

# OSTEOGENESIS IMPERFECTA

## AN OVERVIEW

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### IMPORTANT

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### Introduction

Osteogenesis imperfecta (OI) is a skeletal disease characterised by unusually fragile bones that break easily, often under loads that normal bones daily bear. This inherent weakness of the bones is due to a malfunction in the body's production of the protein collagen.

(Picture below - Blue Sclera is a sign of possible Osteogenesis imperfecta)



Collagen, of which there are at least 10 identifiable subtypes, is found in the connective tissues of the body and makes up a large portion of bone and cartilage. It is the substance that holds the tissues together, providing strength and mass to the bones. For an individual with Osteogenesis imperfecta,

either the amount of collagen being produced is too little, or the quality being produced is poor. Consequently, the bones are less dense and break easily. It is estimated that Osteogenesis imperfecta affects between 20,000 and 50,000 individuals.

### Cause

Osteogenesis imperfecta is a genetic disease, and the inheritance pattern is usually autosomal dominant. This means an affected person will have Osteogenesis imperfecta even though only one faulty gene has been genetically received. This faulty gene can originate from either parent, and it can affect either sex. Each child of an affected parent will have a 50 percent chance of developing Osteogenesis imperfecta.

Occasionally, a person will develop Osteogenesis imperfecta even though neither parent carried the faulty gene. This is called a spontaneous mutation, and an individual who develops the disease in this way will have the same chance of passing it on to a child as does someone who inherited the autosomal dominant gene.

### Types of Osteogenesis Imperfecta

There are four types of OI:

- **Type I:** This is the most common and mildest form of Osteogenesis imperfecta. It is autosomal dominant in its inheritance, but it may also result from a spontaneous mutation. People with Type I Osteogenesis imperfecta average nearly 40 fractures before puberty; however, they experience only a few fractures after puberty.

The collagen in Type I Osteogenesis imperfecta is normal, but the amount produced is less than normal. Some features of Type I Osteogenesis imperfecta include:

- Fragile bones
- Triangular-shaped face
- Blue sclera (whites of the eye) - Picture right
- Hearing loss beginning in their 20s
- Scoliosis (curvature of the spine)
- Thin, smooth skin
- Loose joints
- Low muscle tone
- Brittle teeth



- **Type II:** Affecting approximately 10 percent of individuals with Osteogenesis imperfecta, Type II is the most severe form of this disease. The result of a spontaneous gene mutation, the collagen in Type II Osteogenesis imperfecta is improperly formed.

The bones of people with Type II Osteogenesis imperfecta are extremely fragile and often have severe deformities. Type II Osteogenesis imperfecta frequently causes death at or shortly after birth.

- **Type III:** Affecting approximately 20 percent of the people who have OI. Usually the result of a spontaneous mutation, it is common for a person with this type to have experienced 100 fractures by the time he or she reaches puberty. There are often fractures present at birth and X-rays may even show healed fractures that occurred before birth. The collagen in Type III OI is improperly formed. Some of the characteristics of Type III OI include:
  - Short stature (some people only grow three feet tall)

- Sclera (whites of the eye) have a blue, purple, or gray tint
  - Soft bones that not only break easily but also bend
  - Loose joints
  - Poor muscle development
  - Barrel-shaped ribcage
  - Triangular face
  - Scoliosis
  - Poor tooth development, often causing teeth to be brittle and discoloured
  - Possible hearing loss
  - Possible respiratory problems
- **Type IV:** The severity of this type of Osteogenesis imperfecta falls between Type I and Type III. It is inherited in an autosomal dominant manner, although it can also result from a spontaneous mutation.

Fractures are most common in Osteogenesis imperfecta Type IV before puberty. The exception to this is in women, who after menopause experience resurgence in the number of fractures. Typical characteristics of Osteogenesis imperfecta Type IV include:

- Below average height
- Scoliosis (scoliosis is a sideways curve of the spine that results in an 'S' shape of the back. This is a common defect in childhood, and if discovered early, treatment may prevent it from getting worse. Treatment includes braces, casts, exercises, and corrective surgery)
- Mild to moderate bone deformity
- Triangular face
- Barrel-shaped ribcage
- Possible blue tint to the sclera (whites of the eye)
- Possible hearing loss
- Possible brittle teeth
- Loose, easily overstretched joints

Picture below - Blue Sclera is a sign of possible Osteogenesis imperfecta)



## **Diagnosis**

A practitioner can often make the diagnosis of Osteogenesis imperfecta after a thorough physical examination using the above symptoms as a guide, as well as a review of the family medical history. However, sometimes it is necessary to perform laboratory tests in order to confirm the disease. These tests will either be biochemical to test the collagen, or molecular to examine the genetic pattern of the disease. The results of the lab test often take several weeks.

## **Treatment and Management**

There is no cure for Osteogenesis imperfecta. Consequently, treatment for the disease focuses on managing the symptoms, preventing complications, and developing and maintaining bone mass and muscle strength.

Extreme care must be taken in handling patients, especially infants who have a serious condition, to keep broken bones from happening. Giving the patient magnesium oxide may lower the broken bone rate and the sweating, fever, and constipation linked to the condition.

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