Carrying Haemoglobin S (sickle cell)

A carrier can use this booklet to…

- *help explain carrying haemoglobin S to their partner, blood relatives and others.*
- *show to any health professional (doctor, nurse or midwife) they see about having a family , or pregnancy, or carrying haemoglobin S.*

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Introduction

Haemoglobin S is one of a range of variations in the blood called *haemoglobin gene variants*. Here we call them *haemoglobin variants* for short.

Carriers of haemoglobin S are also sometimes said to *be AS*, or to *have sickle cell trait.*

Haemoglobin S is inherited. That is, it is passed on from parents to their children, like height, hair colour or eye colour. It is passed on equally by men and women. It is present at birth, and it remains the same for life.

It is important to distinguish clearly between people who *carry haemoglobin S* and people who *have a sickle cell disorde.*

Carriers of haemoglobin S…

… inherited haemoglobin S from one of their parents. Carrying haemoglobin S does not affect their own health. However, if their partner is also a carrier they could have children with a serious sickle cell disorder.

People who have a sickle cell disorder.…

… inherited two haemoglobin variants, one from each of their parents. Together, the two variants cause a serious inherited anaemia that can lead to life-long health problems.

This document is about carrying haemoglobin S.

What does it mean to carry haemoglobin S?

Blood is made up of millions of *red blood cells* floating in a fluid called *plasma*. Red blood cells are full of haemoglobin, which is red. This is why blood is red. The heart pumps blood round the body through the blood vessels. The body needs oxygen to function. Haemoglobin picks up oxygen as blood passes through the lungs, and carries it to the rest of the body as the blood circulates.

The usual type of haemoglobin is adult haemoglobin or haemoglobin A. Carriers of haemoglobin S have an unusual haemoglobin called haemoglobin S as well as haemoglobin A.

Haemoglobin S is *inherited* - it is handed on from parents to their children, it is present at birth, and it remains the same for life.

How is haemoglobin S inherited?

It is inherited through *genes.* Every human characteristic, such as eye colour, or height, or type of haemoglobin is controlled by genes that parents pass on to their children. A child inherits two genes for every characteristic, one from each parent. Most people inherit two genes for the usual type of haemoglobin, *haemoglobin A.* Each gene is responsible for making half of the haemoglobin A in each red blood cell.

A carrier of haemoglobin S has inherited a gene for haemoglobin A from one parent and a gene for haemoglobin S from the other. Each of their red blood cells contains both haemoglobin A and haemoglobin S. Haemoglobin S is only slightly different from haemoglobin A. Their blood functions normally, and carrying haemoglobin S does not cause them any health problems.

How do people find out that they carry haemoglobin S?

By having a special blood test “for haemoglobin disorders”. This usually involves two steps.

- The first step is to measure the size of their red blood cells. This shows nothing unusual.
- The second step is to analyse the types of haemoglobin in their blood. This shows that their blood contains haemoglobin S as well as haemoglobin A.

Can carrying haemoglobin S affect your health?

Carriers of haemoglobin S are not ill, and are no more likely to get ill than other people. Carrying haemoglobin S does not make them weak, and they can do any kind of work they choose.

There is a small risk that if a carrier gets extremely short of oxygen, they may have an attack of pain called a "sickle cell crisis". People can get short of oxygen, for example, by competing to hold their breath under water. Such activities should be avoided. People who carry haemoglobin S should also avoid extreme endurance exercises in very hot conditions.

Occasionally a carrier of haemoglobin S may see a little blood in their urine. Usually this is nothing to worry about and stops on its own. If it does happen, they should tell their doctor, to make sure that there is not a more serious cause.

When a carrier sees a health professional (doctor, dentist, nurse or midwife) they should tell them that they carry haemoglobin S, so that they can have appropriate health care. For example if they have an anaesthetic the medical staff need to know.

Is there any treatment to get rid of haemoglobin S?

