

SICKLE CELL

A guide for GPs, nurses and other health professionals



INFORMATION, COUNSELLING
AND CARING FOR THOSE WITH
SICKLE CELL DISORDERS
AND THEIR FAMILIES

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What is this leaflet for?

This leaflet is intended to give you the information you need to recognise sickle cell disease and to help someone who has it to lead a good life.

How common is sickle cell and Thalassaemia

Sickle cell disorders are estimated to affect more than 1 in every 2,400 births in England. It is estimated that 240,000 are healthy carriers of the sickle gene variants and over 12,500 people have a sickle cell disorder. The highest prevalence of the sickle cell disorder is found amongst Black Caribbean, Black Africans and Black British.

The prevalence of Thalassaemia is less common. There are currently an estimated 214,000 healthy carriers of the beta Thalassaemia gene variant in England with over 700 people having beta Thalassaemia. Those with the highest prevalence of Thalassaemia are Cypriots, Italians, Greeks, Indians, Pakistanis, Bangladeshis, Chinese and other south East Asians group. Both genes are also (less commonly) found in the northern European population.

Who gets Sickle Cell Disease?

In Britain, sickle disease is more common in people of African or West Indian (Caribbean) descent. (At least 1 in 10 have sickle cell trait and 1 in 200 have sickle cell disease). It may also occur in people from the eastern Mediterranean, the Middle East, India and Pakistan. This geographical distribution has probably arisen because sickle cell trait offers some protection against malaria. (Sickle cell disease does **not** offer this protection).

What causes Sickle Cell Anaemia?

There are over 300 different types of haemoglobin. The most common type is haemoglobin A (Hb A) and most people inherit Hb A from both parents (Hb AA).

Sickle cell anaemia occurs when most of the haemoglobin in the red cells is **Sickle Haemoglobin** (Hb S). It is given this name because it causes the red blood cells to become sickle or crescent shaped when they give up oxygen.

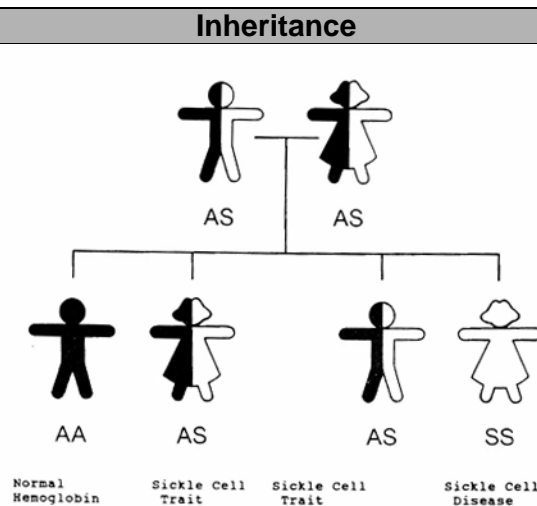


Micrograph of sickle cells in blood

What is Sickle Cell Trait?

Sickle cell trait occurs when one parent passes on the usual haemoglobin (Hb A) and the other parent passes on sickle haemoglobin (Hb S), resulting in Sickle Cell Trait (Hb AS). Sickle cell trait is **not an illness and cannot turn into sickle cell disease** (although very occasionally haematuria may occur).

However, if both parents are carriers of sickle cell trait, there is a **one in four chance** that each of their children could be born with sickle cell disease. You may find the following chart helpful when you explain how sickle haemoglobin is inherited.



Someone who has sickle cell trait (Hb AS) has a 50% chance of passing sickle haemoglobin (Hb S) on to each of their children and a 50% chance of passing on the usual haemoglobin (Hb A).

Someone who has sickle cell anaemia (Hb SS) passes sickle haemoglobin (Hb S) to each of their children.

If the child inherits sickle haemoglobin from both parents he or she will have sickle cell anaemia (Hb SS).

What are haemoglobin SC disease (Hb SC) and Sickle Beta-Thalassaemia (Hb SB-Thal)?

These conditions occur when someone inherits sickle haemoglobin (Hb S) from one of their parents and either haemoglobin C or Beta-Thalassaemia from the other parent. The symptoms of these two conditions are often similar to, but usually less severe than, those of sickle cell disease. People with Sickle Cell Anaemia, Sickle Beta-Thalassaemia and SC disease can sometimes have problems with sight or with thrombosis.

What are the symptoms of sickle cell disease?

Although sickle cell disease is present from birth, symptoms are rare before the age of three to six months, due to the persistence of foetal haemoglobin (Hb F). The main symptoms of sickle cell disease are episodes of anaemia, pain or infection. These are called **crises**. Some people get crises quite often; others may have them only once every few years. In between crises the sickler is usually quite well. Most serious acute complications occur during childhood.

