

OSTEOGENESIS IMPERFECTA

What is it?

Osteogenesis Imperfecta (OI) is a genetic bone disorder affecting bone formation. It is also known as “brittle bone disease” and is caused by a mutation to the gene responsible for producing collagen. OI is characterized by bones that are easy to fracture, often from little or no apparent cause or stress. There are several types of OI and the characteristics can differ from child to child. Type 1 is the mildest form while Type 2 is the most severe. Below are the four well-known types of OI, although 3 additional forms (5-8) have also been identified.

OI Type 1	Most common, mildest form. Bones fracture easily but often before puberty with minimal bone deformity. Fractures are normally spiral. Sclera (white of the eyes) is usually purple, blue or gray tinted in color. Collagen structure is normal, but less than normal amount is
OI Type 2	Most severe, often lethal soon after birth related to respiratory complications. Bones can appear crumpled and fractured even before birth. Severe bone deformity results. Also suffer a narrow chest with underdeveloped lungs and unusually soft skull bones. Tinted
OI Type 3	Fractures and healed fractures are often present at birth. Tinted sclera similar to type 1. Loose joints and poor muscle development in extremities. Barrel shaped rib cage, spinal
OI Type 4	Severity classified between Type 1 and 3. Bones fracture easily but often before puberty. Sclera are white or near white in color (normal). Bone deformity is mild to moderate. Barrel

What are the common effects?

Monitoring for fractures or breaks is important. Assessment of signs and symptoms could include swelling, bruising, tenderness, pain, or limited movement. The child could also appear pale, sweating, or holding the limb close to the body. Children with OI not only suffer broken bones, but they also can experience muscle weakness, loose joints, skeletal deformities, short stature, delayed motor skills, brittle teeth and hearing loss. It is important to monitor for fractures, pain, joint laxity, muscle strength, spinal curvature, and muscle spasms.

What is the treatment?

There is no cure for OI. Instead, multidisciplinary treatment aims to prevent symptoms, develop muscle, strengthen bone mass and maximize independence. Fracture care is similar to any other fracture with splinting, casting and bracing for bone stabilization and healing. PT/OT services will help to strengthen muscle and promote function. For some fractures, surgical rod placement in long bones or “rodding” can be used to help with positioning and to strengthen and prevent fractures.

Some medications are prescribed to treat OI. These include bisphosphonates to increase bone mineral density. Some bisphosphonate medications have also been shown to reduce pain. Other treatments for OI include vitamin D and calcium supplements, oxygen for lung issues, dental care for teeth, and hearing aids for hearing loss.



Kennedy Krieger Institute

The Specialized Health Needs Interagency Collaboration (SHNIC) program is a collaborative partnership between the Kennedy Krieger Institute and the Maryland State Department of Education.

Suggested school accommodations

Planning for the student to attend school will require the team to consider the age of the student, the physical environment of the school, and the child's current medical condition. Supporting students with this condition in the school require educators and parents/guardian to work as a team. Some accommodations to consider for a 504/IEP could include:

- PT/OT/ SLP evaluations
- Assessment of hearing
- Modified or flexible school day
- Plan for absences and make-up work
- Adaptive devices (including technology and seating)
- Preferential setting in classroom
- Rest breaks
- Extra time between classes
- Extra set of books at home
- Clutter free areas
- Mobility concerns/accommodations
- Physical Education restrictions
- Assistance with ADL's
- Staff education/training as appropriate
- Emergency Evacuation Plan (EEP)

Specific health issues for Individualized Healthcare Plan

- Diagnosis including documentation of associated medical problems (hearing loss, scoliosis, etc.)
- Current medication list including side effects
- Past hospitalizations including fracture and surgical history
- Nutrition orders including vitamins and supplements
- Risk assessment prior to start of school
- Orders regarding injury to limbs or body
- Orders for orthotics, braces, splints and other adaptive equipment
- Orders for limitation and/or mobility restrictions
- CPR modifications considering chin lift versus head tilt
- Communicate with school staff, parents/guardian, and provider any changes or concerns about the disease
- Emergency Care Plan(s) (ECP) related to medical needs in the school setting and staff education/training as appropriate for each

Resources & Manuals

Kennedy Krieger Institute: Osteogenesis Imperfecta Clinic

<https://www.kennedykrieger.org/patient-care/centers-and-programs/osteogenesis-imperfecta-clinic>

Osteogenesis Imperfecta Foundation (OIF)

<https://oif.org/>

OIF– Checklist of possible accommodations

https://oif.org/wp-content/uploads/2019/08/College_Checklist_for_Accommodations.pdf

Brittle Bone Society– Including children who have OI in mainstream PE lessons

https://brittlebone.org/wp-content/uploads/2017/01/110798_Including_Children_In_PE_lores.pdf