Information for couples where both partners carry Delta-Beta Thalassaemia

Couple "at risk" for homozygous delta-beta thalassaemia

	Name	Date of Birth
Ms		
Mr		

- ***One of you carries beta thalassaemia and the other carries delta-beta thalassaemia. This means that, as a couple, you could have children with homozygous delta-beta thalassaemia. This booklet explains the implications.***
- ***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 alpha or beta thalassaemia*

Centre where tests were done

Address	
Telephone	Fax
Other	

Issued

Date
Signature of Doctor or Counsellor

Contents

What is an “At Risk Couple”?

You both carry delta-beta thalassaemia. You are both healthy but you could have children with *homozygous delta-beta thalassaemia*. This is why you are called an “at risk couple”. ***In your case this is not a good description, because homozygous delta-beta thalassaemia is not usually a serious condition.***

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

Delta-beta thalassaemia is one of a range of variations in the blood, 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.

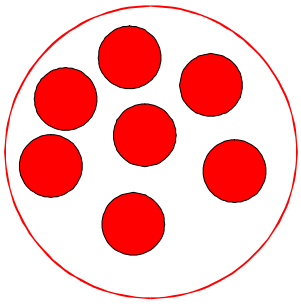
Haemoglobin is a component of the blood. It contains iron, which makes it bright red. This is why your blood is red. Your body needs oxygen to function: as your blood circulates haemoglobin picks up oxygen in your lungs and carries it round to all parts of your body. The usual type of haemoglobin is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

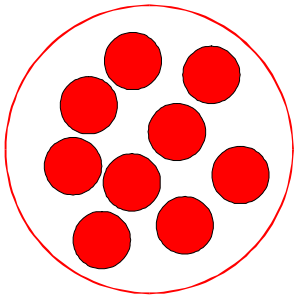
What is delta-beta thalassaemia?

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 a 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).

How is delta-beta thalassaemia inherited?

It is 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.* This is 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.

When a couple both carry delta-beta thalassaemia a child could inherit a delta-beta thalassaemia gene from both parents. This child would have *homozygous delta-beta thalassaemia*. When a person has homozygous delta-beta thalassaemia neither of their haemoglobin genes functions normally. Their red blood cells contain only haemoglobin F, but they cannot make quite enough of it, and they have a moderate form of anaemia, called *mild thalassaemia intermedia*.

Mild thalassaemia intermedia means that the child can manage without regular blood transfusions.

What are your chances of having children with Mild Thalassaemia Intermedia?

