The differential diagnosis for a young child with 1 or more fractures is extensive and should be given careful consideration. Because inflicted trauma is a frequent cause, the question is best rephrased as “What is a reasonable differential diagnosis for the young child with fracture(s) and the cost-effective approach to the evaluation of such a child?” This article focuses on diagnoses introduced into the forensic assessment of the young child with multiple fractures. Is it appropriate to order tests simply to address possible differential diagnoses introduced by the defense? Are physicians who do not test making conclusions about child abuse prematurely? How does the analysis of injury biomechanics and fracture type help us? Does a delay in obtaining medical care affect the assessment of child abuse or neglect?

Defense attorneys may want to create reasonable doubt during a criminal prosecution when child abuse is being alleged and would prefer that testing not be done, so that the possibility of a medical explanation for the findings lingers. Prosecuting attorneys would prefer to eliminate that possibility of doubt, by having definitive tests that have excluded various medical/pathologic causes for the child’s fracture(s). Physicians must be judicial in ordering tests and order them when it is medically reasonable to do so, rather than when they are legally indicated. Which medical conditions may predispose a child to fracture or be mistaken for trauma on imaging studies? At what point and at what cost is testing for these conditions appropriate?

Suspected child abuse and neglect require evaluation by a multidisciplinary team. It is the medical provider’s job to report suspected child abuse while further diagnoses are being considered. There is immunity from civil and criminal liability on the presumption that the reporting party is acting in good faith. However, the inverse is not true; if the responsible party suspects child abuse and does not immediately report it, he or she is civilly and criminally liable for failure to report, which is punishable with a fine and/or imprisonment. Unless
suspected child abuse is reported, the physician may be unaware that there have been previous allegations. Also, if the report to child protective services (CPS) has not also been cross-reported to law enforcement, the team may be unaware of the criminal history of the caregivers.

**CASE 1**

A 3 month old is brought in by ambulance, called by the mother who perceived that the infant had left knee pain and swelling. The mother relates a vague history of trauma, after hearing the results of the leg radiograph. The parents are separated and each has a history of prior arrests. The infant’s examination is remarkable only for crying and perceived pain with movement of the left leg and when the child is lifted or carried. A skeletal survey (SS) reveals an acute proximal tibia fracture and a corner metaphyseal lesion of the distal femur. The radiographs do not reveal any evidence of osteopenia or structural abnormalities of the bones. Laboratory testing shows normal electrolytes, calcium, phosphorus, and alkaline phosphatase levels. A radionucleotide bone scan additionally reveals findings suspicious for 3 right-sided rib fractures, 2 of which are anterolateral and 1 is posterior-medial (Fig. 1). A follow-up chest radiograph 10 days later confirms the 3 rib fractures (Fig. 2). The investigating detective asks whether we have ruled out the possibility of the infant having brittle bone disease. The detective reports that the criminal prosecutor wants to know.

Fig. 1. Bone radionucleotide study shows increased uptake in the right ribs (thin arrows) and proximal left tibia (thick arrow).
CASE 2
A 3 year old is referred to the clinic for suspicion of child abuse after sustaining a proximal ulna fracture at age 2 years (Fig. 3). The case was brought to the attention of CPS when a neighbor called the hotline after observing the child crying while guarding his arm all day. The child is brought in by a new foster parent who has no history on the child or his family. The child also has a history of a tibia/fibula fracture sustained at age 18 months, at which time an obese 12-year-old cousin fell on him (Fig. 4). The child is speech delayed and can offer no explanation for either injury. There have been 5 previous reports for child neglect, emotional abuse, caretaker absence, and physical abuse (3 unfounded and 2 substantiated). The mother is known to abuse crystal methamphetamine, and his youngest sibling was methamphetamine exposed at birth and died at 1 month in an overlay incident.

CASE 3
A baby who was a 25-week preemie was brought to the emergency department at age 8 months by her mother who reported decreased use of her right leg. During the evaluation, a frontal skull fracture was discovered in addition to healing fractures of the distal right tibia and proximal left radius. There were no explanations for any of the fractures. The infant had spent 4 months in the neonatal intensive care unit (NICU) where she was diagnosed with rickets based on increased alkaline phosphatase (1104 IU/L), craniotabes, rachitic rosary, and pathologic radius and rib fractures. Radiographs revealed diffuse osteopenia; periosteal changes; and cupped, frayed metaphyses.
FRACTURES AND ABUSE

National Child Abuse and Neglect Data System data revealed that, in the United States in 2007, an estimated 794,000 children (1% of minors) were reported to CPS and substantiated as victims of maltreatment. More than one-half (59.0%) of victims suffered neglect, 13.1% of victims suffered from more than 1 type of maltreatment, and 10.8% suffered physical abuse [1]. There were many more victims who were not recognized or not reported.

