NIFTY®

Non-Invasive Prenatal Testing

Over 20 Million Tests Conducted

www.bgi.com/global/

BGI

Omics for All

Introduction to NIFTY®

For Reliable and Early Answers about Fetal Genetic Health Without the Miscarriage Risk of Invasive Procedures

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 testing (NIPT) has introduced a highly accurate screening strategy for fetal aneuploidies.

The NIFTY® test (Non-Invasive Fetal TrisomY test) was one of the first NIPTs to enter clinical testing in 2010 and was launched commercially in 2013. Providing screening for the most common trisomies present at birth, as well as testing options for sex chromosomal aneuploidies and certain chromosomal deletions/duplications, NIFTY® provides a significantly stronger risk indication than traditional screening procedures and serves as one of the most comprehensive NIPTs on the market.

To date, over 20 million NIFTY have been performed worldwide. The NIFTY® test is brought to you by BGI, one of the world's leading genomics organizations.

Validated by a Study of 146,958 Pregnancies

Noninvasive 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

Test Overview

Tested Samples

20+ million

Turnaround Time

<7 working days

Test From

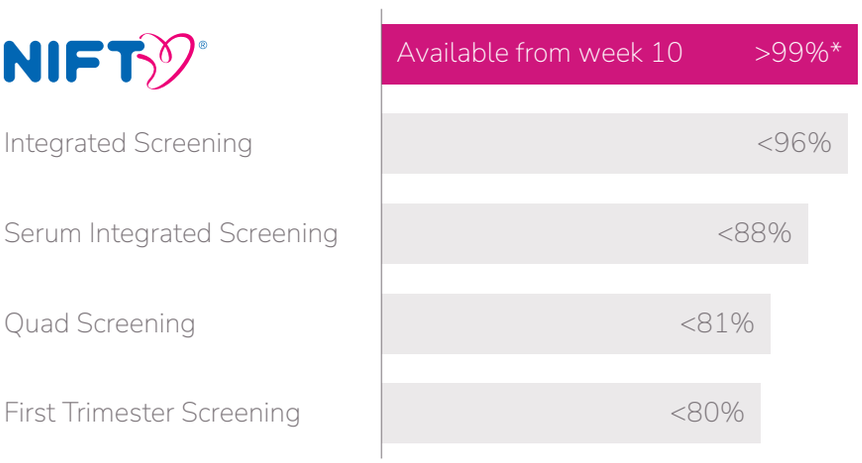
week 10 of pregnancy

- Twin Pregnancy
- IVF Pregnancy
- Egg Donor Pregnancy
- Reports Fetal Fraction
- Reports gender (optional)

Why Non-Invasive Prenatal Testing?

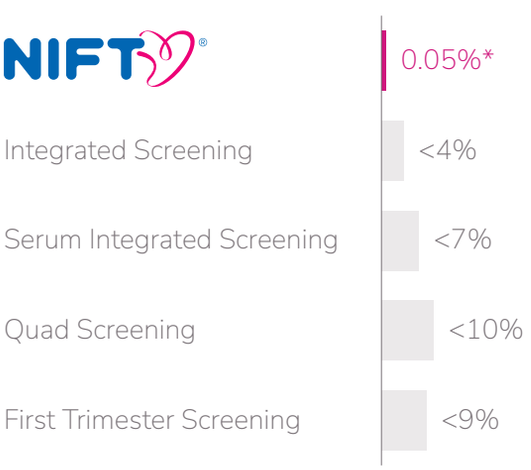
Many prenatal screening options already exist. However, compared to non-invasive prenatal testing (NIPT), traditional screening methods suffer from lower accuracy and higher false positive rates. Invasive diagnostic tests such as amniocentesis or chorionic villus sampling (CVS) are accurate but can carry a risk of miscarriage. NIFTY® poses no risk to the mother or fetus.

Comparison of Detection Rates Between NIFTY® and Traditional Screening Tests*



*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

Comparison of False Positive Rates (FP*R) Between NIFTY® and Traditional Screening Tests*



*Accuracy figures quoted from various publicly available data sources.

