Clinical haematology

About thalassaemia

Information for patients, relatives and carers

Introduction

The purpose of this leaflet is to provide patients who have sickle cell disease (SCD) with a brief guide to the condition and an overview of the services provided by Imperial College Healthcare NHS trust.

What is thalassaemia?

Thalassaemia is a lifelong red blood cell condition caused by a genetic abnormality of a substance in the blood called haemoglobin.

Haemoglobin is a red protein responsible for carrying oxygen around the body in the blood. People with thalassaemia have a gene that results in a decrease in the amount of normal haemoglobin produced.

- Before birth, our bodies usually make a type of haemoglobin known as haemoglobin F (HbF – F is for fetal).
- After birth, production switches to haemoglobin A (HbA – A is for adult). Normally by 18 months of age, very little HbF is produced and most haemoglobin is HbA.

Most babies with thalassaemia are born without any problems, because they make HbF normally. However, as the babies get older, their HbF production stops and there is no HbA being made to take over. This causes a condition called anaemia, which means you do not have enough red blood cells.

The way this happens varies in different people and often depends on the type of thalassaemia. When the haemoglobin is low the body responds by trying harder to make red blood cells. Nowadays, we assess each person individually and start treatment with blood transfusions (when you're given blood from someone else, called a ‘donor’) before they develop the effects.

Thalassaemia complication

Thalassaemia complications may result from:

- the condition itself (anaemia)
- the effect of excess iron from blood transfusions
- the effect of medications taken to reduce the amount of excess iron in the body

There is good evidence to show that sticking to treatment improves the health and well-being of people living with thalassaemia.

Problems related to anaemia:

Anaemia makes people feel tired because there is not enough oxygen being delivered to the body. As a result, the bone marrow (spongy tissue inside some of the bones in the body) tries to make more red blood cells.

During childhood:

- anaemia slows down growth and development
- the child often feeds poorly
- their tummy may swell with the large liver and spleen
- bone marrow can also expand. This causes bone thinning and swelling of the cheekbones and forehead, giving rise to a typical facial appearance.

In children, these features are usually a sign they need to start a transfusion programme. This involves regular blood transfusions to keep the haemoglobin high enough. This is usually needed every 3 to 4 weeks. Regular transfusion can reduce the symptoms of anaemia or get rid of them completely.

If their haemoglobin is kept at a good level, children grow well and look like healthy children. Regular transfusion is equally important for adults, as many of the same complications arise when the haemoglobin is too low. It is important to watch each adult and child carefully to ensure they do not suffer any lasting harm from anaemia.

Problems related to transfusion:

Blood transfusion is lifesaving in thalassaemia patients. Red blood cells contain iron, which is an essential component of haemoglobin. The body has no natural way to get rid of excess iron, so every time a transfusion is given, more iron builds up.

Too much iron (iron overload) is harmful, so drugs are prescribed to get rid of the extra iron and prevent this from building up.

These drugs are called chelators and treatment with them is known as chelation.

Importance of modern chelation therapy

In the past, when chelators first became available in the 1970s, patients often developed serious complications from iron overload, and some died as a result.

The harmful iron would build up around the body in vital organs such as the heart, pancreas, and hormone-producing glands. This resulted in serious problems, including heart disease, diabetes, failure to grow and go through puberty, infertility, an underactive thyroid gland and liver disease.

Heart disease is the most dangerous of these complications. It is the most common cause of death for people with thalassaemia who do not have access to modern chelation treatment, for example, people in developing countries.

Good chelation therapy avoids all these dangers.

Some thalassaemia patients may need transfusions when they have an infection or are ill. Iron can also accumulate during pregnancy and, over time, cause the same problems. Some thalassaemia patients have iron overload due to their bodies 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 respond poorly to infection. They can become very sick very quickly and may get unusual types of infections. It can be particularly difficult to fight infections if iron has built up in the heart.

Other problems:
These do not affect everyone. They tend to be more common as people get older.

