JOHNS HOPKINS DEPARTMENT OF

# Orthopaedic Surgery

Pediatric Orthopaedics

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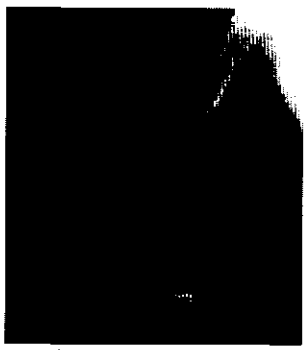
## Patient Guide to Osteogenesis Imperfecta (OI)

***What is osteogenesis imperfecta?***

As the name implies, the condition refers to an imperfect creation ("genesis") of the bones ("osteo"). In short, there is a defect in a protein called "collagen type I" which is a major component of bone and other connective tissues in the body.

Osteogenesis imperfecta is a genetic disorder meaning that it is caused by an abnormality in the gene which instructs the body to make type I collagen. Since different genetic mutations are possible, different people with the condition may be affected more or less severely than others.



***Bones in arm in patient with OI***

***How many children are born with OI?***

Overall, OI affects 1 child per every 10,000 that are born. The more severe forms of the disease are less common.

***How do I know if my child has OI?***

Children with more severe forms of the disease are diagnosed at

birth by the presence of multiple bone fractures. Depending on the severity of the disease, any of the following features may be seen in a patient with OI.

A child may have fragile bones and break them much easier that other children of the same age. Some are short and develop scoliosis (see Patient Guide to Scoliosis). The teeth may not develop in a normal fashion, making them soft and brownish in color. In addition, patients may have deafness due to the improper functioning of the bones of the middle ear. Loose ligaments, blue sclerae (the normally white part of the eye) and mis-shapen skulls are also seen in this condition. The loose ligaments can, in fact, lead to joint dislocations which is another sign of OI. Finally, because of multiple fractures, the bones may take on abnormal shape (see the bowing of the tibia in the image below).

***Radiograph of bowed tibia in patient with OI***

***How do I know what severity of OI my child has?***
The Sillence classification system is most commonly used to place patients into four groups based on the clinical severity of the disease. Each type is designated by a roman numeral: I, II, III, IV.

Type I OI is a mild form of the disease. Type I collagen production is reduced by 50 % compared with normal individuals. Fractures of the bones may occur in later childhood, but they decrease as the child reaches adolescence. These patients do have fragile bone and the blue sclerae as mentioned above . This group of patients is further divided into those who have abnormal teeth and those who have normal teeth.

Type II is the most severe form of the condition and leads to death shortly after birth. These children have very dark blue sclerae indicating the little amount of collagen produced by the body.

Type III OI is the most severe form of OI in patients surviving the neonatal period. Multiple broken bones are often seen at birth. The bones become more and more deformed over time.

Strangely, these patients have normal sclerae (i.e. they are colored white) and hearing (i.e. the bones of the middle ear responsible for hearing are not damaged).

Type IV is a moderately severe from of the condition. The features can be similar to those of both types I and III. These patients also have fragile bones. They have normal sclerae and hearing. They are also divided further into those who have abnormal teeth and those who have normal teeth.

***What is the treatment of OI?***

The care of a patient with osteogenesis imperfecta depends on the type or severity of the condition that a child has. In type I patients, the child may be relatively normal and the pediatric orthopaedic surgeon may not need to be involved to a great extent, except to treat bone fractures that develop. In type II, the child usually dies shortly after birth and therefore gives no time for any treatment to begin. Type III and IV are more difficult. Research is looking into ways to help these patients with medicines.

The pediatric orthopaedic surgeon plays an important role in trying to prevent bone fractures. If and when they do occur, however, they are treated with splints or braces to keep the child from putting too much weight on the broken site. Not only do the bones in a patient with OI break easily, but it is also more difficult to get them to heal. In cases where it is impossible to fix the fracture, surgery may be needed. Plates, screws, and rods can be used inside the body to help them stay together in the proper position so that they can heal well.

Patients with OI can also develop scoliosis (see Patient Guide to Scoliosis). For these patients, bracing is used to help a child maintain a proper upright posture.

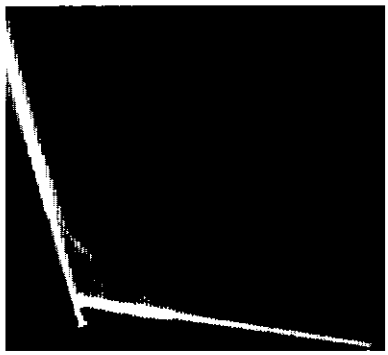
***Bracing in patient with OI***

All in all, it is worthwhile to start an aggressive program of exercises and proceed with appropriate seating, including a wheel

chair if needed.

There a few medications that are currently being tested in patients that have osteogenesis imperfecta. One class of drugs receiving attention is called the "bisphosphonates." This group, which prevents cells called "osteoclasts" from degrading bone (thereby strengthening it), has been used in patients with osteoporosis and bone cancer. The bisphosphonates have shown some promise in children and adults with OI, but research is still being conducted.

***What is the surgery for OI?***
Operations are usually not done until the child is 5 years old. After this time, the surgeon can go in and intentionally break an abnormal bone ("osteotomy") and fix it in place with a rod. There are various types of rods that are currently used.



***Surgically placed rods in patient with OI (radiograph)***

Surgery for scoliosis is quite difficult. The curvature in these patients tends to be severe and bracing (the first line treatment) is often not effective. Also, it is hard to attach a rod to the bone of a patient with OI since it is much softer than normal bone. Nevertheless, a rod can be attached to the spine and tightened so that the curve becomes more normal.

***Should my child need follow-up with a doctor for OI?***
Yes. Seeing a doctor regularly is very important, even from an early age. The orthopaedic surgeon can continue to watch the child for the development or progression of scoliosis. Also, fractures in the skull can cause neurological signs that can be picked up by the physician. A patient with abnormal teeth should see his or her dentist every 3 to 6 months.

***What is the likely future for my child with OI?***
This depends on the type of the disease. Type II patients, as mentioned above, usually die shortly after birth. Type III is the next most severe. These children usually need several orthopaedic (bone) surgeries. Types I and IV are more mild (especially type I). Type I patients may have few fractures and have hearing loss be their most frustrating symptom. Type IV patients have more fractures, but these stop around puberty.

**For further information, contact the following sites:**

- The Osteogenesis Imperfecta Foundation: http://www.oif.org
- The Osteogenesis Clinic and the Kenedy Krieger Institute: http://www.osteogenesisimperfecta.org

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