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Editorial

Osteogenesis Imperfecta

Aakshi Kainthola*

Department of Life Sciences, Graphic Era Deemed to be University, Dehradun, India

EDITORIAL

Osteogenesis imperfecta (OI) is a category of bone-related hereditary diseases. The word "osteogenesis imperfecta" refers to a defect in the creation of bones. Bones in people with this illness are prone to breaking (fracture), often as a result of minor trauma or for no apparent reason. Multiple fractures are common and can occur even before birth in extreme circumstances. Milder cases may only result in a few fractures throughout the course of a person's life.There are 19 different types of osteogenesis imperfecta, ranging from type I to type XIX. Although their indications and symptoms overlap, there are several varieties that can be recognised by their signs and symptoms. Rarer varieties of osteogenesis imperfecta are defined by genetic factors. The mildest form of osteogenesis imperfecta is Type I (also known as classic non-deforming osteogenesis imperfecta with blue sclerae). The most severe form is Type II (also known as perinatally fatal osteogenesis imperfecta).Types III (progressively deforming osteogenesis imperfecta) and IV (common variable osteogenesis imperfecta with normal sclerae) of this disorder feature indications and symptoms that are in the middle of these two extremes.

Bone fractures are common in the milder variants of osteogenesis imperfecta, such as type I, during childhood and adolescence, and are commonly caused by small trauma, such as falling when learning to walk. Adults are less likely to suffer from fractures.

Causes

Mutations in one or more genes can cause osteogenesis imperfecta. Approximately 90% of cases are caused by mutations in the COL1A1 and COL1A2 genes. These genes give instructions for manufacturing proteins that go into putting type I collagen together. Mutations in the COL1A1 or, less commonly, the COL1A2 gene cause osteogenesis imperfecta type I. The amount of type I collagen produced in the body is reduced as a result of these genetic alterations, albeit the molecules produced are normal.

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Diagnosis

Medical imaging, such as plain X-rays, and symptoms are usually used to make a diagnosis. Abnormalities in all extremities and the spine can be seen on medical imaging. An OI diagnosis can be confirmed through DNA or collagen testing, but in many cases, the presence of other clinical symptoms such as blue sclera and the occurrence of bone fractures with little damage are enough to make a diagnosis. To determine the structure and quantity of type I collagen, a skin biopsy can be conducted. Although DNA testing can confirm the diagnosis, it cannot rule out the possibility of OI because not all mutations that cause OI are recognised and/or screened for. When many fractures and other diagnostic signs are present, OI type II is frequently identified by ultrasound during pregnancy.

Treatment

There is no way to stop it. Fractures can be avoided by leading a healthy lifestyle that includes exercise and avoiding smoking. Broken bone care, pain medication, physical therapy, braces or wheelchairs, and surgery are all possible treatments. To strengthen long bones, a sort of surgery that inserts metal rods into them may be performed. Bone infections are treated with the proper antibiotics and antiseptics as they arise.

Correspondence to: Aakshi Kainthola, Department of Life Sciences, Graphic Era Deemed to be University, Dehradun, India, E-mail: aakshi.kan7@gmail.com

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