Haemoglobin E - Beta 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.

The combination of different globin chains with the haem groups produces the different types of haemoglobin present in people. The normal adult haemoglobin is Hb A.

Where a person has inherited Hb A from both parents, they will have the usual Hb combination, Hb AA.

What is Haemoglobin E?

Haemoglobin E (HbE) is a variant Hb and occurs when the structure of the globin chain is altered. It is frequently seen in people of Southeast Asian ancestry.

Heterozygous haemoglobin E (haemoglobin E trait) occurs when a person inherits the normal haemoglobin from one parent and the haemoglobin E from the other (Hb AE).

When a person inherits the haemoglobin E from both parents, they are said to have homozygous haemoglobin E (Hb EE).

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 Haemoglobin E / Beta Thalassaemia?

Haemoglobin E / beta thalassaemia (Hb E/β thalassaemia) occurs when a person inherits a haemoglobin E gene from one parent **and** a beta (β) thalassaemia gene from the other parent.

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

What is the Treatment?

Haemoglobin E/beta thalassaemia is an extremely variable condition, usually requiring significant medical follow-up.

It can present as a condition that is comparable to beta thalassaemia major and require regular blood transfusions and iron chelation therapy.

Or it may follow a milder course with regular haemoglobin checks being the main medical follow-up required.

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

In all instances, folic acid supplements will be required.

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 Hb E/beta thalassaemia will in turn pass on **either** a Hb E gene or a β thalassaemia gene to each of their children.

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

Why is Knowing About Haemoglobin E/Beta Thalassaemia so Important?

Haemoglobin E/beta thalassaemia is a moderately severe condition.

A person with Hb E/beta thalassaemia will pass on **either** a Hb E gene or a β thalassaemia gene to each of their children which will produce a mild disorder in the child.

However, the inheritance of a beta thalassaemia gene from **both** parents produces beta 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 beta thalassaemia can also produce a severe condition.

If you have Hb E / beta 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 Haemoglobin E and/or Thalassemia?

YES ! Blood tests can show whether an individual has haemoglobin E, thalassaemia or a combination of the two.

If you have Hb E/beta thalassaemia it is important to have your partner tested for thalassaemia or a variant Hb before you have children. Other members of your family who are of reproductive age should also be tested.

The documentation of carrier status or of a condition requiring medical follow-up is important as:

1. Patients with Hb E/beta thalassaemia may require regular checks and monitoring to ensure their well-being.
2. It avoids the need for repeat testing in the future.
3. Thalassaemia trait or haemoglobin E are often confused with iron deficiency and iron therapy may be given unnecessarily.
4. Haemoglobin E and/or thalassaemia are inherited disorders which if passed on to a child by **both** parents could result in a severe condition.

If two people who carry a variant haemoglobin or 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 Haemoglobin E or Thalassemia Be Prevented?

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

People who think they may have haemoglobin E or 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.
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On behalf of the Clinical Haematology Team at Christchurch Hospital