University College Hospital

Thalassaemia
(transfusion-dependent and non-transfusion-dependent)

North Central London Haemoglobinopathy Network jointly with Whittington Health, Royal Free London, and Luton and Dunstable NHS Foundation Trust

Joint Red Cell Unit
If you need a large print, audio, braille or translated copy of this booklet, please contact us on 020 3447 9638. We will do our best to meet your needs.

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Introduction

This booklet explains more about thalassaemia and provides an overview of the services offered by University College London Hospitals NHS Foundation Trust (UCLH).

If you would like more information, please ask for our leaflet ‘Guide to services for people with red cell and iron disorders’.

What is thalassaemia?

Thalassaemia is a disorder of the body’s red blood cells that is genetically inherited and lifelong. It is caused by an abnormality in the genes that control haemoglobin – the part of red blood cells that carries oxygen around the body. This results in the reduced amount of normal haemoglobin. Most of the normal haemoglobin produced by the foetus in the womb is Haemoglobin F (HbF – F is for foetal), and shortly before birth the production switches to Haemoglobin A (HbA — A is for adult).

By 18 months of age children with normal haemoglobin production make very little HbF and most haemoglobin is the HbA type. In most cases of thalassaemia, babies are born without any problems as they make the HbF normally. As they get older, the production of HbF stops and they cannot make HbA. The way this happens is different in different people and often depends on what exactly the genetic problem is.

When the haemoglobin gets low, the body will try even harder to produce the red blood cells. However, as they don’t contain much haemoglobin, the person becomes anaemic. This effort can make some of the organs too big, for example the liver and spleen, and change the shape of the face.

Nowadays, we assess each person individually and start treatment before any significant side effects of the disease are present. The timing of this can vary from infancy (previously known as thalassaemia major) to later in life (previously known as thalassaemia intermedia treated as major).
The name of these disorders has now changed and is referred to as transfusion-dependent thalassaemia (TDT).

Some people may not need transfusions until much later in life, or perhaps never. These people have non-transfusion-dependent thalassaemia (NTDT).

**What kind of problems can happen in thalassaemia?**

The problems in thalassaemia may result from the thalassaemia itself, the excess iron following blood transfusions, the excess iron that may be absorbed from the diet, or the medications taken to reduce the excess iron in the body. Closely adhering to treatment plans is linked to better health and survival in thalassaemia.

**Problems related to anaemia**

Anaemia makes people feel tired and can cause a decrease in growth. The body will recognise that it is anaemic and try to make more blood. Once significant anaemia develops, babies may feed poorly. Older children may lose their appetite and their tummy may swell with the large liver and spleen. The bone marrow can also expand to give characteristic bone thinning with swelling of facial bones, which is typical of thalassaemia. These features are usually a sign that the child will need to start a transfusion programme.

Good management will involve regular blood transfusions, usually every three to four weeks. This is to keep the haemoglobin high enough so that these symptoms are reduced to an absolute minimum or are not present at all. If the haemoglobin is kept at a good level (usually higher than 95g/L), the child will grow well and look like any other healthy child.

Patients with NTDT may get to a point where they too need to have regular transfusions for similar reasons as those who started transfusions in early childhood. Sometimes people with NTDT need an occasional or a course of transfusions due to a temporary issue, such as infection or pregnancy.
Problems related to transfusion
Blood transfusion can be lifesaving in thalassaemia patients but it comes at a cost. Each time blood is given the patient will also receive iron which is an essential part of haemoglobin. There is no natural way to get rid of iron from the body, therefore we must use medications to help remove the extra iron. These medications are called iron chelators and the treatment is called iron chelation therapy.

In the 1950s when these medications were not available, patients developed serious complications from iron overload and most died as a result. If the extra iron is not removed, it deposits around the body, mainly in the liver but also in vital organs such as the heart, pancreas and hormone producing glands. This results in serious problems, for example diabetes, failure to grow and go through puberty, infertility, low thyroid function and liver disease. Heart disease is the most dangerous and is usually the cause of death in people who do not have access to chelation treatment.

Even patients with NTDT are at risk of iron overload and the complications associated with it. This may be caused by occasional transfusions which may be needed during periods of infection or illness, or an increased iron absorption from food. For this reason, we monitor all patients carefully for signs of iron overload through a wide variety of tests.

Infection
People with excess iron in their body respond badly to infection and may become very sick very quickly. They also often suffer from more unusual types of infection. If their iron is in their heart, they may find it particularly difficult to tolerate infection.
Other problems
These occur in some people when they are older, or often not at all:

• bone thinning
• infertility
• side effects of chelation
• kidney stones
• extramedullary haematopoiesis – this is when bone marrow tries to grow outside of the bone. This can be a particular problem if it is large or if it presses on something important, for example a nerve.

