CONDITION

Osteogenesis imperfecta

Learn how osteogenesis imperfecta affects bones, teeth, hearing, growth, and movement, and how fractures and long-term health are managed.

WHAT IT IS
Group of genetic conditions causing fragile bones

BODY SYSTEM
Bones and skeleton

MAIN SYMPTOMS
Frequent fractures, bone pain, short stature, hearing loss

WHAT CAUSES IT
Inherited or new genetic changes

HOW IT IS DIAGNOSED
Genetic testing

MAIN TREATMENT
Supportive bone and mobility care

CAN IT BE CURED?
No, genetic change cannot be corrected

IS IT AN EMERGENCY?
Sometimes, suspected fracture with severe pain

LONG-TERM OUTLOOK
Lifelong and widely variable with potentially major complications

Overview

Brittle bone disease usually refers to osteogenesis imperfecta , often shortened to OI. It is a group of genetic conditions in which bones break more easily than expected. "Osteogenesis imperfecta" means imperfect bone formation.

OI often affects collagen, a protein that helps give bones strength and flexibility. Collagen is also present in the skin, teeth, tendons, the whites of the eyes, and other tissues. For this reason, OI can affect more than the skeleton.

The condition varies widely. Some people have only a few fractures and may not learn that they have OI until adulthood. Others have frequent fractures, short stature, curved bones, spine problems, or breathing difficulties. The most severe forms can cause major complications before or shortly after birth.

OI is lifelong, but it does not usually get steadily worse in the same way as a degenerative disease. Fracture rates can change with age, growth, activity, pregnancy, menopause, and overall health. Treatment cannot correct the underlying genetic change, but it can strengthen bones, support movement, reduce complications, and help people remain as independent as possible.

OI is not the same as osteoporosis. Osteoporosis is a loss of bone strength that is more common with aging and certain medical conditions. A person with OI can also develop osteoporosis or other causes of low bone density.

Symptoms

Frequent fractures are the best-known feature of OI. A fracture may happen after a minor fall, a simple movement, or another force that would not usually break a healthy bone. However, the number and timing of fractures differ greatly between people.

Possible signs and symptoms include:

Dental changes caused by OI are called dentinogenesis imperfecta . Teeth may appear gray, blue, yellow-brown, or translucent. They may wear down or break more easily because the layer beneath the enamel is abnormal.

Infants with severe OI may be born with fractures, soft skull bones, short or bowed limbs, or a small chest. Milder OI may not be recognized until a child starts walking, develops repeated fractures, or has an evaluation because another family member is affected.

Pain can come from a new fracture, an older fracture that healed in an altered position, joint strain, muscle fatigue, or spine problems. New or worsening pain should not automatically be assumed to be part of OI.

- Repeated bone fractures or fractures after minor injuries
- Bone pain, muscle weakness, or reduced endurance
- Bowing of the arms or legs
- Short stature or slower growth
- Curvature of the spine, called scoliosis or kyphosis
- Loose joints or flexible ligaments
- A triangular facial shape in some forms
- Blue, gray, or purple coloring of the sclera, which is the white part of the eye

- Hearing loss, often beginning in adolescence or adulthood
- Teeth that are weak, discolored, or easy to damage
- Easy bruising
- Breathing problems related to the shape of the chest or spine

Causes and risk factors

OI is caused by a disease-related change in a gene involved in bone formation or bone strength. Most cases are linked to changes in the COL1A1 or COL1A2 genes. These genes provide instructions for making type I collagen, an important structural protein in bone.

A genetic change may reduce the amount of collagen or alter its structure. Changes that affect collagen structure often cause more significant bone fragility, although genetic results do not always predict exactly how a person will be affected.

Less common forms of OI involve other genes that help produce, process, transport, or organize collagen and bone tissue. These forms may follow different inheritance patterns.

Many COL1A1- and COL1A2-related forms are autosomal dominant . This means a person can have OI after inheriting one altered copy of the gene from either parent. Some people have a new genetic change that was not detected in either parent. This is called a de novo variant.

Other forms may be autosomal recessive, meaning altered copies are inherited from both parents, or X-linked, meaning the gene is on the X chromosome. Rarely, a parent may have the genetic change in only some egg or sperm cells. This is called germline mosaicism and can affect the chance of OI occurring again in a family.

Having a close relative with OI raises the likelihood of inheriting certain forms. However, OI can occur without a known family history. It is not caused by exercise, diet, parenting, or something a person did during pregnancy.

Diagnosis

Diagnosis begins with a medical and family history. A clinician may ask how fractures happened, when they began, whether growth has been affected, and whether relatives have fractures, hearing loss, short stature, or characteristic dental or eye findings.

The physical examination may assess bone shape, joint movement, spine alignment, teeth, hearing, growth, and the whites of the eyes. X-rays can show current or healed fractures, curved bones, compression of spinal bones, or small extra bones within skull sutures known as

wormian bones.

Other tests may include:

Low bone density can support the assessment, but it does not diagnose OI by itself. Some people with genetically confirmed OI do not have extremely low DXA results. Bone density measurements in children and people with short stature also require careful interpretation.

