# Screening tests for you and your baby

Public Health England (PHE) created this leaflet on behalf of the NHS

Important information about the screening choices you will have during and after your pregnancy

This booklet gives you information on the screening tests you will be offered during your pregnancy and after your baby is born. It explains the different types of test and what the tests are looking for.

We hope that reading this booklet will help prepare you for discussions with your midwife or doctor so that you can ask the questions that are important to you. It will be helpful if you have the booklet with you when you see them.

It is your decision whether or not to have any of the tests described in this booklet.

In this booklet, we use the terms ‘woman’ and ‘women’ to refer to anyone able to become pregnant, including trans men. Trans men who are pregnant should be offered the same antenatal and newborn screening tests as other pregnant individuals.

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# Introduction

## What you need to know about screening

### Screening tests

Screening tests are used to find people at higher chance of a health problem. This means they can get earlier, potentially more effective treatment, or make informed decisions about their health. It can be helpful to imagine screening like putting people through a sieve. Most people pass straight through but a small number get caught in the sieve. The people caught in the sieve are those considered to have a higher chance of having the health problem being screened for.

Screening tests are not perfect. Some people will be told that they or their baby have a high chance of having a health problem when in fact they do not have the problem. Also, a few people will be told that they or their baby have a low chance of having a health problem when in fact they do have the problem.

**Partners are welcome to attend screening and information sessions**

### The difference between screening tests and diagnostic tests

A screening test can find out if you, or your baby, have a high or low chance of having a health problem. But it cannot usually tell you for certain, so people found to have a high chance of a problem will often be offered another kind of test. This is called a diagnostic test and gives a more definite ‘yes’ or ‘no’ answer.

## Screening tests during and after pregnancy

Screening tests are offered during pregnancy to try to find any health problems that could affect you or your baby. The tests – ultrasound scans, blood tests and a questionnaire – can help you make choices about care or treatment during pregnancy or after your baby is born.

Watch an animation about the screening tests you'll be offered at www.nhs.uk/pregnancyscreening

Some screening tests are offered for your baby soon after he or she is born. We offer these so your baby can be given appropriate treatment as quickly as possible if needed.

## Some practicalities

If you know that you, the father of your baby, or a family member already has a health problem, please tell your midwife. If you move home while waiting for the results of any screening test, please tell your midwife or health visitor your new address.

## Screening is your choice

Whether or not to have each test is a personal choice and one which only you can make. You can discuss each test you are offered with health professionals and decide, based on your own circumstances, whether or not it is right for you. You can also change your mind at any stage.

Some of the screening tests described in this booklet, such as blood tests for infectious diseases, eye screening if you have diabetes and the newborn checks, are recommended by the NHS. This is because results from these tests can help make sure that you or your baby gets urgent treatment for serious problems.

Screening tests in pregnancy for sickle cell disease and thalassaemia, Down's syndrome, Edwards' syndrome, Patau's syndrome and the 20-week scan can lead to very personal decisions. These could include:

- whether or not to have a diagnostic test with a possible chance of miscarriage
- whether to continue or end your pregnancy

It is important to think carefully about whether or not you want to have these screening tests. Your decisions will be respected and health care professionals will support you. You should speak up if you feel your decisions are not being respected.

## Further support

For details of organisations that can provide support about any of the conditions in this booklet, see www.nhs.uk.

## Screening and the NHS

The NHS decides which screening tests to offer based on good evidence. A group of experts called the UK National Screening Committee (UK NSC) provides advice to the NHS. All screening tests provided by the NHS are free. Some private companies also provide screening tests that you have to pay for. The NHS cannot guarantee the quality of private screening. More advice is available at www.gov.uk/phe/private-screening.

## Confidentiality

Find out how Public Health England and the NHS use and protect your screening information at www.gov.uk/phe/screening-data.

If screening or pre-natal diagnostic (PND) tests suggest your baby has certain conditions we will pass the information to the National Congenital Anomaly and Rare Diseases Registration Service (NCARDRS). This includes:

- sickle cell disease or thalassaemia
- Down's syndrome, Edwards' syndrome or Patau's syndrome
- other conditions picked up by the 20-week scan

This is a secure national register of children who are born with rare conditions. It helps us improve screening and prevent or treat the condition. You can opt out of the register, or find out more, at www.gov.uk/phe/ncardrs. We will never publish information that could identify you or your child.


# Infectious diseases

## Purpose of screening

To find out if you have hepatitis B, HIV (human immunodeficiency virus) or syphilis.

Women already known to have HIV or hepatitis B need early specialist appointments to plan their care in pregnancy.

## About these conditions

Hepatitis B and HIV are passed on in blood and body fluids through sexual contact or infected needles. These viruses can also be passed from mother to baby.

**Hepatitis B** virus affects the liver. It can cause acute (immediate) and chronic (long-term) ill health. Babies infected at birth or during the first year of life have a 9 in 10 (90%) chance of developing lifelong hepatitis B infection. This can lead to illnesses such as liver infections and cancers.

**HIV** weakens the immune system making it difficult to fight off infections. If untreated, it can be passed from mother to baby in pregnancy, when giving birth or by breastfeeding. Treatment in pregnancy greatly reduces the chance of passing on HIV to a baby from 1 in 4 (25%) to less than 1 in 200 (0.5%).

**Syphilis** is an infection passed on through sexual contact. It can be passed from mother to baby during pregnancy. If it is untreated, it can result in serious health problems for a baby or cause miscarriage or stillbirth.



Find more information and details of support

## The screening test

Blood is taken from your arm.

A small amount of this blood is stored in the laboratory. This can be used for tests if you show signs of other infections or rashes during your pregnancy.

## Safety of the test

There are no risks associated with the test.

## Screening is your choice

These tests are strongly recommended to protect your health through early treatment and care. They greatly reduce any chance of passing an infection onto your baby, partner or other family members.

## Not having the test

If you decide not to be tested, you will be reoffered screening later in pregnancy, ideally by 20 weeks.

**England now has its lowest ever rate of mother-to-child HIV transmission**



groups at **www.nhs.uk/infections**

# Infectious diseases

## Negative result

A negative result means you are ‘negative now’. This does not mean you are protected throughout your pregnancy. Protect yourself from infection and report symptoms as soon as possible to your midwife or GP. You and your partner can be tested at any time.

We recommend repeat testing if you change your sexual partner, inject drugs, are a sex worker, have an infected partner, have a partner who is sexually active with other people or are diagnosed with a sexually transmitted infection (STI).

