How does NIFTY® work?

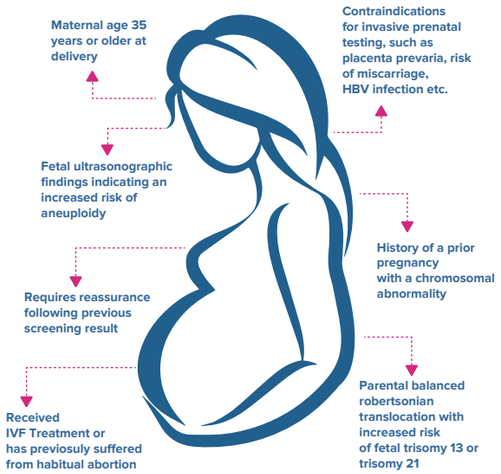
The NIFTY® test requires taking a small maternal blood sample of 10ml. cDNA in the maternal blood is then analysed to detect for chromosomal abnormalities. If an aneuploidy is present, small excesses or deficits in counts of the selected chromosome will be detected.

NIFTY® effectively resolves the difficulty in measuring the small increments in the specific chromosome DNA concentration through use of massively parallel sequencing technology (MPS). This means NIFTY® sequences millions of fragments of both fetal and maternal DNA from each sample. Using whole genome sequencing technology and four different proprietary bioinformatics analysis pipelines, the NIFTY® test is able to analyse data across the entire genome and compare chromosomes in the tested sample against optimal reference chromosomes to accurately determine the presence of a genetic abnormality.

As opposed to the 'targeted sequencing' methods employed by some other NIPS tests, the NIFTY® methodology allows for highly accurate results irrespective of the clinical symptoms of the patient, and a broader range of testing options including testing for trisomy, sex chromosomal aneuploidy and deletion syndromes.

Indications

Prior to undergoing the NIFTY® test, a pregnant women should receive comprehensive information regarding non-invasive screening and non-directive advice on human genetics. The NIFTY® test is available from the 10th week of pregnancy.



Clinical Validation

Large scale validation of the NIFTY® test

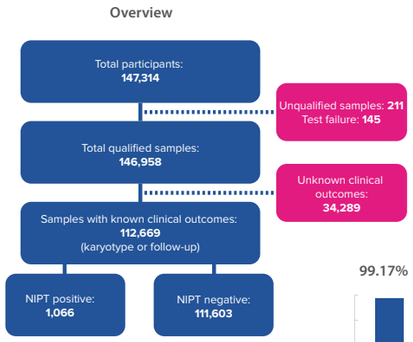
The NIFTY® test has been validated by the world's largest study on the clinical performance of NIPS to date.

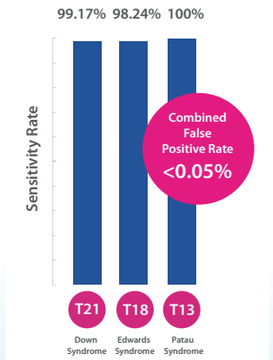
Non-Invasive Prenatal Testing For Trisomy 21, 18 and 13 – Clinical Experience from 146,958 Pregnancies.

Overall Sample Total with Known Pregnancy Outcomes 112,669					
Trisomy	TP	Sensitivity	Specificity	PPV	NPV
T21	720	99.17%	99.95%	92.19%	99.99%
T18	167	98.24%	99.95%	76.61%	100%
T13	22	100%	96.96%	32.84%	100%
TOTAL	909	99.02%	99.86%	85.27%	99.99%

Samples were collected between Jan 2011 and Aug 2013. The study was published in the Journal of Ultrasound in Obstetrics and Gynecology.

Wei Wang et al, Journal of Ultrasound in Obstetrics and Gynecology





SAFE: Non-invasive with no risk of miscarriage

SIMPLE: Test from a small 10ml maternal blood sample as early as week 10 of pregnancy

ACCURATE: Proven >99% sensitivity based on a study of nearly 147,000 pregnancies

TRUSTED: Over 1,000,000 tests carried out to date

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

Prenatal Peace
www.prenatalpeace.com

Based in Brisbane, within the Royal Brisbane Hospital Precinct, our partner laboratory utilises next generation sequencing to perform whole genome sequencing for Non-invasive Prenatal Screening (NIPS). Combining this state-of the art technology with the world's leading database from BGI Health, ensures the highest accuracy for the NIFTY® test.

