



Great Ormond Street Hospital for Children NHS Foundation Trust

# Osteogenesis Imperfecta: information for families

**Osteogenesis Imperfecta (OI) is a genetic condition present from birth. Its primary feature is fractures usually caused by minimal impact. This information sheet from Great Ormond Street Hospital (GOSH) describes osteogenesis imperfecta (OI), what causes it and how it can be managed. It also tells you about the highly specialised service for OI based at GOSH.**

Osteogenesis Imperfecta (OI) is a defect where collagen (the protein that is responsible for bone structure) is missing, reduced or of low quality, so 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?

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.

- 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.
- Joints can be hypermobile or very flexible
- Some degree of joint or bone pain may be present
- Problems with formation of teeth (dentinogenesis, or DI)
- Children with OI may tire easier than other children

- Hearing problems are known to affect people with OI usually after puberty.
- 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, but there is not enough data available to give accurate results for children under five years old. Genetic testing is possible but it is not undertaken routinely. OI remains a clinical diagnosis.

## How is OI treated?

The majority of patients with mild or moderate OI are treated with physiotherapy and management of symptoms as they occur. The main medications used are called pamidronate and zoledronic acid. Both are 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.

Bisphosphonates strengthen the bone by increasing bone density and correcting 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 bisphosphonates.

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. We will start to discuss this when your child is 12 years old.

## The OI Service at GOSH

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 ongoing development and practical day-to-day management.

If there is an orthopaedic or fracture management query, please discuss this with your 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.

## Members of the team

The team have specialist knowledge and expertise in the medical and practical management of children with OI. The team includes the following professionals. Who you see at an appointment will vary depending on your child's needs.

**OI Consultant Paediatric Doctors:** review medical details including investigations, diagnostic information and contribute to the ongoing monitoring of your child. All the consultants have experience in OI and come from different specialities such as endocrinology, neurology, neurodisability or orthopaedics.

**Physiotherapists and Occupational Therapists:** assess how your child is managing on a day-to-day basis with life skills, including physical activity and self-care. They will consider how your child is managing at home and in school. The therapy team offer strategies, activities and exercises to promote independence. Therapists may be involved in rehabilitation, for example following an acute injury or after orthopaedic surgery.

**Clinical Nurse Specialist:** supports families and children/young people through investigations and medical treatments, and assists with any health and medical queries. The Clinical Nurse Specialist is the first port of call for medical OI queries.

**Psychologist:** considers the psychological wellbeing and resources of children, young people and families living with OI and offers support, advice, treatment and onward referral where necessary.

**Social Worker:** provides emotional and practical support for the child and their family, advocating

on a child or family's behalf and provide consultation and advice.

**Administrative staff:** organise appointments and other administrative queries. The OI team may refer your child onto other professionals within GOSH. This could include the dental team, orthotics department or other specialist doctors.

## Who we see

We see children from birth to 18 years of age who have a confirmed diagnosis of OI as well as those who need further specialist assessment to establish diagnosis. We tend to see children from South East England but can see them from any location in the UK if referred.

In most instances where there is a confirmed diagnosis of OI, children will remain under the care of GOSH as a specialist service, alongside support and input from their local team.

## Clinic visits

### Before the first appointment

We will ask the referrer to send results of any investigations your child has had and other relevant information available, such as x-rays. Sometimes we ask for more investigations to be done before your child comes to an appointment with us. These may be done before the appointment or sometimes on the same day. Once all this information has been provided, we will arrange an appointment time with you.

Initial appointments may need you to be at the hospital for most of the day. This is to allow time for any investigations to take place, for your child to meet all members of the team, and have assessments carried out.

The first appointments are held at 09:30 and 10:30 on a Friday morning only. You may be required to attend up to an hour before your appointment time for x-rays to be taken. They are

taken before the appointment so the results can be discussed with you in the clinic appointment.

There are many cafes and restaurants in the area but we advise that you bring snacks, drinks and other essentials such as nappies and toys.

For information on travelling to the hospital and parking, please visit the GOSH website.

