Preimplantation Genetic Testing- