TDL GENETICS

Information for patients

Non-invasive prenatal testing

Genetic testing during pregnancy performed in the UK

Non-invasive prenatal testing (NIPT) can identify a pregnancy in which the baby is likely to have a chromosome condition. These conditions are uncommon, do not usually run in the family, and can happen in any pregnancy.

NIPT can be requested by your doctor from 10 weeks' gestation and we recommend that your doctor arrange for you to have an ultrasound to confirm your gestation, prior to taking your blood sample.

Collecting a mother's blood sample for NIPT poses no threat to the fetus.

Our NIPT routinely screens for these chromosome conditions:

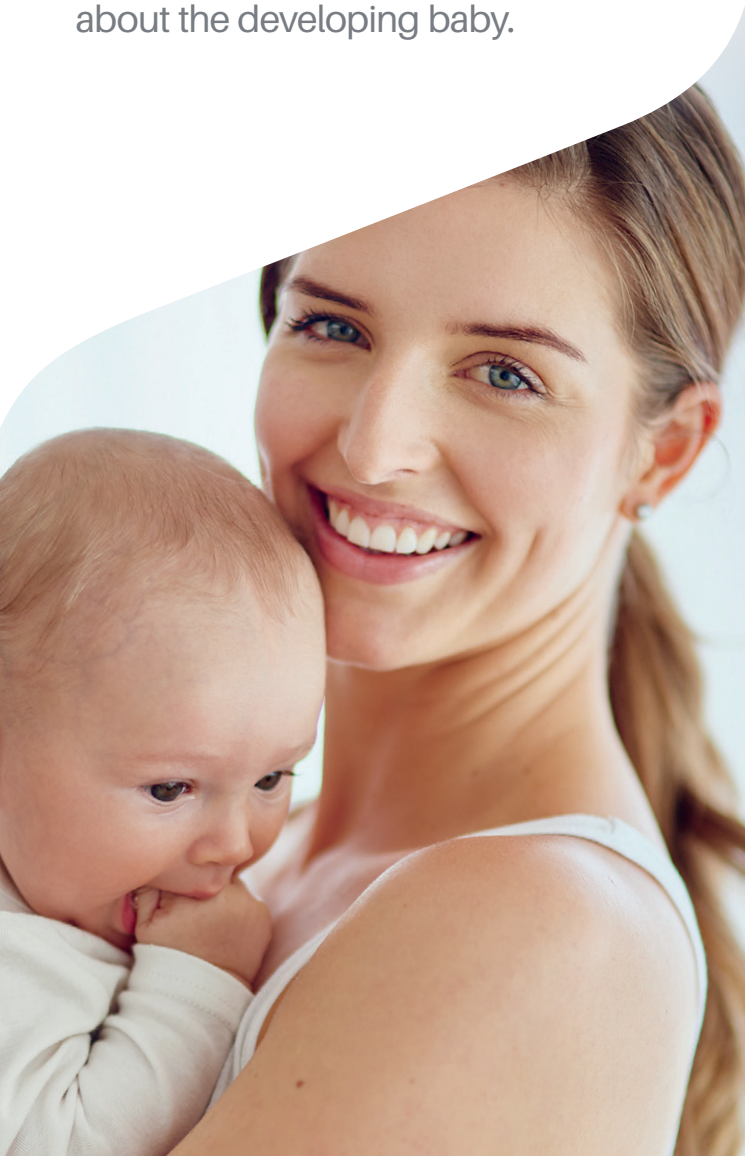
- Down syndrome (trisomy 21)
- Edwards syndrome (trisomy 18)
- Patau syndrome (trisomy 13)

If requested by your doctor, NIPT can also screen for:

- Turner syndrome
- Klinefelter syndrome
- Other sex chromosome anomalies (Triple X syndrome and XYY syndrome)

You also have the option to determine the sex of your baby.

A non-invasive prenatal test (NIPT) from the mother's blood that provides important information about the developing baby.



How accurate is NIPT?

Non-invasive prenatal testing (NIPT) is a screening test – it is a test for women who are unlikely to have a baby with a chromosome condition. It is much more accurate than first trimester blood screening and ultrasound tests. This has reduced the need for invasive tests such as chorionic villus sampling (CVS) or amniocentesis, although these will still be required to confirm a high probability result.

NIPT screening for trisomy 21, 18 and 13 – the most common chromosome conditions – provides the highest accuracy. Screening for other syndromes or for fetal sex is reasonably reliable; however, it is not as accurate as the screening for trisomy 21, 18 and 13.

Who should have NIPT?

Chromosomal conditions like Down syndrome (trisomy 21) do not typically run in families and can happen in any pregnancy. Although the chance of having a baby with Down syndrome increases with age, most babies with Down syndrome are born to women under 35.[2]

You must be at least 10 weeks pregnant for this test. Patients who have received bone marrow or organ transplants, or those who have metastatic cancer are not eligible for the test.

How does NIPT work?

There are small fragments of DNA in the mother's blood that have come from both the mother and the placenta. This is a normal process.

TDL Genetics uses an NIPT assay called Veriseq v2, manufactured by Illumina and processed in our London laboratory. The assay analyses the proportions of the DNA fragments that come from specific chromosomes.

If a particular proportion is too high or too low, this indicates that there may be a chromosome condition involving the placenta and, potentially, the developing baby.

What the NIPT report will include, and how you will receive your test results

Your sample is checked to see if there is sufficient DNA from the developing pregnancy to provide a reliable result. Then, for each condition included on the request form, the report will indicate whether there is a low or high probability of the condition being present.

Results will be sent to your healthcare provider in 2–4 business days.

Your healthcare provider will discuss the report with you and let you know if any other investigations are recommended. In your initial consultation, prior to the test being ordered, we strongly advise that you discuss what you would do with the possible results.

If your report indicates a high probability of a chromosome condition being present, a CVS or amniocentesis procedure can be arranged by your doctor to obtain a definitive result.

Does a low probability result mean that the baby does not have Down Syndrome?

No, any screening test carries a chance of a 'false negative' however the chance of this happening with NIPT is much lower than with conventional screening for Down syndrome and other chromosome conditions.

If I have a high probability result, does that mean that the baby's chromosomes are abnormal?

No, not necessarily. It means that there is a higher chance that your baby may have a chromosome condition, and you will be offered the option of amniocentesis to assess the chromosomes directly. You will be offered support and counselling to help you reach that decision.

What if I have twins?

This test can also be applied to twin pregnancies but is not suitable in the case of a vanishing or demised twin.

The sex of twins will be reported as one result. If male, one or both of the twins will be male. If female, both twins will be female.

Will I always get a result?

Two out of 100 women will require a repeat test. Patients will not be charged if we are unable to obtain a result.

On rare occasions, it is not possible to issue an NIPT result. This is usually due to the complex biology of pregnancy rather than a failure of the test method.

If NIPT cannot provide an assessment for the three most common chromosome conditions (trisomy 21, 18 and 13) after one collection of a blood sample (or two, if recommended by the laboratory), you will not be charged.

Does NIPT have any limitations?

The NIPT assay used by TDL Genetics – VeriSeq NIPT Solution v2 – is not validated for use in pregnancies with more than two fetuses, nor in cases of fetal demise, mosaicism, partial chromosome aneuploidy, triploidy, translocations, maternal aneuploidy, transplant or malignancy. VeriSeq NIPT Solution v2 does not detect neural tube defects. Certain rare biological conditions may also affect the accuracy of the test.

For twin pregnancies, HIGH PROBABILITY test results apply to at least one fetus; male test results apply to one or both fetuses; female test results apply to both fetuses.

Due to the limitations of the test, inaccurate results are possible.

A LOW PROBABILITY result does not guarantee that a fetus is unaffected by a chromosomal or genetic condition. Some fetuses without a trisomy or chromosomes anomaly may have HIGH PROBABILITY results. In cases of HIGH PROBABILITY results and/or other clinical indications of a chromosomal condition, confirmatory testing is necessary for diagnosis.

References

1 https://www.rcog.org.uk/en/news/rcog-response-to-uknsc-recommendation-on-nipt-testingfor-high-risk-women/ [Accessed 31.05.2018]

2 Morris JK, et al BMJ. 2009 Oct 26;339:b3794

3 Position statement from the International Society for Prenatal Diagnosis on the use of non-invasive prenatal testing for the detection of fetal chromosomal conditions in singleton pregnancies. Hui L, Prenatal Diagnosis – Wiley Online Library. https://obgyn.onlinelibrary.wiley.com/doi/10.1002/pd.6357

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Arranging a test

NIPT can be requested by your doctor from 10 weeks' gestation.

Your doctor will complete a Non-invasive prenatal test request form, and should discuss the conditions for which you may be tested, and whether you want to know your baby's sex.

Is this test available through the NHS?

Since 2021, NIPT has been available and funded through the NHS as a follow-up test for women who receive a higher chance result of >1:150 from a previous screening test (combined or quad test). TDL Genetics is one of three laboratories chosen to provide this testing in England.

For further information, please email
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