MEDGENOME | Claria Reproductive Genetics

Carrier Screening Test

Provide the best clarity and reassurance for couples planning a family.

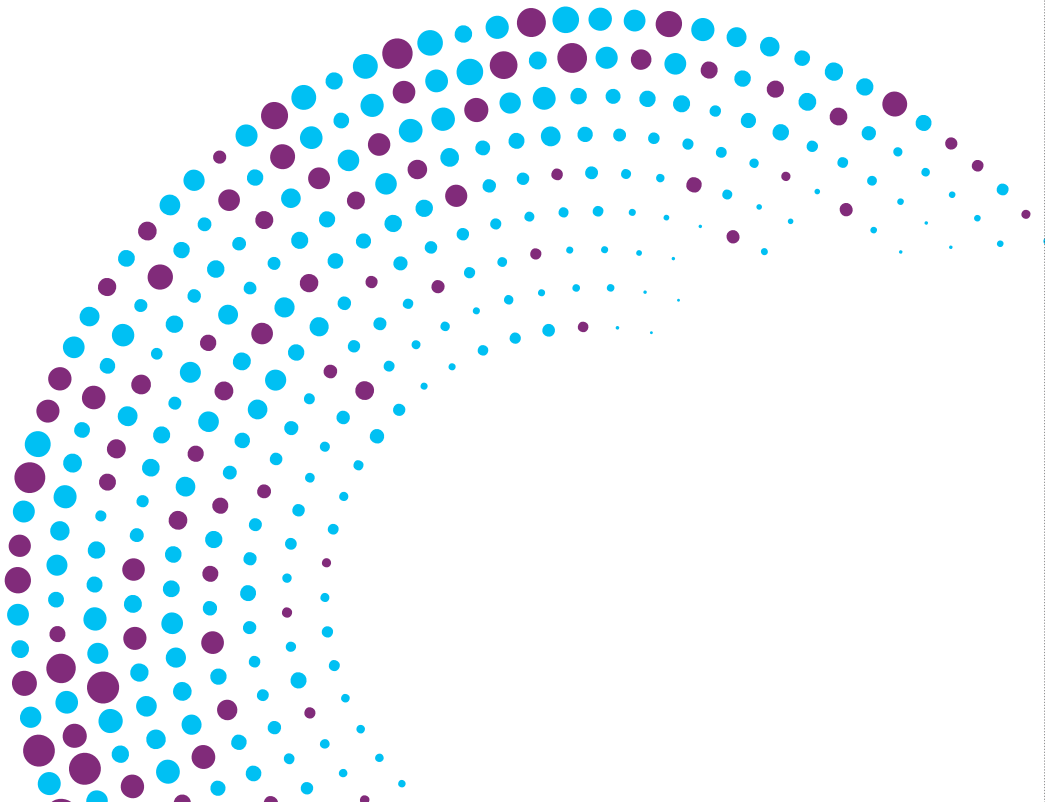
Claria
From MedGenome

MedGenome is driven to empower clinicians to deliver the best outcomes to their patients. Our passion to deliver actionable insights to clinicians has resulted in the development of “Claria” - a suite of NGS (Next-Generation Sequencing) technology-based solutions for reproductive testing.

Claria offers the most accurate Non-Invasive Prenatal Screening Test (NIPT), the Genetic Carrier Screening Test and the Preimplantation Genetic Screening/Diagnosis (PGS/PGD).

We understand your time is valuable, and that’s why Claria has a team of in-house genetic counsellors to help you interpret and explain reports.

Additionally, Claria offers an absolutely free, on-demand pre and post-test genetic counselling to all your patients.



Carrier Screening Test

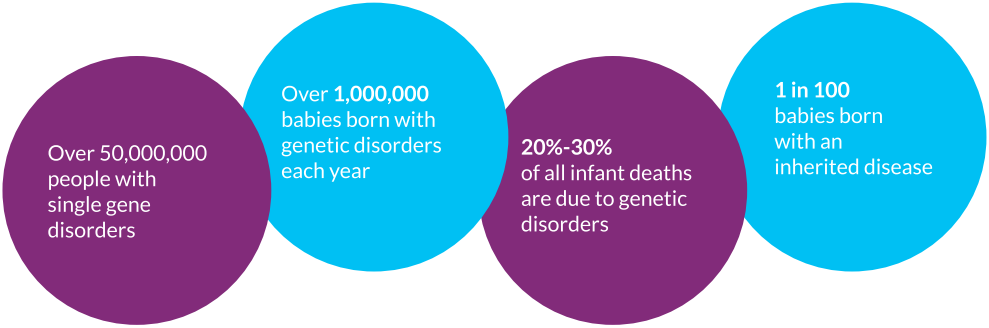
When a couple is planning for a pregnancy, it's important that they know all they can about the wellness of their future family.

