

Non-invasive Prenatal Genetic Testing for Fetal Chromosomal Aneuploidies for Twins Pregnancy

Sample information

Barcode: _____ Patient ID: _____ Patient name : _____
 Estimated date of confinement (d/m/y) : _____ Age: 27 _____ Gestational week by ultrasound: _____
 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.

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

Test Result

Conditions	Risk Assessment
Trisomy21	Low risk
Trisomy18	Low risk
Trisomy13	Low risk

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

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

Reference:

- 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.
- Lau TK, et al. Non-invasive prenatal screening of fetal Down syndrome by maternal plasma DNA sequencing in twin pregnancies. *J Matern Fetal Neonatal Med.* 2013 Mar;26(4):434-7.
- 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.

Disclaimers

- The NIFTY test is NOT a diagnostic test and therefore false positive and false negative results can occur.
- 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.
- Result accuracy for twin pregnancy testing may be affected by 'vanishing twin' syndrome.
- 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.
- Results should always be interpreted by a qualified healthcare professional in the context of clinical and familial data.
- Results are for informational use.
- If the result is "Low Risk", 99% possibilities of both fetuses are at low risk for T21, T18, and T13. If the result is "High Risk", it suggests that at least one of the twin fetuses is at high risk, further diagnostic test for both fetuses are needed. A "high risk" result should not be considered as diagnostic. The finding should be confirmed by conventional tests such as fetal karyotyping.

Approved by: _____

Dated: _____

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

Prenatal Genetic Testing: Opt-In Testing for Y Chromosome Detection Information for Twins Pregnancy

Sample information

Barcode: _____ Patient ID: _____ Patient name: _____
Estimated date of confinement (d/m/y) : _____ Age: 27 Gestational week by ultrasound: _____
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 by molecular genetic testing.
Results should always be reviewed with a qualified healthcare professor.

Test Result

Y Chromosome

NOT Detected

Disclaimers

- 1) The result "Detected" returns as one male fetus at least of the twin pregnancy. The result "NOT Detected" returns as both female fetuses of the twin pregnancy.
- 2) In twin gestational pregnancies, a result that returns as male does not exclude the possibility that other fetus might be female.
- 3) 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 above result of whether Y chromosome detection or not detected .
- 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) 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.
- 6) Results should always be interpreted by a qualified healthcare professional in the context of clinical and familial data.
- 7) Results are for informational use.

Approved by: _____

Dated: _____

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