People with SC disease and some with Sickle Beta-Thalassaemia may never experience a painful crisis but still develop chronic eye, bone or kidney problems.

People who only have sickle cell **trait** do not suffer any of the symptoms of sickle cell **disease**. Sickle cell trait is not an illness and people with trait are perfectly healthy. However, they require extra oxygen during anaesthetic and operation, and are advised against participating in some sports, such as scuba diving or climbing very high mountains, where the oxygen supply may become reduced.

How to recognise a sickle cell crisis

A crisis is a sudden onset of any of the following:

Pain

Sickle cell disease (Hb SS) sometimes causes attacks of pain to the chest, abdomen, back, jaw, legs and arms. They occur because the Sickling of the red blood cells causes them to block up small blood vessels and stop the flow of blood. The 'Sickling' of red blood cells which can cause a crisis is more likely to take place under certain conditions. These include a reduction of the level of oxygen in the blood (after exertion, during some anaesthetics or at very high altitudes), dehydration, and during pregnancy. Painful crises may also occur in association with febrile childhood or adult illness. If pain is very severe, admission to hospital may be necessary.

Anaemia

People with sickle cell disease are not always anaemic. In those with Sickle Cell Anaemia, however, the haemoglobins between 7 and 10 G/l. And the blood picture shows anisocytosis, sickle cells and a raised reticulocyte count due to haemolysis. The anaemia may become worse as a result of acute splenic sequestration or a plastic crisis. If so, emergency treatment with blood exchange transfusions may be necessary.

Infections

People with sickle cell disease are particularly prone to minor infections and also to serious and life-threatening infections like septicaemia, pneumococcal meningitis and osteomyelitis.

Other problems

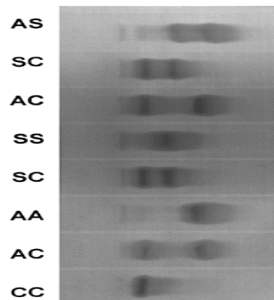
Sickle cell disease is a multi-system disorder. Children may get painful swelling of the hands and feet (called the hand-foot syndrome). They are also prone to enuresis and delayed puberty.

Adults (and sometimes children) can develop stiff and painful joints or ulcers on the lower legs. In general, people with sickle cell disease have an increased incidence of gallstones, jaundice, haematuria, strokes, priapism and difficulties during pregnancy and childbirth.

Diagnosis of Sickle Cell Anaemia and Sickle Cell Trait

The only method of diagnosing both sickle cell **disease** and **trait** is a specific blood test called **haemoglobin electrophoresis** which shows the haemoglobin type (e.g. AA, AS, AC, SS, SC etc..).

The sickle cell test is a simple solubility test which shows the presence of sickle haemoglobin but does not distinguish between sickle cell trait and sickle cell disease, and gives no information about the other abnormal haemoglobin. It should never be used on its own; whatever the result, sickle cell test must always be done with haemoglobin electrophoresis and blood count. Family studies and the measurement of Hb A₂ and Hb F are required for a complete and accurate diagnosis of sickle cell disease.



Haemoglobin Electrophoresis

When to screen for sickle cell trait and disease

Screening, with genetic counselling should be offered.

1. Prior to anaesthesia.
2. Prior to conception whenever possible, e.g. when discussing contraception or sexual health.
3. At antenatal booking clinics.

4. At birth.

Management of people who have sickle cell disease

(**Note:** This section does **not** refer to sickle cell trait).

Although there is no cure for sickle cell disease, doctors and other professionals can help to reduce the frequency and severity of crises and their complications by prompt recognition and treatment. Like any chronic illness, sickle cell disease is sometimes difficult to come to terms with. Practical and sympathetic advice can help affected families and individuals to cope with the inconvenience and painful effects of the condition.

Your treatment of sickle cell disease will obviously depend very much on the condition of the patient. In general, management of sickle cell anaemia falls into the following categories:

Steady State

Aim for the patient to live as normal a life as possible but be prepared to take immediate action if he or she becomes ill.

