

To: Add-On Scans and Labs-Bangalore

No 5, MGR Complex,
Marathahalli -Sarjapur Rd,Above Pizza Hut,
Kaikondrahalli
Bengaluru - 560035
Contact:9900811118

Report Of: Mrs Pallavi Verma

Pt. Contact: 8368258902



Sample ID	2510014538
Patient ID	P25179966/223495
Collected on	29/07/2025 16:04
Received on	30/07/2025 10:37
Registered on	30/07/2025 12:56
Reported on	05/08/2025 13:29
Referred by	Dr.Kumari Deepmala

InsighT-Plus (NIPS) Report (Singleton Pregnancy)

Patient Name: Mrs Pallavi Verma

DOB: 29/10/1995

GA by Ultrasound: 13 weeks + 3 days

Specimen Type: Peripheral Maternal Blood

Referral Reason: Double marker test shows high risk for Trisomy 21 (1:180) and USG at 12 weeks 3 days shows unossified nasal bone.

Methodology

The InsighT-Plus 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-Plus test provides risk assessment for common aneuploidies (T21, T18, and T13), Sex chromosome aneuploidies, and 60 Microdeletion/duplication syndromes. The validation studies have been carried out for all the conditions reported by InsighT-Plus 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 Counselling.

Test Results

Conditions	Risk Assessment
Trisomy 21	Low Risk
Trisomy 18	Low Risk
Trisomy 13	Low Risk

Sex Chromosome Aneuploidies	Risk Assessment
XO (Turner syndrome)	Low Risk
XXY (Klinefelter syndrome)	Low Risk
XXX (Jacob's syndrome)	Low Risk
XXX (Trisomy X)	Low Risk

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

Additional Conditions	Risk Assessment
Microdeletions/Duplications (60 Types)	Low Risk

Fetal cf-DNA Percentage	6.80%
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Fetal fraction is found to be sufficient for analysis.



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Recommendations

1. Genetic Counselling is recommended to understand the test results.

Performance Validation Of The Test

Common Aneuploidies

Conditions	Sensitivity	Specificity	PPV	NPV	Study 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 (Singleton Pregnancy)

Conditions	Sensitivity	Specificity	PPV	Study 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%	
XXY	100%	100%	80%	

Additional Conditions (Singleton Pregnancy)

Conditions		Sensitivity	Specificity	Study Reference
Microdeletions/Duplications (60 Types)	>10Mb	88.89%	99.32%	PLoS One.2016 Jul 14;11(7): e0159233
	<10Mb	72.73%	99.09%	
Total		84.21%	98.42%	



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60 Microdeletion/duplication Syndromes Screened In InsighT-Plus

Disorder	Chromosome	Location
Chromosome 1p36 microdeletion Syndrome	1	1p36
Van der Woude Syndrome I (VWS)	1	1q32.2-q41
Chromosome 1q41 -q42 deletion syndrome	1	1q41-q41
Feingold Syndrome I	2	2p24.3
Split-Hand/Foot Malformation Type 5 (SHFM5)	2	2q31
Glass Syndrome	2	2q33.1
Holoprosencephaly Type 6 (HPE6)	2	2q37.1-q37.3
Dandy-Walker Syndrome(DWS)	3	3q22-q24
Rieger Syndrome Type 1 (RIEG1)	4	4q25
Cornelia de Lange Syndrome I (CDLS)	5	5p13.2
Cri du Chat(5p deletion) Syndrome	5	5p15.2 to the entire short arm
Chromosome 5q21.1-q31.2 deletion Syndrome	5	5q21.1-q31.2
Prader-Willi-like Syndrome	6	6q16.3
Saethre-Chotzen Syndrome(SCS)	7	7p21.1
Chromosome 8p23.1 deletion Syndrome	8	8p23.1
Trichorhinophalangeal Syndrome Type 1(TRPS1)	8	8p23.3
Branchiootorenal dysplasia Syndrome I(BOR)/Melnick-Frazer Syndrome	8	8q13.3
Langer-Giedion Syndrome(LGS)	8	8q23.3-q24.11
Distal Arthrogryposis type 2B (DA2B)	8	9p13.3;11p15.5; 17p13.1
Chromosome 8p23.1 duplication Syndrome	8	8p23.1
Monosomy 9p Syndrome	9	9p22.3-p23
DiGeorge type 2 Syndrome (DGS2)	10	10p14-p13
Chromosome 10q22.3-q23.31 microdeletion Syndrome	10	10q22.3-q23.31
Bannayan-Riley-Ruvalcaba Syndrome(BRRS)	10	10q23.31
Cowden Syndrome(CD)	10	10q23.31
Chromosome 10q deletion Syndrome	10	10q26
Split-hand/foot malformation type 3 (SHFM3)	10	10q24
Aniridia II & WAGR Syndrome	11	11p13
Wilms tumor 1 (WT1)	11	11p13
Leukodystrophy with 11q14.2-q14.3	11	11q14.2-q14.3

