SAMPLE REPORT

Non-Invasive Prenatal Test Report

Personal Information		Specimen Information		Test Information	
Patient Name	Jane Doe	**Sample ID**	202111039712102	**Institution**	Hospital A
		Medical Record No	-	**Ordering physician**	Dr. Smith
Date of Birth	1995/03/08	**Date collected**	2021/10/28	**Test reported**	2021/11/08

PREGNANCY INFORMATION						QUALITY CONTROL		
Gest.Age/Weight	Ultrasound Feature	Nuchal Translucency (NT)	Multiple Marker Screening Test	In-vitro Fertilization	No. of Fetus	DNA Quality	NGS Data Quality	QC Quality
14w+2d/61kg	None	1mm	None	None	Single	Pass	Pass	Pass

FETAL FRACTION	9.1%	FETAL SEX	Male

TEST RESULT	LOW RISK		
Chromosomal Abnormality	**Result**	**Risk after NIPT**[1)]	**Maternal age-specific risk**[2)]
Trisomy 21	LOW RISK	< 1/10000 (0.01%)	1/423
Trisomy 18	LOW RISK	< 1/10000 (0.01%)	1/1301
Trisomy 13	LOW RISK	< 1/10000 (0.01%)	1/4052
XO	LOW RISK	< 1/10000 (0.01%)	1/1000
XXX	LOW RISK	< 1/10000 (0.01%)	1/1400
XXY	LOW RISK	< 1/10000 (0.01%)	1/1200
Trisomy 9	LOW RISK	N/A	
Trisomy 16	LOW RISK		
Trisomy 22	LOW RISK		

Deletion Syndrome	Result
1p36	LOW RISK
2q33.1	LOW RISK
5p15 (Cri-du-chat)	LOW RISK
11qter (Jacobsen)	LOW RISK
Other Microdeletions[3)]	Not Detected

1) Risk after NIPT: Maternal age-specific risk * Relative risk for the corresponding chromosomal results of G-NIPT
2) Maternal age-specific risk: Average risk for the chromosomal aneuploidy in the same age group of pregnant women.
* >7Mb deletion tested

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ADDITIONAL TEST RESULT

Chromosomal Abnormality	Result	Chromosomal Abnormality	Result
Trisomy 1	LOW RISK	**Trisomy 10**	LOW RISK
Trisomy 2	LOW RISK	**Trisomy 11**	LOW RISK
Trisomy 3	LOW RISK	**Trisomy 12**	LOW RISK
Trisomy 4	LOW RISK	**Trisomy 14**	LOW RISK
Trisomy 5	LOW RISK	**Trisomy 15**	LOW RISK
Trisomy 6	LOW RISK	**Trisomy 17**	LOW RISK
Trisomy 7	LOW RISK	**Trisomy 19**	LOW RISK
Trisomy 8	LOW RISK	**Trisomy 20**	LOW RISK

* The clinical sensitivity was not determined due to low incidence.

INTERPRETATION

No fetal chromosomal abnormalities in autosomes and sex chromosomes were found. However, we cannot completely rule out the possibility of false negative results that may be caused by factors such as maternal chromosomal microdeletion/duplication, confined placental mosaicism (CPM), and low fetal fraction. If any fetal abnormalities are found by ultrasonography, it is recommended to perform high-resolution cytogenetic testing regardless of the result of G-NIPT.

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TEST INFORMATION

- Test Method: Next Generation Sequencing (NGS)
- Test Subject: Fetal Trisomy (Chromosome 21, 18, 13, 9, 16, 22), Sex Chromosome Aneuploidy, Microdeletion Syndrome (>7Mb)
- Specimen Type: cfDNA tube WB 10mL
- Bioinformatics Pipeline: NIPT.v1.2

TEST PERFORMANCE

Test Item	Sensitivity	Specificity	NPV	PPV
Trisomy 21	99.66%	99.99%	99.99%	99.32%
Trisomy 18	99.09%	99.98%	99.99%	95.61%
Trisomy 13	99.99%	99.99%	99.99%	71.43%
Sex Chromosome Aneuploidies (XO, XXX, XXY, XYY)	99.99%	99.86%	99.99%	48.21%
Other Chromosomes	The clinical sensitivity was not determined due to low incidence.			
Microdeletion Syndrome	The clinical sensitivity was not determined due to low incidence, and the sensitivity may be significantly affected by factors such as fetal DNA fraction and microdeletion size.			

* Test performance is based on the G-NIPT test result conducted between 2015.12 ~ 2019.02 and may be changed in the future.

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 performance is superior to the existing prenatal multiple marker screening tests.
- This test cannot identify neural tube defects and polyploidy such as trioploidy and tetraploidy.
- This test does not report monosomy. Fetal sex is not reported for twins.
- In case of trisomy 9, 16, 22 and microdeletion syndrome, the clinical sensitivity was not determined due to low incidence, and the sensitivity may be significantly affected by factors such as fetal DNA fraction and microdeletion size.
- 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 rule out 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), etc.

REFERENCE

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- Clin Genet. 2016 May;89(5):523-30. Clinical implementation of NIPT - technical and biological challenges
- Fetal Diagn Ther. 1995 Nov-Dec;10(6):356-67