All about Thalassaemia

Information and advice for patients/parents and carers

Sickle Cell & Thalassaemia Centre

What is thalassaemia?

Thalassaemia is a genetic blood disorder caused by the haemoglobin, a substance that is found in red blood cells and carries oxygen around your body, not being made properly. The gene for thalassaemia is passed on from parents to their children through genes, just like eye colour and hair colour are. If both parents are carriers, they are equally likely to pass it on. Moreover, as it is a genetic condition, you can't catch it and carriers will have not become ill as a result of it. There are two forms of thalassaemia:

- Alpha thalassaemia
- Beta thalassaemia

Haemoglobin is made up of 2 different kinds of protein chains named alpha and beta chains. Both types of thalassaemia are caused by abnormalities in these protein chains. As a result of this, thalassemia can lead to the body making red blood cells that are not very effective, in addition to haemolysis (destruction of red blood cells).

Thalassemia major is an inherited disorder that results from decreased and defective production of haemoglobin.

Alpha thalassaemia

Alpha thalassaemia is less common in the UK compared to other countries. Carrying alpha thalassaemia does not cause any illness. Most people who carry alpha thalassaemia do not know that they have it. They only discover it when they have a special blood test. However, if your partner also carries alpha thalassaemia, there is a chance that this will affect your children. There are 2 types of alpha thalassaemia:

- Alpha plus thalassaemia which is harmless.
- Alpha zero thalassaemia which may have more serious complications.

Alpha plus thalassemia

Alpha plus thalassemia can come in two forms, you may be a carrier of the gene (where you have no symptoms) or you may have the genetic condition. If you have alpha plus thalassemia, you may find that you have some form of anaemia, which can be treated. Despite this, it has been found that both carriers and sufferers of alpha plus thalassemia live normal lives.

Alpha zero Thalassaemia

Alpha zero thalassemia can come have different effects depending on what genes you have inherited. You may be a carrier of the gene (where you have no symptoms) or you may have anaemia (which can be treated) or, in some rare cases, it can lead to the birth of a stillborn baby.

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Beta thalassaemia

Beta thalassemia is the more common form of thalassaemia seen in the UK. There are 2 types of beta thalassaemia:

- Beta thalassaemia intermedia (BTI) which affects different people in different ways.
- Beta thalassaemia major (BTM), also known as Cooley's anaemia, which has more serious side effects.

Beta thalassaemia intermedia (BTI)

BTI is a mild form of beta thalassaemia. BTI affects different patients in different ways: for some it only causes mild anaemia whereas others may need regular blood transfusions (extra blood). In some cases, transfusions may be needed at certain times such as if they are unwell or have infections during pregnancy or when they have operations. People with BTI will need regular haemoglobin checks. To help the body manage iron levels appropriately, you may require regular blood transfusions which we will monitor for you.

Beta thalassaemia major (BTM)

BTM is a severe form of beta thalassaemia. People with BTM will need blood transfusions to provide them with haemoglobin. They will need these regularly throughout their lives. Without transfusions every few weeks, children diagnosed with beta thalassaemia major would usually die by age 7 due to the effects of severe anaemia on the body. They will also need medicines all their lives to help their bodies to control the amount of iron in the blood.

How is thalassaemia inherited?

If 1 parent has the gene and passes it onto their child, then the child is said to have thalassemia trait. Thalassemia trait will not develop into the full-blown disease, has no or few symptoms and no treatment is necessary for someone who has it.

However, genetic counselling is important for families that carry the thalassemia gene. This is because someone with the trait has a 25% (1 in 4) chance of having a child with the disease if his or her partner also has the thalassaemia trait.

Thalassemia affects many people coming from areas around Mediterranean (Italy, Greece, and Turkey), parts of Southeast Asia (India, Pakistan and Bangladesh) Africa, Malaysia, and China.

What are the symptoms?

Most of the time there are no symptoms, however over time some patients may experience some of the symptoms below:

- Fatigue or shortness of breath (especially leading up to a transfusion) are very common.

