

Down's syndrome, Edwards' syndrome and Patau's syndrome: options after a higher chance screening result

Public Health England (PHE) created this information on behalf of the NHS. In this information, the word 'we' refers to the NHS service that provides screening.

Your choices after a higher-chance screening result

Non-invasive prenatal testing (NIPT) is currently being rolled out across England. You may or may not be eligible for NIPT depending on which NHS Trust provides your care during pregnancy. Your midwife will be able to provide more details.

This information is for pregnant women with singleton (having only one baby) and twin pregnancies who have received a higher-chance result from the combined or quadruple screening test. These tests are for Down's syndrome (also known as Trisomy 21 or T21), Edwards' syndrome (Trisomy 18 or T18) and Patau's syndrome (Trisomy 13 or T13).

If you are in this situation, you will have 2 or 3 options depending on whether you are eligible for NIPT. These options are:

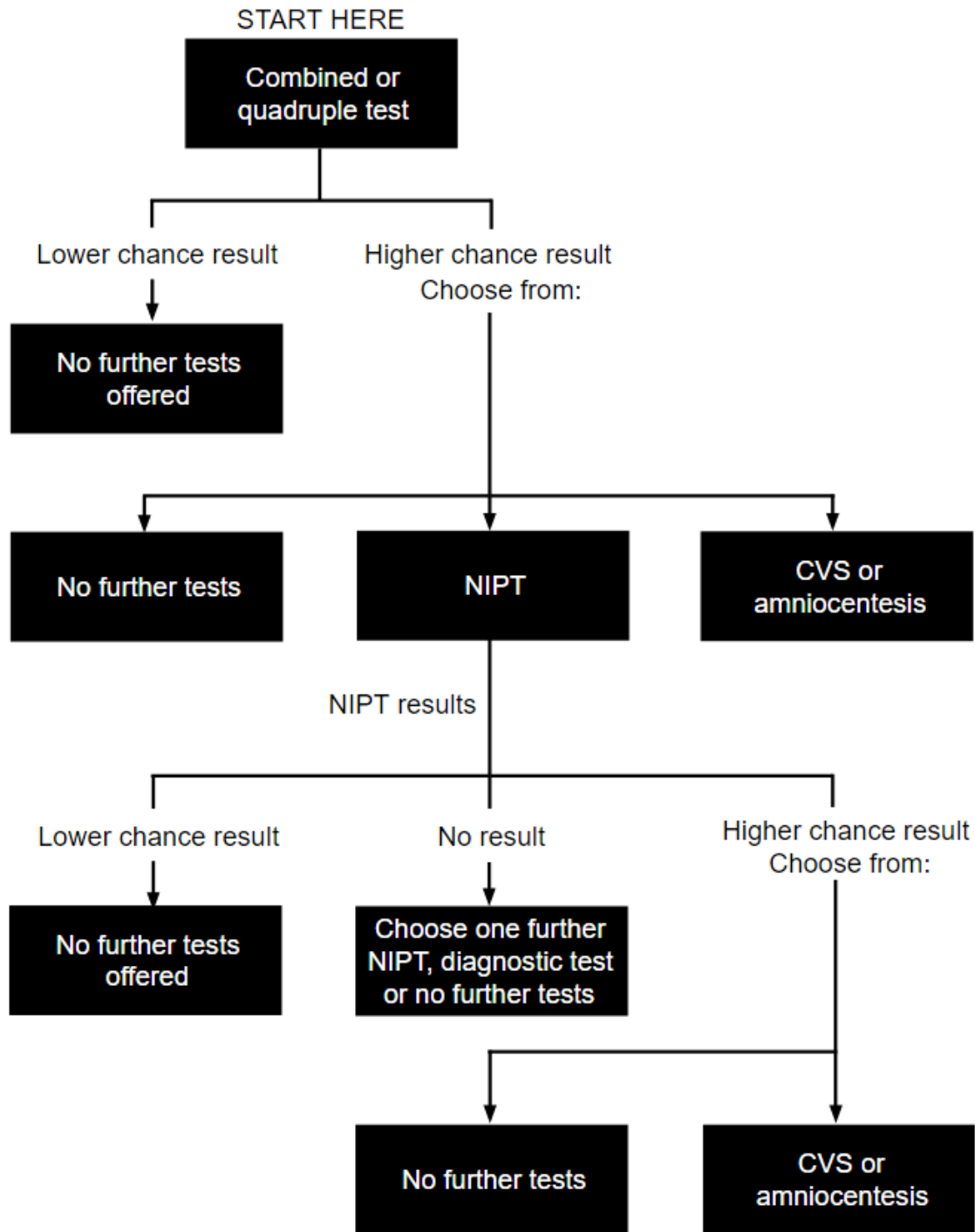
- no further testing
- a more accurate screening test – NIPT – which tells you whether or not your baby is likely to have the condition
- an invasive diagnostic test – chorionic villus sampling (CVS) or amniocentesis – which gives a definite 'yes' or 'no' answer

This leaflet explains these 3 options. They are summarised in the flowchart below. It also provides information about Down's syndrome, Edwards' syndrome and Patau's syndrome. It should support the discussions you have with your healthcare professionals.