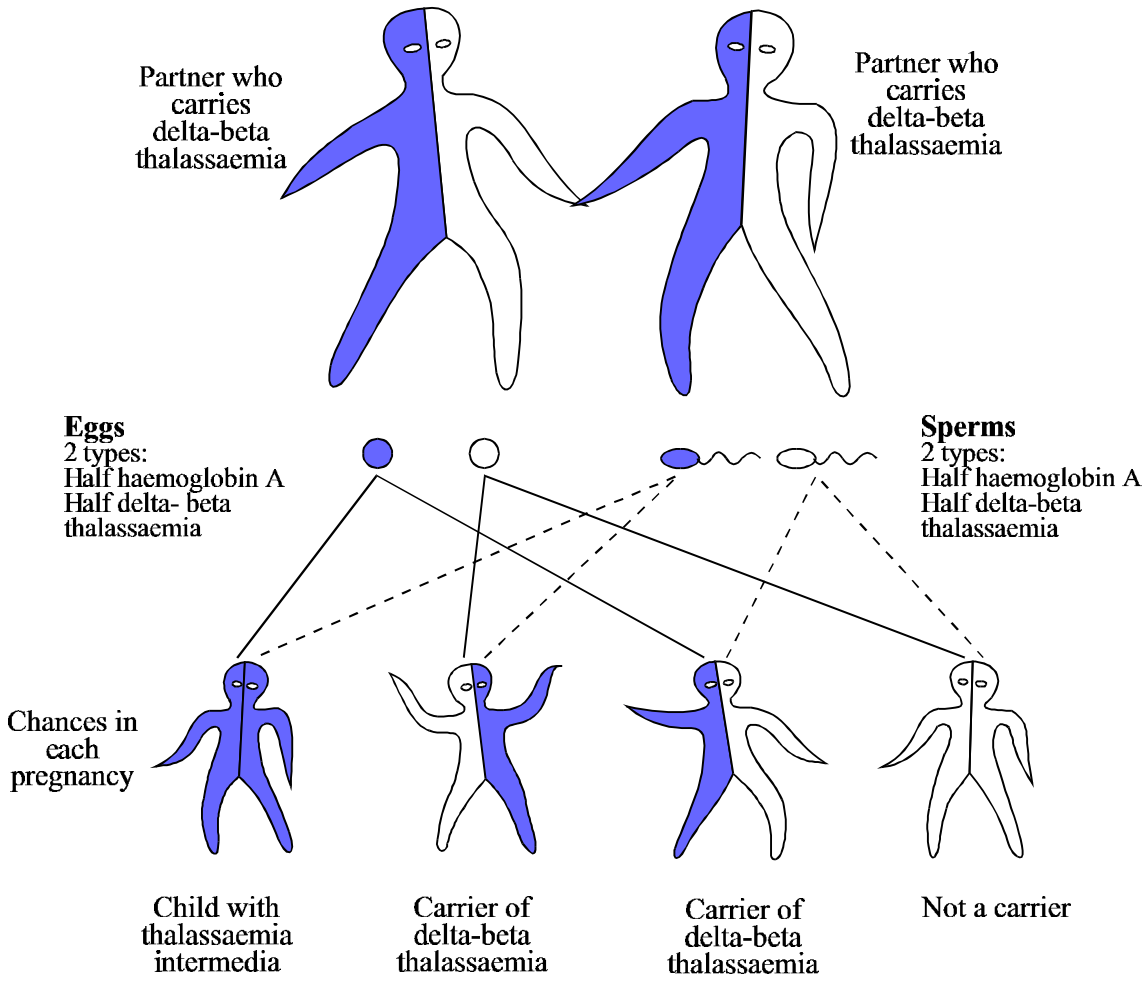
Couples where both partner carry delta-beta thalassaemia have the following chances ***in each pregnancy***

- a 1-in-4 chance of a baby that is not a carrier.
- a 1-in-2 chance of a baby that is a healthy carrier of delta-beta thalassaemia.
- a 1 in 4 risk of a baby with homozygous delta-beta thalassaemia - mild thalassaemia intermedia.

In every pregnancy your chance of having a healthy child is much higher than your risk of having a child with mild thalassaemia intermedia.

How could a child inherit Mild Thalassaemia Intermedia from you?

When a child is conceived, it inherits one gene for haemoglobin from each parent. The picture shows that when both partners carry delta-beta thalassaemia there are four possibilities.



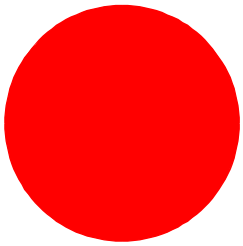
Women usually produce one egg each month. When a woman carries delta-beta thalassaemia, each egg contains either her normal haemoglobin gene or her delta-beta thalassaemia gene, but not both.

Men make sperm all the time. When a man carries delta-beta thalassaemia, each sperm carries either his normal gene or his delta-beta thalassaemia gene, but not both.

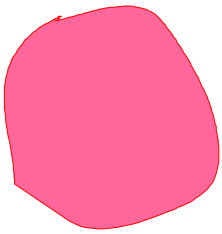
- If a normal egg is fertilised by a normal sperm, the child will not carry any haemoglobin disorder.
- If a normal egg is fertilised by a delta-beta thalassaemia sperm the child will carry delta-beta thalassaemia.
- If a delta-beta thalassaemia egg is fertilised by a normal sperm the child will carry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

What causes Mild Thalassaemia Intermedia?

