



**Screening for sickle cell and
thalassaemia in early pregnancy**

In the first few weeks of your pregnancy, we will offer you a blood test for sickle cell and thalassaemia.

This leaflet describes the screening process. It explains why we offer the test and helps you decide whether to accept it.

What are sickle cell and thalassaemia disorders?

Sickle cell disorder and thalassaemia major are serious, inherited blood disorders. They affect haemoglobin, a part of the blood that carries oxygen around the body. People who have these conditions will need specialist care throughout their lives.

Sickle cell disorders

People with sickle cell disorders:

- can have attacks of very severe pain
- can get serious, life-threatening infections
- are usually anaemic (which means that their bodies have difficulty carrying oxygen), and
- need medicines and injections when they are children and throughout the rest of their lives to prevent infections.

Thalassaemia major

People with thalassaemia major:

- are very anaemic (their bodies have difficulty carrying oxygen)
- need blood transfusions every four to six weeks, and
- need injections and medicines throughout their lives.

There are also other, less common, haemoglobin disorders. Many of these are not as serious.

How are they passed on?

Sickle cell and thalassaemia are inherited disorders that are passed on from parents to children through unusual haemoglobin genes.

People only have these disorders if they inherit **two** unusual haemoglobin genes – one from their mother, and one from their father. People who inherit just **one** unusual gene are known as 'carriers'. (Some people call this having a 'trait'.)

Carriers are healthy and do not have the disorders.

But if a carrier has a baby with someone else who is also a carrier (or who has one of the disorders), there is a chance that their baby could inherit a disorder.

Who can be a carrier?

Anyone can be a healthy carrier. But you are more likely to carry the unusual genes if your ancestors came from places where malaria has been common. This is because being a carrier can help to protect people against malaria.

This means you are more likely to be a carrier if your ancestors came from the Mediterranean (for example Cyprus, Italy, Portugal, Spain), Africa, the Caribbean, the Middle East, India, Pakistan, South America or south and south-east Asia.



"If you're discovered to be a carrier it doesn't affect your general health."

Father who came forward for testing

What tests are involved?

Screening involves a simple blood test. Ideally the best time to have the test is before you are 10 weeks pregnant.

All pregnant women are offered a blood test for thalassaemia. But you will not always be offered a blood test for sickle cell. You may be given a questionnaire to find out where your family – and the family of your baby's father – come from. If this shows you are at low risk, you may not be offered the blood test for sickle cell. But you can always ask for the test if you want it.

Why should I be tested?

The test gives important information for your baby's health

- If the blood test shows that you are a carrier, we will invite your baby's father for a test. If he is also a carrier, your baby has a chance of inheriting a disorder. (The diagram on page 3 shows the different chances for your baby. These include inheriting the disorder, being a carrier or not being affected.)

Finding this out early in your pregnancy gives you the chance to talk to a counsellor and find out more about the disorders and the care available. If you want to, you can have another test to confirm whether your baby has one of the disorders. (See 'Is there a further test?' on page 4).

The test can benefit you and your family

- If the test shows that you are a carrier, there is a chance that other members of your family could be carriers too. You may want to encourage them to ask for a test, especially if they are planning to have a baby themselves.
- Although people who carry sickle cell are healthy, they can experience some problems in rare situations where their bodies might not get enough oxygen (for example, when having an anaesthetic or during deep-sea diving). Knowing that you carry sickle cell can help you manage these situations.

However, people who carry thalassaemia or other unusual haemoglobin genes do not experience these problems.

For all of these reasons, we **strongly recommend** that you have screening. However, you can choose not to be tested, and we will respect your choice at all times.

Are there any risks?

Screening is a simple blood test, with almost no risk to you or your baby.

How will I get my results?

The person doing the test will discuss the arrangements for providing your results.

What will the results tell me?

The most likely result is that you are not a carrier. Your pregnancy should continue as normal.

If the result shows that you are a carrier for sickle cell, thalassaemia or another haemoglobin disorder, we will offer you counselling to talk about what this could mean for you, your baby and your family. We will also offer your baby's father a test to find out whether he is a carrier.

In very rare cases, the test may show that you have a haemoglobin disorder without knowing it. A health professional (for example, a nurse, doctor or midwife) will discuss your options with you, including the care you will need while you are pregnant.

Although the test is between 95% and 99% accurate, in a small number of cases the result may be unclear. If this happens, we will usually offer you another test.

Why should my baby's father have a test?

Babies can only inherit the disorders if **both parents** carry the unusual gene. So, if you are a carrier, it is important to find out whether the baby's father is also a carrier.

If he is not available or does not want to have a test, we may offer another test to find out whether your baby has sickle cell or thalassaemia. (See 'Is there a further test?' on page 4).

The diagram below shows the chances (for each pregnancy) of two carrier parents having a child with a sickle cell or thalassaemia disorder.