NIFTY® Test Options

The NIFTY® test is one of the most comprehensive NIPTs on the market screening across all 23 pairs of chromosomes, and offers a flexible test panel with a variety of different testing options to suit partner or patient need.

Genetic Conditions

Trisomies

- Trisomy 21 (Down syndrome)
- Trisomy 18 (Edwards syndrome)
- Trisomy 13 (Patau syndrome)
- Trisomy 9
- Trisomy 16
- Trisomy 22

Sex Chromosome Aneuploidies

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

92 Deletion/Duplication Syndromes, including

- 5p (Cri-du-Chat syndrome)
- 1p36
- 2q33.1
- Prader-Willi/ Angelman Syndrome (15q11.2)
- Jacobsen Syndrome (11q23)
- DiGeorge Syndrome (22q11.2)
- 16p12

Why Choose NIFTY®?

The Superior Accuracy and Lower False-Positive rate of NIPT Compared to Traditional Screening Tests may Minimize Anxiety and Invasive Procedures Caused by False Positive Results*

Safe

Non-invasive with no risk of miscarriage

Simple

Test from a small >5ml maternal blood sample as early as week 10 of pregnancy

Accurate

Proven >99% sensitivity for T21, 18 & 13, based on a study of nearly 147,000 pregnancies

Trusted

Over 20 million NIFTY® tests carried out to date by clinicians in more than 80 countries

Comprehensive

Detection of 23 pairs of chromosomes

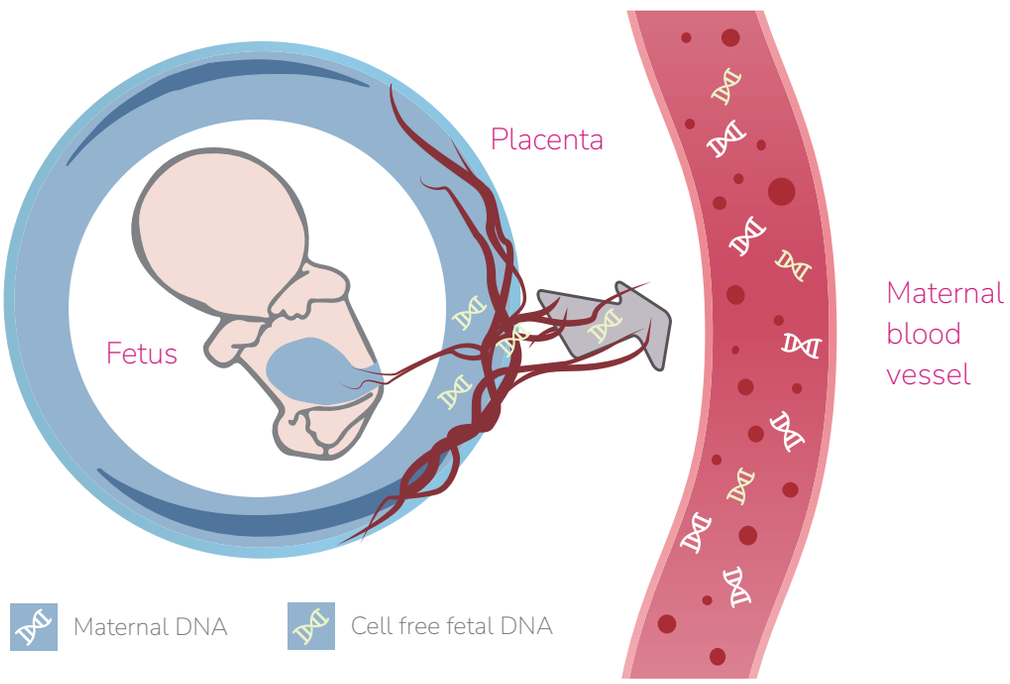
Visit us at www.bgi.com/global/
For enquiries, please email info@bgi.com

*Wax et al. J Clin Ultrasound. 2015 Jan; 43(1): 1-6

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. Fetal cell-free DNA (fetal cfDNA) is present only as a minor component of the total cfDNA in maternal plasma, which poses a significant technical challenge for some NIPT detection methods.



