You're **pregnant!**

Scans and tests

what you need to know

Pregnancy screening **tests**

NHS SCOTLAND

Public Health Scotland

This booklet explains the blood tests, screening tests and diagnostic tests you can have in pregnancy.

Screening in pregnancy involves simple tests that help to find out the chances of you or your baby having a health or chromosomal condition.

The screening tests we look at in this booklet use:

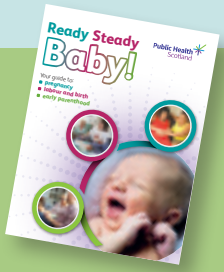
- blood tests
- ultrasound scans.

Screening tests are offered to look for some health conditions and chromosomal conditions. Most screening tests will show that your baby is healthy. If the screening tests suggest there might be a health issue for you or your baby, you'll be offered a diagnostic test to get a definite answer. You do not have to have these tests, it's your choice.

The **diagnostic tests** we look at are:

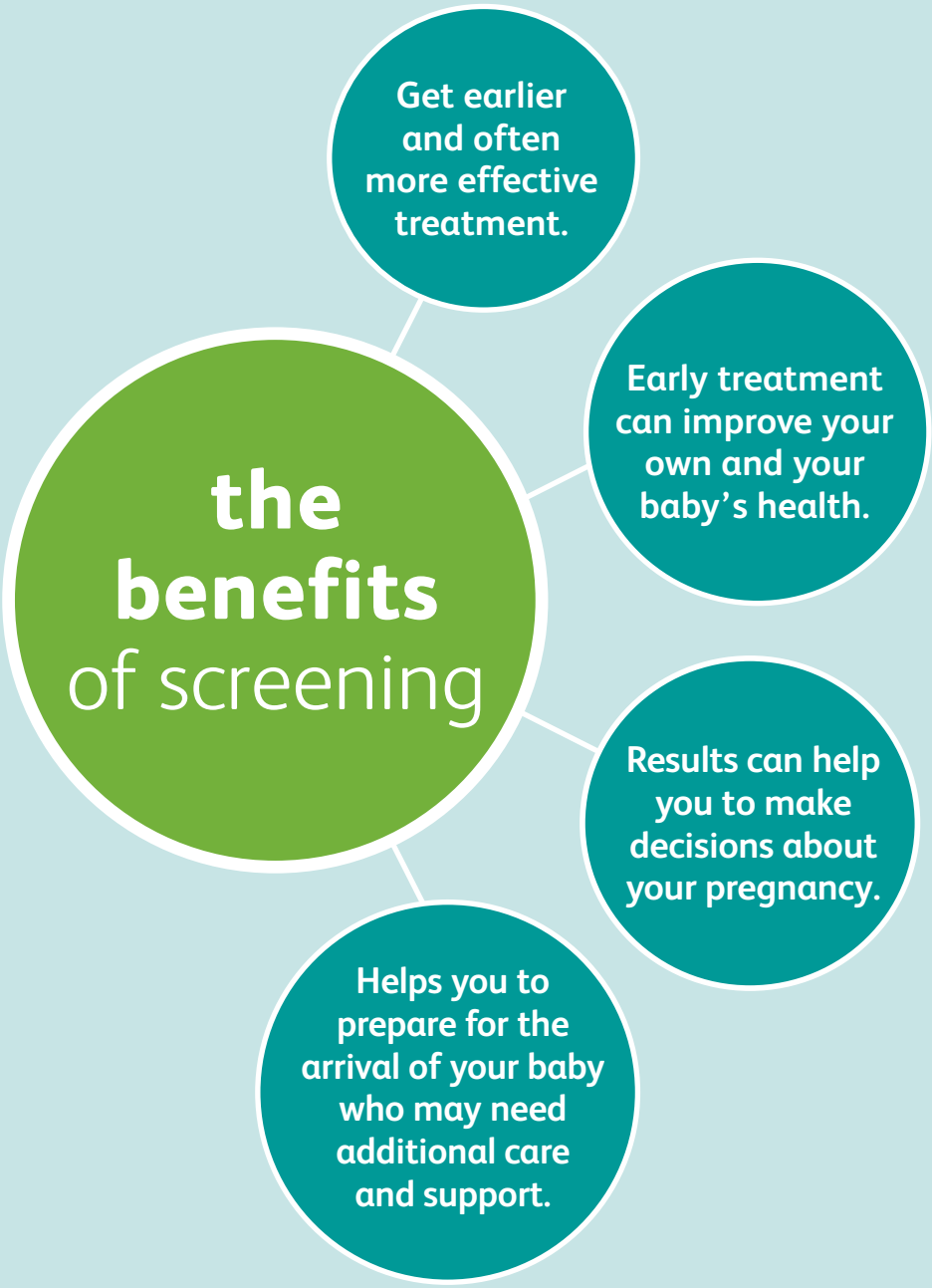
- chorionic villus sampling (CVS)
- amniocentesis.

If you do not speak or understand English, the NHS will provide someone who can translate what is being said into your own language. Let your health professional know if you feel you need an interpreter.



More information on your pregnancy can be found in the Ready Steady Baby book or online at **www.nhsinform.scot/ready-steady-baby**

During pregnancy, you'll be offered free immunisations to protect you and your baby against whooping cough and influenza (flu). For information about immunisations offered during pregnancy, go to **www.nhsinform.scot/vaccinesinpregnancy**



Your midwife will guide you through all the tests and scans offered during your pregnancy.

At a glance

Before 10 weeks

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Between 11 and 14 weeks

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* It's best if these tests are carried out in the early stages of pregnancy, but they can still be done at any point, up to and including labour.

