MEDGENOME

CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

NIPT

Non-invasive Prenatal Test to deliver assurance to expectant parents

What is MedGenome NIPT?

MedGenome Non-invasive Prenatal Testing (NIPT) uses advanced bio-informatics technology to evaluate fetal DNA (of placental origin) in maternal blood to identify genetic variations that can lead to disorders.

Simple, safe, and non-invasive prenatal screening test.

Provides assurance to expectant parents with accurate genetic information about their baby.

Advanced bioinformatics technology to evaluate fetal DNA (of placental origin) in maternal blood.

Powered by iFACT

(Individualized Fetal Aneuploidy Confidence Test)

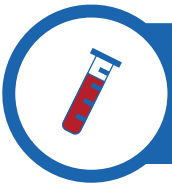
Non - Invasive genetic test for Precise Management

- Assay uses a dynamic threshold metric known as iFACT to make chromosomal aneuploidy calls.
- Indicates whether the system has generated sufficient sequencing coverage, given the fetal fraction estimate for each sample.
- Ensures high accuracy of the result.

MedGenome NIPT: Screens for the Most Common Aneuploidies and rare autosomal aneuplodies

Common Aneuploidies
Trisomy 21
Trisomy 18
Trisomy 13
Sex Chromosome Aneuploidies
XO (Turner Syndrome)
XXX (Trisomy X)
XXY (Klinefelter syndrome)
XYY (Jacob's syndrome)
Rare Autosomal Aneuplodies

Methodology



Cell-free DNA (cfDNA) from both the fetus and the mother is found in the maternal blood.

Next Generation Sequencing (NGS) can detect and measure aneuploidy within this mixed sample.

Quantitative differences in cfDNA in maternal blood can be used to distinguish fetuses affected with abnormalities such as trisomy 21, trisomy 18, trisomy 13 from those that are unaffected.

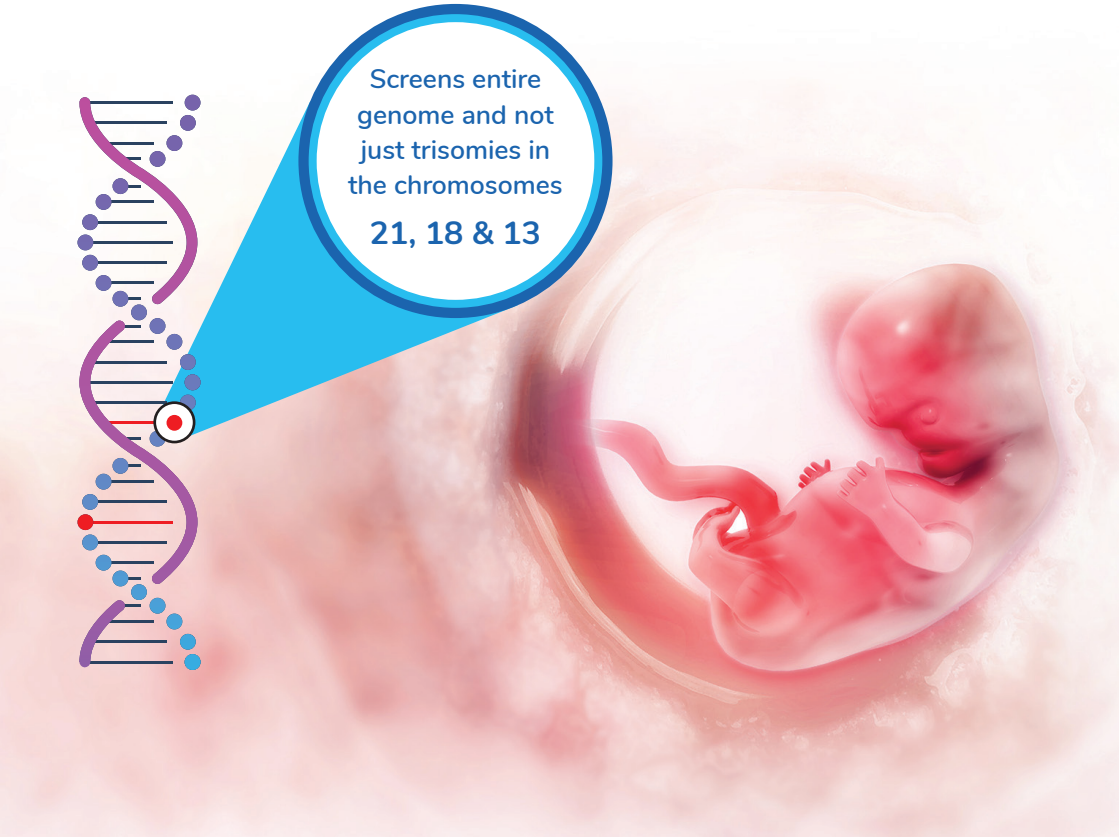
MedGenome NIPT is based on Illumina VeriSeq version 2 Solution to deliver Whole Genome Sequencing (WGS) approach to Non Invasive Prenatal Test (NIPT)

Provides a comprehensive view of the chromosomes.

Offers an enhanced counting technique along with cutting edge algorithms to determine the risk of aneuploidies based on a ratio between chromosomes of interest to multiple reference chromosome.

