

Thalassaemia

Information for you, your family
and carers

What is thalassaemia?

Thalassaemia is a lifelong condition and is caused by faulty genes that children inherit from their parents. It reduces the amount haemoglobin you produce – a substance that helps red blood cells carry oxygen round the body. Having less haemoglobin can cause anaemia, making you feel tired.

Most babies are born without problems as they make the infant version of haemoglobin (HbF) normally. Children can start to display symptoms as they get older and cannot make the adult version of haemoglobin (HbA) they need.

Treatment will often be given as blood transfusions, but it depends on the type of thalassaemia and how severe it is. We assess everyone individually and start treatment before they develop the effects of the disease. Treatment could start in infancy (thalassaemia major) or later in life (thalassaemia intermedia treated as major). Some people never need blood transfusions or only need them occasionally (thalassaemia intermedia).

What kind of problems can happen in thalassaemia?

Thalassaemia is a serious condition and will need close monitoring but treatment has been shown to improve health and can be used to manage any problems.

Problems related to anaemia

Anaemia makes you feel tired because not enough oxygen is delivered to the body. As a result, the bone marrow tries to make more red blood cells. In people with thalassaemia major, this does not work.

Anaemia can cause growth and development to slow down but treatment usually starts before this happens. If left untreated, a child will often feel poorly and their tummy may swell due to the large liver and spleen. The bone marrow can also expand and cause bone thinning and swelling of the cheek bones and forehead, causing a characteristic facial appearance.

With regular blood transfusions to keep haemoglobin at a good level, children with thalassaemia major grow well and look like any other healthy child. In thalassaemia intermedia a child grows and develops at a similar rate to other children even if the haemoglobin is low. Careful monitoring helps to make sure children do not have any lasting harm from anaemia.

Problems related to blood transfusion

Blood transfusion is a lifesaving treatment in thalassaemia major patients but it does cause a build-up of iron in the body. There is no natural way for the body to get rid of the

iron contained in red blood cells so every time a transfusion is given, more iron accumulates.

Too much iron (iron overload) is harmful, and can cause serious problems from iron depositing around the body in vital organs. This can cause diabetes, failure to grow and go through puberty, infertility, an underactive thyroid gland, liver disease and heart disease. Drugs are prescribed to get rid of the extra iron and prevent it from building up. These drugs are called chelators and treatment with them is known as chelation.

In thalassaemia intermedia patients may need transfusions at times of infection or illness and during pregnancy, which can also cause iron to build-up. Even patients who do not require transfusions can develop iron overload. This is due to the body absorbing too much iron from food. We monitor all patients for signs of iron overload using a variety of tests.

Infection

People with too much iron in their body do not respond well to infection. They can become very sick very quickly and may get unusual types of infection. It can be particularly difficult to fight infection if iron has built up in the heart.

If you/your child develop any symptoms that may suggest you have an infection, such as fever, chills, diarrhoea or vomiting, weakness of the limbs or palpitations (heart beating fast or unusually) you should go to the emergency department (A&E) immediately. If you are worried about feeling unwell, get in touch with the team using the contact details at the end of this booklet.

Other problems

There are other problems associated with the condition but they do not affect everyone. They tend to be more common as people get older, and include:

- enlargement of the spleen
- bone thinning
- infertility
- side effects of chelation
- gallstones
- kidney stones
- extramedullary haemopoiesis – this is when bone marrow grows outside of the bone. This can be a particular problem if the growth becomes big or presses on something important like a nerve.

Having treatment at Evelina London

The aim of our care is to keep you/your child out of hospital and make sure life you can lead your life as healthily and normally as possible. Regular reviews in the haemoglobinopathy clinic are an essential part of this, as this helps to identify and treat any problems quickly. You may also need to see other specialists in endocrinology

(for bones and hormone balance), cardiology (for the heart) and hepatology (for the liver).

People with thalassaemia major need regular blood transfusions, usually every 3 to 4 weeks. These are given in the day care unit. The aim is usually to keep the haemoglobin above 90 grams per litre, although this may be adjusted when needed.

If you are having regular transfusions, once the level of iron in the body (ferritin level) approaches a certain amount you will be offered chelation therapy to get rid of the extra iron. A common medicine is deferasirox (also known as Exjade), a tablet that is taken once a day. Desferrioxamine (also known as Desferal), is also commonly used and is given over several hours through an injection under the skin several times a week, is also used by many people with thalassaemia. Deferiprone (also known as Ferriprox or L1), a tablet/liquid that is taken 3 times a day, is used less commonly but may be particularly effective if there is iron overload in the heart. Your doctor will advise on the most suitable choice.

