

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.

Beta Thalassaemia

People with β thalassaemia don't produce enough β globin chains.

Most people have a mild form, called β thalassaemia trait (also called heterozygous β thalassaemia or β thalassaemia minor).

The most severe form of β thalassaemia is β thalassaemia major. This is a severe condition treated with regular blood transfusions from early childhood. Iron chelation therapy is also needed to help the body remove excess iron from the repeat transfusions.

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 β globin, a total of **two** β globin genes are needed; one from each parent.

A person with β thalassaemia trait inherits one normal β globin gene and one β thalassaemia gene which produces no or very few β globin chains.



How Will This Affect My Children?

Every child inherits haemoglobin genes from each parent. The type of haemoglobin gene inherited from the parent determines the type of haemoglobin the child will have.

A person with β thalassaemia trait will pass on either a β thalassaemia gene **or** a normal β globin gene to each of their children.

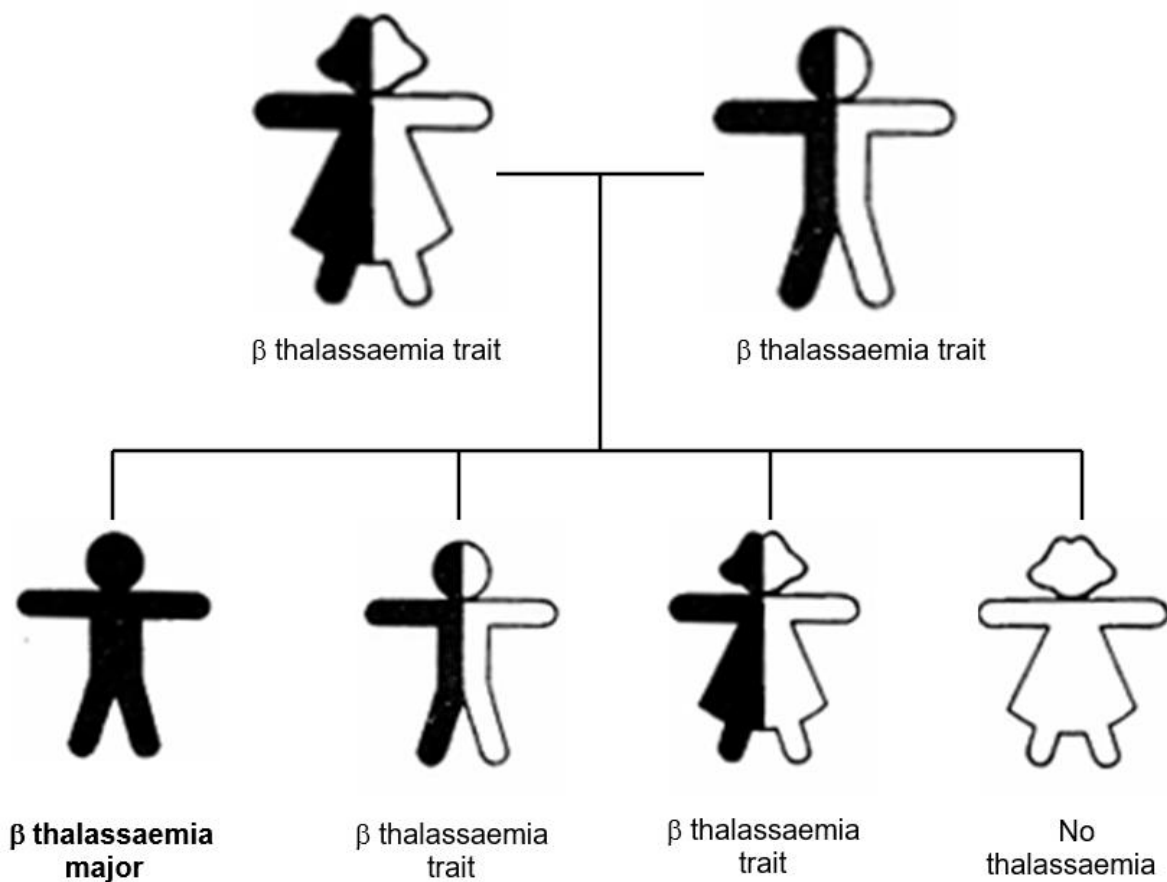
When **both** parents carry a gene for thalassaemia, there is a chance that their children could inherit a severe form of thalassaemia.

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

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

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

BOTH PARENTS WITH β THALASSAEMIA TRAIT



Why is Knowing About Thalassaemia so Important?

The inheritance of β thalassaemia trait produces a mild condition.

However, the inheritance of a β thalassaemia gene from **both** parents produces β thalassaemia major which is a severe condition requiring lifelong medical treatment.

There are other types of thalassaemia or variant haemoglobins which if inherited together with β thalassaemia can also produce a severe condition.

If you have β thalassaemia trait it is important to have your partner tested for the presence of thalassaemia or a variant haemoglobin 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 β 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 thalassaemia do become parents and there is a risk of a severe disorder in the fetus, special prenatal tests can be done to detect or rule this out.

Early diagnosis of a possible severe form of thalassaemia is important so that treatment can 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.

Address: 524 Hagley Ave, Christchurch 8011.

Phone: (03) 364 0300.
