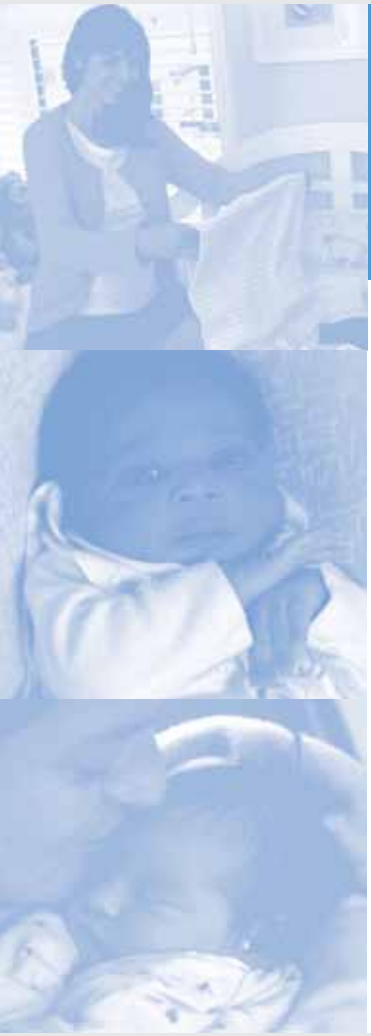
NSC UK National Screening Committee

NHS Screening Programmes

Antenatal and Newborn



Screening tests for you and your baby

Important information for you to keep with your hand-held maternity records

2010 version

Screening in pregnancy: When it

Being screened is entirely your choice. Please ask if there is anything

Pre-conception

Antenatal

Commence Folic Acid

Sickle Cell & Thalassaemia
Blood test

Early Down's Syndrome
Blood test

Haemoglobin; Group & Antibodies; Resus Factor; Syphilis; Hepatitis B; Rubella susceptibility; HIV
Blood test

Week 0 1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 23

Pre-Screening Info
Read & discuss

Dating
Ultrasound scan

Nuchal Translucency
Ultrasound scan

Detailed Ultrasound Anomaly
Ultrasound scan

Contents

happens

about it that you do not understand

Newborn

Later Down's Syndrome
Blood test

Haemoglobin & Antibodies
Blood test

Physical Examination by 72 hours
Check

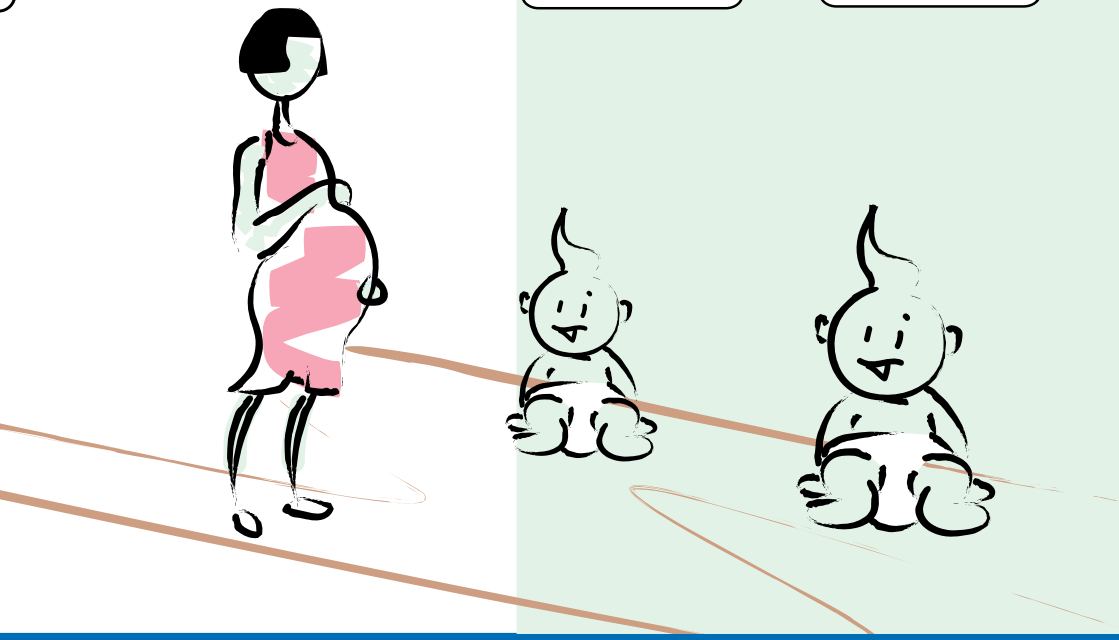
Physical Examination by 8 weeks
Check

24 25 26 27 28 29 30 31 32 33 34 35 36 BIRTH +1 +2 +3 +4 +5 +6

Newborn Screening Info
Read & discuss

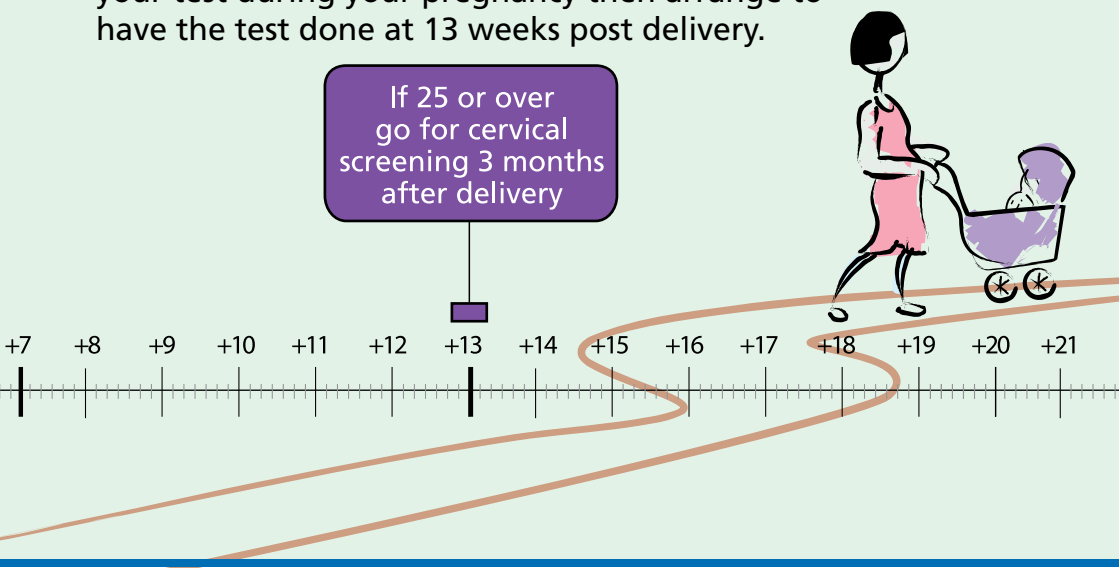
Newborn Blood Spot
Blood test

Newborn Hearing Screen
Check



In the busy time after the baby is born remember to look after your own health too.

If you are not up to date with cervical screening or you missed your test during your pregnancy then arrange to have the test done at 13 weeks post delivery.



Contacts

Your midwife's contact details

Name:

Address:

Phone number:

Your doctor's contact details

Name:

Address:

Phone number:

Using this booklet

It would help if you have this booklet handy when you see health professionals at home or in hospital.

This booklet is about the screening tests you will be offered while your are pregnant and screening tests for your baby in their first few weeks.

It is important you understand the purpose and possible results of the screening tests before you decide whether to have them. To help you the UK National Screening Committee has written this booklet, explaining the screening tests in detail.

The UK National Screening Committee gives the health departments of the four UK countries advice on all aspects of screening.

Please read this booklet as it will help prepare you for discussions with your midwife or doctor and will help you ask the questions that are important to you. It will be helpful if you have the booklet with you when you see them.

Some of the tests need to take place as early as 10 weeks in pregnancy, so you should read this booklet as soon as possible.

Towards the end of your pregnancy your midwife will talk to you about the screening tests recommended for newborn babies. You should look at this booklet again at this stage.

We hope you will find this booklet useful. Please keep it safe with your maternity notes.

For more information on screening tests, talk to your midwife, doctor or health visitor. You can also visit the following websites.

- For antenatal and newborn screening programmes: www.screening.nhs.uk/an
- For antenatal results and choices charity: www.arc-uk.org
- For comments about people's experiences: www.dipex.org/antenatalscreening
- Contact a Family charity: www.cafamily.org.uk

Screening for sickle cell and thalassaemia in early pregnancy

In the first few weeks of your pregnancy, we will offer you a blood test for sickle cell and thalassaemia. This chapter describes the screening process. It explains why we offer the test and helps you decide whether to accept it.

What are sickle cell and thalassaemia disorders?

Sickle cell disease and thalassaemia major are serious, inherited blood disorders. 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.

Sickle cell disease

People with sickle cell disease:

- can have attacks of very severe pain
- can get serious, life-threatening infections
- are usually anaemic (which means that their bodies have difficulty carrying oxygen), and
- need medicines and injections when they are children and throughout the rest of their lives to prevent infections.

Thalassaemia major

People with thalassaemia major:

- are very anaemic (their bodies have difficulty carrying oxygen)
- need blood transfusions every four to six weeks, and
- need injections and medicines throughout their lives.

There are also other, less common, haemoglobin disorders. Many of these are not as serious.

How are they passed on?

Sickle cell and thalassaemia are inherited disorders that are passed on from parents to children through unusual haemoglobin genes.

People only have these disorders if they inherit **two** 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 disorders.

