NIFTY pro™
NON-INVASIVE FETAL TRISOMY TEST

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Sample Information

Patient Name	National ID (for insurance)	NIFTY Barcode	Date of Birth
Clinic Name	**Patient ID (in clinic/hospital)**	**Gestational age**	**EDC**
Zentya a.s		18w+6	
Referring Clinician	**Collection date**	**Sample type**	**Received date**
	28/08/2018	Blood	30/08/2018

Test Result

Fetal Sex: Female **Fetal cfDNA(%):** 12.00% **QC: Passed**

Condition	Result	Probability	Interpretation
Trisomy 21	High risk	>1/20	High possibility for Trisomy 21, please follow up counselling and recommendation with your physician
Trisomy 18	Low risk	1/5830550275	Please review with physician
Trisomy 13	Low risk	1/4170380029	Please review with physician

Condition	Result	Interpretation
Trisomy 9	Low risk	Please review with physician
Trisomy 16	Low risk	Please review with physician
Trisomy 22	Low risk	Please review with physician
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

Other findings include microdeletion/duplication syndrome and incidental finding

Microdeletion/duplication syndrome:
Not detected

Aneuploidy of other chromosomes: Not detected

Chromosomal deletions/duplications: None

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. For gender identification, it 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. 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 microdeletion/duplications is only available for singleton pregnancy. 84 types of del/dup syndromes are detected in this test; the accuracy of del/dup syndrome that the abnormal size of which is over than 10M is validated; simulation experiment shows a detection rate can be over 95% in del/dup syndromes with abnormal size over 5M and around 90% when the abnormal size is smaller than 5M; some of the diseases on the list of microdeletion/duplication 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.

Reference

1. 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.
2. 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.
3. Pan X, Zhang C, Li X, Chen S,et al. Noninvasive fetal sex determination by maternal plasma sequencing and application in X-linked disorder counselingJ. Matern Fetal Neonatal Med. 2014 Dec;27(18):1829-33. doi: 10.3109/14767058.2014.885942. Epub 2014 Feb 20.
4. Jiang et al. Noninvasive Fetal Trisomy (NIFTY) test: an advanced noninvasive prenatal diagnosis methodology for fetal autosomal and sex chromosomal aneuploidies. BMC Medical Genomics. 2012 5:57.
5. Yao H, et al. Detection of fetal sex chromosome aneuploidy by massively parallel sequencing of maternal plasma DNA: initial experience in a Chinese hospital. Ultrasound Obstet Gynecol. 2014 Jul;44(1):17-24. doi:10.1002/uog.13361.

Clinical Data

	Sensitivity	Specificity	PPV	NPV
Trisomy 21	99.17%	99.95%	97.58%	99.99%
Trisomy 18	98.24%	99.95%	97.67%	100%
Trisomy 13	100%	99.96%	83.33%	100%
Fetal Sex	99.53%	99.20%	NA	NA

	Detection Rate	PPV	NPV
XYY	100%	50.00%	100%
XXY	100%	42.86%	100%
XXX	100%	70.00%	100%
XO	100%	40.00%	100%
Microdel/dup	100%	NA	NA

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Dated: 04/09/2018