

What Is Osteogenesis Imperfecta?

Fast Facts: An Easy-to-Read Series of Publications for the Public

Osteogenesis imperfecta (OI) is a disease that causes weak bones that break easily. It is known as brittle bone disease. Sometimes the bones break for no known reason. OI can also cause many other problems such as weak muscles, brittle teeth, and hearing loss. About 20,000 to 50,000 people in the United States have OI.

What Causes Osteogenesis Imperfecta?

OI is caused by one of several genes that aren't working properly. Genes carry our hereditary (family) information. We each have two copies of most genes: one set from each parent. Genes are what make you look like your biological family.

Each of the genes that cause OI plays a role in how the body makes collagen. Collagen is a material in bones that helps make them strong. When these genes aren't working properly, there isn't enough collagen, or the collagen doesn't work properly. This leads to weak bones that break easily.

Most children inherit the gene that doesn't work properly from one parent. Some inherit it from both parents. In some cases, neither parent passes on this gene. Instead, the gene stops working properly soon after the child is conceived.

What Are the Symptoms of Osteogenesis Imperfecta?

All people with osteogenesis imperfecta have brittle bones. OI can range from mild to severe and symptoms vary from person to person. Some of the symptoms that people with OI may have are:

- Malformed bones
- Short, small body
- Loose joints
- Muscle weakness
- Sclera (whites of the eyes) that look blue, purple, or gray
- Triangular face
- Barrel-shaped rib cage
- Curved spine
- Brittle teeth
- Hearing loss (often starting in 20s or 30s)
- Breathing problems
- Type 1 collagen that does not work well
- Not enough collagen.

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What Are Some Types of Osteogenesis Imperfecta?

There are 8 main types of osteogenesis imperfecta. People with types 2, 3, 7, and 8 tend to have severe symptoms. People with types 4, 5, and 6 tend to have more moderate symptoms. People with type 1 tend to have mild symptoms. There used to be only 4 types of OI, but then scientists found that even when symptoms look similar in a group of patients, they can be caused by problems in different genes. This is why there are now 8 main types of OI instead of 4.

How Is Osteogenesis Imperfecta Diagnosed?

No single test can identify osteogenesis imperfecta. To diagnose OI, doctors look at:

- Family history
- Medical history
- Results from a physical exam
- X rays.

Your doctor may also test your collagen (from skin) or genes (from blood). It may take a few weeks to learn the results of the tests. These tests spot OI in 9 out of 10 people who have it.

How Is Osteogenesis Imperfecta Treated?

Although there is no cure for OI, symptoms can be managed. Treatments for OI may include:

- Care for broken bones
- Care for brittle teeth
- Pain medication
- Physical therapy
- Use of wheelchairs, braces, and other aids
- Surgery.

One type of surgery is called “rodding.” Metal rods are put inside the long bones to:

- Strengthen them
- Fix bone malformations
- Prevent bone malformations.

A healthy lifestyle also helps people with OI. You can help prevent broken bones and maintain your health if you:

- Exercise (swimming, water therapy, walking)
- Keep a healthy weight
- Eat a balanced diet
- Do not smoke
- Do not drink a lot of alcohol and caffeine
- Do not take steroid medicines.

Proper care helps children and adults who have OI to:

- Stay active
- Make bones more dense
- Keep muscles strong.

What Research Is Being Done on Osteogenesis Imperfecta?

No medications are approved to treat OI. But, experts are trying to learn more about:

- Genes that cause OI
- Medications to help people with OI grow (growth hormone)
- Drugs to make bones stronger (bisphosphonates and teriparatide)
- Better devices to use in surgery.

For More Information About Osteogenesis Imperfecta and Other Related Conditions:

NIH Osteoporosis and Related Bone Diseases~National Resource Center

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The information in this publication was summarized in easy-to-read format from a more detailed publication. To view, download, or order the full-text version, visit http://www.niams.nih.gov/health_info/bone.

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For Your Information

For updates and for any questions about any medications you are taking, please contact the U.S. Food and Drug Administration at 1-888-INFO-FDA (1-888-463-6332, a toll-free call) or visit their Web site at www.fda.gov.