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Information for patient

Non-Invasive Prenatal testing (NIPT)

What is an NIPT screening test?

Non-Invasive Prenatal testing (NIPT) is a blood test taken from the mother which uses cutting edge DNA technology to evaluate accurately whether a pregnancy has a high chance of having specific chromosomal conditions.

NIPT offers a personalized probability score, which indicates the chance of your baby being affected by Down's syndrome (trisomy 21), but also gives information for two rarer fetal chromosomal imbalances. Edward's syndrome (trisomy 18) and Patau's syndrome (trisomy 13).

NIPT is not a diagnostic test. For a definitive result an invasive test such as a CVS or amniocentesis is recommended

How does it work?

During pregnancy the placenta releases microscopic fragments of cell free fetal DNA (cfDNA) into the mother's bloodstream. As a result, the mother's blood contains a mixture of placental and maternal DNA. By looking at the placental DNA in the mother's blood, an NIPT test is able to predict whether or not the baby is more likely to have a chromosomal condition such as Down's syndrome.

What is Down's syndrome?

The vast majority of babies have a balanced number of chromosomes, inheriting 22 chromosomes from the mother and 22 from the father. There is also an extra pair of chromosomes known as sex chromosomes that determine the baby's gender, making 46 Chromosomes.

Babies born with Down syndrome have an extra copy of chromosome 21 (trisomy 21) making 47.

Down syndrome is a life-long condition typically associated with varying degrees of learning disability. Some health conditions are also more common in people with Down syndrome and while some children and adults will need long term support many adults can live fairly independent and productive lives.

What is the possibility of having a Down's syndrome?

All women, whatever their age, have a small chance of delivering a baby with an imbalance of chromosomes and whilst the chance increases for older mothers, over half of babies born with Down syndrome are born to mothers under the age of 35 years.

What are Edwards and Patau's syndrome?
Babies born with Edward's syndrome have an extra copy of chromosome 18 (trisomy 18) and babies born with Patau's syndrome have an extra copy of chromosome 13 (trisomy 13). Edwards' and Patau's syndromes affect about 1 in 7,000 births and the risk increases with maternal age. Babies born with Edwards Syndrome (trisomy 18) have an extra copy of chromosome 18 and babies born with Patau's syndrome trisomy 13 have an extra copy of chromosome 13.
Edwards' and Patau's syndrome are life limiting conditions and will causes a wide variety of developmental and health difficulties, some of which can be very serious. Around 70% of pregnancies affected by Edwards' or Patau's syndrome will end in miscarriage or stillbirth. Partial forms of Edwards' or Patau's syndrome have a lesser impact upon the child.

Sex aneuploidy, 22q and other microdeletion screening
The Nuffield Council on Bioethics has recommended that such use of NIPT technology must not be undertaken routinely. It may be justified in rare (selected) cases in this context, such as when it is suspected that a fetus has a significant medical condition or an impairment of unknown origin. We would be happy to discuss this further during your consultation. More information is available at nuffieldbioethics.org.

Who can have an NIPT screening test?
The tests can be performed in most pregnancies from 10 weeks. However, the test is not currently recommended for the routine detection of non trisomic conditions as the accuracy of this testing has not been verified.
The SAFE and TDL Test cannot currently be used for multiple pregnancies greater than twins, or if the mother has cancer, chromosomal or genetic conditions (including Down's syndrome). It is also unsuitable for mothers who have undergone a blood transfusion, transplant surgery, immunotherapy or stem cell therapy in the last 3 months.
The Panorama test is not suitable to be used by women carrying three or more babies (triplets and above), women who had used an egg donor or surrogate carrying more than one baby (twins and above), or those who have received a bone marrow transplant.

How do I take the test?
You are invited to take the time to ask all the questions you might have to enable you to make an informed personal decision as to whether you wish to take the NIPT screening test. Once you decide you want to take the test, you will be asked to sign a consent form by one of our doctors or an appropriately trained member of our staff. We will then draw a small amount of blood from a vein in your arm. This may cause some discomfort, but the sample is usually taken very quickly. Sometimes there can be some bruising after a blood sample is taken.
We will send the blood sample to the laboratory for analysis along with your personal information including your name, date of birth and gestation age.

Where is the analysis carried out and how long does it take?

SAFE test	St Georges Hospital in London	Results usually available within 7 days*
TDL test	TDL Genetics in London	Results usually available within 7 days*
Panorama test	California. USA	Results usually available within 10 days*

*Occasionally the laboratory may need to re-run your sample leading to delays in reporting. Such technical laboratory delays do not increase the likelihood of a concerning NIPT result.

How are the tests reported?

The way the result is reported is dependent on the test you have chosen. However, your result will indicate if your baby has a higher or lower chance of having a condition, or if the test has not been able to provide a result.

Low Chance Means that it is very unlikely that your baby is affected by either trisomy 21, 18 or 13 and therefore very unlikely that your baby has Down, Edwards or Patau's Syndrome.

High Chance Means that there is an increased chance that your baby will have either trisomy 21 or 18 or 13 Down, Edwards or Patau's Syndrome and that the result should be confirmed by an invasive diagnostic test.

No Call. A small number of tests will fail to give a result. This is usually due to insufficient levels of fetal cell free DNA being identified in the blood sample. If this happens you will be invited back for a repeat blood test.

What happens if I get a high chance result?

If you receive a high chance result it does not automatically mean that your baby definitely has trisomy 21,18 or 13. It is important to remember that a Non-Invasive Prenatal test is a screening test and not a diagnostic test.

With your permission we will discuss your result with your referring obstetrician, doctor or midwife who will talk to you about the option of having the result confirmed by a diagnostic test such as a CVS or amniocentesis.

Do I need to have any other tests?

The NIPT screening test does not provide information on other rarer chromosomal abnormalities. If your scan shows a high nuchal translucency measurement (greater that 3.5 mm) or major structural anomalies such as heart malformations the indication for other rarer chromosomal imbalances may be high. In such cases you may choose to have a CVS or amniocentesis.

NIPT Screening tests do not provide information on physical differences such as heart or brain malformations, spina bifida or fetal growth. It is therefore advisable that you still have ultrasound scans at 11-13 weeks (nuchal scan) and 20-22 weeks (fetal anomaly scan) to examine the fetal anatomy and at 36 weeks (fetal wellbeing scan) to examine your baby's growth.

Can an NIPT test tell me the sex of the baby?

Each of the tests can tell you the sex of your baby but there are some differences between the brands:

The SAFE test can tell you the sex of the baby for a single or identical twin pregnancy. If a 'vanished twin' has been identified or a non-identical twin pregnancy, sex determination cannot be performed.

The Panorama test can tell you the sex of the baby for a single and for each twin in multiple pregnancies.

The TDL test can tell the sex of your baby in single pregnancies. In twin pregnancies the TDL test may not be able to tell you the gender of both twins.