

(A Unit of KOS Healthcare)



Dr. Vinay Chopra
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Chairman & Consultant Pathologist

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

Name : Mrs.MEENAKSHI SHARMA Centre Details :ONCQUEST PANCHKULA Age : 37 Yrs Sex: Female Accession.ID :PAN2406260051 Collection Date :KOS DIAGNOSTIC LAB : 27/Jun/2024 04:23PM Referred By Received Date : 27/Jun/2024 09:02AM Report Date :06/Jul/2024 03:53PM Registration Date : 26/Jun/2024 Ref. No./TRF No.

### DEPARTMENT OF MOLECULAR DIAGNOSTICS-III

### **#NIPT: All Chromosome Test**

### Indication: -

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

Au	tosomal Aneuploidies	Risk	Test Results	Aneuploidy Risk
Chromosome 13	01001111		Low risk group	<1/10000
Chromosome 18		•	Low risk group	<1/10000
Chromosome 21		•	Low risk group	<1/10000
Other Chromoson	ne	•	Low risk group	

### Test Results for Sex Chromosome Aneuploidies



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Sex Chromosome Aneuploidies	Risk	Test Results	Aneuploidy Risk
xo		Low risk group	<1/10000
XXY		Low risk group	<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	010111111111111111111111111111111111111		Low risk group
Chromosome 15			Low risk group
Chromosome 16			Low risk group
Chromosome 17			Low risk group
Chromosome 19			Low risk group
Chromosome 20			Low risk group
Chromosome 22			Low risk group







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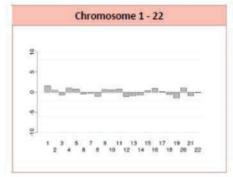
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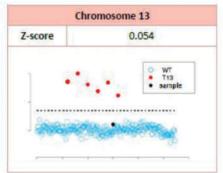
### DEPARTMENT OF MOLECULAR DIAGNOSTICS-III

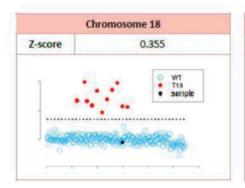
Sample Information		
Fetal DNA fraction	12.39 %	

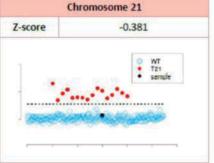
Note: In rare cases when fetal DNA fraction level is low, new blood sample will be requested for retesting.

### **Test Results**









These images are only for representation purposes.





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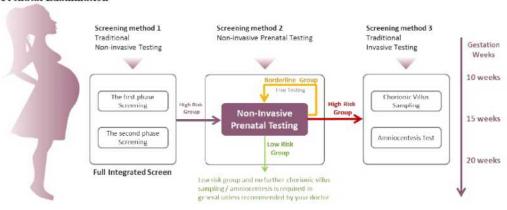
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### DEPARTMENT OF MOLECULAR DIAGNOSTICS-III

#### Prenatal Examination



### 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.

### References

- 1. ObstetGynecol 2012; 119:890-901.
- 2. BMJ 2011; 342:c7401.
- 3. PrenatDiagn 2012; 32:c7401.





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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