Screening involving blood test Screening involving ultrasound scan

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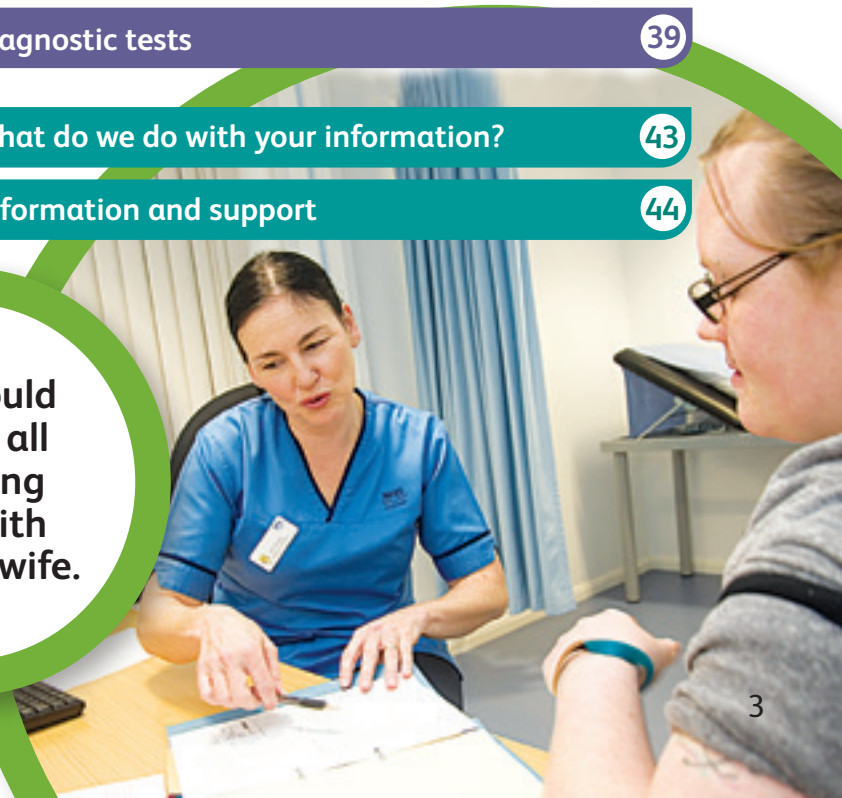
You should discuss all screening tests with your midwife.



Your pregnancy,

It's your choice whether to accept screening and diagnostic tests. You should have a more detailed conversation with your health professional, who will tell you more about screening and what the results can mean.

You can decide at any point that you do not want to be tested, or you only want some of the tests. No one will ever test you without being sure you know what the test is for, how it's done, and that you're prepared to have it.

We talk about 'your health professional' throughout this booklet. This can be a midwife, your GP, a specialist doctor (obstetrician), a sonographer or any other professional involved in your care during your pregnancy.

your choice

People make different decisions about screening and diagnostic testing. They may choose:

- **not to be screened**, because they do not want to know during pregnancy if they have, or their baby has, a health condition or a chromosomal condition
- **to be screened and consider diagnostic testing,** because they want to know during pregnancy if they have, or their baby has, a health condition or a chromosomal condition.

Decisions about whether to accept screening or diagnostic testing can be difficult. You may want to talk with your partner, family or friends. Your health professional and the organisations listed at the end of this booklet can help. But the final decision is yours.

Whatever decision you make, you can agree a pregnancy plan with your midwife that feels right for you.

Facts

All screening and diagnostic tests in this booklet are provided free in Scotland by the NHS. Healthcare and treatment for children in Scotland born with health conditions, chromosomal conditions or disabilities are provided free by the NHS.

Screening

using blood tests

At a glance ...

- Blood tests are an important part of your care during pregnancy.
- They can help to protect your own and your baby's health.
- The blood needed for these tests can usually be taken at one time.
- Your midwife will take blood from your arm at one of your first visits and you should get the results at your next clinic visit.
- Some of the tests may be repeated routinely later in your pregnancy. This will be discussed with you.

What will I be tested for?

Full blood count

This test looks at the red cells, white cells and platelets in your blood. This test will tell if you have anaemia, a condition when you do not have enough healthy red blood cells to carry oxygen around your body. This can be treated with iron tablets and other treatments to support your health and the health of your baby.

Blood group and Rhesus status

The test will tell you what blood group you have. People belong to one of four blood groups, called A, B, O and AB. It's important to know your blood group:

- in case you need a blood transfusion
- because substances in the blood called blood-group antibodies can sometimes affect your baby. If these antibodies are found, your health professional will discuss it with you.

The test will also show if you're Rhesus positive or Rhesus negative. If you're Rhesus positive, you do not need treatment. About one in six women are Rhesus negative. This means they do not have a substance called the Rhesus antigen on their blood cells.

If you're Rhesus negative, there can be issues if your baby is Rhesus positive and their blood enters your blood stream. This is unlikely to be an issue in a first pregnancy, but can be serious in future pregnancies. Your health professional will offer you an injection in your arm – the 'anti-D' injection – that will help to protect your health and that of any future babies you might have.

You can ask your midwife how to find out your blood group, and how and when you'll receive your results. If any health issues are found, your health professional will contact you as soon as possible and give you advice and care.

for sickle cell and thalassaemia

At a glance ...

- Sickle cell and thalassaemia are serious blood disorders that affect haemoglobin.
- Haemoglobin is in red blood cells and carries oxygen around the body.
- People who have these conditions need specialist care throughout their lives.
- Your baby's father might also be offered a test. This will make the results more accurate.

People with sickle cell:

- can have attacks of very severe pain
- may have serious life-threatening infections
- are usually anaemic (which means their blood is not good at carrying oxygen)
- may need medicines and injections throughout their lives to stop them from getting infections.

People with thalassaemia:

- may be very anaemic
- may need blood transfusions every four to six weeks
- may need injections and medicines throughout their lives.

You'll be offered screening tests for sickle cell and thalassaemia at, or shortly after, your first midwife visit. You should decide early whether you want to have the tests, as they should be done as soon as possible in the pregnancy. This is ideally by 10 weeks – although it can still be helpful to have them later.

What causes the conditions?

Sickle cell and thalassaemia are passed from parents to children through **altered haemoglobin genes.**

Genes

Genes determine your characteristics, from the colour of your hair to your blood group. For all our characteristics, we get one gene from each parent.

