QR Code for report verification

Case ID	: **2100128913**	**Sample Type**	: **BLOOD IN STRECK TUBE**
Name	: X	**Date & Time Collected**	: **24-Dec-2021 04:45 PM**
Sex/Age	: **Female/37 Years**	**Date & Time Received**	: **24-Dec-2021 04:56 PM**
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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 aneuplodies will be provided. This test confers an accuracy of up to **99%** on the detection of fetal chromosomal aneuploidy.

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
Sex Chromosome		High risk group	< 1/20
Other Chromosomes		Low risk group	-

Test Results forSex Chromosome Aneuploidies

Sex Chromosome Aneuploidies	Risk	Test Results	Aneuploidy Risk
XO		Low risk group	< 1/10000

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XXY/XYY	🔴	High risk group	< 1/20
XXX	🟢	Low risk group	< 1/10000

※ Risk description: 🟢 Low risk group; 🟠 Borderline group; 🔴 High risk group

ADVISE : Amniocentesis to rule out XXY/XYY.

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
Chromosome 12		🟢	Low risk group
Chromosome 14		🟢	Low risk group

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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

Qubit Fluorometer (ng/uL)	0.81
Volume (uL)	30
Total amount (ng)	24.30
Fetal DNA fraction	11.87%

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

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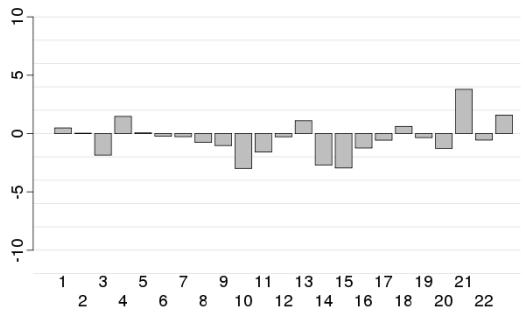
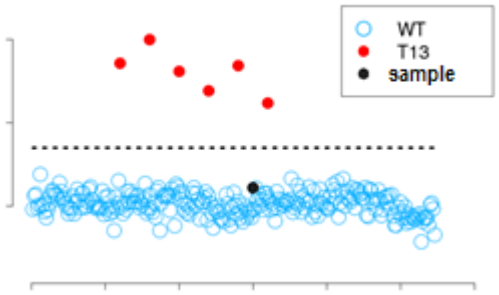
Test Results

Chromosome 1 - 22



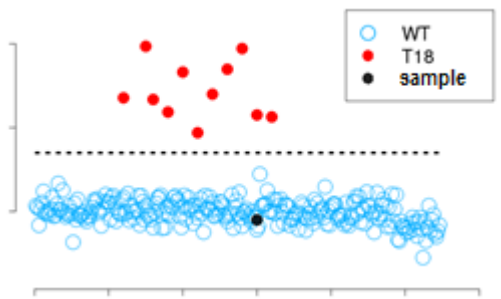
Chromosome 13

Z-score	1.17



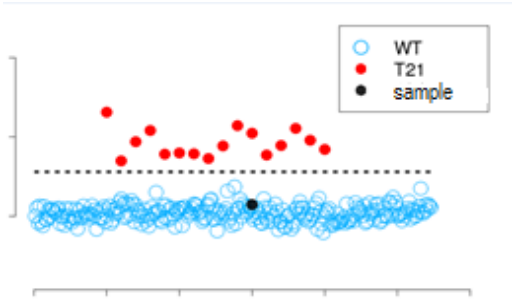
Chromosome 18

Z-score	-0.02



Chromosome 21

Z-score	-1.01



Prenatal Examination

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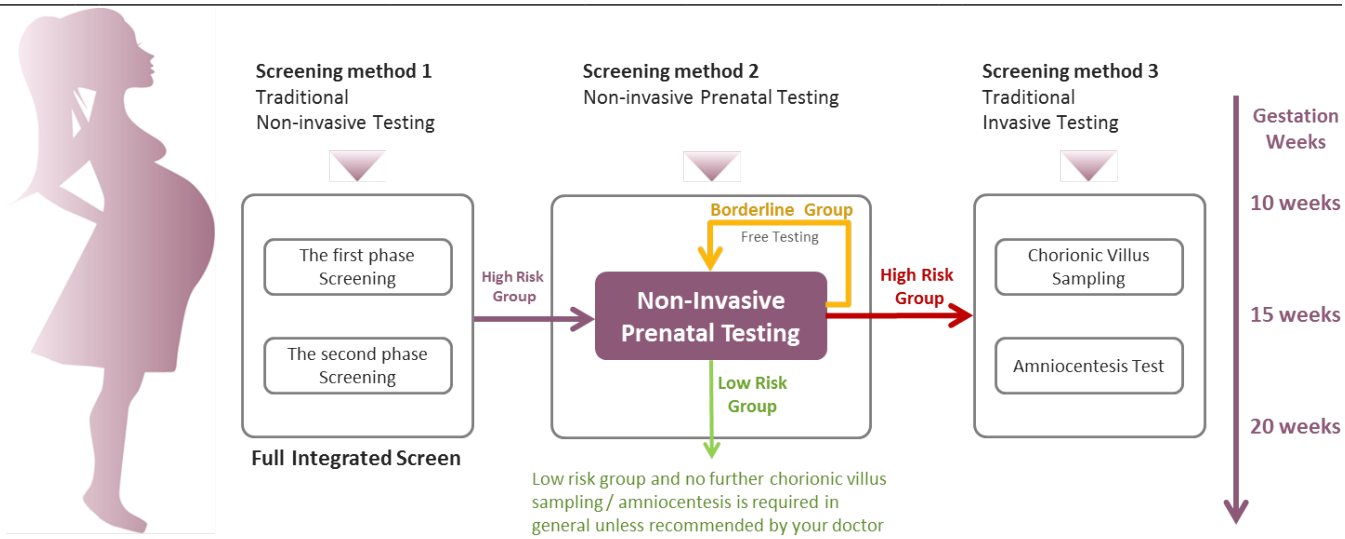

About the Test

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 revealed due to presence of aneuploidy involving sex chromosome.

---------- End Of Report ----------

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