NICE® Test Report for Twin Pregnancy

NICE®
Non Invasive Chromosome Examination

Sample Information
Sample Type: PLASMA
Client Sample ID:
Date of Draw:
Date Received:
Reporting Date:
Resample: Yes

Patient Information
Name:
Date of Birth:
Gest. Age at Draw:
Pregnancy type: Twin
Indication: Other ()
Medical Record/Patient ID:

Provider Information
Hospital:
Physician:
Phone:
Fax:

Quality Test

Sample suitablilty	Pass	**NGS data quality**	Pass
DNA quality	Pass	**Reference material test**	Pass
Library quality	Pass	**Fetal fraction**	17.0%

Results

Ver.4.0

Chromosome	Result	Interpretation
Trisomy 21	Low Risk	Result consistent with two copies for Chromosomes 21,18,13.
Trisomy 18	Low Risk	
Trisomy 13	Low Risk	

Fetal Sex	**Y chromosome detected**

Limitations of Test

This test is designed to screen for fetal chromosome aneuploidies from cell-free DNA analysis, and is validated for detecting chromosome 21, 18, 13 clinical sample testing. The test is validated for singleton pregnancies with gestational age of at least 10 weeks, as estimated by last menstrual period. These results do not eliminate the possibility that this pregnancy may be associated with other chromosomal or subchromosomal abnormalities, birth defects, and other conditions. This test is not intended to identify pregnancies at risk for open neural tube defects. A negative test result does not eliminate the possibility of chromosomal abnormalities such as trisomy 21, trisomy 18, trisomy 13. There is a small possibility that the test results might not reflect the chromosomes of the fetus, but may reflect chromosomal changes of the placenta(confined placental mosaicism) or of the mother(chromosomal mosaicism). Potential sources of an inaccurate test result may include but are not limited to: maternal, fetal and/or placental mosaicism, low fetal fraction, blood transfusion, transplant surgery and stem cell therapy. Especially, fetal deduction including vanishing fetus, fetal demise can result in false negative result or false positive result. If definitive diagnosis is desired, chorionic villus sampling or amniocentesis would be necessary.

■ **Disclaimer :** The manner in which this information is used to guide patient care is the responsibility of the healthcare provider, including advising for the need for genetic counseling or diagnostic testing. Any test should be interpreted in the context of all available clinical findings. The result of this test are not interceded to be used as the sole means for management decisions.

Predictive Value based on Maternal age

(%)

Maternal age	T21		T18		T13	
	PPV	NPV	PPV	NPV	PPV	NPV
27	91	>99	>99	>99	>99	>99

* PPV : Positive Predictive Values, NPV : Negative Predictive Values
* All the probabilities and rates are calculated using below site based on the maternal age and NICE sensitivity/specifcity data. (https://www.perinatalquality.org/Vendors/NSGC/ NIPT/)
* The predictive rates may vary depending on gestational age, ultrasound findings, and biochemical screening.

Performance Metrics

Trisomy	N	Sensitivity %	95% CI	Specificity %	95% CI	PPV %	95% CI	NPV %	95% CI
T21	6,238	100(54/54)	93.40-100	99.98(6,237/6,238)	99.91-100	98.18	88.38-99.74	100	99.92-100
T18	6,238	100(14/14)	76.84-100	100(6,238/6,238)	99.91-100	100	73.23-100	100	99.92-100
T13	6,238	100(2/2)	15.81-100	100(6,238/6,238)	99.91-100	100	19.78-100	100	99.92-100

Method and Disclosure

■ Test Method : Cell-free DNA extraction, massively parallel sequencing, and analysis of sequencing results to determine fetal aneuploidy.
■ Disclosure : This test was developed and its performance characteristics determined by Eone-Diagnomics Genome Center, Co. Ltd. The EDGC laboratory is CAP-accredited and certified under Korean Institute of Genetic Testing Evaluation (KIGTE) as qualified to perform accurate genetic testing. This prenatal test has not been cleared or approved by the FDA. This test is a trademark of by Eone-Diagnomics Genome Center, Co. Ltd. All names. Logos and trademarks are the property of by Eone-Diagnomics Genome Center, Co. Ltd.

References

1. Anthony R et al., Noninvasive prenatal screening for fetal aneuploidy 2016 update: a position statement of the American College of Medical Genetics and Genomics. Genet Med 2016; 18:1056-65.
2. Bianchi DW et al., DNA sequencing versus standard prenatal aneuploidy screening. N Engl J Med. 2014; 370:799-808.
3. Kwon HJ et al., Multiple z-score based method for noninvasive prenatal test using cell-free DNA in maternal plasm. Open Journal of Genetics, 2017; 7:1-8.
4. Wong FC, Lo YM. Prenatal Diagnosis Innovation: Genome Sequencing of Maternal plasma. Annu Rev Med 2016; 67: 419-32.
5. Yun SY et al., Noninvasive prenatal testing for fetal chromosomal abnormalities using massively parallel sequencing: Clinical experience from 7,910 Korean pregnancies. Open Journal of Genetics, 2018; 3:42-53.

EDGC EONE-DIAGNOMICS Genome Center BRIA
CAP & KIGTE Laboratory Director : Jin-Sik Bae, Ph.D. CAP : 9515154 KIGTE : 259
291 Harmony-ro, Yeonsu-gu, Incheon 22014, South Korea / www.edgc.com
6 (Sonthiwattana3) Ladprao 110, Plubpla, Wangtonglang, Bangkok 10310, Thailand.
TEL +662 106 6999