InsighT-Adv Report for Fetal Chromosomal Aneuploidies in **Singleton Pregnancies**

Patient Information	
Name: Mrs.Pranjali Pandey	**Patient ID: 10022134330**
Date of Birth: 15/07/1987	**Sample ID: 2200140982**
Gestation age by Ultrasound: 16 Weeks + 4 Days	**Sample collected on: 07/02/2023**
Referring Doctor: Dr.Meenakshi Tiwari	**Sample received on: 09/02/2023**
Sample Type: Blood	**Report released on: 22/02/2023**

Methodology

InsighT-Adv: Advanced Non-invasive prenatal screening (NIPS) test uses an innovative, patented technology called 'Target Capture Enrichment Technology' (TACS) developed by NIPD Genetics to isolate & analyses the cell-free DNA from maternal plasma. TACS technology is specifically designed to overcome problems associated with other NIPTs and to increase the precision and accuracy through InsighT-Adv test.

Multiplexed parallel analysis of specific regions of interest is applied for the copy number determination of chromosomes 13, 18, 21, aneuploidies of X, Y, select microdeletions including, DiGeorge (22q11 deletion), 1p36 deletion syndrome, Smith-Magenis (17p11.2 deletion), and Wolf Hirschhorn (4p16.3 deletion). Proprietary bioinformatics pipelines are used to analyze the sequencing data produced through high read depth. This increases the sensitivity and specificity of the test for detection of conditions being screened under it. Validation studies have been/ are carried out for the conditions reported by InsighT-Adv NIPS test.

Results of the test should always be reviewed and communicated by a qualified healthcare professional only along with appropriate genetic counselling.

Test results

CONDITIONS	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
XXX (Trisomy X)	Low Risk
XO (Turner syndrome)	Low Risk
XXY (Klinefelter syndrome)	Low Risk
XYY (Jacob's syndrome)	Low Risk
XXYY (XXYY syndrome)	Low Risk

Sex of the fetus cannot be revealed as per PC-PNDT Act 2003.

MICRODELETIONS	RISK ASSESSMENT
DiGeorge (22q11) syndrome	Low Risk
1p36 deletion syndrome	Low Risk
Smith-Magenis (17p11.2) syndrome	Low Risk
Wolf Hirschhorn (4p16.3) syndrome	Low Risk

Fetal cfDNA Percentage	**5.0%**

Fetal fraction is found to be sufficient for analysis.

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Clinical performance of the test

CONDITIONS	TEST SENSITIVITY	TEST SPECIFICITY	PPV	NPV
Trisomy 21	~99.99%	~99.98%	~100%	~100%
Trisomy 18	~99.99%	~99.98%	~100%	~100%
Trisomy 13	~99.99%	~99.98%	~71%	~100%
XO (Turner syndrome)	~99.99%	~99.95%	~57%	~100%
XXX (Trisomy X)	-	~99.95%	-	~100%
XXY (Klinefelter syndrome)	-	~99.95%	-	~100%
XYY (Jacob's syndrome)	-	~99.95%	-	~100%
XXYY (XXYY syndrome)	-	~99.95%	-	~100%
Microdeletions (DiGeorge (22q11.2 deletion)syndrome, 1p36 deletion syndrome, Smith-Magenis (17p11.2 deletion) syndrome, Wolf-Hirschhorn (4p16.3 deletion) syndrome)	~99.99%	-	~100%	~100%

Reference:

1. Koumbaris G, Kypri E, Tsangaras K, Achilleos A, Mina P, Neofytou M, Velissariou V, Christopoulou G, Kallikas I, González-Liñán A, Benusiene E. Cell-free DNA analysis of targeted genomic regions in maternal plasma for non-invasive prenatal testing of trisomy 21, trisomy 18, trisomy 13, and fetal sex. Clinical chemistry. 2016 Jun 1;62(6):848-55.
2. Kypri E, Ioannides M, Touvana E, Neophytou I, Mina P, Velissariou V, Vittas S, Santana A, Alexidis F, Tsangaras K, Achilleos A. Non-invasive prenatal testing of fetal chromosomal aneuploidies: validation and clinical performance of the veracity test. Molecular cytogenetics. 2019 Dec 1;12(1):34.
3. Neofytou MC, Tsangaras K, Kypri E, Loizides C, Ioannides M, Achilleos A, Mina P, Keravnou A, Sismani C, Koumbaris G,Patsalis PC. Targeted capture enrichment assay for non-invasive prenatal testing of large and small size sub-chromosomal deletions and duplications. PLoS One. 2017 Feb 3;12(2):e0171319.
4. For More information: Visit - https://www.nipd.com/products/prenatal/veracity-doctors/

Disclaimer:

1. The InsighT-ADV test is not diagnostic but a screening test and results should be considered in the context of other clinical criteria. Clinical correlation with ultrasound findings, and other clinical data and tests is recommended. If definitive diagnosis is desired, amniocentesis is necessary.
2. The referral clinician is responsible for counselling before and after the test including the provision of advice regarding the need for additional invasive genetic testing.
3. Although this test is highly accurate, there is still a small possibility for false positive or false negative results. This may be caused by technical and/or biological limitations, including but not limited to confined placental mosaicism (CPM) or other types of mosaicism, maternal constitutional or somatic chromosomal abnormalities, residual cfDNA from a vanished twin or other rare molecular events.
4. This test has been validated on full region deletions and maybe unable to detect deletion of smaller regions. The test will not identify all deletions associated with each microdeletion syndrome.
5. Test performance is valid only for full chromosomal aneuploidies for chromosomes 21, 18, and 13 and upon request aneuploidies of X, Y. It does not exclude other chromosomal abnormalities, birth defects or other complications.
6. Sex chromosome aneuploidies are not reportable for twin and vanished twin gestations. Patients with

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malignancy or a history of malignancy, patients with bone marrow or organ transplant, as well as twin and vanished twin pregnancies conceived through in-vitro fertilization (IVF) with egg donation or use of a surrogate mother are not eligible for the test.

7. Validation studies are carried out for all conditions by NIPD Genetics Public Company Limited. The test is not intended and not validated for mosaicism, triploidy, partial trisomy or translocations.
8. 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.
9. 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 communicated in a setting that includes appropriate genetic counselling.
10. 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.
11. A positive result for twin pregnancies indicates high risk for the presence of at least one affected fetus.
12. In case of twin pregnancies, only the lowest fetal fraction is reported.
13. This test has been performed at NIPD Genetics.

Pallavi Kadam

Verified By
Scientific Officer
NGS Department

Pusalkar

Dr. Madhavi Pusalkar, Ph.D.
General Manager: Genomics

********** **End of the Report**********

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