# Thalassaemia

Haematology

## What is Thalassaemia?

Thalassaemia is the name given to a group of inherited blood disorders that affect the body's ability to create red blood cells.

**Red blood cells**
Red blood cells are very important because they carry a protein called haemoglobin around the body. Haemoglobin transports oxygen from the lungs to the rest of the body.

Haemoglobin is produced in the bone marrow (a red spongy material found inside the larger bones). If your body does not receive enough oxygen, you will feel tired, breathless, drowsy and faint. This condition is known as anaemia. The most serious types of thalassaemia can cause other complications, including organ damage, restricted growth, liver disease, and heart failure.

## Types of thalassaemia

Thalassaemia is caused by changes (mutations) in the genes that make haemoglobin.

Haemoglobin is made up of matching chains of proteins (which are named after Greek letters of the alphabet). To work properly, haemoglobin needs both an alpha chain and a beta chain of proteins. A change that affects the alpha chain causes alpha thalassaemia, and a change that affects the beta chain causes beta thalassaemia.

## Alpha thalassaemia

The alpha chain is produced by four genes and the severity of the condition depends on how many of those genes have been mutated.

- If one gene is mutated, there is little or no effect.
- If two genes are mutated, there may be symptoms of mild anaemia. This condition is known as the alpha thalassaemia trait. If two people with the alpha thalassaemia trait have a child, there is a one-in-four chance that their child will inherit the most severe form of alpha thalassaemia
- If three genes are mutated, the result will be a condition called haemoglobin H disease. People with haemoglobin H disease will have lifelong (chronic) anaemia and may require regular blood transfusions

**The prevention of infection is a major priority in all healthcare and everyone has a part to play.**

- **Please decontaminate your hands frequently for 20 seconds using soap and water or alcohol gel if available**
- **If you have symptoms of diarrhoea and/or vomiting, cough or other respiratory symptoms, a temperature or any loss of taste or smell please do not visit the hospital or any other care facility and seek advice from 111**
- **Keep the environment clean and tidy**
- **Let's work together to keep infections out of our hospitals and care homes.**

- If all four genes are mutated, the result will be the most severe form of alpha thalassaemia, known as alpha thalassaemia major. Infants with this condition are unable to produce normal haemoglobin and are unlikely to survive pregnancy. There have been some cases of unborn babies being treated with blood transfusions while still in the womb, but this type of treatment has a low success rate.

## Beta thalassaemia

Beta thalassaemia is the more common form of thalassaemia seen in the UK.

Beta thalassaemia can range from moderate to severe. The most severe form of the condition is known as beta thalassaemia major (BTM). People with BTM will require blood transfusions for the rest of their life.

The more moderate form of the condition is known as beta thalassaemia intermediate (BTI). The symptoms of BTI will vary from person to person. Some will experience only symptoms of mild anaemia while others will require blood transfusions.

People with BTM will need regular blood transfusions to provide them with haemoglobin. They will need these regularly throughout their lives. They will also need medication all their life to help their bodies control the amount of iron in the blood.

## How common is Beta Thalassaemia Major?

It is estimated that there are 1,000 people in the UK living with BTM and most cases are found in people of Mediterranean, Middle Eastern and, in particular, South Asian ancestry. Eight out of 10 babies born with BTM in the UK have parents of Indian, Pakistani or Bangladeshi ancestry.

## What are the Symptoms of Thalassaemia?

- Fatigue or shortness of breath are very common (this is due to the chronic anaemia and can be worse leading up to a blood transfusion)
- Mild jaundice
- Liver and spleen can be enlarged in some patients
- Gallstones
- Poor growth. This can occur as a result of a low haemoglobin and the reduced ability of the blood to carry oxygen to the body
- Bone abnormalities and bone pain. Some patients can experience enlarged cheek bones, forehead and other bones. Bony problems can be caused by a lack of sex hormones and bony changes can cause brittle bones which can be extremely painful

## Complications of Thalassaemia

Most complications of thalassaemia are due to the effect of iron overload in the body. Blood contains iron which can build up in different parts of the body and this can cause damage to these organs.  Patients with thalassaemia need regular lifelong blood transfusions which causes a build- up of iron in the body. This build- up of iron can lead to problems with;

- The Heart: Iron overloading can be mild, moderate or severe. The amount of iron overloading is picked up by a scan called an MRI scan (a scan of the body using strong magnetic fields and radio waves to produce detailed images of inside the body). Large amounts of iron in the heart can lead to irregular heart rhythms and even heart failure. Iron in the heart is dangerous but it can be removed using iron removing medications
- The Liver: iron overload in the liver can cause scarring of the liver known as cirrhosis
- Hormone glands: In women periods can be delayed, irregular or even stop. Later on this can affect fertility. In men less testosterone is made which is needed for male growth in puberty including muscle growth and facial hair
- Pancreas: Too much iron in the pancreas can lead to diabetes which is irreversible and often needs treating with lifelong insulin.

