Osteogenesis Imperfecta / Brittle Bone Disease

Osteogenesis imperfect (OI) stands for an imperfectly formed bone. Those who have the disease have genetic defects that impair the ability of the body to create strong bones. Some individuals struggle with more severe forms of the condition. Bones will break easily and individuals can end up breaking hundreds of bones during their lifetime. Many individuals have a milder form of the disease and can make it through life without a lot of fractures.

In those who have osteogenesis imperfect, one of the genes responsible for telling the body how to create specific protein doesn't function properly. This specific protein is a major contributor to the connective tissue within the bones. Collagen is important to the formation of teeth, ligaments and the whites of the eyeballs. Due to the defective gene, enough collagen isn't produced, or the collagen isn't of exceptional quality. Either way, the bones are fragile and can easily break, but they heal normally.

The condition is a genetic disorder. It occurs in 1:20,000. It is characterized by the easy breaking of bones, sometimes with minimal trauma, and is apparent from an early age in children. Some forms of OI are very severe, with children dying shortly after birth, or having severe structural deformities of the skeleton. And other forms are less severe (although still serious) and cause fractures during childhood which can reduce into adulthood.

Osteogenesis Imperfecta Anatomy

The skeletal system is made up of all the joints and bones in the body. All of the bones are a complex group of living organs that are composed of multiple cells, protein fibers and a range of minerals. The skeleton serves as a scaffold by delivering protection and support for the soft tissues that make up the remainder of the body. The system provides a series of attachment points that allow the muscles to move at the joints.

New blood cells are generated by the red bone marrow that is inside of the bones. The bones act as the warehouse for the body's calcium, energy and iron in the form of fat. The skeleton continues growing throughout childhood and serves as a framework for the remainder of the body to continue growing along with it.

How to Treat Osteogenesis Imperfecta:

1. **Medication**
   Medical bisphosphonates, administered to children by mouth or through an IV, help to slow down the bone reabsorption. Children with severe cases of the disease find that this medication can help to reduce the amount of pain and fractures in the bone. The medications have to be administered by trained professionals and need to be monitored closely.
2. **Therapy**
   Working with a specialist pediatric therapist can significant improve the quality of life of the child. They can have fun doing strengthening exercises and a therapist can help motivate the child and set goals.
3. **Immobilization**
   Bracing, casting or splinting fractures is often needed to make sure the bones stay still and properly in line to promote healing.
4. **Exercise**
   After fracture has occurred, weight bearing and movement are advised once the bone has finished healing. Specific exercises work to increase mobility while decreasing the risk of additional fractures.

Tips:

- Swimming and walking can help to strengthen the bones, as well as the muscles supporting them.
- Adults who have this disease should refrain from drinking, smoking and taking steroids as they have negative impacts on your total bone density.
- Choose an infant car seat that you can recline. It needs to be easy to remove and place the child into and out of the seat.
- Never lift children with this disease underneath their armpits.
- Don't pull on legs or arms in those who have severe forms of the condition. Lift the legs using the ankles to change diapers.