Osteogenesis Imperfecta

What Is Osteogenesis Imperfecta?

Osteogenesis imperfecta (also known as brittle bone disease or OI) is a genetic condition that causes a defect in a protein found in bones—called collagen. The defect leads to fragile bones that can break easily. It is a lifelong condition that varies greatly in severity, affecting bone quality and bone mass.

Types of Osteogenesis Imperfecta

At least eight different forms of osteogenesis imperfecta exist, ranging in severity from a lethal form to a milder form with few visible symptoms. The specific medical problems children encounter depend on the degree of severity.

Up to 95 percent of OI cases fall into four major types of the condition:

Type I – Mild

Type I is the most common form of osteogenesis imperfecta and it’s usually inherited. Children who have Type I osteogenesis imperfecta typically don’t develop bowing deformities in the arms or legs (these are known as long bones), and their height is usually normal.

Common characteristics of children who have Type I osteogenesis imperfecta include:

- Bones that fracture more easily than normal, most often before puberty.
- A near-normal or slightly short stature.
- Whites of the eye (also known as sclera) that are blue or blue-gray.
- Dental problems (also known as dentinogenesis imperfecta).
- Hearing loss beginning around age 20 to 30.
• A curved spine.
• A triangular-shaped face.

Type II – Extremely Severe/Lethal

Children who have Type II osteogenesis imperfecta are born with severe skull, spine, chest-wall and long-bone deformities that develop as they grow in the uterus. Survival past infancy is rare. Usually a new gene mutation—not an inherited gene mutation—causes osteogenesis imperfecta Type II.

Type III – Severe, Progressive Deformity

Children who have Type III osteogenesis imperfecta are born with fractures that cause moderate to severe abnormalities. X-rays might show healing fractures that happened before birth. With growth, further abnormalities develop, leading to limb shortening and bowing. These problems impair upper-extremity motion and are often severe enough that children can’t stand. Children who have Type III osteogenesis imperfecta usually have no family history of the condition.

Children with osteogenesis imperfecta Type III typically have:
• Bones that fracture more easily than normal.
• Severe hearing loss.
• Loose joints and poor muscle development in the arms and legs.
• A barrel-shaped rib cage.
• Short stature.
• A curved spine.
• A triangular-shaped face.
• Dental problems.
• Blue-gray or blue sclera.

Type IV – Moderate to Severe

Although fractures are common for children with Type IV OI, bone deformities are usually not as severe. Type IV is more severe than Type I, but not as severe as Types II or III. Type IV often can be traced through family lines.

Common characteristics of children who have Type IV include:
• Bones that fracture easily, most often before puberty.
• Normal or near-normal colored sclera.
• A curved spine.
• Loose joints.
• Short stature.
• Mild to moderate bone abnormalities.
• Hearing loss.
• A barrel-shaped rib cage.
• A triangular-shaped face.

In some cases, children who have Type IV osteogenesis imperfecta have dental problems.

**What Causes Osteogenesis Imperfecta?**

Osteogenesis imperfecta isn’t caused by too little calcium or poor nutrition. The most common forms of OI are caused by a mutation on collagen genes. The mutation affects the body’s production of the collagen found in bones and other tissues. Other types of osteogenesis imperfecta are caused by a mutation of the cartilage-associated protein gene.

Osteogenesis imperfecta occurs equally in girls and boys and among all racial and ethnic groups, affecting six to seven people in every 100,000. An estimated 20,000 to 50,000 people in the U.S. have the condition. The estimated number varies greatly because milder forms of osteogenesis imperfecta can go undiagnosed.

The greatest risk factor is heredity. If one parent has osteogenesis imperfecta, a child has a 50 percent chance of having the condition. The most common forms of osteogenesis imperfecta are inherited and can usually be traced through the family. Less common forms are passed to children through recessive inheritance.

Approximately 35 percent of children who have osteogenesis imperfecta have no family history of the condition. This occurs when there’s a new mutation on a gene. It isn’t caused by anything parents did before or during pregnancy.

**Osteogenesis Imperfecta Symptoms and Effects**

All forms of osteogenesis imperfecta share a major concern—fragile bones that break frequently. Despite the challenges of brittle bone disease symptoms, most children, teens and adults who have the condition lead full and productive lives.

The effects of osteogenesis imperfecta vary greatly:

• A person who has mild osteogenesis imperfecta symptoms might experience a few fractures, and life expectancy isn’t affected.
- Those with severe types of osteogenesis imperfecta might have hundreds of breaks in a lifetime, and life expectancy might be shortened.

