NIFTY®
Non-Invasive Prenatal Testing

Non-Invasive Prenatal Screening
Test Request Form

Barcode Area

Patient's Details

Patient's Surname:

Given Names:

Date of Birth: D D / M M / Y Y Y Y

Weight(kg): Height(cm): BMI .

Email:

Phone Number:

Patient Informed Consent Statement

☐* I consent to the test of NIFTY, a non-invasive prenatal screening. I confirm that I have acknowledged, understood, and agree to the Informed Consent provided on the **BACK PAGE** of this form. I confirm that I have had the opportunity to discuss the test and ask relevant questions to my physician, and I have fully understood the indication, intended purpose, procedure, eligibility, limitations, and potential risks of this test, as explained to me by my physician.

☐* I consent to the processing of my personal data by means and for purposes defined in the Privacy Policy.

☐* I confirm the personal information I have provided is true and correct.

☐* I consent to the transfer, processing and storage of my sample, information and genetic data abroad or outside the EU, which will comply with applicable laws and policy.

☐ I consent to the preservation and use of my leftover specimens and de-identified test results in the statistics database for research purpose as stated in the Informed Consent.

Patient Signature:

or **Patient Guardian Signature**:

Date: D D / M M / Y Y Y Y

*In accordance with the applicable regulations, we are not permitted to conduct your tests without these consents.

Test Options

☐ **NIFTY®*** - Trisomy 21, Trisomy 18, Trisomy 13

☐ Sex Chromosome Aneuploidies (optional, for singleton only)

☐ **NIFTY® Pro*** - Trisomy 21, Trisomy 18, Trisomy 13, Trisomy 9, Trisomy 16, Trisomy 22, Sex Chromosome Aneuploidies , other Aneuploidies, 84 Types of Microdeletion and Microduplication Syndromes, and Incidental Findings.

☐ **NIFTY® mono*** - screening for disease-causing variants in the following 18 genes - BRAF, CHD7, COL11A1, COL1A1, COL1A2, COL2A1, FGFR1, FGFR2, FGFR3, HRAS, KRAS, LMNA, MAP2K1, MAP2K2, SOX9, STAT3, TSC1, and TSC2.

☐ *Check if you would like to know the information about **Fetal Sex**

Special Sample Concessions

I understand that my sample may not meet the acceptance criteria for the following reason(s):

☐ Samples may arrive in more than 96 hours but not more than 7 days;

☐ Gestational weeks less than 10 weeks or more than 24 weeks;

☐ BMI more than 40;

I am fully aware of the associated risks of my situation, including failed and/or inaccurate test results, and still willing to continue to test and accept the risks.

Patient Signature:

or **Patient Guardian Signature**:

Date: D D / M M / Y Y Y Y

Ordering Healthcare Provider's Details

Doctor's Name:

Healthcare Provider Name:

Healthcare Provider Address:

Phone Number:

Email(for test results):

Doctors Report Copy to and Their Email (if needed):

Clinical Information

Gestational Age: Weeks Days

Estimated Due Date: D D / M M / Y Y Y Y

Number of Fetus: ☐ Singleton ☐ Twins-can only select NIFTY®

First Sampling ☐ Yes

☐ No, code of the first sampling:_____

Patient's Medical Condition

Received allogeneic blood transfusion:
☐ No ☐ Yes, date of receiving: D D / M M / Y Y Y Y

Received heparin therapy:
☐ No ☐ Yes, time of last taken: D D / M M / Y Y Y Y

Received immunotherapy and/or human serum albumin therapy:
☐ No ☐ Yes, date of last injection: D D / M M / Y Y Y Y

Diagnosed with vanishing twin syndrome:
☐ No ☐ Yes, date of vanishing: D D / M M / Y Y Y Y

Received assisted reproductive technology treatment:
☐ No ☐ Yes, please specify:_____

Family history of genetic disease/s or syndromes:
☐ No ☐ Yes, please specify:_____

Abnormal Reproductive History:
☐ No ☐ Yes, please specify:_____

Abnormal results of other prenatal screening tests:
☐ No ☐ Yes, please specify:_____

Please Attach Any Further Relevant Medical History

Healthcare Provider Statement

I confirm that the patient understands the purpose, limitations, potential risks, scope and performance of the test explained by myself. The patient has given full consent for this test.