But if a carrier has a baby with someone else who is also a carrier (or who has one of the disorders), there is a chance that their baby could inherit a disorder.

Who can be a carrier?

Anyone can be a healthy carrier. But you are more likely to carry the unusual genes if your ancestors came from places where malaria has been common. This is because being a carrier can help to protect people against malaria.

This means you are more likely to be a carrier if your ancestors came from the Mediterranean (for example Cyprus, Italy, Portugal, Spain), Africa, the Caribbean, the Middle East, India, Pakistan, South America or South and South-East Asia.

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

Father who came forward for testing

What tests are involved?

Screening involves a simple blood test. Ideally the best time to have the test is before you are 10 weeks pregnant.

All pregnant women are offered a blood test for thalassaemia. But you will not always be offered a blood test for sickle cell. You may be given a questionnaire to find out where your family – and the family of your baby's father – come from. If this shows you are at low risk, you may not be offered the blood test for sickle cell. But you can always ask for the test if you want it.

Why should I be tested?

The test gives important information for your baby's health

- If the blood test shows that you are a carrier, we will invite your baby's father for a test. If he is also a carrier, your baby has a chance of inheriting a disorder. (The diagram on page 9 shows the different chances for your baby. These include inheriting the disorder, being a carrier or not being affected.)

  Finding this out early in your pregnancy gives you the chance to talk to a counsellor and find out more about the disorders and the care available. If you want to, you can have another test to confirm whether your baby has one of the disorders. (See 'Is there a further test?' on page 10).

The test can benefit you and your family

- 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 themselves.
- Although people who carry sickle cell are healthy, they can experience some problems in rare situations where their bodies might not get enough oxygen (for example, when having an anaesthetic or during deep-sea diving). Knowing that you carry sickle cell can help you manage these situations.

People who carry thalassaemia or other unusual haemoglobin genes do not experience these problems.

For all of these reasons, we **strongly recommend** that you have the screening test. However, you can choose not to be tested, and we will respect your choice at all times.

Are there any risks?

Screening is a simple blood test, with almost no risk to you or your baby.

How will I get my results?

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

What will the results tell me?

The most likely result is that you are not a carrier. Your pregnancy should continue as normal.

If the result shows that you are a carrier for sickle cell, thalassaemia or another haemoglobin disorder, we will offer you counselling to talk about what this could mean for you, your baby and your family. We will also offer your baby's father a test to find out whether he is a carrier.

In very rare cases, the test may show that you have a haemoglobin disorder without knowing it. A health professional (for example, a nurse, doctor or midwife) will discuss your options with you, including the care you will need while you are pregnant.

Although the test is between 95% and 99% accurate, in a small number of cases the result may be unclear. If this happens, we will usually offer you another test.

Why should my baby's father have a test?

Babies can only inherit the disorders if **both parents** carry the unusual gene. So, if you are a carrier, it is important to find out whether the baby's father is also a carrier.

If he is not available or does not want to have a test, we may offer another test to find out whether your baby has sickle cell or thalassaemia. (See 'Is there a further test?' on page 10).

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

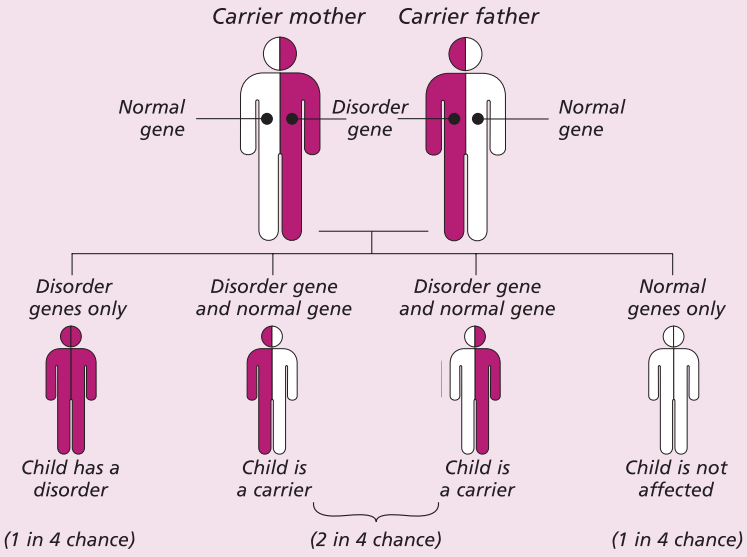
If you and the baby's father both carry the gene for sickle cell, thalassaemia or another haemoglobin disorder, for each baby you have there is:

- a 25% (one in four) chance that your baby will not be affected (that is, it will not have or carry a disorder)
- a 50% (two in four) chance that your baby will be a carrier, and
- a 25% (one in four) chance that your baby will have a disorder.

This is shown in the diagram below.

We will offer you counselling to discuss what this means for your family and what choices you want to make.
If you want, you can choose to have another test to find out if your baby has sickle cell or thalassaemia or another haemoglobin disorder (see 'Is there a further test?' on page 10).

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



"The lady at the centre was really, really helpful. She was so nice and she talked us through everything and the chances of us having a sickle cell baby and everything. And then I made up my mind I was going to have those tests done..."

A pregnant woman who sought counselling about being a carrier

Is there a further test?

If both you and your baby's father are carriers - or if your baby's father does not come for testing - you can choose to have another test while you are pregnant. This is called a 'diagnostic test'. It will show whether your baby has a disorder.

A health professional will explain the different types of diagnostic test and help you decide whether you want the test. If you do want the test, it is important to have it as early as possible in your pregnancy.

If the test shows that your baby has sickle cell, thalassaemia or another haemoglobin disorder, we will offer you more counselling. This will help you to think about what this may mean for your family, the care that is available, and whether you want to continue with your pregnancy.

Testing for new babies

As well as the tests described in this leaflet, all babies in England are offered a heel-prick blood test when they are five to eight days old. This test is done by taking some blood from your baby's heel. It tests for a number of conditions including sickle cell. It will show whether your baby is not affected, is a carrier, or has a disorder. This is called newborn blood-spot screening and a health professional will give you more information later in your pregnancy.

Questions?

If you have any questions about the test, or anything else in this leaflet, please discuss them with your GP, midwife, hospital doctor or specialist counsellor. They will be able to give you advice. They may also have information about other organisations which can give you support. We have listed some of these on the next page.

More information

Visit the website of the NHS Sickle Cell and Thalassaemia Screening Programme at **www.screening.nhs.uk/sickleandthal**
Read about people's real-life experiences of sickle cell and thalassaemia screening like the ones shown in this leaflet.
Visit **www.dipex.org/sicklecellandthalassaemia**

Other organisations

Sickle Cell Society
54 Station Road,
London NW10 4UA
Phone: 020 8961 7795
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**

UK Thalassaemia Society
19 The Broadway, Southgate Circus
London NI4 6PH
Phone: 020 8882 0011
Email: office@ukts.org
Website: **www.ukts.org**

"I think people should definitely ask to have the screening because if both parents are carriers, and if the child does have the disorder, it can have very serious consequences. It would be better to know about your options in advance."

Sarah, who accepted screening when she became pregnant.

Testing for infections in pregnancy

Testing for infections

At your first antenatal visit you will be offered and recommended tests for hepatitis B, Human Immunodeficiency Virus (HIV), rubella and syphilis. The tests can all be done from one blood sample, and are offered to help protect the health of you and your baby. Although the infections may not make you feel ill, if they are not detected they can cause serious damage to your baby. If we know about them, you can receive special care or medicine to reduce the risk of damage to you or your baby. It is better to have the tests as early as possible, but they can be done at any time during your pregnancy. If you decide not to have any of these tests, it will not affect the rest of your care in any way. Your midwife will tell you how the results of these tests will be given to you. If a test result suggests that you have an infection, a second test will be done to check the result. Having any of these tests does not affect your chance of getting certain insurance or the cover provided. It is only if you are actually found to have HIV, syphilis or hepatitis B infection that future insurance cover might be affected.

Confidentiality

The results of these tests will be kept confidential. Your needs and information about you will not be given to anyone outside the health-care team without your permission. Some information is gathered for essential public health purposes. However, those receiving this information will not be able to identify you from the information.

Hepatitis B

If you are infected with hepatitis B, your baby is at risk of being infected when you give birth. An infected baby has a high risk of carrying the hepatitis B virus for life. About a quarter of babies who are infected in this way develop serious liver disease in later life. If you are a carrier of hepatitis B, your baby can receive a course of hepatitis B vaccine which begins soon after birth. It is important that your baby has the full course of vaccine for it to be effective in reducing the baby's risk of becoming a carrier. The baby can have a blood test to check the course of vaccine has been effective. Hepatitis B is an infection of the liver caused by a virus. You can have the virus but feel well. Most women who have hepatitis B probably caught it from their own mother at birth. You can also become infected through unprotected sex (that is, having sex without using a condom) with someone who is already infected, or by sharing needles when injecting drugs, or by having a tattoo or body piercing with needles which have not been sterilised. All blood transfusions in the UK are screened for the virus and so it cannot be passed on in this way. But we cannot be so sure for unscreened blood transfusions abroad or for medical procedures using unsterilised equipment. Although nine out of 10 adults who catch hepatitis B recover fully from the infection, up to one in 10 will carry the virus for life. If you are found to have hepatitis B infection, your doctor should offer the test to your partner and any other children you have so that they can be offered a vaccination if it is appropriate. You may also be referred to a doctor who specialises in this condition.

