

**Information for couples where
one partner carries Delta-Beta
thalassaemia
and one carries Haemoglobin E**

Contacts for expert counselling centre

Couple "at risk" for Haemoglobin E/ Delta-beta thalassaemia

	Name	Date of Birth
Ms		
Mr		

- *One of you carries delta-beta thalassaemia and one carries haemoglobin E. This means that, as a couple, you could have children with haemoglobin E/ delta-beta thalassaemia . This booklet explains what this means.*
- *Your full blood test results are given below.*
- *Keep this booklet with your personal papers, so that you can refer to it again. Show it to your doctor and midwife in every pregnancy.*

Blood test results

Name	Date of Test	Hb	MCH	MCV	Hb A ₂	Electro-phoresis	DNA data*

** this is essential information for all couples at risk for haemoglobin E/ delta-beta thalassaemia*

Centre where tests were done

Address	
Telephone	Fax
Other	

Issued

Date
Signature of Doctor or Counsellor

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What is an “At Risk Couple”?

One of you carries delta-beta thalassaemia and the other carries haemoglobin E. You are both healthy but you could have children with haemoglobin E/ delta-beta thalassaemia. This is why doctors call you an “at risk couple”.

In your case this is not a good description, because haemoglobin E/ delta-beta thalassaemia is not thought to be a serious condition.

Your blood test results are written in the front of this booklet.

Delta-beta thalassaemia and haemoglobin E are variations in the blood, of a kind that doctors call “haemoglobin disorders”.

What are haemoglobin disorders?

Haemoglobin disorders cause changes in the haemoglobin in a person's red blood cells. They are *inherited* - they are handed on from parents to their children, they are present at birth, and they remain the same for life.

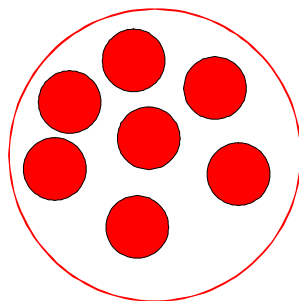
Haemoglobin is a component of the blood. It contains iron, which makes it bright red. This is why your blood is red. Your body needs oxygen to function: as your blood circulates haemoglobin picks up oxygen in your lungs and carries it round to all parts of your body. The usual type of haemoglobin is called haemoglobin A.

Haemoglobin is packed into *red blood cells*. Blood contains millions of red blood cells floating in a slightly yellow fluid called plasma.

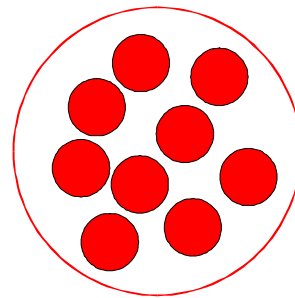
What is delta-beta thalassaemia?

Delta-beta thalassaemia is an unusual type of *beta thalassaemia*. A carrier's red blood cells are slightly smaller than usual. Carriers make up for having small red blood cell by making more of them. Delta-beta thalassaemia carriers also have some *haemoglobin F*, which is the usual haemoglobin of unborn babies. This makes their thalassaemia milder than typical beta thalassaemia. Their blood functions normally, and they are healthy people.

The picture shows the usual kind of red blood cells, and a delta-beta thalassaemia carrier's red blood cells, seen down a microscope.



Usual red blood cells



A delta-beta thalassaemia carrier's red blood cells

How do you find out you carry delta-beta thalassaemia?

People find out they carry delta-beta thalassaemia by a special blood test called a "haemoglobinopathy screen". This shows that:

- they have small red blood cells
- they have between 15 and 30% of haemoglobin F in their blood. Most people have less than 1% of haemoglobin F.
- the haemoglobin F is distributed unevenly among their red blood cells.

They also need a special "DNA" test to confirm that they carry delta-beta thalassaemia.

A DNA test is essential to be sure that they do not carry a similar but harmless condition called "hereditary persistence of fetal haemoglobin" (HPFH for short).

What is haemoglobin E?

Haemoglobin E carriers have an unusual haemoglobin called haemoglobin E, as well as haemoglobin A. They also have smaller red blood cells than usual. They make up for having small red blood cells by making more of them. Their blood functions normally and they are healthy people.

