

Dr. Vinay Chopra
 MD (Pathology & Microbiology)
 Chairman & Consultant Pathologist

Dr. Yugam Chopra
 MD (Pathology)
 CEO & Consultant Pathologist




Gestation Age: 15 weeks As Per LMP

Patient Id: 10793740	Gender: Female	Center Code: KOS Diagnostic Lab (Ambala Cantt)
Patient Name: Mrs ANURADHA SHARMA	Clinician Name: Dr. Yugam Chopra	Sample Collected: 2024-12-16
Patient DOB: 1989-08-12	Pregnancy Type: Singleton	Sample Received: 2024-12-18 , 11:34:19
Age: 35	Hospital Name: KOS Diagnostic Lab	Report Released: 2024-12-26,16:29:15

Test Performed:-NIPT- All Chromosome Test

Clinical Indication:

A 35 year old female with GA of 15 weeks and she was advised to be screened for NIPT.

TEST RESULTS		*Fetal fraction: 4.34%		
ANEUPLOIDIES		RESULTS	Z-Score	Risk
Down syndrome (Trisomy 21)		Low Risk:Result consistent with two copies of chromosome 21	-0.63	● ▼
Edwards syndrome (Trisomy 18)		Low Risk:Result consistent with two copies of chromosome 18	-0.15	● ▼
Patau syndrome (Trisomy 13)		Low Risk:Result consistent with two copies of chromosome 13	1.41	● ▼
Sex Chromosomes		Low Risk	-0.20	● ▼

About this test:

This screening test evaluates whether your pregnancy is at increased risk for certain types of chromosomal disorders. Because this is a screen, false positives and false negatives can occur. The estimated fetal fraction of DNA present in this sample is one component of Redcliffe Lab's non-invasive screening algorithm.

*If the fetal fraction is lower than 3.5%, the accuracy of the test may be reduced. To ensure the accuracy of the results, we would recommend a re-sampling of the maternal blood one or two weeks later.

CLINICAL COMMENTS


This result shows a low risk group for all chromosomes based on the Z score.


EXPECTED TEST RESULTS

NIPT analysis can yield any of the following results:

- Low Risk : The probability that the fetus is affected with the specific chromosomal aneuploidy is low.
- High Risk : The probability that the fetus is affected with the specific chromosomal aneuploidy is high confirmatory testing via amniocentesis/CVS is recommended.
- Borderline: Further confirmatory test recommended (Amniocentesis or other confirmatory tests)
- Inconclusive : Due to unavoidable reasons a result could not be generated on the given maternal sample therefore repeat sampling is advised. Invasive testing is recommended if a NO RESULT is generated again.


 Performed by
 Rishabh Sharma
 Senior Scientific Officer
 Clinical-Genomics


 Reviewed by
 Aayushi Gupta
 DBT-HSSC Certified
 Genetic Counsellor


 Approved by
 Dr. Himani Pandey
 Postdoc-SGPGIMS Lucknow
 Lab Head-Clinical Genomics

NOTE:

This Sample was outsourced

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PRENATAL CHROMOSOMAL ANEUPLOIDY RESULTS FOR OTHER CHROMOSOMES

CHROMOSOME		RISK	Test Results	Z-SCORE	Sensitivity(%)
CHROMOSOME 1		● ▼	Low Risk ▼	-1.39	99.9
CHROMOSOME 2		● ▼	Low Risk ▼	1.27	99.9
CHROMOSOME 3		● ▼	Low Risk ▼	0.14	99.9
CHROMOSOME 4		● ▼	Low Risk ▼	0.37	99.9
CHROMOSOME 5		● ▼	Low Risk ▼	-0.76	99.9
CHROMOSOME 6		● ▼	Low Risk ▼	2.39	99.9
CHROMOSOME 7		● ▼	Low Risk ▼	0.74	99.9
CHROMOSOME 8		● ▼	Low Risk ▼	1.26	99.9
CHROMOSOME 9		● ▼	Low Risk ▼	-1.15	99.9
CHROMOSOME 10		● ▼	Low Risk ▼	-1.68	99.9
CHROMOSOME 11		● ▼	Low Risk ▼	-0.29	99.9
CHROMOSOME 12		● ▼	Low Risk ▼	-1.29	99.9
CHROMOSOME 14		● ▼	Low Risk ▼	0.87	99.9
CHROMOSOME 15		● ▼	Low Risk ▼	-0.21	99.9
CHROMOSOME 16		● ▼	Low Risk ▼	-0.44	99.9
CHROMOSOME 17		● ▼	Low Risk ▼	-0.09	99.9
CHROMOSOME 19		● ▼	Low Risk ▼	0.15	99.9
CHROMOSOME 20		● ▼	Low Risk ▼	-0.76	99.9
CHROMOSOME 22		● ▼	Low Risk ▼	0.05	99.9