People only have these disorders if they get **two** altered haemoglobin genes – one from their mother and one from their father. People who get just one altered gene and do not have either condition are known as **carriers**.

Carriers

Carriers do not have either sickle cell or thalassaemia. But if a carrier has a baby with someone who is also a carrier, or who has sickle cell or thalassaemia, there is a higher chance that the baby could have one of the conditions, or be a carrier.

Anyone can be a carrier. But you're more likely to carry the altered genes if your ancestors (parents, grandparents and those further back in your family line) came from places where malaria was common, such as:

- an African country
- South Asia (India, Pakistan or neighbouring countries)
- the Caribbean
- the Middle East
- South America
- Southern Europe
- East and South-East Asia (China, Hong Kong, Malaysia or their neighbours).

People from Poland may also be affected because some Polish people moved many generations ago from areas where malaria was common.

Facts

Although sickle cell carriers are healthy, they can have issues when their bodies do not get enough oxygen (when having an anaesthetic, for example). Knowing you're a carrier can help you manage these situations.

People who are thalassaemia carriers do not experience these issues.

The Family Origin Questionnaire

To help find out if you or your baby's father have a chance of carrying genes for these conditions, your health professional will ask you questions from the **Family Origin Questionnaire.**

The aim is to find out where your immediate family and your ancestors came from, and if you have a higher chance of carrying the genes.

What will I be tested for?

You will be screened for **sickle cell** and **thalassaemia** by:

- a blood test
- answering questions from the Family Origin Questionnaire.

Your baby's father may be invited for a blood test too, as testing both parents gives more accurate results. If both parents are found to be carriers or are affected by one of the conditions, **or** if it's not possible to test both parents, you may be offered a diagnostic test to confirm whether or not your baby has or is carrying one of the conditions.

Tell your midwife if …

you and your partner are related by blood. If you each have inherited genes from a shared relative, it may be more likely that you're both carriers.

What will the results tell me?

The most likely result is that you and your baby's father are not carriers for sickle cell or thalassaemia. If one of you is, your health professional will talk to you about what this could mean for you, your baby and your family.

Very rarely, the test may show that one or both of you have a blood disorder without knowing it. Your health professional will discuss this with you and give you more information.

The test is very reliable, but if the result is unclear you'll be offered another test to be sure.

Results from your own and your baby's father's tests will then be used to see if your baby has a higher chance of developing one of the conditions.

Facts

If it's not possible to test both parents, you may be offered a diagnostic test to confirm whether or not your baby has or is carrying one of the conditions.

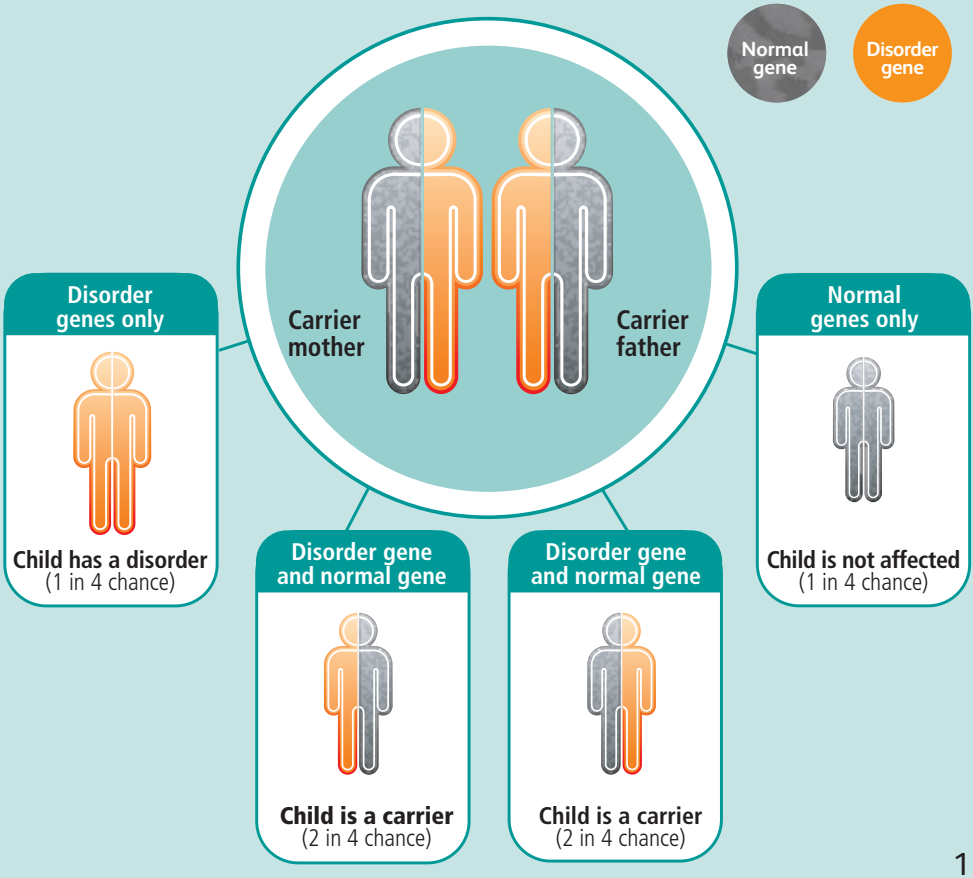
If you and your baby's father both carry the gene for sickle cell, thalassaemia or another blood condition, there is usually:

a 25 % (1 in 4) chance your baby will **not have a disorder**

a 50 % (2 in 4) chance your baby will **be a carrier**

a 25 % (1 in 4) chance your baby will **have a disorder**

The chances stay the same with each pregnancy.



Please give staff as much information as you can if ...

you have an assisted pregnancy (for example IVF – in vitro fertilisation). This could affect your screening result. Your health professional will explain your results.

Can my unborn baby be tested?

As with all screening tests, screening for sickle cell and thalassaemia is not 100% accurate. Diagnostic testing is the only way to know for sure if your baby has one of the conditions. See pages 39–42.

