Osteogenesis Imperfecta (OI)

Etiology

OI is also know as brittle bone disease. The cause of osteogenesis imperfecta is due to a mutation in the collagen.[1] The result is a marked decrease in collagen of both cortical and trabecular bone.[2] This leads to the loss of bone density and bone growth. Mutations in COL1A1 or COL1A2 results in defects in bone, dentin, skin, sclera, and tendon formation. There are many different types of OI depending on the which defects are impacted in the mutation.

Signs and Symptoms

Bone fragility is often mild to moderate with fracture rates ranging from a few fractures to several dozen fractures over person's lifetime. [3] Often there is a history of fractures that occur with little or no trauma. These can start early as infants become mobile, followed with a series of fractures throughout the toddler years. [3] Type I collagen makes up about 75% of total collagen in the adult myocardium, which increases the risk for developing complications. This includes the heart valves, chordae tendineae, annuli fibrosis, interventricular septum, aorta, and arteries.[4] Little is known about the extent of cardiovascular disease and patients with OI. There is an increased risk for heart disease related to valvular dysfunction and a dilated aortic root.[4]

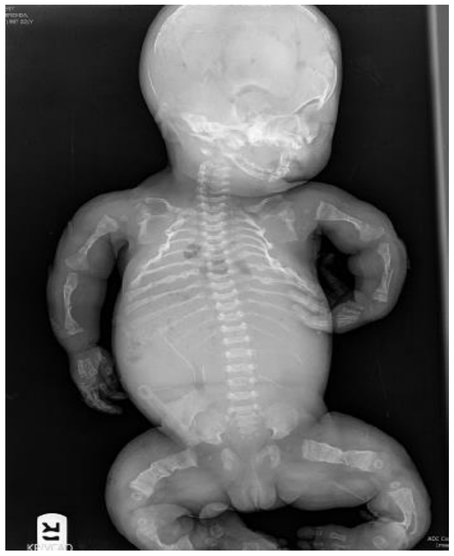

Figure 1: *Postnatal radiography showing typical features of severe lethal OI including rib fractures and crumbled bones.*[2]

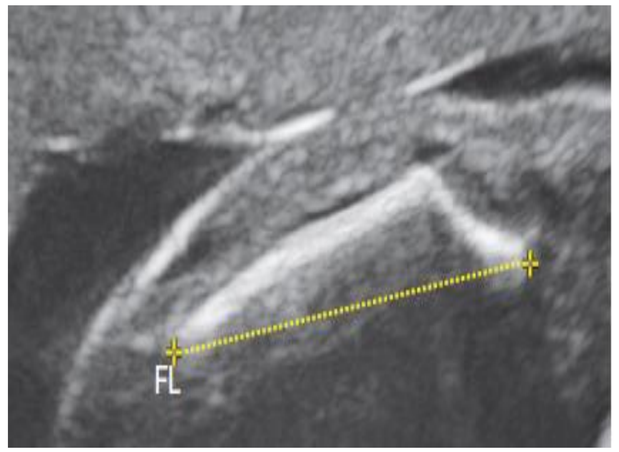

Figure 2: *US showing angulation or bowing of the long bones.*[2]

Diagnosis

Diagnosis is initially done by x-ray examination after birth. Plain radiography enables the assessment of the mechanical axis and deformity of the long bones and spine (see figure 1).[2] Severe deforming OI is often not recognized in utero, but it may be suspected after the mid-second trimester when long bone length falls away from normal length (see figure 2).[2] Both Magnetic resonance imaging (MRI) and computed tomography (CT) have a role in the identification of important basal skull abnormalities in OI such as basilar impression and invagination.[5]

Treatment

There is no cure for OI. Thus, medical therapy aims to reduce fracture rates and bone fragility while maximizing mobility and improving quality of life with the use of bisphosphonates, orthopedic surgical interventions along with physical and occupational therapy.[6] Management should be multidisciplinary and includes rehabilitation, surgical, and pharmacologic treatment. The degree of intervention needed depends on the severity of the clinical phenotype.[7]

Imaging Considerations

Patients with OI have less bone density which may require the radiographic technologist to lower the kilovoltage to compensate for the loss of bone quality.[8]

References
1. Shaker JL, Albert C, Fritz J, Harris G. Recent developments in osteogenesis imperfecta. *F1000Research*. July 2015. doi:10.12688/f1000research.6398.1.
2. Krakow D. Osteogenesis Imperfecta. *Obstetric Imaging: Fetal Diagnosis and Care*. 2018. doi:10.1016/b978-0-323-44548-1.00052-8.
3. Marini J, Smith SM. Osteogenesis imperfecta. In: De Groot LJ, Chrousos G, Dungan K, et al., eds. Endotext. South Dartmouth, MA: MDText.com, Inc. 2015.
4. Ashournia H, Johansen FT, Folkestad L, et al. Heart disease in patients with osteogenesis imperfecta—a systematic review. Int J Cardiol. 2015;196:149-157. 2010;20(5):225-231.doi:10.1016/j.paed.2009.11.008
5. Arundel P, Bishop N. Diagnosing osteogenesis imperfecta. *Paediatrics and Child Health*.
https://doi.org/10.1016/j.ijcard.2015.06.001.
6. Tsimicalis A, Boitor M, Ferland CE, et al. Pain and quality of life of children and adolescents with osteogenesis imperfecta over a bisphosphonate treatment cycle. European Journal of Pediatrics. 2018;177(6):891-902. doi:10.1007/s00431-018-3127-9.
7. Harrington J, Sochett E, Howard A. Update on the Evaluation and Treatment of Osteogenesis Imperfecta. Pediatric Clinics of North America. 2014;61(6):1243-1257. doi:10.1016/j.pcl.2014.08.010.
8. Eisenberg RL, Johnson NM. *Comprehensive Radiographic Pathology*. 6th ed. St. Louis, MO: Mosby; 2012.