A collection service is available in our Brisbane Clinic and through a national network of collection facilities. Contact us for collection sites or visit our website.

We are able to offer referral to a Genetic Counsellor if required.

Please call our Toll Free number 1300 482 165 to make an appointment for blood collection.

Toll Free: 1300 482 165 **Fax:** 02 9475 4330
Email: info@gtldna.com.au
Web: www.gtldna.com.au

Prenatal Peace
www.prenatalpeace.com

Non-Invasive Prenatal Screening (NIPS)

Over 1,000,000 tests performed worldwide.

Validated by a study of nearly 147,000 pregnancies.

targeting your *health* together

Non-Invasive Prenatal Screening (NIPS)

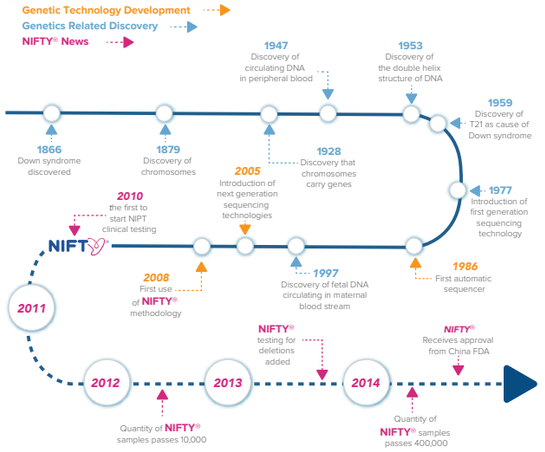
Introduction to NIFTY®

During the last decade, developments in the science of genetics and enormous advances in genetic technologies have altered our capability to understand diseases, make diagnoses and provide effective treatments. Transforming the world of prenatal testing, the advent of new DNA-based non-invasive prenatal screening (NIPS) has introduced a highly accurate screening strategy for fetal aneuploidy.

The NIFTY® test (Non-Invasive Fetal Trisomy test) was the first NIPS to enter clinical testing in 2010 and was launched in Europe in the first quarter of 2013. Providing screening for the most common trisomies present at birth, as well as testing options for gender, sex chromosomal aneuploidies and chromosomal deletions, NIFTY® provides a significantly stronger risk indication than traditional screening procedures.

As of 2016, over 1,00,000 NIFTY® tests have been performed worldwide. The NIFTY® test used technology and bioinformatics developed by BGI Diagnostics

Timeline

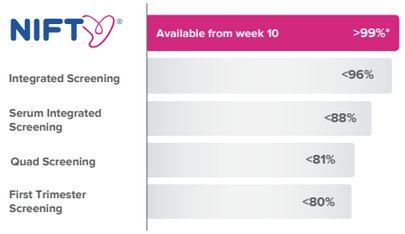


Why Non-Invasive Prenatal Screening?

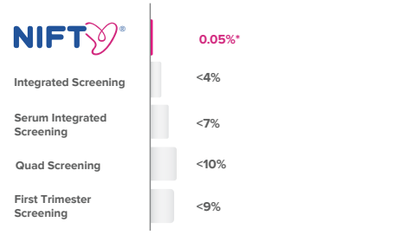
Many prenatal screening options already exist. However, compared to non-invasive prenatal screening (NIPS), traditional screening methods suffer from lower accuracy and higher false positive rates. Invasive diagnostic tests such as amniocentesis or chorionic villus sampling (CVS) are accurate but carry a 0.5-1% risk of miscarriage.

How does NIFTY® compare to traditional screening methods?

A Comparison of Detection Rates



A Comparison of False Positive Rates (FPR)



*Non-Invasive Prenatal Testing For Trisomy 21, 18 and 13 – Clinical Experience from 146,958 Pregnancies, Wei Wang et al, Journal of Ultrasound in Obstetrics and Gynecology

Introduction to Genetic Conditions Tested by NIFTY®

Trisomies

A trisomy is a type of aneuploidy in which there are three chromosomes instead of the usual pair. Trisomy 21 (Down syndrome), Trisomy 18 (Edwards syndrome) and Trisomy 13 (Patau syndrome) are the three most commonly occurring autosomal chromosome aneuploidies in live births. These chromosomal conditions are caused by the presence of an extra copy or partial copy of chromosome 21, 18 or 13 respectively. This additional genetic material can cause dysmorphic features, congenital malformation and different degrees of intellectual disability. We also include testing for Trisomy 9, 16 and 22 as part of the NIFTY Plus test.

