

Sgrinio Cyn Geni Cymru
Antenatal Screening Wales

Screening for sickle cell and thalassaemia in pregnancy

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This leaflet explains the tests you can have during pregnancy to find out if you are a carrier of sickle cell or thalassaemia. If you are a carrier, the father of your baby will also be offered testing. There are a number of different types of carrier. If you and the father of your baby are both carriers of 'important types', your baby could inherit a sickle cell disorder or thalassaemia major.

You can choose whether or not to have these tests.

If you or the father of your baby has been screened before for sickle cell and thalassaemia, and you carry a card showing your result, please tell your midwife and doctor. You may be offered the screening test again.



What are sickle cell disorders and thalassaemia major?

Sickle cell disorders and thalassaemia major are serious inherited blood conditions. They affect the haemoglobin in the red blood cells. Haemoglobin is important because it carries oxygen around the body. People who have these conditions will need specialist care throughout their lives. There are also other, less common haemoglobin disorders. Many of these are not as serious.

Sickle cell disorders

People with a sickle cell disorder can:

- have tissue and organ damage and varying degrees of symptoms;
- have attacks of severe pain where they need to stay in hospital; and
- be more prone to serious infections.

Beta thalassaemia major

People with beta thalassaemia major have:

- severe anaemia and need blood transfusions every four to six weeks as well as other treatments.

How are the disorders inherited?

Sickle cell and thalassaemia are genetic disorders. They are passed

on in families.

If both parents carry a sickle cell or thalassaemia gene, the baby may have a one in four, or 25% chance of having a sickle cell disorder or thalassaemia major.

If only one parent (either the mother or father) has the sickle cell or thalassaemia gene, it is very unlikely that their baby will have a sickle cell disorder or thalassaemia major. But the baby may be a carrier. That means that, like the mother or father, the baby will have the sickle cell or thalassaemia gene, but the gene does not usually cause problems. Women who are carriers sometimes have problems in pregnancy. For example, they are more likely to be anaemic.

Who can be affected?

Anyone can be a carrier of sickle cell or thalassaemia.

The chances of being a carrier of sickle cell or thalassaemia are higher for certain groups of people. This means you are more likely to be a carrier if your family, no matter how many generations back, come from the Mediterranean (for example, Cyprus, Italy, Portugal, Spain), Africa, the Caribbean, the Middle East, South Asia (for example, India, Pakistan, Bangladesh, Sri Lanka), South America or South East Asia (for example, Thailand, Vietnam, China).

The test

The test is a blood test which can be done with other blood tests, usually early in pregnancy.

The test is only offered to women with a higher chance of carrying sickle cell or thalassaemia. See 'Who is offered the test?' to see if you are in this group.

As part of your antenatal care, you will be offered a routine blood test (a full blood count) to check your haemoglobin level to see if you are anaemic. The full blood count can also find some types of thalassaemia.

The midwife will ask you if you would like to have thalassaemia screening as part of the full blood count test. If your full blood count test suggests that you might carry thalassaemia, the laboratory may also screen your blood for sickle cell disorders and thalassaemia.

Who is offered the test?

You should be offered the test if:

- you or your partner has a family history of sickle cell or thalassaemia;
- you, your partner, anyone in your family, or anyone in your partner's family, no matter how many generations back, came from anywhere in the world apart from the UK and the Republic of Ireland;
- you or your partner do not know your family history – for example,

you or your partner were adopted; or

- you have unexplained anaemia.

What are the advantages of having screening for sickle cell and thalassaemia in pregnancy?

If you are a carrier of sickle cell or thalassaemia, it is important to know so you can have the right kind of care during your pregnancy.

Women who know their baby has a high chance of inheriting a sickle cell disorder or thalassaemia major can have a diagnostic test to find out if the baby is affected. This could be done by either chorionic villus sampling (CVS) or amniocentesis. If the baby is affected, the woman can decide whether to continue with the pregnancy or to end it.

What are the disadvantages of having screening for sickle cell and thalassaemia in pregnancy?

Having the test may make you anxious if you find out you carry sickle cell or thalassaemia. Some women would be offered a diagnostic test to see if the baby is affected. Because the diagnostic tests can cause a miscarriage, many women find this a difficult decision. Some women may wish they had not had the screening test because making this decision is difficult.

Should I have the blood test for sickle cell and thalassaemia?

Only you can decide to have the test or not. Some women want to find out if their baby has sickle cell or thalassaemia, and some do not. Having the test may cause anxiety as the result may mean that you are offered further tests.

Where will the blood test be done?

Your midwife will tell you where you can have the test done.

Results

Will my results be confidential?

The NHS keeps the results of all tests confidential. Hospital policies vary on how many people in the NHS have access to your test results. Your midwife will be able to explain the local arrangements to you.

How will I get the result from my screening test?

Your midwife, doctor, or counsellor from the local sickle cell and thalassaemia centre will give you your test result.

What will the results tell me?

If the result shows you are not a carrier, it is very unlikely your baby could have a sickle cell disorder or

thalassaemia major. Although the test is very accurate, a small number of results may be unclear. If this happens, you will be offered another test.

If the test shows you are a carrier or a possible carrier, you will be able to talk to a specialist nurse or doctor and they will give you more information. They will suggest you ask the father of your baby to have a blood test to find out if he is a carrier. If his test result shows he is not a carrier, it is very unlikely your baby will have a sickle cell disorder or thalassaemia major.

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

If the test shows the father of your baby is a carrier, there may be a one in four (or 25%) chance your baby could have a sickle cell disorder or thalassaemia major. You can then decide whether to have more tests to find out if your baby is affected. These tests are called diagnostic tests. If you choose not to have more tests, your baby can be tested at birth for sickle cell disorders or for thalassaemia major. This means that if he or she is affected, treatment can start early.

What are the diagnostic tests?

Chorionic villus sampling (CVS) and amniocentesis

CVS and amniocentesis are diagnostic tests. They are accurate ways of testing for sickle cell disorders and thalassaemia major. If you decide to have a diagnostic test, you and the father of the baby will be asked to give another blood sample to help the laboratory give an accurate diagnosis.

What is a CVS?

CVS is a procedure during which a doctor removes a small amount of tissue from your placenta (afterbirth) during the pregnancy. The cells in this tissue are tested in the laboratory to look at your baby's chromosomes. You can usually have CVS from week 11 to week 14 of your pregnancy. However, in special circumstances you can have it done after 14 weeks.

If you have CVS, there is a 2% risk that you could have a miscarriage (that means one in every 50 women who has the test could lose their baby).

What is an amniocentesis?

An amniocentesis is a procedure to remove 15 to 20 millilitres (about three to four teaspoons) of amniotic fluid from around the baby in the womb. The cells from your baby that are floating in this fluid can be tested in the laboratory.

It can be done after the 15th week of your pregnancy.

Amniocentesis involves some risk. It causes a miscarriage in about one in 100 pregnancies (1%).

What are the possible results from diagnostic tests?

If you have CVS or amniocentesis, the result may show:

- that your baby does not have this problem; or
- your baby has a sickle cell disorder or thalassaemia major. You can then decide whether to prepare for the birth of a baby with sickle cell or thalassaemia major or to end your pregnancy.

CVS and amniocentesis can detect other chromosome abnormalities and you can get more information in the CVS and amniocentesis leaflet.

More information

You can get more information about sickle cell and thalassaemia from your midwife or your hospital doctor (your obstetrician).

Other organisations

Sickle Cell Society

Website: www.sicklecellsociety.org

UK Thalassaemia Society

Website: www.ukts.org