Fractures are one of the leading presentations of child physical abuse, second to soft tissue injuries. Of children less than 12 months of age hospitalized with fractures, approximately 25% were attributed to abuse. Of children less than 36 months with fractures, the overall proportion leading to hospitalization because of abuse was 12% [2]. However, there are children initially believed to have been abused who have fractures that are common from accidents [3]. Kirschner and Stein [4], a forensic pathologist, described 10 cases reported as child abuse by inexperienced physicians that led to false allegations and failure to recognize serious medical conditions. Small but significant numbers of children who are deemed nonabuse have factors predisposing to bone fragility after assessment [5]. Wardinsky and colleagues [6], as part of a child abuse team, analyzed 504 consults and found 7% to have medical conditions, such as osteogenesis imperfecta (OI) or Ehlers-Danlos syndrome initially not considered. Dr Carole Jenny [7], in a clinical report on behalf of the American Academy of Pediatrics, discussed the differential diagnosis and medical evaluation of young children with fractures. She comments that parents of children later diagnosed with medical conditions leading to bone disease were at times mistakenly accused of child abuse [7].
SUSPICION AND REPORTING
Primary care pediatricians underreport and underdiagnose child abuse, especially compared with doctors with access to consultation with child abuse specialists [8–11]. Child abuse specialists in Toronto found that 20.5% of inflicted fracture cases were initially unrecognized as abuse related. The cases more likely to be missed were boys with extremity fractures who presented in the emergency department or to their pediatrician [12]. For many pediatricians, it is unfathomable that a parent could harm a child to such a degree. Some pediatricians deny that abuse is a possibility despite overwhelming signs that a child has been seriously injured, because they are so devoted to the families [13]. It is challenging for a general pediatrician who has an ongoing relationship with a family to suspect or accuse an outwardly loving caregiver of abusing a child. The opinion may be deferred to an unbiased expert in the field who does not have personal loyalties to the family.

WORK-UP AND EVALUATION
How can a price be placed on laboratory testing when the caregiver’s and child’s lives may be affected in such a drastic way? Who should pay for testing? Consequences may involve criminal, personal, and emotional costs that are priceless to the accused. Children are placed in protective custody, criminal charges are pressed, and major financial strains are imposed on the family.
CONSIDERING THE HISTORY OF TRAUMA WITH THE BIOMECHANICS

Recent studies of engineering principles have changed the way fractures are analyzed. When considering the history, the clinician must carefully consider the external forces imposed on the bone with characteristics of the fracture (type, location, number, and age) [14]. To consider the mechanism, a detailed history must be obtained, to translate which loading forces were involved to see whether the history and anticipated forces correlate (Table 1). Factors to consider are the movement of the child at the time of injury, the height of the fall, and the surface onto which the child fell. On occasion, a perpetrator may partially disclose the mechanism of injury. The fewer the details, or the more inconsistencies, the higher the suspicion of inflicted injury or neglect.

There are pure external forces, simply push (compression), pull (tension), and shear. However, most real-life injuries involve a combination of forces. All forces can be seen in accidental or inflicted trauma. A spiral fracture may occur by a twisting mechanism. An accidental spiral femur fracture is seen in toddlers who plant 1 foot as they are running and fall as they change their direction of motion. A torus fracture is a mild fracture caused during a sudden axial compression as with a fall on an outstretched arm from playground equipment. A transverse fracture may occur as a result of a direct blow perpendicular to the bone. One example is a 2-year-old child who was playing in the backyard in the middle of a construction site. The child pulled a large 10 cm \( \times \) 10 cm panel of wood down causing a transverse fracture of the femur. In the same month, a 2 year old was punched in the leg by his father for running in the house. A nearly identical femur fracture resulted.

In case 1, the infant presented with acute rib fractures. The explanation the mother gave for the rib fractures was an incident 3 weeks earlier when the father allegedly closed a door on the mother while she was holding the infant against her chest. The infant being squeezed between the door and the mother’s chest produced anterior-posterior compression and bending forces against the infant’s rib cage. However, although the biomechanics may seem plausible, the timing does not.

<table>
<thead>
<tr>
<th>Loading force</th>
<th>Fracture type</th>
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<tbody>
<tr>
<td>Torsional</td>
<td>Spiral</td>
</tr>
<tr>
<td>Compressive/axial</td>
<td>Buckle/torus</td>
</tr>
<tr>
<td>Bending</td>
<td>Transverse</td>
</tr>
<tr>
<td>Compressive + bending</td>
<td>Oblique</td>
</tr>
<tr>
<td>Shear/tensile</td>
<td>Classic metaphyseal lesion</td>
</tr>
</tbody>
</table>

Intrinsic bone factors are equally important. Certain locations within the bone carry higher specificity for abuse because of the bone’s intrinsic properties (ie, metaphysis vs diaphysis). The metaphysis has a cortex that is thinner and more porous, overlying trabecular bone that is spongy, making it weak compared with the diaphyseal portion of bone. The diaphysis has a much stronger cortex, with dense underlying bone. The transition zone between diaphysis and metaphysis is an inherently weak region, making it also susceptible to fracture [2,14].

