



InsighT (NIPS) Report for Fetal Chromosomal Aneuploidies in Singleton Pregnancies

| Patient Information | | |
|---|---------------------------------|--|
| Name: Mrs. DEEPA SONKAR | Patient ID: 160245277 | |
| Date of Birth: 20/09/1997 | Sample ID: 2400090761 | |
| Gestation age by Ultrasound: 16 Weeks + 6 days | Hospital ID: P030823LKO0008718 | |
| Referring Doctor: Dr. Pawan Yadav | Sample collected on: 03/06/2024 | |
| Hospital Name: Indira Path Lab-Lucknow | Sample received on: 06/06/2024 | |
| Sample Type: Blood | Report released on: 12/06/2024 | |
| Referral Reason: USG at 16 weeks 6 days shows echogenic intracardiac focus in left ventricle. | | |

Methodology

The InsighT (MPSS) test works by isolating the cf-DNA (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 the laboratory's proprietary bioinformatics algorithms and a risk score and/or assessment is produced for the conditions that are tested for. This test confers an accuracy and detection rate (sensitivity) of up to 99%. Results should always be reviewed by a qualified healthcare professional.

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 |
|-----------------------------|-----------------|
| хо | Low Risk |
| XXY/XYY | Low Risk |
| XXX | Low Risk |

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

| Fetal cfDNA Percentage | 12.95% |
|------------------------|--------|
|------------------------|--------|





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Reference:

- 1. Bianchi DW, Platt LD, Goldberg JD, Abuhamad AZ, Sehnert AJ, Rava RP. Genome-wide fetal aneuploidy detection by maternal plasma DNA sequencing. Obstetrics & Gynecology. 2012 May 1;119(5):890-901.
- 2. Chiu RW, Akolekar R, Zheng YW, Leung TY, Sun H, Chan KA, Lun FM, Go AT, Lau ET, To WW, Leung WC. Non-invasive prenatal assessment of trisomy 21 by multiplexed maternal plasma DNA sequencing: large scale validity study. Bmj. 2011 Jan 11;342.
- 3. Kappou D, Papadopoulou E, Sifakis S. Non Invasive Prenatal Diagnosis of Down Syndrome. InPrenatal Diagnosis and Screening for Down Syndrome 2011 Aug 17. IntechOpen.
- 4. 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.

Disclaimer:

- 1. The InsighT (MPSS) test is NOT a diagnostic test. It is a screening test, therefore false-positive and false-negative results can occur.
- 2. 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.
- 3. 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.
- 4. The fetal fraction (%) has been estimated in an algorithm based on global model fitting.
- 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.
- 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.

Pallari Kadam

Verified By

Scientific Officer

Genomics

Dr. Madhavi Pusalkar, Ph.D.

General Manager

Genomics

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