No, a person who is born carrying haemoglobin S will always carry it.

Can carrying haemoglobin S turn into a serious haemoglobin disorder?

It cannot.

Can people catch haemoglobin S from a carrier?

They cannot.

Can a carrier of haemoglobin S be a blood donor?

They can give blood like other people, provided they are not anaemic (do not have a lower haemoglobin level than usual).

However, every unit of blood is now filtered for additional safety. The red blood cells of people who carry haemoglobin S may get stuck in the filter and block it. The blood transfusion service cannot use blood from people who carry haemoglobin S until this technical problem has been solved.

Is it a bad thing to carry haemoglobin S?

It is not. Carriers of haemoglobin S may be healthier than other people in some ways. For example, they have some natural protection against severe forms of malaria.

Malaria parasites live inside red blood cells, and are most comfortable in the red cells of people who do not carry any haemoglobin variant. People who carry haemoglobin S can be infected with malaria like anyone else. However, their red blood cells sickle when a malaria parasite settles inside them. Their spleen then removes the sickled cells from the circulation and kills the parasites. Therefore carriers have less severe infections and less chance of dying from malaria than other people.

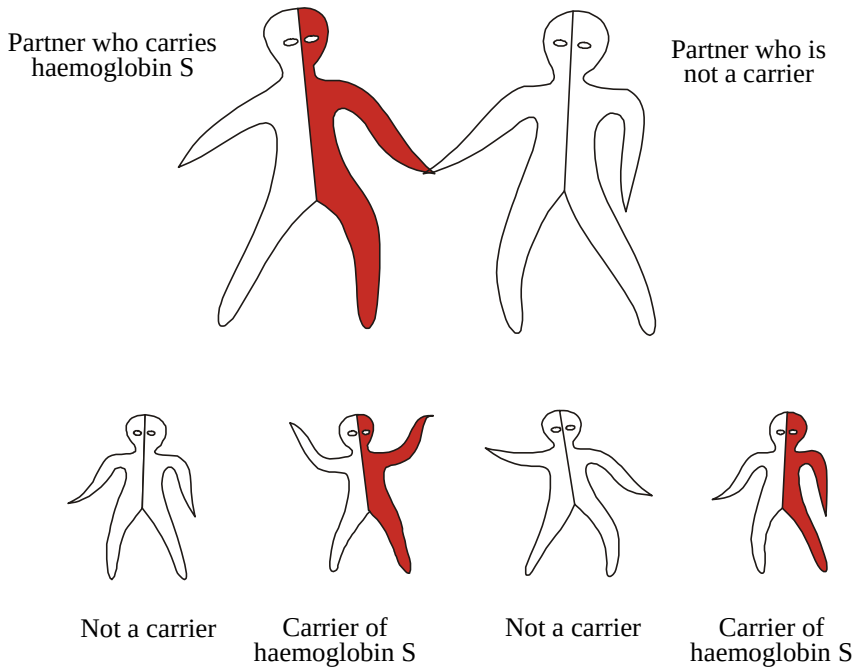
In countries where it was common malaria used to kill many children. Children who carried haemoglobin S survived better than other children and passed haemoglobin S on to their children in turn. As time passed carrying haemoglobin S became very common in such areas, and that is why there are now so many carriers in the world.

Malaria has been eradicated in many countries, so being a carrier is less advantageous than it used to be. Haemoglobin S does not go away when malaria is eradicated, or when a carrier moves to a different part of the world, because it is inherited.

Carriers of haemoglobin S should not rely on their natural protection against malaria when they visit a malarious country. Their protection is limited. They should take antimalarial tablets like everybody else.

Implications for a carrier's children…

If one partner carries haemoglobin S and the other does not carry any haemoglobin variant, their children could not have a serious haemoglobin disorder



In each pregnancy, there are two possibilities:

- The child may not carry any haemoglobin variant.
- The child may carry haemoglobin S. This is harmless.

The couple has the same chance of a healthy family as other couples do.

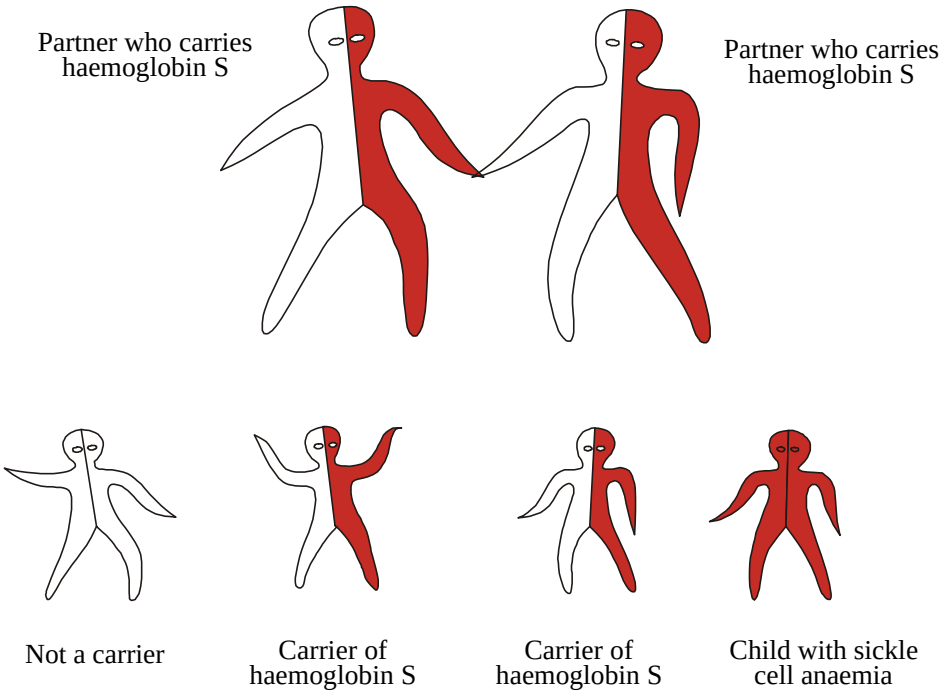
There is also no risk of a serious haemoglobin disorder if one partner carries haemoglobin S and the other carries any of the following:

alpha thalassaemia

haemoglobin D (not Punjab)
haemoglobin E
hereditary persistence of fetal haemoglobin (HPFH)

one of a range of possible rare haemoglobin variants

If both partners carry haemoglobin S, their children could have sickle cell anaemia



In each pregnancy there are three possibilities:

- The child may not carry any haemoglobin variant.
- The child may carry haemoglobin S. This is harmless.
- The child may inherit haemoglobin S from both parents. This child would have sickle cell anaemia.

In each pregnancy there is a 3 in 4 chance of a healthy child, and a 1 in 4 risk of child with sickle cell anaemia.

There is also a known risk of a sickle cell disorder when one partner carries haemoglobin S and the other carries any of the following:

beta thalassaemia (including unusual forms, such as delta-beta thalassaemia or haemoglobin Lepore)

haemoglobin C

haemoglobin D Punjab

haemoglobin O Arab

one of a few rare haemoglobin variants

What is sickle cell anaemia?

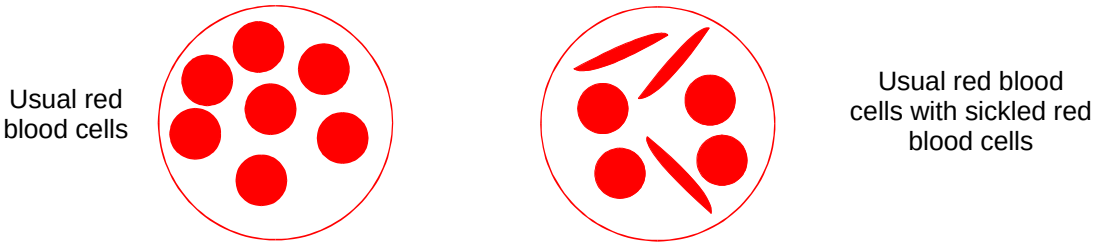
It is a *sickle cell disorder*. Sickle cell disorders can cause anaemia, infections, chest problems and painful crises. A painful crisis is an unpredictable attack of very severe pain, that can occur anywhere in the body. The hands and feet are often affected in young children, the limbs and back in adults.