Disorder	Chromosome	Location
Jacobsen Syndrome	11	11q23
Chromosome 11q11-q13.3 duplication Syndrome	11	11q13.3
Chromosome 12q14 microdeletion Syndrome	12	12q14
Chromosome 14q11-q22 deletion syndrome	14	14q11-q22
Microphthalmia Syndrome Type 6, pituitary hypoplasia	14	14q22.2-q22.3
Angelman Syndrome/PraderWilli Syndrome	15	15q11-q13
Deafness-infertility Syndrome	15	15q15.3
Chromosome 15q26 overgrowth Syndrome	15	15q26
Diaphragmatic hernia, congenital (HCD/DIH1)	15	15q26.1
Chromosome 16p11.2-p12.2 microdeletion Syndrome	16	16p11.2-p12.2
Alpha Thalassemia, Mental Retardation Syndrome	16	16p13.3
Chromosome 16p11.2-p12.2 microduplication Syndrome	16	16p11.2-p12.2
Smith-Magenis Syndrome	17	17p11.2
Chromosome 17q21.31 deletion Syndrome	17	17q21.31
Potocki-Lupski Syndrome (17p11.2 duplication Syndrome)	17	17p11.2
Chromosome 17q21.31 duplication Syndrome	17	17q21.31
Chromosome 18p deletion Syndrome	18	18p
Holoprosencephaly Type 4(HPE4)	18	18p11.31
Chromosome 18q deletion Syndrome	18	18q
Dyggve-Melchior-Clausen Syndrome(DMC)	18	18q21.1
Holoprosencephaly Type 1(HPE1)	21	21q22.3
Cat-eye Syndrome(CES)	22	22q11.21
Duchenne muscular dystrophy (DMD); Duchenne/Becker muscular dystrophy (DMD/BMD)	X	Xp21.2-p21.1
Microphthalmia with linear skin defects	X	Xp22.2
Orofaciodigital Syndrome I	X	Xp22.2
Androgen Insensitivity Syndrome(AIS)	X	Xq12
X-linked lymphoproliferative Syndrome(XLP)	X	Xq25
Chromosome Xp11.22-p11.23 microduplication Syndrome	X	Xp11.22-p11.23
Mental retardation X-linked growth horm. Def (MRGH)	X	Xq26-q27
Panhypopituitarism, X-linked	X	Xq26-q27



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Evidence & Guideline Based NIPS Test Solutions From Lilac Insights

InsighT range of tests exclusively available from Lilac Insights in India, are highly reliable NIPS/NIPT tests. Utilizing the world's most robust and validated technology, these tests are preferred by millions of pregnant women and leading medical experts globally. They offer sensitive and accurate prenatal screening for Down syndrome and other common genetic conditions during pregnancy.

