Non-Invasive Fetal TrisomY Testing

Over 15,000,000+ Tests Conducted

NIFTY® is a world-leading non-invasive prenatal test (NIPT) product independently developed by BGI Genomics. By collecting >5 mL of maternal peripheral blood, extracting cell-free DNA, and using low-depth whole-genome sequencing technology, combined with bioinformatics analysis, the risk of fetal chromosomal abnormalities can be determined.

It is a safe and accurate method for detecting T21, T18 and T13, and can expand detection for other autosomal aneuploidies, sex chromosome aneuploidies, and pathogenic chromosomal deletions/duplications (CNVs).

Advantages

Accurate
Over 99% sensitivity for trisomy 21, 18 and 13.

Safe
Only maternal peripheral blood is needed, **no risk** to mother or fetus.

Trusted
Over 15,000,000 samples processed worldwide.

Comprehensive
NIFTY® Pro detects **over 100 genetic conditions**.

Early
Screen as early as the **10th week of pregnancy**.

Quick
TAT as fast as **5 working days**.

(The delivery time may vary due to different service form, please refer to the actual situation)

Insured
Offering insurance in cases of false positive or false negative.

(Insurance service is limited to samples sent to BGI laboratory for testing only)

Test Options

NIFTY®

Common Autosomal Trisomies

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

Rare Autosomal Trisomies

Trisomy 9
Trisomy 16
Trisomy 22

Sex Chromosome Aneuploidies*

X0 (Turner syndrome)
XXY (Klinefelter syndrome)
XXX (Triple-X syndrome)
XYY (Jacobs syndrome)

Incidental findings*

Other autosomal aneuploidies

Sex Indication*

Y chromosome detection

NIFTY® pro

Common Autosomal Trisomies

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92/10 micro deletion/duplication, including

DiGeorge syndrome (22q11.2 deletion)
1p36 deletion syndrome
Prader-Willi/ Angelman syndrome
Smith-Magenis syndrome
Cri-du-Chat syndrome
4p16.3 deletion syndrome
Distal 18q deletion syndrome
18p deletion syndrome
9p deletion syndrome
Jacobsen Syndrom

Incidental findings

Other autosomal aneuploidies
& ≥5Mb CNVs

Sex Indication*

Y chromosome detection

* The above asterisks are optional detection

The detection of sex chromosomal aneuploidies is limited to singleton pregnancy.

NIFTY® and NIFTY® Pro has completed large-scale validation[1,2].

Chromosome abnormalities	Sensitivity (95%CI)	Specificity (95%CI)	PPV (%)
Common Trisomies	99.02% (98.38%-99.66%)	99.86% (99.84%-99.88%)	85.27%
T21	99.17% (98.52%-99.83%)	99.95% (99.93%-99.96%)	92.19%
T18	98.24% (94.93%-99.63%)	99.95% (99.93%-99.96%)	76.61%
T13	100.00% (84.56%-100.00%)	99.96% (99.95%-99.97%)	32.84%
SCAs	100.00% (90.59%-100.00%)	99.90% (99.85%-99.94%)	68.12%
RATs	100.00% (19.79%-100.00%)	99.88% (99.82%-99.92%)	6.67%
CNVs	100.00% (74.65%-100.00%)	99.94% (99.89%-99.97%)	51.72%
< 10Mb	100.00% (59.77%-100.00%)	99.86% (99.93%-99.98%)	50.00%
≥ 10Mb	100.00% (56.09%-100.00%)	99.97% (99.94%-99.99%)	53.85%

Note: The data in the table comes from historical literatures and only reflects the test results at the time of the studies. In actual applications, deviations may occur due to factors such as the population and sample. They are for reference only and do not represent promised values.

PPV, positive predictive; SCAs, sex chromosome abnormalities.

RATs, rare autosomal aneuploidies; CNVs, copy number variations.

[1]Zhang H, Gao Y, Jiang F, et al. Ultrasound Obstet Gynecol (2015), 45:530-538.

[2]Zou Y, Feng C, Qin J, et al. Front. Genet.(2023), 13:1073851.

Sample Requirements

Sample Type	Quantity	Requirements	Shipment
Mternal peripheral blood	6-10mL	Using authorised cell-free DNA blood collection tubes	Stored and shipped between 6~35 °C within 4 days. Keep the tubes upright during shipping
Plasma	1.8mL	Plasma separation needs to be completed within 96 hours	Stored and shipped in no higher than -20˚C within 7 days

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 a digital copy of sample information sheet to BGI
5. Arrange collection of blood sample with courier
6. Send Consent Form/ Test Request Form with blood sample to BGI laboratory
7. Receive results in 5-7 working days
8. Conduct post-test genetic counseling and provide guidance advice as required

The test is suitable for

- Any age of pregnant women from 10 weeks gestation
- Singleton, twins and vanishing twin syndrome (VTS) pregnancies
- IVF pregnancies

(For the specific applicable, caution, and prohibited population, please conduct genetic counseling before testing)

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