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Osteogenesis Imperfecta (OI)

When most people think about fragile bones, they tend to think of osteoporosis and older ladies whose hips can break with an unexpected step, causing them to fall. Because of bone density testing and new osteoporosis medications, this once feared orthopedic condition is now able to be treated rather successfully.

There is another fragile bone disease that you may not be familiar with. It is Osteogenesis Imperfecta (OI). This is a lifelong genetic brittle bone disease.

Osteogenesis Imperfecta (OI) is a genetic disease where the body does not have properly functioning collagen, which is a protein that helps build bone, as well as skin, teeth, tendons, and ligaments. Either the body just doesn't make enough or it makes a very poor quality of collagen. OI does not occur because either the patient or the patient's mother (during pregnancy) didn't have enough calcium or suffered from poor nutrition.

There are several different levels of OI, ranging from mild to severe. These differences mean the treatment needs to be designed specifically for each patient's unique problem. Proper diagnosis involves the efforts of pediatricians and geneticists, as well as orthopedic surgeons and therapists.

One of the first indicators of brittle bone disease is the patient's history of unusual broken bones at an early age. Any related history in the family is taken into consideration as well. Another indicator may be a young's child's failure to grow properly in height. A bone density test showing poor bone quality may be an additional indicator. Many of these children will have a blueish tint around the pupils of their eyes, or fragile teeth (called Dentogenesis Imperfecta). If the pediatrician, dentist, or orthopedic surgeon suspects OI, genetic testing is a common next step, looking for a variety of DNA disorders or of biochemical disorders of collagen.

While we do not yet have a cure for OI, we are making progress. In previous decades, we could only treat the fractures as they happened, or insert a series of pins through the center of the large bones to add additional strength. Today, largely because of the work done to treat and prevent osteoporosis, we have medicines which can help prevent or markedly reduce fractures in these children. When surgery is necessary, we have developed dramatically better techniques of bone fixation, which have resulted in better functional outcomes and fewer additional surgeries for these special patients.

Exercise is important, and swimming is ideal because it helps to strengthen the muscles while not putting undue stress on the bones.

There are only a few of us who specialize in treating Osteogenesis Imperfecta (OI) in the United States today. We are very fortunate to have a dynamic OI program at the Shriner's Hospital for Children here in Houston, where I have the advantage of working as part of a wonderful team of physicians, nurses, therapists, and social workers. I provide follow up adult OI care in my private practice, once the patient has completed treatment in the Shriner's Hospital program or has become too old to qualify for the Shriner's program. My OI patients are all special, sweet individuals, and I enjoy being able to help them.

Dr. Parr is a limited liability partner in FOUNDATION SURGICAL HOSPITAL and DOCTORS SURGICAL CENTER.

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