DATING FRACTURES
Occasionally, dating the fracture assists with determining the plausibility of the history provided. Child abuse experts are frequently asked to narrow the timing of the injury to evaluate allegations of physical abuse or neglect. Prosser and colleagues [15] reviewed the literature in an attempt to create uniformity in a timetable of radiologic findings in children’s fractures. However, because of the number of studies that included infants and young children, in conjunction with the lack of a uniform language, dating of fractures in children remains a difficult task. Those with a prosecutorial focus often need a much narrower time window than can be offered. Time frames for dating fractures are usually on the order of weeks, not days. For example, soft callus formation is seen between 10 to 21 days after injury, and hard callus may exist as early as 14 days and may persist until 90 days after injury. The studies that have documented radiographic healing processes have dealt with immobilized bones and may not be applicable [16] to fractures caused by abuse that are often left unattended for long periods of time [17]. Repeated injury to an existing abusive fracture extends the time frames and alters the appearance of some radiographic findings. If the abused child expires, histologic examination of the fracture may elucidate the timing of the injury or show a chronic fracture or one that has been repeatedly injured [18].

CORRELATION OF ABUSE BY FRACTURE LOCATION
Some bones carry a higher specificity for abuse (eg, ribs, scapula, sternum, acromion), but no fracture is pathognomonic. A meta-analysis reviewed 32 studies that sought to determine whether some bones are more often associated with abuse than with other mechanisms of accidental injury. Kemp used strict criteria for confirmed abuse that required perpetrator admission, witnessed event, or abuse confirmed through court proceedings or by a multidisciplinary team. Abused children commonly had more than 1 broken bone. Rib fractures (71%) carried the highest probability for abuse. Probability of abuse was lower in femur fractures and skull fractures (28% and 30% respectively), although the probabilities increase in nonambulatory children. The probability of confirmed abuse was higher in the humerus (48%), especially for midshaft fractures [19].

Preambulatory children rarely sustain extremity fractures. Long bone fractures are strongly associated with abuse in infants less than 12 months of age.
However, children who are capable of rolling and crawling do sustain fractures of the skull with a short fall. A simple, linear parietal skull fracture is a familiar sequela after a fall from a bed or couch onto a hardwood or tile floor. Skull fractures can be difficult to judge. In cases of both inflicted and accidental trauma, the parietal bone is the most frequently fractured part of the skull. Characteristics of abusive fractures include those that are multiple, bilateral, and that cross suture lines.

Studies have shown the high specificity of rib fractures for abuse in a young child. One study found that the positive predictive value of rib fractures being inflicted in children less than 3 years old was 95%. Another group retrospectively studied 39 infants with rib fractures: 32 were caused by abuse, 3 were caused by major accidental trauma, 1 was the result of birth trauma, and 3 were attributed to bone fragility. The causes of bone fragility were OI and rickets (1 nutritional and 1 ex-preemie). Although rib fractures may strengthen an argument for abuse, alternative causes must be contemplated. Rib fractures are not obvious on plain chest films, even on follow-up after the standard 2 weeks. In a postmortem study of 11 infants with 84 rib fractures, only 30 healing fractures were discernible on the SS. Twenty-three rib fractures were acute and incognito; a reminder that a repeat SS at 2 weeks follow-up may increase the yield in identifying occult injuries.

DISORDERS OF COLLAGEN FORMATION

OI

OI is a well-recognized inherited clinical entity characterized by bone fragility and predisposition to fractures. The milder forms of OI, types I and IV, may be considered in the differential diagnosis of children with multiple fractures or fractures after seemingly minor trauma. Both OI and abuse may present with multiple fractures in various stages of healing and children with OI may also be abused. Some of the physical findings of OI are not apparent in the young infant, so genetic testing should be performed when it cannot be ruled out. The incidence of OI has been estimated as 1:10,000 to 1:20,000 live births. In children less than 3 years old, abuse is 24 times more likely to be the cause of a fracture than OI. A careful history of the injury should be sought. Ideally, a family member will have witnessed an apparently minor trauma and would be able to explain the details of the timing, location, and other circumstances. The family history may reveal other family members who have sustained fractures after minor trauma, are short or deaf, and who have blue sclera. However, the genetic mutation in collagen is more often de novo and the family history is negative. Physical examination may reveal classic signs of OI in infancy. However, many of the examination findings in mild forms of OI are not evident in infancy. The hearing impairment, short stature, bone deformities, and dentinogenesis imperfecta associated with OI may not be evident until well after infancy but should be inquired about in the family. Alternatively, blue sclerae are a common finding in many infants because their sclerae are thinner and reveal the underlying uveal
tissue. Review of radiographs may raise suspicion of OI if osteopenia is appreciated in the long bones or more than 10 wormian bones on the skull radiograph are noted [26]. With normal bone density, OI is excluded, but bone density can be difficult to ascertain. In the absence of abuse, classic metaphyseal lesions and symmetric rib fractures are rarely seen with OI [27]. In the absence of clinical or radiologic signs of OI or family history in an infant, it has been estimated to have a likelihood of 1 in a million of being the cause of a fracture.

