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## **Chapter 19 Osteogenesis Imperfecta**

Introduction.

Osteogenesis

imperfecta (OI) or

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“brittle bone disease” is characterized by reduced skeletal mass and bone fragility. Over the past recent years, it has evolved from being a collagenopathy caused by mutations in the genes encoding type I collagen, and for which there was no medical treatment to a fascinating heterogeneous group of conditions, caused by numerous different

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book Pediatric Bone,  
made available for  
individual purchase.  
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well as the entire book,  
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## **Chapter 19, Osteogenesis Imperfecta**

Osteogenesis imperfecta (OI) is a group of genetic disorders that mainly affect the bones. The term "osteogenesis imperfecta" means imperfect bone formation. People with this condition have bones that break (fracture) easily, often from mild trauma or with no apparent



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cause. Multiple  
fractures are ...

## **Osteogenesis imperfecta: MedlinePlus Genetics**

Osteogenesis imperfecta (OI), also known as brittle bone disease, is a group of genetic disorders that mainly affect the bones. It results in bones that break easily. The severity may be mild to severe.

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imperfecta  
Other symptoms may include a blue tinge to the whites of the eye, short height, loose joints, hearing loss, breathing problems and problems with the teeth.

## **Osteogenesis imperfecta - Wikipedia**

There are 8 main types of osteogenesis imperfecta People with type I tending to have mild symptoms. People

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with types IV, V, and VI tend to have more moderate symptoms. People with types II, III, VII, and VIII tend to have severe symptoms, with type II being lethal in the perinatal period. Osteopenia and osteoporosis occurs to varying degrees.

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imperfecta (OI) |  
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imperfecta (OI) may be caused by changes (mutations) in any of several genes. OI is most commonly due to a variation (mutation) in either the collagen genes COL1A1 or COL1A2 gene, which cause OI types I through IV. The collagen genes play a role in how the body makes collagen, a material that helps to strengthen the bones.

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## **Osteogenesis imperfecta | Genetic and Rare Diseases ...**

Osteogenesis imperfecta can be caused by mutations in one of several genes. Mutations in the COL1A1 and COL1A2 genes cause approximately 90 percent of all cases. These genes provide instructions for making proteins that are used to assemble type I collagen. This type of

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collagen is the most abundant protein in bone, skin, and other connective tissues that provide structure and strength to the ...

## **Osteogenesis imperfecta - Genetics Home Reference - NIH**

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may be suspected in  
children suffering from  
osteogenesis  
imperfect. T/F. ...  
Osteogenesis  
imperfecta is a  
disorder characterized  
by: a. brittle bones b.  
soft bones c.  
mineralized bones

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multiple fractures

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during childhood can  
lead to the suspicion of  
glass bones What is  
glass bone disease?  
Osteogenesis  
imperfecta is

## **Osteogenesis imperfecta (glass bones): causes and**

...

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Osteogenesis  
Imperfecta have bones  
that break (fracture)  
easily, often from mild  
trauma or with no



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apparent cause.

Multiple fractures are common, and in severe cases, can occur even before birth. Milder cases may involve only a few fractures over a person's lifetime.

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imperfecta - Genetics  
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Osteogenesis  
imperfecta is a  
disorder characterized  
by: a. brittle bones. b.  
soft bones. c.  
mineralized bones. d.  
porous bones. A. 40.

Signs of osteogenesis

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## **Osteogenesis Imperfecta - NORD (National Organization for ...**

Osteogenesis  
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Hopkins Medicine.  
Osteogenesis  
imperfecta (OI), also

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imperfecta  
known as brittle bone disease, is an inherited disorder of the connective tissue. A child born with OI may have soft bones that fracture easily, bones that are not formed normally, and other problems.

Osteogenesis imperfecta (OI), also known as brittle bone disease, is an inherited disorder of the connective tissue.

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## **Osteogenesis**

### **Imperfecta | Johns Hopkins Medicine**

AMA Citation Chapter  
19. Pathology of the  
Bones and Joints. In:  
Kemp WL, Burns DK, ...

Overview: There are  
several different types  
of osteogenesis  
imperfecta, each one  
caused by one of  
several different  
mutations. Some of the  
mutations have an  
autosomal dominant  
inheritance pattern.

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Only type I and type II  
osteogenesis  
imperfecta, two of the  
...

## **Chapter 19. Pathology of the Bones and Joints | Pathology ...**

Osteogenesis  
imperfecta (OI) is a  
genetic disorder, which  
is sometimes referred  
to as “brittle bone  
disease.” The name,  
osteogenesis  
imperfecta, means

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imperfect bone formation. OI is named after its primary clinical feature, which involves bones that fractures easily.

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### **Biochemistry ...**

ICD-10 code Q78.0 for  
Osteogenesis

imperfecta is a medical  
classification as listed  
by WHO under the  
range - Congenital  
malformations,  
deformations and  
chromosomal  
abnormalities .

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