# Treatment for Thalassaemia

Blood Transfusions: Having a blood transfusion regularly (every 3-4 weeks) for life will help to bring haemoglobin levels back to a normal level.

**Management of iron overload:** If you are having blood transfusions for life it is very important that you try to control iron levels at all times, because, if iron is not controlled, it can be very dangerous. Your hospital team will give you a medication which is known as an iron chelator. Iron chelation means medication to remove the excess iron in the body. There is a choice of medication and how it is given (please ask for more information from your hospital doctor or nurse specialist). The hospital will discuss with you the options for iron chelation, and the medication will be chosen for you as an individual.

# Monitoring

Patients with thalassaemia will need regular tests to monitor their blood levels and organs.

Every month a full blood count, liver and kidney tests will be taken. Every 3 months ferritin levels will be taken which is a blood test that gives the hospital a guide to the iron levels in the body.

Every year other tests will be carried out. These include hormone tests, hearing and eye tests to see if the iron reducing medication you are taking is affecting other parts of your body. MRI scans of the liver and heart and glucose tolerance tests to monitor for diabetes. It is very important that you attend your hospital clinic appointments and take your medication regularly so you can be closely monitored for any complications.

# Useful Contact Numbers

**Consultant in Haemoglobinopathies**
01902 695271

**Haematology Clinical Nurse Specialist**
01902 695276

# Where can I get further information?

**The UK Thalassaemia Society**
19 The Broadway
Southgate Circus
London
N14 6PH
Tel: 0208 882 0011
www.ukts.org

**Sickle cell and Thalassaemia support Project**
2nd floor office ST Johns House
St Johns Square
Wolverhampton
WV2 4BH
Tel 01902 444076
www.scstsp.org.uk
email info@sctsp.org.uk

**Sickle Cell Care (Wolverhampton)**
58 Bank Street
Wellington Road
Bilston
WV14 8PD
Tel 01902 498274
www.sicklecellcare.net

## References used for this leaflet

United Kingdom Thalassaemia Society, 'Standards for the clinical care of children and adults with Thalassaemia in the UK', 3rd Edition, (2016).

## English

**If you need information in another way like easy read or a different language please let us know.**

**If you need an interpreter or assistance please let us know.**

## Lithuanian

**Jeigu norėtumėte, kad informacija jums būtų pateikta kitu būdu, pavyzdžiui, supaprastinta forma ar kita kalba, prašome mums apie tai pranešti.**

**Jeigu jums reikia vertėjo ar kitos pagalbos, prašome mums apie tai pranešti.**

## Polish

**Jeżeli chcieliby Państwo otrzymać te informacje w innej postaci, na przykład w wersji łatwej do czytania lub w innym języku, prosimy powiedzieć nam o tym.**

**Prosimy poinformować nas również, jeżeli potrzebowaliby Państwo usługi tłumaczenia ustnego lub innej pomocy.**

## Punjabi

ਜੇ ਤੁਹਾਨੂੰ ਇਹ ਜਾਣਕਾਰੀ ਕਿਸੇ ਹੋਰ ਰੂਪ ਵਿਚ, ਜਿਵੇਂ ਪੜ੍ਹਨ ਵਿਚ ਆਸਾਨ ਰੂਪ ਜਾਂ ਕਿਸੇ ਦੂਜੀ ਭਾਸ਼ਾ ਵਿਚ, ਚਾਹੀਦੀ ਹੈ ਤਾਂ ਕਿਰਪਾ ਕਰਕੇ ਸਾਨੂੰ ਦੱਸੋ।

ਜੇ ਤੁਹਾਨੂੰ ਦੁਭਾਸ਼ੀਏ ਦੀ ਜਾਂ ਸਹਾਇਤਾ ਦੀ ਲੋੜ ਹੈ ਤਾਂ ਕਿਰਪਾ ਕਰਕੇ ਸਾਨੂੰ ਦੱਸੋ।

## Romanian

**Dacă aveți nevoie de informații în alt format, ca de exemplu caractere ușor de citit sau altă limbă, vă rugăm să ne informați.**

**Dacă aveți nevoie de un interpret sau de asistență, vă rugăm să ne informați.**

## Traditional Chinese

如果您需要以其他方式了解信息，如易读或其他语种，请告诉我们。

如果您需要口译人员或帮助，请告诉我们。

Designed & Produced by the Department of Clinical Illustration,
New Cross Hospital, Wolverhampton, WV10 0QP Tel: 01902 695377.

Review Date 08/07/2024 Date Produced 06/15 MI_962314_23.06.21_V_2