BGI HEALTH (HK) CO. LTD
3/F, NO.16 DAI FU STREET
TAI PO INDUSTRIAL ESTATE
N.T., HONG KONG
DX-PD-B

BGI Clinical Report

Non-invasive Prenatal Genetic Testing of Fetal Chromosomal Aneuploidies

Demo report

Sample Information

Patient Name: **Patient ID:** **Lab No.:**

Clinic Ref. No.: **DOB:** **Collection Date:**

Referring Doctor: **Gestation:**

Referring Unit:

Results **Fetal cfDNA Percentage: 9.16%**

Condition	Probability	Result
Trisomy 21	>1/20	High risk
Trisomy 18	1/9538264007	Low risk
Trisomy 13	1/1950395519	Low risk

Condition	Result
Trisomy 9	Low risk
Trisomy 16	Low risk
Trisomy 22	Low risk

Condition	Result
X0	High risk
XXY	Low risk
XXX	Low risk
XYY	Low risk

Condition	Result
84 Types of Chromosomal Micro-dup & del (please refer to the Appendix)	Di George Syndrome is detected; it is resulted from a small part of deletion on chromosome 22q11.21; please follow up counselling and view the result with physicians

Fetal Gender Determination (Sensitivity Rate >99%)	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 (cffDNA>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.

Signed by: _____ ***Dated:***

Wong Sai Wah BSc (Hons), MSc, MLT (HK) Registration No.: MT100861, BGI HEALTH (HK) CO. LTD

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Appendix - Condition list for microdeletions & microduplications

Chromosomal microdeletions and microduplications make up a fraction of copy-number variants (CNVs). CNVs are defined as either the gain (duplication) or loss (deletion) of a stretch of DNA on the chromosomes. Microdeletions and microduplications have been associated with abnormal developmental outcomes. Eighty-four Microdeletions and microduplications have been screened in this test.

	Condition	Region
1	Chromosome 1p31 duplication syndrome (染色體 1p31 重複綜合症)	1p31.3
2	Chromosome 1p32-p31 deletion syndrome (染色體 1p32-p31 缺失綜合症)	1p32-p31
3	Chromosome 1p36 deletion syndrome (染色體 1p36 缺失綜合症)	1p36
4	Chromosome 1q41-q42 deletion syndrome (染色體 1q41-q42 缺失綜合症)	1q41-q42
5	Chromosome 2p12-p11.2 deletion syndrome (染色體 2p12-p11.2 缺失綜合症)	2p12-p11.2
6	Chromosome 2p16.1-p15 deletion syndrome (染色體 2p16.1-p15 缺失綜合症)	2p16.1-p15
7	Split-hand/foot malformation 5 (裂手裂足症 5 型)	2q31
8	Chromosome 2q31.1 duplication syndrome (染色體 2q31.1 重複綜合症)	2q31.1
9	Chromosome 2q33.1 deletion syndrome (染色體 2q33.1 缺失綜合症)	2q33.1
10	Chromosome 2q35 duplication syndrome (染色體 2q35 重複綜合症)	2q34-q36
11	Holoprosencephaly 6 (前腦無裂畸形 6 型)	2q37.1-q37.3
12	Chromosome 3pter-p25 deletion Syndrome (染色體 3p-綜合症)	3pter-p25
13	Chromosome 3q13.31 deletion syndrome (染色體 3q13.31 缺失綜合症)	3q13.31
14	Dandy-Walker syndrome (染色體 3q22-q24 缺失綜合症)	3q22-q24
15	Chromosome 3q29 deletion syndrome (染色體 3q29 缺失綜合症)	3q29
16	Chromosome 3q29 duplication syndrome (染色體 3q29 重複綜合症)	3q29
17	Wolf-Hirschhorn syndrome (沃夫-賀許宏氏症候群)	4p16.3
18	Chromosome 4q21 deletion syndrome (染色體 4q21 缺失綜合症)	4q21
19	Chromosome 4q32.1-q32.2 triplication syndrome (染色體 4q32.1-q32.2 三倍重複綜合症)	4q32.1-q32.2
20	Cri du Chat syndrome (貓鳴綜合症)	5p
21	Chromosome 5q12 deletion syndrome (染色體 5q12 缺失綜合症)	5q12
22	Chromosome 5q14.3 deletion syndrome (染色體 5q14.3 缺失綜合症)	5q14.3-q15
23	Chromosome 6pter-p24 deletion syndrome (染色體 6pter-p24 缺失綜合症)	6pter-p24
24	Chromosome 6q11-q14 deletion syndrome (染色體 6q11-q14 缺失綜合症)	6q11-q14
25	Chromosome 6q24-q25 deletion syndrome (染色體 6q24-q25 缺失綜合症)	6q24-q25
26	CHDM (脊索瘤感受性)	6q27
27	Chromosome 7q deletion (染色體 7q 缺失綜合症)	7q
28	Chromosome 7q11.23 deletion syndrome (染色體 7q11.23 缺失綜合症)	7q11.23
29	Chromosome 7q11.23 duplication syndrome (染色體 7q11.23 重複綜合症)	7q11.23
30	Chromosome 8p23.1 deletion syndrome (染色體 8p23.1 缺失綜合症)	8p23.1
31	Chromosome 8p23.1 duplication syndrome (染色體 8p23.1 重複綜合症)	8p23.1
32	Chromosome 8q12.1-q21.2 deletion syndrome (染色體 8q12.1-q21.2 缺失綜合症)	8q12.2-q21.2
33	Chromosome 8q22.1 duplication syndrome (染色體 8q22.1 重複綜合症)	8q22.1
34	Chromosome 8q22.1 deletion syndrome (染色體 8q22.1 缺失綜合症)	8q22.1
35	Langer-Giedion syndrome (Langer-Giedion 綜合症)	8q24.11-q24.13
36	Chromosome 9p deletion syndrome (染色體 9p 缺失綜合症)	9p
37	DiGeorge syndrome 2 (狄喬治症候群第 2 型)	10p14-p13
38	Chromosome 10q22.3-q23.2 deletion syndrome (染色體 10q22.3-q23.2 缺失綜合症)	10q23
39	Chromosome 10q26 deletion syndrome (染色體 10q26 缺失綜合症)	10q26
40	Potocki-Shaffer syndrome (Potocki-Shaffer 綜合症)	11p11.2
41	WAGR syndrome (WAGR 綜合症)	11p13
42	WAGRO syndrome (WAGRO 綜合症)	11p13-p12