## Positive result

If you have **hepatitis B** it is important for specialist teams to check your health before and after the birth. Your partner, any other children and close family members may need testing and vaccination.

It is very important that your baby has all 6 recommended hepatitis B vaccinations at the correct times to protect their health. Ask your GP, practice nurse or health visitor if any of these do not happen. Vaccinations should take place:

- within 24 hours of birth
- at 4 weeks of age



Find more information and details of support


- at 8, 12 and 16 weeks of age (part of routine childhood immunisation schedule)
- at one year of age

Your baby may also need an injection of antibodies (hepatitis B immunoglobin) at their first vaccination.

They will also have a blood test at their final vaccination to check if infection has been avoided.

If you have **HIV** you can greatly reduce the chance of passing HIV on to your baby with specialist care and treatment, drugs, planned care for your birth, and by not breastfeeding.

If you have **syphilis** urgent referral to a specialist team is needed. Treatment is usually a course of antibiotics. These are safe for your baby. The team will also offer to test your partner to see if they need treatment. Your baby will need an examination and blood tests after birth and may need antibiotics.

## Getting my results

If your result is negative your midwife will discuss it with you. This will happen at or before your next antenatal appointment at about 16 weeks. It will also be recorded in your notes.

If you are positive for HIV, hepatitis B or syphilis a specialist midwife will contact you to arrange an appointment within 10 days. They will discuss the result, carry out further tests and plan your care with you.

# Sickle cell and thalassaemia

## Purpose of screening

To find out if you are a carrier of the sickle cell or thalassaemia gene and therefore likely to pass it on to your baby.

## About these conditions

Sickle cell disease (SCD) and thalassaemia major are serious, inherited blood diseases. 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.

People with SCD can have attacks of very severe pain, get serious, life-threatening infections and are usually anaemic (their bodies have difficulty carrying oxygen). Babies with SCD can receive early treatment, including immunisations and antibiotics, which, along with support from their parents, will help prevent serious illness and allow them to live a healthier life.

**It is best to have the test before you are 10 weeks pregnant**

People with thalassaemia major are very anaemic and need a blood transfusion every 3 to 5 weeks, and injections and medicines throughout their lives.

There are also other, less common, less serious haemoglobin diseases that may be found.



Find more information and details of support

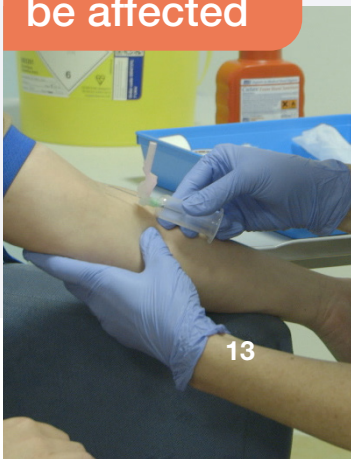
SCD and thalassaemia are inherited diseases that are passed on from parents to children through unusual haemoglobin genes. Genes are the codes in our bodies for things such as eye colour and blood group. Genes work in pairs. For everything that we inherit we get one gene from our mother and one from our father.

People only have SCD or thalassaemia if they inherit 2 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 disease, although they can experience some problems in situations where their bodies might not get enough oxygen, for example, having an anaesthetic.

When **both parents** are carriers the baby has:

- a 1 in 4 (25%) chance of not being affected – the baby will not have or carry a disease
- a 1 in 4 (25%) chance of inheriting both unusual haemoglobin genes and having a haemoglobin disease
- a 2 in 4 (50%) chance of inheriting 1 unusual haemoglobin gene and being a carrier

**Both parents need to be a carrier for your child to be affected**





groups at **www.nhs.uk/sct**

# Sickle cell and thalassaemia

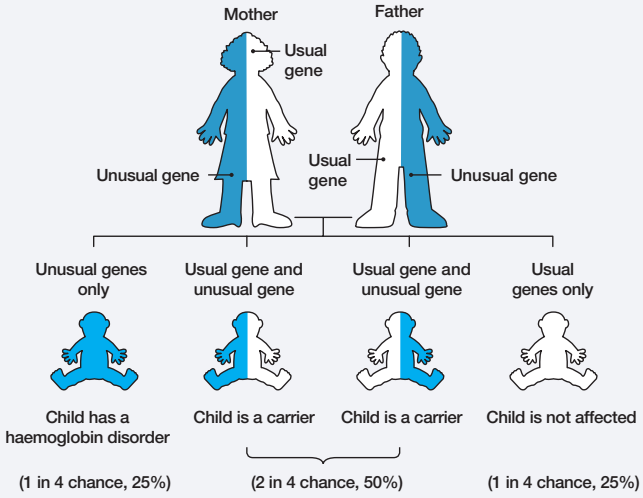
Anyone can be a carrier of a haemoglobin disease. However, it is more common among people whose ancestors come from Africa, the Caribbean, the Mediterranean, India, Pakistan, South and South-East Asia and the Middle East.

## The screening test

Screening in pregnancy for SCD and thalassaemia involves having a blood test. It is best to have the test before you are 10 weeks pregnant.

All pregnant women are offered a test for thalassaemia but not all women are automatically offered a test for SCD. The screening offered depends on where you live.

In areas where haemoglobin diseases are more common, you will be offered a blood test for SCD. In areas where haemoglobin diseases are less common, a questionnaire is used to identify the family origins of the baby's mother and father.



If the questionnaire shows either parent may be a sickle cell carrier, a blood test is offered to the woman.

You can ask to have the blood test even if your family origins do not suggest your baby has a high chance of a haemoglobin disease.

**Dads – if the mum is a carrier it's important for you to get tested too**

## Safety of the test

The screening test cannot harm you or your baby but it is important to consider carefully whether to have this test or not. The screening test can provide information that may mean you have to make further important decisions. For example, you may be offered further tests that have a risk of miscarriage.

## Screening is your choice

You do not need to have the screening test. Some people want to find out if their baby has SCD or thalassaemia and some do not.

## Not having the test

If you choose not to have the screening test in pregnancy, your baby can have newborn blood spot screening for SCD at 5 days of age.



groups at **www.nhs.uk/sct**

# Sickle cell and thalassaemia

## Possible results

The test will tell you if you are a carrier or not, or whether you have the disease yourself.

## Further tests

If you are a carrier of a haemoglobin disease, your baby's father will be offered a blood test. If your baby's father is also a carrier you will be offered diagnostic tests to find out if your baby is affected.

If your baby's father is not available and you have been identified as a carrier, you will be offered the diagnostic test.