Human Immunodeficiency Virus (HIV)

The HIV test is offered and recommended to all pregnant women because if you have HIV, you can pass the virus on to your baby while you are pregnant, when you give birth or by breastfeeding. If you have HIV infection you can receive special care and medicine to greatly reduce the chances of your baby becoming infected. The treatment may also help to keep you in better health. HIV (Human Immunodeficiency Virus) is the virus which causes AIDS (Acquired Immune Deficiency Syndrome). The virus gradually weakens the immune system, which makes it difficult to fight off infections. The most common way of becoming infected with HIV is by having unprotected sex (that is, sex without using a condom) with someone who already has HIV.

You can also become infected with HIV if you share needles when injecting drugs, or have a tattoo or body piercing with needles which have not been sterilised. All blood transfusions in the UK are now screened for HIV. The HIV test looks for antibodies to HIV. If the test is negative, it usually means that you are not infected with HIV. However, it can take up to three months for antibodies to develop, so if you think you may have been at risk of catching HIV recently, it is important to discuss this with your midwife. If you are found to be infected with HIV, your doctor can discuss offering a test to your partner and any children you have so that they can receive the necessary care if appropriate.

Syphilis

Although syphilis is rare in the UK, testing is recommended during pregnancy because of the serious damage it can do to you and your baby. It can be cured by treatment with antibiotics (usually penicillin), which will also treat infection in the unborn baby. If you do have syphilis, your baby will also be treated with antibiotics after birth to make sure he or she is not infected. Although most people who have syphilis are only ill for a short time, it can eventually have very severe effects (such as brain damage) if it is not treated. If it is passed to the unborn baby it often results in miscarriage or stillbirth, or the baby being born early and severely ill. Almost all syphilis infections in the UK are caught by having unprotected sex, with a person who is infected. If you have syphilis, your partner should also be offered a test.

Rubella (German Measles)

The test for rubella is different from the other tests described in this leaflet. It is not to see if you are infected in pregnancy but to see whether you are protected against rubella. If you are not protected, you will be offered the MMR vaccine (which protects against Measles, Mumps and Rubella) after your baby is born to protect you in future pregnancies. Rubella is spread very easily from person to person as the virus is carried in the air. It is usually a mild disease, and it is not always possible to tell if someone has it. However, if you catch rubella in the first 12 weeks of pregnancy, your baby is at very high risk of being born with serious defects of the brain, heart, eyes, and hearing. The vaccine is very effective in protecting against rubella, but about one in 20 people who have had the vaccine will not be fully protected. So even if you have been immunised in the past, it is still important to have the blood test, and it is particularly important if you have never been immunised before.

Testing for Down's syndrome in pregnancy

Choosing whether to have the tests is an important decision, for you and for your baby.
This chapter gives you some information about Down's syndrome and about testing for it, so you can decide whether to have the tests.

- **All pregnant women in England are now offered tests for Down's syndrome. This booklet gives you some information about Down's syndrome and about testing for it, so you can decide whether to have the tests.**
- **Choosing whether or not to have these tests is an important decision for you and your baby. You need to make the decision that is right for you, so please read this booklet carefully.**
- **Your midwife or your GP will talk to you about testing for Down's syndrome. They will be happy to answer your questions – so please do ask if there is anything you are not clear about.**

Data protection and guaranteeing quality in the Down's syndrome screening programme

One of the aims of the screening programme is to make sure that it meets set quality standards and guidance. To do this, hospitals may need to use information about your screening choices to help improve the programme and to tell NHS planners about any extra funding they need. We will store personal information under the Data Protection Act 1999. By law all NHS staff must follow the Act and keep your information confidential.

We will only keep your personal information for as long as it is necessary for monitoring the screening programme. We will not give it to anyone outside the NHS. If you want to discuss any concerns you have about how we will keep the information, please ask your midwife.

If you do not want your personal information stored or used for monitoring please inform your midwife or doctor.

What is Down's syndrome?

There is no such thing as a typical person with Down's syndrome. Like all people, they vary a lot in appearance, personality and ability. People with Down's syndrome have learning difficulties. Some have more serious difficulties than others. It is hard to tell in babies how much they will be affected as children, or when they are grown up. Some adults with Down's syndrome are able to get jobs and live fairly independent lives. However, most people with Down's syndrome need long-term help and support.

A number of health problems are linked to Down's syndrome. But again, people vary, and some people with Down's syndrome enjoy good health. Problems which are linked with Down's syndrome include heart problems and reduced hearing and vision. Many of the problems can be treated, and frequent health checks can make sure that any problems are picked up as early as possible. Most people with Down's syndrome live to be 50 years of age and some live to be over 70. Alzheimer's disease (a form of senile dementia) may affect people with Down's syndrome at an earlier age than other people.

Anyone can have a baby with Down's syndrome.

How common is Down's syndrome?

People do not usually expect to have a baby with Down's syndrome. It does not usually run in families. Some people think that only older women can have a baby with Down's syndrome, but this is not true. Anyone can have a baby with Down's syndrome, but the risk does go up with age.

The older a mother is the more chance she has of having a baby with the condition. For example, the chance of having a baby with Down's syndrome is one in 1500 for women who are 20 years old, one in 900 for women who are 30 years old, and one in 100 for

women who are 40 years old. Some people prefer to think of the risk as a percentage. A risk of one in 1500 is the same as a 0.07% risk. A risk of one in 900 is the same as a 0.1% risk. A risk of one in 100 is the same as a 1% risk.

What causes Down's syndrome?

Inside all of the cells of our bodies there are tiny structures called chromosomes. These chromosomes carry the genes that determine how we develop. Most people have 23 pairs of chromosomes in each of their cells. When our bodies produce the special cells needed to make babies, the chromosome pairs divide and rearrange themselves. Sometimes these pairs of chromosomes do not divide correctly, and this causes the baby's cells to have an extra copy of chromosome number 21. This causes Down's syndrome, (and it is the reason why one of the medical names for Down's syndrome is Trisomy 21). The extra chromosome cannot be removed from the cells, so there is no cure for the condition.

If the chromosomes divide incorrectly, this happens by accident. It is not caused by anything parents have done or have not done.

Testing for Down's syndrome during pregnancy

Should I have the tests for Down's syndrome?

Only you can decide that. Some women want to find out if their unborn baby has Down's syndrome and some do not. Information about the tests and how they work can help you make up your mind. This booklet gives the main facts and tells you how you can get more information if you want to know more.

A screening test carries no risk of miscarriage.

Will the tests tell me for certain if my baby has Down's syndrome?

We do not offer all women a test that will tell them for certain. This section explains why.

There are tests which give definite information. These are called **diagnostic tests.** The problem is that having a diagnostic test increases your risk of miscarriage. This is why we do not offer diagnostic tests to all women. Instead, we offer tests in two stages. We begin by offering all women a test that carries no risk of miscarriage. This type of test is called a **screening test.**

Screening tests **do not** give a definite answer, but they do tell us which babies have a higher risk (increased risk) of having Down's syndrome. We then offer diagnostic tests to the women at higher risk (increased risk). (There is more information on diagnostic tests later in the booklet.) If your screening result shows that you are at higher risk (increased risk), then you will be offered a diagnostic test. It is very important to understand that screening tests cannot tell you whether your baby definitely does or definitely does not have Down's syndrome.

We only use screening tests, and the two-stage process (the screening tests first and then the diagnostic tests), because we do not have a risk-free diagnostic test to offer. If you decide to have a screening test, and we later offer you a diagnostic test, it is your choice whether or not to have that diagnostic test. We explain more about the two-stage process later in the booklet.

Screening tests do not give definite answers.

What information does a screening test give me?

All of the tests described later in this booklet give results in the form of 'one in ...', for example, 'one in 100' or 'one in 1500'. (The same results can also be given as percentages – see page 18.) These numbers tell us

how likely it is that your baby has Down's syndrome. For example, the result 'one in 100' means that there is one chance in 100 that your baby has Down's syndrome. The result 'one in 1500' means that there is one chance in 1500 that your baby has the condition. It is very important to understand that as the second number in the result gets **bigger,** the chance of your baby having Down's syndrome gets smaller.

Next we explain how we use the results from the screening test to decide whether to offer you more tests.

Women who are not offered more tests

If the screening test shows that the risk of your baby having Down's syndrome is lower than the recommended national cut-off, we will not offer you a diagnostic test.

Most screening test results (above 95%) fall into this category. This is known as having a 'low-risk' result. It is important to understand that a low-risk result means exactly that. It does not mean that there is no risk at all that your baby has Down's syndrome, just that it is unlikely.

There is still some risk of your baby having Down's syndrome because some babies with Down's syndrome are not detected by screening tests. This happens if the expected pattern is not seen in the blood test or in the scan measurements because of natural variations. Overall, about a quarter of babies with Down's syndrome are not detected by screening tests.

Women who are offered more tests

If the result of the screening test shows that the risk of your baby having Down's syndrome is greater than the recommended national cut-off we will offer you a diagnostic test. The screening result that leads us to

offer you more tests is sometimes known as the 'higher risk' or 'increased risk' result.

Overall about one in 30 (3%) of women screened have a higher (increased) risk result and are offered a diagnostic test. It is very important to remember how screening tests fit into the two-stage process. If you get a higher risk (increased risk) result from a screening test it means that we will offer you more tests. It does not mean that your baby definitely has Down's syndrome. As we explain later, most of the women who are offered further tests learn that their baby does not have Down's syndrome.

About a quarter of babies with Down's syndrome are not detected by screening tests.

What screening tests will I be offered?