Sickle cell anaemia can cause a range of serious problems, such as stroke in children, damage to bones, joints, eyesight or kidneys, and ankle ulcers in adults.

About one third of people with sickle cell anaemia have few medical problems. Most have two or three infections or painful crises a year, and may need to be admitted to hospital from time to time. About one in 20 have frequent serious problems, and may need regular transfusions to avoid organ damage. There is an increased risk of premature death, even for people with few other problems.

What causes sickle cell anaemia?

When a person has two genes for haemoglobin S their red blood cells contain mainly haemoglobin S. This is called *sickle cell haemoglobin* because it causes red blood cells to change from their normal disk-like shape to a sickle shape (long, curved and pointed), when they are short of oxygen. Sickled red blood cells can become jammed in small blood vessels, block the blood supply, and cause pain in that part of the body.



What is the treatment for sickle cell disorders?

People with a sickle cell disorder, and their family, need to understand the condition so that they can take steps to avoid problems as far as possible. For example they can reduce the risk of painful crises by drinking plenty of fluid and avoiding extremes of heat and cold. They should take penicillin daily to reduce the risk of infections, and their diet should contain adequate vitamins. They should attend a sickle cell clinic regularly so that problems can be detected and treated as early as possible.

They can have a wide range of other problems each requiring different, appropriate treatment.

Is it possible to predict how severe sickle cell anaemia would be?

It is not yet possible to predict how severe sickle cell anaemia will be in any individual case.

Can serious haemoglobin disorders be prevented?

Carrier couples who know of the risk for their children have a number of choices. They can take steps to make sure that they have healthy children, and can make sure that affected children have the best possible care from birth. Their choices are not simple. They need to know their risk early, so that they have enough time to make the decisions that are right for them.

In the UK, it is national policy to identify and inform as many carriers as possible before they have children, so that they can have an informed choice. The NHS is expected to take the following steps.

- *Offer carrier testing*. At present this is usually offered by midwives to pregnant women. In some districts it is offered to all pregnant women, and in others only to pregnant women with ancestors from areas where haemoglobin variants are common.
- *Inform carriers.* They need information on (a) the possible risk to the health of their children, and (b) the need for their partner to have a carrier test.
- *Inform carrier couples*. Couples who are both carriers need to see a specialist counsellor for haemoglobin disorders. The counsellor will find out whether they are "at risk" for having children with a serious haemoglobin disorder, and inform them of the exact nature of the risk and the possibilities for avoiding it.

All at risk couples should be offered both *prenatal diagnosis* and *neonatal diagnosis* (new-born diagnosis).

- Prenatal diagnosis means testing an unborn baby to see whether it has a serious disorder. DNA tests are done on a tiny amount of tissue taken from the developing placenta. This can be done as early as 11 weeks of pregnancy. If the baby is affected, the parents can decide whether to continue the pregnancy and plan the best possible care for the baby, or to terminate the pregnancy and try again for a healthy child.
- Neonatal diagnosis is done after the baby is born, using blood taken from the umbilical cord or by pricking the baby's heel a few days after birth. Neonatal diagnosis for thalassaemia is done by DNA tests. When there is a risk of a sickle cell disorder, early diagnosis and treatment can be life-saving for the child.

It is now recognised that screening during pregnancy often identifies at risk couples too late. Carrier testing should be offered by family doctors either before pregnancy, or as soon as a pregnancy has started.

