

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 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 the twenties or thirties).
- Breathing problems.
- Type 1 collagen that does not work well.
- Not enough collagen.

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.

How Is Osteogenesis Imperfecta Diagnosed?

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

- Family history.
- Medical history.

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- 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.
- Drugs to make bones stronger.
- Better devices to use in surgery.

For More Information About Osteogenesis Imperfecta and Other Related Conditions:

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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 www.niams.nih.gov/health_info/bone.

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For Your Information

This publication may contain information about medications used to treat the health condition discussed here. When this publication was printed, we included the most up-to-date (accurate) information available. Occasionally, new information on medication is released.

For updates and for any questions about any medications you are taking, please contact the U.S. Food and Drug Administration (FDA) toll free at 888-INFO-FDA (888-463-6332) or visit its website at www.fda.gov. For additional information on specific medications, visit Drugs@FDA at www.accessdata.fda.gov/scripts/cder/drugsatfda. Drugs@FDA is a searchable catalog of FDA-approved drug products.