Your **pregnancy**, your choice

What you need to know after getting a higher-chance screening result for Down's syndrome, or Edwards' syndrome or Patau's syndrome

NHS SCOTLAND

Public Health Scotland

This booklet tells you about:

- Down's syndrome, Edwards' syndrome and Patau's syndrome
- what having a baby with one of these conditions could mean
- your choices for further testing.

All tests in this booklet are provided free in Scotland by the NHS.

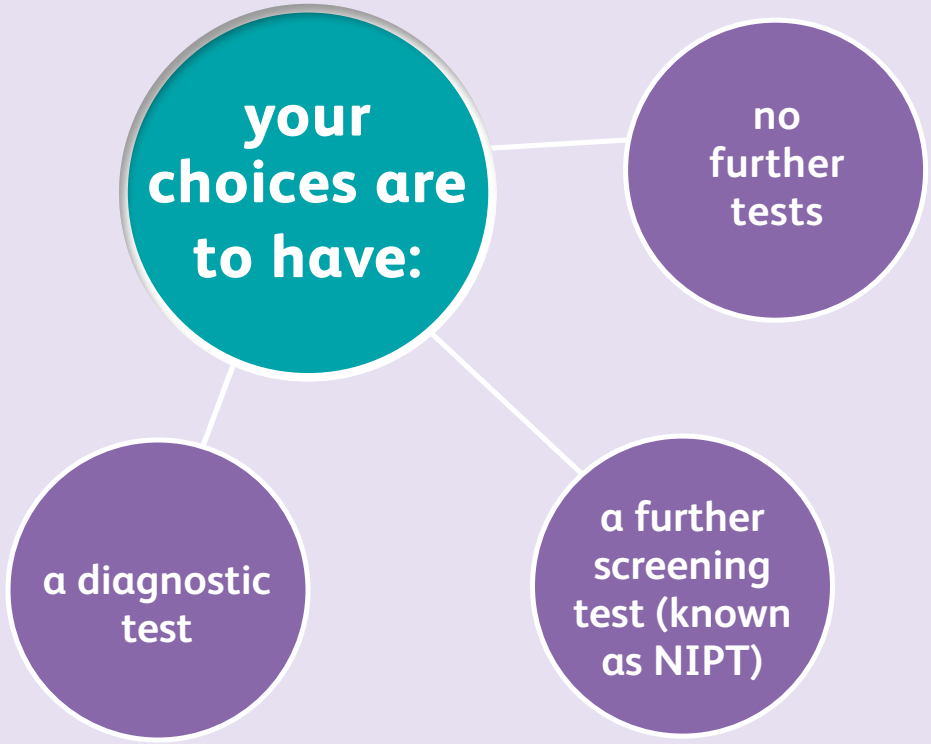
Why you've been given this booklet

Your first screen has shown your baby has a higher chance of having Down's syndrome, or Edwards' syndrome or Patau's syndrome. This does not mean your baby definitely has one of the conditions.

The term 'your midwife' is used throughout this booklet. This could also be your GP, obstetrician (a specialist doctor), sonographer (a professional who carries out ultrasound scans) or any other health professional involved in your care during your pregnancy.

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.



Results from diagnostic testing may affect whether you decide to continue or end your pregnancy. You can find more information about your choices on pages 8 to 19.

Down’s syndrome

- A baby with Down’s syndrome is born with more of chromosome 21 in all or some of their cells. 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 may live independently, have jobs, be in relationships and socialise with minimal support. Evidence suggests most families of people with Down’s syndrome 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 can be less common.

Around 5 in 10 babies born with Down’s syndrome will have heart issues and fewer than 1 in 5 of those may need surgery. They’re more likely to have muscle tone and digestion issues, and may have reduced hearing or vision. As adults, people with Down’s syndrome 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 issues 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 **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 can not 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 are likely to 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 issues, some of which are serious.

Around 9 in 10 babies will have heart issues, 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 **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 are likely to 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 issues, some of which are serious.

Around 8 in 10 babies may have heart issues, 6 in 10 may have issues with brain development and around 6 in 10 may have a cleft lip and palate. Some 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**

Your choices for further testing

Your first screen has shown that your baby has a higher chance of having Down's syndrome, or Edwards' syndrome or Patau's syndrome. This does not mean your baby definitely has one of the conditions.

Your choices are to have:

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

Further testing can give you more accurate information about how likely it is your baby has Down's syndrome, or Edwards' syndrome or Patau's syndrome.

Your midwife will discuss your result with you and explain your choices for further testing

Your choices

Even if the results will not affect whether you decide to continue or end your pregnancy, you can have any of the tests just for information to help you prepare for a baby who may need additional care and support.