How Does NIFTY® Work?

The NIFTY® test requires taking a small maternal blood sample at least 5ml. Fetal cfDNA in the maternal blood is then analysed to detect chromosomal abnormality. If aneuploidy is present, small excesses or deficits in counts of the affected 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 genetic abnormality. As part of the NIFTY® test's quality control procedures, the fetal cfDNA % is listed on every test report.

As opposed to the 'targeted sequencing' methods employed by some other NIPT 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 for trisomy, sex chromosomal aneuploidy and deletion and duplication syndromes.

Introduction to Genetic Conditions Tested by NIFTY®

Trisomies

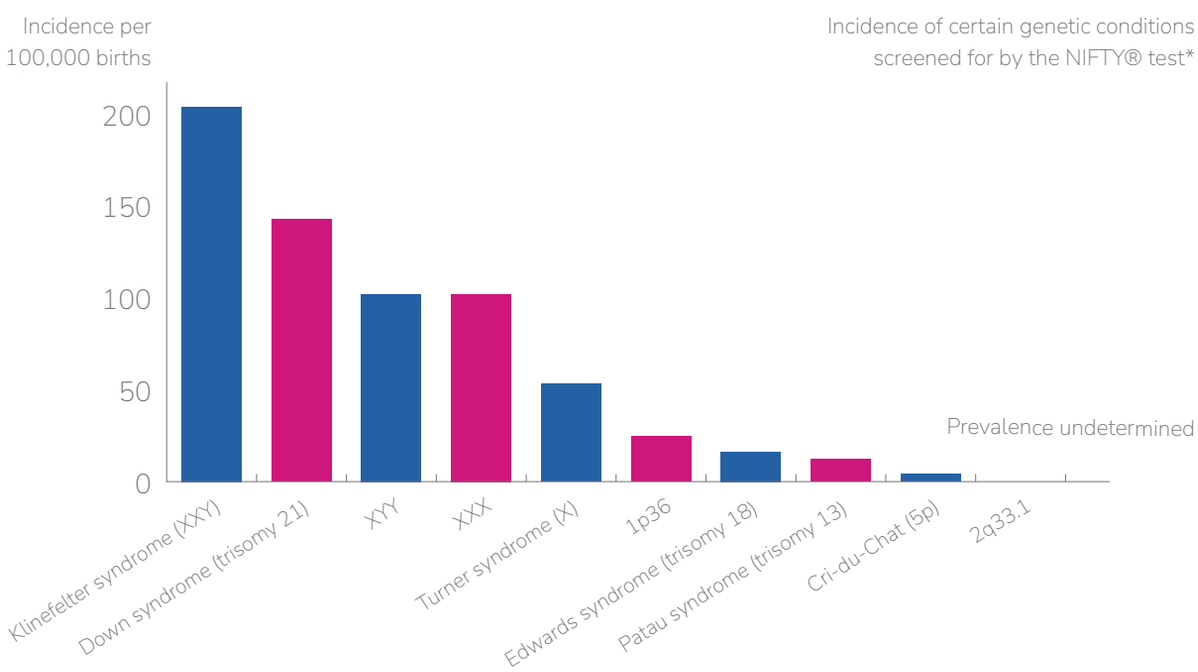
A trisomy is a type of aneuploidy in which there are three chromosomes instead of the usual pair. Trisomy 21 (Down syndrome), Trisomy 18 (Edwards syndrome) and Trisomy 13 (Patau syndrome) are the three most commonly occurring autosomal chromosome aneuploidies in live births. These chromosomal conditions 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.

Deletion/duplication Syndromes

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

Sex Chromosomal Aneuploidies

Sex chromosome aneuploidy is defined as a numeric abnormality of an X or Y chromosome, with 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.