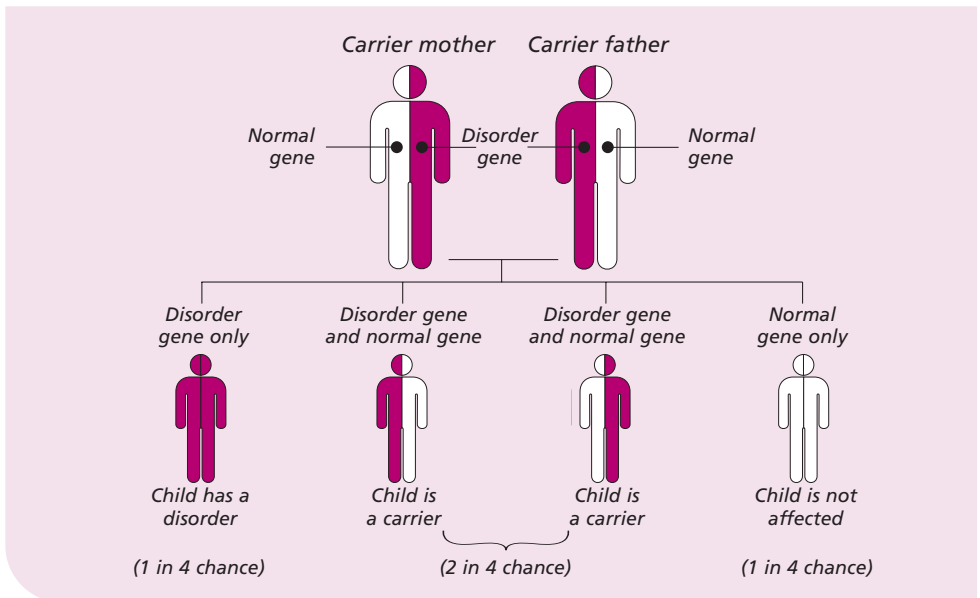
What if my baby's father is also a carrier?

If you and the baby's father both carry the gene for sickle cell, thalassaemia or another haemoglobin disorder, for each baby you have there is:

- a 25% (one in four) a chance that your baby will not be affected (that is, it will not have or carry a disorder)
- a 50% (two in four) chance that your baby will be a carrier, and
- a 25% (one in four) chance that your baby will have a disorder.

This is shown in the diagram below.

We will offer you counselling to discuss what this means for your family and what choices you want to make. If you want, you can choose to have another test to find out if your baby has sickle cell or thalassaemia or another haemoglobin disorder (see 'Is there a further test?' on page 4).



“The lady at the centre was really, really helpful. She was so nice and she talked us through everything and the chances of us having a sickle cell baby and everything. And then I made up my mind I was going to have those tests done...”

A pregnant woman who sought counselling about being a carrier



Is there a further test?

If both you and your baby's father are carriers – or if your baby's father does not come for testing – you can choose to have another test while you are pregnant. This is called a 'diagnostic test'. It will show whether your baby has a disorder.

A health professional will explain the different types of diagnostic test and help you decide whether you want the test. If you do want the test, it is important to have it as early as possible in your pregnancy.

If the test shows that your baby has sickle cell, thalassaemia or another haemoglobin disorder, we will offer you more counselling. This will help you to think about what this may mean for your family, the care that is available, and whether you want to continue with your pregnancy.

Testing for new babies

As well as the tests described in this leaflet, all babies in England are offered a heel-prick blood test when they are five to eight days old. This test is done by taking some blood from your baby's heel. It tests for a number of conditions including sickle cell. It will show whether your baby is not affected, is a carrier, or has a disorder. This is called newborn blood-spot screening and a health professional will give you more information later in your pregnancy.

Questions?

If you have any questions about the test, or anything else in this leaflet, please discuss them with your GP, midwife, hospital doctor or specialist counsellor. They will be able to give you advice. They may also have information about other organisations who can give you support. We have listed some of these on the next page.

More information

Visit the website of the NHS Sickle Cell and Thalassaemia Screening Programme at www.screening.nhs.uk/sickleandthal

Read about people's real-life experiences of sickle cell and thalassaemia screening like the ones shown in this leaflet. Visit www.dipex.org/sicklecellandthalassaemia

Other organisations

Sickle Cell Society

54 Station Road,
London NW10 4UA
Phone: 020 8961 7795
Helpline: 0800 001 5660
Email: info@sicklecellsociety.org
Website: www.sicklecellsociety.org

Sickle and Thalassaemia Association of Counsellors (STAC)

South West London Sickle Cell and Thalassaemia Centre,
Balham Health Centre,
120 Bedford Hill, Balham,
London SW12 9HP
Phone: 020 8700 0615
Email: info@stac.org
Website: www.stacuk.org

UK Thalassaemia Society

19 The Broadway, Southgate Circus,
London N14 6PH
Phone: 020 8882 0011
Freephone advice line:
0800 731 109
Email: office@ukts.org
Website: www.ukts.org



"I think people should definitely ask to have the screening because if both parents are carriers, and if the child does have the disorder, it can have very serious consequences. It would be better to know about your options in advance."

Sarah, who accepted screening when she became pregnant

Your local specialist centre or service is based at:



We aim to treat all records relating to antenatal screening for sickle cell, thalassaemia and other haemoglobin disorders in line with the Data Protection Act 1998.

This leaflet is based on high-quality research and evidence and the views of parents and health professionals. We would like to thank the many people who have been involved. It was produced by the NHS Sickle Cell and Thalassaemia Screening Programme in November 2006. From 2007 you can get this leaflet in other languages, on audio tape, in large print and in Braille. For more details, please see the programme website: www.screening.nhs.uk/sickleandthal

Cover Photo: Marcus Lyon



UK National Screening
Committee

