



Osteogenesis Imperfecta

What is Osteogenesis Imperfecta?

Osteogenesis Imperfecta (OI) is a genetic disorder of collagen formation. Collagen is a protein found in bones and other connective tissue. People with OI have either inadequate collagen or have a poor quality collagen. This leads to weak bones that can fracture easily, thin and transparent skin and muscles that lack tone.

The severity of the symptoms of OI varies among people. Some of the common features are:

- bones that fracture easily
- short stature
- hearing loss
- discolored, brittle teeth
- blue sclerae (blue color in the whites of the eyes)
- skeletal deformities of the limbs, chest and skull
- scoliosis (curvature of the spine)
- respiratory difficulties
- weak muscles
- loose joints and ligaments
- tendency to bruise easily
- high-pitched voice
- excessive sweating
- constipation

How is OI diagnosed?

OI is initially diagnosed because of fractures that occur with little or no trauma or injury. X-rays may show fractures at different stages of healing. Sometimes, newborn babies with OI are born with multiple fractures.

How is it treated?

There is no cure for OI. The main goal of treatment is to limit the frequency of fractures and encourage mobility and independence. Sometimes a fracture can be treated with just a splint or a cast. Fractures of the long bones, such as the femur (thigh bone), may need metal rods. The rods will help with healing and prevention of fractures.

Precautions to take:

Never pull or push on an arm or leg or bend it into an awkward position.

Lift a baby with OI by placing one hand under the buttocks and legs and the other hand under the shoulders, neck and head.

Do not lift under the armpits or lift by the ankles (i.e. when changing a diaper).

Use caution if an I.V. (intravenous line) is inserted or blood pressure is taken. Pressure on the arm or the leg can result in bruising or a fracture.

The Team:

At Texas Scottish Rite Hospital for Children (TSRHC) a team will see your child in the clinic. This team includes the orthopedic surgeon, clinic nurse and therapists (physical and occupational) as needed. Our goal at TSRHC is to help your child function at the highest level possible.

If you suspect that your child has a fracture, please contact your nurse for guidance. You may be told to take your child to the nearest emergency room for evaluation. If your child does have a fracture, please follow up with your nurse so that a plan of care at TSRHC can be discussed.