



Results of Newborn Blood-Spot screening

**Carrier of a sickle cell gene –
sometimes called trait
Hb AS**

Newborn Blood-Spot screening tests have shown that your baby carries a gene for sickle cell. Your baby does not have sickle cell disease and does not need medical treatment.

However, it is important that you read this leaflet. It contains information about what this result may mean in the future for your baby and for your family.

What is my baby's screening result?

When your baby was about a week old, a midwife took some blood from your baby's heel to test for rare conditions including sickle cell disease.

The test shows that your baby has not inherited sickle cell disease.

However, your baby has **inherited one gene for sickle cell**. We call this being a sickle cell carrier. Some people call it having sickle cell trait.

Carriers do not have sickle cell disease. But, they may pass on the gene for sickle cell to their own children – they can carry the gene to the next generation. It will be important for your child to know about being a carrier when they grow up and want to start a family of their own.

This leaflet explains about sickle cell, about being a carrier, and about how this might affect your family.



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

Father who came forward for testing

What action should I take?

We recommend that you:

- keep a copy of your baby's test result so that you can show it to health professionals such as anaesthetists (please ask your health visitor if you do not have a copy)
- write the result in your Child Health Record Book (sometimes called the red book)
- ask your GP or health visitor to write the result in your child's medical health record
- if you do not already know whether you are a carrier, arrange a blood test for yourself and your baby's father (this is especially important if you plan to have another baby)
- talk to other family members and explain why it is useful for them to know if they are carriers before starting a family.

What does it mean to be a sickle cell carrier?

We all have two copies of each gene in our bodies. We inherit one copy of each gene from our mother and one from our father. Genes are the information that our bodies use to shape the way we grow and function.

Some of these genes control haemoglobin – the substance in our blood that carries oxygen around our body. We write the usual haemoglobin as Hb AA. (Hb is the short way of writing haemoglobin).

People who have sickle cell disease have inherited **two** unusual genes for sickle cell – one from their mother

and one from their father. We write their haemoglobin in different ways depending on which genes they have inherited – for example Hb SS for sickle cell anaemia.

People who are carriers – like your baby – have inherited **one** gene that makes usual haemoglobin A and one gene that makes sickle haemoglobin S. So, we write their haemoglobin as Hb AS.

Because your baby makes some usual haemoglobin, they will never have sickle cell disease. They will always be a healthy carrier.

But, if they have a baby with someone else who also has an unusual gene, there is a chance that their child could inherit sickle cell disease.

“In future it's something to sit down and just go through... Just in case there's a health risk to future grandchildren. I'm glad, you know, I'm happy that we know now.”

The father of a baby identified as a carrier



How will being a carrier affect my child?

Your child's health

Your child is healthy and does not need any treatment.

However, people who carry sickle cell can experience problems in rare situations where their bodies might not get enough oxygen. The most important example is having a general anaesthetic. You should make sure that health professionals know that your child is a sickle cell carrier before they have an operation. You should check that a trained anaesthetist is looking after your child and avoid any operations where this is not offered – for example, some dental surgery.

Your child should also take extra care in situations where there might be a lack of oxygen such as deep-sea diving or high-altitude mountaineering. Apart from these very rare situations, your child can live completely normally.

Planning a family in the future

It is important that you tell your child about being a carrier. This will be valuable information when they grow up and want to start a family. Your child

will be able to explain about being a carrier and suggest a blood test for their partner. If the partner is also a carrier, there is a chance that their baby could inherit sickle cell disease. The diagram on page 4 shows the different chances of inheriting sickle cell disease.

How might the carrier result affect our family?

Your baby's test result means that:

- you or the baby's father is a sickle cell carrier (it could be the father or mother)
- both of you could be carriers
- one of you could have sickle cell disease, or
- one of you could be a carrier and the other have sickle cell disease.

If you do not already know whether you are a carrier and you are planning to have another baby, we strongly recommend that both you and the baby's father have a blood test to find out if you are carriers. This is because there is a chance that your next baby could inherit sickle cell disease. The diagram on page 4 shows the different chances of passing on the disease.

Your baby's test result also means that:

- other children in the family might be sickle cell carriers (or have sickle cell disease)
- other people in the wider family might be carriers (or have sickle cell disease).

We recommend that you talk about your baby's screening result with other members of your family, and encourage them to think about having a blood test to see if they are carriers.

As we have already explained, this is important if they are planning to have a family, and for health reasons – especially if they need to have a general anaesthetic.

If you want to arrange blood tests, please talk to your family doctor (GP) or local haemoglobinopathy service. (See the back page of this leaflet for details.)

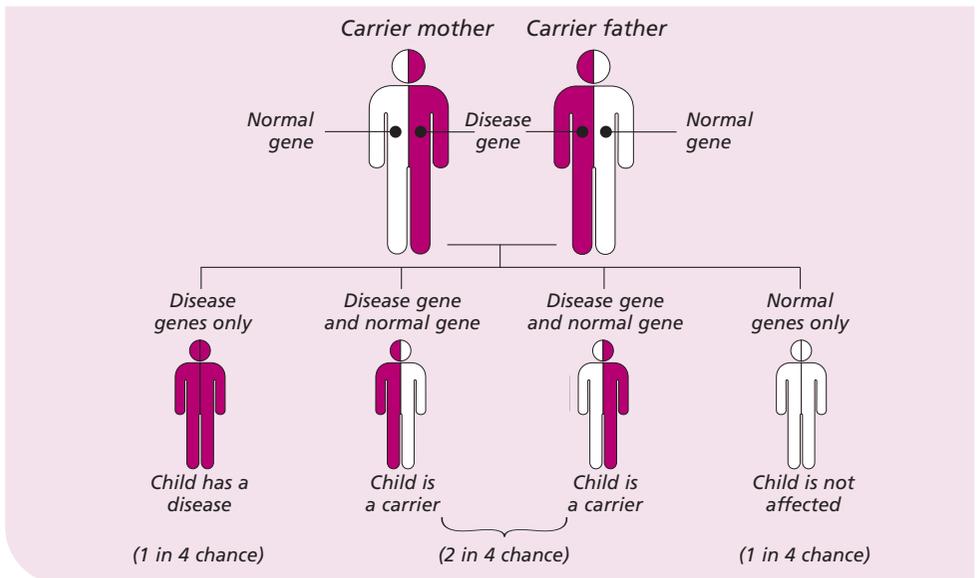
How do babies inherit sickle cell disease?