World's Most



Preferred

>1 Crore Successful Tests

Done Till Date



Validated

Largest Prospective
Validation On

>146900 Pregnancies



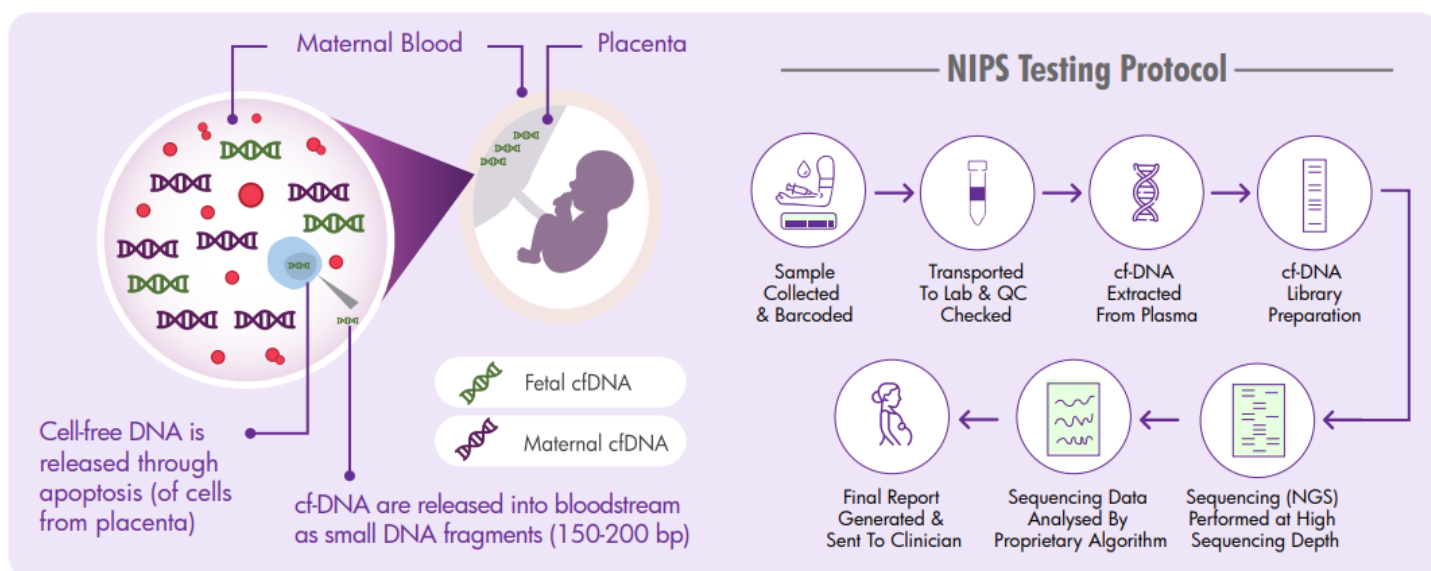
Accurate

Proven Superior
Performance With

>70 Publication Studies

How NIPS Test Is Performed?

The NIPS test analyzes the fetal DNA present in the mother's blood sample to assess whether the developing baby exhibits common chromosomal conditions that could impact their health and development. E.g. T21 (Down syndrome), T18 (Edwards syndrome), T13 (Patau syndrome), Sex chromosome aneuploidies, Microdeletion/duplication syndromes.