State-of-the-art technology, high detection rates and an unparalleled service model can help couples understand and plan better for the future.

The test is a comprehensive screening test that screens for genetic disorders and has the power to detect disease-causing mutations in over 2000 recessive genes.

Claria is based on the best in class NGS and MLPA technologies, and provides your patients with the most accurate and comprehensive information they need when they are preparing for their pregnancy.

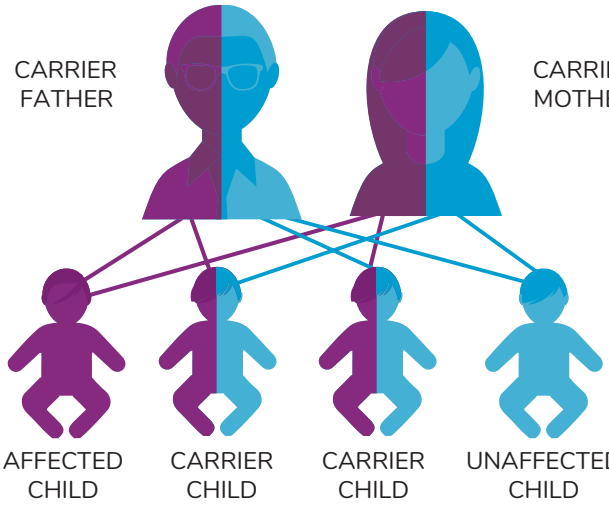
Why is the screening test critical in India?



Most Common Genetic Disorders in India

Autosomal Recessive Disorders	X-linked Recessive Disorders
Beta Thalassemia	Duchenne Muscular Dystrophy
Sickle Cell Anaemia	Haemophilia A/B
Cystic Fibrosis	Hunter Syndrome
Congenital Adrenal Hyperplasia	G6PD Deficiency
Spinal Muscular Atrophy	X-Linked Mental Retardation
Congenital Hypothyroidism	

Autosomal Recessive Disorders



AFFECTED UNAFFECTED CARRIER

If both parents are carriers

- 25% chance of the baby being unaffected
- 25% chance of the baby being affected
- 50% chance of the baby being a carrier

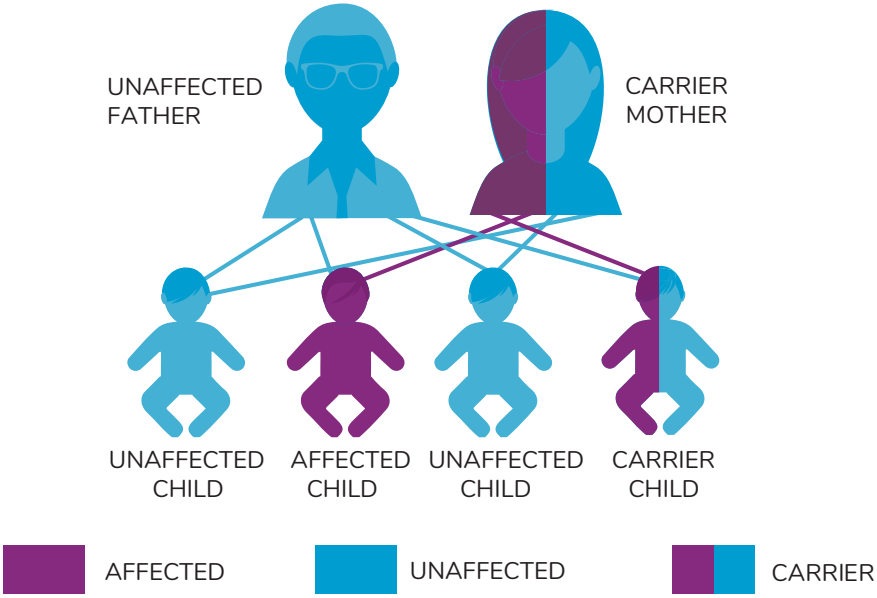
If only one parent is a carrier

- 50% chance of the baby being unaffected
- 50% chance of the baby being a carrier

If one parent is affected and the other parent is a carrier

- 50% chance of the baby being affected
- 50% chance of the baby being a carrier

X-Linked Recessive Disorders



Why screen for inherited genetic conditions?

- Each individual harbours an average of 2.8 known severe recessive mutations.[#]
- Couples may not be aware that they could be carrying a genetic variant, which could lead to their child developing a genetic disorder.
- Carriers are usually healthy or unaffected but, they have a risk of passing on their genetic condition to their children.
- Traditionally, carrier screening has been offered to patients based on their ethnic background or family history.
- However, more than 80% of babies born with inherited genetic diseases have no known family history.*

[#] *Bell CJ, et al. Carrier Testing For Severe Childhood Recessive Diseases by NGS. Sci Transl Med. 2011 Jan 12;3 (650:65ra4)*

[*] *Blythe and Farrell. Advances in the Diagnosis and Management of Cystic Fibrosis. Clinical Biochemistry. 1984*

Who should get tested?

Carrier testing helps determine one's carrier status, and is an important step of the family planning process. The Claria Carrier Screening Test is recommended for all couples planning for a pregnancy. However, the test is strongly recommended in the following cases:

- Either partner is affected by a hereditary disorder
- History of a genetic disorder in the family
- Bad obstetric history
- Couples from consanguineous marriages
- Couples from ethnic groups with high carrier rate of certain genetic disorders
- Congenital anomalies detected in antenatal scans

When should a couple get tested?

A couple can get tested in the following situations:

- Preconception (before pregnancy): when the couple is planning to have a baby (with or without family history)
- Prenatal Diagnosis (early pregnancy): with a history of previous child affected (to know the status of affection in present pregnancy)
- Before a Pre-implantation Genetic Diagnosis (embryo selection)
- Before an IVF procedure (before the use of a donor sperm and/or oocyte)

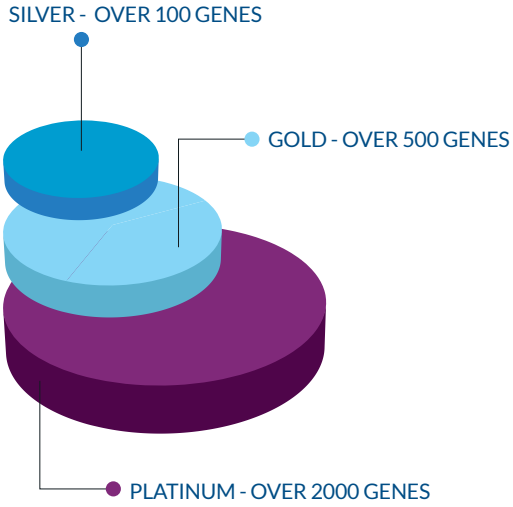
How do we test for these disorders?

The Claria Carrier Screening Test is based on NGS and Multiplex Ligation Probe Amplification (MLPA) technologies. This enables us to detect disease-causing mutations in over 2000 genes which are responsible for the Autosomal Recessive and X-Linked Recessive genetic disorders.

Next-Generation Sequencing uses best in class and superior technology to detect all common and rare disease-causing mutations while MLPA is used to detect deletion and duplication in specific genes (eg. SMN, *CYP21A2*).

Our team of experts adhere to the recommendations of the American College of Medical Genetics and Genomics (ACMG).

With three different panels available (Silver, Gold and Platinum), the Claria Carrier Screening Test offers the option of selecting the version that is best suited to the couple or family.



Representative diagram only

Next-Generation Sequencing vs. Genotyping

	Genotyping	NGS
Usage	Used by many companies for routine carrier screening	Used by a few providers to comprehensively evaluate the gene
Mutation detection	Tests for a limited set of common mutations	Tests for 5-10 times more pathogenic mutations, and detects all common and rare disease-causing mutations
Accuracy	Provides limited utility beyond Caucasian and Jewish ethnicities	Delivers high accuracy across ethnicities
Detection of new pathogenic mutations	Fails	Enables the discovery of rare and novel mutations in a pan-ethnic population
	Fails	Low residual risk, regardless of ethnicity

Why trust the Claria Carrier Screening Test?

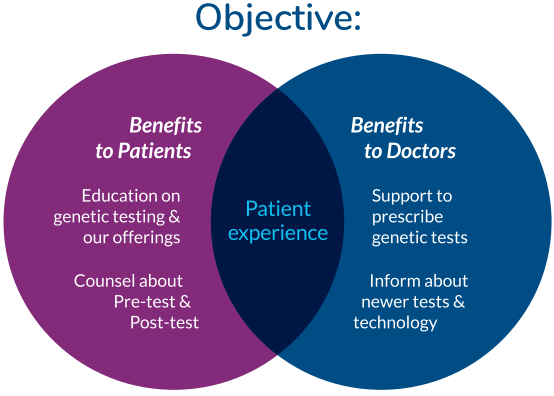
The Claria Carrier Screening Test screens for genetic variations and diseases that are very specific to Indian population.

The unique Claria Carrier Screening Test is based on the NGS technology, a highly focused and cost-effective test to screen for diseases and genetic variations that are very specific to the Indian population.

