Controversies regarding abusive fractures

Michelle Clayton MD, MPH
Child Abuse Pediatrician, CHKD
Associate Professor, EVMS

Fracture evaluation

- There is no single fracture type which is diagnostic of abuse:
  - Rib, metaphyseal fractures highly associated with abuse
- Diagnosis of abuse depends upon age and developmental ability of child:
  - When fractures occur in a child who is not ambulatory (able to get around on their own), there is a heightened suspicion for abuse in the absence of a plausible explanation for the injury.

Fracture evaluation

- Fractures have multiple configurations which can give important information about the type of force causing the fracture.
- Understanding this relationship between type of force and fracture type can permit an experienced clinician to determine whether the fracture history is consistent with the injury.
- Bruises are often not present at the fracture site because many fractures occur via indirect forces.

Type of force applied can be deduced from fracture configuration

Screening for Skeletal Trauma

Skeletal survey:
- Detects occult skeletal injury
- Includes all extremities individually, hands and feet, 2 views of the chest/spine, 2 views of the skull
- Used in infants < 2 years old
- Used occasionally in children 2-5 years, and not indicated over 5 years old
Defense strategies in suspected abuse cases involving fractures

Rickets

What is rickets?

- A bone disorder caused by deficiency of Vitamin D, calcium, or phosphate
  - Calcium and phosphate are the mineral components of bone
- There are several different types of rickets, all characterized by softening and poor mineralization of bones
  - Most common is nutritional rickets (caused by dietary deficiency of Vitamin D, calcium, or phosphate)
  - Most commonly seen in children ages 6 months – 24 months

Vitamin D metabolism

- Body's supply is obtained by dietary intake
- Inactive form (25(OH)D₃) is the most common form present in the body
- Active form is created by skin cells when they are exposed to sunlight
- Activated Vitamin D (1,25(OH)₂D₃) responsible for absorption of calcium from intestine

Categories of Vitamin D deficiency

- Sufficient >30 ng/mL
- Insufficient 20-30 ng/mL
- Deficient <20 ng/mL
  - Mild 10-20 ng/mL
  - Moderate 5-10 ng/mL
  - Severe <5 ng/mL
How often does Vitamin D deficiency occur?

- Frequency in US is high, ranging from 22-58%
- Occurs in healthy infants and children
  - More prevalent among overweight/obese children
- Most common cause is nutritional deficiency
  - Breastfed infants, infants with darker skin at higher risk, especially during winter months

Does Vitamin D deficiency always lead to fractures?

- Fracture risk is low, even among children with bone abnormalities from rickets (0-17%)
- When fractures occur in children with rickets, the affected children have severe rickets with obvious clinical features

Rickets: Laboratory findings

- Can have decreases in:
  - Calcium
  - Phosphorus
  - Calcified (calcified), a precursor which is metabolized to form active vitamin D
  - Activated vitamin D (calcitriol)
  - Urinary calcium
- Can have increases in:
  - Parathyroid hormone
  - Alkaline phosphatase
  - Urinary phosphorus

This young girl had been rescued from a "baby farm" around 1880 or 1891

Widened metaphyses

Rachitic rosary
Rickets: Radiographic appearance

- Radiographic features
  - Metaphyseal cupping and fraying
  - Cortical thinning
  - Periosteal reaction
  - Widened metaphyses
  - Costochondral budding (rachitic rosary; anterior rib enlargement at costochondral junction)
- Generalized distribution of skeletal findings
- Symmetric pattern of skeletal involvement

Metaphyseal cupping and fraying

Metaphyseal cupping and fraying

X-ray of rachitic rosary

Congenital rickets

- Very rare
  - An Indian study looking at 337 million births found 3 cases
- Results when maternal Vitamin D and calcium levels are very low and mothers have osteomalacia
- Maternal calcium stores have to be exhausted in order for this condition to occur

Vitamin D metabolism in fetus

- Fetus obtains calcium from mother by active uptake
  - Calcium and phosphorus levels in fetus higher than in mother
- Calcium levels in the fetus are maintained even when maternal calcium levels are low
- Placental parathyroid related protein (PTHrP) is primary mechanism for fetus obtaining calcium from mother
- Fetal parathyroid hormone (PTH) also controls fetal bone mineralization and responds to maternal hypocalcemia
- Vitamin D is not required for fetal skeletal development or mineralization
Osteogenesis imperfecta (Brittle bone disease)

Vitamin D metabolism in infant

- Newborn is dependent on intestinal absorption of calcium, rather than active uptake of calcium controlled by Vitamin D
- Vitamin D deficiency does not appear to affect the newborn
- Even children with genetic disorders of Vitamin D do not have rickets at birth

What is OI?