Your health professional will help you to understand what it will mean if the results show your baby has a blood disorder. They'll talk with you about the care that is available and whether you wish to continue with your pregnancy.

A genetic counsellor is a professional who is specially trained to give you information and support if you have questions or concerns about a genetic condition. Your midwife or GP will be able to refer you.

Testing new babies for sickle cell

All newborn babies are offered a 'blood spot' screening test, ideally when they are 5 days old. Your midwife will prick your baby's heel and put a few spots of blood onto a card. The card is sent to a laboratory where tests are done for nine conditions, including sickle cell. You'll get the results on or before your baby's check-up at 6–8 weeks.

For more information about the blood spot test and other newborn screening tests

Look out for the booklet 'Your baby! Tests offered' which you'll get later in your pregnancy.

Or you can visit the NHS inform website at **www.nhsinform.scot/newbornscreening**

for infectious diseases

At a glance ...

- Simple treatments can reduce the chances of you and your baby being affected by infectious diseases. You'll be offered blood tests for hepatitis B, syphilis and human immunodeficiency virus (HIV).
- You'll usually get the results at your next clinic visit, but you'll be contacted sooner if an infection is found.
- You can find more information about how infectious diseases are caused, diagnosed, treated and prevented at www.nhsinform.scot/psid
- Ask your midwife about your free whooping cough, flu and COVID-19 vaccines.

What will I be tested for?

Hepatitis B

Hepatitis B is caused by a virus which can be passed from mother to baby during birth. The virus can cause serious liver disease, but women carrying hepatitis B may have no signs of infection. Without a test, they would not know they're infected.

Without immunisation, many babies born to mothers who are infected with hepatitis B will become infected themselves.

If the test shows you're infected with hepatitis B, you'll be offered specialist treatment.

Your baby will be immunised against hepatitis B at birth. This will usually stop them getting hepatitis B and protect them from serious liver disease.

Syphilis

Syphilis can damage your own and your baby's health if it's not found and treated. It can be treated quickly and simply with antibiotics. People can have syphilis without realising it.

HIV

HIV is the virus that causes AIDS. Over time, HIV damages the body's defences against infection and disease. People with HIV cannot be cured, but with treatment they can usually expect to live a full and healthy life.

A woman who has HIV can pass the infection to her baby during pregnancy, birth and through breastfeeding. Like hepatitis B and syphilis, women with HIV may not know they're infected until they have a test – it can take years for HIV to make someone ill.

If your pregnancy screening test shows you may have HIV, you'll be offered another test to check for sure. You'll be offered antiviral medicines to keep you healthy and greatly reduce the chance of you passing HIV to your baby. Your health professional will give you advice about the safest way to deliver and feed your baby. They'll also tell you about medicines your baby can have after they're born to help protect against HIV.

Support and advice

Organisations listed at the end of the booklet can provide information and support on infectious diseases.

Facts

Having a blood test for HIV does not affect your current or future life insurance policies, but if a health condition is found, it could affect your insurance. You might wish to check any policies you have for further details.

It's still recommended that you take the test.

by ultrasound scan

At a glance ...

- Your midwife will offer you two ultrasound screening scans – one between 11 and 14 weeks and a second between 18 and 21 weeks.
- These scans are carried out by trained health professionals called sonographers. Some midwives are also sonographers.
- Scans are not 100% accurate. Sometimes there are conditions that cannot be picked up by the scan.
- Your scan will be a two-dimensional black and white image. Three-dimensional (3D) and colour scans are not routinely used in the NHS.
- Ultrasound scans are safe for mother and baby.

Screening scans look for issues and check if your baby is growing well. Read this section carefully and speak to your health professional before deciding if you want to have the scans.

Early pregnancy screening scan

Your first screening scan is offered between 11 and 14 weeks. The scan:

- checks your baby's heartbeat, growth and development
- estimates the stage of pregnancy
- confirms whether you're having one baby or more
- gives the nuchal-translucency measurement (see page 33).

If a specific issue is found at this stage, your health professional will discuss it with you at the time.

Mid-pregnancy screening scan

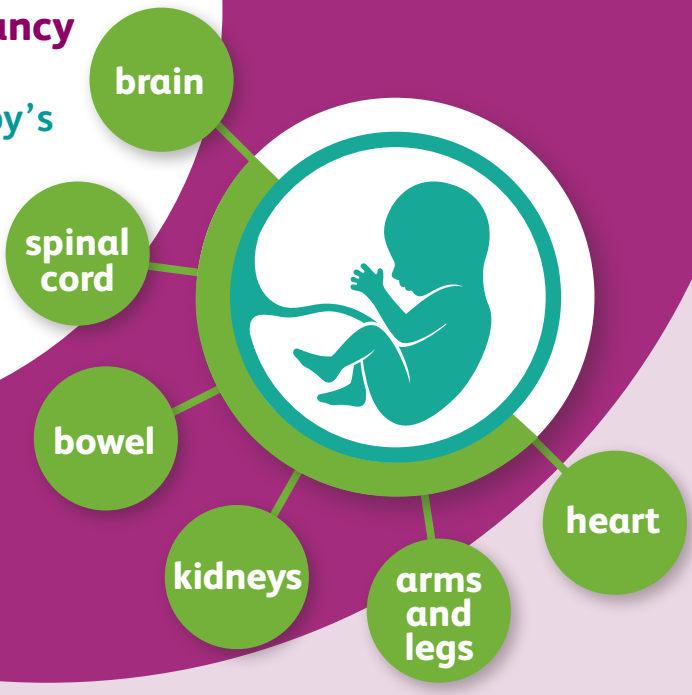
This scan is offered between 18 and 21 weeks. It's also known as a fetal anomaly scan.

The scan will look for the following conditions:

- anencephaly
- open spina bifida
- cleft lip
- diaphragmatic hernia
- gastroschisis
- exomphalos
- bilateral renal agenesis
- serious cardiac abnormalities
- lethal skeletal dysplasia
- Edwards' syndrome, or T18*
- Patau's syndrome, or T13.*

*This scan cannot tell the type of Edwards' syndrome or Patau's syndrome, for example, full or partial.