How do you find out you carry haemoglobin E?

People find out they carry haemoglobin E through a special blood test called a *haemoglobinopathy screen*. This shows that:

- they have small red blood cells.
- they have about 30% of haemoglobin E in their blood.

How are delta-beta thalassaemia and haemoglobin E inherited?

They are inherited through *genes*. Every human characteristic, such as eye colour, or height, or type of haemoglobin is controlled by genes that we inherit from our parents. A child inherits *two* genes for every characteristic, one from each parent. Most people have inherited two genes for *haemoglobin A*, the usual type of haemoglobin.

A ***delta-beta thalassaemia carrier*** has inherited a gene for haemoglobin A from one parent and a gene for delta-beta thalassaemia from the other. Their delta-beta thalassaemia gene cannot make any haemoglobin A, but they make some haemoglobin F instead. Their normal haemoglobin A gene makes enough haemoglobin for their red blood cells to function normally.

A ***haemoglobin E carrier*** has inherited a gene for haemoglobin A from one parent and a gene for haemoglobin E from the other. Their haemoglobin E gene makes less haemoglobin than usual, but their haemoglobin A gene makes enough haemoglobin for their red blood cells to function normally.

When one partner carries delta-beta thalassaemia and the other carries haemoglobin E, a child could inherit both the delta-beta thalassaemia gene and the haemoglobin E gene. This child would have *haemoglobin E/delta-beta thalassaemia*. When a person has haemoglobin E/delta-beta thalassaemia neither of their haemoglobin genes functions fully, and their red blood cells contain less haemoglobin than usual. This causes a mild anaemia.

Couples where one partner carries delta-beta thalassaemia and the other carries haemoglobin E have the following chances ***in each pregnancy***

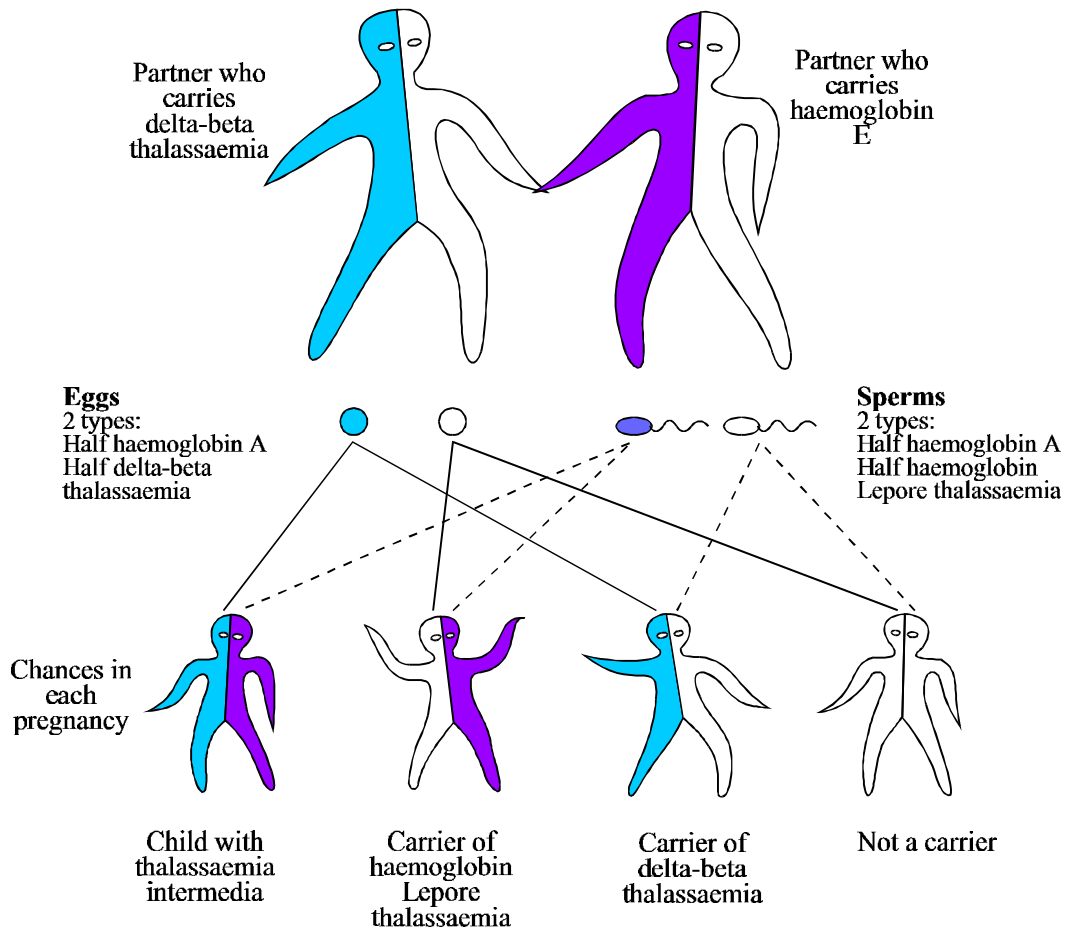
- a 1-in-4 chance of a child that is not a carrier.
- a 1-in-2 chance of a child that is a healthy carrier of delta-beta thalassaemia or haemoglobin E.
- a 1 in 4 chance of a child with haemoglobin E/ delta-beta thalassaemia.