- OI is an inherited disorder characterized by weak bones which break easily
- Caused by defect in collagen
  - Collagen is a protein which is abundant in skin, bone, and other connective tissues which provide strength and structure
- Severely affected people can have fractures before birth; those with milder forms may only have a few fractures during their lifetime
- For the most common types (Types I and IV), fractures occur most often during childhood and adolescence, and less frequently after adulthood

Osteogenesis imperfecta: Diagnosis

- X-rays generally helpful
- Examine child for presence of clinical features
- Collect three-generation pedigree from family regarding relevant historical features
- Skin biopsy to test for normal Type I collagen
- If normal collagen is present, either patient without OI, or previously unidentified mutation is present

Osteogenesis imperfecta: Type I

- Mild phenotype
- 66% of cases
- Mild short stature
- Pre-senile hearing loss in about 50%
- Mild to moderate osteoporosis
- Frequency of fracture decreases at puberty
- Bowing of long bones eventually occurs
- Autosomal dominant
- Wormian bones
- Family history positive
- Ligamentous laxity
- Easy bruising
- Delayed fontanelle closure

Osteogenesis imperfecta: Type I: subtypes

- Group I A
  - Blue sclera
  - Normal teeth
    (most are group I A)
- Group I B
  - Dentinogenesis imperfecta
    (uncommon)
**Blue sclerae**

- More severe bone fragility than in type I, less severe than in type III
- Tilted hearing
- Normal sclera
- Fracture frequency decreases at puberty
- Most are short in stature
- Variable deformity of long bones

**Dentinogenesis imperfecta**

- Premature loss of teeth
- Dentin is not as opaque as in type I
- More teeth are affected
- Height and weight are normal

**Osteogenesis imperfecta**

- Type IV
- Less than 10% of cases of OI
- Less likely to be confused with abuse
- Extremely severe form of the disease
- Infants born with multiple fractures and deformities
- Extreme osteopenia
- Poor mineralization of bones
- Wormian bones
- Triangular faces
- Blue-black sclerae
- Small thorax
- Prenatal death common
- Premature death due to respiratory insufficiency
Triangular facies

Osteogenesis imperfecta
Type II

Osteogenesis imperfecta
Type III
- Most severe nonlethal form
- Severe bone fragility
- Multiple fractures
- Severe bowing and shortening of extremities
- Normal or slightly blue sclerae
- Severe skeletal dysplasia
- Presence of fractures at birth
- Severe progressive deformities of extremities and spine
- Autosomal recessive inheritance (negative family history)

Ol Type III: typical radiographs
- Demineralization of tibia and fibula
- Bowing deformities of tibia and fibula
- Healing fracture of the distal femur diaphysis

Ol Type III: typical radiographs
- Severe bowing of humerus (previous fracture with abnormal healing)
- Thinning of diaphysis of humerus, radius, ulna
- Elongation of radius, ulna

Transverse fracture in Ol
Osteogenesis imperfecta:
Radiographic appearance
• Any type of long bone fracture can occur in OI
• Transverse fractures of bone are by far the most common
• Oblique and spiral fractures are also very common, as in all childhood trauma
• Fractures of metacarpals and phalanges occur in child or toddler with OI
• Rib fractures, accompanied by respiratory insufficiency, only in the most severe variants (Types II and III)

Osteogenesis imperfecta:
Uncommon radiographic features
• Rib fractures are uncommon in OI
  – Might occur with minor trauma in child with osteopnenic ribs (i.e., more severe variants)
• Multiple fractures in various stages of healing not usually seen
• Brain trauma is not part of the OI picture; case reports of SDH with minor trauma

Osteogenesis imperfecta:
Uncommon radiographic features
• Vertebral body compression fractures only seen with severe osteopenia (e.g., Type III)
• Skull fractures are not typical presentation of OI, although can be seen if trauma occurs
• Classic metaphysical lesions (CML) are rare in OI

