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CHAMP Quarterly Case Review on February 11, 2010

Why the case was of interest

Infants and toddlers with multiple unexplained fractures are often the victims of nonaccidental trauma. However, upon presentation it is important to rule out other medical conditions that can result in multiple fractures in children of this age group.

Case Summary

- Healthy 5 month old Caucasian male product of a normal vaginal delivery, no complications at birth
- Developmentally appropriate, exclusively breastfed
- Had previous episode of a “popping sound” in the back with x-rays at that time found to be “negative”
- Presented 2 months later with another episode of a “snap” in the left leg after breastfeeding with subsequent irritability
- Mother took child to MD who referred to ED. X-rays done revealed a non-displaced spiral fracture of the left mid-femur and a questionable old fracture of the left distal tibia
- Subsequent skeletal survey revealed an acute fracture of the left 5th rib posteromedially and prominence of the posterior aspect of multiple ribs particularly the right 4th and 5th ribs
- There was also an “osteopenic appearance” to the bones on x-ray
- Family history notable for maternal history of multiple fractures (x 3) to same wrist and shoulder dislocation x1 and 3 healthy siblings with no history of fractures
- Physical exam was unremarkable except for a subtle blue hue to the sclera of both eyes
- Multiple subspecialists were consulted including Genetics

Key Points

- Osteogenesis imperfecta (OI) is a genetic disorder characterized by fragile bones. A person born with this disorder can sustain frequent fractures throughout their lifetime.
- In addition, people with OI often have muscle weakness, hearing loss, fatigue, joint laxity, curved bones, scoliosis, blue sclerae, brittle teeth, and short stature.
- OI is caused by a mutation on a gene that affects the body’s production of the collagen found in bones, and other tissues.
- OI is variable with 8 different types described in medical literature with the types ranging in severity from a lethal form to a milder form with few visible symptoms.
- A person with mild OI may experience a few fractures while those with the severe forms may have hundreds in a lifetime.
- The number of Americans affected with OI is thought to be 25,000-50,000 with the wide range attributed to the fact that mild OI often goes undiagnosed.
- OI occurs with equal frequency among males and females and among all racial and ethnic groups.
- Approximately 35% of children with OI are born into a family with no family history of OI.

**Web resource: the Osteogenesis Imperfecta Foundation
accessed at www.oif.org**

Relevant Articles

- Carole Jenny and the Committee on Child Abuse and Neglect. Evaluating Infants and Young Children with Multiple Fractures. *Pediatrics* 2006;118:1299-1303
- Gahagan S, Rimsza, ME. Child Abuse or Osteogenesis Imperfecta: How Can We Tell? *Pediatrics* 1991; 88; 987-992
- Olney RC, Mazur JM, Pike LM, Froyen MK, et. al. Healthy Children With Frequent Fractures: How Much Evaluation Is Needed? *Pediatrics* 2008;121:890-897