MEDGENOME | Claria Reproductive Genetics

CAP ACCREDITED
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Genetic testing for

Recurrent Pregnancy Loss

Provide comprehensive clarity on the underlying causes of miscarriage and plan for the future.

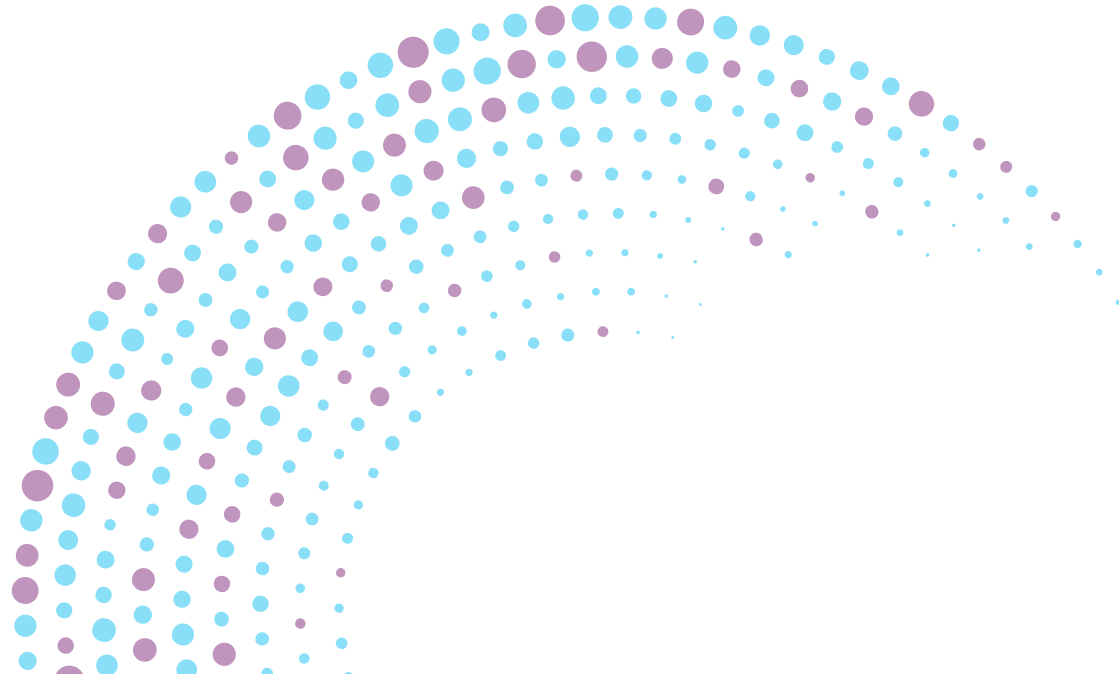
Claria
From MedGenome

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).

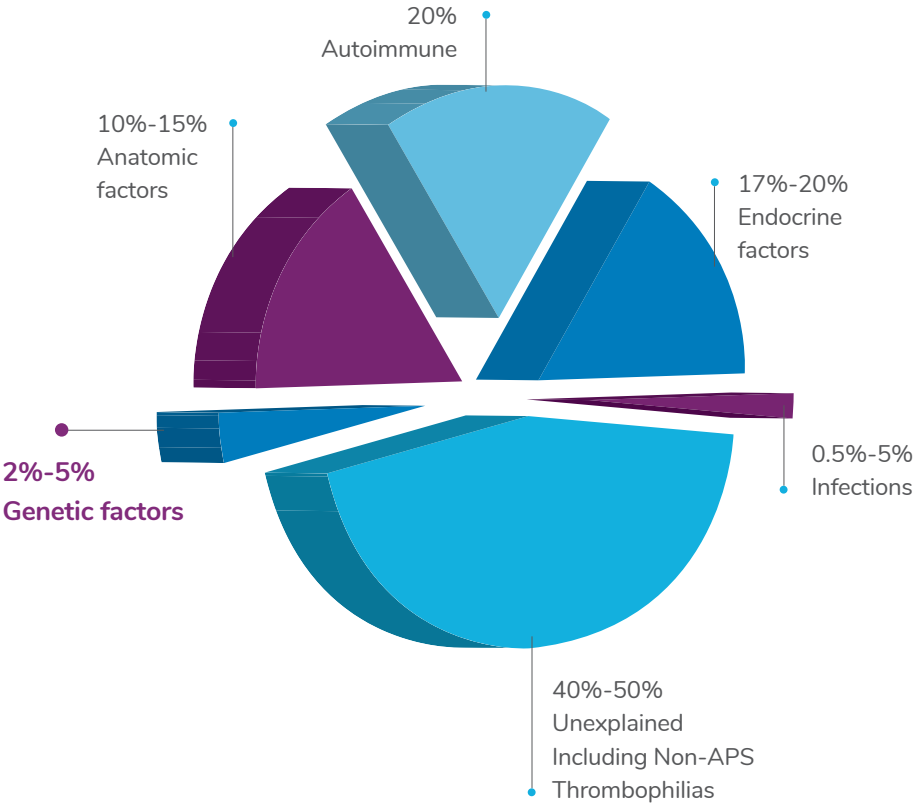
We understand your time is valuable, and that’s why Claria has a team of in-house genetic counsellors to help you interpret and explain reports.