Babies can only inherit sickle cell disease if both parents pass on an unusual gene.

When a mother and a father both carry an unusual gene for sickle cell, every time they have a baby there is:

- a 25% (one in four) chance that their baby will not be a carrier or have sickle cell disease
- a 50% (two in four) chance that their baby will be a carrier
- a 25% (one in four) chance that their baby will have sickle cell disease.

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



"[The fact that our baby is a carrier] is just something that will just sort of sit in the background, up until either we decide to extend the family or we're waiting for grandchildren. And I think that's the only times that it will come up."

The mother of a baby identified as a carrier



Other questions that parents of carriers often ask

Can my baby develop sickle cell disease?

No – your baby will never have sickle cell disease. But they will always be a carrier.

Is being a carrier infectious?

No – you cannot catch sickle cell disease. You can only be a carrier if you inherit the gene from one of your parents.

Why is it important to tell my child about being a carrier?

It is important information for the future. Your child needs to know about being a carrier if they are planning to start a family because, as we have already explained above, their child could inherit sickle cell disease. Also, your child should take care when having a general anaesthetic.

Try to introduce the subject in a calm way and use opportunities that come up naturally – for example learning about blood at school. Do reassure your child that they are healthy and do not need any treatment.

Does being a carrier protect my child from malaria?

Carrying an unusual gene for sickle cell helps to protect people against malaria in childhood. This explains why the gene became common and why you are more likely to carry the sickle cell gene if your ancestors came from places where malaria has been widespread. These places include the countries around the Mediterranean (for example Cyprus, Turkey, Italy, Greece), Africa, the Caribbean, the Middle East, parts of India, Pakistan, South America or south and south-east Asia. But anyone can carry an unusual gene for sickle cell, and people who are carriers should still take precautions and medicines against malaria when they are travelling in countries where malaria is common.

Facts about sickle cell disease

Sickle cell disease is the name we give to a group of related conditions. These conditions affect haemoglobin – the substance in our blood that carries oxygen around our body.

Some sickle cell conditions are more serious than others. We call the most serious form sickle cell anaemia and write it as Hb SS. Other conditions that need treatment include Hb SC and Hb S beta thalassaemia.

People with sickle cell disease:

- can have attacks of severe pain
- can get serious, life-threatening infections
- are usually anaemic. (Their bodies can have less iron than usual because their red blood cells break down more quickly than normal red blood cells. This may cause difficulties in carrying oxygen)
- will need treatment and care throughout their lives.
- Sickle cell disease is the most common genetic disease in England
- There are at least 240,000 healthy sickle cell carriers in England
- Around 12,500 people live with sickle cell disease in England.

We are providing this for information only. Your baby does not have sickle cell disease.

Questions?

If you have questions about any of the information in this leaflet, please talk to your health visitor, GP or local haemoglobinopathy service. They may also know about other organisations which can give you information. We have listed some of these on the next page.

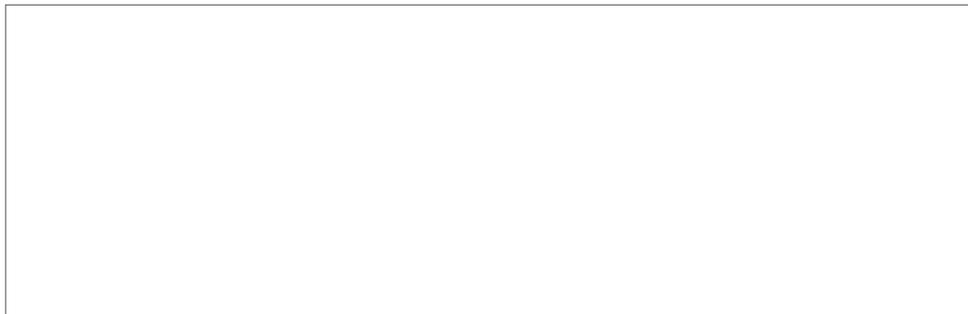
More information

The NHS Sickle Cell and Thalassaemia Screening Programme is developing more detailed information about carrying an unusual gene for sickle cell, sickle cell disease and the chances of babies inheriting sickle cell. Visit www.sickleandthal.org.uk.

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

Find out more detailed information about being a sickle cell carrier and sickle cell disease at www.chime.ucl.ac.uk/APoGI (click on 'APoGI' for haemoglobin gene variants).

Your local haemoglobinopathy service is based at:



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

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

This leaflet was produced by the NHS Sickle Cell and Thalassaemia Screening Programme in September 2007. We will provide versions of this leaflet in other languages and on audio tape, in large print and in Braille during 2007-2008. For more details, please see the programme website at www.sickleandthal.org.uk

Cover Photo: Marcus Lyon



UK National Screening
Committee

