

NAME	: MRS BANDANA	REFERRED BY	: DR PRIYANKA KUNDAL	VISIT NO	: VAMJ25017100
AGE	: 27Y 0M 0D	PATH SQUARE LABS PVT LTD		COLLECTED ON	: 23-06-2025 10:21
GENDER	: Female	LAB MR#	: AAMJ00067544	RECEIVED ON	: 23-06-2025 10:36
OP / IP / DG #	:			APPROVED ON	: 05-07-2025 20:12
				REPORT STATUS	: Final Report



Test Name	Result	Biological Ref. Interval	Unit
Sample Information			
Fetal Fraction	10.24%		
Sample Information	Qubit Fluorometer (ng/ul):2.33; Volume (ul): 50; Total amount (ng): 116.5		

I. Screening results

Chromosomes	Risk	Z score	Test Results	Reference interval
Chromosome 13		-0.18	Low Risk	-6<Z score<2.8
Chromosome 18		-0.73	Low Risk	-6<Z score<2.8
Chromosome 21		-0.23	Low Risk	-6<Z score<2.8
Sex Chromosomes		Part II	Low Risk	Part II
Other Chromosomes		Part III	Low Risk	Part III

Low Risk; High Risk — Further Investigation Recommended

II. Sex Chromosomes

Sex Chromosome Aneuploidies	Risk	Z score	Test Results
XO			Low Risk
XXY/XYY		-0.81	Low Risk
XXX			Low Risk
Low Risk; High Risk — Further Investigation Recommended			

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III. Other Chromosomes				
Chromosome		Risk	Z score	Test Results
Chromosome 1			0.20	Low Risk
Chromosome 2			-0.45	Low Risk
Chromosome 3			-0.10	Low Risk
Chromosome 4			0.98	Low Risk
Chromosome 5			0.73	Low Risk
Chromosome 6			0.46	Low Risk
Chromosome 7			0.38	Low Risk
Chromosome 8			-0.42	Low Risk
Chromosome 9			-0.69	Low Risk
Chromosome 10			-0.76	Low Risk
Chromosome 11			-0.41	Low Risk

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Chromosome 12		0.87	Low Risk
Chromosome 14		0.23	Low Risk
Chromosome 15		0.11	Low Risk
Chromosome 16		-0.13	Low Risk
Chromosome 17		-0.61	Low Risk
Chromosome 19		0.99	Low Risk
Chromosome 20		0.77	Low Risk
Chromosome 22		0.45	Low Risk

Low Risk; High Risk — Further Investigation Recommended

Pipeline version: 97bef21

IV. Supplementary information

- The NIPT test screens a maternal blood sample for chromosome aneuploidy in placental DNA using the following methodology:
 - Extraction of cell-free placental DNA from the maternal blood sample
 - High throughput sequencing of the extracted cell-free placental DNA
 - Calculation of molecular mass of placental DNA in all chromosomes
- The method is intended for use in pregnant women who are at least 10 weeks of pregnancy. The method is suitable for both singleton and twin pregnancies. The accuracy may be slightly lower in twin pregnancies due to multiple sources of fetal DNA.
- Based on the scope, the NIPT test can detect the following:
 - Whole Genome - 23 pairs of human chromosomes
 - Sex chromosomal aneuploidies: XO, XXX, XXY/XXY
- The test is capable of genome-wide aneuploidy detection over the whole fetal genome and gives the results for 23 pairs of chromosomes. This test confers an accuracy of up to 99% on the detection of fetal aneuploidy for chromosomes 13, 18 and 21. In a study of over 2000 samples, 6 samples were determined to be at high-risk of having an autosomal aneuploidy other than 13, 18 and 21. This is a prevalence rate of 0.3%, which is consistent with prevalence in published studies.

Results are indicated for screening, NOT diagnosis. – (Results should be reviewed and discussed with your clinician.)

This test does not report the gender of the fetus.