1 in every 200 women (0.5%) who have a diagnostic test will miscarry as a result of the test. It is up to you whether or not to have the further test. There are 2 types of diagnostic test.

**CVS (chorionic villus sampling)** is usually done from 11 to 14 weeks of pregnancy. A fine needle, usually put through the mother's abdomen, is used to take a tiny sample of tissue from the placenta. The cells from the tissue can be tested for SCD or thalassaemia.

**Amniocentesis** is usually done after 15 weeks of pregnancy. A fine needle is passed through the mother's abdomen into the uterus to collect a small sample of the fluid surrounding the baby. The fluid contains some of the baby's cells, which can be tested for SCD or thalassaemia.

## Possible results of diagnostic tests

If the result shows that your baby has SCD or thalassaemia then you will be offered an appointment with a health professional. You will be able to get information about the condition your baby has inherited and talk through your choices.

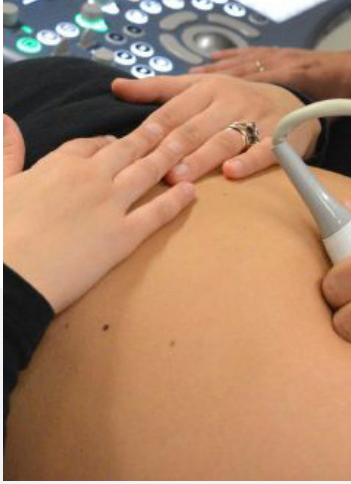
Some conditions are more serious than others. Some women decide to continue with the pregnancy. Others decide they do not want to continue with the pregnancy and have a termination.

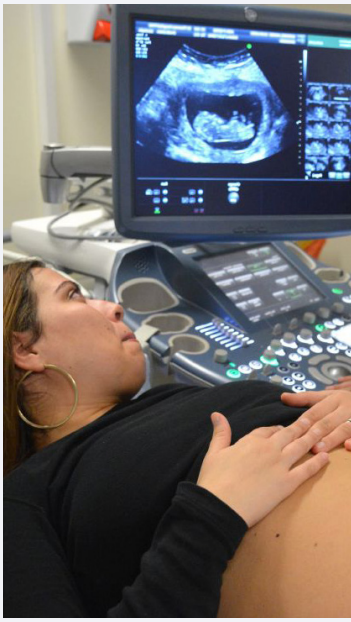
If you are faced with this choice you will get further information about the condition and support from health care professionals to help you make a decision. Information is also available from support groups.

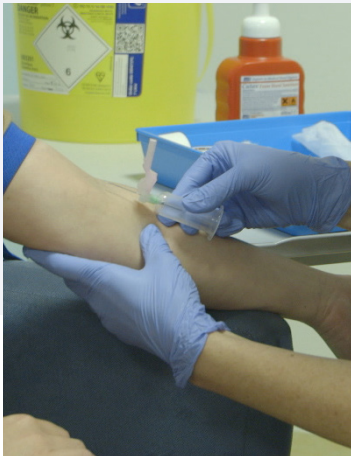
If the test shows that you are a carrier, there is a chance that other members of your family could be carriers too. You may want to encourage them to ask for a test, especially if they are planning to have a baby.

## Getting results of diagnostic tests

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







groups at **www.nhs.uk/sct**

This screening will be offered if you're having one baby or twins

# Down's syndrome, Edwards' syndrome and Patau's syndrome

## Purpose of screening

To find out how likely it is that your baby has Down's syndrome (also known as Trisomy 21 or T21), Edwards' syndrome (Trisomy 18/T18) or Patau's syndrome (Trisomy 13/T13).

## About these conditions

Inside the cells of our bodies there are tiny structures called chromosomes. These chromosomes carry the genes that determine how we develop. There are 23 pairs of chromosomes in each cell. Changes can occur when the sperm or egg cells are produced, which can lead to a baby having an extra chromosome.

Babies with Down's syndrome, Edwards' syndrome and Patau's syndrome are born to mothers of all ages but the chance of having a baby with one of these conditions increases as the age of the mother increases.

### Down's syndrome (T21)

Down's syndrome is caused by an extra copy of chromosome 21 in all or some cells of the body.

A person with Down's syndrome will have some level of learning disability. This means they will find it harder than



Find more information and details of support

most people to understand and to learn new things. They may have communication challenges and difficulty managing some everyday tasks. People with Down's syndrome have distinctive facial features but they do not all look the same.

A child with Down's syndrome

Most children with Down's syndrome attend mainstream schools but will require additional support.

Some health problems are more common in people with Down's syndrome. These include heart conditions and problems with hearing and vision. Many health problems can be treated but unfortunately around 5% of babies will not live past their first birthday.

For babies without serious health problems survival is similar to that of other children and most people with Down's syndrome will live into their 60s or longer.

People with Down's syndrome can have a good quality of life and most say they enjoy their lives. With support, many more people with Down's syndrome are able to get jobs, have relationships and live semi-independently in adulthood.

**Edwards' syndrome (T18) and Patau's syndrome (T13)**

Babies with Edwards' syndrome have an extra copy of chromosome 18 in all or some cells. Babies with Patau's

# Down's syndrome, Edwards' syndrome and Patau's syndrome

syndrome have an extra copy of chromosome 13 in all or some cells.

Sadly the survival rates are low and of those babies born alive only around 13% with Edwards' syndrome and 11% with Patau's syndrome will live past their first birthday. Some babies may survive to adulthood but this is rare.

All babies born with Edwards' syndrome and Patau's syndrome will have a learning disability and a wide range of physical challenges, which can be extremely serious. They may have problems with their heart, respiratory system, kidneys and digestive system.

**Down's syndrome** occurs in 10 in every 10,000 births.

**Edwards' syndrome** occurs in 3 in every 10,000 births.

**Patau's syndrome** occurs in 2 in every 10,000 births.

Around half of babies with Patau's syndrome will also have a cleft lip and palate. Babies with Edwards' syndrome and Patau's syndrome will have a low birthweight.

Despite their difficulties, children can slowly make progress in their development. Older children with either condition would need to attend a specialist school.



Find more information and details of support

## The screening test

A screening test for these conditions, called the "combined test", is available between 10 and 14 weeks of pregnancy.

If you choose to have the combined test, a blood sample is taken from your arm. At the dating ultrasound scan the fluid at the back of your baby's neck is measured (known as the nuchal translucency). The information from these 2 tests is combined to work out the chance of your baby having Down's syndrome, Edwards' syndrome or Patau's syndrome.