- The most severe forms of osteogenesis imperfecta result in death at birth or during infancy.

- People who have serious forms of the condition might have difficulty breathing.

- Respiratory failure and accidental trauma are the most frequent causes of death for people who have osteogenesis imperfecta.

**Osteogenesis Imperfecta Diagnosis**

Sometimes severe cases of osteogenesis imperfecta are detected in infancy. Milder forms are often more difficult to diagnose in infancy and childhood.

Osteogenesis imperfecta is typically diagnosed after reviewing information gathered from a physical exam and from:

- A history of frequent fractures with minimal trauma.
- Genetic testing of a blood sample (DNA blood test).
- A bone density scan using dual-energy X-ray absorptiometry (also known as a **DXA scan**) to assess bone mass.

Findings from the physical exam and **X-rays** might initially suggest osteogenesis imperfecta, but further testing is often required to confirm it. In some cases, a skin biopsy or bone biopsy is done. Some of these tests help to rule out other conditions, such as:

- Hypophosphatasia.
- Nutritional rickets, caused by a vitamin D deficiency.
- Cushing’s disease.
- Calcium deficiency/malabsorption.
- Nonaccidental injury.

**Osteogenesis Imperfecta Treatments**

Because there is no cure for brittle bone disease, the goal of treatment is to minimize the effects of the disorder, including:

- Increasing bone density and muscle mass.
- Minimizing fractures.
- Maintaining or restoring bone alignment.
- Maximizing musculoskeletal function.
• Maintaining optimal growth and well-being.

From childhood to adulthood, Gillette Children’s Specialty Healthcare can provide expert care for your child who has osteogenesis imperfecta. A collaborative team will develop a customized treatment plan for your child that might include:

**Medications**

If your child has Type III or Type IV osteogenesis imperfecta, medications might help. Oral or intravenous bisphosphonate medication is a common treatment option if your child has experienced three to four major fractures within two years.

Your child might experience reduced bone pain within weeks after bisphosphonate medication. The use of bisphosphonates has also been shown to:

• Improve the ability to walk.
• Decrease the number of fractures.
• Increase stature.
• Increase bone density.

Before receiving any medication, you’ll receive information on usage and potential side effects.

**Physical Therapy and Occupational Therapy**

Your child might have delays in gross motor development associated with osteogenesis imperfecta. In this case, you might work with rehabilitation specialists, physical therapists and occupational therapists to safely support ongoing development, mobility and function.

With carefully designed therapy, your family will learn how to safely lift and position your child to encourage healthy development. Your child might need to use adaptive equipment to help with sitting, standing or walking. If your child has a period of immobilization after casting, strength and endurance exercises might be part of therapy.

As your child grows, we routinely evaluate their strength and developmental abilities and adjust therapy as needed.

**Orthopedic Treatments**

**Orthopedic devices**

Braces and splints (also known as orthotics) can help prevent fractures and provide support. If your child fractures a bone, a lightweight cast or splint supports the limb while it heals. Because minor trauma causes many fractures,
the period of immobilization is brief—often two to four weeks—to reduce further bone loss that could occur with prolonged immobilization.

**Orthopedic Surgery**

Surgery can help to manage recurring fractures, bowed bones and sideways curvature of the spine (also known as scoliosis).

If your child needs orthopedic surgery, telescoping rods are implanted to support the long bones of the arms and legs.

- Rods placed are within the canal of the bone to help stabilize fractures and realign deformed bones.
- Rods are preferred to plates and screws, which can cause fragile bones to break.
- Telescoping rods elongate within the bone as your child grows, requiring replacement less often than solid rods.

**Integrated Care**

At Gillette, your child can receive comprehensive care at the interdisciplinary osteogenesis imperfecta clinic in St. Paul—designed specifically to evaluate and treat newly diagnosed children. In one location, you have access to orthopedic surgeons, endocrinologists, geneticists and pediatric rehabilitation medicine specialists who collaborate to develop a custom and coordinated treatment plan for your child.

For bone disorders like osteogenesis imperfecta, treatment might include working with experts in:

- Audiology.
- Casting.
- **Child life.**
- **Endocrinology.**
- **Medical genetics and genetic counseling.**
- **Neurology.**
- **Orthopedics.**
- **Orthotics.**
- **Pulmonology and respiratory care.**
- **Radiology and imaging.**
- **Rehabilitation medicine.**
- **Rehabilitation therapies.**
- **Seating and mobility equipment.**
- **Social work.**