



# What is Osteogenesis Imperfecta?

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The Osteogenesis Imperfecta Foundation, Inc. is the only voluntary national health organization dedicated to helping people cope with the problems associated with osteogenesis imperfecta. The Foundation's mission is to improve the quality of life for people affected by OI through research to find treatments and a cure, education, awareness, and mutual support.

People with OI have brittle bones. Their bones break more easily than other people's bones. People with mild OI may only have a few broken bones in their lifetime. People with more severe OI may have a lot of broken bones.

Some people with OI are small. Some are about the same size as their friends and family. Some people with OI use a wheelchair. Some use crutches or a walker. Some walk without any help. Some people with OI have operations to have metal rods put in their bones, to help make the bones stronger. Some people with OI have trouble hearing, or have fragile teeth, too.

Kids with OI sometimes feel different from their friends. Some kids with OI might not be able to walk, run or play sports like their friends. They might have to stay home from school, or miss a fun party, because of a broken bone.

But kids with OI are just like other kids in lots of other ways. They can learn things in school, play a musical instrument, read books, play games, go to parties, use a computer, and take a vacation with their family. Adults with OI can go to college, get jobs, drive a car, get married, have children, go on trips, and live in their own house or apartment.

## How does OI happen?

People with OI have a problem with a gene in their body. Everyone has genes in his or her body. Genes send messages that tell the body how to grow, and what to do. For example, a boy with brown hair has a gene that tells the body to grow brown hair. A tall girl has a gene that tells her body to grow tall.

A baby is born with a whole set of genes, half from their mother, and half from their father. Most of the time, a person's genes work right. But sometimes a baby is born with a gene that does not work right. There is a mix-up in the gene. A mix-up in the genes is called a *mutation*.

Several of the genes that a baby is born with tell his or her body to make collagen. Type 1 collagen is a protein that makes your bones strong.

Think of collagen like the metal rods that are put in a concrete wall, or the wooden studs in the walls of your house. The hidden support structure makes the

walls strong. If there is a problem with the hidden supports, the walls are weak.

In a person with OI, the genes have a mutation, and are either making poor quality collagen, or are making too little collagen. This makes the support inside the bones weak, and makes the bones very brittle and easy to break.

### **Treatment: General Information**

Treatments for OI involve:

- Caring for the fractures.
- Providing support for weak arms or legs through an operation that puts metal rods inside the bones.
- Providing support through the use of braces.
- Physical therapy or an exercise program to develop strong muscles, and good overall fitness. Swimming is a good exercise for people with OI.
- Medicines that are used to treat osteoporosis in older adults are being studied as treatments for children and adults with moderate and severe OI.
- Good nutrition is important. Getting enough calcium and vitamin D won't cure OI, but it will help make the bones as strong as possible.
- New treatments such as gene therapy might be available in the future.

### **History of OI**

OI has affected people throughout all of history. It has been identified in an Egyptian mummy dating from 1000 BC. Historians believe that OI was probably the medical condition suffered by Ivan the Boneless who lived in 9th century Denmark. Prince Ivan, according to legend, was carried into battle on a shield because he was unable to walk on his soft legs.

Today, people involved in medical research are working to learn more about this complicated medical disorder, to find better treatments and someday to find a cure.

For more information about osteogenesis imperfecta contact:



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