

# SICKLE CELL

A general guide to Sickle Cell Disorders



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

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## Introduction

This leaflet will explain the difference between sickle cell disorders (the illness) and sickle cell trait (the healthy carrier state) and other haemoglobin traits. First of all it might be useful to explain briefly the general term anaemia. ANAEMIA occurs when red blood cells cannot carry as much oxygen as normal, either because there are not enough of them or they are of poor quality. This can make a person feel tired, giddy and breathless after slight exertion. There are many causes and therefore the treatment will vary accordingly. For example, the most common type, iron-deficiency anaemia, can be treated by giving extra iron. **Iron-deficiency anaemia is not the same as sickle cell anaemia.**

## What Is Sickle Cell Disorder

Sickle Cell disorder (SCD) is the name for several related but different inherited disorders associated with sickling of the red blood cell. **Sickle Cell Anaemia** is the most common and best known type include:

### **Haemoglobin SC disease Sickle-Beta Thalassaemia**

People are often confused and disturbed by some of the incorrect information they have received about SCD.

SCD is not Leukaemia  
is not Cancer  
is not white blood cells eating up red blood cells  
is not infectious – you cannot ‘catch it’ as you would catch measles or a Cold.

Sickle Cell Disorder is a condition, which is inherited from both parents. This Means that people are born with it, just as they are born with other Characteristics such as eye colour, hair texture and height.

## Symptoms and Problems Associated with SCD

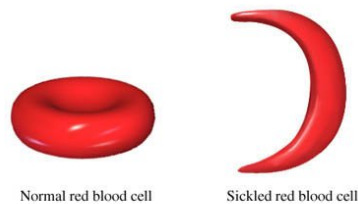
Symptoms very rarely start until the age of four to six months and these could include painful swelling of the hands and feet, infection and anaemia. The illness may cause frequent episodes of pain in the joints, abdomen and any other part of the body. These episodes of pain are commonly called **crises** and may be more frequently experienced because of dehydration, strenuous exercise, infection, pregnancy and anaesthesia. Other difficulties could include problems in the spleen, jaundice, strokes, leg ulcers, blood in the urine, eye, chest, hip and shoulder complaints and delay in growth. Admission to hospital may be necessary. It is important to stress that the symptoms and their effects can vary greatly from one individual to another, and also from time to time in a particular individual.

## Who Gets SCD

The different kinds of SCD and the different traits are found mainly in people whose families come from Africa, the Caribbean the Eastern Mediterranean, Middle East and Asia.\* In Britain SCD is most common in people of African and Caribbean descent (at least 1 in 10-40 have sickle cell trait and 1 in 60-200 have SCD). It is estimated there are over 15,000 adults and children with SCD in Britain at present. There are other inherited conditions that mainly affect other groups, e.g. Cystic Fibrosis in Europeans, and Tay-Sachs disease in Jewish people.

## How is the Blood Affected in a Person with SCD

The disorder affects the red blood cells, which contain a substance called haemoglobin (Hb for short). The function of haemoglobin is to carry oxygen from the lungs to all parts of the body. The type of haemoglobin a person has is determined by a pair of genes, one inherited from each parent (usually haemoglobin AA). In a person with sickle cell anaemia, the red blood cells contain only sickle haemoglobin (Hb S), instead of the usual haemoglobin A. This occurs when **both** parents have passed on a gene for sickle haemoglobin, i.e. (Hb SS). Red blood cells containing normal haemoglobin remain round when they give up oxygen. When a red blood cell containing mostly sickle haemoglobin gives up oxygen, it crystallises and changes its shape to a farmer's sickle or an irregular shape (hence the name sickle cell).



Because of their shape the sickle cells can clump together, preventing the normal flow of blood vessels. This may cause severe pain or damage various parts of the body. The other problem is that such red blood cells do not live as long as the normal 120 days and this results in the chronic state of anaemia. In spite of this, a person with sickle cell disorder can attend school, college and work. People with sickle cell disorder need regular medical attention particularly before and after operations, dental extraction and pregnancy.

Many hospitals arrange follow-up appointments and it is advisable to discuss with their doctors, questions concerning schooling, strenuous exercise, family planning, suitable types of employment and air travel. When a person is found to have sickle cell disorder it is important that all members of the family be tested. They will not necessarily have sickle cell disorder but may be healthy carriers of sickle cell trait.