Key Highlights of MedGenome NIPT

1. Comprehensive view of fetal genome



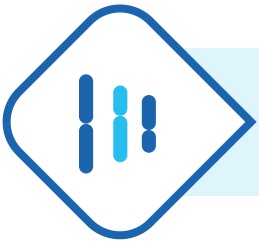
2. Test Performance

- Sensitivity and specificity of >99.9% for trisomy 21, 18, 13
- >99% call rate

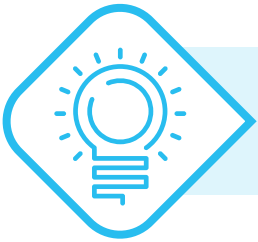
Common Aneuploidies	Sensitivity	Specificity
Trisomy 21	>99.9%	>99.9%
Trisomy 18	>99.9%	>99.9%
Trisomy 13	>99.9%	>99.9%
Any Anomaly	95.5%	99.34%

Accuracy of VeriSeq™ V2 for detecting sex chromosomes aneuploidies.

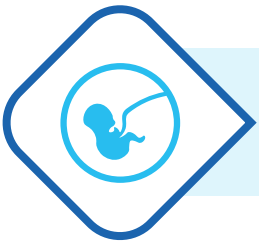
3. Low Test Failure Result



WGS assays provide ample data across the entire diploid genome.

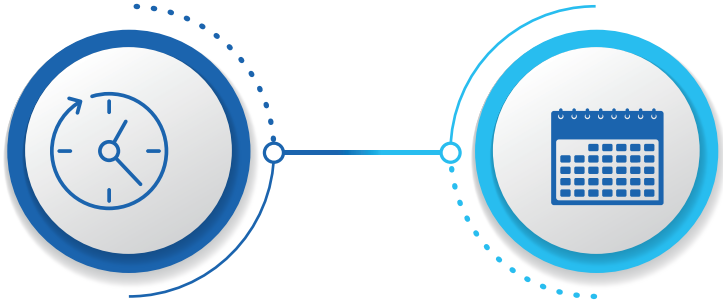


Provides high sensitivity when working with low fetal fraction sample.



Makes correct aneuploidy calls from range of fetal fractions.

4. Fastest Test Result



MedGenome NIPT offers a fast three step automated workflow for NIPT.

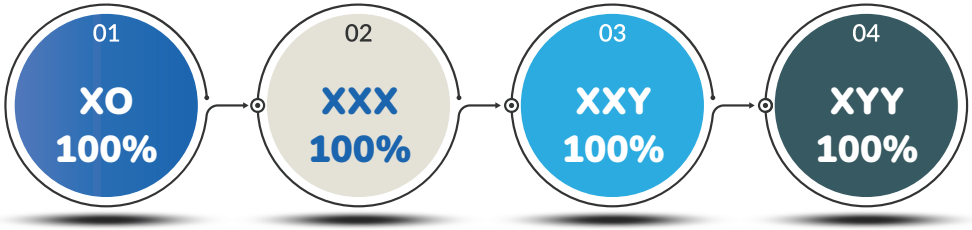
Turn around time is less than or equal to 7 working days.

5. Extensive Validation on Indian Samples

MedGenome NIPT is validated using 303 clinical samples from Indian population.

It includes both high risk samples and samples tested across platform.

Validation successfully identified 51 high risk cases and the low risk cases.



Concordance of MedGenome NIPT for detecting sex chromosomes aneuploidies in MedGenome internal clinical validation.

Why MedGenome NIPT is Superior?

Medgenome Lab Expertise

- Largest CAP Accredited Lab in South Asia
- First Lab in India to introduce NIPT
- Internationally validated technology
- Demonstrates high PPVs
- 100% year on year Proficiency testing since 2017
- ~ 1,00,000 samples analyzed
- High quality report – Fetal fraction, patient's specific risk score always mentioned on the report.

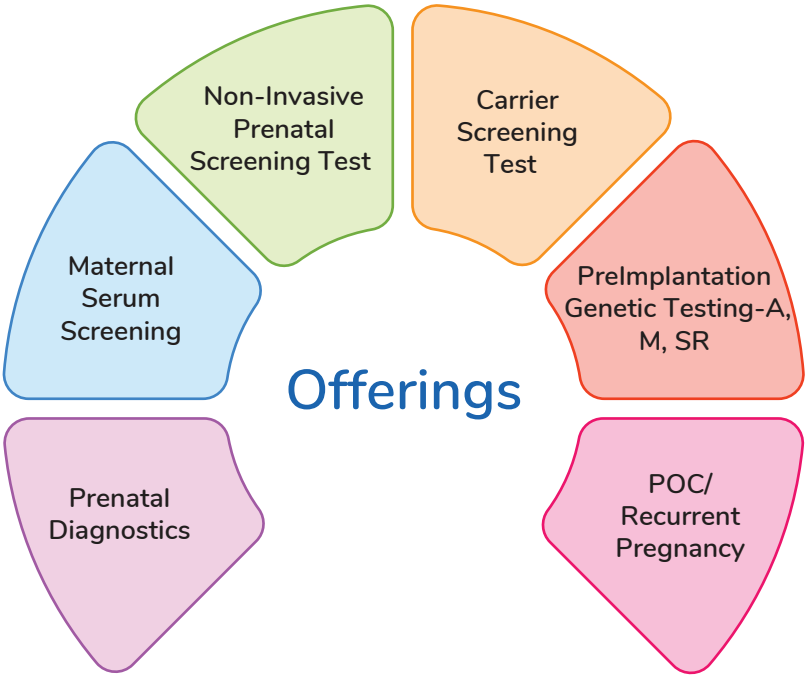
Process

- Active follow up of high-risk cases to maintain quality.
- Detailed analysis for accurate results.

People

- Experienced Analyst across multiple NIPT platform.
- 4 Peer review publications.

Precision in Reproductive Genetic solutions



- QF-PCR
- FISH
- Karyotyping
- MLPA
- Chromosomal Microarray
- Exome Sequencing
- Sanger Sequencing

Talk to the Experts: 1800 296 9696 diagnostics@medgenome.com www.medgenome.com