Information for couples where one partner carries Haemoglobin S (sickle cell) and one carries Haemoglobin E

Contacts for expert counselling centre

Couple at risk for Haemoglobin S/E

	Name	Date of Birth
Ms		
Mr		

- ***One of you carries haemoglobin S and the other carries haemoglobin E. This means that, as a couple, you could have children with haemoglobin S/E . 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*

- *e.g. alpha thalassaemia mutation, Xmn I genotype.*

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 haemoglobin S (sickle cell) and the other carries haemoglobin E. You are both healthy, but you could have children with haemoglobin S/E**.** This is why doctors call you an *at risk couple*.

In your case this is not a good description, because haemoglobin S/E is not a serious condition.

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

Haemoglobin S and haemoglobin E are variations in the blood, of the 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.

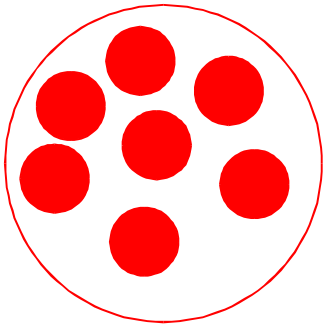
Haemoglobin is packed into *red blood cells*. Blood contains millions of red blood cells floating in a slightly yellow fluid called plasma.

What is haemoglobin S (sickle cell)?

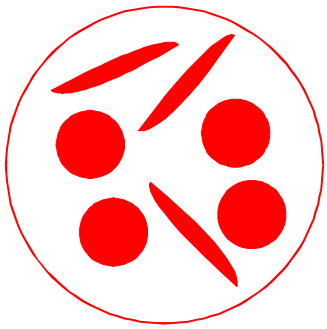
Haemoglobin S carriers have an unusual haemoglobin called haemoglobin S, as well as haemoglobin A. Their red blood cells contain approximately equal amounts of haemoglobin A and haemoglobin S. This is why they are sometimes said to "*be AS*". Their blood functions normally and they are healthy people.

Haemoglobin S is called *sickle cell haemoglobin* because red cells that contain it can sometimes change from their normal disk-like shape, to sickle-shaped (longer, curved and pointed). This change can cause health problems.

Carriers are healthy because their red blood cells contain haemoglobin A as well as haemoglobin S. Such red cells hardly ever sickle.



Usual red blood cells



Usual red blood cells with sickled red blood cells

How do people find out they carry haemoglobin S?

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

- they have an unusual haemoglobin in their blood, and that it is haemoglobin S.
- their red blood cells contain slightly more haemoglobin A than haemoglobin S.

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 haemoglobin S 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.

A haemoglobin S carrier has inherited a gene for haemoglobin A from one parent and a gene for haemoglobin S from the other. This is why they are sometimes said to "*be AS*".

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 haemoglobin S and the other carries haemoglobin E a child could inherit both a haemoglobin S gene and a haemoglobin E gene. This child would have *haemoglobin S/E*. When a person has haemoglobin S/E their red blood cells contain contain mainly haemoglobin S and a smaller amount of haemoglobin E. Very occasionally their red blood cells sickle, and this can cause health problems.

What are your chances of having children with haemoglobin S/E?

A couple where one partner carries haemoglobin S and the other carries haemoglobin E have the following chances ***in each pregnancy***

