

Scoliosis in Children and Osteogenesis Imperfecta (OI) — Symptoms and Treatment

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Scoliosis is the name given to the curvature of the spine, and it is common in the pediatric population. Scoliosis can be classified into 3 types: idiopathic, congenital, and secondary. It is important to diagnose and start immediate treatment because the spine can continue to curve with the growth of a child. Osteogenesis Imperfecta is a congenital disease that causes brittle bones. The manifestation of the condition can vary from mild to severe, and the symptoms can range from weak bones to hearing loss and significant bone deformity.



Definition of Scoliosis in Children

Scoliosis is a deformity of the spine that causes the spine to bend sideways.

Scoliosis is defined as a curvature of 10° or more from a perpendicularly straight spine with rotational deformity of the spine. Normally, the spine is straight, but in scoliosis, the spine takes a C or S shape. A person with scoliosis appears to be in a leaning posture.

Epidemiology of Scoliosis in Children

Scoliosis can occur on the left or right side or on both sides of the spine in different sections. There are many types of scoliosis in the pediatric population, with idiopathic scoliosis representing most cases (80%). The lumbar and thoracic segments are mostly involved in this condition.

Etiology of Scoliosis in Children

Scoliosis is idiopathic in the majority of cases. The condition occurs more commonly in girls. According to a recent data, 3 to 5 children per 1000 are affected and require appropriate treatment. Scoliosis **can be divided into 3 major types:**

Idiopathic Scoliosis

The cause has not been discovered so far. Idiopathic scoliosis is subclassified into the following categories:

- **Infantile:** Occurs from birth up to 3 years of age
 - **Mostly seen in boys, with the spine curved toward the left side**
 - Curvature often improves as the child grows
- **Juvenile:** Occurs between 3 and 10 years of age
- **Adolescent:** Occurs between 10 and 18 years of age
 - A common form
 - Mostly seen in girls

Congenital Scoliosis

Congenital scoliosis **occurs due to defects in the fetal growth process** as a result of the following factors:

- Improper formation of the vertebral column
- Absence of vertebral bodies
- Joined vertebrae
- Incomplete formation of vertebral bones

Secondary Scoliosis

Secondary scoliosis involves forms that develop because of other abnormalities. These cases often occur as a consequence of the following:

- Infections
- Tumors
- Injuries

Neuromuscular conditions

- Spina bifida
- Cerebral palsy
- Muscular dystrophy
- Tumors of the spinal cord
- Neurofibromatosis

- Paralytic conditions

Symptoms of Scoliosis in Children

Symptoms of scoliosis include the following:

- Difference in shoulder heights
- Head position may not be centered with respect to the body axis
- Hip position can be affected by scoliosis
- Shoulder blade can deviate from the normal position
- Arm lengths vary when the person stands straight
- **Other symptoms may include back pain and leg pain**
- Rarely, bowel and bladder problems can arise

Diagnosis of Scoliosis in Children

Early detection is essential for effective treatment. Scoliosis is diagnosed on the basis of history, physical examination, and radiologic investigations.

Clinically, the degree of spinal deviation from the normal axis can be established with the help of various tools, such as a scoliometer, the Adams forward bend test, and the plumb-line test. In the scoliometer technique, the physician stands behind the patient. The patient is asked to bend forward with the feet together, knees extended, and hands hanging freely by the side. The scoliometer is placed over the back, and any measurement greater than 7° demands radiologic investigations.

The shoulder and hip girdle are also observed by the doctor while the patient stands straight with the back facing the doctor. Any discrepancies in limb lengths and neurologic findings are also determined.

Radiography is a primary tool for diagnosing scoliosis. The degree of spinal curvature is measured from the X-ray image. The Cobb angle is measured by drawing 2 lines above and below the curved segment that are parallel with the top and bottom of the tilted vertebrae. A perpendicular line is then drawn to each of the parallel lines. The angle formed where the perpendicular lines intersect called the Cobb angle.

Computed tomography scans and magnetic resonance imaging are also used for diagnosing the condition. These investigations show structures in detail and can be used to detect any other underlying pathology.

Treatment of Scoliosis in Children

The treatment of **scoliosis depends on many factors**, which include the following:

- The child's age and medical history
- Causes
- Extent or severity of the condition
- Expected duration of treatment
- Opinions or preferences

The goal is to stop the progression of the deformity. Treatment plans are discussed in the following sections.

Observations and Examinations

Repeated observation and examination are required when the spine is curved less than 25° degrees and the child is in a phase of rapid growth. This is done to determine whether the spine continues to curve or not. The progression rate depends on skeletal enlargement and maturity, and it stops when the child has reached puberty. After the age of skeletal maturation, the condition is monitored every 8–12 months to determine the status of the spinal deformity.

Bracing

Bracing is used when radiography shows spinal curvature of 25°–30°. It is also used if the child has a curvature of 20°–25° and is growing and the condition is not showing improvement. The types and duration of bracing vary with each case of scoliosis. The aim is to stop the progression of the condition; bracing cannot correct spine segments that are already curved. Bracing also significantly reduces the incidence of surgery.

Surgery

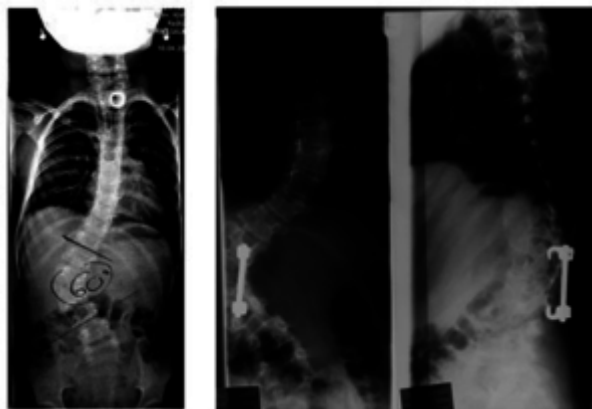


Image: Progression after early operation for severe hemivertebra at the age of 6 years. After surgical intervention at the age of 6 years, there was a significant progression of scoliosis (60° to 90°) and kyphosis as well at the last follow-up at the age of 11 years, the patient still being premenarchial. By Kaspiris A, Grivas TB, Weiss HR, Turnbull D. License: [CC BY 2.0](https://creativecommons.org/licenses/by/2.0/)

Surgery is used when the curvature is more than 45°. Bracing is generally not successful for this magnitude of curvature, so surgery is the only option. A surgical technique called 'spinal fusion' is used to straighten the curvature and fuse the vertebral bodies to align them in a straight fashion.

This helps stop the growth of the affected segment completely. Only the curved vertebral bodies are fused together; the rest are mobile. Metal rods are used to hold the bones until the process of fusion is completed. These are attached with wires, screws, and hooks to the spine.

Definition of Osteogenesis Imperfecta (Brittle Borne Disease)

Osteogenesis imperfecta is a **bone disorder that produces fragile bones that can break easily**. It is present from birth, and it usually occurs in children with a family

history of the disorder. As is indicated by the name, it describes impaired bone formation. Multiple fractures can occur and, in many cases, they occur before birth. The intensity can vary from mild to severe. The mild form results in fractures, whereas the severe form presents more like a syndrome constituting a pattern of disorders. **These include:**

- Hearing loss
- Heart failure
- Spine problems
- Permanent deformities

There are **8 types of osteogenesis imperfecta**, which are distinguished on the basis of different signs and symptoms. Type 1 is characterized as the mildest form and type 2 as the most severe form of osteogenesis imperfecta. The rest of the types fall between these 2 extremes.

Etiology of Osteogenesis Imperfecta

Osteogenesis imperfecta is an inherited disorder that often runs in a family. Spontaneous mutations can also occur at the time of conception. It is **due to defects in the genes that produce type 1 collagen, a protein that gives strength to the bones**. The absence of enough collagen will result in weak bones.