Screening for Down's syndrome is offered to pregnant women of all ages. The tests can provide information about the chance of a baby having Down's syndrome. These tests use blood samples taken from the mother, measurements taken from ultrasound scans or both to work out this chance. The tests you will be offered depend on how many weeks pregnant you are.

All tests offered must meet the national standards. To ensure this happens we audit the data (see page 16 for more information).

The combined test offered in early pregnancy

The combined test uses the results of a blood test and an ultrasound scan to calculate the risk (chance) of the unborn baby having Down's syndrome.

A blood sample taken from the mother between 10 weeks to 13 weeks and 6 days of pregnancy is used to measure the amount of some substances that

are found naturally in the mother's blood. These substances are passed to the mother from the baby.

An ultrasound scan is carried out between 11 weeks and 0 days to 13 weeks and 6 days of pregnancy. This scan measures the amount of fluid lying under the skin at the back of the baby's neck. This is called the nuchal translucency (NT) measurement.
A computer program then uses the results from the blood sample combined with the NT measurement to work out a risk (chance) figure. In addition to the results from the blood sample and the NT measurement, the program also uses the mother's age, weight, weeks of pregnancy, family origin and smoking details to work out this risk (chance) figure.

Screening later in pregnancy

If it has not been possible to have the combined test in early pregnancy, you will be offered a blood test between 15 weeks and 0 days to 20 weeks of pregnancy. This test looks at different substances to those measured in early pregnancy. Like the combined test, a computer program uses the results and the mother's details to work out a risk (chance) figure. This test is known as the quad (or quadruple) test.

What happens next if I have a higher risk (increased risk) result and I am offered more tests?

Your midwife or doctor will discuss the results with you and answer any questions that you have. You will be offered a diagnostic test which can tell you definitely whether your baby has Down's syndrome or not. There are two diagnostic tests available – chorionic villus sampling (CVS) and amniocentesis. CVS can be performed from weeks 10 to 22 of pregnancy although it is usually performed between weeks 11 and 13. Amniocentesis is usually carried out from week 15 of pregnancy.

If you do get a higher risk result from a screening test, your midwife or doctor will give you information and support. You will also have time to make up your mind about what to do next.

If you are in this position it is important to understand that you have a difficult decision to make.

You have two options. You can decide not to have a diagnostic test. This means spending the rest of your pregnancy knowing the screening result, which might be stressful.

The only other option is to have the diagnostic test, knowing that this will slightly increase the risk of miscarriage. You need to think carefully about what you would do if you found yourself in this position. Once you know the result of the screening test, you can't put the clock back. If you would not be happy with either of the above options, you need to consider very carefully whether it would be better for you not to have the screening test in the first place.

Once you know the result of the screening test, you can't put the clock back.

How will I get the result from my screening test?

At the time of your test, ask your midwife or doctor how you will get the results. The test results should be available within two weeks.

What happens if I get a low-risk result, so I am not offered more tests?

Although your risk is low, you may want to discuss your results with your midwife or doctor.

Diagnostic tests for Down's syndrome

What can you tell me about diagnostic tests?

This booklet just gives some basic facts to help you decide whether you would want to have any screening or diagnostic tests you are offered. If you are actually facing a decision about diagnostic testing, your doctor or midwife will give you more detailed information.

What is chorionic villus sampling (CVS)?

CVS is a test carried out during pregnancy that involves removing a small piece of tissue from the placenta. It will generally be carried out between weeks 10 and 22 of pregnancy but is usually done between weeks 11 and 13. Sometimes it is carried out later. It is usually only offered in a specialist centre. The test itself takes around 10 minutes.

Immediately before the test, your abdomen is cleaned to make sure that the test can take place in the most sterile conditions possible. During CVS a

sonographer puts gel on your abdomen. You will then have an ultrasound scan to check the position of your baby. The sonographer or doctor will keep scanning you throughout the procedure. A fine needle is then inserted either through your vagina or through your abdomen and into your womb. A tiny sample of tissue is then removed from your placenta. Your placenta will usually contain tissue that is genetically identical to your baby.

The sample is analysed in the laboratory and the baby's chromosomes are counted. Very occasionally (about two in 100) CVS samples do not produce a result.

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

About one in 30 women screened are offered a diagnostic test.

What is amniocentesis?

Amniocentesis is a test carried out during pregnancy which involves using a fine needle to remove a small amount of the amniotic fluid around the unborn baby. It is a widely used procedure which usually takes about 10 minutes.

Amniocentesis is usually carried out between weeks 15 and 18 of pregnancy. However, the test can be done later in pregnancy.

Immediately before the test, your abdomen is cleaned to make sure that the test can take place in the most sterile conditions possible. During the amniocentesis, a sonographer puts gel on your abdomen. You will then have an ultrasound scan to check the position of your baby. The sonographer or doctor will keep scanning you throughout the procedure. A fine needle is then inserted through your skin, through your abdomen and into your womb. The needle is used to remove a small sample of the amniotic fluid surrounding your baby.

This fluid contains cells from the baby, which will be examined at the laboratory and the baby's chromosomes counted. About one in every 100 samples does not produce a result because the cells do not grow or the results are not clear. If this happens, you will be offered a second amniocentesis.

Are these procedures safe?

These procedures are not completely safe, and this is why we don't offer them to everybody. The overall risk of you having a miscarriage after CVS is about 1 to 2%. In other words, about one or two in every 100 women who have CVS will miscarry. For amniocentesis, the rate is about one in 100. These figures vary slightly from hospital to hospital. If you would like to know the miscarriage rates after CVS or amniocentesis in your hospital, please ask your doctor or midwife.

Are the tests painful?

Many women find the procedures uncomfortable but they should not be painful. For a day or two afterwards you will be advised to take things easy. If possible, you should avoid activities that involve lifting, bending or stretching. You may have some discomfort in your lower abdomen for a day or two after the procedure. This is normal, and you can take paracetamol to relieve the discomfort. Remember, you can only take a maximum of eight tablets in 24 hours. If you are worried about taking painkillers or have any questions, you should talk to your doctor or midwife.

How long does it take to get the results?

It can take up to 18 days to get the results of diagnostic tests. Some hospitals offer a 'molecular test' as part of the diagnostic test. This is usually known by its initials –

PCR. This test provides some information within two to three days.

Waiting for the results can be an anxious time. Do call your midwife or contact one of the support organisations listed on pages 29-31 if you need to talk.

How will I get my results?

It is important that you talk to your midwife about how you want to receive the results. The chances are that your results will show that your baby does not have Down's syndrome, but if the results show that your baby does have Down's syndrome, you need to think carefully about how you want to hear this information. Your midwife can give you the results at your home, at the antenatal clinic, over the phone or in a letter. Discuss what is appropriate for you with your midwife.

For every 100 women who have amniocentesis one will miscarry.

What are the possible results from diagnostic tests?

The baby does not have Down's syndrome

This is the result that most women get. Some women are happy just to get this news. They do not want to talk about tests and test results any more. Others may want to discuss the results with somebody. They want to know how the two tests they have had – the screening test and the diagnostic test – can seem to say different things. If you want more information, we give a very brief explanation on page 28. If you really don't want to read any more details, just remember, the diagnostic test gives the definite result.

We explained earlier that we use screening tests to decide who should be offered a diagnostic test. What the screening tests do is tell us if there is a certain sort of pattern in your blood test or ultrasound results. This pattern is one that can sometimes be seen when a baby

has Down's syndrome. The problem is that the same pattern can also be seen in many normal pregnancies.

Women's blood test and scan results vary for all sorts of normal reasons. Screening tests just detect the pattern, they don't tell us the reason for the pattern. Only diagnostic tests can tell us if the reason for the pattern is that the baby has Down's syndrome. When a woman has a diagnostic test and the result shows her baby does not have Down's syndrome, that woman's earlier screening test result is sometimes called a **'false positive'** result. If this happens and you feel confused or upset about it, please talk to your midwife or doctor.

The baby does not have Down's syndrome, but the tests have shown some other problem

The main purpose of a CVS or amniocentesis is to find out whether a baby has Down's syndrome. But when the baby's chromosomes are examined, very occasionally other chromosome variations are identified. Some of these chromosome variations can be serious, and others will have only a minor effect, or no effect, on the baby. If the tests show there is a problem, you will be referred to a genetic counsellor for specialist information and support.

The baby has Down's syndrome

Most women who have a diagnostic test, learn that their baby does not have Down's syndrome.

A small number of women who have a diagnostic test will learn that their baby has Down's syndrome. They then have three options, and it is entirely the parents' decision which they choose. Some people will decide to continue with the pregnancy, make plans and prepare for any extra challenges they might face bringing up a child with Down's syndrome. Some people may feel unable to bring up their child themselves, and decide on adoption. Some people

will decide they do not want to continue with the pregnancy, and will choose to have a termination. If you are faced with this decision, you need to make sure you reach the right decision for you. We will give you information and support to help you make your decision, but it is up to you to decide what will be best for you. You will have the opportunity to discuss your options with healthcare professionals, and you will also be offered information and support from outside the health service. You will have time to decide what you are going to do and your GP and midwife will support you in your decision.

The diagnostic test gives the definite answer.

You can get more information about screening from the following organisations:

Antenatal Results and Choices (ARC)
www.arc-uk.org
73 Charlotte Street
London
W1T 4PN
Helpline: 0207 631 0285
Email: info@arc-uk.org
Antenatal Results and Choices (ARC) provides impartial information and individual support to parents whether they are going through antenatal screening or whose unborn baby has been diagnosed with an abnormality.

