

Child Abuse or Osteogenesis Imperfecta?

A child is brought into the emergency room with a fractured leg. The parents are unable to explain how the leg fractured. X-rays reveal several other fractures in various stages of healing. The parents say they did not know about these fractures, and cannot explain what might have caused them. Hospital personnel call child welfare services to report a suspected case of child abuse. The child is taken away from the parents and placed in foster care.

Scenes like this occur in emergency rooms every day. But in this case, the cause of the fractures is **not** child abuse. It is **osteogenesis imperfecta**, or **OI**. OI is a genetic disorder characterized by bones that break easily—often from little or no apparent cause. A person with OI may sustain just a few or as many as several hundred fractures in a lifetime.

What Is Osteogenesis Imperfecta?

Osteogenesis imperfecta is a genetic disorder. Most cases involve a defect in type 1 collagen—the protein “scaffolding” of bone and other connective tissues. People with OI have a faulty gene that instructs their bodies to make either *too little* type 1 collagen or *poor quality* type 1 collagen. The result is bones that break easily plus other connective tissue symptoms.

*Most cases of OI are caused by a dominant genetic defect. Most children with OI inherit the disorder from a parent who has OI. Some adults with very mild OI may not have been diagnosed as children. **Approximately 25% of children with OI are born into a family with no history of the disorder.** In these cases, the genetic defect occurred as a spontaneous mutation. Because the genetic defect is usually dominant—whether inherited from a parent or due to a spontaneous mutation—an affected person has a 50% chance of passing on the disorder to each of his or her children.*

Several forms of OI have been described, representing wide variations in appearance and severity from one individual to another. Types of OI range from lethal in the newborn period to very mild. Recent research has identified two new moderately severe types that do not appear to have a type 1 collagen defect. It is estimated that 50,000 people have OI in the United States. It occurs in approximately 1:10,000 births.

Below are the clinical features of the major types of OI. **Clinical features vary widely not only between types, but within types, and even within the same family.** Children with milder OI, in particular, may have few obvious clinical features of OI.

Clinical Features of Osteogenesis Imperfecta

Type I (Mild)

- Most common and mildest type of OI.
- Bones predisposed to fracture. Most fractures occur before puberty.
- Normal or near-normal stature.
- Loose joints and muscle weakness.
- Sclera (whites of the eyes) usually have a blue, purple, or gray tint.
- Triangular face.
- Tendency toward spinal curvature.
- Bone deformity absent or minimal.
- Brittle teeth possible.
- Hearing loss possible, often beginning in early teens.
- Collagen structure is normal, but the amount of collagen is less than normal.

Type II (Perinatal Lethal)

- Most severe form.
- Frequently lethal at or shortly after birth, often due to respiratory problems.
- Numerous fractures and severe bone deformity evident at birth.
- Small stature with underdeveloped lungs.
- Collagen is improperly formed.

Type III (Progressive)

- Progressive bone deformity, often severe.
- Bones fracture easily. Fractures are often present at birth, and x-rays may reveal healed fractures that occurred before birth.
- Short stature.
- Sclera have a blue, purple, or gray tint.
- Loose joints and poor muscle development in arms and legs.
- Barrel-shaped rib cage.
- Triangular face.
- Spinal curvature.
- Respiratory problems possible.
- Brittle teeth possible.
- Hearing loss possible.
- Collagen is improperly formed.

Type IV (Moderate Severe)

- Between Type I and Type III in severity.
- Bones fracture easily, most before puberty.
- Shorter than average stature.
- Sclera are white or near-white (i.e. normal in color)
- Mild to moderate bone deformity.
- Spinal curvature.
- Barrel-shaped rib cage.
- Triangular face.
- Brittle teeth possible.
- Hearing loss possible.
- Collagen is improperly formed.

Type V & VI (Novel Forms)

- Recently identified types of OI.
- At this time no collagen defect has been found.
- Characteristics are similar to Type IV OI.
- Additional Type V characteristics include:
 - Dense band adjacent to the growth plate of long bones.
 - Development of hypertrophic calluses from fracture or surgery.
 - Calcification of the membrane between the radius and the ulna.
- Type VI bones have a “fish scale” appearance when viewed under a microscope.

Other clinical features that may occur in people with all types of OI are thin, smooth skin; loose joints; low muscle strength; and excessive sweating. People with OI may also bruise easily; however, because fractures often occur with very little trauma and during normal activities, it is common for a child to have *no* bruising around the fracture site.