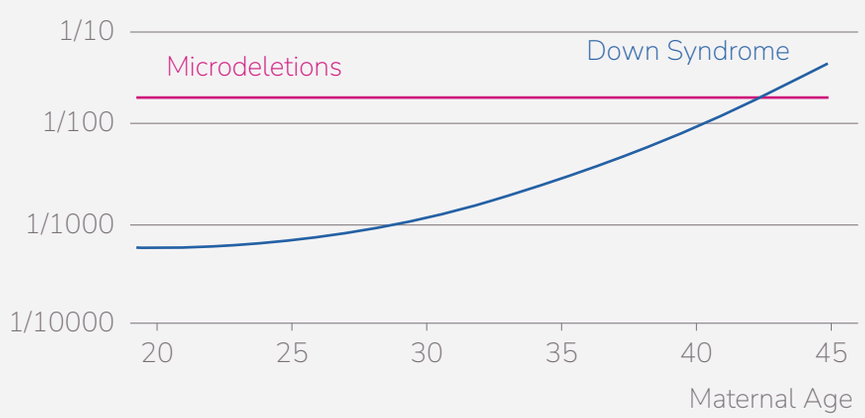
*Data taken from multiple public sources.

Microdeletion/Duplication Syndromes

Screen for Different Microdeletion/Duplication Syndromes

Besides common chromosomal aneuploidies, chromosomal microdeletion/duplication (also called chromosomal copy number variations or CNVs) can also cause serious birth defects and health problems. The prevalence of these conditions ranges from 1/4000 to 1/200,000, with fragment sizes from 100K to over 10M.

The incidence of microdeletions can be more common than Down Syndrome for pregnant women aged under 40



Snijders,et al.ultrasound obstet gynecol 1999

ACMG
American College of Medical Genetics and Genomics

ACMG recommends that healthcare providers inform all pregnant women about the availability of NIPT screening for clinically relevant microdeletions, when counseling about the risks, **benefits and limitations can be provided***

American College of Medical Genetics and Genomics (ACMG), Position Statement, Jul 2016.

Sample Requirements

Sample Type	Quantity	Requirements	Shipment
Maternal Blood	at least 5ml	Gently invert the tube ten times immediately after blood sampling.	Stored and shipped between 6~35 °C within 4 days. Keep the tubes upright during shipping.

The Test Workflow

1

Conduct pre-test genetic counseling and ensure patient provides informed consent for test

2

Discuss and fill in the NIFTY® Consent Form/Test Request Form with the patient

3

Conduct blood draw

4

Send scanned copies of Consent Form/Test Request Form and information sheet to BGI

5

Arrange collection of the blood sample with a courier

6

Send Consent Form/Test Request Form with blood sample to BGI laboratory

7

Receive results back in 10 working days

8

Conduct pre- and post-test genetic counseling and provide drug guidance advice as required

If you wish to request a genetic counseling, please contact us: P_International_GC @genomics.cn

Indications

To undergo the NIFTY® test, a pregnant woman should receive comprehensive information regarding non-invasive prenatal testing and non-directive advice on human genetics from a qualified health professional. The NIFTY® test is available from the 10th week of pregnancy.

The NIFTY® test is suitable for, but not limited to, patients who exhibit any of the following indications

- All age of Pregnancies for screening
- Contraindications for invasive prenatal testing, such as placenta prevaria, risk of miscarriage, HBV infection etc.
- History of a prior pregnancy with a chromosomal abnormality
- Fetal ultrasonographic findings indicating an increased risk of T21, T18 or T13
- Requires reassurance following previous screening result
- Received IVF Treatment or has previosuly suffered from habitual abortion

