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

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Scoliosis is the name given to the curved spine. The condition is common in the pediatric population. It is classified into three types: idiopathic, congenital and secondary. It is important to diagnose and start immediate treatment in many cases as 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 hence, the symptoms can range from weak bones to hearing loss, and massive bone deformities.

Definition of Scoliosis in Children

It is a deformity of the spine which causes it to bend sideways. Scoliosis is defined as a curvature of 10 degrees or more from a perpendicularly straight spine with rotational deformity of the spine. Normally, the spine is in an upright alignment but, in scoliosis, the spine takes the C or S shape. A person with scoliosis appears to be in a leaning posture.
Epidemiology of Scoliosis in Children

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

Etiology of Scoliosis in Children

It is defined as idiopathic in the majority of cases. The condition occurs more commonly in females. According to a recent data, 3 to 5 children per 1000 are affected, which requires an appropriate treatment. Scoliosis can be divided into the three major types:

Idiopathic Scoliosis

The cause has not been discovered so far. It is sub-classified into:

- **Infantile**: The time period for the occurrence of the infantile form is up to 3 years after birth
- **Mostly seen in males with the spine curved towards the left side**: The curvature often gets improved with the growth process of the child
- **Juvenile**: It occurs between 3-10 years of age
- **Adolescent**: This is a common form and occurs mostly in girls with a duration of 10-18 years of age

Congenital Scoliosis

This type occurs due to the defects in the fetal growth process, and happens as a result of the following factors:

- Failure of the proper formation of a vertebral column
- Absence of the vertebral bodies
- Joined vertebrae
- Incomplete formation of vertebral bones

Secondary Scoliosis

The secondary involves other forms that develop due to the other abnormalities. These cases often occur as a consequence of the following abnormalities:

- Infections
- Tumors
- Injuries

**Neuromuscular conditions**

- Spina Bifida
- Cerebral palsy
- Muscular dystrophy
- Tumors of the spinal cord
- Neurofibromatosis
- Paralytic conditions
Symptoms of Scoliosis in Children

The symptoms can be listed as follows:

- Difference in the height of shoulders
- Position of the head may not be central with respect to the body axis
- Hip position also gets affected by scoliosis
- Shoulder blade can also 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

The early detection is essential for an effective treatment. Scoliosis is diagnosed on the basis of history, physical examination, and radiological investigations.

Clinically, the degree of spinal deviation from the normal axis can be established with the help of various tests like ‘Scoliometer,’ ‘Adam’s Forward Bend Test’ and ‘Plumbline 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 should hang freely by the side. The Scoliometer is placed over the back and any measurement greater than 7 degrees demands radiological 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 the length of limb and neurological findings are also determined.

An X-ray is a primary tool used routinely for diagnosing scoliosis. The degree of spinal curvature is measured from the X-ray. The Cobb angle is measured by drawing two parallel lines at the top and bottom of the tilted vertebrae at above and below the curved segment, respectively. A perpendicular line is then drawn to intersect both the parallel lines. The angle formed between the parallel and perpendicular line is called the Cobb angle.

CT scans and MRI are also used for diagnosing the condition. These investigations show the structures in detail and any other underlying pathology can also be detected.

Treatment of Scoliosis in Children

The treatment of scoliosis is dependent on many factors, which include:

- 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 and prevention of the deformity. The treatment plan is discussed under the following sub-headings:
Observations and Examinations

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

Bracing

This is the method used when the spinal curvature is 25–30 degrees on the X-ray. It is also done if the child has a curve of 20–25 degrees, is growing, and the condition is not showing improvement. The types of bracing and the time duration vary with each case of scoliosis. The aim is to stop the progression of the condition; it cannot correct the already-curved segments of the spine. Bracing also significantly reduces the incidences of curvatures from exceeding to the point of surgery.

Surgery

This treatment modality is used when the curvature is more than 45 degrees. The bracing is mostly not successful for this much curvature, so surgery remains the only option. A surgical technique called ‘spinal fusion’ is used to straighten the curvature and fuse the vertebral bodies so they get aligned in a straight fashion.

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

Definition of Osteogenesis Imperfecta (Brittle
Osteogenesis Imperfecta is present from the time of birth. It usually occurs in children having a family history of the disorder. As clear from the name, it means an impaired bone formation. Multiple fractures can occur and, in many cases, they occur before the 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 eight forms of Osteogenesis Imperfecta from type 1 to type 8. The types 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 type of Osteogenesis Imperfecta. All of the rest fall in between these two extremes.

Etiology of Osteogenesis Imperfecta

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

Inheritance of Osteogenesis Imperfecta

**Autosomal Dominant:** In the dominant pattern of inheritance parent is symptomatic, only a single % of a parent’s cells carry the mutation which is transmitted to the offspring. A person with a dominant OI carries a 50% chance of transferring the mutation to the offspring.

**Autosomal Recessive:** In the recessive form, both the parents carry the mutations, but they themselves do not have the disease. Sometimes the genetic mutations are responsible for causing the disease. None of the family members carry any gene copy, but the child gets the disease.

Symptoms Osteogenesis Imperfecta

The bones can break very easily, even with minor activities like the change of the baby’s diapers. Some can have a few bones broken and others may have hundreds broken during their lifetime. Symptoms can be present at birth or at a later stage during the teenage years.

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

- Bleeding from injured areas
- Bruises
- Blue color of 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** such as smaller bone deformity, fewer fractures, normal height, hearing loss can be present, and there is a normal life expectancy. The incidence of fractures decreases after puberty.

**In moderate to severe form, there are more fractures**, children are born with broken bones, short height, deformed spine and breathing problems, which can be life-threatening.

**In the most severe form, babies die in the uterus or soon after birth due to extensive bone fractures and breathing problems.**

**Diagnosis of Osteogenesis Imperfecta**

The **disease can be easily diagnosed with a history and physical examination**. Many laboratory tests of blood and urine are needed to rule out other similar conditions like rickets.

DNA blood testing is the confirmatory test, which can identify the faulty genes with an accuracy of 90%. Collagen analysis is also done, but a negative result is not sufficient to rule out OI. Prenatal chorionic villus sampling and ultrasonography are useful diagnostic tests.

Plain X-rays can demonstrate the fracture lines in the bones. DEXA scans are also
performed to predict the risks of future fractures. A bone biopsy can also be done to study the histological changes, but not routinely performed for OI.

**Management of Osteogenesis Imperfecta**

There is no definitive cure for the disease. The treatment is targeted at improving the disease symptomatology; relief of pain, prevention of bone breakage, improving of patient’s mobility, and assisting the breathing. The disease demands a lifelong treatment and proper monitoring. The various treatment options utilized for Osteogenesis Imperfecta include:

### General treatment

- Casts and splints for the 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 like crutches, canes, wheelchairs, and walkers
- Supplemental oxygen for breathing problems

### Lifestyle Modifications

- Controlled weight as excess can put extra stress on bones
- Safe exercises to improve the muscle and bone strength
- Diet rich in calcium and vitamin D is recommended
- Supplements can also be prescribed
- Avoid intake of alcohol and caffeine
- Steroids should not be used miscellaneously as they reduce the bone density
- Stop the active or passive inhalation of cigarette smoke

### Medications

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

### Surgery

**Surgery is needed to repair broken bones,** more specifically it is used to correct bone deformities, to stabilize the spine and repair the tiny middle ear bones to improve hearing. Rodding is also done to stabilize long bones and to prevent fractures.

### References


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