Children's haematology

Children and thalassaemia

Information for patients, relatives and carers

Introduction

This leaflet has been provided to help answer some of the questions you may have about Thalassaemia in children. It is not meant to replace the consultation between you and the medical team, but aims to help you understand more about what you discussed together.

What is thalassaemia?

Thalassemia is a lifelong genetic condition which affects the body's red blood cells. Haemoglobin is the part of red blood cells that carries oxygen around your body. Thalassemia causes abnormalities in the genes that control how haemoglobin is made. Genes are small parts of DNA. Thalassemia makes you have less haemoglobin than other people.

Most babies with thalassemia are born without any problems. This is because they produce haemoglobin F (fetal) normally before being born. Shortly before they're born, babies without thalassemia will start producing haemoglobin A (adult). But when babies with thalassemia grow older and stop producing haemoglobin F, they can get anaemia, which means they can't produce enough haemoglobin A. The progression of this condition varies among individuals and often depends on the type of thalassemia.

Today, we assess each individual with thalassemia and start treatment with blood transfusions — where you get blood from a donor — before significant side effects of the disease happen. Side effects happen at different times for different people, and of different severities. This means treatment starts at different ages for different people.

When you need blood transfusions from very young, we call it transfusion-dependent thalassemia (TDT). When you need transfusions later in life, or possibly never, we call it non-transfusion-dependent thalassemia (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 absorbed from your 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 causes fatigue because there isn't enough oxygen being delivered to the body. In response, the bone marrow (the spongy tissue inside some bones) tries to make more red blood cells. When babies become very anaemic, they can feed poorly, and this can slow growth and development.

Affected children may have poor appetite, and their abdomen may swell due to an enlarged liver and spleen. Additionally, the bone marrow can expand, leading to bone thinning and swelling in the cheekbones and forehead, resulting in a characteristic facial appearance.

In children these features are usually a sign they need to start a transfusion programme. This involves blood transfusions on a regular basis, usually every three to four weeks, to maintain sufficient haemoglobin levels. With regular transfusion these symptoms can be kept to a minimum or go away completely. When haemoglobin levels are at a healthy level, children grow well and appear healthy. Regular transfusions are equally important for adults, as many of the same complications can occur when haemoglobin is too low. It is crucial to monitor both adults and children closely to ensure they do not experience any lasting harm from anaemia.

People with NTDT may also 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 transfusions are lifesaving for patients with thalassemia. Red blood cells contain iron, an essential component of haemoglobin. However, there is no natural way for the body to eliminate excess iron, leading to accumulation with each transfusion. Too much iron (known as iron overload) can be harmful, so medications called chelators are prescribed to remove the excess iron and prevent it from building up. This treatment is referred to as chelation therapy.

Before chelators became available in the 1970s, patients often developed serious complications from iron overload. Fortunately, these problems can be avoided with effective chelation therapy.

Some thalassemia patients may require transfusions during infections or illnesses, and during pregnancy, iron can accumulate over time, causing similar complications. Additionally, some patients may experience iron overload due to excessive absorption of iron from food. For these reasons, we closely monitor all patients for signs of iron overload using various tests.

People with excess iron in their bodies can become very ill quickly if they develop an infection. They often experience more unusual types of infections as well. If excess iron accumulates in the heart, it can make it especially difficult for them to tolerate infections.

Other problems

The following problems may occur in some people when they are older:

- Bone thinning

- Enlargement of the spleen
- Infertility
- Side effects of chelation
- Kidney stones
- Gallstones
- 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 as soon as possible.

Treatment

The aim of the care we provide for thalassaemia is to keep your child out of hospital and ensure they lead as healthy and normal life as possible. Regular review in the paediatric haemoglobinopathy clinic is essential, so that we can identify and treat any problems at an early stage. They 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 TDT have regular blood transfusions, usually to keep their haemoglobin above 95g/L. Your child's doctor may decide to raise this threshold.

Your child will have their blood transfusions on the paediatric haematology day unit (PHDU) which is located on the sixth floor of the Queen Elizabeth Queen Mother Building and open from 09.00 to 17.00 Monday to Friday. Your child will need to have a special blood test two days before your transfusion. Both appointments will need to be booked in advance.

As the serum ferritin (a measure of iron stores) increases towards 1000, we will offer you chelation therapy. This may be:

- Deferasirox 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 people with thalassaemia who need treatment may also respond to a medicine called hydroxyurea to increase their HbF. This may be suitable for those with a less severe disease.

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

Bone Marrow transplant is currently the curative treatment option. Transplants are often carried out in childhood if the child's sibling is their bone marrow match.

There are also two transplant-type treatment options currently used in clinical trials: gene therapy and gene editing. Gene therapy is when a normal gene is inserted into a person's genes to allow them to produce normal HbA. Gene editing is when a gene is changed to encourage haemoglobin production.

What to do in an emergency

The following symptoms require immediate medical attention:

- Fever: 38°C or above
- Chills
- Coffee coloured urine or blood in urine
- Diarrhoea or vomiting
- Feeling very unwell
- Palpitations
- New weakness/lethargy

When you get to A&E, tell the receptionist your child has TDT. Please go to the Emergency Department (A&E). It's best to ring the CNS before you come in so that they can talk to the team seeing you.

