

Non-Invasive Prenatal Testing – NIPT

NIPT screens a maternal blood sample for chromosomal aneuploidy in fetal DNA using the following methodology:

- (1) Extraction of fetal cell-free DNA from the maternal blood sample
- (2) High throughput sequencing of the extracted fetal cell-free DNA
- (3) Calculation of molecular mass of fetal DNA in all chromosomes








Based on the scope, NIPT can screen the following conditions:

- (a) Whole Genome - 23 pairs of human chromosomes
- (b) Common Chromosomal abnormality:
 - Trisomy 13 (Patau's Syndrome)
 - Trisomy 18 (Edwards' Syndrome)
 - Trisomy 21 (Down's Syndrome)

NIPT is capable of genome-wide aneuploidy detection of the whole fetal genome (23 pairs of chromosomes). Test results with the interpretation of risk for Trisomy 13 Trisomy 18, Trisomy 21 and sex chromosome aneuploidies will be provided. This test confers an accuracy of up to **99%** on the detection of fetal chromosomal aneuploidy.

| Pregnancy Type | Blood ID | Patient ID |
|----------------|----------|------------|
| Singleton | NA | NA |

Test Results Summary

| Autosomal Aneuploidies | Risk | Test Results | Aneuploidy Risk |
|---|---|----------------|-----------------|
| Chromosome 13  |  | Low risk group | < 1/10000 |
| Chromosome 18  |  | Low risk group | < 1/10000 |
| Chromosome 21  |  | Low risk group | < 1/10000 |
| Other Chromosomes |  | Low risk group | - |



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Test Results for Sex Chromosome Aneuploidies

| Sex Chromosome Aneuploidies | Risk | Test Results | Aneuploidy Risk |
|-----------------------------|------|----------------|-----------------|
| XO | ● | Low risk group | < 1/10000 |
| XXY | ● | Low risk group | < 1/10000 |
| XXX | ● | Low risk group | < 1/10000 |
| XYY | ● | Low risk group | < 1/10000 |

✂ Risk description: ● Low risk group; ● Borderline group; ● High risk group

Test Results for Other Chromosomal Aneuploidies

| Other Chromosomal Aneuploidies | Risk | Test Results |
|---|------|----------------|
| Chromosome 1  | ● | Low risk group |
| Chromosome 2  | ● | Low risk group |
| Chromosome 3  | ● | Low risk group |
| Chromosome 4  | ● | Low risk group |
| Chromosome 5  | ● | Low risk group |
| Chromosome 6  | ● | Low risk group |
| Chromosome 7  | ● | Low risk group |
| Chromosome 8  | ● | Low risk group |
| Chromosome 9  | ● | Low risk group |
| Chromosome 10  | ● | Low risk group |
| Chromosome 11  | ● | Low risk group |



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| | | | |
|---------------|--|--|----------------|
| Chromosome 12 | | | Low risk group |
| Chromosome 14 | | | Low risk group |
| Chromosome 15 | | | Low risk group |
| Chromosome 16 | | | Low risk group |
| Chromosome 17 | | | Low risk group |
| Chromosome 19 | | | Low risk group |
| Chromosome 20 | | | Low risk group |
| Chromosome 22 | | | Low risk group |

※ Risk description: Low risk group; Borderline group; High risk group

Sample information

Fetal DNA fraction

10.09%

Note: In rare cases when fetal DNA fraction level is low, new blood sample will be requested for retesting.

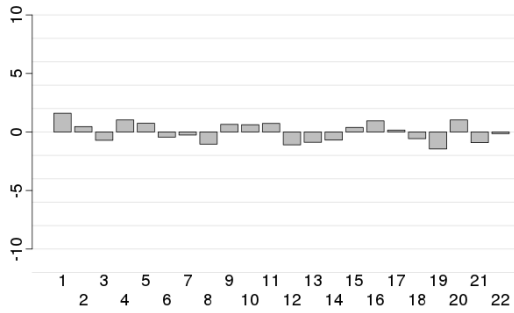
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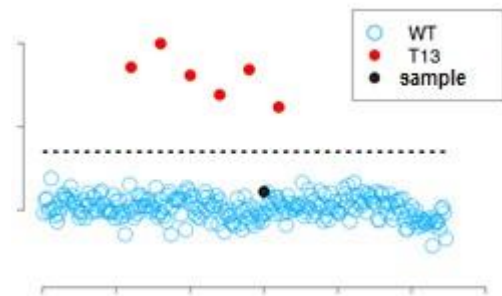
Test Results