In every pregnancy your baby is more likely to be normal or a carrier than to have haemoglobin E/delta-beta thalassaemia.

How could a child inherit Haemoglobin E/ Delta-Beta thalassaemia from you?

When a child is conceived, it inherits one gene for haemoglobin from each parent. The picture shows that when one parent carries delta-beta thalassaemia and the other carries haemoglobin E there are four possibilities.

Here we describe the possibilities when the woman carries delta-beta thalassaemia and the man carries haemoglobin E. Of course, it may also be the other way around.



Women usually produce one egg each month. When a woman carries delta-beta thalassaemia, each egg contains either her normal haemoglobin gene or her delta-beta thalassaemia gene, but not both.

Men make sperm all the time. When a man carries haemoglobin E, each sperm carries either his normal gene or his haemoglobin E gene, but not both.

- If a normal egg is fertilised by a normal sperm, the child will not carry any haemoglobin disorder.
- If a normal egg is fertilised by a haemoglobin E sperm the child will carry haemoglobin E.
- If a delta-beta thalassaemia egg is fertilised by a normal sperm the child will carry delta-beta thalassaemia.
- If a delta-beta thalassaemia egg is fertilised by a haemoglobin E sperm, the child will have haemoglobin E/ delta-beta thalassaemia.

What is Haemoglobin E/ Delta-beta thalassaemia ?

Haemoglobin E/ delta-beta thalassaemia is very uncommon, and we do not know of any case reports in the medical literature. *All available scientific information suggests that haemoglobin E/ delta-beta thalassaemia is an extremely mild condition.*

We expect that people with haemoglobin E/ delta-beta thalassaemia would not be able to make haemoglobin quite as well as usual. As a result their red blood cells would contain a little less haemoglobin than usual, and would be smaller than usual. A person with haemoglobin E/ delta-beta thalassaemia is likely to be more like a thalassaemia carrier than a person with a serious haemoglobin disorder.

If a baby has haemoglobin E/delta-beta thalassaemia, the parents should take the baby to see an expert in haemoglobin disorders every few months for the first year or so. After that, visits should be made every 6 months to every year until the child is grown up.

Finding out whether or not your Baby has Haemoglobin E/ Delta-Beta thalassaemia

You can find out as soon as the baby is born, through *neonatal diagnosis* (new-born diagnosis).

Neonatal diagnosis is done on blood taken from the baby's cord at birth, or from a heel-prick done a few days later.

DNA analysis is the only reliable method for diagnosing haemoglobin E/delta-beta thalassaemia in a new-born baby.

Do you want to know more?

If you want to know more, or you are unsure what to do, you may find it useful to visit an expert centre for haemoglobin disorders. You can discuss your case with an expert who will answer your questions. To make an appointment, phone the number on the front of this booklet directly, or ask your counsellor or family doctor to make an appointment for you.