You can get more information about Down's syndrome from the following organisations:

Contact a Family (CAFAMILY)
www.cafamily.org.uk
209-211 City Road
London
EC1V 1JN
Helpline: 0808 808 3555
Email: info@cafamily.org.uk
Contact a Family is a charity which provides support, advice and information for families with disabled children, no matter what their condition or disability.

Down's Syndrome Association
www.downs-syndrome.org.uk
Langdon Down Centre
2a Langdon Park
Teddington
TW11 9PS

Helpline: 0845 230 0372
Email: info@downs-syndrome.org.uk

The Down's Syndrome Association is a support group for parents and carers of people with Down's syndrome.

Down's Syndrome Medical Interest Group
www.dsmig.org.uk
Children's Centre
City Hospital Campus
Hucknall Road
Nottingham
NG5 1PB

Telephone: 01159 627658 extension: 45667
This website is aimed at health professionals but includes information that parents may find helpful.

NHS Fetal Anomaly Screening Programme
www.fetalanomaly.screening.nhs.uk
Innovation Centre
Rennes Drive
University of Exeter,
Devon,
EX4 4RN
Telephone: 0845 527 7910
Email: enquiries@ansnsc.co.uk
The NHS Fetal Anomaly Screening Programme is responsible for both the NHS Fetal Anomaly Ultrasound Screening Programme and the Down's syndrome Screening Programme for England.

References

Newton R. *The Down's Syndrome Handbook: A Practical Guide for Parents and Carers.* Random House, London, 2004.

Rondal J, Perera J, Nadel L. *Down's Syndrome: A Review of Current Knowledge.* Whurr Publishers, London, 1999.

Selikowitz M. *Down Syndrome The Facts,* 3rd edn. Oxford University Press, New York, 2008.

UK National Screening Committee. *Fetal Anomaly Screening Programme – Screening for Down's Syndrome: UK NSC Policy Recommendations 2007–2010: Model of Best Practice.* Department of Health, London, 2008.

Having a mid-pregnancy ultrasound scan?

This scan (sometimes called the 18^{+0} to 20^{+6} weeks fetal anomaly scan) is offered as part of the NHS 'fetal anomaly' ultrasound screening programme. This chapter gives you some information about the purpose of the ultrasound scan in mid-pregnancy between 18 weeks and 0 days to 20 weeks and 6 days, so you can decide whether or not to have this examination.

What is the purpose of a mid-pregnancy ultrasound scan?

An ultrasound scan is an important examination.

Why am I offered a mid-pregnancy scan?

The main purpose of this scan is to look for abnormalities in your unborn baby.

Is the mid-pregnancy scan safe?

As far as we know, the scan we offer is safe for mothers and babies.

What kind of scan will I be offered?

You will be offered a scan that produces a two-dimensional (2-D) black and white image that gives only a side view of the baby. The 3-D and colour images we sometimes see on television and in magazines are not used in the NHS screening programme.

Does everybody have a scan?

We offer the scan to everybody, but you do not have to have it if you do not want to. Before making up your mind, there are a few things you do need to know, so please read this section carefully.

Giving permission for the scan

Remember that this is a medical examination. You will be asked to give your permission for it to be carried out. Make sure you understand what is going to happen. Feel free to ask any questions.

What can a scan tell me about my baby's health and development?

Before deciding whether or not to have a scan, you need to know what scans can and cannot tell you. During the scan, we take a very close look at your baby. We usually learn that the baby appears to be healthy and is developing well, but sometimes we find a problem. If this happens it will be explained to you.

The table on pages 34-35 shows the 11 problems the NHS Fetal Anomaly Screening Programme (FASP) is audited against. Your sonographer will be looking specifically for these problems during the scan.

Some problems are quite common, others are very rare and they will be explained to you.

What are the chances that we will be able to see it on an ultrasound scan?

Problem	Description	Chance of being seen
Anencephaly	This is abnormal development of the brain and the bones of the skull. Sadly, babies with this problem cannot live once they are born and die soon after birth.	98%
Open spina bifida	Spina bifida is when your baby's spinal cord has not developed properly and there is a gap or split in the spine.	90%
Cleft lip	This happens when certain parts of your baby's face and particularly the lips do not join together properly.	75%
Diaphragmatic hernia	This occurs when your baby's diaphragm does not fully form. The diaphragm is a muscle that helps us breathe and it keeps the heart and lungs separate from the bowel and the rest of the organs in the abdomen.	60%
Gastroschisis	This is a defect or 'hole' in the baby's abdominal wall to one side of the umbilical cord (usually the right side). Some of the bowel escapes through this hole and develops outside of the baby's abdomen.	98%

Problem	Description	Chance of being seen
Exomphalos	Exomphalos occurs when the abdomen fails to close around the base of the umbilical cord during the early development of the baby. This means some organs develop on the outside of the baby's abdomen.	80%
Serious cardiac abnormalities	These include a range of heart abnormalities which mean your baby will need medical attention very soon after birth.	50%
Bilateral renal agenesis	This means that your baby's kidneys have not developed. Sadly, babies with this condition die shortly after birth as they cannot live without kidneys.	84%
Lethal skeletal dysplasia	Lethal skeletal dysplasia is a problem which affects the size and shape of arms, legs, the body or sometimes the skull. The chest and lungs of these babies do not fully develop, which means that they do not survive.	60%
Edwards' syndrome (Trisomy 18)	This rare genetic chromosomal disorder occurs when a baby has three copies of chromosome 18 instead of the usual two.	95%
Patau's syndrome (Trisomy 13)	Trisomy 13 is a chromosomal disorder. It occurs when a baby has three copies of chromosome 13 rather than the usual complement of two.	95%

Scans are not guaranteed to find all problems. Sometimes we have to say there might be a problem, but we cannot say for certain. In a small number of cases, babies are born with abnormalities that were not spotted by the scan.

The rest of this section tells you what it's like to have a scan, and what happens if any kind of problem (or possible problem) is found. Remember that for most people their scan is a happy experience. Unfortunately though this is not true for everybody, which is why we ask you to read this section carefully before you decide whether or not you want a scan. You may find it useful to talk to your midwife before deciding. Remember though that most babies are healthy.

The scan usually takes around 30 minutes.

What is it like to have a scan?

Can I bring family or friends with me when I have the scan?

Most hospitals will recommend that you bring a partner, a friend, or a family member when you have the test because you might be anxious. We suggest that if possible, someone should accompany you to and from the hospital. Most hospitals do not allow children to attend the screening tests as childcare facilities are not usually available. Please ask your hospital for its policy on this before your appointment.

Remember, an ultrasound scan is an important medical examination, and it is treated in the same way as any other hospital investigation.

Do I need to drink water and have a full bladder before my scan?

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.

What will happen when I go into the scan room?

Most scans are carried out by specially trained staff called sonographers. In order for the sonographer to get good images of your baby, the procedure is carried out in a dimly lit room. You will first be asked to lie on a couch. You will then be asked to raise your top to your chest and lower your skirt or trousers to your hips. Tissue paper will be tucked around your clothing to protect it from the ultrasound gel, which will then be put on your tummy. (The gel makes sure there is good contact between the machine and your skin.) The sonographer then passes a hand-held device called a probe over your skin. It is this probe which sends out ultrasound waves and picks them up when they bounce back.

The sonographer will carefully examine your baby's body. Having the scan does not hurt, but the sonographer may need to apply slight pressure to get the best views of the baby. A black and white picture of the 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 the baby – either directly facing them or at an angle.

How long will my scan take?

A scan usually takes around 30 minutes. However, the sonographer may not be able to get good views if your baby is lying in an awkward position or is moving around a lot. If you are overweight, this can reduce the quality of the image, because there is more tissue for the ultrasound waves to get through before they reach the baby. If it is difficult to get a good image, the scan may take longer, or have to be repeated at another time.

If everything appears normal, what happens next?

Most scans show that the baby is developing normally, and no problems are found. This is because most babies are healthy and do not have abnormalities.

Will the sonographer tell me the sex of my baby?

Finding out the sex of your baby is not offered as part of the national screening programme, but this depends on the policy of your hospital.

Can I have a picture of my baby?

You will need to check if your hospital provides this service. If they do, there may be a charge.

Scan results and findings

Will I need another scan?

If everything appears normal, you will probably not need another scan.

Sometimes scan results can be uncertain and the sonographer may ask for a further opinion.

If the sonographer does not see everything clearly, perhaps because you are overweight or your baby is lying in an awkward position, you may have to have the scan repeated again on a different day. This happens quite often and doesn't mean the sonographer has seen anything to worry about. If the sonographer cannot get a good image of the baby after two separate attempts, you may not be offered another scan.

Will the scan say for certain whether or not there is a problem?

Not always. As we explained earlier, not every abnormality can be spotted by a mid-pregnancy scan. This means that in some cases, babies are born with abnormalities when no problem was identified by the scan.

What kind of problems can be seen?

Major structural abnormalities in the development of the baby such as spina bifida, are usually obvious on the scan and the sonographers and doctors can be absolutely certain of the findings. Scans are not so reliable at seeing problems such as some heart defects and we do not expect to pick up every heart condition before birth.

If a problem is found or suspected, you might be offered further tests.

Sometimes minor changes in the baby's body are seen. Usually these mean nothing at all, but sometimes we see a pattern which tells us there could be an underlying problem. Some minor problems may need follow-up care after the baby is born.

What will happen if a problem is found, or suspected, during the scan?

If any problem is found or suspected, the sonographer may ask for a second opinion from another sonographer or clinician. You would then be told what the concerns were, but the exact problem might not be clear at this stage.

