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

Contacts for expert counselling centre

Couple at risk for Haemoglobin O Arab/Lepore

	Name	**Date of Birth**
Ms		
Mr		

- ***One of you carries haemoglobin O Arab and the other carries haemoglobin Lepore thalassaemia. This means that, as a couple, you could have children with haemoglobin O Arab/Lepore. 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 a serious form of thalassaemia*

Centre where tests were done

Address	
Telephone	Fax
Other	

Issued

Date
Signature of Doctor or Counsellor

Contents

Part 1.
What is an “At Risk Couple”?

One of you carries haemoglobin O Arab and the other carries haemoglobin Lepore thalassaemia. You are both healthy, but you could have children with thalassaemia intermedia. This is why you are called an “at risk couple”. Your blood test results are written in the front of this booklet.

Haemoglobin O Arab and haemoglobin Lepore thalassaemia 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.

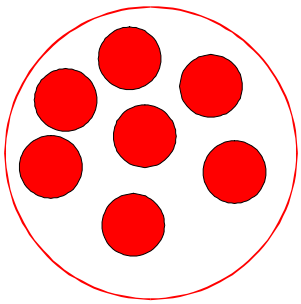
Haemoglobin is a component of the blood. It contains iron, which makes it bright red. This is why your blood is red. Your body needs oxygen to function: as your blood circulates haemoglobin picks up oxygen in your lungs and carries it round to all parts of your body. The usual type of haemoglobin is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

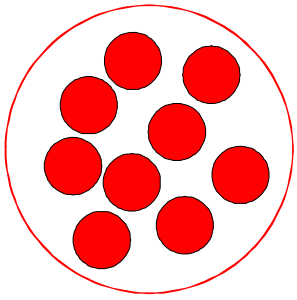
What is haemoglobin O Arab?

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.

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



Usual red blood cells



A haemoglobin O Arab carrier’s red blood cells

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.

What is haemoglobin Lepore thalassaemia?

Haemoglobin Lepore thalassaemia is an unusual kind of beta thalassaemia. People find out they carry haemoglobin Lepore thalassaemia through a haemoglobinopathy screen. This shows that:

- they have small red blood cells.
- they have between 7 and 15% of an unusual haemoglobin called *haemoglobin Lepore* in their blood. Most people have no haemoglobin Lepore.

They need a special "DNA" test to confirm that a they carry haemoglobin Lepore thalassaemia.

People who carry haemoglobin Lepore thalassaemia are often said to carry "haemoglobin Lepore", for short. Here we use the term haemoglobin Lepore.

How are haemoglobin O Arab and haemoglobin Lepore 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.* This is the usual type of haemoglobin.

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.

A haemoglobin Lepore carrier has inherited a gene for haemoglobin A from one parent and a gene for haemoglobin Lepore from the other. Their haemoglobin Lepore gene cannot make any haemoglobin A. Instead, it makes a small amount of haemoglobin Lepore. Their normal haemoglobin A gene makes enough haemoglobin for their red blood cells to function normally.

When one partner carries haemoglobin O Arab and the other carries haemoglobin Lepore, a child could inherit both the haemoglobin O Arab gene and the haemoglobin Lepore gene. This child would have *haemoglobin O Arab/Lepore.* When a person has haemoglobin O Arab/Lepore neither of their haemoglobin genes functions fully, and their red blood cells contain less haemoglobin than usual. This may cause a mild form of *thalassaemia intermedia*.

What are your chances of having children with Haemoglobin O Arab/Lepore?

