Child Abuse & Neglect Tips

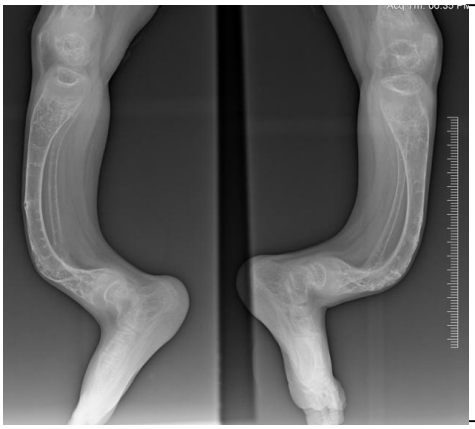
"DID YOU KNOW?"

Osteogenesis Imperfecta *is sometimes called Brittle Bone Disease?*

- Osteogenesis Imperfecta (OI) is a congenital bone disorder which means that it is present at birth. It is characterized by brittle bones that are prone to fracture. People with OI are born with defective connective tissue caused by genes that don't work properly. There is a deficiency of Type-I collagen which is an important building material for healthy bones.
- The diagnosis of OI is based on the **clinical features** and may be confirmed by collagen or DNA testing. OI can range from mild to severe and symptoms vary from person to person. To date, eight types of OI have been recognized.

What Are the Symptoms of Osteogenesis Imperfecta?

- Malformed bones
- Short, small body
- Loose joints
- Muscle weakness
- Sclera (whites of the eyes) that looks blue, purple, or gray
- Triangular shape to the face
- Barrel-shaped rib cage
- Curved spine
- Brittle teeth and other dental abnormalities
- Hearing loss (often starting in 20s or 30s)
- Breathing problems



Severely malformed leg bones due to OI and multiple fractures over time. Not only is the shape of the bone abnormal, but also the bone density and X-ray appearance.

Questions to Ask:

- ☑ Has the child been evaluated by an **expert in OI**? Generally, this is a geneticist. If you need help locating a geneticist, please contact CMDP Provider Services Unit.
- ☑ **X-ray Findings**: Is there evidence of "wormian bones", decreased bone density or other findings consistent with OI? The X-ray reading will need to be performed by a pediatric radiologist or geneticist.
- ☑ Has the infant had **metabolic testing** demonstrating evidence of calcium, phosphorous, alkaline phosphatase, or PTH (hormone) abnormalities that are consistent with bone disease?
- ☑ Are there **other suspected non-accidental trauma (SNAT) findings** (such as retinal hemorrhages or subdurals) that would ***not*** be consistent with OI??