Osteogenesis Imperfecta

Osteogenesis imperfecta is a genetic bone disorder leading to brittle bones, susceptibility to fracture and other manifestations such as hearing abnormalities and dental imperfections. It can be caused by a variety of gene defects, but the most common presentation is inherited in an autosomal dominant manner and yields a defect in Type 1 Collagen production.



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Pathophysiology

Autosomal Dominant
Domino
The most common presentations of osteogenesis imperfecta are inherited in an autosomal dominant manner.

Decreased Type I Collagen Production
Down-arrow Type (1) Wand Cola-gem
People with osteogenesis imperfecta (OI) have defective connective tissue, as they are deficient in Type 1 collagen. In the most common form of OI, there is an issue with the amino acids stabilizing the collagen triple helix, leading to decreased production of Type 1 collagen.

Symptoms

Phenotypically Diverse
Phoenix-types Diverse presentations
OI is known for having phenotypically diverse presentations, as there are over 15 variations of the disease. Some patients may show severe symptoms and major deformities while others may have minor hearing problems.

Brittle Bone Disease
Breaking/Flaking Bones
This is a disease of brittle bones, as type 1 collagen is an integral part of bone structure. Patients may show brittle bones, have multiple fractures before or during birth, and can even suffer from limb shortening from multiple fractures to the same area.

Fractures from Minimal Trauma
Fracture from Mini Trauma-spike
As bone structure is compromised, patients can develop fractures from minimal trauma.

Confused with Child Abuse
Suspicion of Child being Abused
In severe presentations of OI, patients can develop fractures as children from minimal trauma. When presented to the physician, multiple, various fractures can often be a tip-off of child abuse. Thus, OI can sometimes be mistaken for abuse without a thorough history.

Hearing Loss
Deaf-guy
Hearing loss is progressive in these patients and can begin at an early age. Up to 50% of patients are affected by this. The pathophysiology is similar to otosclerosis and is an audio conduction pathology.

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Dental Imperfections

Bad teeth

Commonly, patients develop dental imperfections with this disease, due to a lack of dentin. Findings include obliterated pulp cavities, opalescent teeth, and constricted coronal-radicular junctions.

Blue Sclera

Blue-eyed Skull

As type I collagen formation is defective, the sclera of the eye is much thinner than normal. Because of this, the underlying choroidal veins show through, giving the eye a blue or sometimes grey color.

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