Ninety percent of OI cases are the result of a mutation in 1 of 2 genes: COL1A1 or COL1A2. These genes are the blueprints for the production of type IA1 and IA2 procollagens. A trimer of the 2 types of procollagen forms collagen, the basic structural protein of bone. If there is an error (mutation) in the blueprint or processing of collagen, the structure of bone is not sound. There may be mutations that result in shortened or disfigured proteins; either may lead to bone fragility. The disfigured proteins cause more instability to bone than do the shortened proteins [28,29].

There are 2 types of laboratory tests that are used to diagnose OI: fibroblast tissue culture ($1350) or serum DNA sequencing ($2300), and each identifies about 90% of patients with OI [30,31]. The fibroblast culture may have a

<table>
<thead>
<tr>
<th>OI type</th>
<th>Possible clinical features in infants</th>
<th>Incidence</th>
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</thead>
<tbody>
<tr>
<td>I</td>
<td>Blue sclera</td>
<td>4:100,000</td>
</tr>
<tr>
<td></td>
<td>Wide fontanelle-delayed closure</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Wormian bones</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Normal or near-normal stature</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Thin, easily bruised skin</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Fractures are rare in neonatal period</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Normal teeth</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Mild osteopenia with femoral bowing</td>
<td></td>
</tr>
<tr>
<td>II</td>
<td>Lethal in utero</td>
<td>1:100,000</td>
</tr>
<tr>
<td>III</td>
<td>Born with multiple fractures</td>
<td>1:100,000</td>
</tr>
<tr>
<td></td>
<td>Diagnosed on prenatal ultrasound</td>
<td></td>
</tr>
<tr>
<td>IV</td>
<td>White sclera</td>
<td>4:100,000</td>
</tr>
<tr>
<td></td>
<td>Short stature</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Triangular facies</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Fractures are seen in neonatal period</td>
<td></td>
</tr>
<tr>
<td></td>
<td>DI</td>
<td></td>
</tr>
<tr>
<td>V</td>
<td>Fractures in the first year of life</td>
<td>7 reported cases</td>
</tr>
<tr>
<td></td>
<td>White sclera; normal teeth</td>
<td></td>
</tr>
<tr>
<td>VI</td>
<td>First fractures documented after age 4 months</td>
<td>8 reported cases</td>
</tr>
<tr>
<td></td>
<td>No wormian bones or DI</td>
<td></td>
</tr>
<tr>
<td></td>
<td>White sclera or faintly blue</td>
<td></td>
</tr>
<tr>
<td>VII</td>
<td>Multiple fractures at birth; rhizomelia</td>
<td>8 reported cases</td>
</tr>
<tr>
<td>VIII</td>
<td>Multiple fractures at birth</td>
<td>5 reported cases</td>
</tr>
</tbody>
</table>

OMIM #166200, #166210, #166220, #259420, #610682, #610915, #610967, #610968.

Abbreviation: DI, dentinogenesis imperfecta.
greater usefulness in mild cases with low suspicion; however, it requires a skin biopsy. If there are enough clinical signs, including a family history, a geneticist may make the diagnosis clinically or order the serum DNA sequencing. Both tests give results in approximately 6 to 8 weeks. In practice, DNA sequencing is the test of choice because it is technically much easier to obtain and is less traumatic to the child than a skin biopsy.

Testing is often requested by non–medically trained professionals. Testing may result in few positives, but even a single positive result is significant for the family affected. In a study by Steiner and colleagues [32], 6/48 children had abnormal collagen synthesized by dermal fibroblasts. In 88% of the cases, testing was requested by child protection service workers, attorneys, or courts.

One might expect a greater probability of a positive OI test in children referred for DNA analysis through a geneticist. Byers and colleagues [25] reviewed 262 children who underwent genetic testing during investigations for abuse. Of those, only 11 (4.2%) tested positive for OI. Three of the 11 children had no indication of OI apart from their fractures. The study found that, despite the children being referred by doctors with training in clinical genetics, the predicted incidence of OI was no higher than expected if there had been no clinical screening. One reason for the low percentage of positive OI tests is that a significant number of children were referred by CPS or by the court, and the geneticist’s role was to facilitate testing. In addition, OI is a difficult diagnosis to exclude clinically in infancy. The study did validate the exclusion of children with other findings of abuse (eg, burns, bruises, bites) from genetic testing.

Ten percent of OI cases are not detected by current genetic testing. A geneticist, after reviewing a case, can render a clinical diagnosis if there is enough supporting evidence.

