

PATIENT AND PROVIDER INFORMATION

• SINGLETON

Patient Name: \${patientName}	Hospital/Clinic: \${hospitalName}	Referring Clinician: \${doctorName}
Date of Birth: \${patientBirthDay}	Gestational Age: \${gestationalWeeks}	EDC: \${patientEdd}
Patient ID: \${oldSampleNum}	Sample Collection Date: \${collectDate}	Sample Received Date: \${receivedDate}

RESULTS

Fetal Fraction ($\geq 3.5\%$): \${fetalFraction}%

TRISOMY 21	TRISOMY 18	TRISOMY 13
\${testChr21}	\${testChr18}	\${testChr13}
Probability: \${testChr21P}	Probability: \${testChr18P}	Probability: \${testChr13P}

OTHER FINDINGS

TRISOMY 9	TRISOMY 16	TRISOMY 22	XO	XXX	XXY	XYY
\${testChr9}	\${testChr16}	\${testChr22}	\${XO}	\${XXX}	\${XXY}	\${XYY}
MICRODELETIONS / MICRODUPLICATIONS (92 TYPES)			INCIDENTAL FINDINGS			
\${disease}			\${otherCnv} \${testChrOther}			

ABOUT THE TEST: NIFTY® pro is a non-invasive prenatal screening test that 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 the assessment produced is for the conditions tested only. Only qualified healthcare professionals can order the test, and patients should always be accompanied by them to review the results for further evaluation and diagnosis.

DISCLAIMER: NIFTY® pro is a screening test, NOT a diagnostic test. Posttest counseling is recommended when the NIFTY report indicates that a patient is at high risk. The results are for informational use. The possibility of false positive/negative results cannot be ruled out. Corresponding results will be reported as supplementary information only when opted in for testing. The performance of Other Findings has not been fully validated, but the data in the table below can be used for reference. 92 types of micro-deletions/duplications syndromes are detected in this test, but some of the diseases can also be caused by other genetic factors. Incidental findings will report it if any of fetal microdeletions/duplications greater than 5 Mb and/or autosomal aneuploidies other than trisomy 21, 18, 13, 9, 16, 22 are detected. NIFTY® Pro only detects and analyses the specific fragment according to authorized databases. Potential sources of an inaccurate test result may include but are not limited to: maternal/fetal/placental mosaicism, low fetal fraction, blood transfusion, transplant surgery, stem cell therapy, heparin therapy, and/or abnormal karyotype of biological parents or surrogates. The 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. The test result cannot be used as the sole basis for diagnosis or other pregnancy management decisions.

Performance Only for Reference

CONDITION	SENSITIVITY	SPECIFICITY	REFERENCE
T21	99.17%	99.95%	Ultrasound Obstet Gynecol. 2015 May;45(5):530-8.
T18	98.24%	99.95%	
T13	100%	99.96%	
Fetal Sex	99.53%	99.20%	J Matern Fetal Neonatal Med. 2014 Dec;27(18):1829-33.
Del/Dup	>10Mb	88.89%	PLoS One.2016 Jul 14;11(7):e0159233.
	<10Mb	72.73%	

CONDITION	SENSITIVITY	SPECIFICITY	PPV	REFERENCE
XO	75%	99.9%	23.53%	BMC medical genomics vol. 5 57. 1 Dec. 2012 Chinese medical journal vol. 133,13 (2020): 1617-1619.
XXX	N/A	N/A	70%	
XXY	100%	100%	75%	
XYY	100%	100%	80%	

The data in the table is based on historical literature and internal data, and only reflects past detection, not the actual condition of the tested sample nor the promised value.