Asking a partner to have a blood test

A carrier who is thinking of having children needs to tell their partner that they carry haemoglobin S, and ask him or her to have a blood test "for haemoglobin disorders".

Is it difficult for a carrier to ask their partner to have a blood test?

It can be easy in some cases and difficult in others. It is easier if both the carrier and their partner know that:

- Carrying a haemoglobin variant is common.
- The test will probably show that the partner does not carry a haemoglobin variant.
- If they do carry a haemoglobin variant it will not affect their health or lifestyle in any way.
- A couple who are both carriers can have a healthy family, with medical help.
- Expert counselling is available.
- No-one will try to tell the couple what to do: all the choices are theirs.
- The results of blood tests, and the couples' decisions, are completely confidential.

If a couple are both carriers, can it interfere with their relationship?

It is unusual for a relationship to suffer because one or both of a couple carry a haemoglobin variant. On the contrary, many couples draw closer together to deal with their problem. This is true whether they are just starting their relationship or have been together for a long time.

When is the best time for a carrier to ask their partner to have a blood test?

Ideally as early as possible, because it can take time to arrange a blood test, and a couple who are both carriers need time to decide what to do. Of course, the best timing depends on the couple's relationship with each other. It may also be influenced by their families' views, and the culture they belong to.

In practice a carrier might suggest a blood test to their partner at one of the following points.

- Before they settle down together.
- After they settle down together but before they start a pregnancy.
- As soon as they have started a pregnancy.

Telling the family about haemoglobin S

A carrier inherited haemoglobin S from one of their parents, so their brothers and sisters and other blood relatives could also be carriers. For example a brother or sister has a 1 in 2 chance of being a carrier.

If a carrier has brothers or sisters, or already has children, they need to know that they may carry haemoglobin S. They should ask their GP or practice nurse for a blood test "for haemoglobin disorders".

Haemoglobin S world-wide

- Sickle cell disorders are the commonest serious inherited disorders in the world.
- About one in 50 human beings carry haemoglobin S (2%). World-wide there are over 110 million carriers.
- World-wide about 260,000 children are born each year with sickle cell anaemia, haemoglobin S/C disorder or haemoglobin S/beta thalassaemia.
- In the UK there are about 145,000 carriers of haemoglobin S, and an estimated 9,000 people with sickle cell anaemia, haemoglobin S/C disorder or haemoglobin S/beta thalassaemia. (Estimates at the beginning of the year 2000.)
- Haemoglobin S is very common among people who originate from Africa. It is common in North and South America, the Caribbean area, the Middle East, India and parts of the Mediterranean area. It is uncommon among people who originate from Northern Europe.

The table shows the carrier frequency in selected population groups.

Population group and area of origin	**Frequency of haemoglobin S carriers**	**% of the population carrying haemoglobin S**
West and East Africa	1 in 4	25
Caribbean area	1 in 10	10
Middle East	1 in 1,000 to 1 in 4	0.1 - 25
India		
"caste" populations	1 in 1,000 to 1 in 50	0.1 - 2
tribal populations	1 in 8	12 - 16
Mediterranean area:		
Southern Portugal, Southern Italy, Sicily, Greece, Turkey	1 in 10 to 1 in 100	1 - 10

Carrying haemoglobin S

(also known as being AS, or having sickle cell trait)…

… is not an illness, and will never become an illness. It was passed to you by one of your parents and you could pass it on to your children.

There is nothing to worry about, unless your partner is also a carrier.

- If your partner is also a carrier, together you could have children with a serious inherited illness. However, with medical help, a couple who are both carriers can have healthy children.
- If you are thinking of having children, your partner should have a blood test “for haemoglobin disorders”.
- If you have children or brothers and sisters, they could carry haemoglobin S like you. Encourage them to have the same blood test.

To find out more, see your GP or a specialist counsellor.