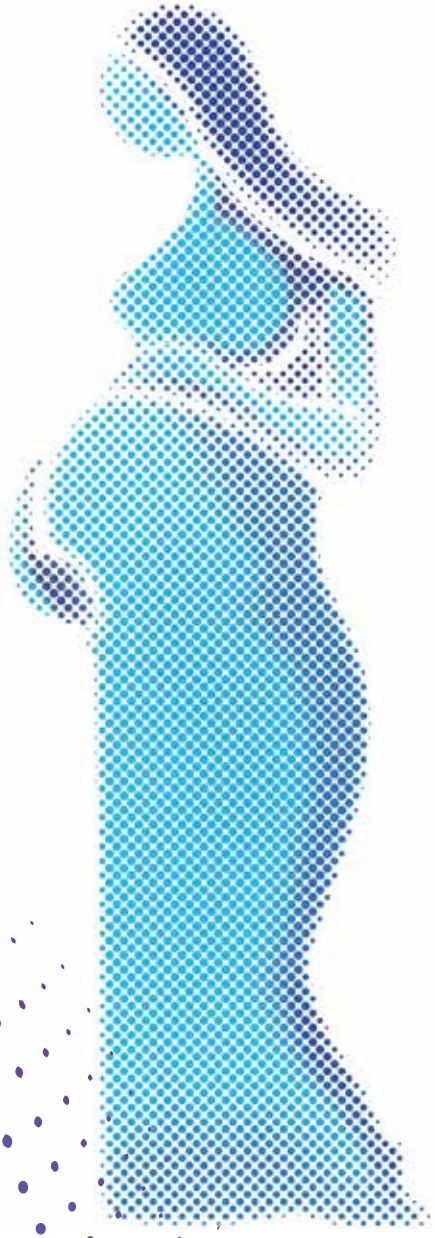
MEDGENOME | Claria Reproductive Genetics

CAP ACCREDITED COLLEGE of AMERICAN PATHOLOGISTS

Claria NIPT

Non-Invasive Prenatal Test

Provide the best clarity and reassurance to your patients



Methodology :

Circulating cell-free DNA (cfDNA), from both the fetus and the mother, is found in maternal blood. On average, ~10% of the cfDNA circulating in maternal blood is from the fetus. Highly sensitive Next Generation Sequencing (NGS), which uses millions of sequence reads per sample, can detect and measure aneuploidy within this mixed sample. Quantitative differences in cfDNA in maternal blood can be used to distinguish fetuses affected with trisomy 21 (and other foetal aneuploidies) from those that are unaffected.

Claria NIPT based on Illumina Veriseq™ Solution v2 brings this whole-genome sequencing (WGS) approach to NIPT. Sequencing of the full fetal genome provides a comprehensive view of the chromosomes. This method 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 Claria NIPT

1. Comprehensive view of the fetal genome

- Screens entire fetal genome and not just trisomies in chromosomes 21, 18, and 13 which represent only a small portion of the genome

2. Test Performance

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

	Trisomy 21	Trisomy 18	Trisomy 13	Any anomaly
Sensitivity (2 sided 95% CI)	>99.9%(130/130) 97.1%, 100%	>99.9% (41/41) 91.4%, 100%	>99.9% (26/26) 87.1%, 100%	95.5% (318/333) 92.7%, 97.3%
Specificity (2 sided 95% CI)	99.9% (1982/1984) 99.63%, 99.97%	99.90% (1995/1997) 99.64%, 99.97%	99.90%(2000/2000) 99.64%, 99.97%	99.34% (1954/1967) 98.87%, 99.61%

Table 1: Accuracy of VeriSeq™ v2 detecting Common Trisomies. (n= 2307 samples, including 7 twin pregnancies)

	XO	XXX	XXY	XYY
Percent Concordant	90.50%	100%	100%	91.70%

Table 2: Concordance of VeriSeq™ v2 for detecting sex chromosome aneuploidies.

3. Low Test Failure Rates

NIPT test failure or no call rates vary significantly based on the test methodology used. Tests that use a targeted approach have demonstrated higher rates of test failure than WGS-based tests, in both validation and clinical experience studies.

WGS assays provide ample data across the entire diploid genome. This coverage produces an analytical reference that current analytical techniques can use to reduce assay- and sample-specific biases. These normalization steps lead to high sensitivity when working with low fetal fraction samples, which means correct aneuploidy calls can be made in the range of fetal fractions that typically requires QC rejection when using targeted approaches[1].

4. Fastest Test Results

- The Claria NIPT offers a fast three-step automated workflow for NIPT
- The turn around time is less than or equal to 7 working days

5. Extensive Validation on Indian Samples

Claria NIPT is validated using 303 clinical samples from Indian population which included both known high risk samples and samples tested with cross platform. The validation successfully identified 51 high risk cases and the low risk cases.

	Trisomy 21	Trisomy 18	Trisomy 13
Sensitivity	>99.99% (6/6)	>99.99% (8/8)	>99.99% (14/14)
Specificity	>99.99% (199/199)	>99.99% (199/199)	>99.99% (199/199)
PPV	>99.99% (6/6)	>99.99% (8/8)	>99.99% (14/14)
NPV	>99.99% (199/199)	>99.99% (199/199)	>99.99% (199/199)

Table 3: Clinical Performance of Claria NIPT in MedGenome internal validation

	XO	XXX	XXY	XYY
Percent Concordant	100% (5/5)	100% (6/6)	100% (6/6)	100% (6/6)

Table 4: Concordance of Claria NIPT for detecting sex chromosome aneuploidies in MedGenome internal clinical validation.

1. Rava RP, Srinivasan A, Sehnert AJ, Bianchi DW. Circulating fetal cell-free DNA fractions differ in autosomal aneuploidies and monosomy X. Clin Chem. 2014;60(1):243-250.

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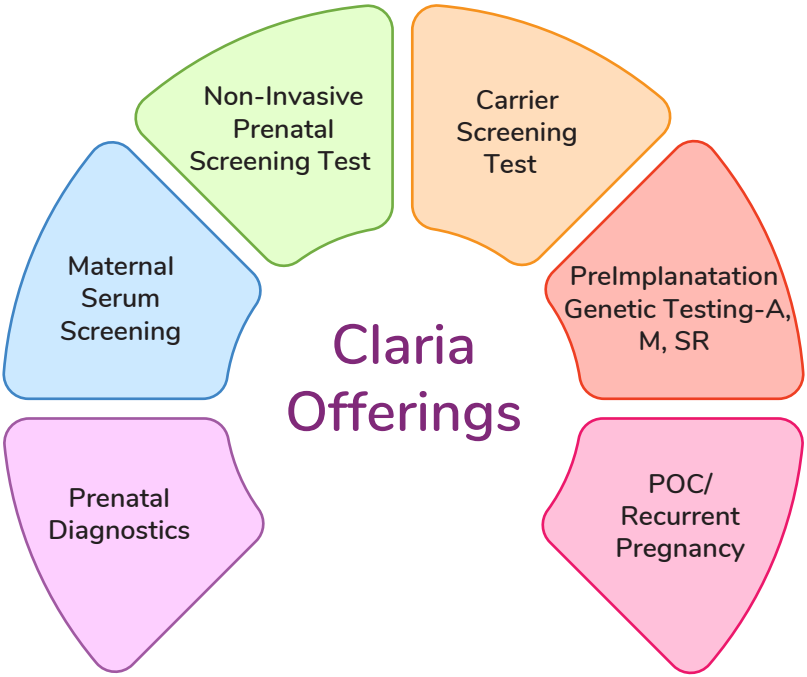
Precision in Reproductive Genetic solutions



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