LABORATORY REPORT

Redcliffe labs

Gestation Age : 21 Weeks 6 days

Patient ID :	5164859	Sex :	Female	Center Code:	GELAB6414
Patient Name :	Mansi	Clinician Name :	NA	Sample Collected :	20 Aug 2023
Patient DOB :	NA	Pregnancy Type :	Singleton	Sample Received :	21 Aug 2023
Age:	39 Years	Hospital Name :	NA	Report Released:	28 Aug 2023

Test Performed:-Non Invasive Prenatal Testing NIPT (Microdeletions)

Clinincal Indication:- A 39 year old female with GA of 21 weeks 6 days, USG scan report shows echogenic bowel and she was advised to be screened for NIPT.

TEST RESULTS

***Fetal fraction: 11.45%**

ANEUPLOIDIES	RESULTS	Z-Score	Risk
Down syndrome (Trisomy 21)	Low Risk	1.10	
Edwards syndrome (Trisomy 18)	Low Risk	0.86	
Patau syndrome (Trisomy 13)	Low Risk	0.75	
Sex Chromosomes	Low Risk		
Microdeletions	Low Risk		

About this test:

NIPT is capable of genome-wide aneuploidy and microdeletion detection of the whole fetal genome (22 pairs of chromosomes). Test results with the interpretation of risk for Trisomy 13, Trisomy 18, Trisomy 21 and sex chromosome aneuploidies and microdeletion will be provided. This test confers an accuracy of up to 99% on the detection of fetal chromosomal aneuploidy along with microdeletion.

*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 & No Microdeletions observed in this report.

EXPECTED TEST RESULTS

NIPT analysis can yield any of the following three 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.**
- **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
Himanshu Saini
Senior Scientific Officer
Clinical-Genomics

Authorized by
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Postdoc-SGPGIMS Lucknow
Lab Head- Clinical Genomics

NIPT

PRENATAL CHROMOSOMAL ANEUPLOIDY RESULTS FOR OTHER CHROMOSOMES

CHROMOSOME	RISK	Sensitivity%
CHROMOSOME 1	●	99
CHROMOSOME 2	●	99
CHROMOSOME 3	●	99
CHROMOSOME 4	●	99
CHROMOSOME 5	●	99
CHROMOSOME 6	●	99
CHROMOSOME 7	●	99
CHROMOSOME 8	●	99
CHROMOSOME 9	●	99
CHROMOSOME 10	●	99
CHROMOSOME 11	●	99
CHROMOSOME 12	●	99
CHROMOSOME 14	●	99
CHROMOSOME 15	●	99
CHROMOSOME 16	●	99
CHROMOSOME 17	●	99
CHROMOSOME 19	●	99
CHROMOSOME 20	●	99
CHROMOSOME 22	●	99

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

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MICRODELETION SYNDROMES

SEX CHROMOSOME ANEUPLOIDIES	RISK	Test Results	Aneuploidy Risk
DiGeorge syndrome	●	**Low risk group**	**< 1/10000**
1p36 deletion syndrome	●	**Low risk group**	**< 1/10000**
Angelman/ Prader-Willi syndrome	●	**Low risk group**	**< 1/10000**
Cri-du-Chat syndrome	●	**Low risk group**	**< 1/10000**
Wolf-Hirschhorn syndrome	●	**Low risk group**	**< 1/10000**

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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, translocat ions, 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. 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.
7. 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.

Bianchi, D.W., et al., Genome-wide fetal aneuploidy detection by maternal plasma DNA sequencing. Obstetrics & Gynecology, 2012. 119(5): p. 890-901.

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.

Chen, M., et al., Validation of fetal DNA fraction estimation and its application in noninvasive prenatal testing for aneuploidy detection in multiple pregnancies. Prenatal Diagnosis, 2019. 39(13): p. 1273-1282.

Lo YM. Non-invasive prenatal diagnosis by massively parallel sequencing of maternal plasma DNA. Open Biol. 2012;2(6):120086.

CONDITIONS FOR REPORTING

It is presumed that the specimen belongs to the patient named or identified, such verification being carried out at the point of generation of said specimen.

A test might not be performed due to following reasons:

1. Specimen Quantity not sufficient (Inadequate collection/spillage during transit).
2. Specimen Quality not acceptable (Hemolysis/clotted/lipemic).
3. Incorrect sample type.

In any of the above cases a fresh specimen will be required for testing and reporting.

Partial representation of the report is not allowed.

The reported tests are for the notification of the referring doctor, only to assist him/her in the diagnosis and management of the patient.

Report with status "Preliminary" means one or more tests are yet to be reported.

This report is not valid for Medico Legal Purpose.

Applicable Jurisdiction will be of "Delhi" for any dispute/claim concerning the test(s) & results of the test(s).

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***AS PER THE ACCORDANCE WITH "PC & PNDT ACT – 1994 & AMENDENTS", SEX OF THE FETUS HAS NEITHER BEEN DETECTED NOR DISCLOSED**

Terms and Conditions of Reporting

1. The presented findings in the Reports are intended solely for informational and interpretational purposes by the referring physician or other qualified medical professionals possessing a comprehensive understanding of reporting units, reference ranges, and technological limitations. The laboratory shall not be held liable for any interpretation or misinterpretation of the results, nor for any consequential or incidental damages arising from such interpretation.

2. It is to be presumed that the tests performed pertain to the specimen/sample attributed to the Customer's name or identification. It is presumed that the verification particulars have been cleared out by the customer or his/her representation at the point of generation of said specimen / sample. It is hereby clarified that the reports furnished are restricted solely to the given specimen only.

3. It is to be noted that variations in results may occur between different laboratories and over time, even for the same parameter for the same Customer. The assays are performed and conducted in accordance with standard procedures, and the reported outcomes are contingent on the specific individual assay methods and equipment(s) used, as well as the quality of the received specimen.

4. This report shall not be deemed valid or admissible for any medico-legal purposes.

5. The Customers assume full responsibility for apprising the Company of any factors that may impact the test finding. These factors, among others, includes dietary intake, alcohol, or medication / drug(s) consumption, or fasting. This list of factors is only representative and not exhaustive.