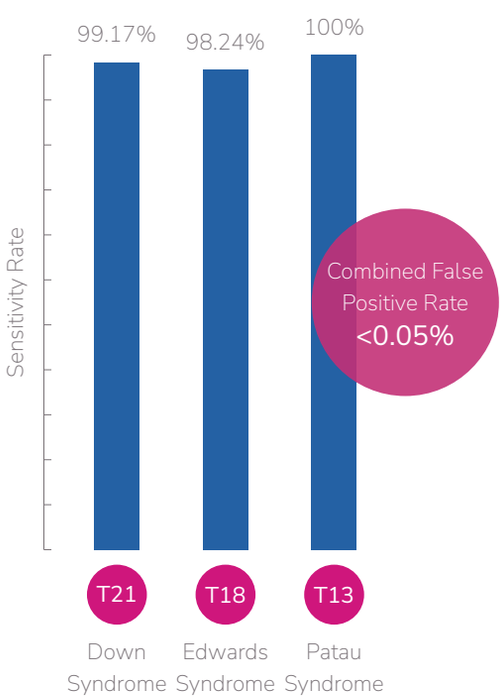
The NIFTY® test is not suitable for patients with the following indications

- Maternal, fetal and/or placental mosaicism (mixtures of chromosomally normal and abnormal cells in the pregnancy)
- Balanced or unbalanced translocation and chromosomal inversion
- Patients who have received a blood transfusion within one year prior to testing date
- Patients who have had transplant surgery
- Patients who have had stem cell therapy
- Vanishing twin syndrome (with developmental arrest identified has having occurred after week 8 of pregnancy and/or within 8 weeks of NIFTY® testing)

Clinical Validation

NIFTY® has been Validated by one of the World's Largest Studies on the Clinical Performance of NIPT and over 100 other Published Papers

With the NIFTY® Test's Stringent Protocols, the High Performance of NIPT Demonstrated by Early Validation Studies can be Maintained in High Volume Clinical Services



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%	99.96%	32.84%	100%
TOTAL	909	99.02%	99.86%	85.27%	99.99%

Data Source: Non-invasive prenatal testing for trisomies 21, 18 and 13: clinical experience from 146,958 pregnancies.Ultrasound Obstet Gynecol. 2015 May;45(5):530-8.doi: 10.1002/uog.14792

ISPD recognizes that cfDNA screening as a primary test to all pregnant women.

Source:Prenatal diagnosis, 35(8), 725–734.

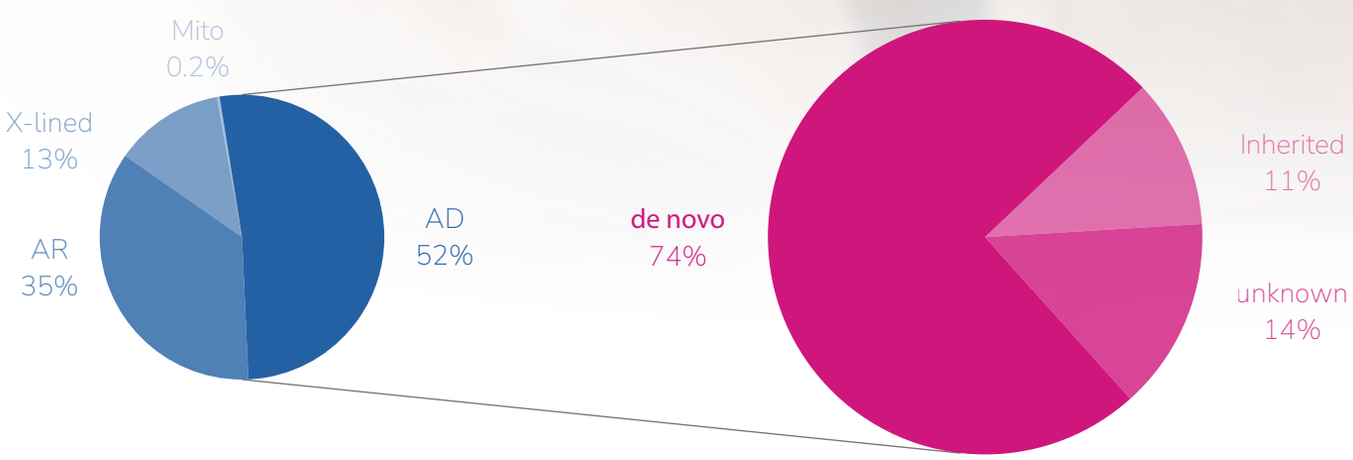
ACMG suggested that NIPS can replace conventional screening for Patau, Edwards, and Down syndromes.