People choose to have further tests or not to have further tests for lots of different reasons and your choices will be personal. 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.

Tell your midwife if at any point you're not sure about the choices you've made or if you'd like more information.

Your midwife will respect your choices and keep them private. They'll look after you and your baby, and will tell you about support organisations that can help and give you more information.

Twin pregnancies

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.

No further tests

- It's your choice whether to have a further screening test or diagnostic test for Down's syndrome, Edwards' syndrome and Patau's syndrome.
- If you choose not to have these tests, you can agree a pregnancy plan with your midwife that feels right for you.
- You'll still be offered other types of tests and scans as part of your routine pregnancy care.
- If your baby does have one of the conditions, it may be found during your routine pregnancy care or after your baby's birth.

What will happen during your pregnancy

If you choose not to have further tests for Down's syndrome, or Edwards' syndrome or Patau's syndrome you'll agree a pregnancy plan with your midwife.

You can still choose to have other types of tests or scans as part of your routine pregnancy care. For example, you'll be offered a mid-pregnancy scan which looks for lots of things, including issues with your baby's growth, heart or kidneys.

Finding out your baby has one of the conditions during routine tests

A mid-pregnancy screening scan (18–21 weeks of pregnancy) is also known as a fetal anomaly scan. If you choose to have this, your midwife may see something during the scan which suggests your baby has one of these conditions, or they may see something during your baby's post-birth examination.

Whether you find out about your baby's condition during your routine pregnancy care or after your baby is born, your midwife will support you to understand and make decisions which feel right for you and your baby.

If your baby has Edwards' syndrome or Patau's syndrome

As miscarriage and stillbirth are more likely for babies with Edwards' syndrome or Patau's syndrome, you may find out about the condition if they do not survive pregnancy or birth.

Babies born alive with Edwards' syndrome or Patau's syndrome are likely to need a lot of medical care and may live a short life. You may need to make decisions about further tests or treatment quickly. Your midwife will support you to understand your options and make choices about caring for your baby. They'll also tell you about organisations that can help.

Non-invasive prenatal testing (NIPT)

- NIPT is a blood test which is more accurate than the first pregnancy screening test you had.
- 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 genetic conditions, or the gender of your baby, as part of NHSScotland's pregnancy screening.

Should I have NIPT?

If you do not want to go straight to having a diagnostic test, your NIPT result may help you to 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.

What result could I get?

Low-chance result

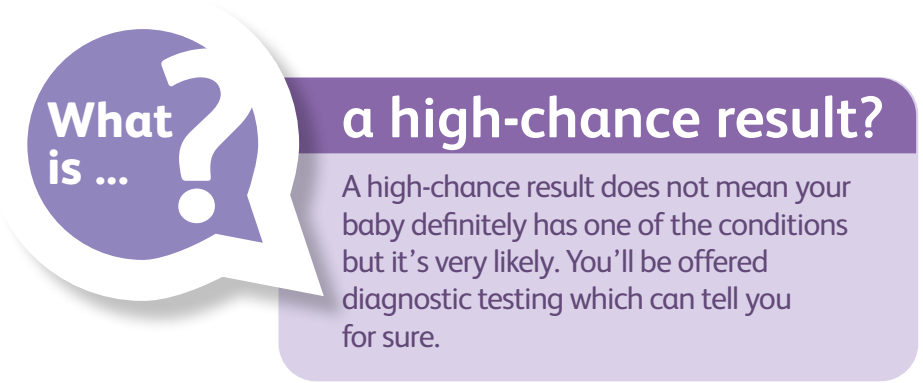
Most women who have NIPT will receive a low-chance result. This means it's unlikely your baby has one of the conditions. If you get this result, you'll not be offered any further tests for these conditions.

There is a small chance that you may receive a low-chance result and your baby does have one of the conditions. This is known as a false negative.

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 is 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 correct:

- 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, like you, who've already had a higher-chance result from your 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 blood transfusion, cancer or have a condition that involves chromosomes 21, 18 or 13.

Diagnostic tests

- There are two types of diagnostic tests: chorionic villus sampling (CVS) and amniocentesis.
- CVS can be carried out between 11 and 14 weeks of pregnancy. Amniocentesis can be carried out after 15 weeks of pregnancy.
- Diagnostic tests can tell you for definite if your baby has Down's syndrome, or Edwards' syndrome or Patau's syndrome.
- 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.

When you'll be offered diagnostic testing

You'll be offered diagnostic testing if you receive a higher chance of having a baby with Down's syndrome, Edwards' syndrome or Patau's syndrome from previous screening tests.

