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

Patient Information	
Name: Mrs. NIMISHA BATRA	**Patient ID: 1602411920**
Date of Birth: 09/03/1988	**Sample ID: 2400178341**
Gestation age by Ultrasound: 12 Weeks + 1 days	**Sample collected on: 26/08/2024**
Referring Doctor: Dr. VIJAYA GUPTA	**Sample received on: 28/08/2024**
Sample Type: Blood	**Report released on: 04/09/2024**
Referral Reason: Advanced maternal age.	

Methodology

The InsighT-Adv 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-Adv test provides risk assessment for common aneuploidies (T21, T18, and T13), Sex chromosome aneuploidies, and 84 Microdeletion/duplication syndromes. The validation studies have been carried out for all the conditions reported by InsighT-Adv NIPS test. With >25 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.

ADDITIONAL CONDITIONS	RISK ASSESSMENT
Microdeletions/Duplications (84 Types)	Low Risk

Fetal cfDNA Percentage	**14.48%**

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84 Microdeletions/duplications syndromes screened in InsighT-Adv

Disorder	Chromosome	Location
DiGeorge syndrome	22	22q11.21
Smith-Magenis syndrome	17	17p11.2
Potocki-Lupski syndrome	17	17p11.2
Wolf-Hirschhorn syndrome	4	4p16.3
Dandy-Walker syndrome	3	3q22-q24
Jacobsen syndrome	11	11q23
Cri du Chat syndrome	5	5p
Cat-Eye syndrome	22	22q11
Prader-Will/Angelman syndrome	15	15q11.2
Chromosome 22q11.2 duplication syndrome	22	22q11.2
Chromosome 22q11.2 deletion syndrome	22	22q11.2
Chromosome 1p36 deletion syndrome	1	1p36
Chromosome 16p11.2-p12.2 microduplication syndrome	16	16p12.2-p11.2
Chromosome 5q12 deletion syndrome	5	5q12
Chromosome 18q deletion syndrome	18	18q
Chromosome 9p deletion syndrome	9	9p
Chromosome 14q11-q22 deletion syndrome	14	14q11-q22
Chromosome 6q11-q14 deletion syndrome	6	6q11-q14
Chromosome 8q12.1-q21.2 deletion syndrome	8	8q12.2-q21.2
Chromosome Xq21 deletion syndrome	X	Xq21
Chromosome 1q41-q42 deletion syndrome	1	1q41-q42
Chromosome 6q24-q25 deletion syndrome	6	6q24-q25
Chromosome 18p deletion syndrome	18	18p
Chromosome 10q26 deletion syndrome	10	10q26
Chromosome 3pter-p25 deletion Syndrome	3	3pter-p25
Chromosome 2p12-p11.2 deletion syndrome	2	2p12-p11.2
Chromosome 5q14.3 deletion syndrome	5	5q14.3-q15
Chromosome 13q14 deletion syndrome	13	13q14
Chromosome 10q22.3-q23.2 deletion syndrome	10	10q23
Chromosome 15q26-qter deletion syndrome	15	15q26-qter
Chromosome 6pter-p24 deletion syndrome	6	6pter-p24
Chromosome 16p12.2-p11.2 deletion syndrome	16	16p12.2-p11.2
Chromosome Xq27.3-q28 duplication syndrome	X	Xq27.3-q28
Chromosome 15q25 deletion syndorme	15	15q25
Split-hand/foot malformation 5	2	2q31
WAGRO syndrome	11	11p13-p12
Chromosome 4q21 deletion syndrome	4	4q21
Yuan-Harel-Lupski syndrome	17	17p12-p11.2
Chromosome 2p16.1-p15 deletion syndrome	2	2p16.1-p15
Chromosome 4q32.1-q32.2 triplication syndrome	4	4q32.1-q32.2
Chromosome Xp21 deletion syndrome	X	Xp21
Chromosome 16p deletion syndrome	16	16pter-p13.3

Disorder	chromosome	Location
CHDM	6	6q27
DiGeorge syndrome 2	10	10p14-p13
Langer-Giedion syndrome	8	8q24.11-q24.13
Holoprosencephaly 1	21	21q22.3
Frias syndrome	14	14q22.1-q22.3
Potocki-Shaffer syndrome	11	11p11.2
HCD	15	15q26.1
Levy-Shanske syndrome	15	15q26-qter
Chromosome 7q deletion	7	7q
Chromosome 1p32-p31 deletion syndrome	1	1p32-p31
Chromosome 1p31 duplication syndrome	1	1p31.3
Chromosome 16q22 deletion syndrome	16	16q22
Chromosome 16p13.3 deletion syndrome	16	16p13.3
Chromosome 15q11-q13 duplication syndrome	15	15q11
Chromosome 15q14 deletion syndrome	15	15q14
Chromosome 17q12 duplication syndrome	17	17q12
Chromosome 17q12 deletion syndrome	17	17q12
Chromosome 3q29 duplication syndrome	3	3q29
Chromosome 3q29 deletion syndrome	3	3q29
Chromosome 8q22.1 duplication syndrome	8	8q22.1
Chromosome 8q22.1 deletion syndrome	8	8q22.1
Chromosome 7q11.23 deletion syndrome	7	7q11.23
Chromosome 7q11.23 duplication syndrome	7	7q11.23
Chromosome Xq22.3 telomeric deletion syndrome	X	Xq22.3
Chromosome 17q21.31 duplication syndrome	17	17q21.31
Chromosome Xp11.3 deletion syndrome	X	Xp11.3
Chromosome 3q13.31 deletion syndrome	3	3q13.31
Chromosome 8p23.1 deletion syndrome	8	8p23.1
Chromosome 8p23.1 duplication syndrome	8	8p23.1
Chromosome 17q23.1-q23.2 deletion syndrome	17	17q23.1-q23.2
Chromosome Xq28 deletion syndrome	X	Xq28
Chromosome 17p13.3 duplication syndrome	17	17p13.3
Chromosome 17p13.3 deletion syndrome	17	17p13.3
Chromosome 19q13.11 deletion syndrome	19	19q13.11
WAGR syndrome	11	11p13
Holoprosencephaly 6	2	2q37.1-q37.3
Chromosome 12q14 microdeletion syndrome	12	12q14
Chromosome Xp11.23-p11.22 duplication syndrome	X	Xp11.23-p11.22
Chromosome 2q31.1 duplication syndrome	2	2q31.1
Chromosome 2q33.1 deletion syndrome	2	2q33.1
Chromosome 2q35 duplication syndrome	2	2q34-q36

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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%	

Additional Conditions

Conditions		Sensitivity	Specificity	Reference
Microdel/Dup (84 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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Disclaimers:

1. The InsighT-Adv 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.

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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.

Pallavi Kadam

Verified By

Scientific Officer

Genomics

Pusalkar

Dr. Madhavi Pusalkar, Ph.D.

General Manager

Genomics

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

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