Information for couples where one partner carries Beta Thalassaemia and one carries Haemoglobin O Arab

Contacts for expert counselling centre

Couple "at risk" for Haemoglobin O Arab/ Beta Thalassaemia

	Name	**Date of Birth**
Ms		
Mr		

- ***One of you carries beta thalassaemia and one carries haemoglobin O Arab. This means that, as a couple, you are at risk for having children with haemoglobin O Arab/ 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 A2	Electro-phoresis	DNA data*

** this is essential information for all couples at risk for haemoglobin O Arab/ 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”?

One of you carries beta thalassaemia and the other carries haemoglobin O Arab. You are both healthy but you could have children with haemoglobin O Arab/ beta thalassaemia. This is why doctors call you an “at risk couple”. ***In your case this is not a good description, because haemoglobin O Arab/ beta thalassaemia is not a serious condition.***

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

Beta thalassaemia and haemoglobin O Arab 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.

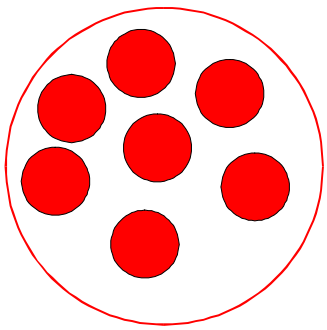
Haemoglobin is a component of the blood. It contains iron, which makes it bright red. This is why your blood is red. Your body needs oxygen to function: as your blood circulates haemoglobin picks up oxygen in your lungs and carries it round to all parts of your body. The usual type of haemoglobin is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

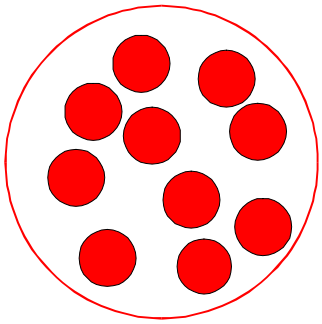
What is beta thalassaemia?

People who carry beta thalassaemia have smaller red blood cells than other people. They make up for having small red blood cells by making more of them. Their blood functions normally, and they are healthy people.

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



Usual Red Blood Cells



A Beta Thalassaemia Carrier’s Red Blood Cells

How do you find out you carry beta thalassaemia?

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

- they have small red blood cells.
- they have more than 3.5% of a haemoglobin called *haemoglobin A2* in their blood. People who do not carry beta thalassaemia have less than 3% of haemoglobin A2 in their blood.

What is haemoglobin O Arab?

Haemoglobin O Arab carriers have an unusual haemoglobin called haemoglobin O Arab, 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 O Arab?

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

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

They also need a special "DNA" test to confirm that a they carry haemoglobin O Arab.

How are beta thalassaemia and haemoglobin O Arab 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 beta thalassaemia carrier has inherited a gene for haemoglobin A from one parent and a gene for beta thalassaemia from the other. Their beta thalassaemia gene can make only a small amount of haemoglobin, or none at all. Their normal haemoglobin A gene makes enough haemoglobin for their red blood cells to function normally.

A haemoglobin O Arab carrier has inherited a gene for haemoglobin A from one parent and a gene for haemoglobin O Arab from the other. Their haemoglobin O Arab 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 beta thalassaemia and the other carries haemoglobin O Arab, a child could inherit both the beta thalassaemia gene and the haemoglobin O Arab gene. This child would have *haemoglobin O Arab/beta thalassaemia.* When a person has haemoglobin O Arab/beta thalassaemia neither of their haemoglobin genes functions normally, and their red blood cells contain less haemoglobin than usual. This causes a mild anaemia.

Couples where one partner carries beta thalassaemia and the other carries haemoglobin O Arab have the following chances ***in each pregnancy***