You should be given time to think through your options. You can ask for more information and do not have to decide straight away.

It is your decision. Your decisions will be respected and healthcare professionals will support you. You should speak up if you feel your decisions are not being respected.

Find more information on the NHS website at www.nhs.uk/pregnancy/your-pregnancy-care/screening-for-downs-edwards-pataus-syndrome.



This flowchart shows your options. A [full text description of this pathway](http://www.gov.uk/government/publications/fetal-anomaly-screening-care-pathways/downs-syndrome-edwards-syndrome-and-patau-syndrome-screening-pathway-combined-or-quadruple-test-taken-on-or-after-1-june-2021) is also available at www.gov.uk/government/publications/fetal-anomaly-screening-care-pathways/downs-syndrome-edwards-syndrome-and-patau-syndrome-screening-pathway-combined-or-quadruple-test-taken-on-or-after-1-june-2021.

Whatever results you get from any of the screening or diagnostic tests, you will get support and care to help you decide what to do next.

You can have any test just for information, even if the result will not affect whether you decide to continue or end your pregnancy.

There are a number of organisations you can contact for support.

About the conditions

All women have a chance of having a baby with Down's syndrome, Edwards' syndrome or Patau's syndrome. Nothing you or your partner has done or not done makes any difference to this chance.

How the conditions affect the baby will depend on a number of factors and cannot be predicted before the baby is born.

Down's syndrome (Trisomy 21)

All people with Down's syndrome have a learning disability. This means that development and learning new things may take longer. There is now a greater understanding of how children with Down's syndrome learn. Support in schools can be provided.

Opportunities for inclusion within society are greater now than ever before. Individuals vary greatly, however, and have different needs. These needs cannot be predicted before the baby is born.

Some health conditions are more common in people with Down's syndrome. These include heart conditions, as well as problems with vision and hearing. Many health conditions can be treated, though unfortunately around 5% of babies with Down's syndrome will not live past their first birthday. For babies without serious health problems survival is similar to that of other children. Most people with Down's syndrome will live into their 60s or longer.

Most people who have Down's syndrome lead healthy and fulfilled lives. Parents say that their children have a positive influence on family life. With support, many more adults are now able to get jobs, have relationships and live in homes of their choosing.

Find out more and see photos of children and adults with Down's syndrome on the NHS website at www.nhs.uk/conditions/Downs-syndrome.

The Down's Syndrome Association also provides information to pregnant women and couples at www.downs-syndrome.org.uk/for-new-parents/being-pregnant. They have the time and expertise to listen and give more information about Down's syndrome. Their helpline is 0333 12 12 300.

Edwards' syndrome (Trisomy 18) and Patau's syndrome (Trisomy 13)

Edwards' syndrome and Patau's syndrome are considered to be life-limiting conditions. This means they affect how long a baby is likely to survive. Sadly the survival rates are low and of those babies born alive, around 13% with Edwards' syndrome and 11% with Patau's syndrome will live past their first birthday. Children surviving to one year have an 80% chance of reaching 5 years. Some will get to adulthood but this is much rarer.

All babies born with Edwards' syndrome and Patau's syndrome will have delayed learning and physical development and a wide range of health challenges, some of which can be extremely serious. They may have problems with their heart, swallowing and feeding difficulties, seizures and breathing difficulties including 'pauses' (apnoea) in their breathing. They will also have a low birthweight.

Despite their difficulties children can slowly make progress in their development. Older babies and children can show some level of communication and some will stand and walk with assistance. Older children would need to attend a specialist school.

Find out more and see photos of children with Edwards' syndrome and Patau's syndrome on the NHS website at www.nhs.uk/conditions/edwards-syndrome and www.nhs.uk/conditions/pataus-syndrome.

SOFT provides support and help for families affected by Edwards' syndrome and Patau's syndrome. See www.soft.org.uk.

No further testing

If you decide to have no further testing you will still be offered all other parts of your routine antenatal care. Your midwife will explain what this means for you.

