

## Patient and Provider Information

Clinic/Hospital Name		Referring Clinician	
Prom-Test LLC			
Gestational Age	EDC	Insurance ID (if relevant)	
Sample Type			
Blood			

NIFTY<sup>®</sup> is a screening test. Genetic counselling and diagnostic testing should be offered to further evaluate these findings.

## Results

Fetal Fraction: 25.38% (≥3.5%)

Test Name	Result	Reference Interval	Probability	Note
Trisomy 21	Low risk	Low risk	1/3116693023	Please review with physician
Trisomy 18	Low risk	Low risk	1/1095024765	Please review with physician
Trisomy 13	Low risk	Low risk	1/7829601763	Please review with physician

## Other Findings

Rare Autosomal Aneuploidies	Result	Note
Trisomy 9	Low risk	Please review with physician
Trisomy 16	Low risk	Please review with physician
Trisomy 22	Low risk	Please review with physician
Sex Chromosomal Aneuploidies	Result	Note
Monosomy X (XO)	Not detected	Please review with physician
XXY	Not detected	Please review with physician
XXX	Not detected	Please review with physician
XYY	Not detected	Please review with physician
Microdeletions/duplications (84 types)	Not detected	
Incidental Findings	Not detected	

**Test Description:** The NIFTY Pro<sup>™</sup> test is a screening test and is not diagnostic. It 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 an assessment is produced for the conditions tested only. Tests should always be ordered by a qualified healthcare professional and results reviewed with the patient. The test must not be used as the sole basis for diagnosis or other pregnancy management decision.

**Disclaimer:** The NIFTY Pro<sup>™</sup> test is NOT a diagnostic test, the results are for informational use and therefore a false positive and false negative results cannot be excluded. The performance of "Other Findings" has not been fully validated, but the data in the table below can be used for reference. 84 types of del/dup syndromes are detected in this test. Some of the diseases on the list of del/dup syndromes can also be caused by other genetic factors, NIFTY Pro<sup>™</sup> only detects and analyzes the specific fragment according to authorized databases. 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 results are specific to the tested sample and should always be interpreted by a qualified professional in the context of clinical and familial data.

Condition	Sensitivity	Specificity	PPV	Reference
T21	99.17%	99.95%	92.19%	UltrasoundObstet Gynecol. 2015 May;45(5):530-8. doi: 10.1002/uog.14792.
T18	98.24%	99.95%	76.61%	
T13	>99.9%	99.96%	32.84%	
CNV	>90%	N/A	N/A	From in-house data. Internal analysis shows a sensitivity of over 90% (cfDNA≥9.5%) in selected del/dup syndromes with abnormal size over 3 Mb.
Condition	Detection Rate	PPV	NPV	Reference
XYY	>99.9%	50.00%	>99.9%	BMC Med Genomics. 2012 Dec 1;5:57. doi: 10.1186/1755-8794-5-57. UltrasoundObstet Gynecol. 2014 Jul;44(1):17-24. doi: 10.1002/uog.13361.
XXY	>99.9%	42.86%	>99.9%	
XXX	>99.9%	70.00%	>99.9%	
XO	>99.9%	40.00%	>99.9%	

Note: 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: .....

Date: 05/12/2022

YUEN Ka Yiu MLT (HK) Registration Number: MT103521

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		Clinic/Hospital Name	
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		Sample Type	
		Blood	

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Y Chromosome	Detected
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**Test Description:** The NIFTY Pro™ test is a screening test and is not diagnostic. Gender identification works by isolating cell free DNA (including both maternal and fetal DNA) from a maternal blood sample, followed by molecular genetic testing to determine the relative quantities of the Y chromosome.

**Disclaimer:** The NIFTY Pro™ test is NOT a diagnostic test, the results are for informational use only. Although the methodology is highly accurate, the test does not provide a result with 100% accuracy. The Y chromosome detection provided in this report cannot be used for diagnosis of fetal sex or gender-related diseases, and is only used as additional information for reference analysis. 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.

Condition	Sensitivity	Specificity	PPV	Reference
Fetal Sex	99.53%	99.20%	N/A	J MaternFetal Neonatal Med. 2014 Dec;27(18):1829-33. doi: 10.3109/14767058.2014.885942.

Note: 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.

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