Not considering the diagnosis of OI can have devastating emotional effects on families. There are reports from patients initially diagnosed with nonaccidental trauma who were subsequently diagnosed with OI [4,33]. Kocher, an orthopedic surgeon, reported on his ongoing review of misdiagnosed child abuse at the annual meeting of the American Academy of Orthopaedic Surgeons in 2007 [34]. Kocher and colleagues gathered retrospective data from self-reported questionnaires of families of children formally diagnosed with OI [34]. The 33 families (who were affiliated with Protect our Families, an organization that represents parents who have been accused of child abuse) answered a questionnaire about their children’s presenting symptoms. The children presented with clinical symptoms of fractures at ages 1 to 23 months (average 7.1 months). There was no known accident in 14 of the 33 children. Multiple fractures (average 7) were found in 28/33 children, often in multiple stages of healing. Twenty-three of the children were removed from their families during the investigation of the fractures. Nearly one-third of the families sued the doctors, hospitals, or CPS. It was not stated whether their lawsuits were successful [34]. Although the removal of the child from the home can be difficult, it is the goal of CPS to prevent another serious injury or the death of the child.
There are descriptions of new types of OI, named types V, VI, VII and VIII, which are subtypes of IV based on the Silence classification system. These newly described types of OI are rare and have unique clinical, histologic, and radiographic features (see Table 2) [35]. Types V to VIII exhibit severe bone deformities. Patients with type VI all had vertebral compression fractures [36]. Type VII is an autosomal recessive form of OI localized to an isolated group of 8 affected family members in Quebec [37]. There are 5 reported cases of type VIII, another form of OI that is autosomal recessive, who exhibit severely affected bones [38]. It is believed that types V, VI, and VII may comprise approximately 8% of patients with OI [24].

Copper Deficiency

Acquired

Acquired copper deficiency has been proposed as predisposing an infant to multiple fractures; however, documented cases are rare. Standard formulas now have adequate copper and there are no reports of a breastfed infant who has suffered from nutritional copper deficiency. Ex-premies are at increased risk, manifesting symptoms at ages 6 to 12 months with a history of prolonged parenteral nutrition. Neutropenia (≤1.5 × 10^9/L) is the first and most common sign of copper deficiency, but a sideroblastic anemia resistant to iron treatment is also prevalent [39]. Less often (11/51 in 1 study), infants with acquired copper deficiency present with fractures [40]. There have only been a few reported cases in full-term babies. Radiographs show changes similar to rickets, with symmetric cupping and fraying of the metaphyses and osteopenia [41]. Low serum copper (<0.45 mg/L) and ceruloplasmin concentrations (<200 mg/L) may not always be evident, but of the 2 values, ceruloplasmin is the first to be reduced [39,42].

Infants who present before the age of 1 year were being fed a nonstandard formula or had a history of prolonged parenteral nutrition. The ex-premature infants present earlier with a copper concentration less than 0.33 mg/L. The full-term infant presents later at approximately 8 months old. Common presenting signs, in addition to the abnormal laboratory values mentioned earlier, are psychomotor retardation hypotonia, pallor, hypopigmented skin and hair, and prominent scalp veins [41].

Menkes disease

Menkes disease (MD) is an X-linked recessive disorder of copper metabolism that is usually lethal by 3 years of age, although there are milder forms. The classic form of MD, with an incidence of 1:300,000, should not be confused with abuse because the fractures are the result of diffuse osteoporosis and there are concomitant clinical features [43]. The radiologic findings overlap with copper deficiency. Seizures develop by 3 months of age and, at about the same time, the diagnostic steel-wool hair is noticed. The mildest form is occipital horn syndrome (OHS), named for characteristic bony protuberances from the occipital bone that appear after age 1 year. Patients with OHS have chronic diarrhea and urinary tract infections [44].
DISORDERS OF MINERALIZATION

Rickets and Vitamin D Deficiency

The clinical and radiographic signs of classic rickets are apparent to an experienced clinician and pediatric radiologist actively considering it in the differential diagnosis. Radiographic findings of rickets should be symmetric and are first evident with cupping and fraying of the costochondral junctions and long bone metaphyses. Subperiosteal new bone forms in both rickets and from trauma. Other findings used to help differentiate the 2 entities are generalized osteopenia and insufficiency fractures, found in rickets but not in child abuse [45]. Dr Paul Kleinman [46] recently acknowledged that a small metaphyseal bony fragment in rickets can look like a classic metaphyseal lesion caused by abuse. The diagnosis may be unclear if the infant has been partially treated with vitamin D. Clinical and laboratory evaluation should clarify this rare scenario. Strouse [47] implores us to create prospective studies to further investigate the question of congenital rickets: “There is no concrete evidence in the literature that vitamin D deficiency in infants younger than 6 months of age renders them susceptible to the same types of fractures as have been accepted to bear high specificity for child abuse” [47].

As was pointed out in a recent commentary, the medical definition of rickets involves radiologic findings. Therefore, deficiency of vitamin D without radiographic signs of rickets is not rickets. There are 3 radiologic findings in rickets: osteopenia, physeal and metaphyseal changes, and osteomalacia [48]. However, in a recent study of healthy infants and toddlers, 7.5% of children who were deficient in vitamin D (25-hydroxyvitamin D [25OHD] <20 ng/mL) had rachitic changes on radiographs, and 32.5% showed demineralization [49].