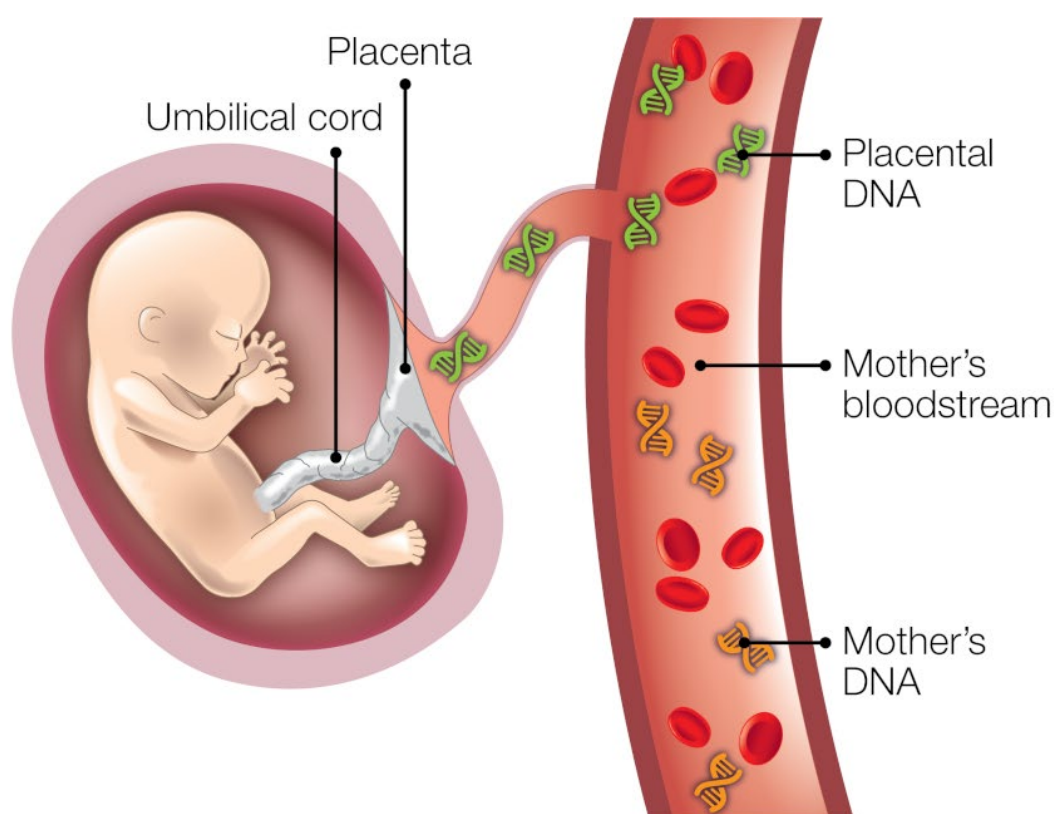
Non-invasive prenatal testing

Non-invasive prenatal testing (NIPT) is a much more accurate screening test than the first screening test (the combined or quadruple test). It involves taking some blood from your arm. It is completely safe and will not harm the baby.

NIPT does not tell you for certain if the baby has Down's syndrome, Edwards' syndrome or Patau's syndrome. Like any screening test, NIPT does not give a definite answer. It will not be used to find other conditions or tell you whether the baby is a boy or a girl. NIPT is offered up to 21⁺⁶ weeks of pregnancy as part of NHS screening.

How NIPT works

NIPT involves measuring the DNA (genetic material) in your blood. Your baby is joined to the placenta in your womb via the umbilical cord. During your pregnancy the placenta releases some DNA into your bloodstream. As a result, your blood contains a mixture of DNA from you and from the baby's placenta.



If NIPT finds more DNA than expected for chromosomes 21, 18 or 13 in your blood it could mean that the baby has one of the conditions.

In most cases, the placental DNA will be the same as the baby's DNA. One of the reasons why NIPT is not 100% accurate is because it tests DNA from the placenta and in rare cases this is not identical to the baby's DNA. This is called confined placental mosaicism.

Note that NIPT blood samples may be kept by the NIPT laboratory for quality purposes for up to 5 years. Please tell your midwife if you do not wish your NIPT blood sample to be kept in this way.

NIPT choices

If you decide to have NIPT, you can have it for:

- all 3 conditions
- Down's syndrome only
- Edwards' syndrome and Patau's syndrome only

Before you make a decision you should discuss your options with your healthcare professional. In particular, if you had a very high chance result ('1 in 2' to '1 in 10') from the first screening test (the combined or quadruple test), the benefits of NIPT are less clear.

NIPT may not be suitable for everyone. You should discuss with your healthcare professional to check it's suitable for you.

NIPT results

You can usually choose whether to receive NIPT results by phone or face-to-face. Discuss your preference with your midwife. Most women will get their result within 2 weeks of having NIPT.