- 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 it becomes big or presses on something important like a nerve

Treatment

The aim of the care we provide for thalassaemia is to:

- keep you out of hospital
- ensure you lead as healthy and normal a life as possible

Regular review in the red cell clinic to identify and treat any problems at an early stage is essential. You may also need to see other specialists, in:

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

Blood transfusions

Blood transfusions are given in the day care unit. This is on the ground floor of the Garry Weston centre. It runs Monday to Sunday, from 08:00 to 20:30.

You will need to have a special blood test 2 days before your transfusion. Both appointments will need to be booked in advance.

MRI scan

MRI (magnetic resonance imaging) scan of your heart and liver are taken at 6 or 12 monthly intervals to monitor the levels of iron there. This will allow your doctor to determine the correct dose of iron chelators you need to remove the iron efficiently.

Drug alternative

For some patients, an alternative to transfusion is the drug hydroxycarbamide (also known as hydroxyurea). If you would like more information on this drug, please ask your nurse. This works best in people with less severe forms of thalassaemia who continue to produce HbF.

What is an emergency?

If you have any of the symptoms listed below or are very unwell and need to be seen straight away, you should go immediately to A&E.

Things that require you to attend immediately are:

- fever of 38oc or above, chills
- diarrhoea or vomiting
- abdominal pain
- feeling very unwell
- palpitations
- new weakness

Where to go in an emergency

- If you have a non-urgent medical problem or a medical problem unrelated to your thalassaemia, for example, a rash or a twisted ankle, then it is appropriate for you to see your GP or go to your local A&E department. They can always discuss your case with us.
- If you have a non-urgent problem related to your thalassaemia, then you can ask for your outpatient appointment to be brought forward. Your clinical nurse specialist can arrange this.
- If you are unwell then you should call the clinical nurse specialist. If it is out of hours, call the red cell registrar through the hospital switchboard for advice. This may be to attend the day care unit, see your GP, go to A&E, or bring forward your clinic appointment.
- Note: if you have an infectious illness, for example, diarrhoea (runny poo), vomiting, or chicken pox, DO NOT visit clinic or the day care unit directly. Please contact the clinical nurse specialist or red cell registrar who will plan for you to be reviewed elsewhere.

Genetic testing

Thalassaemia is an inherited condition.

It most often arises when a child inherits an affected gene from both parents who are carriers (trait). In this situation, the probability of having a child affected by thalassaemia is 1 in 4 (25%) in every pregnancy.

If one parent has the disease the risk is greater – 1 in 2 (50%). If a child inherits a thalassaemia gene from one parent and a sickle gene from the other, this results in a type of sickle cell disease. For more information you can also read the leaflet ‘About sickle cell disease’.

The genetics of thalassaemia are complicated. You can discuss or ask any questions about this when you attend the red cell clinic.

If you are planning to start a family, you should arrange for your partner to be tested before you get pregnant if they do not know whether they are a carrier. If this shows you are risk of having an affected child, the option of prenatal diagnosis will be discussed. If you choose to have this, please tell us immediately you or your partner becomes pregnant.

Obstetric care

It is important that you receive the optimum care during your pregnancy as early as possible. You and your baby will be followed-up and monitored closely at regular intervals in the joint obstetric/haematology clinic.

Women with a history of cardiac complications and iron overload are at particular risk. If you plan to become pregnant, we recommend that you are seen for assessment by a consultant cardiologist before conception. You may need to see other specialists before conceiving to prepare you for pregnancy.

How to stay well as a person with Thalassaemia

- If you need regular blood transfusions, schedule these so that your Hb is kept above 9.5 g/dl.
- 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.
- Continue your folic acid (5mg) once a day.
- Take penicillin twice a day for life if your spleen has been removed.

Make sure you keep your clinic appointments so that we can review your health and monitor you for complications of thalassaemia. These appointments are usually every 3 to 12 months if

you are well but may need to be more frequent in you develop health problems.