Normally, red blood cells are produced in the bone marrow, live for about four months, and then are broken down and replaced by new red blood cells. People with two delta-beta thalassaemia genes cannot make haemoglobin A. However, their haemoglobin F genes are not fully switched off, and they make haemoglobin F instead. Their red blood cells contain *only* haemoglobin F. This functions almost normally but there is not quite enough of it. This causes a mild anaemia.



A normal red blood cell



A homozygous delta-beta thalassaemia red blood cell

What is haemoglobin F?

It is a special type of haemoglobin that unborn babies have. The "F" stands for *fetal* haemoglobin.

Haemoglobin F is different from haemoglobin A. It is produced by different genes. In normal babies the haemoglobin F genes work until birth. After birth they are gradually switched off and the genes for haemoglobin A are gradually switched on. As a result haemoglobin F disappears, and their blood contains maily haemoglobin A by about six months of age. This change is called the "fetal switch".

A delta-beta thalassaemia carrier's haemoglobin F genes do not switch off completely: as a result they make some haemoglobin F life-long.

When a person has homozygous delta-beta thalassaemia they make a large amount of haemoglobin F, life-long. This is why they have mild thalassaemia intermedia.

What is Mild Thalassaemia Intermedia?

People with mild thalassaemia intermedia do not need regular blood transfusions.

Children with mild thalassaemia intermedia grow up normally, and have a normal education. Adults with mild thalassaemia intermedia work normally, find a partner, and can have a family of their own.

A child with mild thalassaemia intermedia is normal at birth, but becomes slightly anaemic by one year of age. The anaemia may become a little more marked up to two to four years of age, but then their haemoglobin usually settles at a steady level.

People with mild thalassaemia intermedia may have some other health problems, but they can all be treated. For example:

- their spleen may enlarge when they are older. This can make their anaemia worse. It can be corrected by taking their spleen out.
- they may develop gallstones when they are adult, and these can cause pain. This can be corrected by removing their gall bladder
- when they are in their 30s or 40s their bones may become thin (this is called *osteoporosis*). This can cause pain. There are new medicines to prevent osteoporosis, if the change is detected early.

How long do people with mild thalassaemia intermedia live?

We expect them to live a normal length of life.

They should attend a specialist clinic for haemoglobin disorders every six months in childhood and every year in adult life, to make sure that any problems are detected and treated early.

What are the hopes for people with mild thalassaemia intermedia the future?

Research is presently going on on the following new developments.

- ***Increasing the amount of fetal haemoglobin in the blood***. If the haemoglobin F genes could be switched back on properly, the anaemia of thalassaemia intermedia would improve. A drug called *hydroxyurea* seems to switch some haemoglobin F back on, in some people with thalassaemia intermedia. DNA studies are necessary to identify people who might respond to this treatment.
- ***"Intra-uterine bone marrow transplantation"***. In the next 10 years or so, it may become possible to treat a fetus with thalassaemia intermedia during early pregnancy. This might make the anaemia milder, so that the child would be more like a carrier than a person with a serious form of thalassaemia.
- ***"Gene therapy"*** may become possible for thalassaemia in the next 20 years. However, it will probably be more complicated and expensive than it sounds.

Finding out if your Baby is Healthy or has Mild Thalassaemia Intermedia

Most "at risk" couples wish to know the answer to this question as early as possible.

Finding out when the baby is born

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. ***The only reliable method for diagnosing delta-beta thalassaemia in a new-born baby is by DNA analysis.***

Finding out during pregnancy

It is also possible to find out during pregnancy, through ***prenatal diagnosis***. This test can be done at any time after 11 weeks of pregnancy. If the baby does not have thalassaemia intermedia this is reassuring. If the baby has thalassaemia intermedia you can take further advice.

Couples who both carry delta-beta thalassaemia may decide not to have a prenatal diagnosis, because the condition is very mild and there is a small risk to the pregnancy.

However, if a couple wishes to have prenatal diagnosis to find out if their child has homozygous delta-beta thalassaemia or not, this is perfectly acceptable.

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 that does prenatal diagnosis for haemoglobin disorders. You can discuss your case with an expert who will answer your questions and help you make the right decisions for yourselves and your family.

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.