

## Osteogenesis Imperfecta (OI)

### WHAT IS OI?

Osteogenesis (pronounced os-tea-yo-jen-er-sis) Imperfecta is a genetic condition which affects the development of collagen (pronounced koll-er-jen), a protein found in bone, skin and many other tissues in the body. In OI, defective collagen means that the bones are extremely fragile and can fracture as a result of everyday movements (e.g. opening a door or turning over in one's sleep).

**HOW IS OI PASSED ON?** The pattern of inheritance in many families with OI is autosomal dominant. Each and every child of an affected parent has a 50% (1 in 2) chance of inheriting the faulty gene and of having OI. However, there are several hundred different mutations which can give rise to OI and many people with OI have no family history of the condition. Their OI may have occurred as a result of a spontaneous genetic mutation or been inherited in a different way.

### A PERSON WITH OI MAY BE AFFECTED IN SOME, OR ALL, OF THE FOLLOWING WAYS:

- Bone fractures are a feature of everyday life. Some babies are born with fractures. Others have their first break soon after birth or when they start to move around. A child may have between 50 and 100 fractures. Each fracture is painful and can mean long periods in plaster at home or in hospital.
- The peak time for fractures is the first 15 years of life.
- In children with severe OI, a specially adapted wheelchair may be their only chance of mobility as they are likely to stand or walk. They may also be of very short stature.
- People with OI may have blue whites to their eyes (because of thin collagen) discoloured or fragile teeth (because dentine is made partly of collagen), loose joints (because joint capsules are made of collagen) and bruising and
- nose-bleeds (because small blood vessels contain collagen). They may also have problems with their hearing, with their spine later in life, and have respiratory problems (chest infections, asthma, etc..)
- Intelligence is not affected and most children with brittle bones should attend ordinary primary and secondary schools.

### OTHER INFORMATION

OI is known to have existed for at least 5,000 years and to occur in every race. In the UK, about 1 in every 10,000 babies born will have OI. Bone fractures are a feature of everyday life. Some babies are born with fractures. Others have their first break soon after birth or when they start to move around. A child may have between 50 and 100 fractures. Each fracture is painful and can mean long periods in plaster at home or in hospital. The peak time for fractures is the first 15 years of life.

OI is also known as 'brittle bones', but we prefer to use the term OI because another bone condition called osteoporosis (in which the bones become thinner and more fragile with age) is also sometimes referred to as 'brittle bones'. Some parents who take a child with OI to hospital with fractures which they cannot explain are wrongly accused of abuse. This is deeply upsetting and can have very serious consequences for the family. There is now a support group for such families within the Brittle Bone Society.

**If you are interested in finding out more about OI, you can write (enclosing an A5 stamped addressed envelope) to: Brittle Bone Society, 30 Guthrie Street, Dundee, DD1 5BS Tel: 01382-204446, Fax: 01382-206-771 [E-mail-bbs@brittlebone.org](mailto:E-mail-bbs@brittlebone.org)  
Freephone helpline: 08000 282459**