Non-Invasive Prenatal Test Report

Patient Information	Specimen Information	Test Information
Patient name: Date of Birth: Sex:	Sample ID: Work No.: Date collected:	Test reported: Ordering physician: Institution:

Patient Information

Quality Control

Gest. Age / Weight	Ultrasound Feature	Multiple Marker Screening Test	In-vitro Fertilization	No. of Fetus	DNA Quality	NGS Data Quality	Material Quality
12+1 / 55kg	None	Low Risk	None	Single	Pass	Pass	Pass

NEGATIVE RESULT

TEST RESULT

Fetal Fraction: 10.9% (+)

Chromosomal Abnormality	Result	Positive Predictive Value (PPV)	False Positive Rate (FPR)
Trisomy 21	**Negative**	-	-
Trisomy 18	**Negative**	-	-
Trisomy 13	**Negative**	-	-
Sex Aneuploidy (X, XXX, XXY)	**Negative**	-	-
Additional Findings			

INTERPRETATION

As the G-NIPT result, any fetal chromosomal abnormalities in the autosomes and sex chromosomes did not detected. However, we cannot completely rule out the possibility of false negative due to maternal chromosomal microdeletion/duplication, confined placental mosaicism (CPM), low fetal fraction and low level fetal mosaicism. It is recommended to perform a high resolution cytogenetic testing if any fetal abnormalities are found on ultrasonography regardless of G-NIPT result.

TEST INFORMATION

- Test Method: Massively Parallel Sequencing(MPS) Test
- Subject: Fetal Trisomy (Chromosome 21, 18, 13), Sex Chromosome Aneuploidy
- Specimen Type : EDTA Plasma 5mL or cfDNA tube WB 10mL

TEST PERFORMANCE

Test Item	Sensitivity	Specificity	PPV	NPV
Trisomy 21	>99.99%	>99.99%	>99.99%	>99.99%
Trisomy 18	>99.99%	>99.99%	>99.99%	>99.99%
Trisomy 13	>99.99%	>99.99%	>99.99%	>99.99%
Sex Aneuploidy	>99.99%	>99.99%	>99.99%	>99.99%

METHOD AND LIMITATIONS

- The purpose of this test is for risk assessment of common fetal trisomies 21, 18, 13 and sex chromosome aneuploidies. This test is performed by massively parallel sequencing for whole-genome using circulating cell-free fetal DNA in maternal plasma and it is possible to detect abnormalities in all chromosomes as well as chromosome 21, 18 and 13. NIPT have a higher performance result compared to existing prenatal multiple marker screening tests.
- This test is not to verify fetal karyotypes but is to determine the risk of fetal aneuploidies. If the result is positive, confirmatory test such as fetal karyotyping should be performed. Moreover, this is not a diagnostic test which does not eliminate probability of false positive or false negative results.
- The factors affecting accuracy of this test are as follows: low fetal DNA fraction (early gestational weeks and high maternal BMI), undetermined maternal chromosomal abnormalities, confined placental mosaicism, fetal chromosomal mosaicism, multiple gestation, arithmetic error of calculating fetal DNA fraction, and maternal status (cancer, blood transfusion, transplantation, chemotherapy, stem cell treatment, or autoimmune disease).

CAP ACCREDITED

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