HK·DNA® 香港基因檢測中心化驗所有限公司
DIAGNOSTICS CENTRE LABORATORY LTD.

Non-invasive Prenatal Genetic Testing of Fetal Chromosomal Aneuploidies

Sample Information

Patient Name: [redacted] **Patient ID:** [redacted] **Lab No.:** [redacted]

Clinic Ref. No.: [redacted] **DOB:** [redacted] **Collection Date:** 15/09/2018

Referring Doctor: DR. TAI HOK LEUNG **Gestation:** 13w+5

Referring Unit: HK DNA DIAGNOSTICS CENTRE LTD

Results **Fetal cfDNA Percentage:** 12.20%

Condition	Probability	Risk Assessment
Trisomy 21	1/8614993772	Low risk
Trisomy 18	1/1028388259	Low risk
Trisomy 13	1/7675654312	Low risk

Condition	Risk Assessment
Trisomy 9	Low risk
Trisomy 16	Low risk
Trisomy 22	Low risk

Condition	Risk Assessment
X0	Low risk
XXY	Low risk
XXX	Low risk
XYY	Low risk

Condition	Result
84 Types of Chromosomal Micro-dup & del (please refer to the Appendix)	Not detected

Fetal Gender Determination (Sensitivity Rate >98%)	Female

Test Description

The NIFTY test works by isolating the cfDNA (including both maternal and fetal DNA) from a maternal blood sample and performing low coverage whole genome sequencing using Next Generation Sequencing technology. The unique reads of each chromosome are calculated and compared to an optimal reference control sample. Data is analyzed using BGI's proprietary bioinformatics algorithms and a risk score and/or assessment is produced for the conditions tested for. Results should always be reviewed with a qualified healthcare professional. It is advised that high-risk results are followed by confirmatory diagnostic testing.

Disclaimers

The NIFTY test is NOT a diagnostic test; the results are for informational use and therefore a false positive and false negative result cannot be excluded. Testing for other chromosomal aneuploidies (except T21, T18, T13) and chromosomal microdeletions & microduplications is only available for singleton pregnancy. 84 types of microdeletions & microduplications are detected in this test; the accuracy of microdeletions & microduplications that the abnormal size of which is over than 10M is validated; simulation experiment shows a detection rate of over 95% in microdelections & microduplications with abnormal size over 5M (cfDNA>15%) and around 90% when the abnormal size is smaller than 5M; some of the diseases on the list of microdeletion & microduplication syndrome can also be caused by other genetic reasons, NIFTY only detects and analyzes the specific fragment according to authorized database. Fetal sex provided in this report cannot be used to diagnose the sex-linked diseases. 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, stem cell therapy, heparin therapy and the abnormal karyotype of biological parents or surrogate. Test result is specific to the tested sample and should always be interpreted by a qualified professional in the context of clinical and familial data.

Approved Signatory: Sum Chi Kwan ***Dated:*** 26/09/2018

Sum Chi Kwan (MT101575)

Registered Med Lab Technologist

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