Screening for sickle cell disease and thalassaemia

An **easy guide** to screening tests when you are pregnant

Inside this booklet

Your choice

This booklet has information about a test you can have.

The test can find out if your baby may have sickle cell disease or thalassaemia. These are serious conditions.

The test can find problems with your baby before he or she is born. The test may not find every problem.

If we find a problem early we can give you information or tell you about treatment for you or your baby.

You can choose if you want to have the test.

The test can show if you or your baby might need extra care.

The test can help you make choices about your care when you are pregnant.

If you are worried, you can talk to your midwife or doctor.

If you say no to the test, your baby can have a test for sickle cell disease when they are born.

About this test

We only show the results of the tests to health staff who need to see them.

It is important to tell your midwife if you or any of your children have sickle cell disease or thalassaemia.

It is also important to tell your midwife if your baby's father or any of his children has one of the conditions.

You will be asked questions about which country you and your baby's father come from.

You will be asked other questions to help health staff decide if you need a test and to do the test properly.

All women are offered screening for sickle cell and thalassaemia.

This test takes a small bit of your blood to find out if you might pass on sickle cell disease or thalassaemia to your baby.

You can have the blood test at any time but it is best to have it with your other blood tests in early pregnancy.

Sickle cell disease and thalassaemia

Sickle cell disease and thalassaemia are serious blood conditions.

You get them from your parents. You have them for life.

Sickle cell disease can cause anaemia. Anaemia means you have problems carrying oxygen in your blood. This makes you pale, very tired and weak.

Sickle cell disease can give you very bad pain and very serious illnesses.
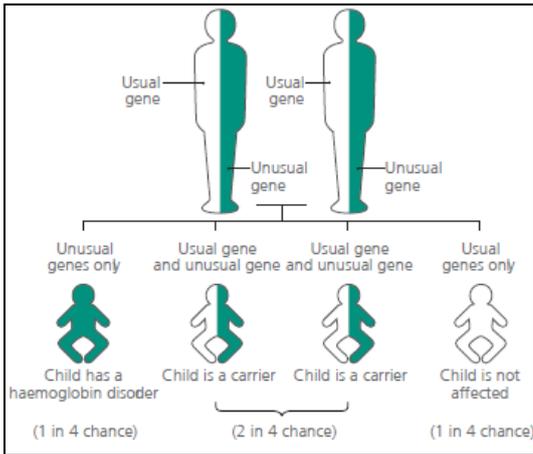
A baby with sickle cell disease can have medicines after they are born. Medicines will help them to be healthy.

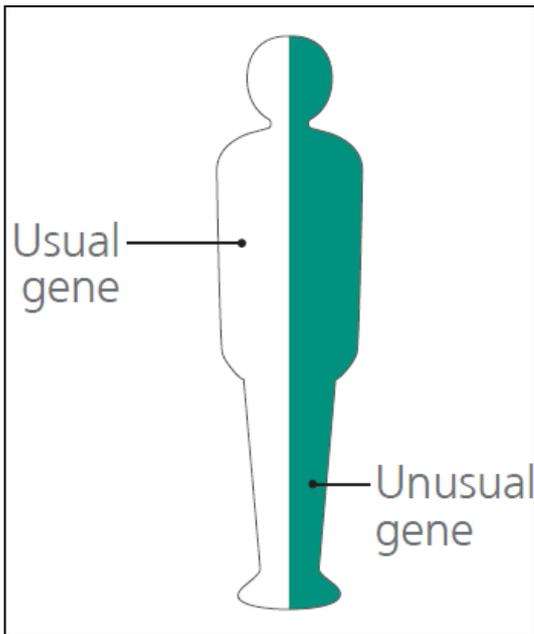Thalassaemia gives you very serious anaemia.

If you have thalassaemia you need blood transfusions every 4 to 6 weeks and you need medicine for life.

A blood transfusion is when blood is taken from a healthy person and put into an ill person.

Sickle cell disease and thalassaemia are passed on through genes.

Genes are like a code for your body.

People who have just one unusual gene are known as carriers.

Carriers are healthy and do not have the blood condition.

If you and the baby's father are carriers, your baby could be born with the condition.

