Public Health England

NHS

NHS Sickle Cell and Thalassaemia Screening Programme

Information and choices for women and couples at risk of having a baby with sickle cell disease

Who is this leaflet for?

We have given you this leaflet because the baby you are expecting may have sickle cell disease.

We know this because your blood tests showed one of the following results.

1. You and the baby's father both have one unusual haemoglobin gene and one normal haemoglobin gene.
2. You have one unusual haemoglobin gene and we don't have test results for the baby's father.

Haemoglobin is a substance in the blood that carries oxygen around the body. People who have one unusual gene are known as carriers.

This leaflet explains:

- what it's like to live with sickle cell disease
- the chances of your baby inheriting sickle cell disease
- possible follow-up tests
- the choices available to you

This information should support, but not replace, the discussions you have with your healthcare professional. Your healthcare professional will support you throughout your choices.

Sickle cell disease

Sickle cell disease is the name for a group of conditions inherited from parents that affect the haemoglobin in red blood cells. The most serious type is called sickle cell anaemia.

In the UK, sickle cell disease is most common in people with an African or Caribbean family background.

People with sickle cell disease produce unusually shaped red blood cells. This can cause problems because they can block small blood vessels. In a person with sickle cell disease, the red blood cells don't last as long as healthy red blood cells.

Sickle cell disease is a serious and lifelong condition, but long-term treatment can help manage many of the symptoms. People with sickle cell disease can lead long, active and fulfilling lives if they manage their condition well and have the right care and support.

Symptoms

The main symptoms of sickle cell disease are:

- painful episodes called sickle cell crises, which can be very severe and last up to a week
- an increased risk of serious infections
- anaemia (a reduction in red blood cells so not enough oxygen is carried around the body), which can cause tiredness and shortness of breath

Other symptoms can include delayed growth, strokes and lung problems.

There are different types of sickle cell disease because there are different types of unusual haemoglobin gene. The most common and most serious sickle cell disease is sickle cell anaemia. Babies inherit sickle cell anaemia if both parents are sickle cell carriers.

If one parent is a sickle cell carrier and the other parent carries another unusual haemoglobin gene then their baby could inherit a different type of sickle cell disease.

Possible treatment

People with sickle cell disease need specialist care throughout their lives. Daily antibiotics and regular vaccinations can reduce the risk of infections. Some children with sickle cell disease benefit from taking a medicine called hydroxycarbamide which helps prevent many complications.

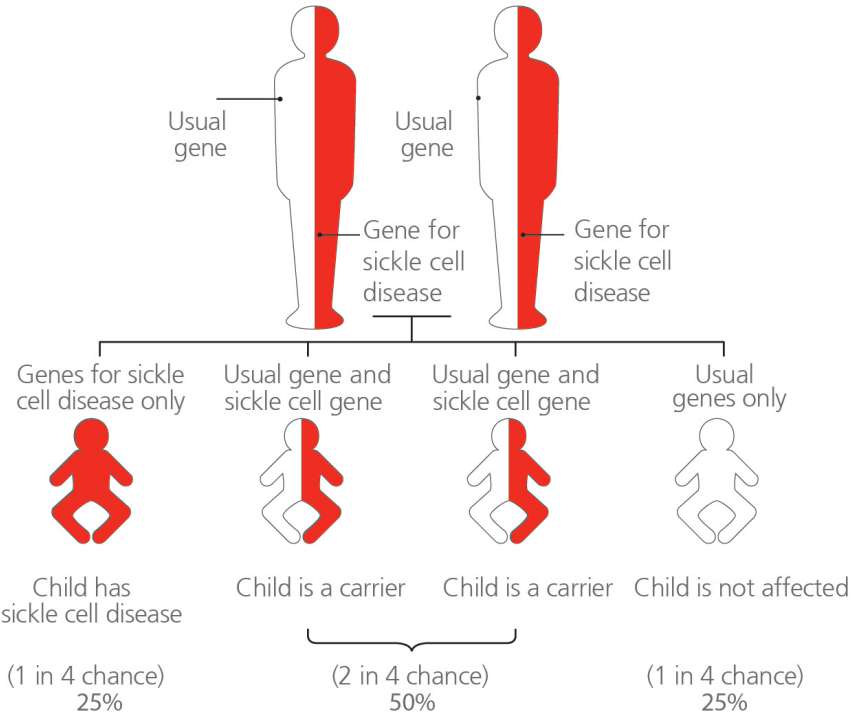
There are a number of things people with sickle cell disease can do to manage pain, avoid infections and stay as healthy as possible. Your healthcare professional can give you more advice about living with sickle cell disease.

