Non-invasive prenatal testing: a new test for
Down syndrome
Edwards syndrome
Patau syndrome
Fetal sex

(A private test not currently funded by the NHS)

In conjunction with:
This leaflet is designed to give you information about a new non-invasive prenatal test (NIPT) in pregnancy.

NIPT is more accurate than the standard antenatal screening tests for Down, Edwards and Patau syndrome that are currently available via the NHS.

If you are considering choosing NIPT please read this leaflet carefully. If you would like further information or would like an appointment, please contact the specialist screening midwife on 01895 279453 between 8am and 4pm Monday to Friday.

Please note all appointments must be pre-arranged.
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What are Down, Edwards and Patau syndromes?

Down syndrome is a life-long condition that causes delays in learning and development and can also cause medical conditions such as heart problems. It is a variable condition and 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.

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 increase as women get older. For more information on Down syndrome please contact the Down Syndrome Association. Their contact details are provided at the end of this leaflet.

Edwards and Patau syndrome are both life limiting conditions and are also caused by the presence of an extra chromosome. Edwards syndrome is caused by an extra copy of chromosome 18 and Patau syndrome by an extra copy of chromosome 13.

For more information on Edwards and Patau please contact www.soft.org.uk
What is NIPT?

We now know that some of a baby’s DNA circulates in the mother’s blood during pregnancy. DNA is the substance that our chromosomes are made of and chromosomes contain our individual genetic information. By looking at a baby’s DNA we are able to identify whether the baby is affected by a chromosomal condition such as Down syndrome. The baby’s DNA is lost from the mother’s blood stream within a few hours of birth 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 10ml (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 baby DNA present in the mother’s blood to conduct the test from 10 weeks of pregnancy onwards, so the test cannot be performed before this time. You will need to have had an ultrasound scan before testing is performed. This is to find out exactly how many weeks pregnant you are, to check whether there is more than one baby in the womb (eg twins) and to exclude any fetal abnormalities. At present we are NOT able to offer NIPT to women with a twin or triplet pregnancy.
How accurate is NIPT?

NIPT is 99% accurate in detecting Down, Edwards, and Patau syndrome. This means that the test detects 99 out of every 100 babies with one of these syndromes but there is a very small chance that 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 a chromosomal anomaly when it does not. Therefore, if the result of your NIPT predicts that the baby has a chromosomal anomaly you will be offered an invasive test to confirm the result. Invasive tests are discussed later in this leaflet.

It is also possible to predict the sex of a baby via NIPT. So you also have the option to choose an NIPT test that will predict the sex of your baby.

We offer a choice of tests:

**Prenatest** which tests for Down Syndrome only - £250

**PrenatalSafe*3** which tests for Down / Edwards / Patau syndrome - £360

**PrenatalSafe*5** which tests for Down / Edwards / Patau syndrome plus fetal sex - £380
How long does it take to get the results from NIPT?

Results will be available within seven business days and you will be informed of your result by the specialist midwife over the phone, with a confirmation letter to follow. In a very small number of cases the laboratory may be unable to provide a result. 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 is no additional cost for this repeat testing.

Please note that blood samples are sent to the laboratory by first class post in a secure biological transport box.

What are the possible results and how do they appear in the report?

- The report will state: that the baby shows evidence or no evidence of the syndromes we have tested for and if you have requested fetal sexing the report will confirm either a male or female result.

- Inconclusive or failed result: We occasionally get an inconclusive or failed result. If we cannot provide a clear result we will inform you and offer you another test. There will be no additional charge for this however the NIPT will only be repeated once.
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 with all blood tests, there may be some bruising around the area of the blood draw.

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 75-85% of babies with Down, Edwards or Patau syndrome and does not tell you the sex of the baby.

Why should I have NIPT?

There are two main circumstances where couples might opt for NIPT:

- As an alternative to the screening tests currently available on the NHS, because it is a more accurate test
- As a second screening test, where the ‘traditional’ screening test has given a high risk for trisomy in the pregnancy.

NIPT can give you a more accurate risk assessment on which to base decisions around further management.
What happens if the NIPT result predicts the baby has Down, Edwards or Patau syndrome?

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

Both procedures involve using a fine needle to collect either a small sample of cells from the placenta (CVS) or a small amount of the amniotic fluid that surrounds the baby (amniocentesis) These tests carry a small risk of miscarriage 0.5% to 1% in the UK. This means that for every 200 women who have a CVS or amniocentesis 2 will miscarry as a result of the test. Therefore, it is important to weigh up how important it is for you to know for certain whether your 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.

How much does the NIPT test cost?

The cost of NIPT at The Hillingdon Hospital ranges from £250-£380 depending on the chosen test. It is important that you keep your receipt in case a refund is indicated. This fee covers the costs of the test itself, information and counselling about the tests, alongside the cost of transporting the specimens, and of getting the result back to you.
Any money generated by offering this test privately will be used to improve the care of pregnant women at our hospital.

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 anomaly 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 this with your healthcare professional.

Where can I get further information and support?

For more information or to book an appointment for this test please contact the specialist midwives on 01895 279 453 between 8am and 4pm Monday to Friday. Other useful sources of information are listed overleaf.
**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

[WWW.soft.org.uk](www.soft.org.uk) supporting families affected by T13/T18

**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](www.rapid.nhs.uk)