The Leeds Teaching Hospitals NHS Trust

Health advice for adults with Thalassaemia

Information for patients

This leaflet provides emergency contact details and health information for adult patients with thalassaemia who are registered at Leeds Teaching Hospitals.

What is thalassaemia?

Thalassaemia is 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 carry oxygen around the body. This results in a reduction of the amount of normal haemoglobin produced.

Normal haemoglobin prior to being born is Haemoglobin F or HbF (F is for foetal) and shortly before birth the production switches to Haemoglobin A or HbA (A is for adult). By 18 months of age very little HbF is produced and most haemoglobin is HbA in people with normal haemoglobin production.

In most cases of thalassamia, babies are born without any problems as they make the HbF normally. As they get older, the HbF stops being produced and then 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 person will become anaemic and the body will respond by trying even harder to produce the red blood cells. This can make some of the organs too big e.g. the liver and spleen as well as produce a characteristic appearance to the face.

Nowadays, we assess each person separately and start treatment before having any of the side effects of the disease. The timing of this can vary from infancy (thalassaemia major) to later in life (thalassaemia intermedia treated as major). Some people may not need transfusions until much later in life, or perhaps never, these people have thalassaemia intermedia.

What kind of problems can happen in thalassaemia?

The problems in thalassaemia may result from the thalassaemia itself, the effect of excess iron from blood transfusions and the effect of medications taken to reduce the amount of excess iron in the body. There is much evidence to show that adhering to treatment plans results in improved health and survival for patients with thalassaemia.

Problems related to anaemia

Anaemia makes people feel tired and as a result the body tries to make more blood. This is usually fruitless in patients with thalassaemia major and so the growth and development of the child will slow down, they often will feed poorly and the 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 that are typical of thalassaemia. These features are usually a sign that the child will need to start on a transfusion programme.

Good management of patients will involve blood transfusions on a regular basis usually every three to four weeks to keep the haemoglobin high enough so these symptoms are reduced to an absolute minimum and are often not present at all. If the haemoglobin is kept at a good level the child will grow well and look like any other healthy child.

Good transfusion is equally important in adults as all these complications develop when transfusion is insufficient even in adult life. In patients with thalassaemia intermedia even with a low haemoglobin they continue to grow and develop at a similar rate to other non-anaemic children. We watch every patient carefully to ensure that they do not suffer any lasting harm from the anaemia.

Problems related to transfusion

Blood transfusion is lifesaving in thalassaemia major patients but 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 and as a result of this we must use drugs to help get rid of the extra iron. The drugs are called chelators and the therapy is called chelation. If the iron was not taken out as was the case in the 1950s when these drugs were not available, patients would start to develop serious complications from iron overload and most died as a result.

The harmful iron would deposit around the body often in vital organs such as the heart, pancreas and hormone producing glands resulting 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 the death in people who do not have access to chelation treatment e.g. in developing countries. These problems are avoidable with good chelation therapy. In thalassaemia intermedia patients occasional transfusions may be needed to cover periods of infection or illness. Iron still accumulates and these complications may develop.

Even in non-transfused patients with thalassaemia intermedia we can still see iron overload from increased iron absorption through food. We monitor all patients for signs of iron overload using a wide variety of tests.

Infection

People with excess iron in their body respond badly to infection and may become very sick very quickly often with more unusual types of infection. If their iron is in their heart, they may find it particularly difficult to fight infection.

Other problems

These occur in some people when they are older and 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 this is large or if it presses on something important e.g. a nerve.

If you develop any symptoms suggestive of infection (e.g. feeling unwell, fever, chills, diarrhoea, vomiting) or weakness of the limbs or palpitations (hear beating fast or unusually) you should contact us urgently for review.

Treatment

The aim of our management is to keep people 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 clinic. Patients will often also see an endocrine specialist several times a year to monitor the function of their bones and hormone producing organs.

Most patients receive blood transfusion during the day but there is also some capacity to transfuse blood out of hours, for example at the weekend.

People with thalassamia major have regular blood transfusions, usually to keep their Hb above 95 g/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 a choice of treatment known as chelation therapy. Usually this is deferasirox (also known as EXJADE®) which is a once daily tablet, although many people use desferrioxamine (also known as Desferal®) which is administered through an injection under the skin over several hours several days a week. Some people may take deferiprone which is a tablet taken three times a day. Your doctor will advise what may be most suitable for you. Sometimes, if iron levels are high, a combination of two iron chelators may be considered. In these circumstances, another option that can be considered is a continuous intravenous infusion of desferal.