Blood transfusions can be given to treat serious cases of anaemia. The only cure for sickle cell disease is a bone marrow (or stem cell) transplant, which replaces damaged blood cells with healthy ones. This is a complex and risky procedure which is only suitable for people with serious complications from sickle cell disease.

What are the chances your baby will inherit the disease?

If you and the baby's father are both carriers then there is a 1 in 4 (25%) chance the baby will inherit sickle cell disease. The diagram below shows how genetic inheritance works.

Both parents in the diagram are carriers for sickle cell disease. They are drawn in 2 colours to show they have one usual haemoglobin gene (white) and one unusual gene (red).



These chances are the same in each and every pregnancy when both parents are carriers.

Follow-up tests

You can choose if you want a test to find out for sure if your unborn baby has inherited sickle cell disease or not. This is called pre-natal diagnosis (PND). It is your decision to have this test or not.

If you decide not to have PND then your baby will still be offered the newborn blood spot test for sickle cell disease at 5 days old.

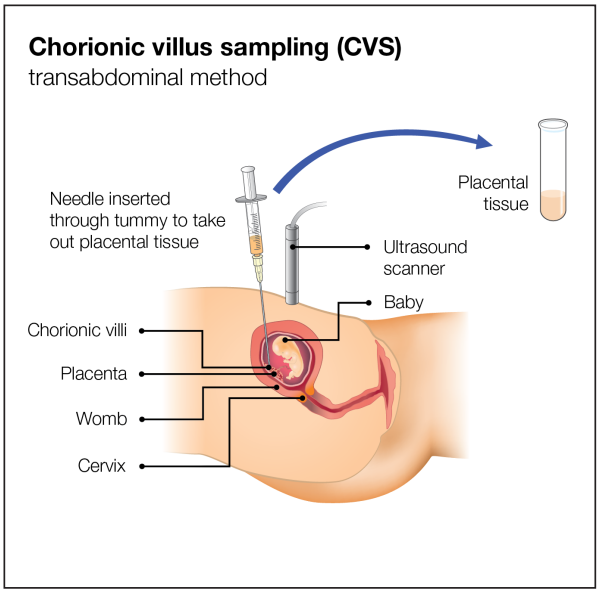
There are 2 main types of PND test depending on the stage of your pregnancy – **chorionic villus sampling** (CVS) and **amniocentesis**. Up to one out of every 100 women who have a CVS or amniocentesis will miscarry due to the test.

CVS is usually done from 11 to 14 weeks of pregnancy but can be done later. CVS can be performed in 2 ways:

- through the abdomen (tummy) – transabdominal
- through the cervix (neck of the womb) – transcervical

Amniocentesis is usually done between 15 and 20 weeks of pregnancy but can be done later.