If you go to an A&E in another hospital and are admitted, we might be able to transfer your child to St. Mary's Hospital, depending on how unwell they are. Though we will always support other teams, we would not be able to manage your child's care at another hospital.

Note: If your child has an infectious illness, such as cough, cold, diarrhoea, vomiting or chicken pox, please inform the A&E team as soon as you arrive.

If you think your child's condition may be life threatening **call 999**.

For a non-urgent medical problem or a medical problem unrelated TDT, for example a rash or a twisted ankle, you should contact your GP or take your child to your local Emergency Department (A&E). If necessary, they can contact us.

How to prevent thalassaemia

Thalassaemia is an inherited condition so it can't be prevented, because it is passed on from parent to child. A parent can have the gene for thalassemia but not have the disease. This parent would be called a carrier. Any parent that is a carrier can pass on the thalassemia gene to their child. If both parents are carriers, the chance their child will have thalassemia is one in four. If one parent has the disease and the other is a carrier, the chance is one in two.

A thalassaemia gene combined with a sickle gene may result in sickle cell disease. The genetics of thalassaemia are complicated so please speak to us if you'd like more information.

If you or your partner become pregnant, it's very important that testing is done as early as possible. Please contact us immediately if this happens and we will organise counselling and assessment for you. We also ask our 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 keep my child 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 advised – this may be every day.
- Make sure infections are treated quickly.
- Maintain a good balance of nutrition and activity.
- Make sure your child's vaccinations are up to date.
- Your child should take penicillin twice a day for life if they don't have a spleen.
- Attend your clinic appointments so that we can review your child's health and monitor them for any complications.
- These appointments are every six to 12 months in healthy people but may be more frequent if they have health problems.
- If the appointment is inconvenient, please **change it as soon as you can**. We will do our best to schedule an appointment that is convenient.

Travel advice

Check if any travel vaccines are needed for the country you are travelling to. 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 talk to your hospital specialist, as we might recommend that you don't have it.

For more information, please see our leaflet: Vaccinations, medication and travel for children with thalassaemia and sickle cell disease. If you don't have this, let us know.

Transition

As your child gets older, we will support them to take more responsibility for their health. This is to make sure that they can look after themselves when they leave home to live independently.

It's as important for them to learn the skills needed for a day-to-day adult life, such as paying bills or working, as it is to be able to navigate their own health needs. This process is called transitioning; we will help and guide them throughout that time. Transitioning happens gradually over their teenage years so that by the time your child is 18, they will feel confident in:

- Knowledge about their condition
- Attending clinic appointments on their own if they want to
- Ordering their medicines
- Organising their vaccinations.

As your child gets older, they may want to talk to the team looking after them in private. They may have questions they would like to ask but may feel uncomfortable to discuss them with you. This is normal and we will be happy to see them on their own.

Contact details

- **Haemoglobinopathies admin team:** 0203 312 6157

  Haemoglobinopathies clinical nurse specialist team (09.00 to 17.00): 07795651153
  imperial.paediatrichaemoglobinopathies@nhs.net
- **Paediatric haematology day unit (PHDU):** 0203 312 5080/5081
- **Paediatric Site Practitioner:** 07824625419
- **Address:** Imperial College Healthcare NHS Trust, St Mary's Hospital, Praed Street, London W2 1NY

The team

- Dr Kirstin Lund, consultant paediatric haematologist,
- Professor Josu de la Fuente, consultant paediatric haematologist
- Dr Adam Gassas, consultant paediatric haematologist
- Dr Toni Petterson, consultant paediatric haematologist
- Dr Leena Karnik, consultant paediatric haematologist
- Miss Catherine Mkandawire, matron (Haemoglobinopathies and apheresis children and young people)
- Miss Kajal Rai, clinical nurse specialist (Haemoglobinopathies children and young people)
- Ms Nancy Huntley, clinical nurse specialist (Apheresis children and young people)
- Miss Ristell Fernandes, clinical nurse specialist (Apheresis children and young people)
- Mr Renzo Tiong, clinical nurse specialist (Apheresis children and young people)

Further information

https://www.imperial.nhs.uk/our-services/adolescent-and-young-adult-healthcare---11to25
https://www.readysteadygo.net/

Sickle Cell Society
020 8861 7795
sicklecellsociety.org

UK Thalassaemia Society
020 8882 0011
office@ukts.org
www.ukts.org

Contact
The charity for families with disabled children
https://contact.org.uk/

Family Fund
https://www.familyfund.org.uk/
Grants are available for families raising a child or young person with a long-term disability, disabling condition or life-limiting illness, and living on a low income.

The Hope Project Scotland
07756 266691
via website
thehopeprojectscotland.org.uk

Citizens Advice Bureau.
https://www.citizensadvice.org.uk/

The Care Information Exchange (CIE)

CIE gives secure online access to information from the hospital such as test results, appointments and letters. Young people and their parents can also add things like symptoms the young person is experiencing to give a more complete record. To register and access the record QR code below to download the application.

Everyone registering will need to complete a form, show proof of identification, and have an email address. The email address will be the login name, and the address for notifications when something new is added to the record. Go to www.careinformationexchange-nwl.nhs.uk/ or scan this QR code with your phone camera to find out more and register:

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 3312 7777** (10.00 – 16.00, Monday to Friday). 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

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