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



What is Recurrent Pregnancy Loss (RPL)?

- RPL is defined as three consecutive pregnancy losses prior to 20 weeks from the last menstrual period[1]
- It is reported in approximately 1% to 2% of pregnant women
- Current techniques can identify up to 50% of couples suffering from RPL.[2]
- Genetic causes account for about 2- 5% of RPL.[1]



1. Ford HB, Schust DJ. Recurrent pregnancy loss: etiology, diagnosis, and therapy. Rev Obstet Gynecol 2009;2: 76-83.

What are the cause of RPL?

- 30% to 50% of all miscarriages are due to Chromosomal abnormalities
- 2% to 4% of RPL is associated with a parental balanced structural chromosome rearrangement, most commonly balanced reciprocal or Robertsonian translocations[1]
- Other reasons include chromosomal inversions, insertions, and mosaicism[1]

Genetic testing for RPL

Consists of testing the products of conception and/or the parents with appropriate pre and post-test genetic counselling.

What is Products of Conception (POC) testing?

POC testing can detect whether the pregnancy had an abnormal chromosome number (aneuploidy) that might have caused a miscarriage. The information from POC testing can be helpful for patients and physicians to understand the cause of miscarriage and to develop a plan to support a future successful pregnancy.

Who should undergo POC testing?

- Any couple who have a pregnancy loss with fetal abnormalities
- Couples who have had a miscarriage
- Couples with recurrent pregnancy loss
- Couples undergoing IVF who have had repeated miscarriage

How is POC testing done?

POC testing was done earlier using Karyotyping and FISH. At MedGenome, we offer the following POC tests:

Test name	Specimen type	Methodology	TAT
POC (All Trimester)	Minimum 20mg of Products of Conception in sterile container in RPMI1640 + 1% Antibiotic. Cardiac/Cord or cord blood in Sodium Heparin Vacutainer	FISH*	4 Working days
		Karyotyping	12 Working days
		QF-PCR	2 Working days
		CMA	10 Working days
		Exome	21 Working days
Parents	2-3 ml blood in Heparin Vial	Karyotyping	12 Working days

*FISH is only performed as a reflex test for POC samples, if cell culture fails.

Parental Karyotype Testing

About 5% of couples with RPL have been known to carry Robertsonian translocations and balanced reciprocal translocations. Both the specific affected chromosome(s) and the types of rearrangement influence the success of pregnancy

Conventional karyotyping can be used in such cases. Standard method can detect most chromosomal abnormalities. Pre and post-test genetic counselling are essential.[4]

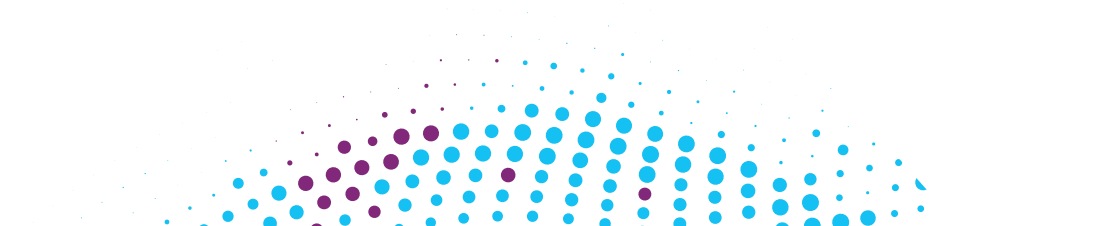
POC using Chromosomal Microarray (CMA)

