

(A Unit of KOS Healthcare)



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

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

Name : Mrs.REKHA DEVI W.O YASH PAUL Centre Details :KOS DIAGNOSTIC LAB
Age : 27 Yrs Sex: Female Accession.ID :PAN2409080020

Collection Date : 08/Sep/2024 02:52PM Referred By :SELF

Received Date : 09/Sep/2024 03:18PM Report Date :17/Sep/2024 05:13PM

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DEPARTMENT OF MOLECULAR DIAGNOSTICS-III

#NIPT: All Chromosome Test

NIPT screens a maternal blood sample for chromosomal aneulpoidy in fetal DNA using the following methodology:

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

Based on the scope, NIPT can screen the following conditions:

- (a) Whole Genome 23 pairs of human chromosomes
- (b) Common Chromosomal abnormality:
 - Trisomy 13 (Patau's Syndrome)
 - Trisomy 18 (Edwards' Syndrome)
 - Trisomy 21 (Down's Syndrome)

NIPT is capable of genome-wide aneuploidy detection of the whole foetal genome (23 pairs of chromosomes). Test results with the interpretation of risk for Trisomy 13 Trisomy 18, Trisomy 21 and sex chromosome aneuploidies will be provided. This test confers an accuracy of up to 99% on the detection of foetal chromosomal aneuploidy.

Pregnancy Type	Blood ID	Patient ID
Singleton	NA	NA

Test Results Summary

Autos	omal Aneuploidies	Risk	Test Results	Aneuploidy Risk
Chromosome 13			Low risk group	<1/10000
Chromosome 18			Low risk group	<1/10000
Chromosome 21			Low risk group	<1/10000
Other Chromosomes			Low risk group	



Dr. Vinay Bhatia Ph.D. Head- Molecular Biology and Genomics

Dr. Shivali Ahlawat MD. D.N.B (Path) Head- National Reference Lab HMC RG-No.17038

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Test Results for Sex Chromosome Aneuploidies

Sex Chromosome Aneuploidies	Risk	Test Results	Aneuploidy Risk	
хо		Low risk group	<1/10000	
XXY		Low risk grou <mark>p</mark>	<1/10000	
xxx		Low risk group	<1/10000	
XYY		Low risk group	<1/10000	

※ Risk description: ■ Low risk group; ■ Borderline group, ■ High risk group

Test Results for Other Chromosomal Aneuploidies

Other Chromosomal Aneuploidies		Risk	Test Results	
Chromosome 1			Low risk group	
Chromosome 2			Low risk group	
Chromosome 3			Low risk group	
Chromosome 4			Low risk group	
Chromosome 5			Low risk group	
Chromosome 6			Low risk group	
Chromosome 7			Low risk group	
Chromosome 8			Low risk group	
Chromosome 9			Low risk group	
Chromosome 10			Low risk group	
Chromosome 11			Low risk group	
Chromosome 12			Low risk group	
Chromosome 14	010		Low risk group	
Chromosome 15	000000000000000000000000000000000000000		Low risk group	
Chromosome 16			Low risk group	
Chromosome 17			Low risk group	
Chromosome 19	3%		Low risk group	





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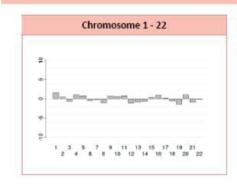
Chromosome 20		Low risk group
Chromosome 22	01001111	Low risk group

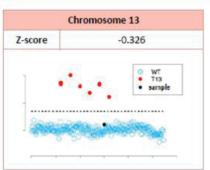


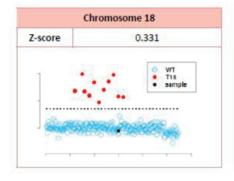
Sample Information					
Fetal DNA fraction			12.35 %		

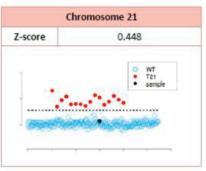
Note: In rare cases when fetal DNA fraction level is low, new blood sample will be requested for retesting.

Test Results



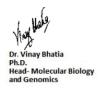






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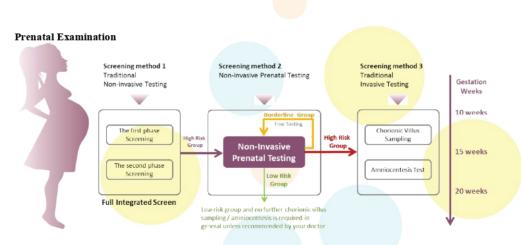
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About the Test

NIPT analyzes circulating fetal cell-free DNA extracted from a maternal blood sample, and is offered to pregnant women with a pre-test risk of an euploidy in chromosomes such as 13, 18, 21, X or Y. The chance that a fetus is affected with chromosomal an euploidy can be estimated using bioinformatics analyses, by which the accuracy rate and sensitivity are over 99%. The accuracy and quality of the test may be affected by low fetal fraction, high data noise due to improper blood sample collection, handling, storage, or transportation.

Limitations of the test

Non-invasive prenatal testing should only be considered a screening test. The screening test of fetal cell-free DNA cannot compare with the prenatal diagnosis with Amniocentesis or Chorionic Villus Sampling (CVS). Pregnant women with a positive NIPT screening result should be given an invasive prenatal diagnosis and referred further for genetic counselling to confirm conditions. On the other hand, a negative test result does not ensure an unaffected pregnancy. Even though NIPT provides reliable results, it does not apply to all cases of chromosomal abnormalities, for example, cases due to placental, maternal, or fetal mosaicism, or other causes (e.g. micro-deletions, chromosome rearrangements, translocation, inversions, unbalanced translocation, uni parental disomy, etc.). NIPT is also not applicable for cases with a diagnosed multiple gestation, or with gestational age that is less than 10 weeks. In rare cases when a borderline screening result is reported, retesting is required to confirm conditions.

Test Method

NIPT applies a non-invasive and low-risk procedure to collect fetal DNA samples. Circulating fetal cell-free DNA is purified from the plasma component of 10mL anti-coagulated maternal whole blood. It is then converted into a genomic DNA library for Next Generation Sequencing to determine Trisomy 21, 18 and 13 and other chromosomal abnormalities.





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References

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

Note: This test is developed, validated & performed at the third party lab.

Note: The sex of fetus is not revealed due to PNDT Act.

*** End Of Report ***

Disclaimer: All Results released pertain to the specimen submitted to the lab

- 1. Test results are dependent on the quality of the sample received by the lab
- 2. Tests are performed as per schedule given in the test listing and in any unforeseen circumstances, report delivery may be delayed 3. Test results may show interlaboratory variations
- 4. All dispute and claims are subjected to local jurisdiction only. Clinical correlation advised.
- 5. Test results are not valid for medico legal purposes
- 6. For all queries, feedbacks, suggestions, and complaints, please contact customer care support +0124 665 0000





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