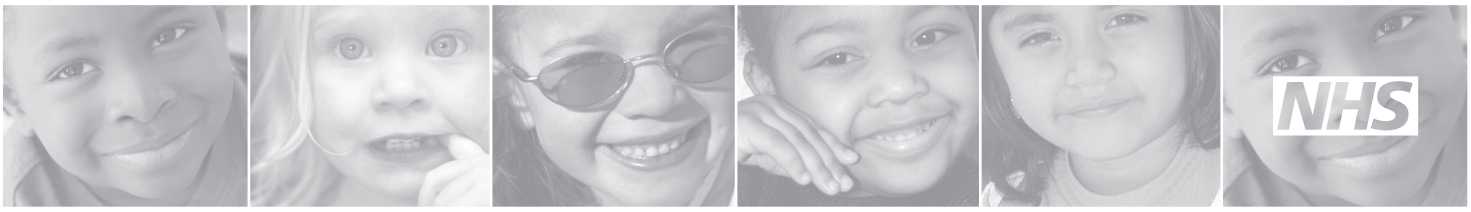

Osteogenesis Imperfecta

This information sheet from Great Ormond Street Hospital (GOSH) Osteogenesis Imperfecta (OI), what causes it, how it can be treated and where to get help.

What is Osteogenesis Imperfecta?

Osteogenesis Imperfecta (OI) is a genetic condition present from birth. Its primary feature is fractures usually caused by minimal impact.

Symptoms experienced by children affected by OI can range from very mild to severe. The severity may vary between affected members of the same family.

What causes OI?

OI is a defect where collagen (the protein that is responsible for bone structure) is missing, low or of low quality, which is not enough to support the minerals in the bone. This makes the bone weak, which in turn makes the bones easy to fracture.

What are the signs and symptoms of OI?

Below is a list of signs and symptoms that may or may not be present.

- Fractures can occur with minimal force – this varies from child to child.
- Bones may have an altered shape, for example, they may be shortened or bowed.
- The whites of the eyes may appear more blue or grey than normal.
- Children with OI can experience symptoms such as hypermobility (very flexible joints).
- Children with OI may have some degree of joint or bone pain.
- Problems with children's teeth (dentinogenesis) are common in OI.
- Children with OI may tire easier than other children.
- Hearing problems are known to affect people with OI usually after puberty.
- Small stature - Children with OI tend to be shorter than other children.

How is OI diagnosed?

No single test can identify OI in all cases. OI is diagnosed clinically in the majority of cases, that is, the doctor will carry out a physical examination of your child and take a full medical history. Imaging such as x-rays is usually suggested to check for fractures and bone changes. Bone density scans (DEXA) can be carried out on children weighing more than 10kg. Genetic testing is possible but it is not undertaken routinely. OI remains a clinical diagnosis.

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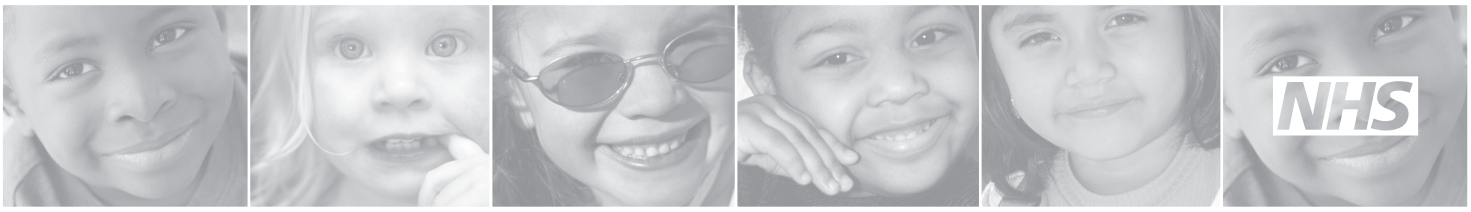

How is OI treated?

The OI Service at GOSH is a multi-disciplinary team that offers specialist advice, on-going management, and support for children and families, where there is a diagnosis of OI.

We contribute towards the diagnosis of OI, and discuss with you and your child what it might mean to have OI. Through a process of assessment and consultation, we make recommendations that will support your child in their on-going development and practical day-to-day management.

If there is an orthopaedic or fracture management query, discuss this with the local team in the first instance. If they need advice or an opinion, they can call the OI Team at GOSH or the orthopaedic registrar on call.

The service is one of four highly specialised services in England for children with OI.

The main treatment is a drug called pamidronate. Pamidronate is a type of bisphosphonate, which is a medicine that prevents loss of bone mass. Bisphosphonates have been used for a variety of conditions in childhood where there is bone pain and immobility in association with osteoporosis and/or abnormal bone formation. Pamidronate strengthens the bone by increasing bone density and corrects the imbalance between bone reabsorption and bone formation which occurs in OI.

Your child may not require treatment soon after diagnosis but could benefit from it a few months or years later. Some children never need pamidronate.

Vitamin D supplements are recommended to all patients with OI, as Vitamin D is necessary to help the body absorb calcium and make bone.

With some adaptations, children and young people can have a near-normal lifestyle, attending school and college and starting work.

What happens next?

Your child will continue to need regular check-ups from the OI service throughout childhood and adolescence, before moving on to an adult OI service around 16 to 18 years old.

Further information and support

At Great Ormond Street Hospital (GOSH), contact the OI Team via our Administration Coordinator on 020 7405 9200 ext 5293. If you have a clinical question, please contact our Clinical Nurse Specialist (CNS) for OI on 020 7405 9200 ext 5824.

The **Brittle Bone Society** offers support and advice to parents of children with all types of OI. Telephone them on 01382 204 446 or visit their website at www.brittlebone.org.

Compiled by the Osteogenesis Imperfecta team in collaboration with the Child and Family Information Group

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www.gosh.nhs.uk

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