on the legs and buttocks.
Phytophotodermatitis

- Occurs when certain biologicals (furocoumarins) react with ultraviolet light
  - *Found in parsley, celery, carrots, limes and a few other plants*
- Usually becomes apparent approx. 24 hours after exposure and peaks 48-72 hours after.
  - *May be intensified by wet skin, sweating and heat*
  - *Think margaritas at the beach (but never in any of our patients, right?)*
- Area may blister and can give a burning sensation to the patient.
- Typically this is self limited and needs no treatment.
  - *Severe cases involving > 30% of skin may need burn unit treatment.*
Phytophotodermatitis vs bruising

- Both may be patterned but patterned bruises are usually an outline.
Case 7

- 7 y/o boy is brought to the PCP due to a cold. While examining his chest large bruises are found over his back.
Cupping

- Marks formed from suction created by applying a heated cup to the skin.
  - Cup cools and creates a suction
  - Also can be created by cupping devises

- Theorized to pull toxins and stagnant blood to the surface allowing new “healthy” blood to enter the deeper tissues.
Various cupping kits for sale online
Coining – Cao Gio
(pronounced Gow-Yaw)

- Created by rubbing a coin or back of a spoon against the skin.
- Traditional healing method often practiced in southeast Asia.
- Current theory is that it induces an inflammatory response causing the immune system to “rev up” and fight infection.
Case 8

- 17 month old girl is brought to ER after his mother found her laying in the hallway crying and refusing to walk.

- She reports she was in the other room and heard her playing then crying.

- ER evaluation reveals a spiral femur fracture of the right leg. She is admitted for placement of a spica cast and further evaluation.

- Child abuse?
Likely a plausible accident

- Similar to a toddler’s fracture of the tibia
- In ambulatory, prepubertal children the muscles are often stronger than bones
- Mechanism is a planting/twisting of the leg (often after running)
- Studies suggest that between 10-30% of femur fractures (including spiral) in ambulatory children less than 3 yrs are abusive
  - >90% of spiral fractures are abusive in non-ambulatory children
Case 9

- Mother brings 2 y/o daughter to urgent care after finding blood in her underwear.
- Mother and father are divorced and pt. just came back from fathers after the weekend.
- Mother found blood in underwear and called dad asking what happened – he reports no knowledge of injury other than a fall in the tub.
- Mother suggests abuse – “I knew that @*!($ would do something to her”
- In UC pt is noted to have large bruise to the vulvar area. Transferred to ER.
Exam finding
Urethral prolapse

- Exact mechanism not known
- More common in prepubertal girls
  - Less estrogen thought to cause weaker tissue
- More common in black and Hispanic girls
- Associated with heavy cough, constipation, obesity, UTI’s, trauma
  - Increased intra-abdominal pressure
Urethral prolapse treatment

- Several methods have been hypothesized
- Sitz baths help with symptomatic control and may help resolution.
- Topical antibiotics have been postulated to help in involution; however, one study showed a high rate of persistence of prolapse 3 years later despite resolution of symptoms.
- Current thought is to treat with topical estrogen cream 2-3 times daily for 2 weeks. Showed a high rate of involution over 3-6 weeks with little recurrence.
- For medical management failure or for areas of thrombosis/necrosis, surgery is recommended sooner rather than later.
Case 10

- 2 y/o girl is brought to the PCP after she slipped and fell, straddling the bathtub wall. PCP noted labial bruise but also a perineal laceration that was concerning so a report was made to LE who then requested an evaluation at the Children’s Advocacy Center. There is no history of abuse reported.
A quick word about CAC’s

- Children’s advocacy centers are set up to bring all services to a single place and minimize trauma to the patient.
  - *Child can have forensic interview and exam all at once.*
  - *Interview is done by forensic interviewer and recorded so pt. doesn’t have to continue to repeat the story over and over.*
  - *Need to be old enough to understand and answer questions – usually age 3-4 is the earliest a child can be interviewed.*

- Generally cannot refer directly to the CAC.
  - *Referral must come from law enforcement*
Exam findings
Failure of midline fusion

- Also known as a perineal groove
- Always found in the midline.
- Note the vascularity of the lesion.
- Non-painful
- Resolves at puberty
- Follow-up exam in 1-2 weeks would show persistence of lesion.
Can also occur around the anus

- Still will always be in the midline
- Typically is shiny in appearance with raised borders
Straddle injury

- If there is deeper injury there should also be more superficial injury.
- Rarely ever injury the hymen
  - No hymenal injury seen in this image (yellow arrow)
Case 11

- 6 y/o girl presents to the ER with a laceration of the anal opening. She reports she fell in the shower.
- She lives in Pennsylvania and is traveling with her grandparents. They are staying in a hotel as they travel to California to go to Disneyland.
Exam findings
The abuser
After suggestion of mechanism that could occur by falling in the shower, LE evaluated the hotel room and found blood on the water spigot and diverter. Her parents were contacted and verified that she was, indeed, with her grandparents on the way to Disneyland.

Laceration was sutured and patient was monitored for 24 hours post-operatively. She was discharged in good condition.

Disneyland trip got canceled unfortunately and she flew back home with her grandma.
Case 12

- 18 month old boy presents to ER unresponsive. Parents called 911 after he had a seizure and stopped breathing. EMS arrived and found patient with severe bradycardia and agonal breathing. CPR was initiated. He had 10 minutes of CPR in route. In ED heart rate recovered and CPR was stopped. Pt. was intubated and admitted to the PICU.

- Evaluation reveals bilateral subdural hematomas, retinal hemorrhages bilaterally (TNC extending to the ora serrata bilaterally), and postero-medial acute rib fractures.

- Intensive care team consults child abuse pediatrician and asks could this all be due to seizure and CPR?
Do seizures cause subdurals?

- Unlikely to occur
- SDH’s may reduce seizure threshold and a seizure may follow.
- SDH may cause some increased intracranial pressure that can also produce a few retinal hemorrhages but not in the number or extent of that seen in AHT.
Posterior rib fractures

- Occur when the infant is grabbed under the chest with fingers at the costovertebral junction.
- Front-back compression “bends” ribs over the extensions of the spine.
- 90% of abusive rib fractures are in children < 2 years old.
  - Rarely seen in older children as it’s harder for adults to hold child in the position required for fracture.
- PPV for abuse is essentially 100% in kids < 2 yrs without history of major trauma.
Posterior rib fractures

- Healing (red arrows)
- New (blue arrows)
In contrast, 2 y/o run over by SUV twice across the chest

-No rib fractures
Does CPR cause rib fracture?

- Exceedingly rare in children
  - *Ribs are still spongy*
- Rib fractures seen in adults are typically anterior and in a line.
- With the new “hand encircling-thumb” compression technique there does seem to be a slight increase in occurrences of rib fractures; however, they are still anterior or antero-lateral, not posterior.
Does CPR cause RH’s?

- If present there may be a few RH’s in the posterior pole.
- RH’s as seen in AHT are not seen due to CPR.
- Similarly, hypoxia from failure to start CPR does not cause SDH’s or RH’s as they are seen in AHT.
Case wrap up

- Findings are consistent with abusive head trauma.
Questions?
Thank you!