Source:Genet Med. 2016; 18(10): 1056-1065.

Introduction to NIFTY® mono

NIFTY® mono is a non-invasive prenatal test for single-gene conditions. It screens for various clinically significant and life-altering genetic conditions in a fetus that are undetectable by current NIPT technology, allowing for a more complete picture of the associated risks when a pregnancy is being adversely affected by a genetic condition.

Previous studies have demonstrated that



Although the occurrence of dominant single-gene conditions is relatively rare, its cumulative rate is more than 1/1500 [1].

Single-gene conditions are responsible for a heavy loss of life, among which dominant inheritance accounts for more than 50% of all single-gene conditions [2].

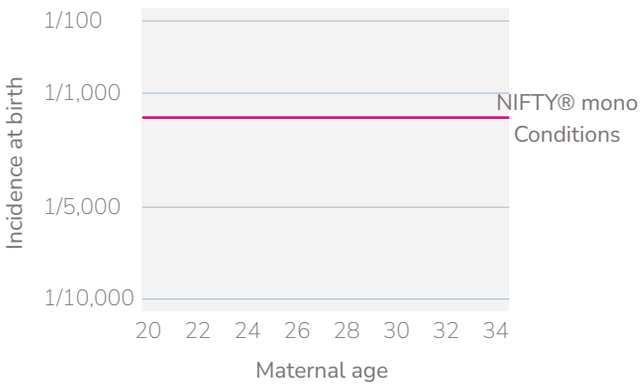
74% of dominant single-gene conditions are caused by *de novo* mutations (a gene mutation that is not inherited) [2]. Family history is typically not a good indicator of probability.

Some conditions in NIFTY® mono are not typically associated with abnormal prenatal ultrasound findings (especially in the first trimester), or may not be evident until the late second/third trimester or after delivery. Therefore, this test is the next leap in the evolution of screening for genetic disorders during pregnancy, providing valuable information for medical decisions, preparation, and peace of mind for families and physicians.

References:
[1] https://www.ncbi.nlm.nih.gov/books/NBK1116/.
[2] Yang, Y. et al. Molecular Findings Among Patients Referred for Clinical Whole-Exome Sequencing[J]. JAMA, 2014, 312(18): 1870-1879.

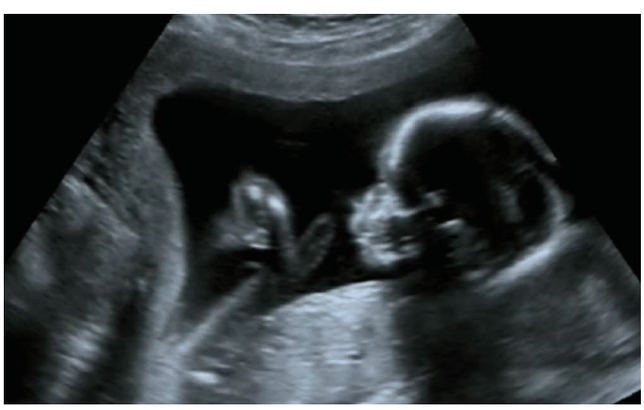
Why NIFTY® mono

NIFTY® mono identifies fetal conditions that may not otherwise be detected by traditional prenatal screening.