If you develop any signs of infection (such as feeling unwell, fever, chills, diarrhoea, vomiting), weakness of the limbs or palpitations (heart beating fast or unusually), you should contact us urgently for review.

Treatment
The aim of treatment is to keep patients out of hospital and to have as healthy and normal lives as possible. To help with this we review patients on a regular basis in our clinics, some of which run in the evening.

We also have additional support for:

• bone health, reproductive health, cardiology (heart) and psychology in our adult clinics, and
• growth in our paediatric clinics.

Children will often have their blood transfusions at the weekend and adults can have them overnight or at the weekend, if previously arranged. However, most people prefer to come to supportive care (adults) or day care (children).

People with TDT have regular blood transfusions, usually to keep their Hb above 95g/L, although your doctor may decide to raise this threshold.
As the ferritin (a measure of iron stores) increases towards 1000, you will be offered chelation therapy. This may be:

- deferasirox (also known as EXJADE®) which comes as a tablet taken once a day
- desferrioxamine (also known as Desferal®) which is given through an injection under the skin over several hours, a few days a week
- deferiprone (also known as Ferriprox®) which comes as a tablet taken three times a day
- a combination of these.

Your doctor will advise you on the most suitable treatment for you and offer you a leaflet with more information.

Some patients with thalassaemia who need treatment may also respond to a medicine called hydroxyurea. This may apply to those with less severe disease who can increase their HbF with this treatment.

Stem cell transplant is currently the curative treatment option. Transplants are often carried out in childhood if the child has a brother or sister who is the right bone marrow match for them. There have only been very few transplants where children received bone marrow from a parent so accurate information about the risks is not available yet.

Gene therapy is another treatment option currently used in clinical trials. This is when a normal gene is inserted into the patient’s genes to allow them to produce normal HbA. We hope that this will offer a cure to more patients in the future.

Other treatment options depend on the complications. It is important that you discuss your concerns in clinic where a thorough assessment can be performed, and the treatment and complications of the disease are monitored.
What to do in an emergency (adults)

If you have a non-urgent medical problem or a medical problem unrelated to your thalassaemia, for example a rash or a twisted ankle, you can contact your GP or go to your local Emergency Department (A&E). If necessary, or if they are unsure whether your problem is related to your thalassaemia, they can discuss it with us.

If you have a non-urgent problem related to your thalassaemia, you can ask for your outpatient appointment to be brought forward. It may also be appropriate to have a review in day care.

If you are unwell, you should call the haematology advice line (office hours). If it is out of hours, please call adult haematology advice line or paediatric helpline – contact details are at the end of this booklet. The haematology advice line is manned by an administrative staff member who will contact your clinical team. The adult haematology advice line and paediatric helpline are manned by a senior haematology ward nurse who may contact the on-call team for further advice.

You may be advised to attend the rapid access service in haematology day care, go to your GP or Emergency Department at UCLH, or to bring forward your clinic appointment. Sometimes the nurse will speak to one of the doctors to get further advice.

The day care rapid access service is only open for assessments between 9am and 4pm, Monday to Friday, after previous discussion with the CNS or medical team. Outside of these times, you will need to go to the Emergency Department.

Even if you are going to the Emergency Department, it is always best to call ahead so that we can try to streamline your assessment as much as possible. We would also ask the doctor assessing you to discuss your problem with the attending consultant to ensure that your symptoms are considered with an understanding of thalassaemia.
If you go to an Emergency Department in another hospital, it may take up to a few days to transfer you to UCLH. Although we are always happy to support other teams, we would not be able to coordinate your care at another hospital. For this reason, we advise that you come to the Emergency Department at UCLH if you think the delay in getting there is not going to be dangerous.

**Note:** If you have an infectious illness, such as cough, cold, diarrhoea, vomiting or chicken pox, **do not** come to clinic or haematology supportive care unit. You should go to the Emergency Department (A&E) and we will see you there. These seemingly minor illnesses can be very serious for some haematology patients with very poor immune systems, for example those recovering from chemotherapy or stem cell transplants.

**What to do in an emergency (children)**

Please go to the Emergency Department (A&E) with your ‘paediatric passport’ and a paediatric specialist will review you. They should discuss your problems with the haematologist on call.

Please note that it is best to ring the CNS before you come in so that they can liaise with the team seeing you. If you attend an Emergency Department (A&E) in another hospital, it may take up to a few days to transfer you to UCLH. Although we are always happy to support other teams, we would not be able to coordinate your care at another hospital. For this reason, we advise that you come to the Emergency Department (A&E) at UCLH if you think the delay in getting there is not going to be dangerous.