Doctor Signature:

Date: D D / M M / Y Y Y Y

Collection Information

Collect Data: D D / M M / Y Y Y Y	Time(AM/PM):
Sample Type: ☐ Whole Blood ☐ Plasma	Collector Signature:

E: info@niftytest.com W: www.bgi.com/global/

NIFTY®
Non-Invasive Prenatal Testing

Non-Invasive Prenatal Screening
Informed Consent

PURPOSE

NIFTY® & NIFTY® Pro are intended to screen fetal trisomy 21, 18, and 13 for pregnancies. Depending on your choice, further details about the clinical condition of the fetus can also be provided, including information on trisomy 22, 9, and 16, sex chromosome aneuploidy, other autosomal trisomy, 84 kinds of microdeletion/microduplication syndromes, Y chromosome conditions (for sex inference), and incidental findings. Incidental findings may not be directly related to your test purpose. However, it may contain information about potential fetal diseases that could be valuable for medical care. Opting NIFTY® Pro and signing this form means that you consent to receive this information.

NIFTY® mono is a non-invasive single-gene disease prenatal test used to screen the fetus for specific disease-causing variants in 18 genes according to authorised databases (see the full list on our website: www.niftytest.com).

TEST PROCEDURE

For each test option you choose, a tube of blood will be drawn and sent to a BGI-owned and/or a partnered laboratory, which will then analyse your DNA using molecular genetics technology. Before and after undertaking the tests, you should consult with the healthcare professionals regarding any risks, diagnoses, treatment and/or any other potentially relevant healthcare issues.

ELIGIBILITY

Patients should be at least 10 weeks' gestational age and with a BMI < 40. Perform the test before 24 gestational weeks of pregnancy to have enough time for further diagnosis or procedure.

Patients who have the following situations are NOT eligible for any of **NIFTY®**, **NIFTY® Pro** or **NIFTY® mono**:

- Have chromosomal abnormality;
- Pregnancy with triplets or more fetuses;
- Have jaundice and/or hyperlipidaemia;
- Have blood-borne infectious diseases;
- Have malignant tumours and/or had a history of malignant tumours or benign tumours (except benign uterine fibroids);
- Maternal, fetal and/or placental mosaicism;
- Received transplant surgery or stem cell therapy;
- Received allogeneic blood transfusion within one year;
- Received human serum albumin therapy and/or exogenous DNA cells introduced immunotherapy within four weeks;
- Received heparin therapy within 24 hours;
- One fetus in twins has developmental defects;
- Have vanishing twin syndrome, unless it has been identified that the developmental arrest occurred within the first eight weeks of pregnancy and more than eight weeks prior to the date of the test;

Patients who have the following situations are NOT eligible for NIFTY® mono:

- Egg donation recipients;
- Single-gene genetic disease;

Patients with the following conditions have a high risk of maternal genetic background for abnormalities which may cause inaccurate test results. Please check genetic abnormalities prior to their test. In these cases, the decision to undergo the tests or not should be made by the patients and/or their health provider in conjunction with local medical and bioethics guidelines, laws and/or regulations. Signing this form means you are fully aware of and willing to accept the risks:

- Patients received Assisted Reproductive Technology therapy (including *In-Vitro* Fertilization & Embryo Transfer, Intracytoplasmic Sperm Injection, *In-Vitro* maturation, *In-Vitro* Gametogenesis, Germinal Vesicle Transfer, Egg/Sperm Donation, Surrogacy);
- Patients who have a history of abnormal pregnancy, or family history of genetic disease or abnormal phenotype;
- Another screening result indicates fetal abnormalities.

TEST LIMITATION

The tests are NOT intended nor validated for diagnostic purposes; thus, the result cannot be used as the sole evidence for a diagnostic conclusion. The sensitivity and specificity of the tests are based on singleton pregnancies. According to studies and theory, the tests perform similarly in twin and singleton pregnancies. The tests cannot be used to predict diseases that are not in the test scope, or rule out risks in patients' families. NIFTY® mono may not cover all pathogenic or likely pathogenic variants in tested genes.