Unusual
genes only

Child has a
haemoglobin disoder

(1 in 4 chance)

If you and the baby's father are both carriers, the baby has a:

- 1 in 4 chance of having sickle cell disease or thalassaemia

Usual gene
and unusual gene

Usual gene
and unusual gene

Child is a carrier   Child is a carrier

(2 in 4 chance)

- 2 in 4 chance of also being a carrier – this means they may then pass the gene on to their children when they are an adult

Usual
genes only

Child is not
affected

(1 in 4 chance)

- 1 in 4 chance of not having sickle cell disease or thalassaemia

Anyone may have this gene. But it is more common for people whose family comes from:

- Africa

- the Caribbean

- the Mediterranean

- South America

- South and South East Asia, or

- the Middle East.

Test results

You will usually get the results at your next appointment.

If the test shows you are a carrier for sickle cell disease or thalassaemia we will contact you.

If you are a carrier and have one of the unusual genes, it is really important for your baby's father to get tested too.

If the baby's father also has one of the unusual genes, you will be offered a second test. This is called an invasive test.
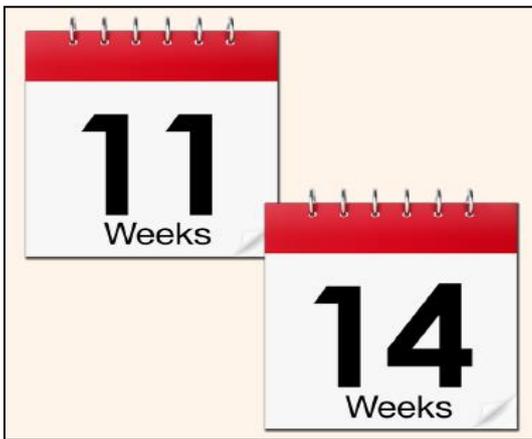
The invasive test has a small chance of about 1 in 200 of making you miscarry.

If you miscarry, the baby will not live.

You will be able to talk about the test with a midwife.

You can say no to this test.

There are 2 types of invasive test.

If you are between 11 and 14 weeks pregnant you can have a chorionic villus sampling test.

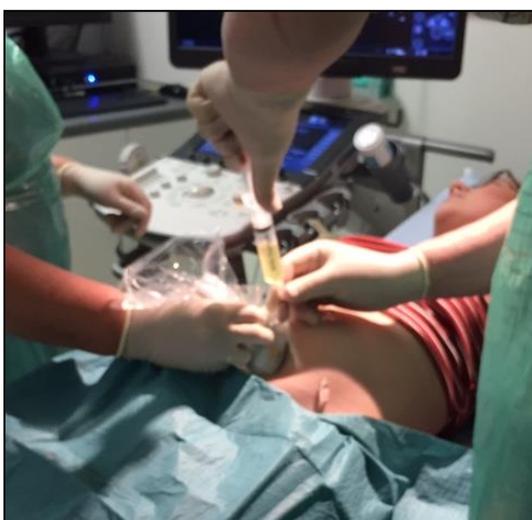This is sometimes called a CVS test.

If you have a CVS test a very thin needle is put into your tummy.

This takes away a tiny bit of the placenta. The placenta links you to the baby.

We can test this for sickle cell disease or thalassaemia.

If you are 15 weeks pregnant (or more) you can have an amniocentesis.

If you have an amniocentesis a very thin needle is put into your tummy.

This takes a tiny bit of fluid from around the baby.

We can test this for sickle cell disease or thalassaemia.

Only a small number of babies have one of these conditions.

This second test may show if your baby has sickle cell disease or thalassaemia. If your baby has sickle cell disease or thalassaemia you will be given information about the condition to help you decide what to do.

Some babies have the condition worse than others. The test cannot tell you how serious the condition will be.

Some women who are told that their baby has sickle cell disease or thalassaemia decide to carry on with the pregnancy.

Some women who are told that their baby has sickle cell disease or thalassaemia decide they do not want to carry on with the pregnancy. They have an abortion. This means the baby does not live.

If your baby has sickle cell disease or thalassaemia you will be given support to decide whether or not to have an abortion.

This is your decision.

It is important to tell your midwife if you move home.