Kuon Healthcare

Non-Invasive Prenatal Test

Non-invasive prenatal screening test performed in a UK laboratory

The safest pre-natal screening for your peace of mind

NIPT helps in identifying if your baby is likely to have a chromosomal anomaly.

It is a safe and non-Invasive screening test that uses cell free DNA (cfDNA) found in maternal blood to detect prenatal chromosomal anomalies accurately, starting from WEEK 10 of pregnancy.

Who can take the test?

All pregnant women, irrespective of age or risk, who are 10 weeks or over into their pregnancy. (NIPT is endorsed by American College of Obstetricians and Gynaecologists (ACOG) /Society for Maternal-Fetal Medicine (SMFM) for all pregnant mothers regardless of age or risk).

Chromosomal conditions do not typically, passed down generations in families and can occur in any pregnancy. Therefore, it is advisable to go for this test after an ultrasound scan to confirm your gestation , after you have completed 10 weeks of pregnancy.

What does NIPT test for?

Basic NIPT routinely tests for the following chromosomal anomalies:

- Down Syndrome (Trisomy 21)
- Edwards Syndrome (Trisomy 18)
- Patau Syndrome (Trisomy 13)

Advanced NIPT is a more comprehensive version of the standard NIPT test. In addition to screening for the conditions covered by basic NIPT, it also includes an analysis for sex chromosome anomalies:

- X(O) -Turners Syndrome
- XXY - Klinefelter's syndrome
- XYY- Jacob's syndrome
- XXX- Triple X syndrome

Gender Detection

Apart from above, both basic and advanced NIPT also offer gender determination, identifying the fetus as male (XY) or female (XX).

Why should I take the test?

Fetal chromosome abnormalities, specifically aneuploidy, which is an abnormal number of chromosomes, are a common cause of reproduc tive failure, congenital anomalies, developmental delay, and intellec tual disabilities.

NIPT detects these abnormalities at a very early stage from week 10, with a high degree of accuracy and a low false-positive rate.

How does the test work?

The NIPT test involves a blood draw from the mother's arm, which contains both maternal and fetal DNA. It analyzes cell-free DNA (cfDNA) from the placenta, naturally found in the mother's bloodstream, to detect potential genetic conditions.

NIPT is powered by Illumina (VeriSeq NIPT Solution v2) and is tested in our laboratory based in Watford, U.K. VeriSeq NIPT Solution v2 utilises whole-genome sequencing of cfDNA fragments in the maternal blood sample to detect chromosomal aneuploidies.

How accurate is it?

NIPT has >99% detection rate for aneuploidies.

NIPT provides fewer false-positive and false-negative results than combined first trimester screening for trisomy 21, 18 and 13.

It is important to note that NIPT is a screening test does not provide a definitive genetic diagnosis, as NIPT cannot differentiate potential chromosome differences between the placenta and fetus. A definitive genetic diagnosis of the fetus requires invasive techniques such as chorionic villus sampling (CVS).

KUON NIPT

Accuracy (T21, T18, T13	Sensitivity*	False-Positive rate*
Combined first trimester screening	82%	1 in 26
NIPT	>99%	<1 in 1,000

* Proportion fetuses with trisomy correctly identified by the test as high probability of disorder.

* Proportion of normal fetuses incorrectly identified by the test as high probability of disorder.

NIPT Performance date in a general screening population

	Detection/ sensitivity	Specificity
Trisomy 21	>99.9% (95% CI:97.1%)	>99.90% (95% CI:99.63)
Trisomy 18	>99.9% (95% CI:91.4%)	>99.90% (99.64% c1:9?.1%J
Trisomy 13	>99.9% (95% CI:87.1%)	>99.90% (95% CI:99.64%)

The VeriSeq NIPT Solution v2 is a screening test and should not be considered in isolation from other clinical findings and test results. Conclusions about the fetal condition and pregnancy management decisions should not be based on the results of the NIPT screening alone.

Is NIPT valid for twin pregnancies?

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.

What does the report look like and how will I receive it?

The maternal blood sample is first checked to see if there is sufficient cfDNA to provide a reasonably accurate result. Then, for each condition (as chosen by the parent(s) on their consent form), the high or low risk of a chromosomal anomaly is detected and reported.

The final result is emailed to the email address provided on the consent form (which could belong to patient/ caregiver or their healthcare provider in up to 5 business days.

Please note:

NIPT is a screening test. Any positive/high-risk results need to be confirmed with a diagnostics test through your OBG/midwife/healthcare provider.

What does a low-risk result mean?

A low risk means - no anomaly detected for that specific condition which is under study. A LOW-RISK result does not guarantee that a fetus is unaffected by a chromosomal or genetic condition as False Negatives are possible, though the probability is quite low with VeriSeq NIPT Solution v2

What does a high-risk result mean?

A high risk means - An anomaly is detected for that specific condition which is under study. Some non-aneuploid fetuses may have HIGH PROBABILITY results i.e., false positives may occur, though the probability of this occurrence is very low. In the event of a HIGH-RISK result and/or other clinical indications of a chromosomal condition, confirmatory testing is necessary for diagnosis.

What happens if there is no result/invalid test result?

Sometimes, no conclusion can be drawn from the NIPT test, or the result is invalid. This happens due to the complex biology of a pregnancy and no deficiencies in the test method itself. In such cases you may choose to have a retest (at no extra cost) or get a refund.

What are the limitations of NIPT?

The VeriSeq NIPT Solution v2 is not suitable for patients with:

- Recent maternal blood transfusion
- Maternal mosaicism
- Maternal prior organ transplant/ stem cell transplant
- Maternal copy number variations
- Maternal autoimmune disease
- Fetoplacental mosaicism / confined placental mosaicism
- Maternal neoplasms (benign and malignant)
- Fetal demise/ vanishing twin
- Sex chromosome anomalies cannot be reported for twin pregnancies.

The VeriSeq NIPT Solution v2 is not validated for use in pregnancies with more than two fetuses, demised or vanishing fetus, mosaicism, partial chromosome aneuploidy, triploidy, translocations, maternal aneuploidy, transplant or malignancy. VeriSeq NIPT Solution v2 does not detect neural tube defects.

Twin pregnancy/imitations: 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.