Is it OI or Child Abuse?

When health care professionals, child welfare workers, and law enforcement officials see an unexplained fracture in a child who appears “normal” and whose bones appear otherwise normal on x-ray, they often suspect child abuse. Tragically, however, OI is often mistaken for child abuse.

When a child has osteogenesis imperfecta:

- ✓ **Fractures may occur during ordinary activities, such as changing a diaper or burping the baby, or when an infant tries to crawl or pull to a stand. There may be no obvious indication that a fracture has occurred, other than the child crying or refusing to put weight on a limb.**
- ✓ **All types of fractures may occur, including rib fractures and spiral fractures, with little or no apparent trauma.**
- ✓ **The child may bruise easily, again with little or no apparent cause.**
- ✓ **There may be no history of OI in the family, as some cases of OI occur due to a spontaneous genetic mutation. In other cases, a parent’s case of mild OI may have gone undiagnosed.**
- ✓ **X-rays may reveal old fractures in various stages of healing that went undetected.**
- ✓ **The OI child may not exhibit the hallmark clinical features of OI, such as blue sclera, bone deformity, or brittle teeth.**

- ✓ **Infants and children with mild or moderate OI may have bones that appear normal on x-rays.**

Fortunately, there are several steps that professionals can take to help determine whether fractures are due to undiagnosed osteogenesis imperfecta.

- ✓ **Obtain a family history.** Was either parent ever diagnosed with OI or any brittle bone disorder? Do either parent, siblings, or extended family members have a history of childhood fractures, spinal curvature, brittle teeth, hearing loss, or other clinical features that might indicate that they have a mild case of OI that was never diagnosed? It is not uncommon for the severity of OI to vary even within the same family.

Look for clinical features of OI in the child--blue sclera; translucent, opalescent, or discolored teeth (even in unerupted teeth in babies); a triangular shaped face; barrel-shaped rib cage; easy bruising; thin skin; excessive sweating; and other features. **However, it is possible for children with OI to exhibit none or few of the outward clinical features.** Some signs such as dentinogenesis imperfecta, hearing loss, bone deformity and short stature are age-dependent and may not be evident in an infant. In its mildest form, OI may exhibit only as unexplained bone fractures in childhood.

Consult a health care professional who has experience diagnosing or treating children with OI. OI remains primarily a clinical diagnosis. Generally, clinical geneticists are familiar with OI diagnosis. They have available to them biochemical (collagen) and molecular (DNA) tests that can help confirm a diagnosis of OI in some situations. A skin biopsy can be analyzed to determine if the quantity or quality of type 1 collagen is abnormal. This approach identifies almost 90% of persons known to have OI. A DNA test can be done on a blood sample to try to locate the mutations that cause OI. Several hundred mutations have been identified. This test identifies about 90% of people with OI. A few individuals test positive for OI on one test and not the other. Approximately 10% of individuals with mild or moderate OI test negative for OI through collagen or DNA testing, despite having the disorder.

It has been estimated that 7% of children who have signs suggesting abuse have an underlying medical condition that explains the injuries (Wardinsky). Besides OI, other conditions that feature fragile bones and bruising include Ehlers-Danlos syndrome, glutaric acidemia type 1, hypophosphatasia, disorders of vitamin D metabolism, disorders of copper metabolism such as Menkes syndrome, and premature birth (Marlowe). OI is *not* normally associated with calcium or phosphate deficiency, so it cannot be diagnosed by measuring the levels of these substances in the blood. The national Osteogenesis Imperfecta Foundation maintains a list of orthopedists and geneticists who are reputed to have experience with all types of OI. Foundation staff can suggest appropriate professionals as needed.

Resources

Marlowe A, Pepin MG, Byers PH. Testing for osteogenesis imperfecta in cases of suspected non-accidental injury. *J Med Genet* 2002;39:382-6.

Wardinsky TD, Vizacarrondo FE, Cruz BK. The mistaken diagnosis of child abuse: a three-year USAF Medical Center analysis and literature review. *Mil Med* 1995;160:15-20.

The Osteogenesis Imperfecta Foundation, Inc., working with the Osteoporosis and Related Bone Diseases ~National Resource Center, prepared these materials to familiarize health care and social service professionals with osteogenesis imperfecta and its clinical manifestations. The Foundation's goal in disseminating this information is to help professionals differentiate between cases of child abuse and cases of OI, so that children in every situation will receive the care and intervention they need. If you are interested in learning more about OI, please contact the OI Foundation.

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