Approved by: Sara El Bisari, PhD, M.Sc. (GT18084)
Medical Laboratory Technologist Cytogeneticist
UK HCPC Registered Clinical Cytogeneticist
Genetics Laboratory Manager

Report Date: \${createTime}



Microdeletion and Microduplication Syndromes List (92 TYPES)

Chromosome 1p36 deletion syndrome	Chromosome 10q26 deletion syndrome
Chromosome 1q41-q42 deletion syndrome	Chromosome 10p12-p11 deletion syndrome
Chromosome 1p32-p31 deletion syndrome	Chromosome 10p duplication
Chromosome 2p16.1-p15 deletion syndrome	Chromosome 11p13 deletion syndrome
Chromosome 2q33.1 deletion syndrome	Chromosome 11p11.2 deletion syndrome
Chromosome 2q31.1 duplication syndrome	Jacobsen syndrome
Chromosome 2q37 deletion syndrome	Chromosome 11q23 deletion syndrome
Chromosome 2q31.1 microdeletion syndrome	Chromosome 12q14 microdeletion syndrome
Chromosome 2q duplication	Chromosome 12p12.1 microdeletion syndrome
Chromosome 3pter-p25 deletion syndrome	Chromosome 12p duplication
Dandy-Walker syndrome	Chromosome 13q14 deletion syndrome
Chromosome 3q13.31 deletion syndrome	Distal chromosome 13q deletion
Distal chromosome 3p duplication	Chromosome 14q11-q22 deletion syndrome
Chromosome 3q duplication	Chromosome 14q22 deletion syndrome
Chromosome 4p16.3 deletion syndrome	Proximal chromosome 14q deletion
Chromosome 4q21 deletion syndrome	Chromosome 14q duplication
Chromosome 4p duplication	Prader-Willi syndrome
Distal chromosome 4q duplication	Angelman syndrome
Distal chromosome 4q deletion	Chromosome 15q26-qter deletion syndrome
Cri-du-Chat syndrome	Levy-Shanske syndrome
Chromosome 5q14.3 deletion syndrome	Chromosome 15q14 deletion syndrome
Chromosome 5q12 deletion syndrome	Chromosome 15q24 microdeletion syndrome
Chromosome 5p13 duplication syndrome	Chromosome 15q26 overgrowth syndrome
Chromosome 5p duplication	Distal chromosome 15q deletion
Chromosome 6pter-p24 deletion syndrome	Chromosome 16p12.2-p11.2 deletion syndrome
Chromosome 6q24-q25 deletion syndrome	Chromosome 16p12.2-p11.2 duplication syndrome
Chromosome 6q11-q14 deletion syndrome	Chromosome 16p13.3 deletion syndrome
Chromosome 6p deletion	Chromosome 16p13.3 duplication syndrome
Chromosome 6q15-q23 deletion syndrome	Proximal chromosome 16q duplication
Chromosome 6q25-qter deletion syndrome	Smith-Magenis syndrome
Chromosome 6q26-q27 deletion syndrome	Chromosome 17p13.3 deletion syndrome
Chromosome 7q deletion	Potocki-Lupski syndrome
Chromosome 7q11.23 deletion syndrome	Chromosome 17p13.3 duplication syndrome
Chromosome 7q21-q32 deletion	Yuan-Harel-Lupski syndrome
Chromosome 7q31-q32 deletion	Chromosome 17p duplication
Chromosome 8p23.1 deletion syndrome	Chromosome 18p deletion syndrome
Chromosome 8p23.1 duplication syndrome	Distal chromosome 18q deletion syndrome
Langer-Giedion syndrome	Alagille syndrome 1
Chromosome 8q22.1 deletion syndrome	Chromosome 20p duplication
Chromosome 8q22.1 duplication syndrome	Chromosome 21q22 deletion
Chromosome 8p duplication	Chromosome 22q11.2 deletion syndrome
Chromosome 8q duplication	Chromosome Xp11.23-p11.22 duplication syndrome
Chromosome 9p deletion syndrome	Chromosome Xp21 deletion syndrome
Chromosome 9p duplication	Chromosome Xq27.3-q28 duplication syndrome
DiGeorge syndrome 2	Chromosome Xq21 deletion syndrome
Chromosome 10q22.3-q23.2 deletion syndrome	Chromosome Xq22.3 deletion syndrome