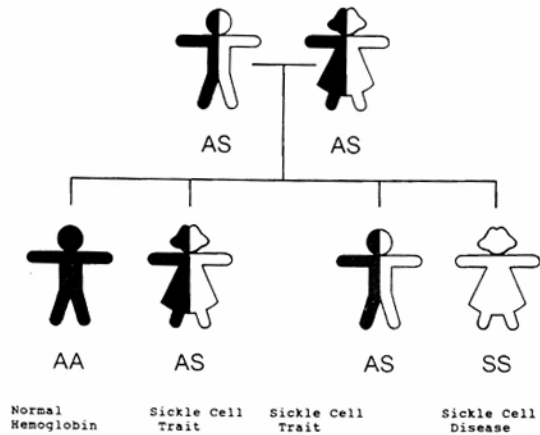
\* The geographical distribution has probably arisen because sickle cell trait (and probably Thalassaemia trait) may offer some protection against malaria.

The same is not true for sickle cell disorders. Always consult your doctor if travelling to areas where Malaria is endemic.

How are Sickle Cell Anaemia and Sickle Cell Trait Inherited?

With Sickle Cell, no one can guess which haemoglobin genes a child will inherit from his or her parents (unless both parents are HbAA or HbSS). However, if you know the types of haemoglobin you and your partner have, you will know the possibilities.

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 a sickle cell disorder. You may find the following diagram helpful when you explain how sickle haemoglobin is inherited.



Someone who has sickle cell trait (HbAS) has a 50% chance of passing sickle haemoglobin (HbS) on to each of their children and a 50% chance of passing the usual haemoglobin (HbA).

Someone who has sickle cell anaemia (HbSS) passes sickle haemoglobin (HbS) to each of their children.

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

What are haemoglobin SC disease (HbSC) and Sickle Beta-Thalassaemia (HbSB-thal)

This conditions occur when an individual inherits sickle haemoglobin (HbS) from one of there 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.

## Types of Haemoglobin

Haemoglobin is the protein molecule, which gives the red colour to blood. The most usual type is haemoglobin A (A stands for Adult). The kinds which are most important in sickle cell disorder are sickle haemoglobin and haemoglobin C. Thalassaemia, a condition in which the body cannot make enough haemoglobin, is also important. The type of haemoglobin a person has is determined by a pair of genes, one inherited from each parent.

**For example if you inherited two genes for the usual haemoglobin A (HbA you will have haemoglobin AA (HbAA)**

**If you inherited one gene for haemoglobin A) HbA) and one for sickle haemoglobin (HbS), you will have sickle cell trait (HbAS). Sickle cell trait is a healthy carrier state and will not make you feel ill.**

The other main types of trait are:

**Haemoglobin AC (C trait)**

**Haemoglobin a Beta-Thalassaemia (Beta-Thalassaemia trait)**

Both of these are healthy carrier states but individuals who carry either pair of genes can also pass one or the other of these genes onto their children.

**Sickle Cell Trait** occurs when a person inherits the usual haemoglobin (HbA) from one parent and a sickle haemoglobin gene (HbS) from the other (ie HbAS). Sickle Cell Trait is not an illness; it is not infectious and does not change to sickle cell anaemia. It is a silent, healthy carrier state and in fact offers some protection (in early childhood only) against one of the serious forms of malaria. Occasionally it may cause blood to appear in the urine.

There are other types of SCD such as 'S' with hereditary persistence of foetal Haemoglobin (S/HbFH), SD Punjab and SO Arab diseases. For further information on these, please contact the Society, nearest Sickle Cell & Thalassaemia Centre or local haematology department.

## Testing for SCD

A special blood test (haemoglobin electrophoresis) can tell you whether you have a sickle cell disorder or are a healthy carrier, e.g. for sickle cell trait. Routine screening should take place for people in the groups listed (see page 4) during pregnancy and before anaesthesia, either at hospital or dental clinics.

Tests can be arranged by your general practitioner or at our local sickle cell centre. (Contact us for an up-to-date list of centres in the UK). After a blood test you may be told that you or your child is 'sickle positive' or has 'sickle cell'. It is important to ask whether this means sickle cell trait or a type of sickle cell disorder.

## Testing unborn babies

It is now possible to test the unborn child to detect a haemoglobin disorder from as early as 11 weeks of pregnancy. For more information contact your local sickle cell centre, local obstetrician, haematologist or general practitioner.

## Can SCD be treated

Various forms of treatment that either prevent or relieve the symptoms are available for example; a person with a sickle cell disorder should always drink plenty of fluids and avoid over-exertion. When a person is admitted to hospital for a painful crisis they may also be given antibiotics for infection, fluids into the veins and occasionally a blood transfusion. If regular blood transfusions are given, **desferal** may also be given to remove excess iron from the patient's body. In order to prevent certain complications children are prescribed a daily dose of penicillin. Folic acid is prescribed to supplement the diet.

Iron tablets are not advised for sickle cell disorders unless special tests inform the doctor that they are necessary; in fact, iron tablets can do some damage if they are taken when they are not needed.

Research has improved the treatment now available, for instance, bone marrow transplants and Hydroxyurea are currently being evaluated, and however the search for a universal cure for sickle cell disorder continues.

**For further information on the work of the  
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