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Prenatal Screening Pathway			
Screening Method 1	Screening Method 2	Screening Method 3	
Traditional First Trimester Non invasive screening Accuracy 85%	Non Invasive Prenatal Testing (NIPT) Accuracy >99%	Invasive Prenatal Diagnostic Testing Accuracy ~100%	
<div style="border: 1px solid black; padding: 10px; width: 150px;"> <div style="border: 1px solid black; padding: 5px; margin-bottom: 10px;">First Phase Screening</div> <div style="border: 1px solid black; padding: 5px;">Second Phase Screening</div> </div>	<div style="border: 1px solid black; padding: 10px; width: 150px;"> <p>High Risk Group</p> <p>Non Invasive Prenatal Testing (NIPT) 10 weeks gestation onwards</p> </div>	<div style="border: 1px solid black; padding: 10px; width: 150px;"> <p>High Risk Group</p> <div style="border: 1px solid black; padding: 5px; margin-bottom: 10px;">Chorionic Villus Sampling (14 weeks gestation onwards)</div> <div style="border: 1px solid black; padding: 5px;">Amniocentesis (16 weeks gestation onwards)</div> </div>	
		<p>Low Risk Group- No further invasive testing required</p>	

About NIPT prenatal screen

The NIPT prenatal screen is a new advanced non-invasive prenatal screening solution using the latest developments in DNA technology to detect placental DNA in maternal blood. It offers a menu-based chromosome analysis to estimate the risk of a fetus having Down's syndrome and other genetic disorders. Enabling pregnant women and their families fast, safe and reliable results and reducing the need for invasive tests and the associated risks, stress and anxiety. NIPT Prenatal screen is indicated for use in pregnant women who are at least 10-week pregnant. Chromosomal aneuploidy can then be detected using bioinformatics analyses, where the detection rate and sensitivity are over 99%.

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Limitations

NIPT prenatal screen is a screening test and all high-risk results should be confirmed through further investigation which may include tests such as amniocentesis or Chorionic Villus Sampling (CVS). Pregnant women with a high-risk result should be referred for genetic counseling and offered invasive prenatal diagnosis for confirmation of test results. Pregnant women with a negative test result do not ensure an unaffected pregnancy. While results of this testing are highly accurate, not all chromosomal abnormalities may be detected due to placental, maternal or fetal mosaicism, or other causes (micro-deletions, chromosome re-arrangements, translocations, inversions, unbalanced translocations, uniparental disomy). The test is not reportable for known multiple gestations, or if the gestational age is less than 10 weeks.

Test method

A simple maternal blood sample is taken from the pregnant mother from 10-week gestation with no risk to the fetus. Circulating cell-free placental DNA was purified from the plasma component of anti-coagulated 10mL of maternal whole blood. It was then converted into a genomic DNA library for Next Generation Sequencing and then determination of chromosomal aneuploidy.

References:

1. Obstet Gynecol 2012;119:890-901.
2. BMJ 2011;342:c7401.
3. Prenat Diagn 2012;32:c7401.
4. ACOG/SMFM Joint Committee Opinion No. 545, Dec 2012.

#Test performed at referral lab

Dr.Kiran K G
Scientist
Molecular biology (Ph.D)

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Disclaimer:

1. All results released pertain to the specimen as received by the lab for testing and under the assumption that the patient indicated or identified on the bill/test requisition form is the owner of the specimen.
2. Clinical details and consent forms, especially in Genetic testing, histopathology, as well as wherever applicable, are mandatory to be accompanied with the test requisition form. The non-availability of such information may lead to delay in reporting as well as misinterpretation of test results. The lab will not be responsible for any such delays or misinterpretations thereof.
3. Test results are dependent on the quality of the sample received by the lab. In case the samples are preprocessed elsewhere (e.g., paraffin blocks), results may be compromised.
4. Tests are performed as per the schedule given in the test listing and in any unforeseen circumstances, report delivery may be affected.
5. Test results may show inter-laboratory as well as intra-laboratory variations as per the acceptable norms.
6. Genetic reports as well as reports of other tests should be correlated with clinical details and other available test reports by a qualified medical practitioner. Genetic counselling is advised in genetic test reports by a qualified genetic counsellor, medical practitioner or both.
7. Samples will be discarded post processing after a specified period as per the laboratory's retention policy. Kindly get in touch with the lab for more information.
8. If accidental damage, loss, or destruction of the specimen is not attributable to any direct or negligent act or omission on the part of Ampath Labs or its employees, Ampath shall in no event be liable. Ampath lab's liability for a lack of services, or other mistakes and omissions, shall be restricted to the amount of the patient's payment for the pertinent laboratory services.