Osteogenesis Imperfecta *Program*

Every child is born with great potential.
Shouldn't every child have the chance to achieve it?

Kennedy Krieger Institute

What is Osteogenesis Imperfecta?

Affecting an estimated 50,000 people in the US alone, Osteogenesis Imperfecta (OI) is a genetic disorder characterized by abnormally brittle bones. Those who have it are far more susceptible to fractures than the average person. It usually originates from a mutation in one of the two genes responsible for making Type 1 collagen, the protein in connective tissue, cartilage, and bone. As a result of this mutation, affected individuals develop either collagen of poor quality or an insufficient amount of collagen. Many people are not even aware that OI exists—in fact, some parents who are not aware that their child has OI have been accused of child abuse when they are unable to explain the cause of their child's fractures.

How severe is OI?

Its severity varies widely from case to case. Individuals with OI can experience as few as ten fractures in a lifetime or several hundred. Some are completely ambulatory, others rely entirely on a wheelchair for mobility. Those with the mildest form of OI might not even discover they have it until later in life. In the most severe cases, OI can affect children before they are born. In these instances, trauma to the skull during delivery can cause brain damage or stillbirth. While there is no cure for OI, quality care helps ensure the best quality of life and the least discomfort possible for those affected by the disorder.

In my mind, I can do anything.

What is life like for individuals with OI?

Many people with OI enjoy productive and fulfilling lives well into their adult years. They attend school, pursue exciting careers and nurture hobbies—some even develop strong athletic abilities. As public awareness of OI improves and greater support is provided for research into the disorder, the future of individuals with OI will grow brighter.

Program Overview

At the Osteogenesis Imperfecta (OI) Program at Kennedy Krieger Institute, our interdisciplinary team of medical professionals work together to evaluate and address our patients' individual needs. Osteogenesis Imperfecta can vary dramatically in severity, so our model emphasizes developing unique therapy regimes tailored to each patient.

Interdisciplinary Evaluations

The first time a patient arrives at the Kennedy Krieger Institute Osteogenesis Imperfecta Program, he or she will receive a thorough evaluation by several physicians and therapists. Patients can opt to receive consultation at Kennedy Krieger or, in some cases, the Johns Hopkins Hospital, on a one-time or as-needed basis. However, most choose to receive continuous follow-up care. Initial evaluations involve the following medical experts, all of whom are available to provide ongoing care when indicated.

- An endocrinologist assesses patients' medical status and makes treatment and medication recommendations.
- An orthopedic surgeon determines surgical needs and, when necessary, performs corrective surgery.
- A radiologist measures patients' bone density.
- A physical therapist offers therapeutic interventions and assists with mobility issues and activities of daily living.
- A physiatrist assesses the need for improving function and offers recommendations for bracing, therapies, and exercises.

Other services available include biomedical testing, genetic analysis and counseling, measurement of growth parameters, referrals for bone densitometry, X-rays and bisphosphonates of different types. In addition to the specialists who participate in an initial evaluation, Kennedy Krieger has many others on the faculty who can assist patients with individual needs. These include audiologists to test for the hearing loss common in OI, nutritionists who

can make dietary recommendations designed to maximize bone density, orthotics specialists to fit patients with braces, social workers to help patients and families who need help coping with OI as well as a consultation service that works with patients' local physicians to develop treatments.

OI Research at Kennedy Krieger Institute

In addition to the state-of-the-art care it provides, the Osteogenesis Imperfecta Program at Kennedy Krieger Institute also serves as one of the world's premier research centers for the disorder. Current research projects focus on improving current treatment regimens and learning more about how certain changes in the body affect the course of OI.

Specifically, these studies include:

- Exploring how certain drugs help increase bone density.
- Examining the effects of diet on bone stability in children with OI. Researchers have found that bone mass and diet may be related in OI.

Understanding more about topics like these helps shape more effective treatments for OI and could someday reveal ways to prevent or reverse the disorder.

In addition to its continuing formal research projects, Kennedy Krieger Institute, in collaboration with the Osteogenesis Imperfecta Foundation, is developing the first-ever OI Registry, a database designed to encourage OI research by consolidating information and connecting people for specific studies and projects. The Registry will also increase public awareness of OI, which is often poorly understood in the general community.

Contact Information

To make a referral or schedule an appointment with the program, please call: 443-923-2703 or 443-923-9400 TTY (for Speech or Hearing Impaired): 443-923-2645 or visit www.kennedykrieger.org.

For Physician referrals, please call our Physicians Assistance Group at: 443-923-9403.

The OI Registry can be accessed through: www.osteogenesisimperfecta.org

Referral Specialists are available:
Monday – Friday, 8:30 a.m. – 5 p.m.

Kennedy Krieger Institute
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Our Mission

We at the Kennedy Krieger Institute dedicate ourselves to helping children and adolescents with disorders of the brain, spinal cord, and musculoskeletal system achieve their potential and participate as fully as possible in family, school, and community life.