Depending on your NIPT screening choice, you will have up to 3 results reported. You could get:

- one for Down's syndrome, one for Edwards' syndrome and one for Patau's syndrome, or
- one for Down's syndrome, or
- one for Edwards' syndrome and one for Patau's syndrome

It is important to remember that NIPT is still a screening test. The result will be reported as either higher-chance or lower-chance for the conditions screened for.

NIPT will give an accurate result for most women who choose to have it but there can also be false positive and false negative results. A false positive result means getting a higher-chance result when the baby does not have the condition. A false negative result is getting a lower-chance result when the baby does have the condition.

Evidence suggests NIPT is less accurate for Edwards' syndrome and Patau's syndrome than it is for Down's syndrome. While it has been suggested that this is because these babies have smaller placentas, the reasons are not fully understood. NIPT might also be less accurate in twin pregnancies.

We always advise that you discuss your individual NIPT result with your healthcare professional.

Lower-chance result

Most women who have NIPT will get a lower-chance result. NIPT is a screening test so is not 100% accurate but it has very few false negative results. If you had a very high chance result from the combined or quadruple test (such as between '1 in 2' and '1 in 10') then a false negative result is more common. So there will be some women who receive a lower-chance NIPT result who do have a baby with one of the conditions.

You will not be offered a diagnostic test after a lower-chance NIPT result. You will continue to receive usual antenatal care.

Higher-chance result

Having a higher-chance NIPT result does not mean that the baby definitely has the condition but it is very likely. At least 90% (9 out of 10) of women who receive a higher-chance NIPT result for Down's syndrome will be carrying a baby who has the condition.

After a higher-chance NIPT result, you will be offered diagnostic testing. But you may prefer to have no further tests.

No result

In a small number of cases, NIPT will not give a result. If you do not get a result you can then choose between one further NIPT, a diagnostic test or having no further testing. Getting no result could be caused by a technical issue relating to the test. It could also happen if there is not enough DNA present in the blood sample, for example because you have a body mass index (BMI) over 30 or you are having twins.

If you get a higher-chance result or no result, healthcare professionals will provide more information and support.

7. Diagnostic testing

You can choose to have diagnostic testing straight away or after a higher-chance NIPT result.

Diagnostic tests give a definite answer. They test cells from the placenta or fluid surrounding the baby. Diagnostic tests can find conditions other than Down's syndrome, Edwards' syndrome or Patau's syndrome but this is rare.

One out of every 200 (0.5% of) women who have a diagnostic test will miscarry as a result of the test.

There are 2 types of diagnostic test: chorionic villus sampling (CVS) and amniocentesis. Your health professional will discuss your options with you. See www.gov.uk/government/publications/cvs-and-amniocentesis-diagnostic-tests-description-in-brief.

CVS (chorionic villus sampling)

This is usually done from 11 to 14 weeks of pregnancy but can be done later. A fine needle, usually put through your abdomen, is used to take a tiny sample of tissue from the placenta.

You will get the first CVS result in about 3 days. If this confirms the NIPT result, and there were relevant scan findings, then your doctor will discuss your options with you straight away. However, if your scan did not show anything relevant then we recommend waiting for the second CVS result, within 2 weeks, before making a decision about whether or not to end your pregnancy.

This is because the second result reflects the DNA of the baby rather than the placenta and will not be affected by confined placental mosaicism.

Amniocentesis

This is usually done after 15 weeks of pregnancy. A fine needle is passed through your abdomen into the uterus to collect a small sample of the fluid surrounding the baby.

Amniocentesis results are usually available in around 3 days. They are a true reflection of the baby's DNA. In some circumstances it may be more appropriate to wait for an amniocentesis rather than having a CVS.

Continuing support and care

Following the diagnostic test result, you may need to decide what to do next. Only you know what is the best decision for you and your family.

You might want to learn more about Down's syndrome, Edwards' syndrome or Patau's syndrome. It can be helpful to speak to a support organisation for parents. If

you feel it would help, these groups may be able put you in touch with someone who has been in a similar situation. Contact details are on the back cover.

Deciding whether or not to continue with the pregnancy will be very personal to you. If you decide to continue with your pregnancy, your doctor or midwife will discuss your care and how best to care for your baby during pregnancy and after birth.

If you decide to end your pregnancy, you will be given information about what this involves and how you will be supported. This will include the choices you have about the method of termination.

Whatever decision you make, your healthcare professionals will support you.

Support organisations

Antenatal Results and Choices (ARC) is a national charity that provides support for people making decisions about screening and diagnosis and whether or not to continue a pregnancy. You can call them on 020 713 7486. See www.arc-uk.org/.

The Down's Syndrome Association (DSA) provides information and support for parents. You can call them on 0333 12 12 300 or visit www.downs-syndrome.org.uk.

SOFT UK provides information and support to families about Edwards' syndrome and Patau's syndrome. See www.soft.org.uk.