It is your decision whether or not to have screening

If you are too far on in your pregnancy to have the combined test for Down's syndrome, you will be offered a blood test called the quadruple test between 14 and 20 weeks of pregnancy. The quadruple test is not quite as accurate as the combined test. If you are too far on in your pregnancy to have the combined test for Edwards' syndrome and Patau's syndrome, you will be offered a 20-week scan.

## Safety of the test

The screening test cannot harm you or your baby but it is important to consider carefully whether to have this test or not. This test cannot tell you if your baby definitely has Down's syndrome, Edwards' syndrome or Patau's syndrome or not. The screening test can provide information that may lead to further important decisions. For example, you may be offered diagnostic tests that have a risk of miscarriage.

# Down's syndrome, Edwards' syndrome and Patau's syndrome

## Screening is your choice

You do not have to have the screening test. Some people want to find out if their baby has Down's syndrome, Edwards' syndrome or Patau's syndrome and some do not.

You can choose to have screening for:

- all 3 conditions
- Down's syndrome only
- Edwards' syndrome and Patau's syndrome only
- none of the conditions

## Not having the test

If you choose not to have the screening test for Down's syndrome, Edwards' syndrome and Patau's syndrome, the rest of your antenatal care will be unaffected.

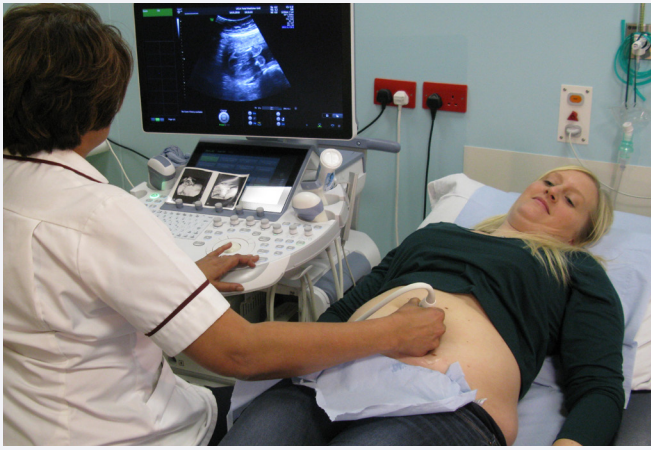
Any scan during your pregnancy may pick up physical problems with your baby which could be related to these conditions. You will always be told if anything unexpected is found during a scan.

## Possible results

You will be given 2 results: one for Down's syndrome and a joint one for Edwards' syndrome and Patau's syndrome.

If the screening test shows that the chance of your baby

having Down's syndrome, Edwards' syndrome or Patau's syndrome is lower than 1 in 150 this is called a "lower-chance" result. Over 95 out of 100 (95%) screening test results will be lower chance.



A lower-chance result does not mean that there is no chance at all of your baby having Down's syndrome, Edwards' syndrome or Patau's syndrome.

If the screening test shows that the chance of your baby having Down's syndrome, Edwards' syndrome or Patau's syndrome is higher than 1 in 150 – that is from 1 in 2 to 1 in 150 – this is called a "higher-chance" result. Fewer than 1 in 20 (5%) of screening test results will be higher chance.

A higher-chance result does not mean your baby definitely has Down's syndrome, Edwards' syndrome or Patau's syndrome.

## Further tests

If you have a lower-chance result, you will not be offered a further test.



groups at **www.nhs.uk/depscreening**

# Down's syndrome, Edwards' syndrome and Patau's syndrome

If you have a higher-chance result, you will be offered a diagnostic test to find out for certain whether or not your baby has Down's syndrome, Edwards' syndrome or Patau's syndrome.

Diagnostic tests for Down's syndrome will also look at chromosomes 18 and 13, so will also tell you whether or not your baby has Edwards' syndrome or Patau's syndrome. Similarly, diagnostic tests for Edwards' syndrome and Patau's syndrome will also look at chromosome 21 for Down's syndrome.

1 in every 200 women (0.5%) who have a diagnostic test will miscarry as a result of the test. It is up to you whether or not to have the further test.

Screening does not give definite answers. That is why you may be offered a diagnostic test

There are 2 types of diagnostic test.

**CVS (chorionic villus sampling)** is usually done from 11 to 14 weeks of pregnancy. A fine needle, usually put through the mother's abdomen, is used to take a tiny sample of tissue from the placenta.

The cells from the tissue are then tested for Down's syndrome, Edwards' syndrome and Patau's syndrome.

**Amniocentesis** is usually done after 15 weeks of pregnancy. A fine needle is passed through the mother's abdomen into the uterus to collect a small sample of the fluid surrounding the baby. The fluid contains cells from the baby, which are tested for Down's syndrome, Edwards' syndrome and Patau's syndrome.

A small number of women who have the diagnostic test will find out their baby has one of the conditions. They then have 2 options. Some women decide to continue with the pregnancy and prepare for their child with the condition; others decide they do not want to continue with the pregnancy and have a termination.

If you are faced with this choice you will get support to help you make a decision.

## Getting my results

If your screening test shows a lower-chance result, you should be told within 2 weeks of the test being taken.

If your screening test shows a higher-chance result, you should be told within 3 working days of the blood test result being available. You will be offered an appointment to discuss the test results and the further options you will have.

# 11 physical conditions (20-week scan)

## Purpose of screening

This scan takes place between 18 weeks and 20 weeks 6 days of pregnancy and is commonly called the 20-week scan. It is also sometimes referred to as the mid-pregnancy scan.

The scan looks for 11 different conditions in your baby and cannot find everything that might be wrong.

## About these conditions

The scan will look in detail at your baby's bones, heart, brain, spinal cord, face, kidneys and abdomen.

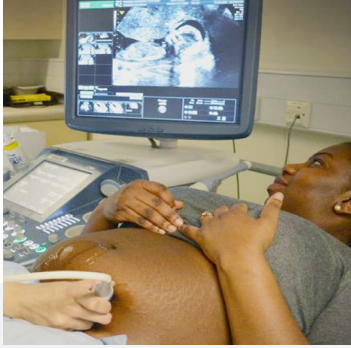
In most cases the scan will show that your baby appears to be developing as expected but sometimes a condition is found or suspected. Some things can be seen more clearly than others. For example, some babies have a condition called open spina bifida, which affects the spinal cord.

Spina bifida can usually be seen clearly on a scan and of those babies who have this condition, around 9 out of 10 (90%) will be detected.