Inheritance of Osteogenesis Imperfecta

Autosomal dominant: In the autosomal dominant pattern of inheritance, one parent is symptomatic; that parent's cells carry the mutation, which is transmitted to the child. A person with a dominant osteogenesis imperfecta carries a 50% chance of passing on the mutation to the child.

Autosomal recessive: In the autosomal recessive pattern of inheritance, both parents carry one copy of the mutation, but they themselves do not have symptoms of the disease.

Sometimes other genetic mutations are responsible for the disease. None of the family members carry any gene copy, but the child has the disease.

Symptoms Osteogenesis Imperfecta

In osteogenesis imperfecta, **bones can break very easily, even with low-impact activities such as a diaper change**. Some individuals with the disease will have a few broken bones during their lifetime, and others may have hundreds. Symptoms can be present at birth or may appear at a later stage, such as during the teenage years.

Generally, the symptoms of brittle bones can be described as follows:

- Bleeding from injured areas



Image: Osteogenesis Imperfecta Type V in an Adult, by ShakataGaNai. License: [CC BY-SA 2.5](https://creativecommons.org/licenses/by-sa/2.5/)

- Bruises
- Blue color of the sclera
- Breathing problems
- Bowing of legs
- Scoliosis or curved spine
- Early hearing loss and deafness
- Fatigue
- Fragile skin
- Weak muscles
- Loose joints
- Short stature

In the mild form, fewer signs are present. There may be smaller bone deformity, fewer fractures, normal height, some hearing loss, and their normal life expectancy. The incidence of fractures decreases after puberty.

In the moderate to severe form, there are more fractures; short height; deformed spine; and breathing problems, which can be life-threatening, and children can be born with broken bones. In the most severe form, babies die in the uterus or soon after birth because of extensive bone fractures and breathing problems.

Diagnosis of Osteogenesis Imperfecta

The **disease can be easily diagnosed with a history and physical examination.** Laboratory blood and urine tests are needed to rule out other similar conditions, such as rickets. DNA blood testing is used for confirmation and can identify faulty genes with an accuracy of 90%. Collagen analysis can also be done, but a negative result is not sufficient to rule out osteogenesis imperfecta. Prenatal chorionic villus sampling and ultrasonography are useful diagnostic tests.

Plain X-ray imaging can show fracture lines in the bones. DEXA (dual-energy X-ray absorptiometry) scans can be performed to predict the risk of future fractures. A bone biopsy can be done to study histologic changes, but this not routinely performed for osteogenesis imperfecta.

Management of Osteogenesis Imperfecta

There is **no definitive cure for the disease**. **Treatment is targeted at improving disease symptomatology**: pain relief, prevention of bone breakage, mobility improvement, and breathing assistance. The disease demands life-long treatment and proper monitoring. Various treatment options used for osteogenesis imperfecta include the following:

General treatment

- Casts and splints for broken bones
- Braces for legs, knees, wrists, and ankles
- Physical therapy
- Implantation of rods in legs or arms
- Dental work-up
- Hearing aids
- Crowns for brittle teeth
- Mobility aids such as crutches, canes, wheelchairs, and walkers
- Supplemental oxygen for breathing problems

Lifestyle Modifications

- Controlled weight, as excess weight can put extra stress on bones
- Safe exercises to improve muscle and bone strength
- Diet rich in calcium and vitamin D is recommended
- Supplements can be prescribed
- Avoidance of alcohol and caffeine
- Steroids should not be used without serious cause because they reduce the bone density
- Avoidance of active or passive inhalation of cigarette smoke

Medications

Bisphosphonates are given to improve bone density. This is helpful in moderate to severe osteogenesis imperfecta. Teriparatide, which is a recombinant form of parathyroid hormone, is also administered. Growth hormone replacement therapy and gene manipulation therapy is in the research phase.

Surgery

Surgery is needed to repair broken bones. More specifically, it is used to correct bone deformities, stabilize the spine, and repair the tiny middle ear bones to improve hearing. Rods are also placed to stabilize long bones and prevent fractures.

References

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