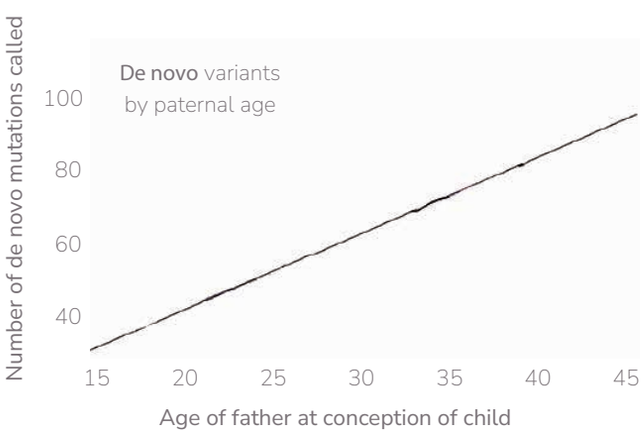
Screens for conditions common across all maternal ages

All pregnant women-regardless of age–are at equal risk of the genetic conditions screened by this test. Furthermore, family history is typically not a good indicator of probability.



Identifies conditions that may have otherwise gone undetected prenatally

These conditions are not typically associated with abnormal prenatal ultrasound findings (especially in the first trimester), or may not be evident until the late second/third trimester, or cannot be detected with other molecular screening methods.



Screens for genetic disorders associated with advanced paternal age

NIFTY® mono screens for genetic conditions (e.g. Crouzon syndrome, Apert syndrome, Osteogenesis imperfecta, etc.) which are associated with advanced paternal age (men who are >40 years of age) [1], ensuring a comprehensive screen for couples of advanced age.

References:
[1] Kong, Augustine, Frigge, et al, Rate of de novo mutations and the importance of father's age to disease risk[J]. Nature. 2012, 488:471-475.

Why choose NIFTY® mono

Safe

High sensitivity and specificity, both greater than 99%

Simple

Only one tube of blood from the mother is required, which can be collected as early as 10 weeks of gestation

Reliable

It is a non-invasive test, no risk to the fetus or the mother

Fast

Turnaround time of 14 working days

Indication for Testing

NIFTY® mono offers a comprehensive screening for several skeletal, cardiac, and syndrome conditions. NIFTY® mono can provide important information for people who:

- Are of advanced paternal age [1-3]
- Have ultrasound anomalies, such as shortened long bones and increased NT, which is suggestive of monogenic disorders
- Would like to avoid an invasive diagnostic procedure
- Want to know “everything”
- Are at risk for the genetic conditions being screened for

References:
[1] Toriello H V, Meck J M. Statement on guidance for genetic counseling in advanced paternal age[J]. Genetics in Medicine, 2008, 10(6):457-460.
[2] Kong A, Frigge M L, Masson G, et al. Rate of de novo mutations, father’s age, and disease risk[J]. Nature, 2012, 488(7412):471-475.
[3] Neville MDC, Lawson ARJ, Sanghvi R, et al. Sperm sequencing reveals extensive positive selection in the male germline. Nature. 2025 Nov;647(8089):421-428.

Sample Requirements

Sample type

Maternal blood.

Quantity

10 ml.

Requirements

Gently invert the STRECK BCT tube ten times immediately after sampling.

Shipment

Stored and shipped between 6~35˚C within 4 days.

Service Workflow

14 working days after receipt of samples.

1 

Conduct pre-test genetic counseling and ensure patient provides informed consent for test

2 

Discuss and fill in the NIFTY® Consent Form/Test Request Form with the patient

3 

Conduct blood draw

4 

Send scanned copies of Consent Form/Test Request Form and information sheet to BGI

5 

Arrange collection of the blood sample with a courier

6 

Send Consent Form/Test Request Form with blood sample to BGI laboratory

7 

Receive results back in 14 working days

8 

Conduct pre- and post-test genetic counseling and provide drug guidance advice as required

Becoming a NIFTY® provider is a quick and simple process.

What is Reported Through NIFTY® mono Results

The test results will show whether pathogenic or likely pathogenic single-gene variants have been identified.

Positive

One or more mutations in one or more genes were detected in this test. Genetic counseling and confirmatory diagnostic testing are recommended.

Negative

No disease-causing mutation in the targeted genes screened was detected in this test.

