# Non-invasive prenatal testing (NIPT) for previous trisomy (R445 pathway)

Information for parents about tests in pregnancy following a previous pregnancy with Down's syndrome (Trisomy 21 or T21), Edwards' syndrome (Trisomy 18 or T18) or Patau's syndrome (Trisomy 13 or T13).

Developed by:
NHS England R445 Working Group
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# Contents

# Guidance

This leaflet provides information about the screening tests offered during pregnancy to anyone who has had a previous pregnancy or baby with Down's syndrome (Trisomy 21 or T21), Edwards' syndrome (Trisomy 18 or T18) or Patau's syndrome (Trisomy 13 or T13). Please speak with your midwife, doctor or genetic counsellor to see which, if any, is the right test for you.

## What are my testing options if I have previously had a pregnancy or child with Down's syndrome, Edwards' syndrome or Patau's syndrome?

We understand that you might need different information for your current pregnancy when you have previously had a pregnancy or baby with Down's syndrome (also known as Trisomy 21 or T21), Edwards' syndrome (Trisomy 18 or T18) or Patau's syndrome (Trisomy 13 or T13). This leaflet will outline what options you have for screening and diagnostic testing in your current pregnancy.

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

You may have had information about these conditions in previous pregnancies or have lost a baby with or have a child or family member living with either Down's syndrome, Edwards' syndrome or Patau's syndrome. However, to support you when making choices about screening or diagnostic tests in pregnancy, we have created a summary of the most up to date information about these three conditions and provided links to organisations that can offer further information and support.

## Down's syndrome (Trisomy 21)

Down's syndrome is caused by an extra copy of chromosome 21 in all or some cells of the body. A person with Down's syndrome will have some level of learning disability. This means they will find it harder than most people to understand and to learn new things. They may have communication challenges and difficulty managing some everyday tasks. People with Down's syndrome have distinctive facial features but they do not all look the same.

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

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

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

For more information about Down’s syndrome please go to:
Down's syndrome (trisomy 21) - NHS (www.nhs.uk)

## Edwards’ syndrome (Trisomy 18) and Patau’s syndrome (Trisomy 13)

Babies with Edwards’ syndrome have an extra copy of chromosome 18 in all or some cells. Babies with Patau’s syndrome have an extra copy of chromosome 13 in all or some cells.

All babies born with Edwards’ syndrome and Patau’s syndrome will have a learning disability and a wide range of physical challenges, which can be extremely serious and present at birth.

Sadly, the survival rates are low and of those babies born alive only around 13% with Edwards’ syndrome and 11% with Patau’s syndrome will live past their first birthday. Some babies may survive to adulthood, but this is rare. Despite their difficulties, children can slowly make progress in their development.

For more information about Edwards’ syndrome please go to:
Edwards' syndrome (trisomy 18) - NHS (www.nhs.uk)

For more information about Patau’s syndrome please go to:
Patau's syndrome - NHS (www.nhs.uk)

## Screening tests for Down’s syndrome, Edwards’ syndrome and Patau’s syndrome for women **without** a history of a previous pregnancy with Trisomy 21, Trisomy 18 or Trisomy 13

All eligible women are offered the choice of screening tests to work out the chance of their baby having Down’s, Edwards’ or Patau’s Syndrome.  These tests are known as either the *Combined test* or *Quadruple test* depending on how many weeks pregnant you are. They involve taking a sample of blood from the pregnant woman’s arm to check specific hormone levels and an ultrasound scan to get measurements from the baby.

Further information about the screening tests, Down’s syndrome, Edwards’ syndrome and Patau’s syndrome can be found in *Screening Tests for you and your baby* leaflet provided by your healthcare professional and available online.

**You are offered a different testing pathway to the combined test or quadruple test as you have had a previous pregnancy or baby with one of these conditions. The options available to you are explained in more detail in the next section.**

## Your testing options

There are three options available to you:

1. **No testing**
   Continue with usual antenatal care.
2. **Non-Invasive Prenatal Testing (NIPT)**
   A blood test which can tell you whether or not your baby is more likely to have the condition.
3. **An invasive diagnostic test (chorionic villus sampling (CVS) or amniocentesis)** Usually provides a ‘yes’ or ‘no’ answer if the baby has the condition.