The mid-pregnancy screening scan looks at your baby's development, including their:



Some conditions may not be picked up by the mid-pregnancy scan. If the sonographer thinks there could be an issue, they may ask for a second opinion from another health professional. If this happens, they'll tell you what they're checking for but they may not be sure yet. If the sonographer finds something that means there's a higher chance of your baby having a chromosomal condition, you may be offered a diagnostic test.

If no issues are found on the mid-pregnancy screening scan, you probably will not need another scan during your pregnancy. But you may be asked to come back on another day for a repeat scan if the sonographer has not been able to see your baby clearly.

Before, during and after your scan

- You'll be asked to drink some water (about a pint/500 ml) an hour before the early pregnancy screening scan. Having water in your bladder will help the sonographer to see your baby more clearly.

- You do not need a full bladder before the mid-pregnancy screening scan, but drinking a glass or two of water will help the sonographer.
- Most hospitals are happy for you to have one person with you during the scan. Young children may not be allowed in with you because they can distract the sonographer. It's a good idea to check beforehand.
- The sonographer will ask you to lie on a couch, raise your top to your chest and lower your skirt or trousers to your hips. They'll squeeze some gel onto your abdomen (tummy) then gently pass a hand-held device across it. The device sends and picks up ultrasound waves that allow a computer to build an image of your baby.
- The scan does not hurt at all, but the gel might be a little cold at first. Sometimes the sonographer needs to press your tummy if some parts of your baby are difficult to see.

Finding out about a condition before birth can help parents to plan and prepare. For example, if your baby may need treatment soon after birth, health professionals can help you plan to give birth in a hospital where you and your baby can have the care you'll need.

Facts

Screening tests cannot find all conditions.

- **The sonographer might not be able to get a clear view of your baby.**
- **Some conditions develop after 21 weeks.**
- **Some conditions cannot be seen on a scan because they do not affect your baby's appearance.**

A sonographer carrying out a scan

A sonographer's screen with an image of a baby

- The scan will take up to 30 minutes.
- You should be able to recognise parts of your baby's body on the screen as the sonographer does the scan – they'll point them out to you.
- Finding out the sex of your baby is not the purpose of the scan unless there's a medical reason for doing so. Often it's impossible to tell because of the position of your baby. It's not completely reliable and can sometimes be wrong.
- The vast majority of scans show that babies are healthy and no issues are found.

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

You'll be offered screening to find out how likely it is that your baby has Down's syndrome, Edwards' syndrome or Patau's syndrome. Whether you want to know how likely it is will be personal to you. It's your choice.

During the first trimester you can choose to be screened for:

- Down's syndrome only
- Edwards' syndrome and Patau's syndrome only
- All three conditions.

You can choose not to have any of these screening tests if you do not want them.

In the UK, around one baby in every 700 births has Down's syndrome. It's the most common chromosomal condition.

In the UK, around one baby in every 5,000 births has Edwards' syndrome. It's the second most common chromosomal condition.

In the UK, around one baby in every 16,000 births has Patau's syndrome.

Chromosomes

Our bodies are made from millions of cells, and inside every cell are chromosomes. Chromosomes carry genes that determine how we develop. People usually have 46 chromosomes, 23 from the mother and 23 from the father. If babies have an extra full or partial chromosome in their cells, they'll have a chromosomal condition. If this is only in some of their cells, it may be called mosaicism.

If you get a higher-chance screening result for one of the chromosomal conditions, you can choose to get a further screening test (known as NIPT, which stands for non-invasive prenatal testing), a diagnostic test, or have no further tests. Down's syndrome, Edwards' syndrome and Patau's syndrome happen by chance. They're not caused by anything parents do before or during pregnancy.

Older mothers are more likely to have a baby with one of the conditions and the chance increases with the mother's age at pregnancy. But Down's syndrome, Edwards' syndrome and Patau's syndrome can occur in pregnancy for women of any age. All pregnant women, no matter what age, can have the test.

Down's syndrome

- A baby with Down's syndrome is born with an extra chromosome 21 in every cell. Down's syndrome is sometimes known as trisomy 21.
- People with Down's syndrome can lead active, healthy and fairly independent lives into their 60s, 70s and beyond.
- Most people with Down's syndrome will have mild to moderate learning disabilities and some may have more complex needs. This cannot be known before birth.
- Some health issues are more common in people with Down's syndrome, but some health issues are less common.

Life with Down's syndrome

There are increasing opportunities and support in education, work and housing for people with Down's syndrome. People with the condition can live healthy and fulfilling lives as part of their families and communities. Some people with Down's syndrome may live independently, have jobs, be in relationships and socialise with minimal support. Evidence suggests that most people with Down's syndrome are happy and most families feel satisfied with their lives.

Children with Down's syndrome can go to a mainstream school and get additional support if they need it. It cannot be known before birth if a person will need any additional help and support.

Down's syndrome and health

Many children with Down's syndrome have similar health issues to all children. Some health issues can be more common in people with Down's syndrome but some health issues can be less common.

Around 5 in 10 babies born with Down's syndrome will have heart conditions and fewer than 1 in 5 of those may need surgery. People with Down's syndrome are more likely to have muscle tone and digestion issues, and may have reduced hearing or vision. As adults, if you have Down's syndrome you may be more likely to develop early-onset dementia but are less likely to develop some cancers and types of heart disease. Regular health checks can make sure any health concerns are picked up early and managed.

Down's syndrome and life expectancy

People with Down's syndrome can live into their 60s, 70s and beyond.

More information

You can get more information and support at Down's Syndrome Scotland: **www.dsscotland.org.uk**

Edwards' syndrome

- Babies with Edwards' syndrome have more of chromosome 18 in all or some of their cells. Edwards' syndrome is sometimes known as trisomy 18.
- Miscarriage and stillbirth are more likely if your baby has Edwards' syndrome.
- Edwards' syndrome affects how long your baby is likely to live.
- Babies with a partial or mosaic form can have less serious health issues than babies with a full form of the condition. Screening tests cannot predict how serious health issues will be before they're born.