If necessary, you will be referred to a specialist, possibly in another hospital. You should be given an appointment within a few days.

In most cases, further tests do not find a problem. However, any extra test can cause great anxiety for parents, and for some people the anxiety can last throughout the rest of the pregnancy.

You might be offered another test, such as an 'amniocentesis', to find out for certain if there is a problem. If you are offered further tests you will be given more information about these tests. You can then choose whether or not you want to have them.

You may want to ask questions and to talk about your worries with your own midwife or consultant. In many hospitals, a specialist screening midwife is available. Other sources of information and support are listed at the end of this section.

What will happen if a type of abnormality is definitely found?

This depends on the type of abnormality and how serious it is. Some abnormalities may turn out not to be serious and some get better on their own. In either of these cases you may be offered further scans throughout your pregnancy to monitor the condition.

Not every problem can be seen on a mid-pregnancy scan.

If the condition is serious someone will talk you through your options, which may include ending the pregnancy. If you need to make any decision, your midwife and the hospital team will give you time, support and information and they will respect your choice. Details of organisations and groups that can give you help and support are given on pages 41 and 42.

Can anything be done before the birth?

Finding out about a condition before birth can help parents prepare themselves, and sometimes it can help to plan treatment after the baby is born. For example, if your baby is known to have a problem that will need an operation soon after birth, such as the repair of a hernia in your baby's tummy, arrangements can be made to deliver your baby in a hospital where this can be done within the first few hours after birth.

Can the baby have an operation before it is born?

Unfortunately, only a very few problems can be treated in this way.

If you would prefer not to know, you need to think carefully about whether you should have a scan at all.

Who can I talk to if I have any questions or concerns about the mid-pregnancy scan?

You can contact your midwife or doctor and you can get more information about screening from the following organisations:

Antenatal Results and Choices (ARC) www.arc-uk.org
73 Charlotte Street
London
W1T 4PN
Helpline: 0207 631 0285
Email: info@arc-uk.org

Antenatal Results and Choices (ARC) provides impartial information and individual support to parents whether they are going through antenatal screening or whose unborn baby has been diagnosed with an abnormality.

Contact a Family (CAFAMILY)
www.cafamily.org.uk
209-211 City Road
London
EC1V 1JN
Helpline: 0808 808 3555
Email: info@cafamily.org.uk

Contact a Family is a charity which provides support, advice and information for families with disabled children, no matter what their condition or disability.

NHS Fetal Anomaly Screening Programme
www.fetalanomaly.screening.nhs.uk
Innovation Centre
Rennes Drive
University of Exeter
Devon EX4 4RN
Telephone: 0845 527 7910
Email: enquiries@ansnsc.co.uk

The NHS Fetal Anomaly Screening Programme is responsible for both the NHS Fetal Anomaly Ultrasound Screening Programme and the Down's syndrome Screening Programme for England.

References

In writing this booklet we referred to the following documents:

Bricker L, Garcia J, Henderson J, Mugford M, Neilson J, Roberts T, Martin MA. Ultrasound screening in pregnancy: a systematic review of the clinical effectiveness, cost effectiveness and women's views. *Health Technol Assess 2000; 4: 1–193.*

Kirwan D and the NHS Fetal Anomaly Screening Programme (FASP). *18^{+0} to 20^{+6} Weeks Fetal Anomaly Scan – National Standards and Guidance for England.* NHS FASP, Exeter, 2010.

National Institute for Health and Clinical Excellence (NICE). *Antenatal Care: Routine Care for the Healthy Pregnant Woman – Clinical Guideline 6.* National Collaborating Centre for Women's and Children's Health NHS, October 2003.

Royal College of Obstetricians and Gynaecologists (RCOG). *Ultrasound Screening for Fetal Abnormalities – Report of the RCOG Working Party.* RCOG Press, London, 1997.

Royal College of Obstetricians and Gynaecologists (RCOG). *Routine Ultrasound Screening in Pregnancy: Protocol, Standards and Training – Supplement to Ultrasound Screening for Fetal Abnormalities – Report of the RCOG Working Party.* RCOG Press, London, 2000.

An introduction to physical examinations of newborn babies and those aged six to eight weeks

What is the physical examination?

When your baby is born, the midwife will carry out some checks. You will then be offered a more detailed physical examination of your baby **within 72 hours of birth and again at six to eight weeks old.** These examinations include a screening examination to find those babies who may have a problem with their eyes, heart, hips and, in boys, testicles. Your baby will experience a lot of physical changes in the first two months of life and this is why the examination is repeated at six to eight weeks.

This section gives you information about:

- why the physical examinations are carried out;
- who will carry them out;
- where the examinations will be carried out;
- how the examinations are carried out;
- how to prepare for the examinations;
- what the results may mean for parents and babies;
- what happens after the examinations;
- where the results will be recorded; and
- where you can go for more information and advice.

Why should I have my baby examined?

The purpose of the examination is to identify babies more likely to have conditions that need to be investigated. However, screening will not always pick

up every problem. Some conditions may only become apparent after several weeks or months and a few may still not be found at the six to eight week check.

The physical examinations can help identify health concerns at an early stage. Most babies who have the physical examinations will be healthy and will not have any health problems. In some cases the findings may suggest a problem, but further investigations often show there is nothing to be concerned about. Most of the problems babies have are minor and do not need treatment.

Health professionals such as GPs, midwives or health visitors are happy to see parents who may have worries about the health and development of their babies.

For the small number of babies who do have serious problems, there are a lot of benefits of having these identified as soon as possible. Early treatment can improve the health of the baby and prevent disability. If further investigations or treatments are needed, an appointment with a specialist will be arranged.

It is recommended that you have your baby examined, but if you are not sure, discuss it with your midwife or other health professional. Also if you think your baby might not have been examined, speak to your midwife, health visitor or GP.

Who will do the examinations?

A doctor, midwife, health visitor or nurse will carry out the examinations. All health professionals carrying out the examination have been specially trained.

Where will the examinations be carried out?

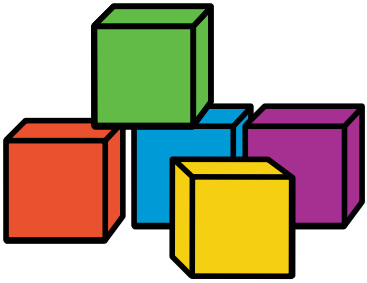
Depending on the health professional doing the examination, and the age of the baby, the examinations may take place in a hospital, GP's surgery, clinic, children's centre or at home.

How are the examinations carried out?

The health professional will introduce themselves and explain the examination. They will ask you about your pregnancy, and the birth, and will check your family history. They will also ask you about your own health and how you are feeling. This is an opportunity for you to talk about the general care of your baby and aspects such as feeding, crying or sleeping and to discuss anything that might be worrying you.

The examinations are normally done when your baby is calm and comfortable. The health professional will carry out an overall physical examination which includes a head-to-toe examination of your baby, looking at their development, feeding, weight, alertness and general wellbeing.

The health professional will look at your baby's eyes, heart, hips and, in boys, his testicles. They will listen to your baby's heart with a stethoscope and will look at your baby's eyes using an instrument called an ophthalmoscope.



Other parts of the examination involve gently handling your baby and moving their legs to check the hips. This should not hurt, although testing your baby's hips can sometimes be a little uncomfortable. If necessary, you can comfort your baby during and after the examination.

How can I prepare for the examinations?

You will be with your baby during the examination but you do not need to prepare anything special. Your baby will need to be undressed for part of the examination, but will be kept warm.

For the six to eight week examination it will be useful for you to think about the growth and development checklist in your baby's child-health record (sometimes known as the 'red book') before talking with your health visitor or doctor.

The checklist asks you to think about a number of questions and discuss them at the 6-8 week examination.

The checklist asks you to think about the following.

- Whether you feel well yourself
- Any worries you have about feeding your baby
- Any concerns you have about your baby's weight gain
- Whether your baby watches your face and follows it with his/her eyes
- Whether your baby turns towards the light
- Whether your baby smiles at you
- Whether you think your baby can hear you
- Whether your baby is startled by loud noises
- Any problems you have looking after your baby
- Whether you have any worries about your baby

What the results may mean for parents and babies

This section contains general information about conditions that may be found by the physical examination. It is not possible to go into detail here about further referrals or treatments in this booklet.
If there is a problem, what happens next will depend on what has been found during the examination. The health professional will be able to discuss this with the parent. Most babies will benefit from the treatments available.

As well as an overall physical examination, the following four screening examinations will be carried out.

Eyes:

The health professional will examine the baby's eyes, focusing on how they look and move. If the eye looks cloudy, this may mean the baby has a cataract and this may affect how well the baby can see. Babies who may have problems will be referred to an eye specialist (an ophthalmologist). About two or three in 10,000 babies have problems eye that need treatment.

Heart:

A general examination of the baby's heart is done by listening with a stethoscope. Sometimes murmurs are picked up. This can be worrying for the parent. A murmur is an extra noise made by blood as it passes through the heart. Murmurs are common in babies and do not necessarily mean that there is a heart problem. In nearly all cases the heart is actually normal. If the health professional finds something that suggests there may be a heart problem, another examination and further tests will be arranged. Around one in 200 babies have a heart problem that needs treatment.

Hips:

Babies can be born with hip joints that are not formed properly. If untreated this can lead to a limp and joint problems. Babies who could benefit from further investigation may have an ultrasound scan of the hips followed by an appointment with a specialist to check the hips again. About one or two in 1,000 babies have hip problems that need treatment.