**The scan looks for certain physical conditions but cannot find everything that could be wrong**

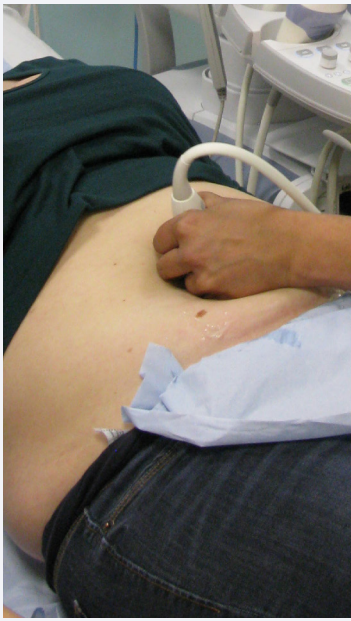


Find more information and details of support



Some of the other conditions, such as heart defects, are more difficult to see. The scan will find about half (50%) of those babies who have heart defects.

Some of the conditions that can be seen on the scan will mean the baby may need treatment or surgery after it is born, for example cleft lip. In a small number of cases some very serious conditions are found. For example, the baby's brain, kidneys, internal organs or bones may not have developed properly. In some very serious rare cases, where no treatment is possible, the baby will die soon after it is born or during pregnancy.

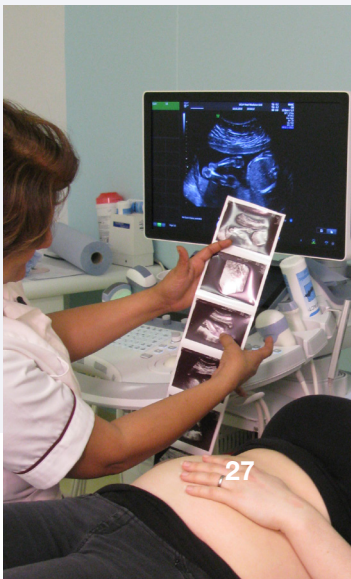


For more detailed information about the main conditions that are looked for during this scan please see **www.nhs.uk/20weekscan**.

## The screening test

Most scans are carried out by specially trained staff called sonographers.

In order for the sonographer to get good images of your baby, the scan is carried out in a dimly lit room.





groups at **www.nhs.uk/20weekscan**

# 11 physical conditions (20-week scan)

1. You will be asked to lie on a couch.
2. You will then be asked to raise your top to your chest and lower your skirt or trousers to your hips.
3. Tissue paper will be tucked around your clothing to protect it from the ultrasound gel, which will then be put on your tummy.
4. The sonographer then passes a hand-held probe over your skin to examine the baby's body. The gel makes sure there is good contact between the probe and your skin.

**You may be offered further tests if a condition is suspected**

Having the scan does not hurt but the sonographer may need to apply slight pressure to get the best views of your baby. This might be uncomfortable. A black and white picture of your baby will then be seen on the ultrasound screen. During the examination, sonographers need to keep the screen in a position that gives them a good view of your baby. The screen may be directly facing them or at an angle.

The appointment usually takes around 30 minutes.

You may need to have a full bladder when you come for the appointment. The doctor or midwife looking after you will let you know before you come. If you are not sure, you can contact them and ask.

You may like someone to come with you to the scan appointment. Most hospitals do not allow children to attend

scans as childcare is not usually available. Please ask your hospital about this before your appointment.

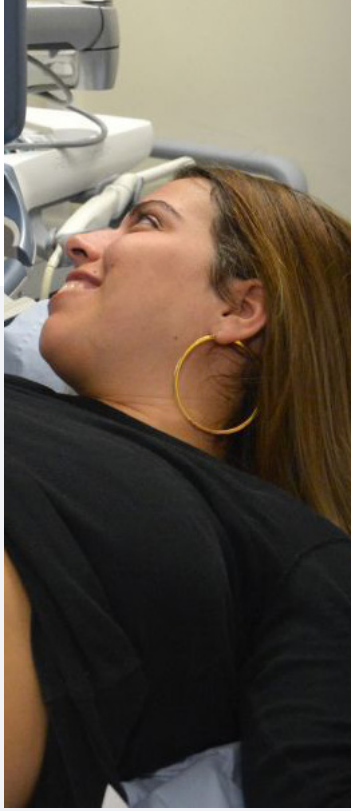
Sometimes it is difficult to get good views of a baby. This does not mean there is anything to worry about. If this happens, you will be offered one further scan by 23 weeks of pregnancy. Very occasionally this second scan cannot be completed, for example because:

- a baby is lying in an awkward position
- you are above average weight

In this case you will not be offered another screening scan but you will offered an all over physical examination for your baby after birth.

## Safety of the test

There are no known risks to baby or mother from having an ultrasound scan but it is important that you consider carefully whether or not to have the 20-week scan. The scan can provide information that may mean you have to make further, important decisions. For example, you may be offered further tests that have a risk of miscarriage.





groups at **www.nhs.uk/20weekscan**

# 11 physical conditions (20-week scan)

## Screening is your choice

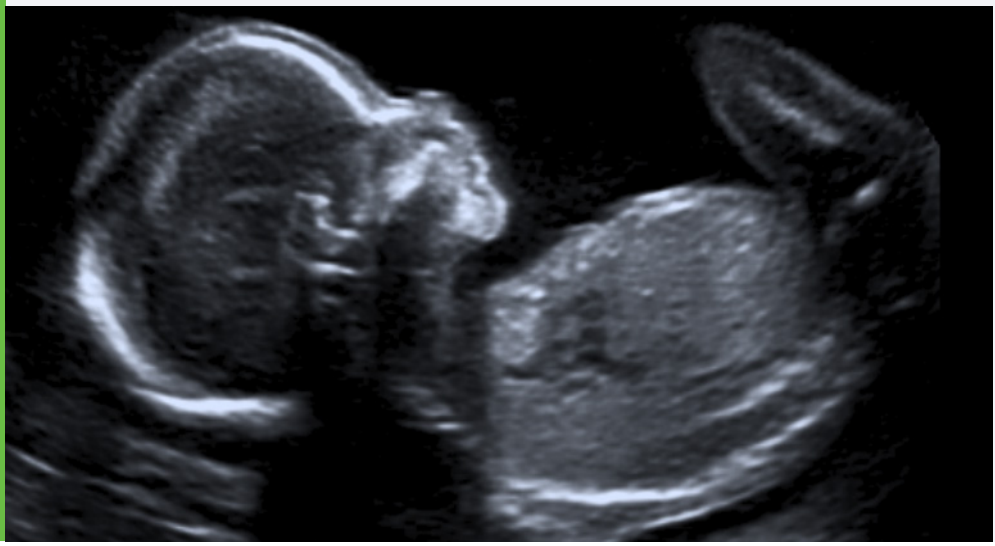
You do not have to have the scan. Some people want to find out if their baby has one of the 11 conditions and some do not.