Clinical Validation

A total of 750 samples were tested in the clinical validation of non-invasive prenatal screening testing for 200+ dominant single-gene conditions. The sensitivity and specificity of the test were both more than 99%[1].

In clinical trials conducted in cooperation with Obstetrics and Gynecology Hospital of Fudan University, Among 750 high-risk pregnancies, non-invasive prenatal screening for 200+ dominant single-gene disorders identified 32 positive cases. Subsequent prenatal diagnosis confirmed one false positive and no false negatives, yielding a sensitivity of 100.0% and a specificity of 99.9% . The positive predictive value and negative predictive value were 96.9% and 100.0%, respectively.

References:[1] Chen S, Xu W, Zhang L, et al. Expanded noninvasive prenatal screening for dominant single-gene disorders: proof-of-concept, performance, and challenges. Am J Obstet Gynecol. 2025 Dec;233(6):673.e1-673.e21.

Disclaimer: NIFTY® mono is a screening test. Pregnancy decisions should not be based solely on the results of NIFTY® mono.The purpose of NIFTY® mono is to indicate if the baby is at increased risk for a genetic disorder allowing for follow-up invasive prenatal studies or newborn studies.

Performing this screening allows for an assessment for known pathogenic and likely pathogenic variants in select genes associated with select disorders. NIFTY® mono should be offered in conjunction with genetic counseling, including a review of family history, to help determine the most appropriate prenatal studies for any pregnant woman.

BGI Introduction

BGI is the world's leading provider of genomic sequencing and proteomic services.

We allow scientists and researchers to reach their full potential by providing them with fully integrated genomic sequencing, proteomic services, and high-quality solutions across a range of applications.

We are committed to enabling and accelerating scientific innovation, strengthening the prevention and control of genetic diseases, and empowering health professionals with advanced diagnostic tools. We endeavor to make meaningful contributions to the development of precision medicine and diagnoses.

Relying on cutting-edge sequencing and bioinformatics technology, we provide our customers with expert and affordable clinical molecular diagnostic solutions and high-throughput sequencing (NGS) research services. These solutions have enabled cutting-edge research that was not even imaginable just a few years ago. Our services cover more than 100 countries and regions, involving more than 2,300 medical institutions.

The BGI Group was founded in 1999 as a research organization to support the Human Genome Project. Over the years, our research has significantly contributed to the development of the genomics field, as evidenced by a wealth of peer-reviewed publications in prestigious scientific journals.

We are passionate about transforming people's lives for the better. We don't hesitate to take bold decisions that can help us to achieve new breakthroughs. We believe that the success of our business will mean a better world for people to live in. We believe in a better tomorrow.

Request for Information or Quotation

www.bgi.com/global/
info@bgi.com

@BGI Genomics

Information is for qualified healthcare professionals only.

Information is not meant to substitute qualified medical advice and is for reference only.

The NIFTY® test is not a diagnostic test. The NIFTY® test screens for the specific genetic conditions listed on the testing panel (as selected for testing). The purpose of the NIFTY® test is to identify pregnancies as more likely to be affected by one of the listed genetic conditions. If the test result returns as high risk, further confirmatory diagnostic testing should be performed for final diagnosis of any condition by a qualified healthcare professional.

Any patient treatment plans should only be recommended and provided by a qualified healthcare professional.

BGI recommends that non-directive genetic counseling and guidance always be provided to patients prior to undertaking any genetic testing and when reviewing results with the patient.

Accuracy of genetic testing may be affected by certain clinical factors. Therefore, test results should always be interpreted in the context of other clinical and family information of the patient.

Informed consent should always be obtained from the patient prior to testing.

Unless otherwise informed, all sequencers and sequencing reagents are not available in Germany, USA, Spain, UK, Hong Kong, Sweden, Belgium or Italy. Certain sequencing services are not available in USA and Hong Kong. Please contact a representative for regional availability. The company reserves the right of final interpretation.

Information, descriptions and specifications in this publication are subject to change without notice.

Published January 2026.

BROCHURE-NIFTY-20260121