1. Maintain general health and nutrition. Make sure that the patient keeps warm and dry.
2. Avoid situations likely to precipitate Sickling e.g. dehydration, acidosis, general anaesthesia, and sports such as skydiving and scuba diving.
3. Treat infections early.
4. Arrange regular blood tests. These are needed for reference in a crisis.
5. Consider folic acid supplements.
6. All patients with sickle cell disease should be offered prophylactic penicillin.
7. Pneumococcal vaccination should be offered although it does not protect against all strains of pneumococci.
8. Ensure adequate malaria prophylaxis as sickle cell disease offers no protection – but see 9 below.
9. Beware of coexistent Glucose-6-phosphate dehydrogenase (G6PD) deficiency. There is a high incidence in the population groups prone to sickle cell disease. Acute haemolytic crisis may be provoked in sicklers by eating broad beans (fava) or by the administration of certain drugs including Anti malarial.

10. Offer genetic counselling.
11. Give the patient a haemoglobinopathy card or a letter giving details of his or her condition.

Crisis

Minor crises can be safely managed at home with pain killers and increased fluid intake. If the patient has high fever, severe pain requiring opiate analgesia, pain in the chest, spine or abdomen, or neurological signs, he or she should be admitted to hospital. Small children, who are always at risk from severe infections, should ideally be seen by a paediatrician with experience in sickle cell disease.

In hospital the following management is usually offered:

1. Immediate, usually opiate analgesia.
2. Intravenous fluids.
3. Prophylactic antibiotics.
4. Oxygen treatment is required in chest syndrome (Sickling in the blood vessels of the lungs).
5. Blood transfusion, often an exchange transfusion in the most severe crises.

Pregnancy

Pregnancy in sickle cell disease is a dangerous time for both the mother and the baby. Affected pregnant women should be looked after by a unit experienced in the care of women with this condition.

Blood transfusion may be needed in some women with poor obstetric history or a severe form of sickle cell disease. Regular folic acid, prompt treatment of infections and crisis, and an increased fluid intake make it possible for most women to have successful pregnancies.

Screening for sickle cell and Thalassaemia

The NHS and Thalassaemia screening Programme is the result of the government's commitment to improve the provision of screening services in England, as outlined in the 2000 'NHS Plan'. The NHS sickle cell and Thalassaemia screening programme is in the process of rolling out the screening programmes throughout England and all areas will be required to offer newborn screening by March 2005 and an antenatal screening programme for sickle cell and thalassaemia by March 2006.

What is the newborn screening policy

By March 2005 all the newborn babies are to be offered screening to detect sickle cell disorders. The newborn sickle cell screening is part of the existing bloodspot (formerly Guthrie) programme for phenylketonuria (PKU) and congenital hypothyroidism (CHT)

What is screening offered to newborn babies

Every year about 300 babies are born in England with sickle cell disorder. These babies are at high risk of death or complications, often arising from treatable infections, in the first few years of life. It is therefore important to offer screening via the bloodspot programme to identify babies with the condition so that treatment, including penicillin and pneumovax vaccine, can be started before 3 months of age.

What is screening offered to newborn babies

Screening in the antenatal period for sickle cell and thalassaemia will become an integral part of the screening package offered to all eligible pregnant women and couples by March 2006. The aim of the programme is to facilitate informed choices in screening, identify women/couples at risk of a pregnancy with sickle cell or thalassaemia disorders and provide appropriate referral and care for prenatal diagnosis with continuation of pregnancy or termination according to women's choice.

Thalassaemia screening using routine blood indices is to be offered to all women in England by March 2006. Screening for other haemoglobin variants will depend on the prevalence of the condition in particular geographical areas. Low prevalence trusts, as a minimum will be required to offer women laboratory testing based on an assessment of risk, which will be determined by the ethnic origin of the women and her partner. Universal laboratory screening will be offered to all women in high prevalence trusts.

GLOSSARY

Haemoglobin type and Description

Hb A	Usual haemoglobin, also called adult haemoglobin.
Hb AA	The inheritance of Hb A from both parents.
Hb S	Sickle Haemoglobin.
Hb AS	Sickle cell trait. The inheritance of Hb A from one parent and Hb S from the other parent.

- Hb SS** Sickle cell disease. The inheritance of Hb S from both parents.
- Hb AC** Hb C trait. The inheritance of Hb A from one parent and Hb C from the other parent.
- Hb SC** SC disease. The inheritance of Hb S from one parent and Hb C from the other parent.
- Hb CC** Hb C disease. The inheritance of Hb C from both parents.
- Hb AB-Thal** Beta-Thalassaemia trait. The inheritance of Hb A from one parent and B-Thal from the other parent.
- Hb SB-Thal** Sickle B-Thalassaemia. The inheritance of Hb A from one parent and B-Thal from the other parent.
- Hb A2 and Hb F** Haemoglobins usually only present in small amounts but present in increased amounts in Beta-Thalassaemia trait. Quantisation of these haemoglobin can therefore be useful in the diagnosis of this condition.

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