Testicles:

Baby boys will be checked to make sure their testicles are in the right place. It can take several months for them to drop down into the scrotum. If this does not happen, a specialist may advise a small operation when the boy is one or two years old. About one in 100 baby boys have problems that need treatment.

What happens after the examinations?

The health professional who does the examination will discuss the results with you immediately. If the examination shows that everything seems to be all right with your baby, there will be no need for any further action.

The examination may highlight concerns with your baby. If this is the case, the health professional will either ask to see you and your baby again, or will offer you an appointment with a specialist. The specialist will give you a detailed explanation about the concerns identified, any further investigations and possible treatment.

Waiting to see the specialist can be an anxious time. Don't hesitate to talk to your midwife, GP or health visitor about your concerns.

Where will the results be recorded?

The outcome of the examination carried out within the first 72 hours of birth will be recorded in both your maternity notes and in your baby's child-health record.

The outcome of the six to eight week examination will be recorded in your baby's child-health record. You need to keep this record safe and take it with you whenever you and your baby see a health professional.

Your baby's hearing screening test

In the first few weeks of your baby's life, you will be offered another routine health check for your baby – a hearing screening test. The test uses quick and simple methods to check the hearing of all newborn babies.

"The test is straightforward and quick. She slept right through it."
(Mother of screened baby)

Why screen my baby's hearing?

One to two babies in every 1000 are born with a hearing loss in one or both ears. This hearing screening test will allow those babies who do have a hearing loss to be identified early.

Early identification is known to be important for the development of the child. It also means that support and information can be provided to parents at an early stage.

No one in my family has a hearing loss. Does my baby still need to have the hearing screening test?

Yes. It is important to screen all babies. Most babies born with a hearing loss are born into families with no history of hearing loss.

When will the hearing screening test be done?

Your baby will be offered the hearing screening test within the first few weeks of life. The hearing screen is usually done before you leave the maternity unit. In some areas it will be carried out at home. Your midwife or the person that gave you this leaflet will be able to tell you where and when the screening test will happen. If your baby's hearing is not screened, ask your health visitor, midwife, local audiology department or family doctor to arrange an appointment.

Will the hearing screening test be painful for my baby?

No. It does not hurt and is not uncomfortable. The screening test will usually be done while your baby is asleep or settled.

What does the hearing screening test involve?

A trained hearing screener or your health visitor carries out the hearing screening test. They place a small soft tipped earpiece in the outer part of your baby's ear which sends clicking sounds down the ear. When an ear receives sound, the inner part, known as the 'cochlea' usually produces an echo. The screening equipment can pick up this response. This is called the Automated Otoacoustic Emission (AOAE) screening test. The AOAE screening test only takes a few minutes. You can stay with your baby while the screening test is done.

When will I get the results of the hearing screening test?

The results will be given to you at the time of the screening test. If you have any concerns or questions about your baby's result contact the hospital where the screening test was done or if your baby was screened at home, contact your health visitor.

What do the results mean if the hearing screening test shows a clear response from both of my baby's ears?

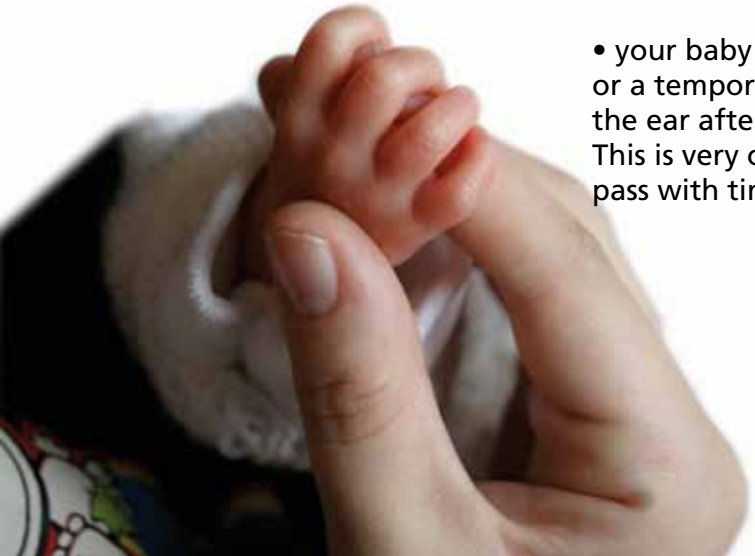
This means that your baby is unlikely to have a hearing loss. The NHS Newborn Hearing Screening Programme is a very reliable way of detecting hearing loss early.

Children can develop or acquire a hearing loss later on, so it is important to check your child's hearing as they grow up. Two checklists of the sounds that your baby should react to and the types of sounds they should make as they grow older are included on page 60. If you have any concerns about your child's hearing, discuss them with your health

visitor or family doctor. Your child's hearing can be tested at any age.

What do the results mean if the hearing screening test does not show a clear response from one or both of my baby's ears?

This means your baby needs a second hearing screening test.



Why does my baby need a second hearing screening test?

A lot of babies need to have a second hearing screening test because the first screen didn't show a clear response from one or both of the baby's ears. This does not necessarily mean that your baby has a hearing loss.

Some common reasons, other than hearing loss, for having a second screening test are:

- your baby may have been unsettled at the time of screening;
- there may have been background noise when the screening test was carried out; or
- your baby may have fluid or a temporary blockage in the ear after birth. This is very common and will pass with time.

Where will the second screening test be done?

Your baby's second screen will usually be done before you leave the hospital. In some areas it may be done in a local surgery or health clinic.

If your baby's first screening test was carried out at home, your health visitor will arrange a further appointment with you.

What does the second screening test involve?

The second screening test may be the same as the first screening test, the Automated Otoacoustic Emission (AOAE) screening test. Your baby may also have another type of screening test. This is known as the Automated Auditory Brainstem Response (AABR) screening test.

This involves three small sensors being placed on your baby's head and neck. Soft headphones, specially made for babies, are placed over your baby's ears and a series of clicking sounds are played.

The hearing screening equipment tells us how well your baby's ears respond to sound. The AOAE screening test takes a few minutes.

The AABR screening test can take between 5 and 30 minutes. You can stay with your baby while the screening test is done.

Neither of these screening tests will hurt or be uncomfortable for your baby. They will usually be carried out while your baby is asleep or is very settled.

What can I do to prepare my baby for the second screening test?

The screening test is easier to carry out if your baby is asleep. Don't worry if your baby will not settle. The Hearing Screener will understand that it is difficult to get a young baby to sleep. The following may help your baby to settle during the screening test.

• If possible, feed your baby shortly before the hearing screening test.

• Ensure you have the things that you may need to make your baby comfortable and happy.

My baby seems to be responding to sound. Does my baby need a second hearing screening test?

Most babies are found to have no hearing loss after the second screening test but it is still very important that your baby has the second screen. This is because babies who have a hearing loss will usually react to some sounds. If your baby does have a hearing loss it is important that you find out as soon as possible.

What do the results mean if the second hearing screening test shows a clear response from both of my baby's ears?

This means that your baby is unlikely to have a hearing loss.

Remember that children can develop or acquire a hearing loss later on, so it is important to check your child's hearing as they grow up.

You can use the two checklists of the sounds that your baby should react to and the types of sounds they should make as they grow older. These checklists are on pages 60 and 61.

If you have any concerns about your child's hearing, discuss them with your health visitor or family doctor. Your child's hearing can be tested at any age.

What happens if the second screening test does not show a clear response from one or both of my baby's ears?

If the second screening test does not show a clear response from one or both of your baby's ears you will be referred to your local audiology department. They will carry out special tests to measure your baby's hearing. Again, this often happens and does not necessarily mean your baby has a hearing loss.

There may be a number of other reasons why the second screen could not record a clear response from one or both of your baby's ears. Further tests by an audiologist will give you better information about your baby's hearing.

You will be given a leaflet explaining what this involves.

What is the likelihood of my baby having a hearing loss if the second screening test does not record clear responses?

Most babies will record clear responses to sound at the second screening test and at the further tests carried out by an audiologist. However, there is a possibility that your baby may have a hearing loss.

Nationally, about one in 25 babies whose second screening test does not record clear responses may have a hearing loss in one or both ears. Finding out that your baby has a hearing loss early means that you and your baby will get advice and support right from the start.

"Daniel is my second little boy and it was amazing. He had his hearing check really soon after I had him."
(Mother of screened baby)

Where can I get further information?
If you would like more information about your baby's hearing screening test, you can contact the hospital that will carry out the test.

You can also find out more from your health visitor, midwife or local audiology department.

The NHS Newborn Hearing Screening Programme has a website that provides further information:
www.hearing.screening.nhs.uk

For information and support about your child's hearing, contact the National Deaf Children's Society Freephone Helpline on 0808 800 8880 (10am to 5pm Mon-Fri), or send an email to helpline@ndcs.org.uk or visit the website: www.ndcs.org.uk

For more information about your baby's hearing screening test contact:

These two lists give pointers about what to look and listen out for as your baby grows to check if he/she can hear. Babies do differ in what they can do at any given age, The ages presented here are approximate only.

REACTION TO SOUNDS CHECKLIST

Shortly after birth - a baby is startled by a sudden loud noise such as a hand clap or a door slamming. Blinks or opens eyes widely to such sounds or stops sucking or starts to cry.

1 month - a baby starts to notice sudden prolonged sounds like the noise of a vacuum cleaner and may turn towards the noise. Pauses and listens to the noises when they begin.

