Alpha (α) Thalassaemia: Information Sheet

Produced by Specialist Haematology, Canterbury Health Laboratories

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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 Thalassaemia

People with α thalassaemia don't produce enough α globin chains. 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 South East 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.

A diagnosis of α^0 thalassaemia is an important factor when planning to have children. When **two** people with α^0 thalassaemia trait have children, they have a 25% chance of producing a child with the most severe form of α thalassaemia resulting in foetal or newborn death.

What is the Treatment?

*Thalassaemia trait is **not** an illness and requires no regular medical follow-up or treatment.*

Thalassaemia trait may be mistaken for iron deficiency anaemia and iron tablets may be given unnecessarily.

Iron tablets should only be taken if your doctor has done a special blood test to check that you lack iron.

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.

To produce normal amounts of α globin, a total of **four** α globin genes are needed; two from each parent.

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.

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 α thalassaemia, that is, when all four α globin genes are missing.

People who have α^+ thalassaemia are **not** at risk of having a child with the severe form of α thalassaemia. It is therefore unnecessary to test your partner or other family members for thalassaemia.

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

When **two** people with α^0 thalassaemia trait, become parents, there are several possible outcomes:

- 25% chance that the child will inherit the α^0 thalassaemia from each parent, resulting in the severe form of α thalassaemia.
- 25% chance that the child will inherit the normal α globin genes from each parent and will not have thalassaemia.
- 50% chance that the child will inherit the normal α globin gene from one parent and the α^0 thalassaemia gene from the other and have α^0 thalassaemia trait like its parents.

These odds are the same for each pregnancy when **both** parents have α^0 thalassaemia trait.

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 condition of variable severity.

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

If you have α^0 thalassaemia trait 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 a person has thalassaemia.

If you have α^0 thalassaemia trait 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. It avoids the need for repeat testing in the future.
2. Thalassaemia trait is often confused with iron deficiency and iron therapy may be given unnecessarily.
3. 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 α^0 thalassaemia do become parents, there are special prenatal tests that can be done to detect a severe alpha thalassaemia in the fetus.

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

Can Thalassemia Be Prevented?

Thalassaemia can not 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

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On behalf of the Clinical Haematology Team at Christchurch Hospital