Some patients with thalassaemia who need treatment may respond to a medicine called hydroxycarbamide rather than transfusions. This is usually in people with less severe disease who can continue to produce HbF.

Other treatment depends on the complications. It is important to discuss your concerns in clinic where a thorough assessment can be performed and complications of the disease and the treatment are monitored.

How to prevent thalassaemia and 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 per cent) in every pregnancy.

If one parent has the disease the risk is greater - 1 in 2 (50 per cent). 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. The genetics of thalassaemia are complicated and 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 at risk of having an affected child the option of prenatal diagnosis may be available. If you choose to have this please tell us immediately if you or your partner becomes pregnant.

Fertility

The reproductive medicine unit offers a comprehensive range of investigations and treatments for couples presenting with fertility problems, recurrent miscarriage and reproductive endocrinology. Pre-implantation genetic diagnosis (PGD) may be offered when both genetic parents have, or are carriers of, a known genetic abnormality.

If a pregnancy is being considered, it is best to discuss your plans in clinic at least 3 months before trying to conceive so that we can optimise your health in preparation for this and ensure that the appropriate referrals are made.

There is a risk that iron chelation treatment or other medicines may be harmful to the developing baby so it is important that both men and women discuss plans to conceive at least 3 months prior as treatment may need to be stopped or changed during this time.

You should see your thalassaemia team regularly for review, whether you are planning a pregnancy or not. The checks may include:

- A detailed scan of your heart (echocardiogram).
- Blood pressure, urine and blood tests.
- MRI scans for measurement of iron.
- Bone density scans.
- Diabetes tests (oral glucose tolerance test).

Obstetric care

It is important that you receive the best care during your pregnancy as early as possible. You and your baby will be followed-up and regularly monitored in the joint obstetric/ haematology clinic. Women with a history of cardiac complications and iron overload are at particular risk. You may need to see other specialists before conceiving to prepare you for pregnancy.

Having a Healthy Lifestyle can help you to stay well with thalassaemia

To help reduce your chances of developing some of the problems associated with thalassaemia it is a good idea to:

- Have a healthy balanced diet you do not usually need a special diet although sometimes you may be advised to take supplements such as folic acid calcium or vitamin D.
- Exercise regularly regular exercise particularly weight bearing anaerobic exercise can help strengthen bones and reduce the risk of osteoporosis.
- Avoid smoking and drinking excessive amounts of alcohol this can help keep your bones and heart healthy.
- Take penicillin twice a day for life if you do not have a spleen.
- Try to avoid infection, wash your hands with soap and water regularly, avoid close contact with sick people when possible and ensure all your vaccinations are up to date.
- Because a number of the complications of thalassaemia are caused by iron build-up in the body, it would be advisable to limit the intake of iron supplemented or iron rich foods.

- If fasting is part of an individuals religious practice they should discuss this with their doctor or specialist nurse and seek advice on how to practice their religion without putting their health at risk (for example managing iron chelation).
- It is advisable to avoid self medication even herbal remedies and the use of these and other over the counter medications, tonics and remedies should be discussed with your doctor before taking them in case they disagree with any prescribed medications.

Hints and tips for keeping well whilst travelling

- Plan in advance and research where you are travelling to and ensure you have had the vaccines required for that area or country (if necessary) and also know how to access healthcare and where it is located.
- Ensure you have been prescribed anti-malarials by your doctor if your destination is a risk for malaria and that you have started the course as advised by your doctor.
- Make sure that you take a supply of your regular medicines for the duration of the trip.
- Keep all of your medicines and health information in your hand luggage.
- Get a summary of your healthcare information such as a recent clinic letter so that you can pass it on to a hospital in case you need treatment abroad - take a photo and keep it on your phone.
- Travel when you are free from infection.

- 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 which can be purchased from your local pharmacy. If you develop a fever or notice blood in the stools you should seek immediate medical advice.
- Get a good night's sleep on the night before your departure.
- Drink plenty of water and non- alcoholic drinks prior to and during your flight and continue to do this for a few days after your arrival.
- Avoid smoking.
- 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 result in dehydration. Wear flight socks to reduce the risk of thrombosis - these can be bought in most large pharmacy stores.
- Watch out for any unusual symptoms of pain or breathlessness and inform the cabin crew immediately if you feel unwell.
- Ensure you have bought travel insurance to cover you during your trip.

Urgent Contact Details

What should I do if I need an urgent appointment?

