NHS SCOTLAND

SPAH
SCOTTISH PAEDIATRIC AND ADULT HAEMOGLOBINOPATHIES NETWORK

Information for new parents and carers

Transfusion dependent thalassaemia

This document has been prepared by NHS National Services Scotland (NSS) on behalf of SPAH. Accountable to Scottish Government, NSS works at the heart of the health service providing national strategic services to the rest of NHS Scotland and other public sector organisations to help them deliver their services more efficiently and effectively. The SPAH Network is a collaboration of stakeholders involved in care of patients with haemoglobinopathies, who are supported by an NSS Programme Team to drive improvement across the care pathway.

Review: September 2028
NSD610-017.34 V2

What is thalassaemia?

Thalassaemia is an inherited blood condition. The name comes from Greek and means “anaemia by the sea”. Thalassaemia is more common in Asia, India, and parts of Southern Europe.

Children with thalassaemia can’t make enough healthy mature red blood cells. This lack of red blood cells is known as anaemia. Transfusion dependent thalassaemia can affect a child’s growth and overall health.

Without treatment, a child with this condition would not survive childhood.

What is blood made of?

Blood is made up of millions of red blood cells in liquid called plasma. Plasma also carries white cells which can fight infection, and platelets which 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 type is caused by not enough iron.

Thalassaemia is a different type of anaemia and has nothing to do with how much iron is in your diet**.** Thalassaemia is caused by faulty genes that affect the production of haemoglobin. This causes red blood cells to break down too quickly and leads to fewer red blood cells (anaemia).

How is transfusion dependent thalassaemia diagnosed?

If your baby was born in the UK, a few days after birth they may have had a blood test taken from their heel. From this test several different conditions can usually be identified. One of them is thalassaemia.

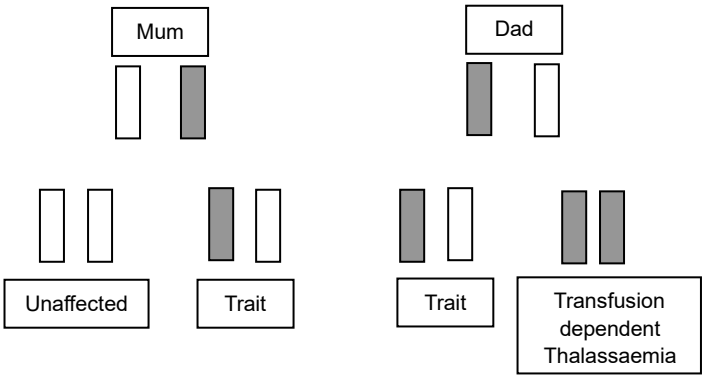
If the first test showed signs of thalassaemia, you would have been invited to speak with a haematologist (a doctor who is a blood specialist) or a paediatrician (a doctor who looks after children).

At that appointment, the doctor would have explained the results. They would also have taken another small sample of blood to confirm the diagnosis.

If your child wasn’t tested as a baby, thalassaemia might have been diagnosed later if they were unwell or not growing properly.

How does my child get thalassaemia?

Thalassaemia is passed down from both **mother and father**. It's shown in the diagram below.



If both parents are carriers of thalassaemia then each time you have a baby there is a:

- 1 in 4 chance of having a baby with transfusion dependent thalassaemia
- 2 in 4 chance of having a baby that carries the thalassaemia gene (thalassaemia trait). That normally causes **no** health problems for the baby
- 1 in 4 chance of having a baby that doesn't carry an abnormal gene and doesn't have thalassaemia

It is important to understand these chances are the same with every pregnancy. It doesn't mean that if you have four children, only one will have the condition.

Understanding the genetics of thalassaemia can be confusing. You may find it helpful to speak with a genetic counsellor who can explain it more clearly. They can also help you think about how this might affect your choices for future pregnancies. This can be arranged at your first appointment or at a later stage if you prefer.

How is transfusion dependent thalassaemia treated?

Thalassaemia is treated with regular blood transfusions, usually every four 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 your baby is born, their blood contains fetal or baby haemoglobin called Haemoglobin F (HbF). As the baby grows the amount of baby HbF decreases usually over the first few months.

Normally at this time adult blood would be produced. 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 check their haemoglobin levels. Your child's weight and general wellbeing will also be carefully monitored. All this will be considered before the doctor recommends that your child needs to start blood transfusions, which is usually around 3-6 months of age.

Are blood transfusions safe?

Yes, they are generally very safe. But like all medical treatments, there are some risks. You will be given more information about this before your child starts blood transfusions. If you are worried, talk to the doctor or nurse.

What is iron overload?

Each blood transfusion adds iron to the body. Over time this extra iron can build up and cause damage.

To stop this, your child will need an additional treatment to help remove the iron from the body. This will usually start at around 12-18 months
but this can vary from child to child and may start earlier.

There are three types of treatment:

- Desferrioxamine (Desferal): this is an infusion usually given overnight at home
- Deferasirox (Exjade): this is a tablet taken once a day
- Deferiprone (Deferiprox) – this is a tablet usually taken 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.

Will my child develop normally?

Yes. If your child has the transfusions and medicines the doctor recommends, they should grow and develop normally.

Is there any other treatment?

Right now, there is no other recommended treatment for children. There are many researchers looking at treatments for thalassaemia. Your haematologist will tell you about treatments relevant to your child, and of course you are always welcome to ask.

Is there a cure for thalassaemia?

Right now, the only cure for thalassaemia is a stem cell transplant from a suitably matched donor.

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 stop? the production of their own bone marrow. Their own bone marrow can then be replaced by the donor's stem cells to allow their 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.

Gene therapy

Gene therapy for patients with transfusion dependent thalassaemia is a new and developing treatment. Talk to your doctor if you are interested in finding out more.

Need more support?

We hope this information will help you to understand your child's thalassaemia.

You may find dealing with the news of your child's condition difficult and may need extra support. The doctors and nurses understand this can be hard, but they are there to help you if you have any questions or need advice.

Please discuss any worries or concerns you have when you attend your child's appointments. You can also contact them by telephone between appointments if you need to.

Local contact details

	Name	Contact number
Consultant Haematologist or Paediatrician		
Clinical Nurse Specialist		

Further information

Additional information can be found on the Scottish Paediatric and Adult Haemoglobinopathies Network (SPAH) website. spah.scot.nhs.uk

If you require an alternative format, please contact nss.equalitydiversity@nhs.scot, telephone: 0131 275 600

British Sign Language, please contact Scotland BSL: Contact Scotland (contactscotland-bsl.org)