Genetic testing can often confirm the diagnosis and identify the inheritance pattern. A negative result does not always rule out OI because current testing may not detect every disease-causing change. Genetic counseling can help patients and families understand results and discuss testing for relatives.

During pregnancy, severe skeletal findings may be visible on ultrasound. If a familial genetic change is known, prenatal or preimplantation genetic testing may be possible. These are personal decisions that require specialist counseling.

Clinicians may also consider other explanations for fractures, including osteoporosis, vitamin or mineral disorders, other genetic bone conditions, and injury. In children, the evaluation must carefully consider both medical causes and possible nonaccidental injury. OI and abuse are separate issues, and neither should be assumed without a thorough assessment.

- Blood or urine tests to look for other causes of fragile bones
- A DXA scan to measure bone mineral density
- Hearing and dental evaluations
- Genetic testing for genes associated with OI
- Imaging of the spine, skull, chest, or limbs when symptoms require it

Treatment and management

Care is usually coordinated by clinicians familiar with inherited bone disorders. Depending on a person's needs, the team may include primary care clinicians, endocrinologists, geneticists, orthopedic surgeons, physical and occupational therapists, dentists, hearing specialists, pain clinicians, and respiratory specialists.

Fractures are treated with appropriate pain control, support, casting, or surgery. Because long periods without movement can weaken bone and muscle, the care team may favor the lightest effective support and safe return to movement when possible. Fractures should still be stabilized properly; activity should not resume before the treating clinician advises it.

Orthopedic surgery may correct severe bowing, stabilize repeated fractures, or place rods

inside long bones. Expandable rods can sometimes be used in growing children. Surgery is individualized according to age, bone size, symptoms, and functional goals.

Physical therapy supports muscle strength, balance, joint stability, and safe movement. Water-based exercise may reduce stress on bones while allowing strengthening. Occupational therapy can recommend safer ways to bathe, dress, study, work, or perform household tasks. Mobility aids and home modifications may improve independence rather than limit it.

Bisphosphonate medicines slow the breakdown of bone. Specialists may use them, particularly for children or adults with moderate to severe OI, vertebral compression, bone pain, or reduced mobility. They can increase bone mineral density and may improve the shape of compressed spinal bones in growing children. Their effect on fracture frequency varies across studies. Treatment decisions require discussion of potential benefits, side effects, dental health, kidney function, and the limited evidence about very long-term use.

Calcium and vitamin D are needed for normal bone health, but supplements do not correct the genetic cause of OI. A clinician may recommend testing or supplementation when intake is inadequate or a deficiency is present. Excessive supplementation can be harmful.

Additional management may include:

Handling techniques are especially important for infants and children. Caregivers can be taught how to lift, position, dress, and transfer a child without pulling or twisting the limbs. Routine movement and affectionate contact remain important.

- Regular dental care and protective treatment for fragile teeth
- Hearing tests and hearing aids or other treatment when needed
- Pain management that combines medical, physical, and practical approaches
- Monitoring and treatment for scoliosis or chest-wall problems
- Healthy nutrition and weight management to reduce avoidable strain on bones
- Avoiding smoking and excessive alcohol because both can harm bone health

Complications

Repeated fractures may heal with angulation, shortening, or deformity. This can affect walking, arm function, daily activities, and comfort. Reduced activity after fractures can also lead to muscle loss and further loss of bone strength.

Compression fractures of the spine can cause pain, height loss, or increasing spinal curvature. Severe scoliosis or chest deformity may reduce the space available for the lungs. Some people

develop restrictive lung disease, meaning the lungs cannot expand fully.

Hearing loss can result from changes in the small bones or other structures of the ear. Dental problems can affect chewing and may require specialized restorative care.

Less common but important complications include pressure or structural changes where the skull meets the spine. These may cause persistent headache, neck pain, swallowing problems, weakness, balance changes, or changes in sensation and require specialist evaluation.

Pregnancy can place added stress on bones, joints, the spine, heart, and lungs. People with OI may benefit from pre-pregnancy counseling and coordinated obstetric, anesthesia, orthopedic, and respiratory planning.

Follow up

Routine follow-up should match the person's age and OI severity. Visits may review fractures, pain, mobility, growth, spine alignment, bone density, nutrition, teeth, hearing, breathing, and emotional well-being. Children may need more frequent review during rapid growth.

Contact the care team promptly for new bone pain, reduced ability to use a limb, increasing back pain, worsening spine curvature, new hearing changes, persistent headaches, or a decline in walking or daily function.

Seek urgent or emergency care for:

When emergency treatment is needed, tell the team about the OI diagnosis and any known spine, airway, bleeding, or anesthesia concerns. Necessary imaging and treatment should not be delayed because of fear of causing another fracture.

- A suspected fracture with severe pain, obvious deformity, an open wound, or loss of feeling or circulation
- A significant injury to the head, neck, or back
- New weakness, numbness, loss of coordination, or loss of bladder or bowel control
- Sudden or severe breathing difficulty
- Chest pain, blue or gray lips, fainting, or inability to stay awake

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