

Osteogenesis imperfecta

What is Osteogenesis imperfecta?

Osteogenesis imperfecta (OI) is a congenital bone disorder characterized by brittle bones that are prone to fracture. OI usually results from abnormalities of the genes that control the production of a protein called collagen which is the main protein in bone and essential for its strength. The fragility of bone in OI is due to the collagen problems. It has nothing to do with the calcium component of bone, which is what shows up on X-rays.

Fractures are difficult to predict, especially in childhood. Some occur with normal handling. Some occur spontaneously or with so little trauma that the usual signs of a fracture may not be seen and the fracture is not identified till some weeks or months later, when an X-ray is done for another reason.

In both sexes and in almost all types of OI the fracture rate diminishes during the teenage years and remains low in adult life. The reason for this is not known.

How your child may present

Besides fractures, there may be problems in other parts of the body. Most of these are, like the fractures, the result of the defects of collagen.

- The joints may be lax.
- The whites of the eyes may be blue or grey.
- The teeth may be discoloured and fragile.
- There may be an increased liability to bruising (thought to be due to the defective collagen in small blood vessels).
- Deafness may occur.
- Hernias are more common in people with OI.
- Excessive sweating or intolerance of heat are common complaints. The cause of this is not known.
- Growth may be impaired.

How can a physiotherapist help?

A physiotherapist will be involved with your child, particularly after any fractures and will issue/order suitable equipment where appropriate e.g. crutches, wheelchair etc. They will be able to advise families and schools regarding participation in sport and exercise.

Participation in sports and exercise is recommended for children and young people with osteogenesis imperfecta (OI) to promote general fitness, muscle strengthening and bones becoming stronger but careful consideration needs to be made on activities encouraged.

Higher risk activities not recommended when you have OI:

- contact sports which involve tackles and unpredictable collisions or falls (for example, rugby, basketball, judo, football)
- trampolining
- certain gymnastic moves such as forward rolls, backward rolls, handstands and cartwheels
- jumping down from heights, for example, off gym apparatus and climbing frames
- horse-riding (this causes repetitive impact though the spine and should be carried out with caution – please discuss with your medical team before taking part)

Recommendations

- Swimming is a non-contact activity and an excellent way to improve fitness and muscle strength.
- Dance is a safe activity to participate in, but repetitive high impact and break-dancing moves should be avoided. Dancing with a partner can be unpredictable; tugging and pulling should be minimised. Holding onto a rope rather than hand-holding will enable children with OI to let go if their partner is pulling too firmly on their arms.
- Racket sports such as tennis, badminton and table tennis are recommended. Some children/young people may need a lighter racket or use a softer ball and should be encouraged to hit the ball holding the racket with two hands.
- Children/young people should be allowed to participate in the training skills of contact sports. For example, dribbling or bouncing the ball, shooting practice, throwing and catching. Children may prefer using a lighter or softer, spongier ball.
- Gym sessions should be supervised. Children/young people may need lightweight apparatus and be shown how to climb on/off equipment safely. Some activities can be carried out at floor level, for example, walking along a line on the floor instead of walking along beams/benches.
- Wearing a bright or different coloured T-shirt/top may make children/young people more visible and help avoid collisions.
- Children/young people with OI often have hypermobile (very flexible) joints. This means they may be more prone to sprains and may have weakness around their

joints - wearing supportive shoes and trainers rather than plimsolls are recommended when exercising.

- Many sports and games can be adapted for wheelchair users – please contact your physiotherapist or OT for details.

Children/young people with OI can get more tired and experience more aches and pains compared to their peers – having rest breaks during activities and not over-doing things will help to manage this.

These are just some ideas on how to promote participation in PE and sports in children/young people with OI. However every child/young person is different and strategies can be put in place to facilitate participating in specific activities/sports. If you are unsure of what activities a child/young person with OI should join in with, or wish to discuss a specific activity, you are advised to speak with your physiotherapist.