## Not having the test

If you choose not to have the scan you can still have all other parts of your routine antenatal care.

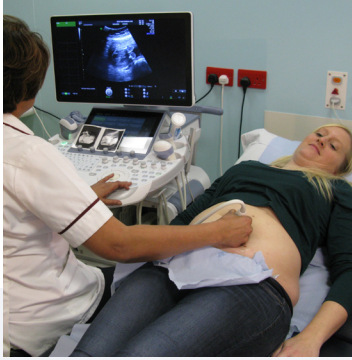
## Possible results

Most scans show that babies seem to be developing as expected, and none of the 11 conditions are found.



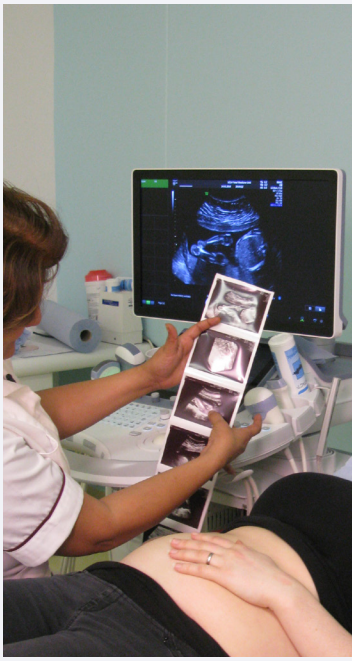


Find more information and details of support



If one of the conditions is found or suspected, the sonographer may ask for a second opinion from another member of staff.

Scans cannot find all conditions. There is always a chance that a baby may be born with a health issue that scans could not have identified.



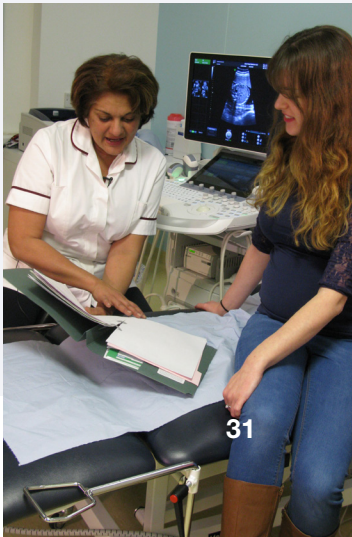
## Further tests

You might be offered another test to find out for certain if your baby has one of the conditions.

If you are offered further tests, you will be given more information about them so that you can decide whether or not you want to have them. You will be able to discuss this with your midwife or consultant. If necessary, you will be referred to a specialist, possibly in another hospital.

## Getting my results

The sonographer will be able to tell you the results of the scan at the time.





groups at **www.nhs.uk/20weekscan**

# Diabetic eye screening

## Purpose of screening

To check for signs of diabetic retinopathy and other eye problems caused by diabetes and monitor the health of your eyes if you are pregnant and have type 1 or type 2 diabetes.

Some women develop gestational diabetes in later pregnancy (28 weeks or more). Gestational diabetes can be treated through changes in diet and usually goes away once the baby is born. You will not be offered diabetic eye screening if you develop gestational diabetes.

## About this condition

Diabetic retinopathy is caused when diabetes affects the small blood vessels in the retina at the back of the eye.

Everyone with diabetes is offered eye screening but screening is very important when you are pregnant because the risk of serious eye problems is greater.

**Screening will only be offered if you already had diabetes before you were pregnant**



Find more information and details of support

## The screening test

You will be offered screening at, or soon after, your first antenatal clinic visit and also after 28 weeks of pregnancy. If early stages of retinopathy are found at the first screening, you will be offered another test between 16 and 20 weeks of pregnancy. If significant retinopathy is found at any screening, you will be referred to an eye specialist.

The screening test during pregnancy is the same as routine screening at all other times.

**Diabetic retinopathy is treatable, especially if caught early**

Screening staff will record your details and level of sight. They will put drops in your eyes to make your pupils larger so the retinas can be seen more clearly. They then take digital photographs of your retinas.

If the images are not clear enough you will be referred to an eye specialist for a different test.

groups at **www.nhs.uk/diabeticeye**

# Diabetic eye screening

## Safety of the test

The photograph is painless and the camera does not come into contact with the eye. The eye drops may sting for a few seconds and cause blurred vision for 2 to 6 hours after the test. Take all your usual spectacles with you. Take sunglasses with you to wear home, as everything may look very bright after the screening test.

Arrange to use public transport or get a lift for the journey home. **You should not drive after screening as the eye drops can blur vision**.

Very rarely, the drops can cause a sudden, dramatic rise in pressure within the eye. Symptoms of pressure rise include:

- pain or severe discomfort in the eye
- redness of the white of the eye
- constantly blurred sight

If you experience any of these symptoms after screening, you should return to the eye unit or go to an accident and emergency department.

## Screening is your choice

This test is strongly recommended if you already had diabetes before getting pregnant.

Eye screening is a part of managing your diabetes and diabetic retinopathy is treatable, especially if it is caught early.



Find more information and details of support

## Not having the test

People with diabetes can decide not to attend eye screening appointments.

If you decide not to have the test you should notify the clinician looking after your diabetes care during pregnancy.

## Possible results

If you have the screening test, possible results are:

- no retinopathy
- early signs of retinopathy
- more serious retinopathy that requires referral to a specialist

If your test shows early signs of retinopathy, your health professional will give you advice about looking after your diabetes during pregnancy. You would then be invited for additional screening tests during your pregnancy. If you are a smoker, seek help to stop or at least cut down.

If the test shows you have referable retinopathy you will receive an appointment with an eye specialist.

## Getting my results

A letter will be sent to you and your GP within 6 weeks following your screening appointment.


# Eyes, heart, hips and testes (physical examination)

## Purpose of screening

Within 72 hours of giving birth, you will be offered a top-to-toe physical examination for your baby. This will include 4 specific screening tests to find out whether your baby has a suspected problem with their eyes, heart, hips or, in boys, their testes which would benefit from early investigation and possible treatment.

The screening tests are carried out again between 6 and 8 weeks of age as some conditions do not develop or become apparent until then.

## About the conditions

Each part of the physical examination is looking for different conditions.

