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Towards a perfect understanding

The Brittle Bone Society supports people affected by osteogenesis imperfecta. The charity's chief executive, **Patricia Osborne**, discusses what radiographers need to know about this often disabling disease

Osteogenesis imperfecta (OI) is a group of genetic bone disorders characterised by fragile bones that break easily. It is also known as brittle bone disease. A person is born with the disorder and is affected throughout their lifetime. It is a rare condition and it is estimated that the number of people born with OI is approximately one in every 15,000, equating to around 5,000 individuals in the UK.

OI is a disorder of collagen, a protein that forms the framework for bone structure. The collagen may be of poor quality, or there just might not be enough to support the mineral structure of the bones and other parts of the body, which makes them weak and fragile.

OI is caused by genetic changes that affect the amount and quality of collagen. While up to a third happen for the first time in an individual, OI may be inherited from an affected parent in the remainder.

OI varies widely in severity and there is a classification system to describe its different types. Some people with OI have hardly any concerns but, in others, OI may lead to physical disability requiring the use of a wheelchair.

OI makes bones liable to fracture at any time, even without trauma. The ligaments stretch more easily. Joint hypermobility results in fatigue of many muscle groups, impairing mobility and the performance of everyday tasks. Those with OI can also have curved bones, scoliosis, spinal cord injury, short stature, hearing loss, brittle teeth and blue sclerae.

Skeletal survey required

It is vital to diagnose OI as soon as possible so treatment can start. However, due to OI being a rare condition, getting a precise diagnosis may be challenging.

Fractures are sometimes confused with those following child abuse, given that the symptoms of OI are not always evident to healthcare professionals who are not trained in recognising the condition. Challenges in diagnosing OI can lead to inappropriate or delayed management or even unnecessary intervention.

It is important to perform X-rays of the whole body, called a "skeletal survey" that includes the skull and spine, and to have these reviewed by a paediatric radiologist looking for features such as Wormian bones (accessory skull bones), rib and vertebral fractures and bowing deformity of long bones.

Additional imaging of the cervical spine and brain may be indicated in severely affected infants for the presence of cervical spine abnormalities, which might compromise spinal cord function or hindbrain abnormalities, such as basilar invagination.

Differential diagnosis

Although both OI and child abuse are diagnostic considerations in a child with unexplained fractures, other disorders that may be considered in the neonatal period include prematurity, hypophosphatasia, mucolipidosis type II (I-cell condition) and Menkes syndrome. In children and adolescents with fractures, the differential diagnosis may include idiopathic juvenile osteoporosis, hypophosphatasia and coeliac disease, among the more commonly encountered disorders.

Visits to A&E

The nature of the condition means it is almost inevitable that all individuals with OI will present to A&E and require X-rays at some point in their lifetime.

Osteogenesis Imperfecta Federation Europe, the patient group serving and supporting OI groups across Europe, has developed a useful OI Passport (*https://oife.org/news-resources/oife-passport/*) that individuals can present at the A&E department. The passport is translated into 22 languages and covers topics such as moving and handling, pain, surgery and anaesthesia.

It is not uncommon for individuals with OI to tell the charity's support officer that their fractures have gone undiagnosed, which can lead to long-term complications and lengthy recovery time.

In a Brittle Bone Society survey on the effects of the Covid pandemic and patient experiences, 20% of individuals with OI told us they did not go to A&E to have a suspected fracture diagnosis confirmed.

Philip Plant, chair of the CoR Patient Advisory Group (PAG), says: "I am most grateful to Patricia for writing this article on behalf of the Brittle Bone Society and for highlighting the effects of this condition and the implications for both the patient and the practitioners when dealing with patients living with OI.

"An important part of the PAG's workplan is to develop relationships with relevant groups looking to benefit and improve patient experience and this relationship clearly has the potential of giving our Staff a much better understanding of this less common condition and thereby ensuring a better outcome for the patient. I look forward to meeting the Staff from the BBS Charity when more normal times return and gaining a better understanding of how we can both benefit from this relationship."

Living with osteogenesis imperfecta

What patients have told the Brittle Bone Society

"A lot of clinicians do not know what I have or have never heard of OI. It makes going to the hospital as a female frustrating because they accuse my partner of domestic abuse on a regular basis – it's got to the point that I very rarely go to A&E."

"Because we may not scream, cry or have typical swelling, we are not believed to have fractured a bone."

"OI is not recognised well enough by some professionals. For example, fractures are not picked up on X-ray because they are not obvious; joint or ligament pain is dismissed because the way the injury occurred would not affect most people. It's frustrating."

SoR comment

Lynda Johnson

SoR professional officer for the College of Radiographers Patient Advisory Group

Radiographers will be familiar with the appearances of common bone abnormalities and fractures. However, it is important not to make assumptions when someone presents with an unusual history and to be aware of the less common causes and appearances of fractures. Having an enquiring attitude and continually extending our knowledge of a wide range of conditions is an important part of continuous professional development for radiographers. Radiographers who develop their knowledge and skills in this way will contribute to the knowledge base of others, and to the improved care and quicker diagnoses for people with OI.

The College of Radiographers welcomes the opportunity to work with the Brittle Bone Society, raising awareness of the needs of people with OI and encouraging radiographers to learn more about the imaging appearances. Together we can positively impact the experience of patients coming into our care.

Contact the Brittle Bone Society

For more information about osteogenesis imperfecta and the Brittle Bone Society, visit www.brittlebone.org or contact Patricia Osborne on 01382 204446 or email at admin@brittlebone.org

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Professor Amaka C Offiah (University of Sheffield)
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OI varies widely and there is a classification system to describe the different types, ranging from mild to severe