



Great Ormond Street Hospital for Children NHS Foundation Trust: Information for Families

Highly Specialised Osteogenesis Imperfecta Service

What is Osteogenesis Imperfecta?

Osteogenesis Imperfecta (OI) is a rare disorder of collagen which makes the bones weak and fragile, and results in bones being more likely to fracture at any time sometimes without trauma. It is a multi-system disorder which may result in bony deformity, can affect growth, and may cause pain and low levels of stamina. Tooth development and hearing may be affected. OI can be divided into different types, and the severity of OI can vary significantly between children.

Motor development and functional abilities can be affected and ongoing monitoring and advice regarding management of OI is essential the best long term outcome.

What is the Osteogenesis Imperfecta Service?

The OI Service at Great Ormond Street Hospital (GOSH) is a multidisciplinary team that offers specialist advice, ongoing 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.

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.

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, and 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 through investigations and medical treatments, and assists with any health and medical queries.

Psychologist: considers the psychological well-being and resources of children 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 are a point of contact if you need to speak with another member of the team.

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.

The clinic visit

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 require 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.

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 refer to the GOSH website.

Follow-up appointments may last up to three hours but are shorter when no investigations are required.

Assessment:

Your child will be required 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.

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 – you may find the table below useful to record fractures. 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 you and 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 and/or psychologist may spend a little 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.

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.

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.

Further information and support

If you have any queries, please contact our Medical PA on 020 7405 9200 ext. 5293. Further information about the OI service is available online at www.gosh.nhs.uk/medical-information/clinical-specialties/neurodisability-information-for-parents-and-visitors/clinics-and-services/osteogenesis-imperfecta-service.

The Brittle Bone Society (BBS) is a charity set up to address the needs of people born with Osteogenesis Imperfecta (OI) in the UK and Republic of Ireland. The Charity work towards improving the quality of life for people diagnosed with OI in the UK and the Republic of Ireland, providing advice, signposting and information, providing financial support for wheelchairs and equipment and raising awareness of the condition. The BBS and the Highly specialized OI service at Great Ormond Street work together and meet at least annually. Telephone them on 01382 204 446 or visit their website at www.brittlebone.org.



Fracture Number Since last OI Appointment	Date	Hospital X-ray and Treatment taken at	Fracture Site	Fracture management
1				
2				
3				
4				
5				
6				
7				
8				
9				
10				

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