Haemoglobin H Disease - Alpha(α) Thalassaemia:

Information Sheet

Produced by Specialist Haematology, Canterbury Health Laboratories

What is Haemoglobin?

Haemoglobin (Hb) is a protein found in red blood cells; it carries oxygen from the lungs to all parts of the body and gives blood its red colour. Hb is made up of haem groups, which contain iron and globin chains which are made of protein.

What is Thalassaemia?

Thalassaemia is a type of inherited anaemia (red blood cell deficiency).

People with thalassaemia don't produce enough globin chains. The two main types are called alpha (α) and beta (β) thalassaemia, depending on which globin chains are affected.

Thalassaemia occurs most frequently in people of

- Southern Asian
- Mediterranean, Middle Eastern
- Indian, African and Polynesian ancestry.

Alpha (α) Thalassaemia

People with α thalassaemia don't produce enough α globin chains. To produce normal amounts of α globin, a total of **four** α globin genes are needed; two from each parent.

α thalassaemia can range from a very mild condition through to the most severe form which results in fetal or newborn death.

There are two types of α thalassaemia:

- α^+ thalassaemia
- α^0 thalassaemia.

α^+ thalassaemia

α^+ thalassaemia is commonly seen in Polynesian, Asian and African people, it is a very mild condition with no associated health issues. This type of thalassaemia produces small red blood cells which may be mistaken as a sign of iron deficiency anaemia.

People with α^+ thalassaemia are **not** at risk of producing a child with the most severe form of alpha thalassaemia.

α^0 thalassaemia

α^0 thalassaemia trait is commonly seen in Southeast Asian, Chinese, Filipino and Mediterranean people. It is a mild condition not associated with any health issues although does produce small red blood cells which may be mistaken as a sign of iron deficiency anaemia.

People with α^0 thalassaemia trait **are** at risk of producing a child with the most severe form of α thalassaemia.

What is Haemoglobin H Disease?

Haemoglobin H (Hb H) disease is a type of α thalassaemia which occurs when a person inherits only **one** out of the four α globin genes and produces a disorder of variable severity.

Hb H disease can occur if one parent has α^0 thalassaemia trait and one parent has α^+ thalassaemia.

People with Hb H disease **are** at risk of producing a child with the most severe form of α thalassaemia.

What is the Treatment?

Hb H disease is an extremely variable condition which may require significant medical follow-up.

Anaemia can be moderate to severe and may worsen during pregnancy, infection or exposure to some drugs. Splenomegaly (enlargement of the spleen) occurs in most patients. The degree of anaemia can be monitored by blood tests, and if the anaemia is severe a splenectomy (removal of the spleen) may help.

However, young children without a spleen may develop very severe infections and surgery should be delayed until 5 years of age. A special vaccination to help prevent infections is always given before splenectomy.

Jaundice (yellowing of the skin and eyes) is usually present.

Children need to be monitored to ensure their growth and development is progressing as expected.

Folic acid supplements may be needed in times of increased demand on red blood cell production e.g pregnancy.

Exposure to oxidant drugs should also be avoided. For example, some sulphonamides, which are a type of antibiotic, **may** be harmful. Discuss this with your doctor.

How is Thalassemia inherited?

Thalassaemia is inherited from either one or both parents who carry the thalassaemia gene on the DNA in their cells.

If a person is born with thalassaemia, they will have it all their life. It is **not** contagious or infectious.

A person with α thalassaemia inherits and in turn passes on to their children a combination of normal α globin genes and α thalassaemia genes which produce no or very few α globin chains. There are different types of α thalassaemia depending on the number of functioning α globin genes.

Hb H disease occurs when a person inherits only one out of the four α globin genes and produces a disorder of variable severity.

The most severe form of α thalassaemia occurs when all four α globin genes are missing and results in fetal or newborn death.

How Will This Affect My Children?

The type of alpha thalassaemia each parent has determines the risk of a child having the severe form of alpha thalassaemia, that is, when all four alpha globin genes are missing.

For a child to be at risk of the severe form of α thalassaemia **both** parents would need to have a gene for α^0 thalassaemia.

If your partner has a form of α^+ thalassaemia, you are at risk of having a child with Hb H disease.

If your partner carries a gene for α^0 thalassaemia there would be a 25% chance of a child having the severe form of alpha thalassaemia.

If you have haemoglobin H disease it is important to have your partner tested for thalassaemia before you have children.

Is There a Test for Thalassemia?

YES ! Blood tests and family genetic studies can show whether an individual has thalassaemia.

If you have Hb H disease it is important to have your partner tested for thalassaemia before you have children. Other members of your family who are of reproductive age should also be tested for thalassaemia.

The documentation of thalassaemia status is important as:

1. Patients with Hb H disease may require regular checks and monitoring to ensure their well-being.
2. It avoids the need for repeat testing in the future.
3. Alpha thalassaemia can be confused with iron deficiency and iron therapy may be given unnecessarily.
4. Thalassaemia is an inherited disorder which if passed on to a child by **both** parents could result in a severe condition.

If two people with alpha thalassaemia do become parents and there is a risk of a severe disorder in the foetus, special prenatal tests can be done to detect or rule this out.

Early diagnosis of a possible severe form of alpha thalassaemia is important to prevent as many complications as possible.

Can Thalassemia Be Prevented?

Thalassaemia cannot be prevented at this time, but a programme of health education, testing for the trait, genetic counselling and prenatal diagnosis can provide families with full medical information to help them have healthy children.

People who think they may have thalassaemia should go to their family doctor for advice and testing.

Your family doctor can contact a Haematologist at the Community Laboratory or the Haematology Department at Canterbury Health Laboratories on (03) 364 0300 if further clinical input is required.

The Specialist Haematology Laboratory at Canterbury Health Laboratories provides a comprehensive diagnostic service in conjunction with the family doctor.

Contact for Canterbury Health Laboratories

Laboratory Services: Specialist Haematology Laboratory, Canterbury Health Laboratories.
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On behalf of the Clinical Haematology Team at Christchurch Hospital