Which, if any, testing to have in your pregnancy is a personal decision. Your health care team will support you whatever you decide. The next sections give more information about each of these options.

## 1. No Testing

If you decide to have no further testing, you will still be offered all other parts of your usual antenatal care. Your midwife will explain what this means for you.
If you have any questions/concerns, please contact your healthcare provider. For example, Obstetrician (doctor for pregnancy), Midwife or GP.

## 2. Non-Invasive Prenatal Testing

Non-invasive prenatal testing (NIPT) is a screening test which is more accurate than the Combined or Quadruple test. This test involves measuring the DNA (genetic material) in your blood by taking a blood sample from your arm. It is completely safe and will not harm your baby.

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.

## How NIPT works

NIPT involves measuring the DNA (genetic material) in your blood. Your baby is joined to the placenta in your womb by 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 chromosome 21 in your blood, it could mean that the baby has Down's syndrome.
- If NIPT finds more DNA than expected for chromosome 18 in your blood, it could mean that the baby has Edwards' syndrome.
- If NIPT finds more DNA than expected for chromosome 13 in your blood, it could mean that the baby has Patau's syndrome.

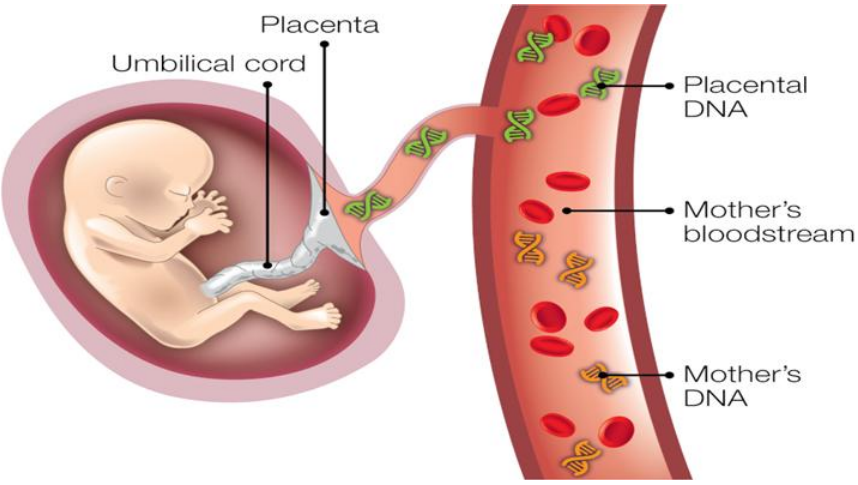

Figure one: Image shows the mother's and placental DNA in the mother's bloodstream. This figure is from: [Your choices after a higher-chance screening result - GOV.UK (www.gov.uk)]

Like any screening test, NIPT does not give a definite answer, and it cannot tell you for certain if the baby has Down's syndrome, Edwards' syndrome or Patau's syndrome.

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 in rare cases the DNA from the placenta is not the same as the baby's DNA. This is called confined placental mosaicism. Your healthcare provider will talk to you about this in more detail if you decide on this option.

NIPT is a more accurate screening test than the combined or quadruple test, as it is looking at DNA within your bloodstream. However, it will not be able to give you any information about the chance of your baby having any other conditions or tell you whether the baby is a boy or a girl.

Evidence suggests NIPT is less accurate for Edwards' syndrome and Patau's syndrome than it is for Down's syndrome, although the reasons are currently not fully understood. NIPT might also be less accurate in twin pregnancies.

## Who can have NIPT on the R445 pathway?

NIPT can be offered to women who are at least 10 weeks pregnant, confirmed by an ultrasound scan. NIPT can be performed in singleton (carrying one baby) and twin pregnancies up to and including 21 weeks and 6 days ($21^{+6}$).