For some patients an alternative to transfusion is the drug hydroxycarbamide (also known as hydroxyurea). This works best in people with less severe forms of thalassaemia.

Where should I go in an emergency?

If you/your child are very unwell or have any of the symptoms listed below, you should go to the Emergency Department (A&E) immediately:

- fever 38 degrees or above, chills
- diarrhoea or vomiting
- palpitations
- new weakness.

If you/your child is unwell and you are worried then call the clinical nurse specialist or, the haematology registrar outside of working hours, for advice. They may suggest you go to the day care unit, see your GP, go to A&E or bring forward your clinic appointment. Contact details are on p11.

Please note: if you have an infectious illness, for example diarrhoea, vomiting, chicken pox and so on, do not come to clinic or the day care unit directly. Please contact the clinical nurse specialist or haematology registrar who will make arrangements for you to be reviewed elsewhere.

What can you do to stay well?

- If you need regular blood transfusions, schedule these so that your haemoglobin is kept above 90 g/L
- Take your chelation therapy
- Make sure infections are treated quickly
- Maintain a good balance of nutrition and activity
- Ensure your vaccinations are up to date

- Avoid smoking
- Take penicillin twice a day for life if your spleen has been removed
- Keep your clinic appointments so that we can review your health and monitor you for complications of thalassaemia. This is a requirement stipulated in the National Standards produced by the UK Thalassaemia Society in conjunction with the Department of Health. These appointments are usually every 3 to 12 months if you are well but may need to be more frequent if you develop health problems.

If your/ child's appointment is not convenient, please change it. We understand that many of you have busy lives with other commitments and will accommodate this wherever possible.

Notes

Further information

Sickle Cell and Thalassaemia Conference

This annual international conference is held at Guy's and St Thomas', with world experts in haemoglobinopathies. For more information visit the websites below. If you are interested in attending, please email Dr Baba Inusa.

website: www.scorecharity.com

website: www.ststn.co.uk

email: baba.inusa@gstt.nhs.uk

Sickle Cell Society

Provides help and support for people affected by sickle cell disorders and their carers.

phone: 0800 001 5660

website: www.sicklecellsociety.org

The Sickle Cell & Thalassaemia Centre

Provides information about sickle cell disease.

Thalassaemia counsellors can also offer support and information. The centre can also direct you to local services.

phone: 020 7414 1363

Contact us

Lead consultant for haemoglobin disorders, and consultant in haematology, phone: 020 7188 6203 (secretary), **email:** Dudhma.Banu@gstt.nhs.uk

Lead nurse, phone: 020 7188 9432 or 07918 338730
email: HaemoglobinopathyCNS@gstt.nhs.uk

Nurse specialist, phone: 07771 345847 or 07517 592436 **email:** HaemoglobinopathyCNS@gstt.nhs.uk

Research nurse, phone: 020 7188 6637 or 07517 592436

Specialist nurse – community, phone: 020 3049 5993 or 07880 781545

Therapy and psychology team, phone: 020 7188 9125 or 07817 905844

Help and advice will always be available if you have any questions or concerns. Out of hours, please call the hospital switchboard, **phone:** 020 7188 7188 and ask them to bleep the haematology registrar.

If you need to come to emergency department (A&E) at St Thomas' Hospital, please telephone the department before you arrive, **phone:** 020 7188 2111 and let them know about your child's condition. This will help to improve your child's care when you arrive.

Evelina London Medicines Helpline

If you have any questions or concerns about your child's medicines, please speak to the staff caring for them or contact our helpline.

phone: 020 7188 3003, Monday to Friday, 10am to 5pm

email: letstalkmedicines@gstt.nhs.uk

Your comments and concerns

For advice, support or to raise a concern, contact our Patient Advice and Liaison Service (PALS), **phone** 020 7188 8801 **email** pals@gstt.nhs.uk. To make a complaint contact the resolution department **phone** 020 7188 3514 **email** complaints2@gstt.nhs.uk

Language and accessible support services

If you need an interpreter or information about your care in a different language or format, please contact the department your appointment is with.

NHS 111

This service offers medical help and advice from fully trained advisers supported by experienced nurses and paramedics. Available over the phone 24 hours a day.

phone: 111 **website:** www.111.nhs.uk

NHS website

This website gives information and guidance on all aspects of health and healthcare, to help you take control of your health and wellbeing.

website: www.nhs.uk

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