- Best in class accuracy and easily interpretable reporting
- Detection of all known common and rare disease-causing mutations
- Free expert genetic counselling sessions with certified, multi-linguistic genetic counsellors
- Option of three customized screening panels, covering over 2000 genes associated with AR/XLR disorders
- State-of-the-art labs based out of India, to process all samples with immediate access to the status of the sample
- Higher accuracy and low residual risk, regardless of ethnicity
- Pan-India presence for easy sample collection
- Turnaround time of just 21 days for Silver and Gold panels and 28 days for the Platinum panel

Assistance beyond screening tests. Counselling to all your patients

Free, on-demand pre & post-test genetic counselling to all patients. An information, education & service hub for patients & doctors.



Case Study

Carrier Screening Test aids in confirming the genetic diagnosis

Mr Satish and Ms. Mahima (names changed), a non-consanguineous couple, lost one male child at the age of 1½ years. Based on the clinical and laboratory findings, he was suspected to be affected with leukocyte adhesion deficiency or chronic granulomatous disease. Since the child was no longer available for genetic testing, the couple was advised to go for Carrier Screening Test.

On analysing the couple's DNA sample, a significant mutation in the CYBB gene, was detected in the mother. The CYBB gene causes X-linked recessive Chronic Granulomatous Disease due to CYBB deficiency .

Conclusion

1. Carrier Screening Test helped identify the genetic disorder that caused the death of a child.
2. Carrier Screening Test can help get a final genetic diagnosis even if the affected child is not available for genetic testing. This final confirmation can help in prenatal diagnosis in the next pregnancy. Such couples can screen future pregnancies for the presence of the defective genes in the foetus and make appropriate informed decisions.

Carrier Screening Testing for Premarital counselling

Mr. Arjun and Ms. Shilpa (names changed) are maternal cousins planning to get married. There is no significant clinical history suggestive of any genetic disorder in the family apart from hypertension in their grandparents. Premarital genetic counselling was done, explaining the risk of genetic disorder in their children, since they are first cousins. They agreed to getting themselves tested.

On performing the Carrier Screening Test on Mr. Arjun and Ms. Shilpa, they were each found to be carriers of three deleterious mutations. These mutations cause autosomal recessive primary microcephaly (CEP135 gene), xeroderma pigmentosum (ERCC3 gene) and reticular dysgenesis (AK2 gene). All the three genetic conditions are very severe and the affected child would need medical intervention that could alter the life of the couple.

Conclusion

Premarital Carrier Screening tests and counselling can give insight into the possibility of having a child affected with a genetic disorder even in the absence of a family history. Such couples can make informed decisions regarding their future, as well as decisions regarding family planning after marriage.

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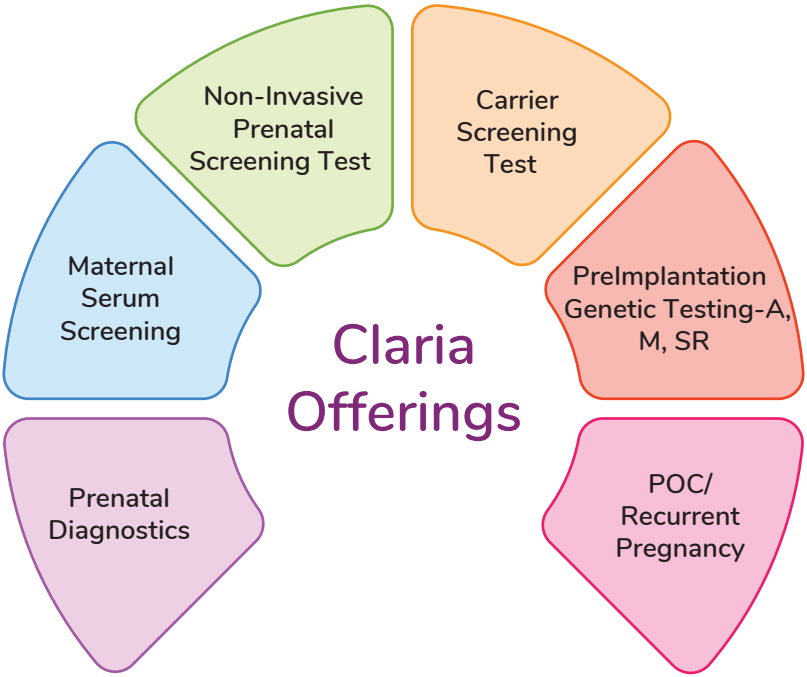
Reproductive Genetics

Precision in Reproductive Genetic solutions



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

Talk to the Experts

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