*Risk Description: ● Low Risk Group ● Borderline Group ● High Risk Group

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METHODOLOGY

NIPT is a simple, non-invasive and low-risk method which offers screening of maternal blood sample for genome-wide aneuploidy detection over the whole fetal DNA (23 pairs of chromosomes) and offers an interpretation of the results for Trisomy 13, Trisomy 18, Trisomy 21, sex chromosomes using following methodology.

1. Extraction of cell free fetal DNA from the component plasma of maternal blood sample.
2. High throughput sequencing of the extracted cell free fetal DNA.
3. Calculation of molecular mass of fetal DNA in all chromosomes.

TEST LIMITATIONS

1. The results of this test are for reference only, not for the final diagnosis. Cell-free fetal DNA does not replace the accuracy and precision of prenatal diagnosis with Amniocentesis or Chorionic Villus Sampling (CVS).
2. If the test result is at high risk, genetic counseling and invasive prenatal diagnosis are needed.
3. If the test result is at low risk, the fetus has a low risk of developing the target disease of this screening, hence unaffected pregnancy. However, the possibility of other abnormalities cannot be excluded, and systematic ultrasound examinations and other prenatal examinations should be conducted.
4. The accuracy and quality of the test may be affected by low fetal fraction <3.5%, maternal or fetal mosaicism, or other causes (micro-deletions, chromosome re-arrangements, translocations, inversions, unbalanced translocations, uniparental disomy). The possibility of false positive or false negative cannot be ruled out.
5. The accuracy and quality of the test may be also be affected by high data noise due to improper blood sample collection, handling, storage, or transportation.
6. This test is not applicable for cases with gestational age <10+0 weeks, received allogeneic blood transfusion, A family history of genetic diseases or a high risk of genetic diseases in the fetus, transplantation and allogeneic cell therapy within 1 year or a pregnancy with malignant tumor.
7. The patient must provide complete, accurate and detailed personal information. Redcliffe labs shall not be responsible for the interruption of testing services and inaccurate results caused by inaccurate information or other misleading factors provided by the patient.
8. The test results in this report are only responsible for the samples submitted for inspection.

REFERENCES

- Jiang, Fetal., Noninvasive Fetal Trisomy (NIFTY) test: an advanced noninvasive prenatal diagnosis methodology for fetal autosomal and sex chromosomal aneuploidies. BMC Med Genomics, 2012. 5: p. 57.
- Chiu, R.W., et al., Noninvasive prenatal diagnosis of fetal chromosomal aneuploidy by massively parallel genomic sequencing of DNA in maternal plasma. BMJ. 2011;342:c7401.
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- Chen, S., et al., A method for noninvasive detection of fetal large deletions/duplications by low coverage massively parallel sequencing. Prenatal Diagnosis, 2013. 33(6): p. 584-590.
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Conditions for Reporting

1. It is presumed that specimen belongs to patient named or identified, such verification being carried out at the point of generation of said specimen.
2. A test might not be performed due to following reasons:
 - a. Specimen Quantity not sufficient (Inadequate collection/spillage during transit).
 - b. Specimen Quality not acceptable (Hemolysis/clotted/lipemic).
 - c. Incorrect sample type.
3. In any of the above case a fresh specimen will be required for testing and reporting.
4. Partial representation of report is not allowed.
5. The reported tests are for the notification of the referring doctor, only to assist him/her in the diagnosis and management of the patient.
6. This report is not valid for Medico Legal Purpose.
7. Applicable Jurisdiction will be of "Delhi" for any dispute/claim concerning the test(s) & results of the test(s).