



Osteogenesis Imperfecta

Osteogenesis Imperfecta (OI) is a genetic condition present from birth. Its most striking feature is that fractures may occur easily. Therefore it is frequently called “brittle bone disease”.

Description of OI

OI is a disorder of collagen, a protein which forms the framework for the bone structure as well as other parts of the body. In OI the collagen may be of poor quality, or there may just not be enough to support the mineral structure of the bones. This makes the bones weak and fragile and results in the bones being liable to fracture at any time even without trauma.

Some people with OI, however, hardly have any symptoms but in others OI may lead to physical disability requiring the use of walking sticks, walking frames or wheelchairs.

As the composition of collagen in the bone is not correct, even when there are no fractures there will be other problems connected to the condition; such as the ligaments stretch more easily, allowing dislocation of joints. The joint hypermobility can significantly affect the quality of life as it results in fatigue of many muscle groups. As a result the mobility and performance of ordinary day tasks of everyday living are impaired. The majority of individuals with OI suffer from bone and muscle pain.



How many people have OI?

The number of people with OI in the UK and Ireland is unknown. This is due to it being difficult to diagnose. Many doctors are not familiar with the condition and many cases go undetected. The best estimate suggests approximately 1 in every 15,000 people will have OI.

How Is Osteogenesis Imperfecta Diagnosed?

No single test can identify OI. To diagnose OI, doctors look at:

- Family history.
- Medical history.
- Results from a physical exam.
- X-rays and Dexa scans to assess bone density are usually done.
- If deemed necessary genetic testing can also be carried out.

Clinical Features

OI is generally subdivided into Types. Not everyone will know what type they are and there is no such thing as a 'typical person' within the types. The main types are as follows:

Type I

This is the least severe and most common form of the disorder. The number of fractures varies greatly. Mild OI has been described as an "invisible disorder" as often the person appears to have nothing wrong with them to the casual observer. Despite appearances it is important that individual with mild OI takes care to prevent fractures.

A Child with Type I OI:

- May appear clumsy as hypermobility may increase the chance of falling/tripping.
- May tire more easily than others of their age.
- Might find as they get older that fractures sustained months or years before, have a major effect on their day to day mobility and other activities.
- May find that they have fractures more frequently.
- Ongoing dental issues.

An Adult with Type I OI:

- Generally normal height with straight limbs.
- Will generally appear physically in good health with fewer fractures from puberty to age of 50 years. This can lead to problems in people misunderstanding the condition.
- Might find as they get older that fractures sustained months or years before, have a major effect on their day to day mobility and other activities.
- Hearing problems can affect some (usually becoming evident in early twenties).
- Ongoing dental issues.

Type II

Type II is the most severe form of OI. Babies tend not to survive beyond the first few months.

- Problems with the bones in the rib cage can lead to the lungs not being fully formed. Often this leads to respiratory complications.
- Parents may have been informed of problems at an ante-natal scan, but may have had no warning that their baby has OI.
- Support needs to be immediate and ranges from providing information and a listening ear, help with financial costs i.e. equipment, hospital travel.

Type III

This is a severe form of the condition. Fractures may occur in the womb and the baby is often born with fractures. The height will be very small, arms and legs will also be bent and short.

A Child with Type III OI:

- May develop spinal curvature at an early age F Can experience a high number of fractures.
- Will normally go to mainstream schooling but commonly will need additional support to help prevent fractures.
- Babies require adapted car seats and buggies.
- Will need specially adapted wheelchairs.
- Will do all the things a child can do, but might find different ways to do them!

An Adult with Type III OI:

- Will still have fractures, but they may not have as many as when they were younger.
- The rib cage may not be properly developed leading to respiratory problems.
- Ligament problems may be evident. This can cause joint problems such as dislocations.
- Spinal curvature (where the shape of the spine is not right) must be monitored and can cause a great deal of pain as well as restriction of breathing.
- May experience pain with no obvious cause (ligament and spinal problems can both cause pain).
- Bending of long bones may be an issue.
- Hearing problems can affect some (usually becoming evident in early twenties).
- Heart problems affecting the valves and muscle may occur.

Type IV

This type falls between I and III in severity. There is huge variation in the number of fractures. Diagnosis is often not made until the person is older as the symptoms are easily missed or misdiagnosed.

A Child with Type IV OI:

- Ligament problems may be evident. This can cause joint problems such as dislocations.
- Spinal curvature (where the shape of the spine is not right) must be monitored and can cause a great deal of pain.

An Adult with Type IV OI:

- Shorter and more frequent and severe fractures in adulthood.
- May experience pain with no obvious cause (ligament and spinal problems can both cause pain).
- Bending of long bones may be an issue.
- Hearing problems can affect some (usually becoming evident in early twenties).

There are other types of OI that have been clinically identified. Type V is the commonest “unusual” OI and is important because there may need to be additional treatment when fractures occur or when bone surgery is performed. This is because a lot of extra bone (callus) can be formed compared to “normal.”

Treatments

The structure of the bone means cells are continually being replaced. Normally there is a balance between the number of old bone cells removed and new bone cells being formed. Osteoblasts are responsible for new bone formation whilst osteoclasts are responsible for removing old bone, a process known as bone resorption.

In individuals with Osteogenesis Imperfecta there is evidence of an imbalance in this process. Although there is no cure for OI, symptoms can be managed.

Treatments for OI may include:

- Care for broken bones.
- Care for brittle teeth.
- Pain medication.
- Physical therapy.
- Use of wheelchairs, braces, and other aids.
- Surgery such as rodding.
- Bisphosphonates (refer to dedicated factsheet).



Physiotherapy

Physiotherapy can help improve muscle tone and fitness. This is crucial as fractures, surgery etc can lead to periods of inactivity. Building up strength can help with pain management and also may shorten the recovery time after a fracture.

Occupational Therapy

Occupational therapy also plays an important role in terms of the practicalities of day to day living. Good quality properly assessed and fitted equipment can make big differences to individuals. Poor seating posture for example (either at a school desk or in a wheelchair) can lead to back pain, hip pain and potentially fractures in the back.

A healthy lifestyle also helps people with OI:

- Exercise (swimming, water therapy, walking).
- Keep a healthy weight.
- Eat a balanced diet.
- Do not smoke.
- Do not drink a lot of alcohol and caffeine.



Prognosis

Despite numerous fractures, restricted physical activity, and short stature, most adults and children with OI lead productive and successful lives. They attend school and further education, develop friendships and other relationships, have careers, raise families, participate in sports and other recreational activities and are active members of their communities.

Notes

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Compiled by the Brittle Bone Society in collaboration with BBS Medical Advisory Board and POINT (Paediatric Osteogenesis Imperfecta National Team). The information in this leaflet is correct as at 31st July 2021 but we cannot guarantee that it will be accurate and current at any given time. This leaflet is not intended in any way to replace the advice of your doctor or other medical professional. Leaflets are available online at www.brittlebone.org. This information is available in accessible formats on request.

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