PATIENT INFORMATION AND DATA

YES-IN-GENE: Non-Invasive Prenatal Testing – NIPT

YES-IN-GENE: 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
(1) High throughput sequencing of the extracted fetal cell-free DNA
(1) 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
(a) 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.

Test Results Summary:

Chromosome	Category	Test result			
		Group			Risk of aneuploidy
		Low risk	Borderline	High risk	
Chromosome 13	Autosomal	●	☒	☒	< 1/10000
Chromosome 18	Autosomal	●	☒	☒	< 1/10000
Chromosome 21	Autosomal	●	☒	☒	< 1/10000
Other chromosomes	Autosomal	●	☒	☒	

PATIENT INFORMATION AND DATA

Test Results for Sex Chromosome Aneuploidies:

Chromosome	Category	Test result			
		Group			Risk of aneuploidy
		Low risk	Borderline	High risk	
XO	Sex chromosome	●	☒	☒	< 1/10000
XXY	Sex chromosome	●	☒	☒	< 1/10000
XXX	Sex chromosome	●	☒	☒	< 1/10000
XYY	Sex chromosome	●	☒	☒	< 1/10000

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

Sample details:

Parameter	Unit	Value
DNA conc.	Nanogram per microliter (ng/µL)	0.71
Volume	Microliter (µL)	30
Total amount	Nanogram (ng)	21.3
Fetal DNA fraction	Percentage (%)	8.24%

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

Z-Score:

Chromosome	Z-Score
Chromosome 13	1.24
Chromosome 18	-0.94
Chromosome 21	0.16

PATIENT INFORMATION AND DATA

Test Description:

YES-IN-GENE : 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 fetalmosaicism, or other causes (e.g. micro-deletions, chromosome re-arrangements, translocations, inversions, unbalanced translocations, uniparentaldisomy, 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.

Testing Methodology:

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.
Samples for NIPT are processed with Ion Chef™ or Ion OneTouch™ 2 System and the sequencing is performed on the Ion Proton™ or S5™ Sequencer (ThermoFisher Scientific, Inc.).

References

1. *ObstetGynecol 2012; 119:890-901.*
2. *BMJ 2011; 342:c7401.*
3. *PrenatDiagn 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.

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