Compound Heterozygous Delta-Beta (δβ)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. Less commonly, two globin chains are affected as in delta-beta (δβ) thalassaemia.

Delta-Beta Thalassaemia

δβ thalassaemia usually occurs in one of two ways; either a person inherits a combined δβ thalassaemia gene from one parent or they inherit a β thalassaemia gene from one parent **and** a δ thalassaemia gene from the other parent.

Most people with δβ thalassaemia have a mild form, called **δβ thalassaemia trait** (sometimes called heterozygous δβ thalassaemia),

δβ thalassaemia occurs most frequently in people of the

- **Central Mediterranean:** Italy, Sicily, and Malta
- **Southeastern Europe:** The Balkans, Greece, and Hungary
- **Eastern Mediterranean & Near East:** Turkey, Israel, and Egypt

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.

Often people with δβ thalassaemia continue to produce large amounts of foetal Hb (Hb F) into adult life. Hb F is the main Hb produced during the foetal period, but from birth is gradually replaced by the production of adult Hb (Hb A). During pregnancy the increased levels of Hb F seen in δβ thalassaemia could be mistaken as an indication of a fetal-maternal bleed.

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 delta and beta globin, a total of **two** delta (δ) and **two** beta (β) globin genes are needed, one from each parent.

A person with compound heterozygous δβ thalassaemia inherits a β thalassaemia gene and a normal δ gene from one parent, and a normal β gene and a δ thalassaemia gene from the other parent.

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.

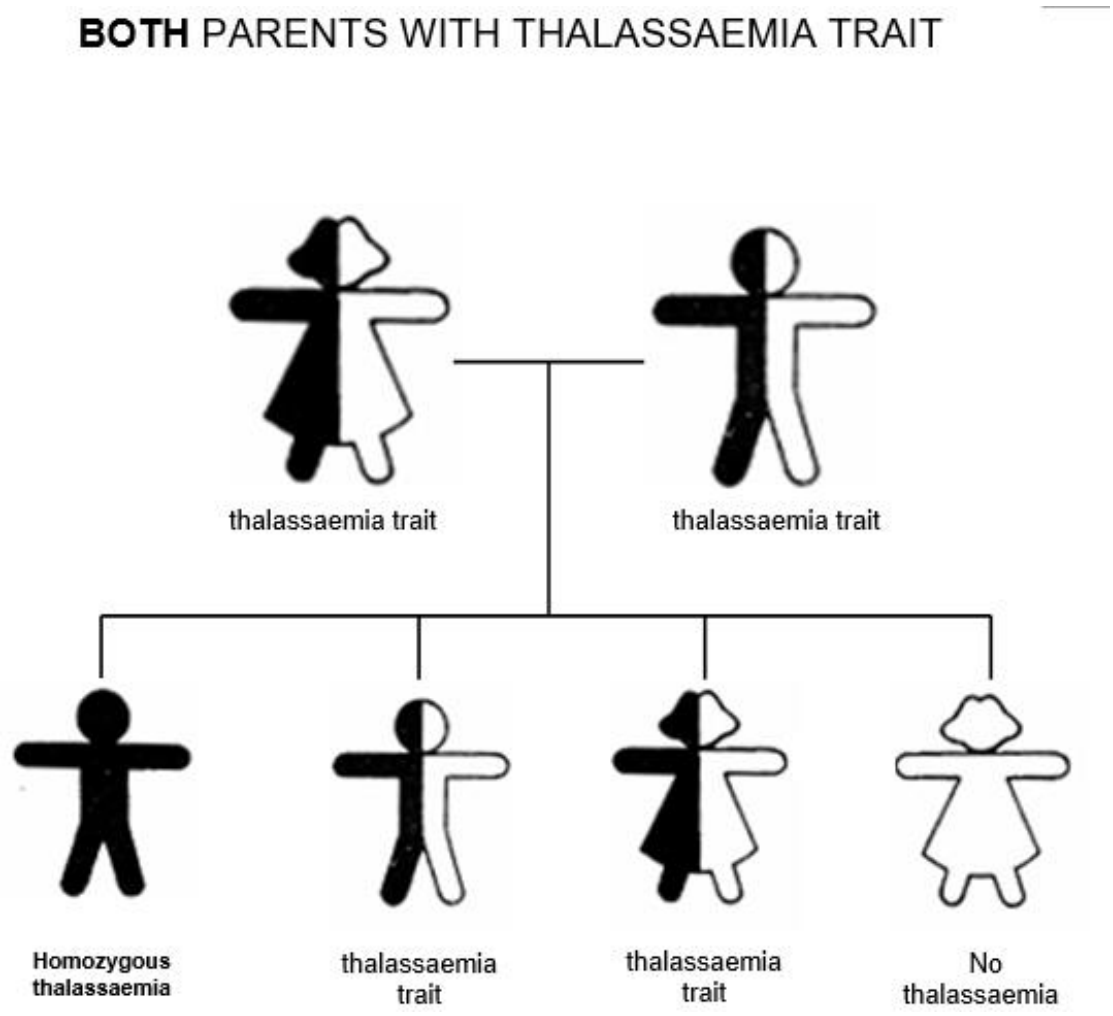
A person with compound heterozygous δβ thalassaemia will pass on either a δ thalassaemia gene **or** a β thalassaemia gene to each of their children.

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

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

- 25% chance that the child will inherit a thalassaemia gene from each parent, resulting in a more serious condition.
- 50% chance that the child will inherit one normal haemoglobin gene and one thalassaemia gene and have thalassaemia trait like its parents.
- 25% chance that the child will inherit a normal haemoglobin gene from each parent and will not have thalassaemia.

These odds are the same for each pregnancy when **both** parents carry a gene for thalassaemia.



Why is Knowing About Thalassaemia so Important?

The inheritance of a β thalassaemia gene from the parent with δβ thalassaemia **and** a second beta (β) thalassaemia gene (another type of inherited anaemia frequent in the Mediterranean) from the other parent, produces β thalassaemia major which is a severe condition requiring life long medical treatment.

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

If you have heterozygous δβ thalassaemia 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 a person has thalassaemia.

If you have δβ 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.
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Phone: (03) 364 0300.

On behalf of the Clinical Haematology Team at Christchurch Hospital