

**Insight Report for Fetal Chromosomal Aneuploidies in Singleton Pregnancies**

Patient Information	
<b>Name: Mrs.Kalaivani .</b>	<b>Patient ID: 10022134474</b>
<b>Date of Birth: 24/07/1994</b>	<b>Sample ID: 2310007171</b>
<b>Gestation age by Ultrasound: 16 Weeks + 3 Days</b>	<b>Sample collected on: 08/02/2023</b>
<b>Referring Doctor: Dr.Kavitha</b>	<b>Sample received on: 09/02/2023</b>
<b>Sample Type: Blood</b>	<b>Report released on: 22/02/2023</b>

**Methodology**

**Insight: Non-invasive prenatal screening (NIPS) test uses an innovative, patented technology called 'Target Capture Enrichment Technology' (TACS) developed by NIPD Genetics to isolate & analyze 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 InsightT 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. 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 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.

<b>Fetal cfDNA Percentage</b>	<b>6.6%</b>
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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%

#### 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. For More information: Visit - <https://www.nipd.com/products/prenatal/veracity-doctors/>

#### Disclaimer:

1. The InsightT 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. 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.
5. Sex chromosome aneuploidies are not reportable for twin and vanished twin gestations. Patients with 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.
6. 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.
7. 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.

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8. 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.
9. 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.
10. A positive result for twin pregnancies indicates high risk for the presence of at least one affected fetus.
11. In case of twin pregnancies, only the lowest fetal fraction is reported.
12. 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 \*\*\*\*\*