**You cannot have NIPT if:**

- your previous trisomy pregnancy was not a full trisomy 21,18 or 13 (e.g. it was a translocation or mosaicism)
- you are over 21 weeks and 6 days pregnant
- you are pregnant with more than two babies
- you have received a blood transfusion in the last 4 months
- you have ever had donor stem cell therapy, bone marrow or organ transplant
- your current pregnancy is a vanished twin pregnancy or is a twin pregnancy where one of the babies has miscarried
- you have Down's syndrome or mosaicism trisomy 21,18 or 13
- you carry a balanced translocation involving chromosomes 21,18 or 13
- your current pregnancy was conceived using a donor egg *(unless the egg for this pregnancy is from the same egg donor used in a previous pregnancy diagnosed with Down's, Edwards' or Patau's syndrome).*

**It may not be possible to offer NIPT if:**

- You have a cancer diagnosis in your current pregnancy
- You have had immunotherapy in your current pregnancy
- You have any other chromosomal condition

These cases require discussion with the NIPT laboratory to assess whether NIPT is possible.

## NIPT results

If you decide to have NIPT, the result will be reported as either ‘higher-chance’ or ‘lower-chance’ for each of the three conditions.

You can usually choose whether to receive NIPT results by phone or face-to-face. Discuss your preference with your healthcare professional. Most women will get their result within two weeks of having NIPT, however you should be contacted if there are any delays. It is important to remember that NIPT is still a screening test. In a small number of cases, it won’t be possible to give a result to you. The reasons why you might not get a result are explained under the section **‘no result’** (see below page 8).

NIPT is a highly sensitive screening test and will identify the presence or absence of the three conditions for most women. However, there can also be *false positive* and *false negative* results, therefore it is not a diagnostic test.

- A false positive result means getting a higher-chance result when the baby does not have the condition.
- A false negative result means getting a lower-chance result when the baby does have the condition.

## Lower-chance result

Receiving a lower chance NIPT result means it is very unlikely that your baby has either Down’s syndrome, Edwards’ syndrome or Patau’s syndrome

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

A higher-chance NIPT result will state which condition the baby has a higher chance of having. For example, it will state if the baby has a higher chance of Down’s syndrome or a higher chance of Edward’s syndrome or a higher chance of Patau’s syndrome.

Having a higher-chance NIPT result does not mean that the baby 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 have a baby with 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. Reasons for not getting a result might include:

- a technical issue with the test
- not enough DNA in the blood sample
- a twin pregnancy

If the first attempt at NIPT does not provide a result, you can then choose between:

- one further NIPT
- a diagnostic test (see the next section)
- no further testing.

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

## 3. Diagnostic Testing

Invasive diagnostic testing involves taking a sample from the placenta (chorionic villus sample - CVS) **or** amniotic fluid (amniocentesis).

The sample is taken using ultrasound to guide a needle **either** into the placenta **or** into the amniotic fluid. It is performed by specialists trained in fetal medicine.

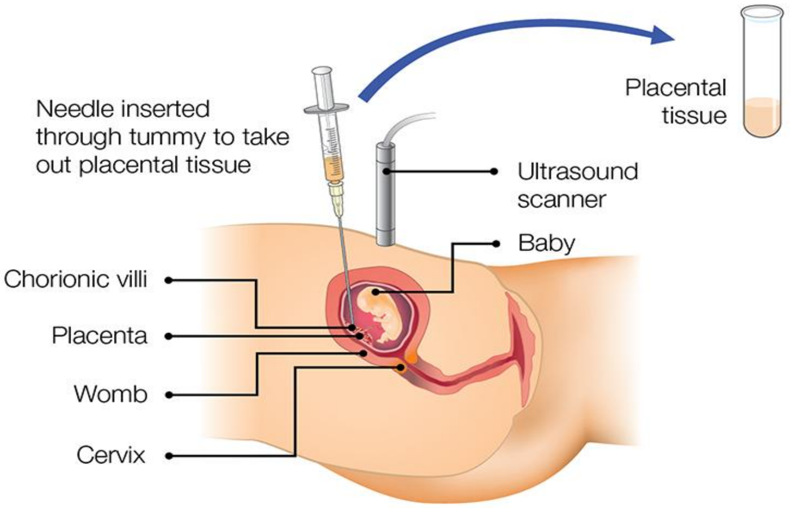

**Figure Two: Chorionic villus sampling (CVS).** This image shows one of the techniques (trans-abdominal) of taking a sample from the placenta.