Life with Edwards' syndrome

How Edwards' syndrome could affect your baby depends on a number of things. Children with full Edwards' syndrome will have significant delays in learning and physical development and will likely need lifelong support with health, care and learning. Children with a partial or mosaic form may be less affected.

Edwards' syndrome and health

Some physical signs of the condition may be seen during the mid-pregnancy screening scan (18–21 weeks of pregnancy). Babies with the full form of Edwards' syndrome are likely to have a wide range of health conditions, some of which are serious.

Around 9 in 10 babies born with Edwards' syndrome will have heart conditions, 5 in 10 may have hearing loss and 5 in 10 may have issues with their muscles and joints. Some babies need help with feeding, swallowing and breathing. These babies usually have a low birth weight and are also more likely to get infections and to need hospital care.

Edwards' syndrome and life expectancy

Around 7 in 10 pregnancies diagnosed with Edwards' syndrome at 12 weeks will end in miscarriage or stillbirth. This is more likely in early pregnancy, and the chance gets less as pregnancy progresses.

Edwards' syndrome will affect how long your baby is likely to live. Of all babies born with Edwards' syndrome, around 5 in 10 will live longer than 1 week and around 1 in 10 will live longer than 5 years. Some babies with full Edwards' syndrome may live into adulthood. This is more likely for babies with partial or mosaic forms.

More information

You can get more information and support at Support Organisation for Trisomy 13 and Trisomy 18 (SOFT): **www.soft.org.uk**

Patau's syndrome

- Babies with Patau's syndrome have more of chromosome 13 in all or some of their cells. Patau's syndrome is sometimes known as trisomy 13.
- Miscarriage and stillbirth are more likely if your baby has Patau's syndrome.
- Patau's syndrome affects how long your baby is likely to live.
- Babies with a partial or mosaic form can have less serious health issues than babies with a full form of the condition. Screening tests cannot predict how serious health issues will be before they're born.

Life with Patau's syndrome

How Patau's syndrome could affect your baby depends on a number of things. Children with full Patau's syndrome will have significant delays in learning and physical development and will likely need lifelong support with health, care and learning. Children with a partial or mosaic form may be less affected.

Patau's syndrome and health

Some physical signs of the condition may be seen during the mid-pregnancy screening scan (18–21 weeks of pregnancy). Babies with the full form of Patau's syndrome are likely to have a wide range of health conditions, some of which are serious.

Around 8 in 10 babies born with Patau's syndrome may have heart conditions, 6 in 10 may have issues with their brain development and around 6 in 10 may have a cleft lip and palate. Some babies will have eye issues, kidney issues, seizures or be born with organs outside their body. Some babies need help with feeding, swallowing and breathing. These babies usually have a low birth weight and are also more likely to get infections and to need hospital care.

Patau's syndrome and life expectancy

Around 7 in 10 pregnancies diagnosed with Patau's syndrome at 12 weeks will end in miscarriage or stillbirth. This is more likely in early pregnancy, and the chance gets less as pregnancy progresses.

Patau's syndrome will affect how long your baby is likely to live. Of all babies born with Patau's syndrome, around 4 in 10 will live longer than 1 week and 1 in 10 will live longer than 5 years. Some babies with full Patau's syndrome may live into adulthood. This is more likely for babies with partial or mosaic forms.

More information

You can get more information and support at **www.soft.org.uk**

What type of screening test will I be offered for Down's syndrome and/or Edwards' syndrome and Patau's syndrome?

You'll be offered either:

First trimester

a blood test combined with an NT ultrasound scan

(if you're between 11 weeks and 14 weeks pregnant)

Second trimester

a blood test on its own for Down's syndrome only

(if you're between 14 weeks and 20 weeks pregnant)

Some tests can only be done at certain times during pregnancy. If your early pregnancy scan shows you're at a different stage of pregnancy than you thought, your midwife will explain which tests you can have.

If you're not able to have first trimester screening for Down's syndrome and/or Edwards' syndrome and Patau's syndrome you can only have screening for Down's syndrome during the second trimester.

Blood tests

The blood test measures substances that have passed between you and your baby. If you decide to have the test, a sample of your blood will be taken between 11 and 20 weeks.

Speak to your health professional if ...

- you smoke
- you have an assisted pregnancy (for example IVF – in vitro fertilisation). Your age and (if relevant) the age of the egg donor are used in the calculation. Having this information can give a more accurate screening result.

NT ultrasound scan

The NT (standing for 'nuchal translucency') ultrasound scan is carried out between 11 and 14 weeks of pregnancy, usually as part of your early pregnancy screening scan. You might hear it called the 'combined' test, because it combines the results from the scan with your blood test.

The ultrasound measures the amount of fluid lying under the skin at the back of your baby's neck. The results of your NT ultrasound scan and blood test, along with your age, weight, stage of pregnancy and some other information (like whether you smoke or not), are put into a computer to work out the chance of your baby having Down's syndrome, or a combined chance of Edwards' syndrome and Patau's syndrome. You'll be offered the same screening choices if you're pregnant with twins as you would be if you were pregnant with one baby. Screening tests may be less accurate with twin pregnancies. Your midwife will help you to understand what this means and support you to decide if choosing further tests feels right for you and your babies.

What happens if I choose to be screened for Down's syndrome and/or Edwards' syndrome and Patau's syndrome?

You may have chosen to be screened for Down's syndrome only, Edwards' syndrome and Patau's syndrome only, or all three conditions. You'll only receive the results you've asked for.

If I get a lower-chance result

Most women will get a lower-chance result. This means it's unlikely your baby has one of the conditions. It usually means your baby has a chance lower than 1 in 150 of having one of the conditions.

If the screening test shows the chance of your baby having Down's syndrome, Edwards' syndrome or Patau's syndrome is low, you'll not be offered further tests.