### Follow up appointments

These can last up to three hours but are shorter when no investigations are required. They are run on Fridays with additional joint OI and orthopaedic clinics held once a month on a Wednesday.

Your child will need to have regular investigations, which we try to organise on the same day as your appointment. The administrator will inform you, usually two to three days before the appointment, what time to attend these.

If you have two children who have been referred at the same time, we will, wherever possible, give you an appointment for both children on the same day to avoid you needing to attend the hospital more than needed.

### What to bring to appointments

It would be helpful if you could bring:

- A list of any medications (including vitamins and supplements) your child takes and the doses. You can take a photo on your mobile phone of the medications to show us if this is easier.
- The names of any doctors involved in your child's care
- The names and addresses of anyone else you would like to receive a copy of your clinic letter.
- A list of how many fractures your child has had since birth, or for follow up patients since we last saw you.

## **Assessment**

When you meet the team, a history or update of how your child has been will be taken. This will include information about any fractures or pain your child may have had. It may be helpful to keep a note of this for the appointment.

The doctors and nurse will carry out a general examination of your child, discuss any questions or concerns you may have and will explain the results of any investigations that have taken place. The use of drug treatments may be discussed.

When your child is older, they will be encouraged to spend a short time on their own with the doctor to discuss any questions they may have as part of the transition process from paediatric to adult health services.

As part of the assessment and for your child's clinical record we may ask you for permission to video your child.

The Occupational Therapist and Physiotherapist will assess your child's physical and practical abilities. The therapists are very experienced in how to assess and handle children who have OI. However, therapists will be guided by you and your child about the best way to manage this.

The therapists will look at how your child is moving, how strong they are and how they are getting around. They will also assess how your child manages activities such as dressing, writing, and playing. Children may like to bring a pair of shorts for this assessment.

For young babies, the therapists will advise on handling and management. They will be able to discuss strategies that you may find useful at home, to make activities such as bathing and dressing easier. Therapists also advise on activities to promote your child's motor development.

For older children, the therapists will be able to provide advice around posture, mobility, and daily living skills.

The Psychologist will want to better understand the impact of the OI on your child and family, and may talk with you about your family system and the strengths, resources and challenges it brings. If you prefer to have these conversations separately from your children this can be arranged – please ask the administrator before your clinic appointment. The Psychologist may use questionnaire measures to help identify children and families who could benefit from additional support.

A social worker can be available for practical and emotional support if required.

As GOSH is a teaching hospital, there may be visiting professionals present during your appointment; however we will consult with and seek your agreement for this.

## **Feedback**

Following completion of the assessment the team will discuss their findings with you and your child. A follow-up plan will be made. Children are usually followed up six monthly or annually depending on their needs.

Following feedback, the therapists, nurse and/or psychologist may spend longer with some children to give specific advice on activities, or to spend a little more time in discussion.

A multidisciplinary report will be written and sent to you, your family doctor (GP) and paediatrician. If it is beneficial to your child, the report can also be sent to any other professional as advised by you. The aim is to send this out within three weeks of the appointment.

## **Outreach**

Some team members are able to offer an outreach service to come to see your child at home, school or when in another hospital. This occurs in collaboration with your local team members where possible.

Outreach appointments tend to be offered to babies with a new diagnosis, for children needing additional support/advice in the nursery/school setting, and for one-off problem solving. They do not replace the routine follow up appointments at GOSH, and often do not include review by a doctor.

## **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.

There are four national services commissioned by NHS England for children with OI. GOSH is one of these and the others are Birmingham Children's Hospital, Bristol Children's Hospital and Sheffield Children's Hospital.

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](http://www.brittlebone.org)

## **Working with local teams**

The OI service supports your child by offering specialist advice and information to you and your local team. All children remain under the care of their local paediatrician, therapy and support services.

## **Emergencies**

Any emergencies, such as fractures, need to be managed by your local hospital's Accident and Emergency Department. If staff at the local hospital have questions about your child's fracture management, the local Orthopaedic Doctor will need to call the on-call orthopaedic registrar at GOSH via the Switchboard on 020 7405 9200.