

Distinguishing Clinical Features of OI by Type

Osteogenesis imperfecta exhibits wide variation in appearance and severity. The primary symptom is frequent bone fractures. Other clinical features include short stature, bone deformity, hearing loss, scoliosis, brittle teeth, tendon laxity and cardiopulmonary complications. All features may not be found in a specific patient. Below are some of the distinguishing features.

Type I (Mild)

- Most common and mildest type of OI; few obvious symptoms.
- Reduced quantity of type 1 collagen.
- Stature may be average or slightly shorter than average when compared with unaffected family members, but within the normal ranges for age.

Type II (Lethal/Most Severe)

- Infants may die within weeks from respiratory or heart complications.
- Numerous fractures, beaded ribs, and severe bone deformity are evident at birth.
- Underdeveloped lungs, severe pulmonary insufficiency and low birth weight.

Type III (Severe)

- Most severe non-lethal form.
- Progressive bone deformity is often seen.
- Rib cage is barrel-shaped and may restrict respiration.
- Fractures are present at birth and x-rays may reveal healed fractures that occurred before birth.
- Short stature – adult height is less than 4 feet.
- Spinal curvature and compression fractures of vertebrae are common
- Mobility problems range from unable to walk to walking with an assistive device.

Type IV (Moderate)

- Between Type I and Type III in severity and height.
- Moderate fracture number and bone deformity.
- Spinal curvature and compression fractures of vertebrae are common.
- Rib cage is barrel-shaped and may restrict respiration.

Type V (Moderate)

- Similar to Type IV in appearance and symptoms of OI.
- Large hypertrophic calluses form at fracture or surgical procedure sites.
- Calcification of the membrane between the radius and ulna restricts forearm rotation.

Type VI (Moderate)

- Extremely rare; similar to Type IV in appearance.
- Distinguished by a characteristic mineralization defect-- “fish scale” bone lamellae -- seen in biopsied bone.

Type XI (Bruck Syndrome)

- Congenital joint contractures and fragile bones. Severity may be mild, moderate or severe.

Thank you to Dr. Michael Bober, A.I. duPont Hospital for Children and Dr. Francis Glorieux, Shriners Hospital for Children - Canada for reviewing this page. February 2015