

NON-INVASIVE PRENATAL GENETIC SCREEN

What is NIFTY™?

NIFTY™ is a highly accurate, Non-Invasive Prenatal Screening (NIPS) test for detecting fetal aneuploidies (chromosomal abnormalities). NIFTY™ uses a blood sample from the pregnant mother to analyze the fetal DNA for chromosomal conditions, such as Down Syndrome (Trisomy 21) and Edwards Syndrome (Trisomy 18), that could affect a baby's health.

Many prenatal screening options already exist. However, compared to NIFTY™, traditional screening methods have lower accuracy and higher false positive rates. Invasive diagnostic tests, such as amniocentesis or chorionic villus sampling (CVS) are accurate but carry a 1 - 2% risk of miscarriage. NIFTY™ is non-invasive with testing performed from a maternal blood sample, carrying no risk to the mother or fetus.

How does NIFTY™ Work?

Cell-free DNA fragments (cfDNA) are short fragments of DNA which can be found circulating in the blood. During pregnancy, cfDNA fragments from both the mother and fetus are present in the maternal blood. The NIFTY™ test requires taking a blood sample from the mother, which is then analysed to detect chromosomal abnormalities in the fetus.

Using sequencing technologies, the fetal and maternal DNA from each sample are analysed. The entire genome is sequenced and compared against optimal reference chromosomes to accurately determine the presence of genetic abnormalities.

Using this methodology, the NIFTY™ test is able to produce highly accurate results and screen for a broader range of abnormalities, including trisomy, sex chromosomal aneuploidy and deletion/duplication syndromes.

Read all the NIFTY™ test's published clinical data at www.niftytest.com/healthcare-providers/clinical-data/

Is NIFTY™ Suitable For Me?

Prior to undertaking any non-invasive prenatal testing, you should consult a qualified healthcare professional regarding any risks, diagnoses, treatment and/or any other potentially relevant healthcare issues. The NIFTY™ test is available from the 10th week of pregnancy and may be particularly suitable for pregnant women who exhibit certain indications, such as:

- Aged 35 years or older at delivery
- Ultrasound findings indicate an increased risk of aneuploidy
- History of a prior pregnancy with trisomy
- Received IVF treatment or have previously suffered from recurrent pregnancy loss
- Require reassurance following previous screening results



SAFE

Non-invasive with no risk of miscarriage.



SIMPLE

Test from a maternal blood sample as early as week 10 of pregnancy.



ACCURATE

Proven >99% sensitivity from a study of nearly 147,000 pregnancies.



TRUSTED

Over 3 million tests carried out worldwide.

What Genetic Conditions are Screened for by NIFTY™?

Trisomies

- These conditions are caused by the presence of an extra copy or partial copy of a chromosome, instead of the usual pair. This additional genetic material can cause dysmorphic features, congenital malformation and different degrees of intellectual disability.

Sex Chromosomal Aneuploidies

- Sex chromosome aneuploidy is a numeric abnormality of an X or Y chromosome, with the addition or loss of an entire X or Y chromosome. Although most cases of sex chromosome aneuploidies are generally mild without intellectual disability, some have a well-established phenotype that can include physical abnormalities, learning delays and infertility.

Deletion and Duplication Syndromes

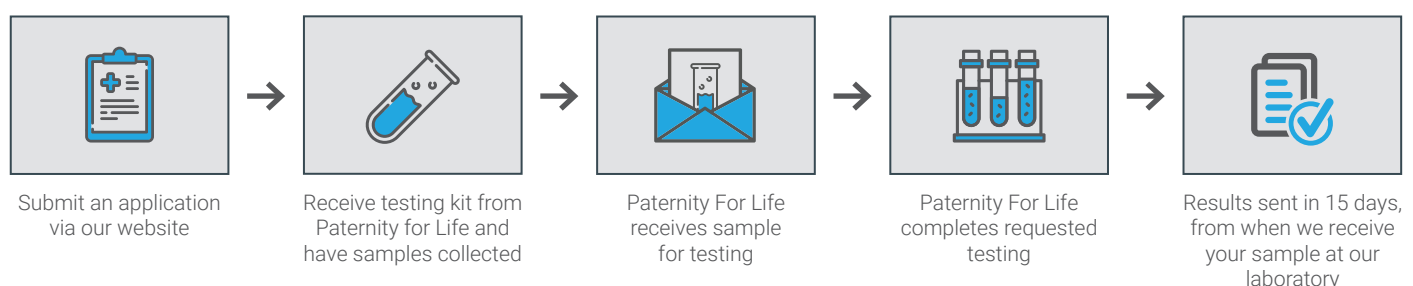
- Deletion and duplication syndromes are disorders characterised by small deletions or duplications of a chromosomal segment. The size and position of the deletion or duplication determine which clinical features are manifested and how severe they are. Clinical features may include developmental delays and intellectual disability, growth differences, behavioural problems, feeding difficulties, low muscle tone, seizures, dysmorphic features and a pattern of varying malformations.

Results

Results will be sent to your referring doctor within 10 -15 working days, from receipt of your sample at our laboratory. Results may sometimes be delayed due to incorrect specimen handling or laboratory processes requiring repeat testing to ensure accurate results. **Delayed results are not indicative of abnormal results.**

Please note that NIFTY™ is a screening test, and not a diagnostic test. A screening test indicates the risk of having a condition, and while they may be highly accurate, do not test with 100% accuracy. NIFTY™ is a screening test, so while it has a very high level of accuracy at 99.5% for detection of Down Syndrome (T21), Edwards Syndrome (T18) and Patau Syndrome (T13), it is not a diagnostic test and it does not screen for all chromosomal conditions. If the test result returns as high risk, further confirmatory diagnostic testing should be performed for final diagnosis by a qualified healthcare professional.

How to Organise Testing



NIFTY™ Plus
Gender Identification
• Male / Female
Trisomies
• Trisomy 21 (Down syndrome) • Trisomy 18 (Edwards syndrome) • Trisomy 13 (Patau syndrome) • Trisomy 9 • Trisomy 16 • Trisomy 22
Sex Chromosome Aneuploidies
• Turner Syndrome (Monosomy X) • XXX (Triple-X) • XXY (Klinefelter syndrome) • XYY Karyotype
Deletion Syndromes
• Cri-du-Chat Syndrome (5p) • 1p36 Deletion Syndrome • 2q33.1 Deletion Syndrome • Prader-Willi/ Angelman Syndrome (15q11.2) • DiGeorge Syndrome II (10p14-p13) • Jacobsen Syndrome (11q23) • 16p12 • Van der Woude Syndrome (1q32.2)