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NIFTY® BGI technology inside

DX-PD-B347 V1.0

Non-invasive Prenatal Genetic Testing for Fetal Chromosomal Aneuploidies for Singleton Pregnancies

Sample information

Barcode:

Patient ID:

Patient name:

Estimated date of confinement (d/m/y) :

Age: 35

Gestational week by ultrasound: 15

Referring Doctor:

Sample type: Blood

Sample received (d/m/y) :

Shipment condition: Room Temperature

Methodology

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 analysed 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. Further information regarding the conditions tested for and support groups can be found at www.niftytest.com.

Test result

Conditions	Probability	Risk Assessment
Trisomy 21	1/1950164323	Low risk
Trisomy 18	1/7633271000	Low risk
Trisomy 13	1/2094121674	Low risk

It is advised that high-risk results should be followed by confirmatory diagnostic testing.

Fetal cfDNA Percentage	13.75%

Reference:

1. Dan S, et al.Clinical application of massively parallel sequencing-based prenatal noninvasive fetal trisomy test for trisomies 21 and 18 in 11,105 pregnancies with mixed risk factors. Prenat Diagn. 2012 Dec;32(13):1225-32.
2. Zhang H, et al. Non-invasive prenatal testing for trisomies 21, 18 and 13: clinical experience from 146,958 pregnancies. Ultrasound Obstet Gynecol. 2015 Jan 19. doi: 10.1002/uog.14792.

Condition	Test Sensitivity
Trisomies (21, 18, 13)	99.12%

Disclaimers

1) The NIFTY test is NOT a diagnostic test and therefore false positive and false negative results can occur.
2) 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 and the abnormal karyotype of biological parents or surrogate.
3) This test assumes that the blood and DNA samples belong to the specified patient as it is claimed, the result is therefore specific to the tested sample.
4) Test results should always be interpreted by a qualified healthcare professional in the context of other clinical and/or family information of the patient. Results should be communciated in a setting that includes appropriate genetic counseling.
5) The result of the test does not eliminate the possibility of other abnormalities of the tested chromosomes and/or other genetic disorders or birth defects.
6) Fetal cfDNA percentage is only available for singleton pregnancy.

Approved by: ***Dated:***

Wong Sai Wah BSc (Hons), MSc MLT (HK) Registration Number: MT100861

Non-invasive Prenatal Genetic Testing: Opt-In Testing for Chromosome 9, 16, 22 Aneuploidies, Sex Chromosome Aneuploidies, Deletion/Duplication Syndromes, Incidental Findings

Sample information

Barcode:
Patient ID:
Patient name:
Estimated date of confinement (d/m/y) :
Age: 35
Gestational week by ultrasound: 15
Referring Doctor:
Sample type: Blood
Sample received (d/m/y) :
Shipment condition: Room Temperature

Methodology

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 analysed 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. Further information regarding the conditions tested for and support groups can be found at www.niftytest.com. Besides the aneuploidies mentioned above, There may be some other incidential findings includes but not limited microdeletion, duplication, and other chromosomal aneupliody.

Test result

Autosome Trisomies	Risk Assessment
Trisomy 9	Low risk
Trisomy 16	Low risk
Trisomy 22	Low risk

Sex Chromosome Aneuplodies	Result	Interpretation
XO	Not detected	None
XXY	Not detected	None
XXX	Not detected	None
XYY	Not detected	None

Deletion/Duplication Syndromes

Not detected

Incidental Findings

Aneuploidy of other chromosomes: Not detected

Chromosomal deletions/duplications: del(3p26.3-p26.1,8.45M)

Conditions	Test sensitivity
Sex Chromomsomal Aneuploidies	95%
Deletion/Duplication Syndrome	Not validated
Autosome Trisomies (9, 16, 22)	Not validated
Incidental Findings	Not validated

Reference:

1. Chen S, Lau TK, Zhang C, Xu C, et al. A method for noninvasive detection of fetal large deletions/duplications by low coverage massively parallel sequencing.Prenat Diagn. 2013 Jun;33(6):584-90.
2. Pan X1, Zhang C, Li X, Chen S,et al. Noninvasive fetal sex determination by maternal plasma sequencing and application in X-linked disorder counseling.J Matern Fetal Neonatal Med. 2014 Dec;27(18):1829-33.

Disclaimers

1) Testing for sex chromosomal aneuploidies, deletion syndromes and gender information is only available for singleton pregnancy.
2) The NIFTY test is NOT a diagnostic test, information on this opt-in report could only be used as reference. BGI has no clinical responsibilities on false positive or false negative results occurred on those syndromes above.
3) 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 and the abnormal karyotype of biological parents or surrogate.
4) This test assumes that the blood and DNA samples belong to the specified patient as it is claimed, the result is therefore specific to the tested sample.
5) Results should always be interpreted by a qualified healthcare professional in the context of clinical and familial data.
6) Results are for informational use.

Dated:

Prenatal Genetic Testing: Opt-In Testing for Gender Information

Sample information

Barcode:
Patient ID:
Patient name:
Estimated date of confinement (d/m/y) :
Age: 35
Gestational week by ultrasound: 15
Referring Doctor:
Sample type: Blood
Sample received (d/m/y) :
Shipment condition: Room Temperature

Methodology

The test works by isolating cell free DNA (including both maternal and fetal DNA) from a maternal blood sample, followed with molecular genetic testing.
Results should always be reviewed with a qualified healthcare professional.

Test result:

Gender	Male

Test sensitivity	98%

Disclaimers

1) Testing for sex chromosomal aneuploidies and gender information is only available for singleton pregnancy. Results for maternal sex chromosome abnormalities will only be reported if a patient as opted-in for reporting of sex chromosomal aneuploidies and/or reporting of incidental findings on the test request form and declaration of consent form.
2) The test is NOT a diagnostic test, information on this opt-in report could only be used as reference. BGI has no clinical responsibilities on false positive or false negative results occurred on the gender above.
3) 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 and the abnormal karyotype of biological parents or surrogate.
4) This test assumes that the blood and DNA samples belong to the specified patient as it is claimed, the result is therefore specific to the tested sample.
5) Results should always be interpreted by a qualified healthcare professional in the context of clinical and familial data.
6) Results are for informational use.

Approved by: _____ ***Dated:*** _____

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