As with OI, there are anecdotal and case reports of infants supposedly misdiagnosed with abuse-related fractures when vitamin D deficiency rickets was apparent [50–52]. Most of these reports by individuals who frequently testify provide selectively sparse and inaccurate details of history, tests, and images [53]. However, many of the children in these cases had unexplained bruising, a finding not commonly attributable to vitamin D deficiency. In 1 case, a 6-week-old infant presented with a seizure and widened sutures without mention of evaluation for intracranial hemorrhage or retinal hemorrhages [54]. The clinical record should be evaluated in its entirety with an attempt to explain all findings, some of which may be highly associated with inflicted trauma. Carole Jenny [55], in an extensive report on her experience, found no children (out of several thousand) who were thoroughly evaluated for maltreatment to have rickets or vitamin D deficiency when they presented exclusively with fractures [55].

Vitamin D deficiency and associated rickets is a significant issue in infants who are solely breastfed, especially by mothers with inadequate ultraviolet B radiation and diet. The American Academy of Pediatrics recommends vitamin D supplementation of 400 IU for all infants and children, beginning soon after birth [56]. Testing for rickets must be considered if dietary history, clinical examination, or radiographs raise suspicion. The serum phosphorus level is usually less than 4 mg/dL, but the alkaline phosphatase level is often increased for age in both rickets and
fractures. A serum 25OHD less than 20 ng/mL is suggestive, and parathyroid hormone levels reactively increase only in older infants and children in the presence of rickets.

**Osteopenia of Prematurity**

Osteopenia of prematurity is a well-described clinical entity with a peak incidence of 6 to 12 weeks postnatal age known to predispose infants to fracture. Laboratory studies of calcium, inorganic phosphate, and alkaline phosphatase are routinely performed beginning at 6 weeks postnatal age to screen for early rickets while the infant is in the NICU. Rickets tests should be repeated on a weekly or bimonthly basis. Birth records and laboratory studies should be obtained when considering whether an infant’s fractures may be the result of prematurity. Premature infants in the very low birth weight (VLBW; birth weight <1500 g) and extremely low birth weight (ELBW; birth weight <1000 g) groups have increased susceptibility to fractures for a variety of reasons, all stemming from poor bone mineralization. For various periods of time, these infants are sustained on parenteral nutrition [57], are deprived of physical stimulation [58], and are given diuretics to treat lung disease associated with prematurity. In addition, 80% of an infant’s calcium and phosphorus deposition occurs in the third trimester, leaving preterm infants seriously deficient in total body store [57]. It has been estimated that up to 24% of VLBW infants sustain a fracture in the first year of life because of osteopenia of prematurity [59,60].

**Nutritional deficits**

When evaluating a preemie with suspected osteopenia, information about calcium, phosphate, alkaline phosphatase, or vitamin D levels ordered in the newborn period should be compared with known normal values for term and preterm babies. Backstrom and colleagues [61] showed that using a minimum alkaline phosphatase level of 900 IU/L and a maximum inorganic phosphate level of 1.8 mmol/L detects all premature infants with osteopenia. However, nutritional supplementation does not overcome all of the influences leading to weaker bones of the premature infant. Chronic lung disease and related use of diuretics further depletes the infant of calcium stores. An orthopedic group assessed 247 VLBW infants for rickets and discovered that 10.5% sustained fractures at around 2.5 months of age. Infants averaged about 4 fractures each, most commonly in the ribs. As mentioned earlier, risk factors for sustaining a fracture included parenteral nutrition, diuretic use, cholestatic disease, chest physical therapy, and passive range-of-motion exercises. However, nutritional supplementation does not overcome all of the external influences leading to weaker bones of the premature infant [60,62].

**Mechanical deprivation**

Mechanical stimulation occurs in the third trimester as the infant kicks and pushes against the uterus. The stress and strain on the bones induced by movement stimulates bone growth and development [63]. The immobility and lack of external (uterine) stress causes increased bone resorption and
demineralization. Premature infants do exhibit some adaptive catch-up mineralization when bone density increases more rapidly than in full-term infants from 40 to 60 weeks after menstruation, so that, between 65 and 100 week after menstruation, all infants have comparable bone mineral content [64]. Therefore, up to 65 weeks after menstruation must be considered as a time for increased susceptibility for fractures in preterm infants. However, each case should be considered for its unique risk factors [60].

Chronic lung disease
Another consideration in ex-preemies may be prolonged mechanical ventilation and use of diuretics. In the past, steroid use further exacerbated bone demineralization, but steroids are no longer commonly used in premature infants as part of standard therapy for chronic lung disease [60].

BIRTH-RELATED FRACTURES
The forceful process of delivery is a common cause for fractures in the newborn period. It is important to understand the characteristics of such birth trauma. Commonly fractured bones include clavicle, humerus, and femur. Skull fractures and rib fractures have been documented after traumatic vaginal deliveries [65]. Cumming studied the radiologic characteristics of 23 neonates with clavicle, humerus, and femur fractures that resulted from delivery and found that the earliest calcification occurred at 7 days after birth, but all had radiologic evidence of periosteal reaction by 11 days [66]. Some parents may have videorecorded the infant during the first examination in the newborn nursery. At times it is helpful for the consultant to view the video because it may provide insight as to whether a fracture may have been present at birth (ie, asymmetric moro reflex may imply a clavicle or humerus fracture).