A negative result cannot totally exclude the possibility of fetal abnormality. Certain rare biological conditions may also affect the accuracy of the tests. Potential sources of false positive or false negative results include, but are not limited to: maternal, fetal and/or placental mosaicism, low fetal fraction, blood transfusion, organ transplant, immunotherapy and/or stem cell therapy. 'Vanishing twin syndrome' may also cause test inaccuracy in the event of twin pregnancy testing. Abnormalities caused by chromosomal polyploid (triploid, tetraploid, etc.), chromosomal balanced translocation, inversion, ring, UPD, monogenic/polygenic disease, etc., cannot be detected by this test; this test cannot exclude the fetal chimeric chromosomal abnormalities.

	Sensitivity Rate (singleton pregnancies)	Available for Twin Pregnancy
Trisomy 21	>99%	YES
Trisomy 18		
Trisomy 13		
Gender Identification	99.53%	YES
Rare Autosomal Trisomy	>99%	NO
Sex Chromosome Aneuploidies	>99%	NO
Microdeletions/ Microduplications	90%	NO

* From in-house data. Internal analysis shows a detection rate of over 90% when cffDNA over 9.5% in selected del/dup syndromes with abnormal size over 3M.

Reference:

- Hong Y, GAO Y, Jia Z, et al. Genome-wide detection of additional fetal chromosomal abnormalities by cell-free DNA testing of 15,626 consecutive pregnant women[J]. SCIENCE CHINA Life Sciences.
- Pan X, Zhang C, Li X, et al. Non-invasive fetal sex determination by maternal plasma sequencing and application in X-linked disorder counseling[J]. The Journal of Maternal-Fetal & Neonatal Medicine, 2014, 27(18): 1829-1833 Jiang et al. Noninvasive Fetal Trisomy (NIFTY) test: an advanced noninvasive prenatal diagnosis methodology for fetal autosomal and sex chromosomal aneuploidies. BMC Medical Genomics. 2012 5:57.
- Yao H, et al. Detection of fetal sex chromosome aneuploidy by massively parallel sequencing of maternal plasma DNA: initial experience in a Chinese hospital. Ultrasound Obstet Gynecol. 2014 Jul;44(1):17-24. doi:10.1002/uog.13361

RESULTS

Reports will be available within 7-12 days from the time the laboratory receives the sample. Results will be sent only to the undersigned healthcare provider due to their complexity and implications. Patients should contact their healthcare provider for test results and interpretation. High-risk results should be followed by confirmatory diagnostic tests.

Occasionally samples fail quality control and/or the initial analysis cannot reach a conclusion. This may require resampling and/or reanalysis, which will be offered free but may delay your report. You will be notified by your healthcare provider if this happens.

INTERNATIONAL DATA TRANSFER

The sample and filled information (including first name, last name, address, date of birth, disease, symptoms and other medical information) will be sent to BGI and/or their partnered laboratory for testing, which may be located in Hungary, Denmark, Australia, Uruguay, Thailand, the UK, Hong Kong (China), etc. We have taken legally required appropriate safeguards to ensure the data protection when transferring your personal data abroad. In principle, samples, information and data of the patient in the European Union, where GDPR provisions apply, will only be processed within the EU. In some situations, your samples, information and data may need to be transferred outside your country or the EU. This transfer will only take place with your consent.

PRIVACY POLICY

The information and test results of the patient are kept confidential and all data will remain anonymous during analysis. Only your healthcare provider will receive your test results unless required or authorised by applicable law.

For your test, we need your clinical information about your pregnancy , such as ultrasound/other screening/diagnostic tests performed during the pregnancy. Auditing, quality assurance, and research may use clinical information. Please read the BGI Privacy Policy (available on: the website: www.niftytest.com/privacy-policy/), which is in every case considered as part of this consent.

USE OF LEFTOVER SPECIMENS AND INFORMATION

In compliance with better practices, your de-identified specimens and genetic and other information obtained from your tests may be utilised for scientific purposes, technological development, and/or clinical research. Personal information will be removed before reports and publications. All written uses will comply with applicable laws. If you do not agree, your leftover samples will be destroyed after expiry in accordance with international clinical laboratory standards.

The specimens and data will be destroyed if you revoke the test (de- identified data cannot be removed or traced). If you have any questions about your rights as a research subject or concerns, requests or complaints regarding this research, please contact: info@niftytest.com

RIGHT OF REVOCATION

You may revoke your consent to the test in full or in part at any time, without providing a reason. You have the right not to be informed of test results (right not to know), to halt testing processes at any time prior to receiving the results, and to request the destruction of all test materials and results collected up until that point.

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