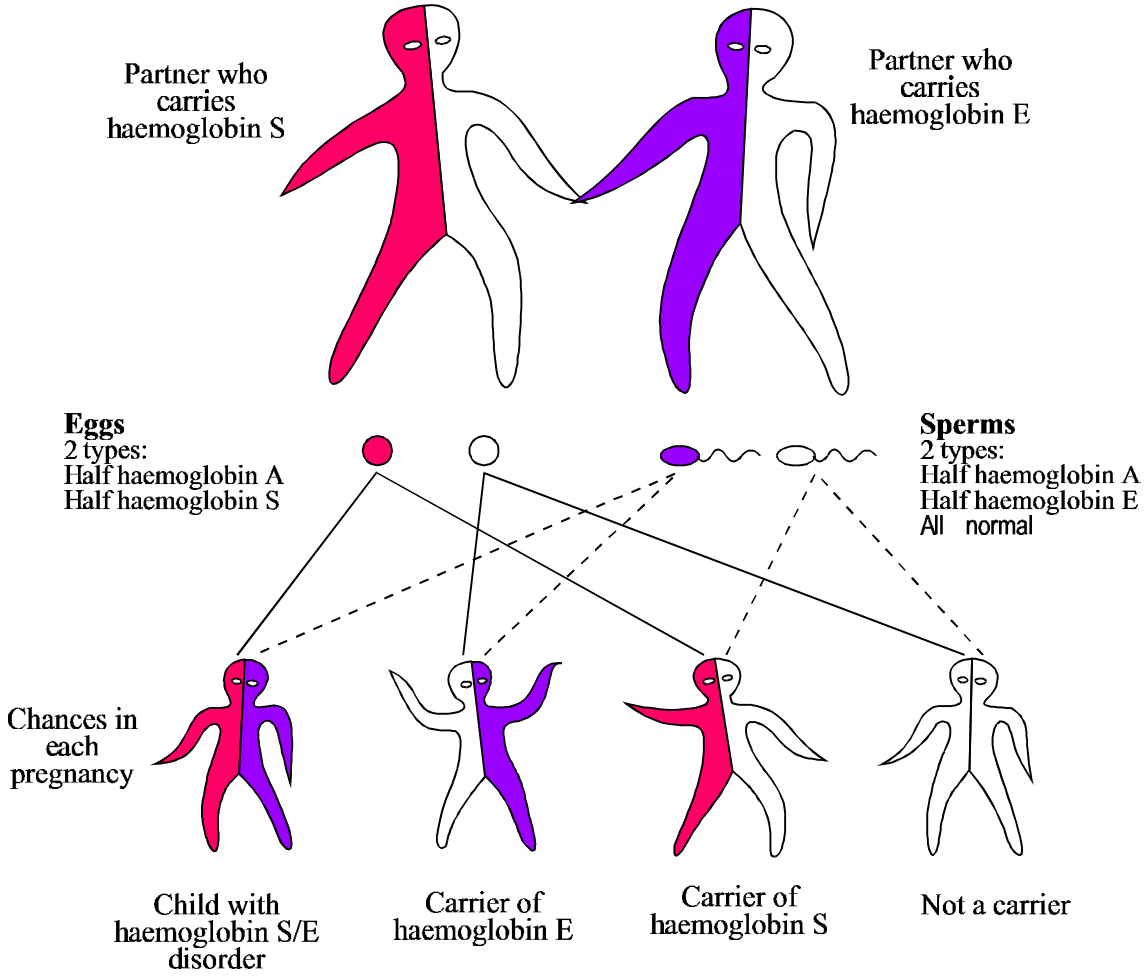
- a 1-in-4 chance of a child that is not a carrier.
- a 1-in-2 chance of a child that is a healthy carrier of haemoglobin S or E.
- a 1 in 4 risk of a child with haemoglobin S/E.

In every pregnancy your chance of having a child that is normal or a carrier is higher than your chance of having a child with haemoglobin S/E.

How could a Child inherit Haemoglobin S/E from you?

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

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



Women usually produce one egg each month. When the woman carries haemoglobin S, each egg contains either her normal haemoglobin gene or her haemoglobin S 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 haemoglobin S egg is fertilised by a normal sperm the child will carry haemoglobin S.
- If a haemoglobin S egg is fertilised by a haemoglobin E sperm, the child will have haemoglobin S/E.

What is Haemoglobin S/E?

Haemoglobin S/E 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 S/E is an extremely mild condition.***

Haemoglobin S/E is a type of *sickle cell disorder*. Most sickle cell disorders cause anaemia and an increased risk of serious infections, especially in childhood. There may also be painful crises and other problems.

In haemoglobin S/E there is usually no anaemia, or very mild anaemia. There is no evidence of an increased risk of serious infections or of any other problems in childhood. However, it may be wise for children with haemoglobin S/E to be given antibiotics regularly to protect them against the risk of infections.

Some adults with haemoglobin S/E have an enlarged spleen, or have joint pains. Probably most have slight thinning of the bones, called *osteoporosis.* A few have occasional painful crises. All these problems can be treated successfully.

Women with haemoglobin S/E who are pregnant should be looked after by an expert experienced in treating sickle cell disorders.

What is the life-expectancy in haemoglobin S/E?

We expect it to be normal. However, older people with haemoglobin S/E should see a specialist haematologist once every few years, for a check-up, to make sure that any possible problems are detected early and treated.

What can parents do, to make sure a child with haemoglobin S/E stays healthy?

When a couple carry haemoglobin S and haemoglobin E, it is usual to test each baby at birth to see if it has haemoglobin S/E of not. If a baby has haemoglobin S/E, the parents and child should attend a sickle cell clinic for the first few years. The baby may be given antibiotics to prevent infections. The parents can get more information from the clinic. If the child becomes ill, they can contact the clinic directly for advice.

Other types of Sickle Cell Disorder

It is important not to confuse haemoglobin S/E with other, more serious, types of sickle cell disorder. The other types of sickle cell disorder include:

- Sickle cell anaemia (haemoglobin SS)
- Haemoglobin S/beta thalassaemia
- Haemoglobin S/C disorder
- Haemoglobin S/D disorder

Sickle cell anaemia and haemoglobin S/D disorder are the severest sickle cell disorders. Some forms of haemoglobin S/beta thalassaemia are milder, and Haemoglobin S/C disorder can be very mild.

Finding out whether or not Your Baby has Haemoglobin S/E

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 best method for diagnosing haemoglobin S/E 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 haemoglobin S/E this is reassuring. If the baby has haemoglobin S/E you can take further advice.

We expect couples at risk for haemoglobin S/E to decide not to have a prenatal diagnosis, because there is a small risk to the pregnancy, and haemoglobin S/E is a very mild disorder.

However, if a couple wishes to have prenatal diagnosis to find out if their child has haemoglobin S/E 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 meet 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.