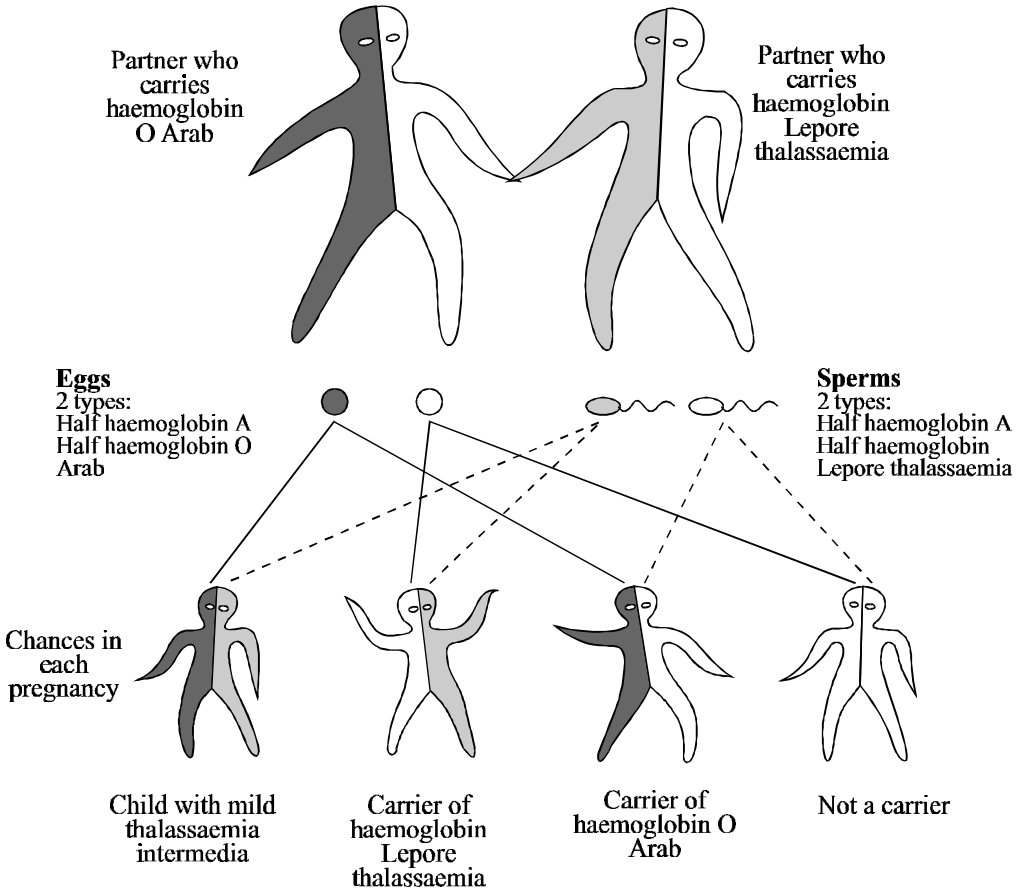
Couples where one partner carries haemoglobin O Arab and the other carries haemoglobin Lepore thalassaemia have the following chances ***in each pregnancy***

- 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 haemoglobin O Arab or haemoglobin Lepore.
- a 1 in 4 chance of a baby with haemoglobin O Arab/Lepore.

How could a child inherit Haemoglobin O Arab/Lepore from you?

When a child is conceived, it inherits one gene for haemoglobin from each parent. The picture shows that when one partner carries haemoglobin O Arab and the other carries haemoglobin Lepore there are four possibilities.

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



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

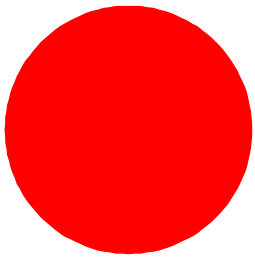
Men make sperm all the time. When a man carries haemoglobin Lepore thalassaemia, each sperm carries either his normal gene or his haemoglobin Lepore gene, but not both.

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

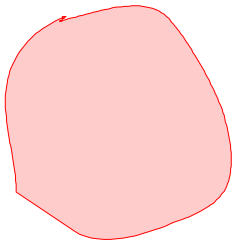
What causes Mild Thalassaemia Intermedia?

Haemoglobin O Arab/Lepore is very uncommon, and we do not know of any case reports in the medical literature. ***However, currrent scientific knowledge suggests that Haemoglobin O Arab/Lepore would cause only a very mild form of 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 one haemoglobin O Arab gene and one haemoglobin Lepore gene cannot make haemoglobin A normally, and their bone marrow cannot make red blood cells completely normally. They make mainly haemoglobin O Arab and haemoglobin Lepore. These haemoglobins function almost normally, but there is not quite enough of them. This can cause a mild anaemia.



A normal red blood cell



A thalassaemia intermedia 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.

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 haemoglobin O Arab/Lepore 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 could have children with haemoglobin O Arab/Lepore may decide not to have a prenatal diagnosis, because the condition is probably 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/Lepore 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.