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

Choosing between CVS or amniocentesis

Your midwife will help you decide which test is right for you, based on your stage of pregnancy. Diagnostic tests sometimes find other conditions, but this is rare.

How a CVS test works

An ultrasound scan will check your baby's position in the womb. A specialist doctor (obstetrician) will guide a fine needle through your tummy and take a small sample of tissue from the placenta. The chromosomes in your placenta can be counted from this sample. Your obstetrician will help you understand what your results mean.

How amniocentesis works

An ultrasound scan will check your baby's position in the womb. A specialist doctor (obstetrician) will guide a fine needle through your tummy and take a small sample of the amniotic fluid which is around your baby. Your baby's chromosomes can be counted from this sample. Your obstetrician will help you understand what your results mean.

Are diagnostic tests painful?

Many women find diagnostic testing uncomfortable, sometimes painful. You might have some discomfort in your lower abdomen which should pass in a few days.

Is diagnostic testing completely safe?

No. Diagnostic testing is not completely safe. 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.

What result could I get?

Your result can come in two parts. It's usually best to wait for all parts before making any decisions about your pregnancy.

The test shows that your baby does not have any of the conditions

If the diagnostic test shows that your baby does not have Down's syndrome, or Edwards' syndrome or Patau's syndrome you'll not usually be offered any further tests for these conditions. You'll continue to get the routine tests, scans and care offered during all pregnancies.

The test shows that your baby does have one of the conditions

If the diagnostic test shows that 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. If you asked to receive your result by phone, the midwife or obstetrician who calls you will arrange a face-to-face appointment to discuss your result and your choices.

Further support if your baby has Down's syndrome, or Edwards' syndrome or Patau's syndrome

You may have found out your baby has a high chance of having one of the conditions by having NIPT or during routine scans. Or, you may have chosen diagnostic tests and know for sure.

If either of these things has happened, your midwife will tell you which support organisations can give you more information about living with the condition your baby has, or is very likely to have. Some contact details are included in this leaflet.

Your midwife will also offer to refer you to a specialist team which includes specialist doctors, midwives and other health professionals. The team may be in another hospital. Your midwife will help you to understand who the specialists are and what they do.

If you need to choose what to do next, support organisations and health professionals can answer any questions you have and support you in reaching decisions that feel best for you and your baby.

Whether you choose to continue with your pregnancy or not, your choices will be respected and you'll get the care and support you need.

Information and support

Down's Syndrome Scotland

Down's Syndrome Scotland is the only charity in Scotland dedicated to supporting people with Down's syndrome and their parents, carers and families. Down's Syndrome Scotland provides 'all through life' support nationally across Scotland.

Phone: 0300 030 2121
www.dsscotland.org.uk

SOFT UK

SOFT UK supports families affected by Patau's syndrome, Edwards' syndrome or related disorders.

www.soft.org.uk

Antenatal Results and Choices (ARC)

ARC offers information and support to parents who are making decisions around antenatal testing and whether to continue pregnancy or end pregnancy.

Phone: 0845 077 2290 or **0207 713 7486** from a mobile.
www.arc-uk.org

NHS inform

NHS inform is for anyone in Scotland looking for evidence-based health and care information.

www.nhsinform.scot

Information about NIPT

Following UK National Screening Committee recommendations, NHSScotland has rolled out NIPT for an evaluation period of at least three years. During this time, NHSScotland will collect information to check how well NIPT performs in a higher-chance population. This means people, like you, who've already had a higher-chance result from your first screening test. They will use this information to contribute to a UK-wide report.

What happens to my information after screening?

Your personal health information will be kept private, which means it's only shared with other staff involved in your care. Processes are reviewed regularly to make sure you're offered the best service possible.

You have rights in relation to the access and the use of your personal health information.

Contact the NHS inform helpline free on **0800 22 44 88** (textphone **18001 0800 22 44 88**) or visit **www.nhsinform.scot/confidentiality** or **www.nhsinform.scot/data-protection**

Translations | Easy read | BSL | Audio | Large print | Braille

Other formats available at

www.nhsinform.scot/otherformatsypyc

0131 314 5300

phs.phs-otherformats@nhs.net

Scan me

If you have questions about your pregnancy or the information in this leaflet, please contact:

Published by Public Health Scotland
1 South Gyle Crescent
Edinburgh EH12 9EB

Established on 1 April 2020, Public Health Scotland is Scotland's national public agency for improving and protecting the health and wellbeing of Scotland's people.

Corporate member of Plain English Campaign
Committed to clearer communication
489

www.publichealthscotland.scot

0098 8/2020