InsighT Report for Fetal Chromosomal Aneuploidies in Singleton Pregnancies

Patient Information	
Name: Mrs. AASHNA SABHARWAL	**Patient ID: 1002429198**
Date of Birth: 23/04/1990	**Sample ID: 2400107790**
Gestation age by Ultrasound: 13 Weeks + 1 days	**Hospital ID: NA**
Referring Doctor: Dr. Feroze Soonawala	**Sample collected on: 05/06/2024**
Hospital Name: Soonawala Clinic	**Sample received on: 06/06/2024**
Sample Type: Blood	**Report released on: 12/06/2024**
Referral Reason: Routine aneuploidy screening.	

Methodology

The InsighT test is a Non-invasive Prenatal Screening test. It works by isolating the cfDNA (including both maternal and fetal DNA) from a maternal peripheral blood sample and performing an extensive analysis using Next-Generation Sequencing technology. This robust data is further analyzed using a proprietary bioinformatics algorithms (software). A final risk assessment is produced for the conditions tested only, as recommended by the latest scientific guidelines for NIPS testing i.e. American College of Medical Genetics and Genomics (ACMG) Guidelines. The InsighT test provides risk assessment for common aneuploidies (T21, T18, and T13), Sex chromosome aneuploidies. The validation studies have been carried out for all the conditions reported by InsighT NIPS test. With >6 Million Reads/Sample the test is able to deliver an unmatched accuracy to ensure informed decision by clinician and couple. Results of the test should always be reviewed and communicated by a qualified healthcare professional only along with appropriate Genetic Counseling.

Test Results

COMMON ANEUPLOIDIES	RISK ASSESSMENT
Trisomy 21	Low Risk
Trisomy 18	Low Risk
Trisomy 13	Low Risk

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

SEX CHROMOSOME ANEUPLOIDIES	RISK ASSESSMENT
XO	Low Risk
XXY	Low Risk
XYY	Low Risk
XXX	Low Risk

Sex of the Fetus cannot be revealed as per PCPNDT Act 2003.

Fetal cfDNA Percentage	22.62%

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Performance validation of the test

Common Aneuplodies					
Conditions	**Sensitivity**	**Specificity**	**PPV**	**NPV**	**Reference**
Trisomy 21	99.17%	99.95%	92.19%	99.99%	UltrasoundObstet Gynecol. 2015 May;45(5):530-8.
Trisomy 18	98.24%	99.95%	76.61%	100%	
Trisomy 13	100%	99.96%	32.84%	100%	
Total	**99.02%**	**99.86%**	**85.27%**	**99.99%**	

Sex Chromosome Aneuploidies				
Conditions	**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%	

References:

1. Rose NC, Kaimal AJ, Dugoff L, Norton ME, American College of Obstetricians and Gynecologists. Screening for fetal chromosomal abnormalities: ACOG practice bulletin, number 226. Obstetrics & Gynecology. 2020 Oct 1;136(4):e48-69.
2. Zhang H, Gao Y, Jiang F, Fu M, Yuan Y, Guo Y, Zhu Z, Lin M, Liu Q, Tian Z, Zhang H, Chen F, Lau TK, Zhao L, Yi X, Yin Y, Wang W. Non-invasive prenatal testing for trisomies 21, 18 and 13: clinical experience from 146,958 pregnancies. Ultrasound Obstet Gynecol. 2015 May;45(5):530-8.
3. Song JP, Jiang YF, Gao TX, Yao YY, Liu LJ, Xu RH, Yi MQ, Yu CJ, Wang WP, Li H. Performance of non-invasive prenatal screening for sex chromosome aneuploidies and parental decision-making. Chin Med J (Engl). 2020 Jul 5;133(13):1617-1619.
4. Dungan JS, Klugman S, Darilek S, Malinowski J, Akkari YM, Monaghan KG, Erwin A, Best RG, ACMG Board of Directors. Noninvasive prenatal screening (NIPS) for fetal chromosome abnormalities in a general-risk population: An evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine. 2023 Feb 1;25(2):100336.
5. Cheung SW, Patel A, Leung TY. Accurate description of DNA-based noninvasive prenatal screening. N Engl J Med. 2015 Apr 23;372(17):1675-7. doi:10.1056/NEJMc1412222. Epub 2015 Apr 1. PMID: 25830325.

Disclaimers:

1. The InsighT test is NOT a diagnostic test. It is a screening test, therefore false-positive and false-negative results can occur.
2. Sex of the fetus cannot be revealed as per PC-PNDT act 2003.
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 and stem cell therapy.
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. This test is not intended to identify pregnancies at risk for open neural tube defects.
6. Test results should always be interpreted by a qualified healthcare professional in the context of other clinical and/or family information of the patient.
7. The results should be communicated in a setting that includes appropriate genetic counseling.

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8. The results of the test do not eliminate the possibility of other abnormalities of the tested chromosomes and/or other genetic disorders or birth defects.
9. This test has been performed at our partner lab.

Pallavi Kadam

Verified By

Scientific Officer

Genomics

Pusalkar

Dr. Madhavi Pusalkar, Ph.D.

General Manager

Genomics

----------***End of The Report***----------

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