Chromosome 1 - 22



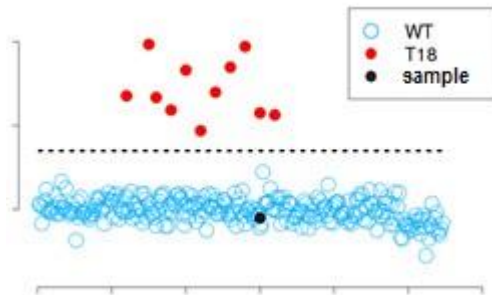
Chromosome 13

Z-score -0.208



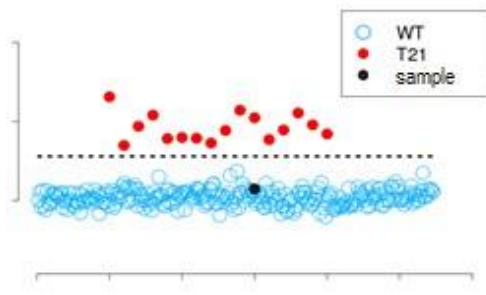
Chromosome 18

Z-score -0.219



Chromosome 21

Z-score -0.482



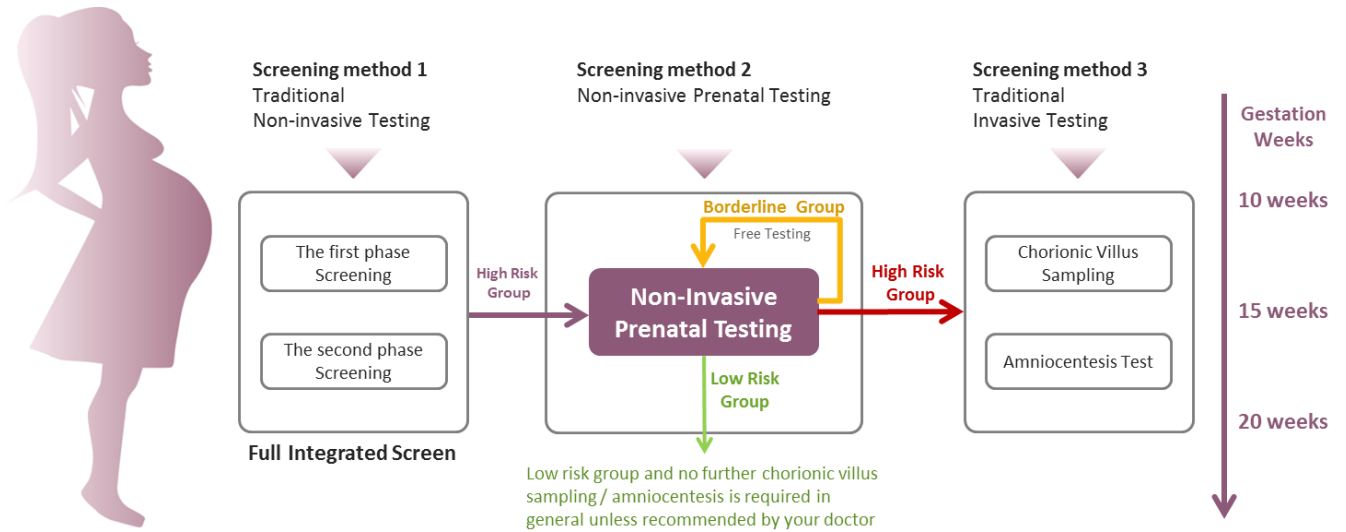
- These images are only for representation puposes.

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Prenatal Examination



About the Test

YES-IN-GENE specializes in research development and high-quality professional services in clinical genetic testing. NIPT analyzes circulating fetal cell-free DNA extracted from a maternal blood sample, and is offered to pregnant women with a pre-test risk of aneuploidy in chromosomes such as 13, 18, 21, X or Y. The chance that a fetus is affected with chromosomal aneuploidy can be estimated using bioinformatics analyses, by which the accuracy rate and sensitivity are over 99%. The accuracy and quality of the test may be affected by low fetal fraction, high data noise due to improper blood sample collection, handling, storage, or transportation.

Limitations of the Test

Non-invasive prenatal testing should only be considered a screening test. The screening test of fetal cell-free DNA cannot compare with the prenatal diagnosis with Amniocentesis or Chorionic Villus Sampling (CVS). Pregnant women with a positive NIPT screening result should be given an invasive prenatal diagnosis and referred further for genetic counselling to confirm conditions. On the other hand, a negative test result does not ensure an unaffected pregnancy. Even though NIPT provides reliable results, it does not apply to all cases of chromosomal abnormalities, for example, cases due to placental, maternal, or fetal mosaicism, or other causes (e.g. micro-deletions, chromosome re-arrangements, translocations, inversions, unbalanced translocations, uniparental disomy, etc.). NIPT is also not applicable for cases with a diagnosed multiple gestation, or with gestational age that is less than 10 weeks. In rare cases when a borderline screening result is reported, retesting is required to confirm conditions.

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Test Method

NIPT applies a non-invasive and low-risk procedure to collect fetal DNA samples. Circulating fetal cell-free DNA is purified from the plasma component of 10mL anti-coagulated maternal whole blood. It is then converted into a genomic DNA library for Next Generation Sequencing to determine Trisomy 21, 18 and 13 and other chromosomal abnormalities.

References

1. Obstet Gynecol 2012; 119:890-901.
2. BMJ 2011; 342:c7401.
3. Prenat Diagn 2012; 32:c7401.
4. ACOG/SMFM Joint Committee Opinion No. 545, Dec 2012.

Note: The sex of fetus is not revealed due to PNDT Act.

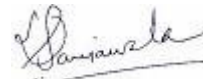
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