**Figure Three: Amniocentesis**

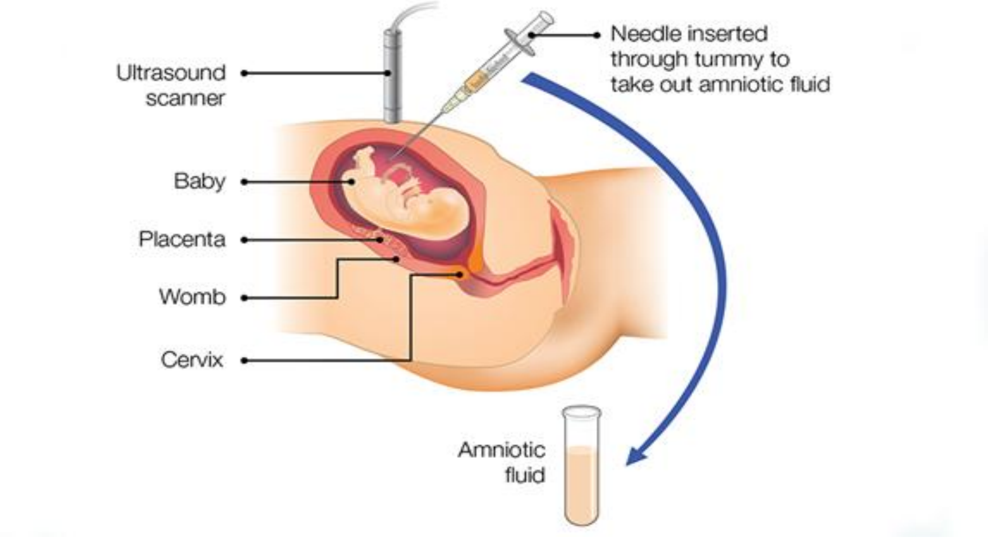

Figure two (chorionic villus sampling (CVS) and figure three (Amniocentesis) are taken from: [Screening in pregnancy: CVS and amniocentesis information for parents - GOV.UK (www.gov.uk)]()

The type of test (either CVS or amniocentesis) depends on how many weeks pregnant you are, any other ultrasound findings and where the placenta is in relation to your womb.

A CVS can usually be performed from around 11 weeks of pregnancy and amniocentesis from around 15 weeks of pregnancy. Your fetal medicine specialist will be able to advise further during your consultation. A sample of your blood is often needed to help provide a result for the pregnancy.

The chance of miscarriage from either of these tests is around 0.5% (1 in 200) and there is a risk of less than 1 in 1,000 that CVS or amniocentesis will cause a serious infection.

Your result from CVS or amniocentesis usually takes 2-3 days after your test. This will confirm if your baby does or does not have Down’s syndrome, Edwards’ syndrome or Patau’s syndrome. It may also tell you the sex of your baby. It will not test for any other conditions. Some results can be delayed due to issues with testing the sample. Rarely, we are unable to provide a result at all from that sample and your options would be discussed.

Your results will be given either by telephone or face-to-face, depending on what you have arranged with the team caring for you.

Storage of any DNA sample will be discussed with you at the time of the procedure.

Which, if any, testing to have in your pregnancy is a personal decision. Your health care team will support you whatever you decide.

Please be aware that this is general information and options outlined may not be suitable for all women with a previous child or pregnancy with Down’s syndrome, Edwards’ syndrome or Patau’s syndrome.

**Always discuss your options with an appropriate healthcare provider**

# Appendix 1: Support organisations

**Downs Syndrome Association:** https://www.downs-syndrome.org.uk/

**SOFT UK:** Trisomy 13 (Patau's Syndrome) and Trisomy 18 (Edwards' Syndrome)
https://www.soft.org.uk/

**Antenatal Results and Choices:** support around antenatal tests and results
https://www.arc-uk.org/

# Appendix 2: Further information for parents

Screening tests for you and your baby (STFYAYB) - GOV.UK (www.gov.uk)

Your choices after a higher-chance screening result - GOV.UK (www.gov.uk)

Screening in pregnancy: CVS and amniocentesis information for parents - GOV.UK (www.gov.uk)

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