Facts

More than 95% of screening test results show the chance of the baby having Down's syndrome, or Edwards' syndrome or Patau's syndrome is low. It does not mean there's no chance at all that the baby has one of these conditions, just that it's unlikely.

If I get a higher-chance result

If you get a higher-chance result, it does not mean your baby definitely has one of the conditions but it's more likely. It usually means your baby has a chance higher than 1 in 150 of having one of the conditions.

You may receive a higher chance result for one of the conditions but the baby could have a different condition. There is a very small chance you could receive a higher chance result for Down's syndrome, but there may be a chance that the baby actually has Patau's syndrome.

If you receive a very high chance result (between '1 in 2' and '1 in 10') for Edwards' syndrome or Patau's syndrome, NIPT is less accurate and may affect your next screening choice.

I have a higher-chance result, what happens next?

Your midwife will discuss your results with you and explain what they mean. They'll then tell you what your further choices are. You'll be given time to think through your choices and reach decisions which feel best for you and your baby. You do not need to make any decisions straight away.

After a higher-chance result, you'll be offered a choice of having:

- no further tests
- a further screening test (non-invasive prenatal testing, known as NIPT)
- a diagnostic test (chorionic villus sampling (CVS) or amniocentesis).

More information

Pages 44 and 45 have a list of organisations where you can find more information and support following a positive screening result.

Non-invasive prenatal testing (NIPT)

- NIPT is a blood test which is more accurate for women who received a higher-chance result from the earlier screening for Down's syndrome and/or Edwards' syndrome and Patau's syndrome.
- No screening test is 100% accurate, so NIPT cannot tell for definite if your baby has one of the conditions.
- Like other pregnancy screening tests, it's completely safe and will not harm you or your baby.
- NIPT will give results about all three conditions. You cannot choose to be screened for Down's syndrome only, or Edwards' syndrome and Patau's syndrome only, like in earlier screening.

How NIPT works

During pregnancy, the placenta releases some of its DNA into your bloodstream, so your blood has both your DNA and some from the placenta. This is what NIPT measures. If NIPT finds more DNA than expected for chromosomes 21, 18 or 13 in your blood it could mean that your baby has one of the conditions.

NIPT will not be used to find other health or chromosomal conditions, or the sex of your baby, as part of NHSScotland's pregnancy screening.

If you do not want to go straight to having a diagnostic test, your NIPT result may help you decide whether to have one or not. It can also help you prepare for the arrival of a baby who may need additional care and support.

If you receive a very high chance result (between '1 in 2' and '1 in 10') from your first screening test, you should speak with your healthcare professional about your options. This is because NIPT is less accurate when you receive a very high chance result for Edwards' syndrome or Patau's syndrome.

Low-chance result

Most women who have NIPT will get a low-chance result. This means it's unlikely your baby has one of the conditions. If you get this result, you'll not be offered any further tests for these conditions.

There's a small chance that you may get a low-chance result and your baby does have one of the conditions. This is known as a false negative. Your midwife will be able to give you more information about this.

High-chance result

If you get a high-chance NIPT result, it does not mean your baby definitely has one of the conditions but it's very likely. You'll be offered diagnostic testing which can tell you for definite if your baby has one of the conditions. Whatever you choose, health professionals will give you information and support.

There's a small chance that you may receive a high-chance result and your baby does not have one of the conditions. This is known as a false positive.

No result

NIPT can sometimes give no result if there's not enough DNA in the blood sample or if there's been a technical issue with the testing. If you do not get a result you can choose to have a repeat NIPT, go straight to diagnostic testing or to have no further tests.

How accurate is NIPT?

If you get a high-chance result that your baby has one of the conditions, this will be confirmed in your baby:

- 91 times out of 100 for Down's syndrome
- 84 times out of 100 for Edwards' syndrome
- 87 times out of 100 for Patau's syndrome.

Research shows that NIPT is better at finding babies who have Down's syndrome than finding babies with Edwards' syndrome or Patau's syndrome. This may be because babies with Edwards' syndrome or Patau's syndrome are likely to be smaller and have smaller placentas. This may mean less DNA from the placenta can be found in your bloodstream.

NIPT is more accurate for women who've already had a higher-chance result from their first screening test. However, no screening test is 100% accurate.

NIPT can be as accurate in identical twin pregnancies as if you were pregnant with one baby. NIPT may be less accurate in non-identical twin pregnancies because there are two placentas releasing their own DNA. It may not be possible to know what type of twin pregnancy you're having.

NIPT is not suitable for everyone. Your midwife will explain to you if there's a reason you cannot have NIPT, for example if you've had a recent blood transfusion, cancer or have a condition that involves chromosomes 21, 18 or 13.

Diagnostic tests

- These tests are offered to women whose screening tests show they have a higher chance of being a carrier for (or having) sickle cell or thalassaemia. Or whose baby has a higher chance of having Down's syndrome, Edwards' syndrome or Patau's syndrome.
- There are two types of diagnostic tests: chorionic villus sampling (CVS) and amniocentesis.
- Diagnostic tests can tell you for definite if your baby has one of these conditions.
- Diagnostic tests increase the chance of miscarriage.

It's very important you know that **it's your choice whether you have the diagnostic tests or not**.

Your health professional will talk it through with you and answer any questions you have. They'll support you to make decisions that feel right for you. Other sources of information and support are listed at the end of this booklet.

Diagnostic testing is not usually recommended after 22 weeks of pregnancy.

CVS (chorionic villus sampling)

CVS can be done from 11 weeks of pregnancy. It's usually only offered in a specialist centre.

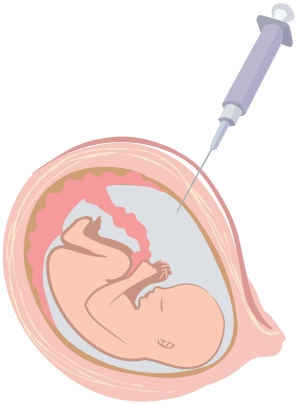
With the help of an ultrasound scan, a specialist doctor (obstetrician) will guide a fine needle through your abdomen (tummy) and will take a small sample of tissue from the placenta.