If you would like to be seen in clinic earlier than usual, please telephone 0113 206 7966 to rearrange your appointment or it may be appropriate to be seen on the supportive care unit (J87).

If you are not sure whether to go to Accident & Emergency (A&E) please telephone SJUH switchboard on 0113 243 3144 and ask for the oncology bleep holder. The nurse practitioners on our haematology/oncology assessment area are available 24 hours a day and can arrange admission to ward J95 and give advice.

In an emergency, dial 999 or go straight to A&E.

You can contact the nurse specialist for advice on telephone number 0113 206 8236 but if your health concern is urgent and related to your red cell condition please call the 24 hour assessment area contact as above.

Note: if you have an infectious illness, e.g. cough, cold, diarrhoea, vomiting, chicken pox etc. do not come directly to clinic or haematology day care without discussion. These seemingly minor illnesses can be catastrophic in some haematology patients.

What is an emergency?

The things that require immediate attendance are:

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

Contact us

The contact details below are non-emergency and available during office hours only, excluding Bank Holidays.

- **Outpatient clinic appointments: 0113 206 7966**
  (for appointment questions)
- **Haematology Supportive Care Unit: 0113 206 9187**
  (for appointment questions or if you are running out of treatment before your next red cell clinic appointment)
- **Clinical nurse specialist: 0113 206 8236**
  (for health related questions)
- **Community Nurse: 0113 206 8236**
  (for health related questions)
- **Haematology secretaries: 0113 206 8497**
  (for general questions)

Other Useful Information

UK Thalassaemia Society

https://ukts.org/

Thalassaemia International Federation

https://thalassaemia.org.cy/

The National Haemoglobinopathy Registry (NHR)

https://www.mdsas.com/

The National Haemoglobinopathy Registry (NHR) is a confidential database of patients with red cell disorders living in the UK. All patients with sickle cell disease, thalassaemia and rare inherited anaemias such as Diamond Blackfan anaemia, sideroblastic anaemia, red cell enzyme disorders are included. Patients with these disorders are managed by specialised haemoglobinopathy services and teams.

The aim of the registry is to improve patient care by ensuring clinically important information related to your health, treatment and complications is available to specialist haemoglobinopathy teams who are involved in your clinical care or may become involved in your care.

UK National Haemoglobinopathy Panel

https://www.nationalhaempanel-nhs.net

The panel hold a monthly Multi-Disciplinary Team Meeting (MDT) that brings together clinical specialists that will consist of professionals from a core panel representing all the Haemoglobinopathy coordinating centres (HCC) across the country representing all professional groups involved in the care of patients with disorders described with the specialist haemoglobinopathies services – sickle cell disease, thalassaemia and rare anaemias.

NHS - Screening for sickle cell and thalassaemia

https://www.nhs.uk/pregnancy/your-pregnancy-care/screening-for-sickle-cell-and-thalassaemia/

The NHS website

https://www.nhs.uk/

- Information about how to find a GP, pharmacy, dentist, urgent or emergency.
- Where to get urgent help for mental health.
- Your complete guide to conditions, symptoms and treatments, including what to do and when to get help.
- Find out how your medicine works, how and when to take it, possible side effects and answers to your common questions.
- NHS advice about healthy living, including eating a balanced diet, healthy weight, exercise, quitting smoking and drinking less alcohol.
- Social care and support guide.

NHS 111

https://111.nhs.uk/

If you think you need medical help right now you can telephone 111 or use the online service link above.

NHS - Better Health

https://www.nhs.uk/better-health/get-active/

NHS - Benefits if you're under State Pension age

https://www.nhs.uk/conditions/social-care-and-support-guide/money-work-and-benefits/benefits-if-you-are-under-state-pension-age/

NHS - Benefits if you're over State Pension age

https://www.nhs.uk/conditions/social-care-and-support-guide/money-work-and-benefits/benefits-if-you-are-over-state-pension-age/

Adult social care

https://www.leeds.gov.uk/adult-social-care

UK Visas and Immigration

https://www.gov.uk/government/organisations/uk-visas-and-immigration

Citizens Advice

https://www.citizensadvice.org.uk/

Leeds Sexual Health

https://www.leedssexualhealth.com/

Or telephone: 0113 843 4495

Leeds Sexual Health - a service that offers Sexually Transmitted Infection (STI) screening and treatment, HIV testing and contraception services.

Questions / Notes

What did you think of your care?

Scan the QR code or visit **bit.ly/nhsleedsfft**

Your views matter

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