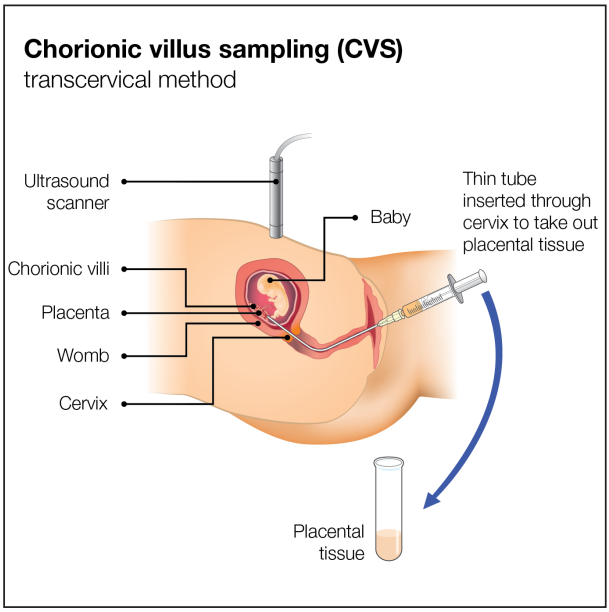
Transabdominal CVS



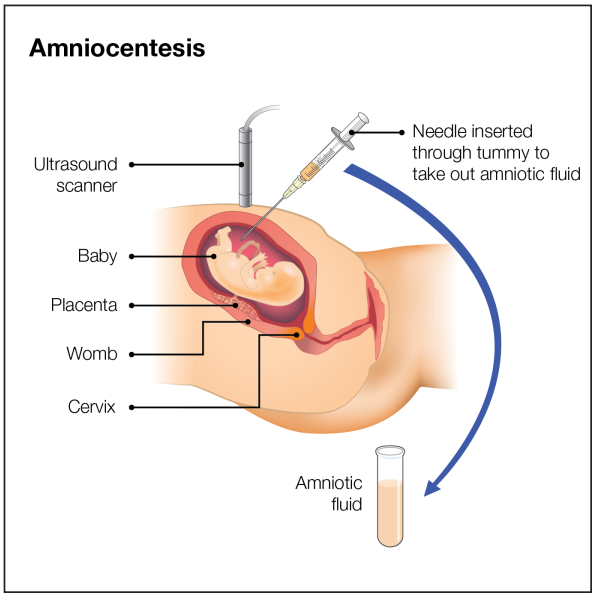
We clean the abdomen with antiseptic and may use a local anaesthetic injection to numb a small area. We insert a fine needle through the abdomen and into the uterus to take the sample. We use an ultrasound probe to guide the direction of the needle.

Transcervical CVS

We insert a thin tube attached to a syringe or small forceps through the vagina and cervix, and guide it towards the placenta using the ultrasound scan. We collect a tiny sample of placental tissue through the cervix.



Amniocentesis



We take a small amount of amniotic fluid (the water around the baby inside the uterus) for testing. The sample contains some of the baby's cells, which contain genetic information.

We clean the abdomen with antiseptic and may use a local anaesthetic injection to numb a small area. We insert a fine needle through the abdomen and into the uterus to take the sample. We use an ultrasound probe to guide the direction of the needle.

Occasionally, for **fewer than 7 in every 100** women, we cannot take enough fluid at the first attempt and have to re-insert the needle. This is usually due to the position of the baby.

If a second attempt fails, we will offer an appointment to have the amniocentesis again on another day.

After taking the needle out, the baby is observed for a short time on ultrasound.

Possible results and choices

There are 3 possible results from PND. Your baby could:

- **inherit sickle cell disease (1 in 4 or 25% chance)**
- **be a carrier of sickle cell disease (2 in 4 or 50% chance)**
- **have normal haemoglobin (1 in 4 or 25% chance)**

In rare cases the screening laboratory cannot give a result. If this happens, you will be contacted and offered a repeat PND test.

If the PND test shows your baby has normal haemoglobin or is a carrier then your pregnancy will continue as usual.

If the PND test shows the baby has inherited sickle cell disease, your healthcare professional will talk to you and offer support. You should also have the chance to talk to a specialist. You may choose to:

- **continue with your pregnancy**
- **end the pregnancy (have a termination)**

If you choose to **continue with the pregnancy** your healthcare professional will provide information and advice about caring for a child with sickle cell disease. The Sickle Cell Society provides support and information and may be able to put you in touch with groups and parents who have children with the same condition.

Your baby will be offered routine newborn blood spot screening (the heel prick test) at 5 days old, which should confirm sickle cell disease. You should get the newborn screening test result before your baby is 28 days old. Your baby will then be referred to a hospital specialist for treatment and care within 90 days of birth.

If you choose to **end the pregnancy** you will be given as much support as you need to help you come to terms with this difficult decision.

Future pregnancies

If you want to have another baby with the same partner and you are both carriers then the chances of the baby inheriting sickle cell disease will be exactly the same as now, 1 in 4 or 25%.

These chances are the same in each and every pregnancy when both parents are carriers. However, there are some choices you can make for any future pregnancies.

You can ask your GP, midwife or sickle cell specialist counsellor for PND early in the pregnancy and this can be performed after 11 weeks. This gives you more time to consider your choices if the baby has sickle cell disease. You'll need to see your GP or midwife as soon as you know you are pregnant.

You can consider pre-implantation genetic diagnosis (PGD). This means having an assisted pregnancy using in-vitro fertilisation (IVF). You can ask to see a genetic counsellor to discuss this option.

You can consider an assisted pregnancy using donated eggs or sperm from people who are not carriers. This means either you or your partner will not be the biological parent of your child. You can discuss this with your healthcare professional.

You can decide not to have any more children.

More information and support

NHS Choices: www.nhs.uk/sct

Sickle Cell Society: www.sicklecellsociety.org
E: info@sicklecellsociety.org T: 0208 9617795

Antenatal Results and Choices: www.arc-uk.org
E: info@arc-uk.org T: 0845 0772290; 020 77137356

Contact a Family: www.cafamily.org
E: info@cafamily.org.uk T: 0808 8083555

For information on how NHS screening programmes use patient information safely and securely, visit
www.gov.uk/phe/screening-data

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More information about sickle cell and thalassaemia screening: www.nhs.uk/sct

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