Chromosomes from the placenta can be counted from the sample. CVS does not give a clear result in around two in every 100 samples. If this happens you may be offered a repeat test. Your obstetrician will help you understand what your results mean.

Placenta

The placenta is inside your womb. It links your blood to your baby and provides nourishment.

Amniocentesis



Amniocentesis (you might hear it shortened to 'amnio') can be carried out after 15 weeks of pregnancy. It usually takes about 10 minutes.

An ultrasound scan will check your baby's position in the womb. A specialist doctor (obstetrician) will guide a fine needle through your abdomen (tummy) into your womb. The doctor can then take a sample of the fluid surrounding your baby (called amniotic fluid).

Your baby's chromosomes can be counted from the sample. Amniocentesis does not give a clear result in around one in every 100 samples. If this happens, you may be offered a repeat test.

Referral to the fetal medicine team

Your health professional may refer you to the fetal medicine team.

The team includes a specialist doctor, midwife and other health professionals. The team, which could be based in another hospital, may offer you further tests and will give information and advice about any health issues, chromosomal conditions or disabilities you or your baby might have. You'll usually have an appointment within a few days.

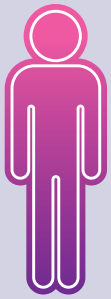
How safe are diagnostic tests?

CVS and amniocentesis are not completely safe but they're the only way to know for sure if your baby has a condition. It's your choice and health professionals will support you whatever you decide.

Support

Organisations listed at the back of this booklet can also provide further information and support.

Diagnostic tests have some risks. Around 1 in every 200 (0.5%) women who have a diagnostic test will miscarry as a result of the test. The risk may be higher in twin pregnancies.



Are diagnostic tests painful?

Many women find the tests uncomfortable, sometimes painful.

Some discomfort in your lower abdomen (tummy) for a couple of days is usual, and you can take paracetamol for this. You should take things easy and avoid hard exercise for a day or two afterwards. If the discomfort carries on beyond this, or if you have any other worries, please contact your midwife.

What happens if the diagnostic test finds a condition?

If the diagnostic test shows your baby does have one of the conditions, your midwife or obstetrician will discuss your result with you and give you time to understand what it means.

Some parents may decide to continue with the pregnancy, while others will feel that ending the pregnancy is right for them. Only you can decide what is best for you and your family.

Your midwife can also tell you information about organisations that can help provide emotional and practical support.

What do we do with your information?

We keep a record of your personal screening information, including test results. Your personal health information will be kept private, which means it is only shared with other staff involved in your care. We regularly review what we do to make sure we offer the best service possible.

The results of some screening and tests in pregnancy are shared with Public Health Scotland, which is part of the NHS. Public Health Scotland uses the information to support ongoing monitoring and improvement of pregnancy screening. For more information, visit **www.publichealthscotland.scot/our-privacy-notice**

Babies with some of the conditions that are screened for in pregnancy are included in the secure congenital and rare condition register, which is maintained by Public Health Scotland. This register monitors how many babies have these conditions and supports the planning and improvement of health, care and other public services. Information about the register can be found at **www.publichealthscotland.scot/cardriss**

You have rights in relation to the access and the use of your personal health information. For more information about your rights or how the NHS uses your personal information, contact the NHS inform helpline free on **0800 22 44 88** (textphone **18001 0800 22 44 88**) or visit **www.nhsinform.scot/confidentiality** and **www.nhsinform.scot/data-protection**

Information and support

The information about screening tests in pregnancy can be a lot to take in. Please talk to your health professional if you have any questions or worries.

You can ask them which organisations may be able to give you support that's right for you and your family. The following contact details may be useful.

NHS inform

For information on screening tests in pregnancy please visit **www.nhsinform.scot/pregnancyscreening** or phone: **0800 22 44 88 (textphone: 18001 0800 22 44 88)**

Antenatal Results and Choices (ARC)

Offers information and support to parents who are making decisions around antenatal testing and whether to continue pregnancy or end pregnancy.
Phone: **0207 713 7486** from a mobile or visit:
www.arc-uk.org

Contact

Provides information and support to parents and carers of children with any additional need or disability.
Phone: **0808 808 3555** (voice and text) or visit:
http://contact.org.uk

Down's Syndrome Scotland

Supports expectant couples and new parents throughout Scotland to make informed decisions by providing up-to-date, accurate and balanced information about people living with Down's syndrome.
Phone **0300 030 2121** or visit:
www.dsscotland.org.uk

Positively UK

Offers a range of peer support, advice, information and advocacy services for HIV-positive women and men.

Phone: **0207 713 0444**

http://positivelyuk.org

SOFT UK

Supports families affected by Patau's syndrome, Edwards' syndrome and related disorders.

Phone: **0300 102 7638**

www.soft.org.uk

Spina Bifida Hydrocephalus Scotland

A family support service for those affected by spina bifida, hydrocephalus and allied conditions.

Helpline: **0345 521 1300**

www.sbhscotland.org.uk

UK Thalassaemia Society

Phone: **0208 882 0011**

http://ukts.org

Waverley Care

Provides care and support to people living with HIV and hepatitis C and their partners, families and carers.

Phone: **0131 558 1425**

www.waverleycare.org

Sickle Cell Society

The Sickle Cell Society supports and represents people affected by a sickle cell disorder to improve their overall quality of life.

Phone: **0208 961 7795**

www.sicklecellsociety.org

British Pregnancy Advisory Service (BPAS)

BPAS provides help to women with an unplanned pregnancy or a pregnancy they choose not to continue with.

Phone: **0345 730 4030**

Email: **info@bpas.org**

www.bpas.org


Translations


Easy read


BSL


Audio


Large print


Braille

For more information, or for translations and other formats:

www.nhsinform.scot/otherformatspregnancyscreening

phs.otherformats@phs.scot

0131 314 5300

Public Health Scotland is Scotland's national agency for improving and protecting the health and wellbeing of Scotland's people.

Corporate member of Plain English Campaign
Committed to clearer communication
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