Ehlers-Danlos Syndrome
Definition

- Group of connective tissue disorders caused by defects in collagen structure, production, or processing
  - Can also be caused by such defects in proteins interacting with collagen
- Collagen is an abundant protein found in most body areas, including skin, muscle, tendons, ligaments, blood vessels, organs, gums, eyes
- Most forms inherited in autosomal dominant pattern; a minority are inherited in autosomal recessive pattern, or as sporadic mutation

Classification

- Previously classified into 10 types
- Nomenclature system revised in 1997 to create six types
  - Classical (formerly types I and II)
  - Hypermobility (formerly type III)
  - Vascular (formerly type IV)
  - Kyphoscoliosis (formerly type V)
  - Arthodactyly (formerly types VIa and VIb)
  - Dermatosparaxis (formerly type VII)

Clinical manifestations

- Most have an unusually large range of joint movement, especially with hypermobility subtype
- Many affected have soft, velvety skin which is elastic, fragile, and bruises easily; some also have abnormal scarring
- Infants affected by EDS can present with weak muscle tone, causing delays in motor skills (e.g., sitting, pulling to stand, walking)
- Often have unstable joints which are prone to dislocation, chronic pain

Hypermobility type (type III)

- Affects 1 in 10,000 to 15,000
- Autosomal dominant or autosomal recessive inheritance
- Mutations in TNSB or COL3A1 genes
- Joint hypermobility is a frequent manifestation
  - Frequent dislocations and subluxations result
  - Osteoarthritis is common, and often occurs relatively early in life
- Chronic severe pain is a common symptom
- There is contradictory evidence regarding the occurrence of decreased bone mineral density in EDS patients with hypermobility type

Hypermobility type (type III): Major diagnostic criteria

- Joint hypermobility
  - Confirmed by a score of 5 or more on the joint hypermobility scale
  - Young children (up to 10 years) tend to be very flexible and are therefore difficult to assess
- Soft skin with normal or only slightly increased extensibility
- Absence of fragility or other significant skin or soft tissue abnormalities
  - For example, no spontaneous or easily induced tears of skin, organs, blood vessels; no surgical complications such as subscapularis/trapezius wound dehiscence; no thin, translucent skin; no atrophic scars

These criteria should all be met to establish a diagnosis of EDS, hypermobility type.
Hypermobility type (type III):
Minor diagnostic criteria

- Positive family history of EDS, hypermobility type, or family history of joint laxity
- Recurrent dislocation or subluxation of multiple joints
- Chronic joint pain, limited range of motion in hands, shoulders, or ankles
- Fatigue or easy bruising
- Gastrointestinal symptoms (gastritis, GERD, delayed gastric emptying)
- Neuromuscular symptoms such as positional orthostatic tachycardia
- High, narrow palate
- Dental crowding

The presence of these criteria is supportive of, but not sufficient to, establish a diagnosis of EDS, hypermobility type.

Classic type (types I and II):
Minor diagnostic criteria

- Frequency of 1 in 20,000 to 40,000 people
- Autosomal dominant
- In the majority of affected families, the variant is localized to COL5A1 and COL5A2
- Diagnostic testing can be done by electron microscopy of a skin biopsy; biochemical testing of cultured fibroblasts; genetic testing

Classic type (types I and II):
Major diagnostic criteria

- Skin hyperextensibility
  - Skin extends easily and snaps back after release
- Widened areolar scars
  - Often have stretching of scars after primary healing
- Joint hypermobility
  - Can be assessed using Beighton scale
- Positive family history

A combination of the first three diagnostic criteria should have a high specificity for EDS, classic type.

Classic type (types I and II):
Minor diagnostic criteria

- Smooth velvety skin
- Molluscoid pseudotumors
  - Fleshly lesions associated with scars over pressure points
- Subcutaneous spheroids
  - Small cyst-like nodules which are freely moveable, located over bony prominences of legs and arms
- Complications of joint hypermobility
  - Sprains, dislocations, subluxations, pes planus

Classic type (types I and II):
Minor diagnostic criteria

- Muscle hypotonia, delayed gross motor development
- Easy bruising
- Manifestations of tissue extensibility and fragility
  - Hiatal hernia, anal prolapse in childhood, cervical insufficiency
- Surgical complications
- Postoperative hernias

The presence of one or more minor criteria contributes to the diagnosis of EDS, classic type, but is not sufficient to establish the diagnosis.

