What is Thalassaemia?

Thalassaemia is an inherited blood condition. The name is derived from Greek and means “anaemia by the sea”. People with Thalassaemia come from around the world, mainly Asia, India and some parts of Southern Europe.

People with Thalassaemia are unable to make enough healthy mature red blood cells. This lack of red blood cells is known as anaemia, and in thalassaemia major, this anaemia prevents a child growing or feeling well.

Sadly, without treatment, a child with Thalassaemia will not survive childhood.

What is blood made of?

Blood is made up of millions of red blood cells in liquid called plasma. There are other cells in the plasma. White cells fight infection and platelets help blood to clot.

Blood is red because the red blood cells contain a substance called Haemoglobin. Haemoglobin is very important as it carries oxygen around your body releasing it wherever it is needed.

What is Anaemia?

There are different types of anaemia. The most common in the world is because of a shortage of iron.

Thalassaemia is a different type of anaemia and has nothing to do with how much iron is in your **diet.**

In thalassaemia there is a mix up in the genetic information that produces haemoglobin. This is the part of the red cell that carries the oxygen. This means that the red cells are more easily broken down and results in lower number of red cells (anaemia).

How is Thalassaemia Major diagnosed?

If your baby was born in the UK, when your baby was a few days old he/she may have had a blood test taken from his/her heel. From this test several different conditions can usually be identified. One of these is Thalassaemia.

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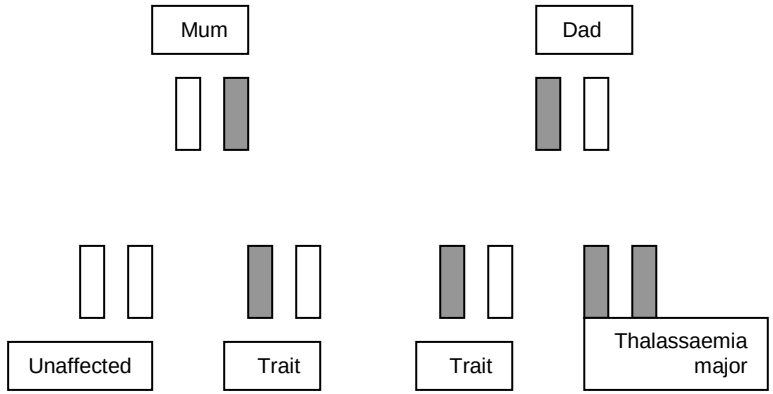
After the first result from your baby’s heel stick test, you will have been invited to speak with a Haematologist. This is a doctor who is a specialist in red blood cell disorders such as Thalassaemia.

At this appointment, the doctor will have discussed your child’s diagnosis and another small sample of blood will have been taken to confirm this.

Sometimes, in babies who have not been tested at birth, thalassemia is diagnosed later in the first year when the child has blood tests done because they are unwell or not growing properly.

How do people get Thalassaemia?

Thalassaemia is an inherited condition passed down from both **mother and father**. It is shown in the diagram below.



Assuming a situation where both parents are carriers of Thalassaemia then, each time you have a baby

- you have a 1 in 4 chance of having a baby with Thalassaemia major.
- you have a 2 in 4 chance of having a baby that carries the Thalassaemia gene (Thalassaemia trait). This normally causes NO health problems for the baby
- You have a 1 in 4 chance of having a baby that does not carry the Thalassaemia gene and does not have Thalassaemia.

It is important to understand that can happen at each pregnancy and does not mean if you have 4 children, only 1 would have the condition.

The genetics of Thalassaemia can be confusing. You may find it helpful to speak with a genetic counsellor who can help you to understand it. They can help you understand the genetics of Thalassaemia, how it may affect your decision in planning future pregnancies. This can be arranged at your first appointment or at a later stage if you wish.

What is the treatment for Thalassaemia?

Thalassaemia is treated with regular blood transfusions, usually every 4 weeks. These blood transfusions allow the body to function normally.

Your child will need these transfusions throughout their life.

When will my child start blood transfusions?

When a baby is born it has fetal or baby haemoglobin called Haemoglobin F (HbF). As the baby grows the amount of baby HbF decreases usually over the first few months of life.

Normally at this time adult blood would be produced but, a child with Thalassaemia is unable to make normal adult blood and that is why they need blood transfusions.

Your child will have a blood test every few weeks to see if their haemoglobin level is dropping. Your child's weight and general well being will also be carefully monitored. All of these factors will be considered before the doctor recommends that your child needs to start blood transfusions.

Are blood transfusions safe?

Almost always yes, but no medical treatment is completely risk free. You will be given more information about this before your child starts blood transfusions. You can discuss any worries you may have with your child's doctor and/or nurse.

Iron Overload

When your child receives a blood transfusion the red blood cells are broken down. Red blood cells normally survive in the body for around 90 days.

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Usually when red blood cells get broken down in the body, we re-use the iron in the blood to make new red blood cells.

Unfortunately, for a child with Thalassaemia, the iron is not re-used and so gets stored in different parts of the body. Over time this iron can start to cause damage.

To prevent this from happening, your child will need an additional treatment to help remove the iron from the body.

At present there are 3 types of treatment used

- Desferrioxamine (Desferal) – this is an infusion which is given through a small needle inserted under the skin. It is usually given overnight, at home, up to 6 nights per week.
- Deferasirox (Exjade) – this is a tablet that is given once a day.
- Deferiprone (Deferiprox) – this is a tablet usually given three times a day.

The doctor will advise which is the most suitable treatment for your child and will discuss this with you before their treatment begins.

When will this treatment start?

This treatment will start when your child is around 12 – 18 months old but this can vary from child to child and may start earlier.

Will my child develop normally?

If your child has the transfusions and medicines that the Haematologist has ordered, they should grow and develop normally. Many people with Thalassaemia go on to university/college, get married and have children.

Is there any other treatment my child can have?

There is no other recommended treatment at present. There are many researchers looking at treatments for thalassaemia. Your haematologist will tell you about them if they may be relevant to you, and of course you are always welcome to ask.

Is there a cure for Thalassaemia?

The only cure for Thalassaemia at present is a stem cell transplant from someone else.

If your child has a full genetic match, it is possible to have a bone marrow transplant. They will need drug therapy before the transplant to shut down the production of their own bone marrow.

Their own bone marrow can then be replaced by the donor's stem cells to allow the body to make normal red blood cells.

There are many possible complications when having a bone marrow transplant and you can discuss these with your child's doctor.

Further information

We hope this information will help you to understand your Thalassaemia.

Some parents can find dealing with the news of their child's condition difficult and you may need extra support. The doctors and nurses will try to understand the feelings you may have and are there to help you if you have any questions or need advice.

Please discuss any worries/concerns you may have when you attend with your child for appointments. You can contact them by telephone between appointments if you need to.

Contact numbers

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Other useful contacts

Scottish Paediatric & Adult Haemoglobinopathy (SPAH)
www.spah.scot.nhs.uk

UK Thalassaemia Society, 19 Broadway, Southgate Circus, N14 6PH.
Tel: 020 8882 0011 www.ukts.org

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SPAH
SCOTTISH PAEDIATRIC AND ADULT HAEMOGLOBINOPATHIES NETWORK

NHS SCOTLAND

Thalassaemia Major

Information for New Parents / Carers