NONFRACTURE MIMICS
Scurvy
Scurvy is a nutritional vitamin deficiency rarely seen in children from the United States or other developed countries. There are few case reports in infants from developed countries in recent decades [67–69]. The usual age at presentation is 6 to 24 months, and the affected infant seems ill, with significant pain leading to pseudoparalysis. Surface examination may reveal petechiae, ecchymoses, and mucosal hemorrhage. It is a clinical and radiologic diagnosis in which a diet deficient in vitamin C should be documented [70]. Getting a detailed diet history at presentation eliminates confusion later. Practitioners should be alert for future cases of congenital scurvy in solely breastfed babies whose mothers have a restricted diet. The recent case reports have exhibited unique diets: 1 ketogenic diet to control a severe seizure disorder and 1 prescribed by the Church of Scientology, both devoid of fruits and vegetables. Vitamin C levels are not routinely sent because of the protein’s lability. Vitamin C is heat labile and has been deficient in infants whose milk was boiled. Radiologic findings in scurvy are subtle but distinct from abusive injuries. Cortices are notably thin and the
epiphyses appear osteopenic with denser outlines. During the repair phase of scurvy, as vitamin C is replenished, subperiosteal and soft tissue hemorrhage appear similar to healing trauma. In addition, the metaphyseal irregularities are similar to those seen in abused children.

**Congenital Syphilitic Periostitis**

Prenatal clinical testing does not rule out congenital syphilis. Untreated primary syphilis infection is almost always transmitted to the fetus. About two-thirds of infants are asymptomatic at birth, but untreated infants develop symptoms within weeks to months. The common early signs include jaundice, hepatosplenomegaly, transaminitis, anemia, and thrombocytopenia, but radiologic findings may precede clinical manifestations in up to 20% of those asymptomatic infants [49]. Radiologic findings are diffuse but irregular and commonly involve the proximal tibia, distal femur, and humerus [31]. Bony lesions show metaphyseal erosions or rat bite lesions, which may be confused with classic metaphyseal lesions.

**Vitamin A Toxicity and Other Causes of Periosteal Reactions**

A careful history of diet, medications, and supplements may alert the treating physician to the possibility of hypervitaminosis A in the face of pathologic periosteal bone formation. Additional radiologic findings with excess vitamin A intake include premature closure of the epiphyses and cortical hyperostosis, which occurs after 6 months of age [71]. Other causes of periosteal reactions include Caffey disease, prostaglandin administration, physiologic, leukemia, and osteomyelitis. The children with excess vitamin A and Caffey disease should have normal metaphyses and no fractures [42]. Osteomyelitis can appear as metaphyseal lesions, but not corner fractures, and young infants may not have systemic signs of infection. Osteomyelitis has been shown in the setting of fatal child abuse [72].

**TEMPORARY BRITTLE BONE DISEASE**

Although there has been controversy around a condition called temporary brittle bone disease (TBBD), there is no accepted medical evidence that such an entity exists. The impetus for such a condition seems to be the legal motivation to explain multiple fractures in a young infant that spontaneously resolve when a child is removed from the home environment. Paterson [73] first described this hypothetical entity of transient copper deficiency. The fractures sustained by the 39 infants in the article had fractures highly specific for abuse, such as ribs and metaphyseal lesions. There was little mention of the results of evaluations for nonaccidental trauma in the infants. The retrospective review did not adhere to any scientific method. A second group has supported a similar idea of TBBD in which the bones are briefly demineralized, and reported 26 cases attributed to decreased and restricted fetal movement, as reported by the mothers. Again, there was no mention of the evaluations for inflicted injury. Their methods for measuring bone density have not been validated. Many
experts from a wide background of subspecialties have dissected TBBD and exposed it as a sham [74–76].

**IMAGING OF FRACTURES**

The complete medical assessment of suspected child physical abuse cases was delivered in a clinical report by Nancy Kellogg in 2007 [77]. Studies have shown that use of the SS in conjunction with bone scinitigraphy (BS) is the best method of detecting occult fractures in a young child [78].

**SS**

When considering radiation, the SS is a low quantity. It is recommended that a SS be done on all children less than 2 years old with a suspicious injury or with any fracture. The current recommended views are skull, chest (including oblique views), humeri, forearm, femurs, lower legs, hands, feet, cervical and lumbar spine, and pelvis. A babygram (single view of the entire skeleton) is suboptimal and should never suffice as a method for detecting occult fractures. The yield on SSs revealing other healing fractures is significant. A follow-up SS 2 weeks after the initial study increases the yield of additional findings [79]. Kleinman and colleagues [80] detected new findings in 61% of cases in which a repeat SS was performed. When head trauma is suspected, a computed tomography (CT) scan of the head is the preferred method of detecting skull fractures and underlying acute injury. The three-dimensional reconstruction of the head CT may help delineate the skull fracture.

**Pros:** detects epiphyseal, metaphyseal, and skull fractures

**Cons:** misses acute and healing rib fractures.

**Bone Scan**

Acute rib fractures may be missed on a chest radiographs. A repeat chest radiograph in 10 to 14 days may reveal some, but not all, occult fractures. Bone scintigraphy should be performed when the suspicion of abuse is great and a complete assessment is needed [81].