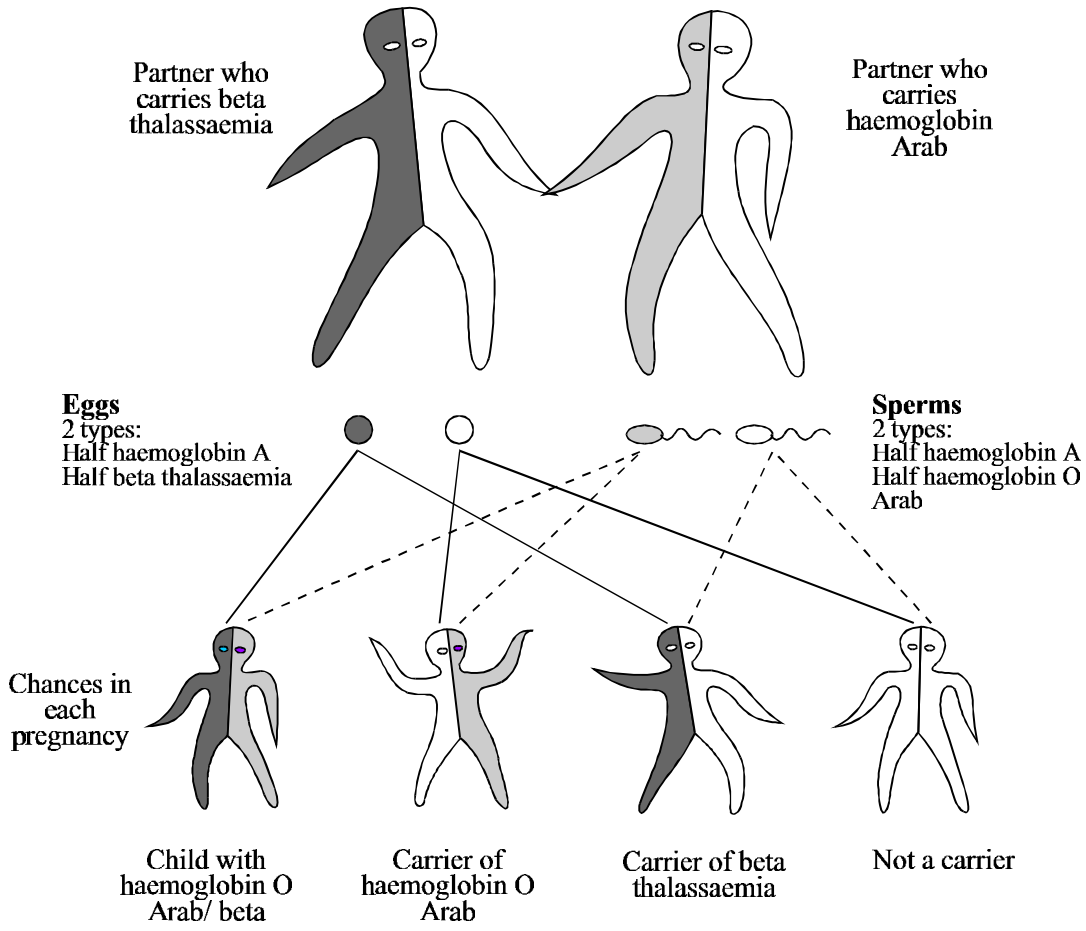
- a 1-in-4 chance of a child that is not a carrier.
- a 1-in-2 chance of a child that is a healthy carrier of beta thalassaemia or haemoglobin O Arab.
- a 1 in 4 chance of a child with haemoglobin O Arab/ beta thalassaemia.

In every pregnancy your baby is more likely to be normal or a carrer than to have haemoglobin O Arab/beta thalassaemia.

How could a child inherit Haemoglobin O Arab/ 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 beta thalassaemia and the other carries haemoglobin O Arab there are four possibilities.

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



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

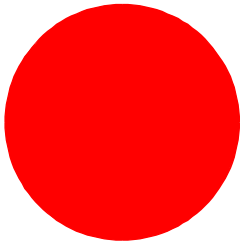
Men make sperm all the time. When a man carries haemoglobin O Arab, each sperm carries either his normal gene or his haemoglobin O Arab gene, but not both.

- 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 O Arab sperm the child will carry haemoglobin O Arab.
- If a beta thalassaemia egg is fertilised by a normal sperm the child will carry beta thalassaemia.
- If a beta thalassaemia egg is fertilised by a haemoglobin O Arab sperm, the child will have haemoglobin O Arab/ beta thalassaemia.

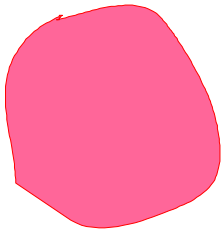
What is Haemoglobin O Arab/ Beta Thalassaemia?

Haemoglobin O Arab/ beta thalassaemia is very uncommon. The description given here is based on a handful of cases reported in the medical literature. ***The reports all suggest that haemoglobin O Arab/ beta thalassaemia is an extremely mild condition.***

People with haemoglobin O Arab/ beta thalassaemia cannot make haemoglobin quite as well as usual, so their red blood cells contain a little less haemoglobin than usual. They have a mild form of anaemia, called mild thalassaemia intermedia.



A normal red blood cell



A haemoglobin O Arab/ beta thalassaemia red blood cell

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.

Can you be sure that a child with haemoglobin O Arab/ beta thalassaemia will not have serious haemoglobin disorder?

Most known people with haemoglobin O Arab/beta thalassaemia have mild thalassaemia intermedia. Current scientific knowledge suggests that it is extremely unlikely that anyone with this condition will need blood transfusions or any other complicated treatment.

However, we cannot be completely sure that the anaemia will be mild in every case.

The partner who carries beta thalassaemia should have DNA studies to find out what type of thalassaemia they carry. A mild beta thalassaemia gene would certainly cause a mild thalassaemia intermedia. A severe beta thalassaemia gene might lead to a more serious anaemia.

Finding out whether or not your Baby has Haemoglobin O Arab/ Beta Thalassaemia

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

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 haemoglobin O Arab/ beta thalassaemia this is reassuring. If the baby has haemoglobin O Arab/ beta thalassaemia you can take further advice.

Couples at risk for haemoglobin O Arab/beta thalassaemia may to 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 haemoglobin O Arab/ 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.