The national standards, produced by the UK Thalassaemia Society with the Department of Health and Social Care, say you must have these appointments.

If your 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.

Vaccination advice

Make sure you are up to date with routine immunisations and hepatitis B vaccine.

Check any necessary travel vaccines and arrange these through your GP practice or a travel clinic at least six weeks in advance. If you are visiting an area that requires yellow fever vaccination and are currently taking the drug hydroxycarbamide, please discuss this with your hospital specialist as it may not be recommended.

If you have thalassaemia and have had your spleen removed, check you have received the following vaccines:

- Pneumovax (given every 5 years)
- Hib – haemophilus influenzae type
- MenC – meningococcal C
- MenACWY – meningococcal ACWY conjugate
- Hepatitis B
- Influenza (given by your GP in the autumn or winter months every year)

Travel advice

- **Malaria** – It is vital you take effective precautions if you travel to or stop over in a country where malaria occurs. Your GP or hospital specialist can advise on the choice of antimalarial medication. Thalassaemia does not protect you from malaria. You should take the relevant antimalarial tablets prescribed to the country or area where you are going to stay.
- **Diarrhoea** – Diarrhoea is common among travellers abroad. If you have thalassaemia this can cause dehydration, which can cause you to become unwell. To prevent this, it is advisable to carry a supply of oral rehydration salt sachets. You can buy these from your local pharmacy. If you develop fever or notice blood in the stools you should seek immediate medical advice.
- **Other infections** – Get treatment urgently for any bites (especially dog) bites as these can lead to serious infections. Any bites should be kept clean. Seek medical advice if

there are any signs of fever or infection. Ensure you use insect repellent containing DEET, wear protective clothing and use mosquito nets.

- **When flying** (especially for 6 hours or more) make sure that you:
    - keep mobile, warm, and well hydrated throughout the flight
    - drink plenty of fluid, preferably water
    - avoid alcohol, as drinking it could make you dehydrated.
    - carry warm clothing or use a blanket to prevent chilling – cabin temperatures are often cold
    - wear flight socks to reduce the risk of thrombosis. These can be bought in most large pharmacies

Reproductive and sexual health

You can discuss contraception with your GP, local family planning clinic or clinical nurse specialist. Please remember to tell us of any changes in medication.

Some people with thalassaemia develop fertility problems due to iron overload. These require specialised assessment and treatment for which you will be referred to the obstetrician at the assisted conception unit.

Sexual health advice is available through the Jefferiss Wing Sexual Health Clinic at St Mary's Hospital, Praed Street, London W2 1NY (Tel: 020 3312 1697).

This offers a walk-in service.

Contact details

Hospital switchboard	020 3313 1000
Clinical nurse specialists	020 3313 8372, 020 3313 4655
Specialist social worker	020 3313 3713
Specialist psychologist	020 3313 3357

How do I make a comment about my visit?

We aim to provide the best possible service and staff will be happy to answer any of the questions you may have. If you have any **suggestions** or **comments** about your visit, please either speak to a member of staff or contact the patient advice and liaison service (**PALS**) on **020 3313 0088** (Charing Cross, Hammersmith and Queen Charlotte's & Chelsea hospitals), or **020 3312 7777** (St Mary's and Western Eye hospitals). You can also email PALS at

imperial.pals@nhs.net The PALS team will listen to your concerns, suggestions or queries and is often able to help solve problems on your behalf.

Alternatively, you may wish to complain by contacting our complaints department:

Complaints department, fourth floor, Salton House, St Mary's Hospital, Praed Street
London W2 1NY

Email: ICHC-tr.Complaints@nhs.net

Telephone: **020 3312 1337 / 1349**

Alternative formats

This leaflet can be provided on request in large print or easy read, as a sound recording, in Braille or in alternative languages. Please email the communications team: imperial.communications@nhs.net

Wi-fi

Wi-fi is available at our Trust. For more information visit our website: www.imperial.nhs.uk

Clinical Haematology
Published: December 2023
December 2026
Reference no: 160