Non-invasive prenatal testing: a new test for Down syndrome

Lucina NIPT at the West Midlands Regional Genetics Laboratory
This leaflet is designed to give you information about a new non-invasive prenatal test (NIPT) for Down syndrome. NIPT is much more accurate than the standard screening tests that are available. An invasive test (CVS or amniocentesis) is still necessary however to confirm diagnosis following a positive NIPT result. If you are considering using NIPT, please read this leaflet carefully. If anything is unclear please contact your antenatal screening midwife via the switchboard at your local maternity hospital.

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What is Down syndrome?

Down syndrome is a life-long condition that causes delays in learning and development, and can cause certain medical problems such as heart problems. It is a variable condition in that some people are more seriously affected than others. Some adults are able to get jobs and live fairly independent lives; however, most people do need long-term help and support. The life expectancy of a person with Down syndrome is 60-65 years, although many live longer.

Chromosomes are present in almost all human cells and store our genetic information. People usually have 46 chromosomes in each cell, but occasionally extra copies of chromosomes can be present, as is the case in Down syndrome.

Down syndrome is caused by an extra copy of chromosome 21. That is why it is sometimes called trisomy 21: trisomy means 3 copies of a chromosome. For every 1,000 babies born, one will have Down syndrome. Anyone can have a baby with Down syndrome, but we do know that the chances of having a baby with Down syndrome increases as women get older.

For more information on Down syndrome please contact the Down’s Syndrome Association. Their contact details are provided at the end of this leaflet.

What is NIPT for Down syndrome?

We now know that some of the baby's DNA circulates in the mother’s blood during pregnancy. DNA is the substance that our chromosomes are made of and contains our genetic information. By looking at the baby’s DNA found in the mother’s blood, we are able to identify whether or not the baby is affected by chromosomal conditions like Down syndrome. The baby’s DNA is lost from the mother’s blood stream within a few hours of the baby being born and so testing is specific to the baby in that pregnancy.
How is NIPT done?

The test is performed on a sample of the mother’s blood. About 10mls (roughly one tablespoon) is taken from the arm like a normal blood test. The blood is then sent to the laboratory for testing.

When is NIPT done?

There is only enough DNA present in the mother’s blood to conduct the test from 10 weeks of pregnancy, so the test cannot be done before then. You will need to have an ultrasound scan first to find out exactly how many weeks pregnant you are and whether there is more than one baby in the womb (such as twins) and to exclude fetal abnormalities. At present we are not offering NIPT to women with twin or triplet pregnancies.

How accurate is NIPT for Down syndrome?

NIPT for Down syndrome is more than 99% accurate. This means that the test detects more than 99 out of 100 cases of Down syndrome so there is a very small chance that the test will not detect an affected pregnancy. There is also a small chance that the test will incorrectly show that the baby has Down syndrome when it does not. Even though the test is highly accurate there is still the very small chance (around 1 in 300) of an incorrect result. Therefore, if the result of your NIPT predicts that the baby has Down syndrome you will be offered an invasive test to confirm the result. Invasive tests are discussed further below.
Does NIPT look for conditions other than Down syndrome?

Down syndrome is not the only condition that NIPT can look for, although it is the most common. NIPT can also look for two rarer chromosome conditions: Edwards syndrome and Patau syndrome. Edwards syndrome is caused by an extra copy of chromosome number 18 and Patau syndrome is caused by an extra copy of chromosome number 13. These conditions are very serious and many affected babies die before or soon after birth.

NIPT is more than 97% accurate for Edwards syndrome, and more than 87% accurate for Patau syndrome. Like Down syndrome, if the NIPT test predicts the baby is affected with any of these conditions an invasive test is recommended to confirm the result.

How long does it take to get the results from NIPT?

