Evelina London

NHS
Guy's and St Thomas'
NHS Foundation Trust

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).

Problems that can happen with 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 have transfusions can develop iron overload. This is because the body can absorb 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 (when bone marrow grows outside of the bone, which can press on something important like a nerve)

Having treatment at Evelina London

We try to keep your child out of hospital and make sure life you can lead your life as healthily and normally as possible. Regular haemoglobinopathy reviews are an essential part of this, as they help us 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 one time each 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 the emergency department 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 them 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 2 times 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. 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 you have busy lives, and will accommodate this wherever possible.

Support and more information

Sickle Cell Society
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
Information, and thalassaemia counsellors. They 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@nhs.net

Lead nurse, **phone:** 020 7188 9432 or 07918 338730 **email:** gstt.haemoglobinopathycns@nhs.net

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

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

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