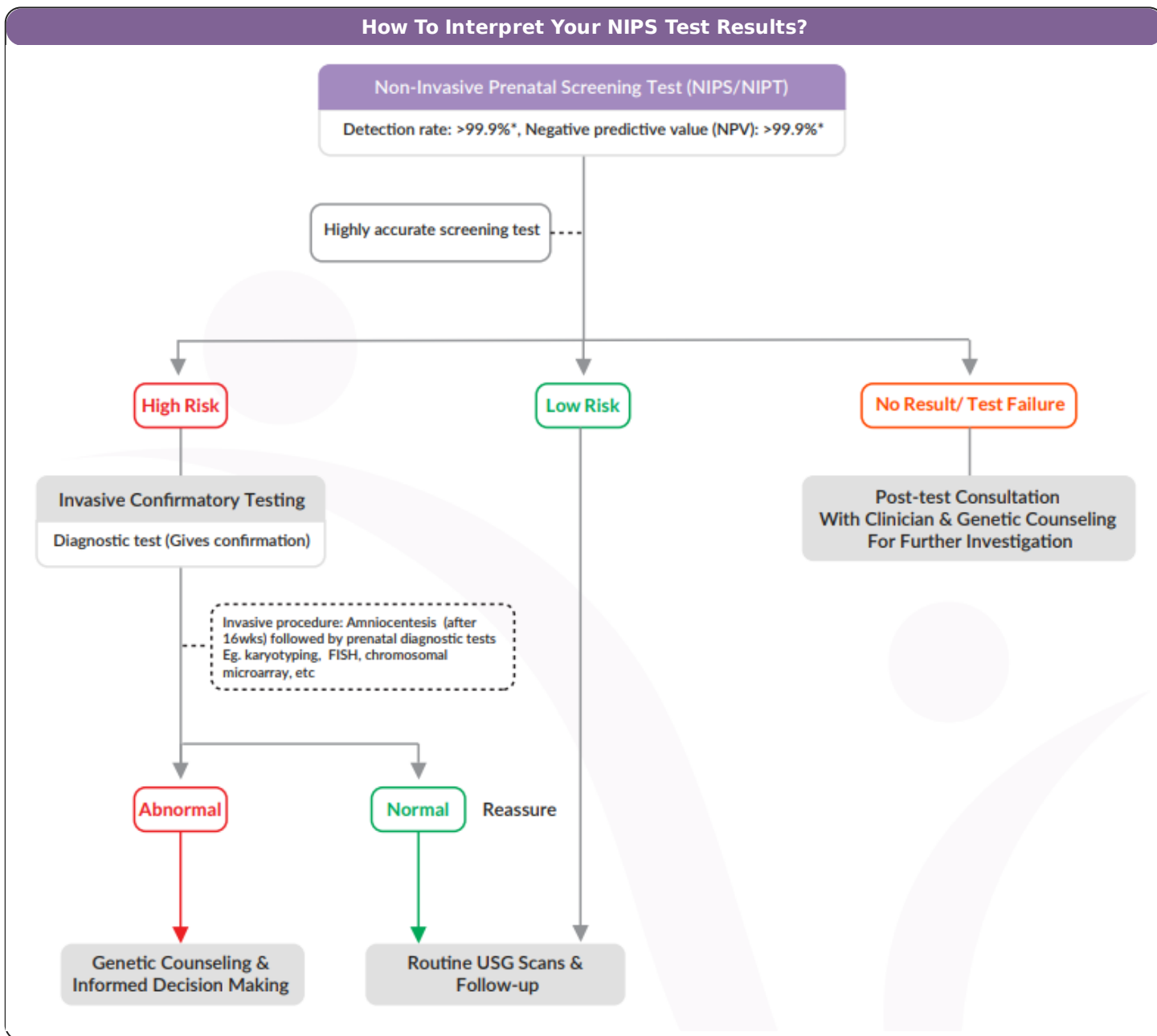
What Is Fetal Fraction (FF%)?

It is the amount of fetal DNA released & circulating into mother's blood stream. Only adequate quantity of fetal DNA can ensure the accuracy and reliability of the results.

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How To Interpret Your NIPS Test Results?



*As Per Validation Studies

cfDNA: Cell-free DNA; NIPS: Non-Invasive Prenatal Screening; FF%: Fetal Fraction %; FISH: Fluorescence In-Situ Hybridisation; KT: Karyotyping; CMA: Chromosomal Microarray; CVS: Chorionic Villus Sampling; SCAs: Sex Chromosome Aneuploidies; USG: Ultrasonography; GA: Gestational Age.

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Disclaimer

1. The Insight range of tests (Insight, Insight-Plus, Insight-Adv) are NOT diagnostic tests. They are screening tests, therefore false-positive and false-negative results can occur. The 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. Sex of the fetus cannot be revealed as per PC-PNDT act 2003. PC-PNDT registration no.:NMMC/PNDT/168.
3. Potential sources of an inaccurate test result 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. 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.
5. Test results should always be interpreted by a qualified healthcare professional in the context of other clinical and/or family information of the patient.
6. The results should be communicated in a setting that includes appropriate genetic Counselling.
7. Validation studies are carried out for all conditions by Beijing Genomics Institute (BGI).
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 (eg. open neural tube defects).
9. Patients with malignancy or a history of malignancy, patients with bone marrow or organ transplant, as well as vanished twin & Twin reduction pregnancies (vanishing/reduction after 8 weeks of GA) are not eligible for these tests.
10. Sex chromosome aneuploidies are not reportable for twin gestations.
11. A positive result for twin pregnancies indicates high risk for the presence of at least one affected fetus.
12. 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
13. This test has been performed at our partner lab.


Verified by
Pallavi Kadam
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