Recently POC testing is being performed using molecular tests such as Chromosomal Microarray. Claria POC testing is done using the Affymetrix Optima Chromosomal Microarray (CMA)

Advantages of using CMA

- No cell culture is necessary
- Highly sensitive platform with >99% sensitivity for detection of chromosomal deletion/duplications
- The array has whole genome coverage and increase coverage targeting 396 regions relevant for prenatal analysis (18,018 CNV and 148,450 SNP)
- Increased coverage density (25 markers/100 kb) in 396 empirically selected regions relevant for prenatal research
- It can detect low levels of Mosaicism in the sample
- A minimum resolution of 1 MB for losses, 2 MB for gains, and 5 MB for LOH/AOH (Loss/Absence of Heterozygosity)

Specimen Type	Volume
Peripheral blood/purified genomic DNA (RNAse treated)/chorionic villus sample (CVS)/amniotic fluid	Minimum 3ml of peripheral blood; minimum 1 microgram of DNA (concentration of 50-100ng/ microliters); 300-500mg of CVS; Amniotic fluid of 15-20ml/T25 culture flask with 100% confluency



Test sample requirement

Minimum 100mg of Product of Conception (POC)

Taken in a sterile 15ml falcon tube either in a saline solution +1% antibiotic or RPMI1640+10%FBS+1% antibiotic

Formalin fixed or degraded samples will not be accepted

Also provide 3ml of maternal whole blood in a EDTA vaccutainer for Maternal Cell Contamination check.

References:

1. Ford HB, Schust DJ. Recurrent pregnancy loss: etiology, diagnosis, and therapy. Rev Obstet Gynecol 2009;2: 76-83.
2. Jaslow CR, Carney JL, KuttehWH. Diagnostic factors identified in 1020 women with two versus three or more recurrent pregnancy losses. Fertil Steril. 2010;93(4):1234–43.
3. Ogasawara M, Aoki K, Okada S, et al. Embryonic karyotype of abortuses in relation to the numberof previous miscarriages. Fertil Steril. 2000;73: 300–304
4. Regan L, Backos M, Rai R. 2011. Green-top GuidelineNo 17. The investigation and treatment of couples with recurrent first-trimester and second-trimester miscarriage. Royal College of Obstetricians and Gynaecologists (RCOG), London.

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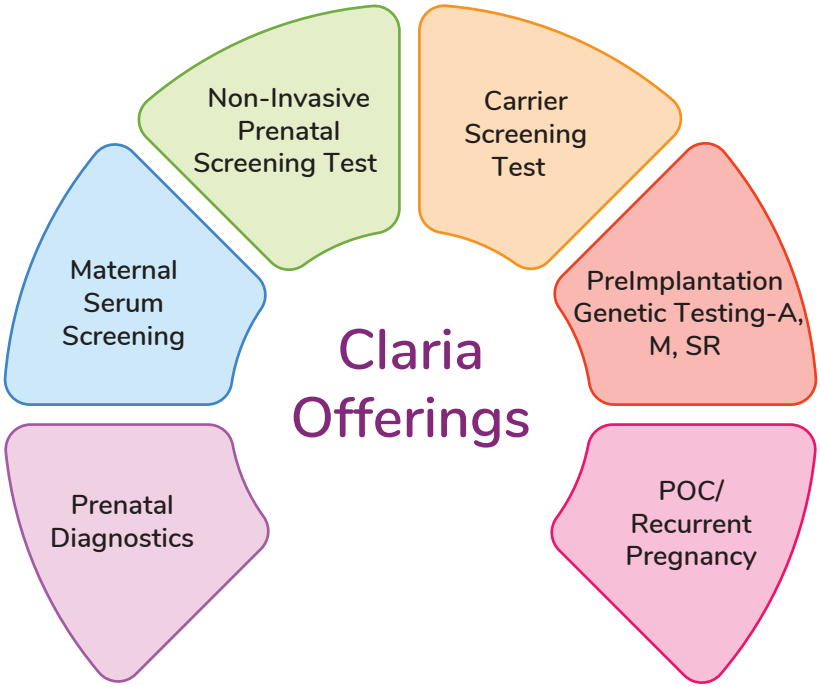
Precision in Reproductive Genetic solutions



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

Talk to the Experts

1800 103 3691 diagnostics@medgenome.com www.medgenome.com