**Pros:** detects acute and healing occult rib fractures

**Cons:** requires intravenous line placement, sedation, radiation exposure; findings are nonspecific for trauma (may be positive in areas of increased metabolic demand (ie, infection).

**VARIABILITY IN DIAGNOSING CHILD ABUSE**

Knowledgeable opinions are variable, even among child abuse experts [82]. What constitutes medical certainty, the standard set in court for an expert opinion? Child abuse specialists use varying degrees of vague language to impart their wisdom. The terms possible, probable, consistent with, indicative of, and more likely than not are frequently used in medical assessments. Less frequently used are terms like not concerning for, definite, or diagnostic of inflicted injury. It is important to consider the choice of language and how it may relate to the
burden of proof in each type of hearing. In a civil hearing, in which dependency
court decides who will care for the children, the burden of proof is a preponder-
ance of evidence, whereas in criminal court the accused may be on trial for conse-
quences that may include a death sentence or life in prison. There, the burden of
proof is much greater; it must be beyond a reasonable doubt.

CASE RESOLUTIONS
Case 1: the calcium, alkaline phosphatase, and phosphorus were within normal
limits for age. The opinion of the pediatric radiologist was that the bones
appear to be of normal density and did not appear suspicious for any metabolic
disease or vitamin deficiency. The infant was otherwise thriving on an appro-
priate standard formula, which provides approximately 400 IU of vitamin D
daily. Multiple altercations between the parents were witnessed by nurses
and staff during the infant’s hospitalization. The father was arrested for assault-
ing the mother outside the hospital. No further testing was done. The infant
was placed in protective custody and has sustained no further fractures.

Case 2: on examining the child, blue sclerae were noted. Further history re-
vealed an autosomal dominant pattern of fractures with minor trauma on the
father’s side. The child was referred to genetics clinic where the clinical diag-
nosis of OI was made and DNA sequencing confirmed a null mutation in
the COL1A1 gene. OI type I was confirmed. Allegations of neglect were
substantiated because of the delay in seeking medical care.

Case 3: because of the infant’s well-documented history of severe rickets, the
team reasoned that much less force would be required to cause fractures in this
susceptible infant. As an outpatient, the infant was being treated with 12,000 IU
of vitamin D daily. It would have been interesting to check a 25OHD level on
the infant as a measure of compliance with therapy. The skull fracture was
caused by an impact trauma to the head that was unexplained, but believed
to have required less force given the history of craniotabes. It is often difficult
to know whether neglect or abuse has played a role. However, the head CT
and ophthalmologic examinations did not reveal other signs of inflicted trauma.

SUMMARY
It is important for the general pediatrician to consider abuse in the differential diag-
nosis each time a child presents with a fracture. When appropriate, the primary
care doctor should order ancillary tests to rule out conditions that may predispose
the young child to fracture, interpret the results of the tests, and refer the case to
a child abuse specialist if expert opinion is sought. Child abuse specialists may be
used to objectively evaluate cases of suspected abuse and neglect [83]. General
pediatricians should not be asked to perform forensic examinations on children
they have known and parents they have befriended. Knowingly or not, their opin-
ions are biased. Suspected cases should be referred to forensic centers by law
enforcement, CPS, and generalists.

Fractures and many other forms of abuse must be evaluated in the consider-
atation of broad differentials, but ordering of tests should be judicious. The
specialist must take a careful history, perform the forensic examination, and consider prudent testing. When evaluating the history and injury, consideration must be given to the biomechanics of both the potential forces involved and the tissues injured. Home investigations are done on families after a fractured bone cannot be explained in a way that is biomechanically rational. The need to protect children must be balanced with the emotional turmoil of disrupting families or making false allegations. The child abuse expert must impart medical wisdom to law enforcement, attorneys, and CPS, keeping in mind the burden of proof in each court. Child abuse and pathologic disease are not mutually exclusive causes of fractures in young children. Children with known disability may be at greater risk for victimization by their caregivers [84].

Future efforts in prevention of child physical abuse are needed. Parents need education regarding age-appropriate discipline and guidance for anger management. High-risk parents should receive training to prepare them for various scenarios that may lead to frustration, anger, and physical harm of their child. In addition, general pediatricians should continue education in detecting the warning signs and symptoms of an abused child.

TAKE-HOME POINTS

- A thorough multidisciplinary team approach is essential to every case of suspected child abuse
- Every case of suspected physical abuse should be reported to CPS and cross-reported to law enforcement
- Obtain a detailed diet history on presentation, from birth, including maternal sun exposure
- Ex-premature infants have several comorbidities that may predispose them to fractures; examine the NICU records
- Judicious testing to rule out diagnoses should be used when the pediatrician (in collaboration with the pediatric radiologist) deems it reasonable to do so
- OI can present in infancy with few signs
- Consider whether neglect played a role in the injury
- No differential diagnosis is mutually exclusive of maltreatment.

References


EVALUATION OF YOUNG CHILDREN WITH FRACTURES