Ehlers-Danlos Syndrome (EDS) and fractures

- Some experts have been diagnosing parents of children with fractures as having EDS, and relating this to an increased fracture tendency
- EDS is connective tissue disorder with hyperextensible skin, joint laxity, easy bruising; it is inherited in autosomal dominant (more common) or autosomal recessive pattern
- Study of 23 patients with EDS (age range 18-64 years) showed that, when compared with healthy staff at a hospital, they were ten times more likely to have a past history of low-impact fracture
- Fractures occur on extremities, humerus, ribs, tibia, femur, clavicle, scaphoid
- Bone density of patients reduced compared to controls
- These findings have limited applicability to child abuse

Temporary Brittle Bone Disease (TBBD)

- Theorized to be caused by temporary defect in collagen formation
  - Collagen is a structural component of bone, skin, and blood vessels
- Suggested that a metabolic defect in collagen processing exists
- Diagnosis is subject of controversy
- No defect has ever been proven
- No scientific basis for TBBD

TBBD features

- Child with multiple unexplained fractures in first year of life, and none thereafter
- Caregivers denied wrongdoing throughout investigation
- No apparent episode of trauma
- Lack of cutaneous injury
- No other findings suggestive of child abuse (i.e., subdural or retinal hemorrhages)
- Laboratory evaluation without evidence of metabolic condition
- Radiographs without evidence of metabolic disease
- Associated features include twins, prematurity, anemia, coeliac, anemia family history of hyperextensibility

Study design

- Studies can be descriptive or analytic
  - Descriptive studies are designed to generate hypotheses (theories about disease causation)
    - Cross-sectional studies
      - Case-control and case series
    - Analytical studies are designed to test hypotheses (determine whether or not a hypothesis is likely to be true)
      - Observational
        - Ecological
          - Temporal
          - Propective
          - Retrospective
          - Case-control
          - Group data

Descriptive studies

Designed for hypothesis generation

- Cross-sectional studies
  - Use data from entire populations to compare disease frequency between different populations at the same time, or in the same population at different points in time
  - For example, rates of colon cancer have a correlation with per capita meat consumption
- Case reports, case series
  - Case reports are the most basic descriptive study of an individual
    - Consists of data that report regarding a single patient
  - Case series are a case report of a case series
    - Does not have characteristics of a number of patients with a given disease
Descriptive studies (con’t)

- Cross-sectional surveys
  - Collects information from individuals regarding disease and exposure to risk factors
  - Information is explored for relationships between disease and various risk factors

Analytical studies

- Designed to answer a research question of interest
- Observational studies
  - Case-control study
    - Patients are selected on the basis of whether or not they have disease and are compared with a group of people who do not have the disease to determine their exposure to risk factors of interest
    - Groups are compared with each other regarding proportion with risk factor

Analytical studies (con’t)

- Observational studies
  - Cohort
    - Patients selected on basis of whether they are exposed or not exposed to the risk factor of interest
    - They are followed for a specified period of time (sometimes several years) to determine whether disease develops
    - Retrospective
      - Exposure and outcome of interest has already occurred at the start of the study
    - Prospective
      - Exposure has occurred, but outcome of interest has not yet occurred at start of study

Analytical studies (con’t)

- Intervention studies [clinical trials]
  - Community trial
    - Non-random assignment to intervention and control groups
  - Clinical trials
    - Random assignment to intervention and control groups

Approach to bogus defenses

- Help the finder of fact keep the big picture in mind
  - Keep referring back to the totality of the injuries when you’re being asked questions which pick apart the causes of injury
- Speak clearly and unambiguously
  - Do not introduce alternative theories into your testimony (but of course, clearly address any theories you are asked about)
  - Don’t waver in your assessment of clearly abusive injuries – remember why you diagnosed the injury as abusive

Literature reviews

- Discusses published information in a particular subject area, sometimes within a certain time period
  - Biased literature reviews may exclude a large number of relevant articles
- Can be simple summary, recap of important information, but is often a summary and synthesis (reorganization or reinterpretation of older information, or designed to show progression in a field)
- The focus is to summarize and synthesize information without adding new contribution
Approach to bogus defenses

- Do not allow inaccurate information to be given to the finder of fact without challenging it (for example, a defense attorney saying that bruises are a presenting feature of IBD)
- Ask to read papers which are shown to you in court (or ask for the citation if they are making outrageous conclusions from a paper they will not show you)
- Be prepared to explain why genetic testing was/was not performed
- Be prepared to explain why a bleeding disorder diagnosis was/was not pursued