Deletion Syndromes

Deletion syndromes are defined as a group of clinically recognisable disorders characterised by a small deletion of a chromosomal segment. The size and position of the deletion determine which clinical features are manifested and how severe they are.

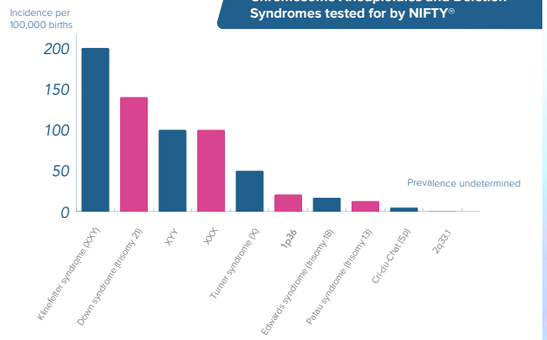
Clinical features of deletions can include developmental delays and intellectual disability, growth differences, behavioural problems, feeding difficulties, low muscle tone, seizures, dysmorphic features and a pattern of varying malformations. The following syndromes are included in our testing; Cri-du-chat Syndrome, 1p36, 2q33.1, Prader-Willi/Angelman Syndrome (15q11.2), Jacobsen Syndrome (11q23), DiGeorge Syndrome II (10p14-p13), 16p12, Van der Woude Syndrome (1q32.2) as part of the NIFTY® Plus test.

Sex Chromosomal Aneuploidies

Sex chromosome aneuploidy is defined as 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. Testing includes; Turner Syndrome, Klinefelter Syndrome, XXX, XYY.

Testing services for trisomy conditions 21, 18 and 13 are available for twin pregnancies, egg donor pregnancies and IVF pregnancies.

Incidence of Trisomy Conditions, Sex Chromosome Aneuploidies and Deletion Syndromes tested for by NIFTY®



"ISPD® recognises that NIPS can be useful as a screening test for women who are at high risk of Trisomy 21, with suitable genetic counselling. A positive test should be confirmed through invasive testing." ***Source:*** *ISPD (International Society of Prenatal Diagnosis)*

"The NSGC® supports NIPS as an option for patients whose pregnancies are considered to be at an increased risk of certain chromosome abnormalities. Patients whose NIPS results are abnormal, or who have other factors suggestive of a chromosome abnormality, should receive genetic counselling and be given the option of standard confirmatory diagnostic testing." ***Source:*** *NSCG (National Society of Genetic Counselors)*

Test Options

Trisomies

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

Sex Chromosome Aneuploidies

- Monosomy X (Turner syndrome)
- XXY (Klinefelter syndrome)
- XXX (Triple-X)
- XYY Karyotype

Additional Trisomies and Deletion Syndromes

- Trisomy 9, Trisomy 16, Trisomy 22, 5p, 1p36, 2q33.1, Di George Syndrome 2 (10p14-p13) 16p12.2 Jacobsen, Van der Woude Syndromes. 1 (1q32.2), Prader-Willi/Angelman

Gender Identification

- Male/Female

Test Information

- Twin pregnancies (Trisomy 21, 18 and 13 only)
- IVF Pregnancy
- Egg Donor Pregnancy
- Tested Samples: **>1,000,000**
- Turnaround time **10 working days**
- Available from **week 10** of pregnancy

Advantages

- Whole genome sequencing and high coverage improving accuracy. NIFTY® price competitive against all other NIPS providers.
- The option to perform an expanded NIPS with the largest range of testing for microdeletion syndromes and additional trisomies.
- Most validated NIPS on the market with a published study based on the outcomes of approximately 147,000 pregnancies and over 1 million NIFTY® tests performed worldwide to date.
- NIFTY® was the first NIPS test to be implemented into clinical use.

NIFTY® Methodology

Cell-Free DNA and Cell-Free Fetal DNA

Cell-free DNA fragments (cfDNA) are short fragments of DNA which can be found circulating in the blood. During pregnancy, cfDNA fragments originating from both the mother and fetus are present in maternal blood circulation. Cell-free fetal DNA (cDNA) is present only as a minority component of the total cfDNA in maternal plasma, which poses a significant technical challenge for some NIPS detection methods.