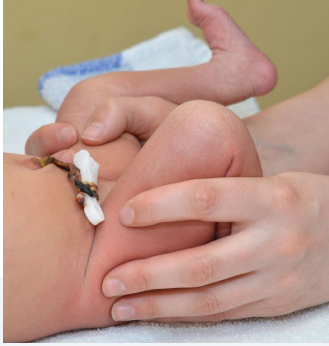
**Eyes** – about 2 or 3 in 10,000 babies have problems with their eyes that need treatment. The examination checks the appearance and movement of the eyes. The main reason for screening is to identify a condition called cataracts (a clouding of the transparent lens inside the eye). The examination cannot tell how well your baby can see.

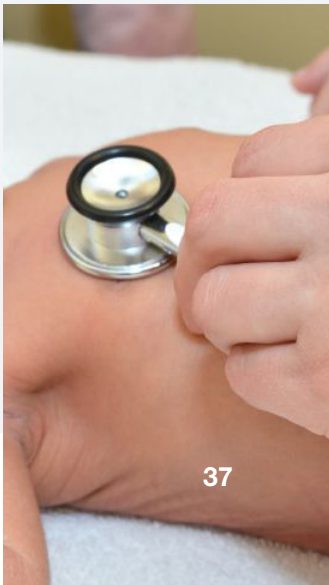
**Heart** – there is a general examination of your baby's heart

**Screening is part of a top-to-toe physical examination of your baby**



Find more information and details of support





and sometimes murmurs are picked up. A murmur is a noise made by the blood as it passes through the heart. In almost all cases when a murmur is heard, the heart is normal. Murmurs are common in babies and this does not always mean there is a problem. However, around 1 in 200 babies have a heart problem that needs treatment.

**Hips** – babies can be born with hip joints that are not formed properly and if untreated this can lead to a limp and to joint problems. About 1 or 2 in 1,000 babies have hip problems that need treatment.

**Testes** – baby boys are checked to make sure their testes are in the right place. It can take several months for testes to drop down (descend) into the scrotum. About 1 in 100 baby boys have problems with undescended testes that need treatment to reduce the risk of problems later in life, such as reduced fertility.

## The screening test

A trained health professional will carry out a physical examination of your baby and ask you questions about your baby's general wellbeing.



groups at **www.nhs.uk/newborninfantexam**


# Eyes, heart, hips and testes (physical examination)

Your baby will need to be undressed for part of the examination. During the examination the health professional will:

- look into your baby's eyes focusing on how their eyes look and move
- listen to their heart using a stethoscope to detect heart sounds
- examine their hips to ensure the joints are working properly
- examine baby boys to check if their testes are in the right place

These screening tests are carried out within 72 hours of birth and again when your baby is between 6 and 8 weeks of age.

**Screening is offered within 72 hours of birth and again at 6 to 8 weeks**

## Safety of the test

There are no risks associated with having this test.

## Screening is your choice

The examination is intended to identify any of the problems early so that treatment can be started as soon as possible.


The general physical examination including this screening is therefore recommended for your baby.

## Not having the test

You can decide to have your baby examined and screened for any or all of the conditions. If you have any concerns you should discuss them with your midwife and the health professional who offers the examination.

**Early detection of problems means quicker treatment and better health outcomes**

## Possible results

Usually there will be nothing of concern found. If the health professional finds a possible problem, your baby will be referred for further assessment and tests if appropriate.

## Getting my results

The health professional carrying out the examination will give you the results straight away. If a referral for further assessment is needed, this will also be discussed with you at the time of the examination.

The results will be recorded in your baby's case notes and personal child health record ('red book'). You will need to keep this record safe and make sure it is available whenever your baby sees a health professional.



groups at **www.nhs.uk/newborninfantexam**

# Hearing loss

## Purpose of screening

To find babies who have a hearing loss so that support and advice can be offered right from the start.

## About this condition

One to 2 babies in every 1,000 are born with a permanent hearing loss in one or both ears. Most of these babies are born into families with no history of hearing loss.

Permanent hearing loss can significantly affect a baby's development. Finding out early can give these babies a better chance of developing speech and language skills. It will help babies make the most of relationships with their family and carers from an early age.

## The screening test

The hearing screening test will be offered to you in hospital before discharge, or when you are at home, or you will be invited to attend a clinic appointment. In some areas, screening will be done by the health visitor within the first few weeks. Ideally, the test should be done in the first 4 to 5 weeks, but it can be done at up to 3 months of age.

The test called the AOAE (automated otoacoustic emission) test takes a few minutes. A small soft tipped earpiece is placed in your

**Most babies with hearing loss are born into families with no previous history of the condition**



Find more information and details of support

baby's ear and soft clicking sounds are played. When an ear receives sound, the inner part (called the cochlea) responds and this can be picked up by the screening equipment.

It is not always possible to get clear responses from the first test. This does not necessarily mean your baby has a hearing loss. It can mean:

- your baby was unsettled when the test was done
- there was background noise
- your baby has fluid or a temporary blockage in their ear – this is very common and passes with time
- your baby has a hearing loss

In these cases your baby will be offered another test. This may be the same as the first test, or another type called the AABR (automated auditory brainstem response) test. This involves 3 small sensors being placed on your baby's head and neck. Soft headphones are placed over your baby's ears and soft clicking sounds are played. This test takes between 5 and 15 minutes.

**The hearing test is straightforward and most babies sleep right through it**



groups at **www.nhs.uk/newbornhearing**

# Hearing loss

## Safety of the test

There are no risks associated with having this test.

## Screening is your choice

This screening test is recommended for your baby. Finding hearing loss early is important for your baby's development.

## Not having the test

If you decide not to have the newborn hearing screening test you will be given checklists to help you check on your baby's hearing as they grow older. If you have any concerns you should speak to your health visitor or GP.

## Possible results

If your baby has a clear response in both ears then they are unlikely to have a permanent hearing loss. However, newborn hearing screening does not pick up all types of hearing loss and children can develop hearing loss later on. It is important to check your child's hearing as they grow up. The checklist in your baby's personal child health record ('red book') tells you how to do this. If you have any concerns about your child's hearing tell your health visitor or GP.

**The hearing test is simple and most babies sleep right through it**

If the screening test results do not show a clear response from one or both of your baby's ears an



Find more information and details of support

appointment will be made with audiology to see a hearing specialist.

About 2 to 3 babies in every 100 do not show a clear response on the screening tests. Being sent for further tests does not necessarily mean your baby has a hearing loss.

A hearing specialist should see you within 4 weeks of having your baby's hearing screening test. It is very important that you attend the appointment in case your baby has a hearing loss.

