



KOS Diagnostic Lab
(A Unit of KOS Healthcare)



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

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

Name : Mrs. KARAMJEET
Age/Gender : 38 Yrs/Female
P. ID No. : 1006202430134
Accession No : **100620243020020**
Referring Doctor : KOS DIAGNOTIC
Referred By :

Billing Date : 02/03/2024 02:14:29 PM
Sample Collected on : 02/03/2024 02:14:31 PM
Sample Received on : 03/03/2024 12:18:56 PM
Report Released on : 11/03/2024 03:19:44 PM

Report Status -Final

MOLECULAR BIOLOGY

NIPS Advanced (Screening for Aneuploidy of 13, 18, 21 & Sex Chromosomes)

Sample : Blood ccfDNA Tube
Method : NGS

NIPS Advanced NIPS Advanced (Screening for Aneuploidy of 13, 18, 21 & Sex Chromosomes)

See Attachment

** End of Report **

Authenticated By

Dr. Sarjana Dutt
Director-NRL Molecular Biology &
Cytogenetics



NOTE:

This Sample was outsourced




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Patient Details			
Accession No.	1006202430134	Clinician Name:	Dr. Sugandha Chopra
Patient Name:	Karamjeet	Hospital/Clinic Name	Loomba Maternity & E.N.T. Hospital
Age/ Gender:	38/F	Sample Collected On:	02/03/2024
Maternal Weight:	61	Sample Received On:	03/03/2024
Pregnancy Type:	Singleton	Report Released On:	11/03/2024 07:56
Gestational Age:	18 Wk 1 Day	Barcode No:	1006019619

Indication: Screening for Aneuploidy

I. Screening Results

Chromosomes	Risk	Z score	Test Results	Reference interval
Chromosome 21 	●	0.82	Low Risk	-6<Z score<2.8
Chromosome 18 	●	1.82	Low Risk	-6<Z score<2.8
Chromosome 13 	●	0.77	Low Risk	-6<Z score<2.8
Sex Chromosomes				
XO	●		Low Risk	
XXY/XXY	●	0.66	Low Risk	Male -3<Z score<3 Female -2.8<Z score<2.8
XXX	●		Low Risk	

Fetal fraction:6.18%

Comments: Low Risk of Aneuploidy for chromosomes 13, 18 & 21 and Sex chromosomes was observed. Further follow up with your Healthcare provider is advised.

Clinical Information:

The Sage™ prenatal screen is an advanced non-invasive prenatal screening solution using the latest developments in DNA technology to detect placental DNA in maternal blood. Sage™ 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. Sage™ is indicated for use in pregnant women who are at least 10-week pregnant.

Based on the scope, this NIPS test can detect the following in the human genome:

- Trisomy 21, 13 and 18
- Sex chromosomal aneuploidies: XO, XXX, XXY/XXY

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

Since this is a Screening Test (pipeline version: 97bef21), based solely on the results of this Test, no irreversible clinical decision should be taken. Clinical correlation and follow up Test using Invasive procedure is mandated in such cases.

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Test Methodology:

The test is capable of aneuploidy detection in the fetal DNA and offers an interpretation of the results for Trisomy 13, Trisomy 18, Trisomy 21, sex chromosomes. The Test is based on Whole Genome Sequencing and confers an accuracy of up to 99% on the detection of fetal chromosome aneuploidy. The Test process consists of-

- Extraction of cell free fetal DNA from the maternal blood sample
- Preparation of genomic DNA library and high throughput Next Generation Sequencing
- Calculation of fetal DNA percentage
- Determination of Chromosomal aneuploidy is done using bioinformatics analyses
- Reporting of Results for aneuploidies as a personalized Risk score, a calculated level of accuracy of a High or Low Risk call.

Limitations:

- Cell-free fetal (placental) DNA Testing does not replace the accuracy and precision of prenatal diagnosis with Amniocentesis or Chorionic Villus Sampling (CVS).
- Pregnant women with a Positive test result should be referred for genetic counseling and offered invasive prenatal diagnosis for confirmation of test results.
- A Negative test result does not ensure an unaffected pregnancy. While results of this testing are highly accurate (>99%), 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). Results may be compromised in case of fetal demise and /or vanishing twin.
- The accuracy and quality of the test may be affected by improper blood sample collection, storage and transportation.
- In Samples where the fetal fraction is less than 3.5%, the sensitivity of the NIPS test is reduced. Fetal fraction decreases with increasing maternal age and weight; with parity and presence of autoimmune disease in the pregnant woman.
- The test is reportable for only for Singleton and twin pregnancies.
- NIPS is not suitable if the pregnant woman has cancer or chromosomal abnormalities.
- NIPS should not be performed if the pregnant woman has had an organ transplant or received stem cell therapy or immunotherapy within the last 12 months; or has had a blood transfusion within the last 3 months. NIPS is not suitable if maternal blood and oocyte are not of the same genetic lineage; as in pregnancy achieved with donor egg or surrogacy.
- Samples with gestational age less than 10 weeks are not reportable.

Note: Results are indicated for screening, NOT diagnosis. – (Results should be reviewed and discussed with your healthcare provider.)

References:

1. Obstet Gynecol 2012;119:890-901.
2. F1000Research 2019, 8(F1000 Faculty Rev) May 2019 :764
3. Practice Guideline Obstet Gynecol 2020 Oct;136(4): e48-e69
4. Prenatal Diagnosis. 2020; 40:155–163.

In accordance with "Pre Natal Diagnostic Techniques (Regulation and Prevention of Misuse) Rules 1994", the laboratory does not disclose the sex of the fetus.



Dr Sarjana Dutt
Director- Molecular Biology & Cytogenetics

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