4 months - a baby quietens or smiles to the sound of familiar voice even when unable to see speaker and turns eyes or head towards voice. Shows excitement at sounds (e.g.: voices, footsteps etc).

7 months - a baby turns immediately to a familiar voice across the room or to very quiet noises made on each side (if not too occupied with other things).

9 months - a baby listens attentively to familiar everyday sounds and searches for very quiet sounds made out of sight.

12 months - a baby shows some response to own name. May also respond to expressions like 'no' and 'bye bye' even when any accompanying gesture cannot be seen.

If at any stage in the baby or child's development you think he/she may have difficulties hearing, contact your health visitor or family doctor.

Adapted from: The 'Can Your Baby Hear You' form, B. McCormick, 1982,Chiidren's Hearing Assessment Centre, Nottingham, UK.

MAKING SOUNDS CHECKLIST

4 months - a baby
makes soft sounds when awake. Gurgles and coos.

6 months - a baby
makes laughter-like sounds. Starts to make sing-song vowel sounds (e.g.: a-a,muh, goo, der, aroo, adah).

9 months - a baby
makes sounds to communicate in friendliness or annoyance. Babbles (e.g.: 'dada da', 'ma ma ma', 'ba ba ba'). Shows pleasure in babbling loudly and tunefully. Starts to imitate other sounds like coughing or smacking lips.

12 months - a baby
babbles loudly, often in a conversational-type rhythm. May start to use one or two recognisable words.

15 months - a baby
makes lots of speech-like sounds. Uses 2-6 recognisable words meaningfully (e.g.: 'teddy' when seeing or wanting the teddy bear).

18 months - a baby
makes speech-like sounds with conversational-type rhythm when playing. Uses 6-20 recognisable words. Tries to join in nursery rhymes and songs.

24 months - a child
uses 50 or more recognisable words appropriately. Puts 2 or more words together to make simple sentences (for example: more milk). Joins in nursery rhymes and songs. Talks to self during play (may be incomprehensible to others).

30 months - a child
uses 200 or more recognisable words. Uses pronouns (e.g.: I, me, you). Uses sentences but many will lack adult structure. Talks intelligibly to self during play. Asks questions. Says a few nursery rhymes.

36 months - a child
has a large vocabulary intelligible to everyone.

Adapted from: M. D. Sheridan (Revised by M. Frost and A. Sharma), 1997, Routledge, London, New York.

Blood spot screening for your newborn baby

In the first week after birth, you will be offered a blood spot screening test for your baby.

Why should I have my baby screened?

Newborn blood spot screening identifies babies who may have rare but serious conditions.

Most babies screened will not have any of the conditions but, for the small number that do, the benefits of screening are enormous. Early treatment can improve their health and prevent severe disability or even death.

What are newborn babies screened for?

All babies are offered screening for phenylketonuria, congenital hypothyroidism, cystic fibrosis, sickle cell diseases and medium-chain acyl-CoA dehydrogenase deficiency (MCADD).

If a baby is thought to have one of the conditions, he or she will need further tests to confirm this. These conditions are explained below.

Phenylketonuria

About one in 10,000 babies born in the UK has phenylketonuria (PKU). Babies with this inherited condition cannot process a substance in their food called phenylalanine. If untreated, they will develop a serious permanent mental disability.

Screening means that babies with the condition can be treated early through a special diet which will prevent severe disability and allow them to lead a normal life.

If babies are not screened, but are later found to have PKU, it may be too late for the special diet to make a real difference.

Congenital hypothyroidism

About one in 4,000 babies born in the UK has congenital hypothyroidism (CHT). Babies with CHT do not have enough of the hormone thyroxine. Without this hormone, they do not grow properly and can develop a serious, permanent, physical and mental disability.

Screening means that babies with CHT can be treated early with thyroxine tablets, which will prevent serious disability and allow them to develop normally.

If babies are not screened and are later found to have CHT, it may be too late to prevent them becoming seriously disabled.

Sickle cell diseases

About one in 2,000 babies born in the UK has a sickle cell disease (SCD). These are inherited diseases that affect the red blood cells. If a baby has a sickle cell disease, their red blood cells can change to a sickle shape and become stuck in the small blood vessels. This can cause pain and damage to the baby's body, serious infection, or even death.

Screening means that babies with SCD can receive early treatment, including immunisations and antibiotics, which, along with parent education, will help prevent serious illness and allow the child to live a healthier life.

What happens to your baby's data after screening? You will be told your baby's result and the result will be sent to your healthcare professional. Identifiable data may be stored by the screening programme and used to monitor and improve screening for sickle cell and thalassaemia. The use of data obtained from sickle cell and thalassaemia screening is governed by the National Information Governance Board[1].

If you do not wish your baby's screening data to be used in this way please contact the sickle cell and thalassaemia screening programme:

email at Radoslav.latinovic@nhs.net or by telephone on 020 7848 6634 where you will be able to speak to someone in a language of your choice or at sct.screening.nhs.uk/contactus

More information on this work is available at sct.screening.nhs.uk/newborn

Cystic fibrosis

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

Screening means that babies with CF can be treated early with a high-energy diet, medicines and physiotherapy. Although a child with CF may still become very ill, early treatment is thought to help them live longer, healthier lives.

If babies are not screened for CF and they do have the condition, they can be tested later but parents may have an anxious time before CF is recognised.

[1] Section 251 of the NHS Act 2006. Section 251 approval has been given for our audit team to access sickle cell and thalassaemia screening data under strict conditions. You can find more information on this at www.nigb.nhs.uk/ecc.

MCADD (medium-chain acyl-CoA dehydrogenase deficiency)

About one in 10,000 babies born in the UK has MCADD. Babies with this inherited condition have problems breaking down fats to make energy for the body. This can lead to serious illness, or even death.

Screening means that most babies who have MCADD can be recognised early. This allows special attention to be given to their diet, including making sure they eat regularly. This care can prevent serious illness and allow babies with MCADD to develop normally.

Screening babies for MCADD is important so the condition can be identified before the babies become suddenly and seriously ill.

Will screening for these conditions show up anything else?

Screening for cystic fibrosis (CF) includes testing some babies for the most common gene changes that cause CF. This means screening may identify some babies who are likely to be genetic carriers of cystic fibrosis. These babies may need further testing to find out if they are a healthy carrier, or have CF.

Screening identifies babies who are genetic carriers of sickle cell or other unusual red blood cell disorders. Carriers of sickle cell disorders are healthy and will not be affected by the condition.

Occasionally, other medical conditions might be identified through these screening tests.

How will the midwife take the blood spots?

About a week after your baby's birth the midwife will prick your baby's heel using a special device to collect some drops of blood onto a card. The heel prick may be uncomfortable and your baby may cry.

You can help by

- making sure your baby is warm and comfortable
- cuddling and being ready to feed your baby

Are repeat blood samples ever needed?

Occasionally the midwife or health visitor will contact you and ask to take a second blood sample from your baby's heel. This may be because there was not enough blood collected, your baby's NHS number was not recorded on the blood spot card, the result was unclear, or your baby was born early or had a blood transfusion before the test. Your midwife or health visitor will explain the reason to you. It is important that a repeat test (if needed) is done so that all the tests have been completed. Usually the repeat results are normal.

Screening is recommended

Screening your baby for all these conditions is strongly recommended, but you do not have to have it carried out. If you do not want your baby screened for any or all of these conditions, discuss it with your midwife. All your decisions will be recorded in your notes.

If you have any concerns about your baby's screening, please ask your midwife or GP.

How will I hear about the results?

- Most babies will have normal results, indicating that they probably do not have any of the conditions. You should receive the screening test results from a health professional by the time your baby is six to eight weeks old. The result of the screening should be recorded in your baby's personal child health record (often referred to as their 'red book'). Please keep this safe.
- If a baby is thought to have phenylketonuria (PKU), congenital hypothyroidism (CHT) or MCADD parents will usually be contacted before the baby is 3 weeks old and given an appointment to see a specialist.
- If a baby is thought to have cystic fibrosis (CF), parents will usually be contacted before the baby is 4 weeks old.
- If a baby is thought to have sickle cell disease (SCD), parents will usually be contacted before the baby is 6 weeks old.
- Some babies are found to be carriers. Their parents will usually be told by the time the child is 6-8 weeks old.

If you move home while you are waiting for the results of your baby's screening test, please tell your midwife or health visitor your new address.

The purpose of screening is to identify babies more likely to have these conditions. Screening is not 100% accurate.

What happens to your baby's blood spots after screening?

After screening, newborn babies' blood spots are stored for at least five years and may be used for the following:

- 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 at:
www.newbornbloodspot.screening.nhs.uk

In the future 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.

Where can I go for more information?

Don't hesitate to talk to a health professional at any time, before or after the examinations or tests, if you are worried about your baby.

Your midwife, health visitor, GP or screener will be able to provide you with more information. You may find the following websites useful:

- For information about all aspects of antenatal and newborn screening and the sources of information available to parents **www.screening.nhs.uk/an**
- For more information about Hearing screening see the information panel on page 58
- For general health advice and information, see the following.
    - Phone NHS Direct on 0845 4647 or visit **www.nhsdirect.nhs.uk**
    - For a directory of personal experiences visit **www.dipex.org**
    - For Contact a Family charity visit **www.cafamily.org.uk**
    - For Antenatal Results and Choices visit **www.arc-uk.org**
    - For information from the UK Newborn Screening Programme Centre, visit **www.newbornbloodspot.screening.nhs.uk**

Notes

Notes

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