## Getting my results

You will be given your baby's results as soon as the hearing test is done.



groups at **www.nhs.uk/newbornhearing**

# Blood spot

## Purpose of screening

To find out if your baby has any of 9 rare but serious health conditions.

Early treatment can improve your baby's health and prevent severe disability or even death. If you, the baby's father, or a family member already has one of these conditions, please tell your health professional straight away.

**Blood spot screening is recommended as it can save your baby's life**

## About these conditions

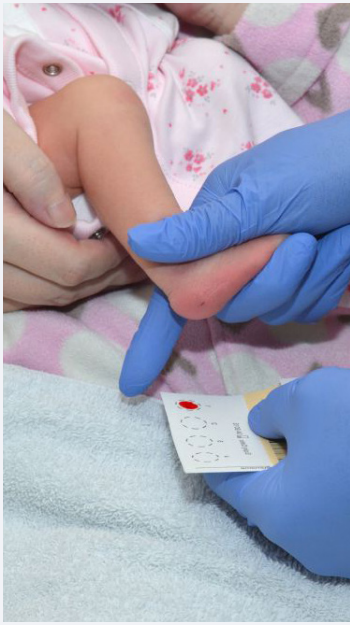
### Sickle cell disease

About 1 in 2,800 babies born in the UK has a sickle cell disease (SCD). These are serious, inherited blood diseases. They affect haemoglobin, a part of the blood that carries oxygen around the body. Babies who have SCD will need specialist care throughout their lives.

People with SCD can have attacks of severe pain, get serious, life-threatening infections and are usually anaemic (their bodies have difficulty carrying oxygen). Babies with SCD can receive early treatment, including vaccinations and antibiotics, which, along with support from their parents, will help prevent serious illness and allow them to live a healthier life.



Find more information and details of support



## Cystic fibrosis

About 1 in 2,500 babies born in the UK has cystic fibrosis (CF). This inherited condition affects the digestion and lungs. Babies with CF may not gain weight well and frequently have chest infections.

Babies with CF can be treated early with a high-energy diet, medicines and physiotherapy. Although children with CF may still become very ill, early treatment can help them live longer, healthier lives.

## Congenital hypothyroidism

About 1 in 2,000 babies born in the UK has congenital hypothyroidism (CHT). Babies with CHT do not have enough of the hormone thyroxine. Without thyroxine babies do not grow properly and they can develop permanent serious physical problems and learning disabilities.

Babies with CHT can be treated early with thyroxine tablets and this will allow them to develop normally.

## Inherited metabolic diseases

It is important to let your health professional know if you have a family history of a




groups at **www.nhs.uk/bloodspot**

# Blood spot

metabolic condition. Babies are screened for 6 inherited metabolic diseases (IMDs).

These are:

- phenylketonuria (PKU)
- medium-chain acyl-CoA dehydrogenase deficiency (MCADD)
- maple syrup urine disease (MSUD)
- isovaleric acidaemia (IVA)
- glutaric aciduria type 1 (GA1)
- homocystinuria (pyridoxine unresponsive) (HCU)

About 1 in 10,000 babies born in the UK has PKU or MCADD. The other conditions are rarer, occurring in 1 in 150,000 babies to 1 in 300,000 babies.

Babies with these inherited conditions cannot process certain substances in their food. Without treatment babies with some of these conditions can become suddenly and seriously ill. The symptoms of the conditions are different; some may be life threatening or lead to severe developmental problems.

**The information collected on your baby's blood spot card is important – make sure all the details are correct**

They can all be treated by a carefully managed diet, which is different for each condition and may include additional medicines.


## The screening test

When your baby is 5 days old the health professional will prick your baby's heel using a special device to collect some drops of blood onto a card. Occasionally this may be later than 5 days. The heel prick may be uncomfortable and your baby may cry. You can help by making sure your baby is warm and comfortable, and by cuddling and feeding them.

Sometimes, a second blood spot sample is required later on. If so, the reason will be explained. It does not necessarily mean there is something wrong with your baby.

## Safety of the test

There are no known risks to your baby associated with having the test.

## Screening is your choice

Screening your baby for all these conditions is recommended because it could save your baby's life but it is not compulsory. You can choose to have screening for SCD, CF or CHT individually but can only choose to have screening for all 6 IMDs or none at all. If you do not want your baby screened for any of the conditions or have any concerns about the test, please talk to your midwife.

Early screening is best as treatment can be started as soon as possible if needed. But if you choose not to have screening, your baby can have the test later if you change your mind. Babies can be screened up to 12 months of age for all the conditions except CF (only up to 8 weeks of age).



groups at **www.nhs.uk/bloodspot**

# Blood spot

## Possible results

Most babies will have normal results indicating that it is unlikely that they have any of the conditions. A small number of babies will screen positive for one of the conditions. This does not mean they have the condition but they are more likely to have it. They will be referred to a specialist for further tests.

Screening for cystic fibrosis finds some babies who may be genetic carriers of the condition. These babies may need further testing. Screening does not detect all carriers.

Occasionally, other medical conditions might be identified through these screening tests. For example, babies with beta thalassaemia major (a serious blood disease) will usually be detected. These children also need to be referred for lifelong treatment and care.

Screening for sickle cell disease also finds babies who are genetic carriers of these or other red blood cell diseases. Carriers are healthy although they can experience some problems in situations where their bodies might not get enough oxygen, for example, if they are having an anaesthetic.

## Getting my results

You should receive the results from a health professional by the time your baby is 6 weeks old. The results should be recorded in your baby's personal child health record ('red book'). Please keep this safe and bring it with you to any further appointments. You will be contacted sooner if there is thought to be any problem with your baby.

## My baby's blood spot card and data after screening

After screening blood spot cards are stored for at least 5 years and may be used:

- to check the result or for other tests recommended by your doctor
- to improve the screening programme
- for research to help improve the health of babies and their families in the UK

This research will not identify your baby and you will not be contacted. The use of these blood spots is governed by the code of practice available from your midwife, or on the website.

There is a small chance researchers may want to invite you or your child to take part in research linked to this screening programme. If you do not want to be invited to take part in research, please let your midwife know.

# Notes

You can write down any notes here from discussions with your health professionals

A phone-friendly HTML version of this leaflet is available. You can view and download it in large print, and use a screen reader for an audio version. Scan the QR code above or visit: **www.gov.uk/phe/pregnancy-newborn-screening**

We can provide a braille version.
Email: **phe.screeninghelpdesk@nhs.net**

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**PHE Gateway number:** GW-



**Web address:** www.gov.uk/phe/screening-leaflets

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