	Condition	Region
43	Jacobsen syndrome (雅各森症候群)	11q23
44	Chromosome 12q14 microdeletion syndrome (染色體 12q14 微缺失綜合症)	12q14
45	Chromosome 13q14 deletion syndrome (染色體 13q14 缺失綜合症)	13q14
46	Chromosome 14q11-q22 deletion syndrome (染色體 14q11-q22 缺失綜合症)	14q11-q22
47	Frias syndrome (Frias 綜合症)	14q22.1-q22.3
48	Chromosome 15q11-q13 duplication syndrome (染色體 15q11-q13 重複綜合症)	15q11
49	Angelman syndrome (普瑞德威利症)	15q11.2
50	Prader-Willi syndrome (小胖威利症)	15q11.2
51	Chromosome 15q14 deletion syndrome (染色體 15q14 缺失綜合症)	15q14
52	Chromosome 15q25 deletion syndrome (染色體 15q25 缺失綜合症)	15q25
53	HCD (先天性橫膈膜疝氣疾病)	15q26.1
54	Chromosome 15q26-qter deletion syndrome (染色體 15q26-qter 缺失綜合症)	15q26-qter
55	Levy-Shanske syndrome (Levy -Shanske 綜合症)	15q26-qter
56	Chromosome 16p deletion syndrome (染色體 16p 缺失綜合症)	16p
57	Chromosome 16p11.2-p12.2 microduplication syndrome (染色體 16p12.2-p11.2 重複綜合症)	16p12.2-p11.2
58	Chromosome 16p12.2-p11.2 deletion syndrome (染色體 16p12.2-p11.2 缺失綜合症)	16p12.2-p11.2
59	Chromosome 16p13.3 deletion syndrome (染色體 16p13.3 缺失綜合症)	16p13.3
60	Chromosome 16q22 deletion syndrome (染色體 16q22 缺失綜合症)	16q22
61	Potocki-Lupski syndrome (Potocki-Lupski 綜合症)	17p11.2
62	Smith-Magenis syndrome (史密斯-馬吉利氏症候群)	17p11.2
63	Yuan-Harel-Lupski syndrome (Yuan-Harel-Lupski 綜合症)	17p12-p11.2
64	Chromosome 17p13.3 duplication syndrome (染色體 17p13.3 重複綜合症)	17p13.3
65	Chromosome 17p13.3 deletion syndrome (染色體 17p13.3 缺失綜合症)	17p13.3
66	Chromosome 17q12 deletion syndrome (染色體 17q12 缺失綜合症)	17q12
67	Chromosome 17q12 duplication syndrome (染色體 17q12 重複綜合症)	17q12
68	Chromosome 17q21.31 duplication syndrome (染色體 17q21.31 重複綜合症)	17q21.31
69	Chromosome 17q23.1-q23.2 deletion syndrome (染色體 17q23.1-q23.2 缺失綜合症)	17q23.1-q23.2
70	Chromosome 18p deletion syndrome (染色體 18p 缺失綜合症)	18p
71	Chromosome 18q deletion syndrome (染色體 18q 缺失綜合症)	18q
72	Chromosome 19q13.11 deletion syndrome (染色體 19q13.11 缺失綜合症)	19q13.11
73	Holoprosencephaly 1 (前腦無裂畸形 1 型)	21q22.3
74	Cat-Eye syndrome (貓眼綜合症)	22q11
75	Chromosome 22q11.2 deletion syndrome (染色體 22q11.2 缺失綜合症)	22q11.2
76	Chromosome 22q11.2 duplication syndrome (染色體 22q11.2 重複綜合症)	22q11.2
77	DiGeorge syndrome (狄喬治症候群)	22q11.21
78	Chromosome Xp11.23-p11.22 duplication syndrome (染色體 Xp11.23-p11.22 重複綜合症)	Xp11.23-p11.22
79	Chromosome Xp11.3 deletion syndrome (染色體 Xp11.3 缺失綜合症)	Xp11.3
80	Chromosome Xp21 deletion syndrome (染色體 Xp21 缺失綜合症)	Xp21
81	Chromosome Xq21 deletion syndrome (染色體 Xq21 缺失綜合症)	Xq21
82	Chromosome Xq22.3 telomeric deletion syndrome (染色體 Xq22.3 端粒缺失綜合症)	Xq22.3
83	Chromosome Xq27.3-q28 duplication syndrome (染色體 Xq27.3-q28 重複綜合症)	Xq27.3-q28
84	Chromosome Xq28 deletion syndrome (染色體 Xq28 缺失綜合症)	Xq28