**What is an emergency?**

The following symptoms require immediate medical attention:

- Fever: 38 °C or above, chills
- Diarrhoea or vomiting
- Feeling very unwell
- Palpitations
- New weakness.
**How to prevent thalassaemia**

As thalassaemia is an inherited condition, each carrier (trait) parent may pass on an affected gene to their child. If both parents are carriers of the beta thalassaemia gene, the chance that their child will have thalassaemia is one in four. If one person has the disease and the other is a carrier, the risk is greater – one in two.

A thalassaemia gene combined with a sickle gene may result in sickle cell disease. The genetics of thalassaemia are complicated and you may wish to speak with us or with our dedicated haemoglobinopathy geneticist.

If you or your partner becomes pregnant, it is extremely important that testing is done as early as possible. Please contact us immediately if this happens and we will organise further counselling and assessment. We also advise patients to tell us as soon as they have met their partner so that we can offer testing at an early stage.

**What can I do to stay well?**

- If you have blood transfusions, schedule them in a way that the levels of your Hb are kept higher than 95 g/dL.
- Take your chelation therapy exactly as you have been advised – this may be every day.
- 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 you don’t have a spleen.
- Ensure that you attend your clinic appointments so that we can review your health and monitor you for any complications.
Attending your clinic appointments is a requirement stipulated in the National Standards produced by the Sickle Cell Society together with the Department of Health and Social Care. These appointments are every six to 12 months in healthy people but may be more frequent if you have health problems.

If you don’t have an appointment, please ask your GP to refer you.

If your appointment is inconvenient, please change it. We understand that many of you have busy lives with many commitments and we will do our best to schedule an appointment that is convenient for you.

**National Haemoglobinopathy Registry (NHR)**

The NHR is a database of patients with red cell disorders (mainly sickle cell and thalassaemia) living in the UK. This database collects data that is required by the NHS England from haemoglobinopathy centres. The central aim of the registry is to improve patient care.

Your red cell team will speak to you about this in detail and offer you an information leaflet.

**Sexual health**

Mortimer Market Centre offers STI (STD) testing, HIV tests and a wide range of sexual health services.

You need to book an appointment to attend the clinic, with the exception of walk-in HIV testing clinics. You can phone the clinic for advice if you have an urgent problem or to find out the opening times.

**Mortimer Market Centre**

Address: Mortimer Market Centre  
(off Tottenham Court Road)  
London WC1E 6JB

Appointments telephone: 020 3317 5100

Website: www.camdenproviderservices.nhs.uk/clinic/mortimer-market-centre
Fertility
The reproductive medicine unit offers a wide range of tests and treatments for couples who experience fertility problems, recurrent miscarriage and reproductive endocrinology. These issues can be discussed in the evening clinic where we have support from a dedicated consultant. They will initiate tests and refer on as necessary.

Family planning
You may wish to discuss contraception with your GP, local family planning clinic or community sickle cell and thalassaemia nurse. You should tell them you have thalassaemia and any other associated medical problems, if applicable.

If there are any changes to your medication, please let the red cell team know.

Stop smoking clinic
UCLH has joined forces with Smokefree Camden to establish two clinics a week. Accredited advisers from the National Centre for Smoking Cessation and Training (NCSCT) offer behavioural support and a range of nicotine replacement therapy patches. If you are interested, please ask your consultant to refer you.
Contact details

Haematology advice line (office hours, adults and children):
020 3447 7359

Adult haematology advice line (out of hours):
07852 220 900

Paediatric helpline (out of hours):
• nurse in charge 07961 081 645
• ward T11 south 020 3456 7890 ext. 71103 or 71143

Apheresis:
020 3447 1803

Address: The Joint Red Cell Unit
Department of Haematology
3rd Floor West, 250 Euston Rd
London NW1 2PG

Website: www.uclh.nhs.uk/JRCU

Haematology consultants:
Professor John Porter
Dr Sara Trompeter
Dr Perla Eleftheriou
Dr Emma Drasar
Dr Bernard Davis
Dr Farrukh Shah

Specialist nurses:
Bernadette Hylton (adults)
Nancy Huntley (apheresis)
Nina Gorman (children)
Where can I get more information?

Sickle Cell Society
Tel: 020 8861 7795
Website: www.sicklecellsociety.org

NHS Sickle Cell and Thalassaemia Screening Programme
Website: www.gov.uk/guidance/sickle-cell-and-thalassaemia-screening-programme-overview

UK Thalassaemia Society
Tel: 020 8882 0011
Fax: 020 8882 8618
Email: office@ukts.org
Website: www.ukts.org

University College London Hospitals NHS Foundation Trust cannot accept responsibility for information provided by external organisations.
Space for notes and questions