



Points To Remember About Osteogenesis Imperfecta (OI)

- Osteogenesis imperfecta (OI) is a disease that causes your bones to break easily. OI is also called brittle bone disease.
- Symptoms may be mild or severe, depending on the type of OI you have.
- OI is caused by a gene that doesn't work correctly. There is no cure for OI.
- Treatment can include physical or occupational therapy, medications, surgery, and aids to help you move around safely.
- You can do things to help you stay healthy and prevent broken bones.

What is osteogenesis imperfecta?

Osteogenesis imperfecta (OI) is a disease that causes your bones to break (fracture) easily. OI is also called brittle bone disease. Your symptoms may be mild or severe, depending on the type of OI you have.

Who gets osteogenesis imperfecta?

Anyone can be born with OI, but people who have family members that have it are more likely to get it.

What are the types of osteogenesis imperfecta?

There are many types of OI. Symptoms vary depending on the type you have. Type I is the mildest and most common form of OI. Type II is the most severe form. Other types of OI have symptoms that fall between Type I and Type II.

What are the symptoms of osteogenesis imperfecta?

All people with OI have weak, brittle bones. Some people with OI may have only a few broken bones over their lifetime. Others may have hundreds of broken bones.

People with OI may have other symptoms, such as:

- Misshapen or bowing of long bones.
- Height that is short, below average.
- Skin that bruises easily.
- Loose joints.
- Weak muscles.
- Whites of the eyes that look blue, purple, or gray.
- A face shaped like a triangle.
- A rib cage shaped like a barrel.
- A curved spine.
- Collapse of the vertebrae in the spine.
- Brittle, misshapen, or discolored teeth.
- Hearing loss.
- Breathing problems.
- A malformed hip joint.

What causes osteogenesis imperfecta?

A change in a gene causes OI. People with OI have a gene that carries the wrong instructions for making collagen, a substance that makes bones strong. You might get this changed gene from one or both parents, or the gene might stop working properly on its own.

Is there a test for osteogenesis imperfecta?

To figure out if you have OI, doctors may:

- Ask about family and medical history.
- Do a physical exam.
- Order x-rays, other tests to check your bones, or blood tests.

How is osteogenesis imperfecta treated?

There is no cure for OI. The goals of treatment are to:

- Prevent or control symptoms.
- Increase bone mass (density) and muscle strength.
- Help you to be independent.

These treatments include:

- Physical or occupational therapy.
- Medicines.
- Bone care, such as a cast or brace.
- Surgery to correct bones.
- Walkers, crutches, or wheelchairs to help you move around.
- Dental care if your teeth are affected by OI.
- Hearing aids and other types of ear care.

Who treats osteogenesis imperfecta?

Many different doctors and health care providers may treat OI, including:

- Primary care physicians, who diagnose and treat adults.
- Pediatricians, who diagnose and treat children.
- Clinical geneticists, who diagnose and treat children and adults with genetic disorders.
- Orthopaedists, who treat and perform surgery for bone and joint diseases.
- Occupational therapists, who teach how to safely perform daily activities.
- Physical therapists, who teach ways to build muscle strength, recover from broken bones, and prevent broken bones.
- Dental providers such as orthodontists and surgeons.

Living with osteogenesis imperfecta

If you have a OI, you can do some things to help you stay healthy and prevent broken bones, such as:

- Eat a diet rich in fruits, vegetables, and whole grains.
- Exercise as much as your doctor recommends. Swimming and water therapy are common choices for people with OI.
- Keep a healthy weight.
- Don't smoke, and avoid secondhand smoke.
- Do not drink a lot of alcohol or caffeine.
- Talk to a health care professional if you feel depressed or anxious about OI and its symptoms.

For more info

Genetic and Rare Diseases Information Center

Website: <https://rarediseases.info.nih.gov/diseases/1017/osteogenesis-imperfecta>

Genetics Home Reference

Website: <https://ghr.nlm.nih.gov/condition/osteogenesis-imperfecta>

U.S. Food and Drug Administration

Toll free: 888-INFO-FDA (888-463-6332)

Website: <https://www.fda.gov>

[Drugs@FDA](https://www.accessdata.fda.gov/scripts/cder/daf) at <https://www.accessdata.fda.gov/scripts/cder/daf> [Drugs@FDA](https://www.accessdata.fda.gov/scripts/cder/daf) is a searchable catalog of FDA-approved drug products.

Centers for Disease Control and Prevention, National Center for Health Statistics

Website: <https://www.cdc.gov/nchs>

NIH Osteoporosis and Related Bone Diseases ~ National Resource Center

Website: <https://www.bones.nih.gov>

Osteogenesis Imperfecta Foundation

Website: <https://www.oif.org>

If you need more information about available resources in your language or other languages, please visit our webpages below or contact the NIAMS Information Clearinghouse at NIAMSInfo@mail.nih.gov.

- [Asian Language Health Information](#)
- [Spanish Language Health Information](#)