Osteogenesis Imperfecta or ‘OI’ is a genetic bone disorder which causes brittle bones that break easily. It is more commonly called ‘brittle bone disease.’ The term taken literally means ‘bone that is imperfectly made from the beginning of life.’ A person is born with this disorder and is affected throughout his or her lifetime. It is thought that there are anywhere from 25,000 to 50,000 people in the United States that have this disorder. More precise numbers are difficult to calculate as many patients with mild OI probably go undiagnosed. Diagnosis of OI is usually based on clinical exam but genetic testing can be done for confirmation.

OI is caused by a genetic mutation that affects the body’s production of the collagen which is found in bones as well as other tissues. The mutation may be inherited or spontaneous (~35%) with no family history of OI. It is evenly divided among both males and females and found in all races. Besides having an increased number of fractures, people with OI often have muscle weakness, hearing loss, fatigue, joint laxity, curved bones, scoliosis, blue sclerae, dentinogenesis imperfecta (brittle teeth), and short stature. In severe cases restrictive pulmonary disease may occur.

Initially OI was divided into 4 types (I-IV) but new types were subsequently added and now there are felt to be 8 types. Even individuals with the same type may have different characteristic features.

Type I is the mildest form. These individuals may have very few fractures and not even realize they have the condition. They commonly have blue sclerae though.

Type IV is next in severity and is a moderate form. These patients have more fractures but even within this type it can vary considerably from characteristics similar to type I and to the other extreme similar to type III. Type IV patients often have short stature.

Type III is the most severe form of OI among children that survive beyond the neonatal period. These infants generally have mildly shortened and bowed limbs, small chests, and a soft calvarium (skull bones). Respiratory and swallowing problems are common in newborns and there may be multiple long bone fractures including ribs. Adults with type III are usually no taller than 3 ½ feet.

Type II Osteogenesis Imperfecta is the most severe form. These infants usually die within weeks of delivery. The cause of death is often from respiratory and cardiac complications.

Dr. Beck has had extensive experience with the use of Fassier-Duval rods in the treatment of children with long bone deformities due to osteogenesis imperfecta. The Fassier-Duval system is specifically designed for use in young children with OI to correct and prevent deformity, and improve development and function. He is one of the few orthopedic surgeons in Florida who can provide this type of care.
Treatment of IO is aimed at reducing the number of fractures and improving the quality of life. Medical treatment includes the use of bisphosphonates which decrease the amount of bone turnover, growth hormone, and vitamin D. Physical activity is also important in the well-being of these patients.

Surgical treatment is aimed at straightening the deformed bones to improve strength, thereby decreasing the number of fractures. (See Figure 1).

Studies also show that straightening the bones and mobilizing the patients greatly improves their overall development.

There is no cure for Osteogenesis Imperfecta but with proper care and treatment, children and adults with OI can participate in sports and other activities and lead productive fulfilling lives.

For more information please visit www.chortho.com

Meet our team of Physicians

For appointments, please call (727) 898– BONE (2663) or (813) 879 BONE (2663)

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