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- Gallstones are common.
- Mild jaundice or the liver and the spleen may be enlarged in some patients.
- Bone abnormalities and bone pain. Patients can experience enlargement of their cheek bones, foreheads, and other bones.
- Poor growth may occur (as a result of low haemoglobin and reduced ability of the blood to carry oxygen to the body).

Complications
Thalassaemia can lead to different complications. It can affect the bones; bony problems can arise due to a lack of sex hormones and the bony changes can cause brittle bones which can be painful and may even cause fractures.

However, most complications of thalassaemia arise from the effects of iron overload. Blood contains iron which can build up in different parts of the body and this can cause damage to these parts:

- **The heart:** This can be mild, moderate or severe and is picked up using a special type of scan called an MRI scan. Large amounts of iron in the heart can lead to heart failure and irregular heart rhythms. Iron in the heart is dangerous but it can be removed by using strong iron-removing medications.
- **The liver:** Iron overload in the liver can result in scarring of the liver which is also known as cirrhosis
- **Pancreas:** A large amount of iron in the pancreas can lead to diabetes. Diabetes is irreversible and is treated with insulin
- **Hormone glands:**
  - Underactive thyroid which can cause tiredness.
  - Sex hormone glands: In women periods may be delayed or irregular or stop. Later on this can affect fertility. In men, less testosterone is made, (testosterone is needed for muscle bulk, secondary sexual characteristics such as facial hair etc.).

How is it diagnosed?

A complete blood count, haemoglobin and a specific blood test called a haemoglobin electrophoresis are used to screen for all types of thalassaemia and can be done in infancy.

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Treatment of thalassaemia

1. **Blood transfusions**
   This can help to bring haemoglobin levels back to normal. Haemoglobin levels before a transfusion should be 90-100g/L.

2. **Management of iron overload**
   It is much better to try to control iron at all times as this can help prevent build-up of dangerous levels of iron. Iron chelation refers to medication to remove excess iron. There is a choice of medication, and what is chosen depends on each person's individual circumstances.

For further information on either of these treatments, please contact the Sickle Cell and Thalassaemia Centre for a leaflet.

What are the risks of not getting treatment?

If you do not have treatment for thalassaemia, depending on the type of thalassaemia, you may experience these symptoms:

- Fatigue or shortness of breath (very common)
- Gallstones
- Mild jaundice
- Liver or spleen enlargen
- Bony abnormalities and bone pain
- Anaemia

Monitoring

Patients with thalassaemia need regular tests to keep a check on their blood count and organs.

- **Each month:** full blood count (FBC), liver and kidney function tests
- **Every 3 months:** ferritin levels which is a rough indication of iron levels
- **Every year:**
  - Glucose tolerance test to check for diabetes
  - Hormone tests (thyroid, testosterone, oestrogen)
  - Check for viruses from the transfusions e.g HIV, hepatitis
  - Hearing tests and eye tests to check whether the iron chelating medications are affecting other parts of the body
  - MRI scans of the heart and liver

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Contact details

If you have any questions or concerns please contact the Sickle Cell and Thalassaemia Centre.

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Monday, Wednesday & Thursday 9am – 5pm
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For further information

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For more information about our hospitals and services please see our websites *www.swbh.nhs.uk* and *www.swbhengage.com*, follow us on Twitter *@SWBHnhs* and like us on Facebook *www.facebook.com/SWBHnhs*.

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Sources used for the information in this leaflet

- Okpala I. and Stephens A.D., Oxford: Blackwell, 'Practical Management of Haemoglobinopathies – The genetics and multiple phenotypes of beta thalassaemia', 2004.
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- Sickle Cell Society, 'Standards for the Clinical Care of Adults with Sickle Cell Disease in the UK', 2008.
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If you would like to suggest any amendments or improvements to this leaflet please contact the communications department on 0121 507 5495 or email: **swb-tr.swbh-gm-patient-information@nhs.net**

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