You will be contacted by a midwife with your results within 2 weeks of the lab receiving your sample. In a very small number of cases the laboratory may be unable to obtain your results. This might be because there was not enough of the baby’s DNA present in the blood sample to perform the test. If this happens you will be asked for a second blood sample so that the test can be repeated; there will not be any additional cost for this repeat testing.
What are the possible results?

- **Highly unlikely to be affected**
  Your baby is very unlikely to have Down syndrome, Edwards syndrome or Patau syndrome. You will then continue with your normal care in your pregnancy.

- **Predicted to be affected**
  It is very likely that the baby is affected with Down syndrome, Edwards syndrome or Patau syndrome. You will be offered an invasive test to confirm this result. This is offered because very occasionally, in around 1 in 300 cases, NIPT may not accurately reflect the result in the baby.

- **Inconclusive result**
  As this is a new test we occasionally get an inconclusive or ‘unclear’ result. If we cannot detect a clear result we will inform you and offer you another NIPT test.

- **Failed result**
  Occasionally we get a failed result, if this happens we will offer you a repeat NIPT test. There will be no additional cost to you for this additional test.

How safe is NIPT?

The test is a blood test taken from the arm like a normal blood test. Therefore, the test carries no significant risk to you or your baby. As is the case with all blood tests, there may be some bruising around the area where the blood sample was taken.

How does NIPT compare with other screening tests currently offered during pregnancy?

The traditional screening test offered during pregnancy, which consists of an ultrasound scan and a blood test (or in some cases only a blood test), is less accurate than NIPT as it only detects 84% – 90% of babies with Down syndrome.
What happens if the NIPT result predicts the baby has Down, Edwards or Patau syndrome?

If the NIPT result shows the baby has one of these chromosomal problems, you will be offered an invasive test. Invasive tests give a more definitive ‘yes’ or ‘no’ result as to whether the baby has Down syndrome. There are two types of invasive test available: amniocentesis is usually performed from 15 weeks of pregnancy, and chorionic villus sampling (CVS) is usually performed between 11 and 13 weeks.

Both procedures involve using a fine needle to collect a small sample of either the amniotic fluid that surrounds the baby (amniocentesis) or a small sample of cells from the placenta (CVS). These tests carry a small risk of miscarriage of 0.5% to 1% in the UK. This means that for every 200 women who have a CVS or amniocentesis, 1 or 2 of them will miscarry due to the test. Therefore, it is important to weigh up how important it is for you to know for certain whether the baby has a chromosome condition and how you feel about this compared to the risk of miscarriage. If these tests show the baby definitely has one of these conditions, you will have the chance to talk to your healthcare professionals about the options for your pregnancy.

What else should I consider before taking this test?

Before making a decision about NIPT, you may want to take some time to consider the test and discuss it with your partner. Think about how you might feel about the test results and how important the information would be for you and your family. It can help to consider whether the results would change the way you feel in your pregnancy or change anything about the management of your pregnancy. Some couples feel that they may not continue with the pregnancy if a chromosome disorder was found. Others would continue whatever the results but would like to know in advance in order to prepare for the arrival of their baby. If you are unsure about anything, it can be helpful to discuss it with your healthcare professional.
Where can I get further information and support?

To get more information you should contact your local maternity unit or named midwife. Other useful sources of information are listed below.

The Down’s Syndrome Association

A charity supporting people affected by Down syndrome.
Telephone: 0333 1212 300 Website: www.downs-syndrome.org.uk
Email: info@downs-syndrome.org.uk

NHS Choices

www.nhs.uk/conditions/Edwards-syndrome/Pages/Introduction.aspx

Antenatal Results and Choices

A charity offering non-directive support and information throughout the antenatal screening and testing process. Telephone: 0845 077 2290 or 020 7713 7486 from a mobile phone
Website: www.arc-uk.org Email: info@arc-uk.org
Hours: Monday – Friday, 10:00am – 5:30pm

UK National Screening Committee

www.gov.uk/topic/population-screening-programmes/fetal-anomaly

RAPID study website

www.rapid.nhs.uk

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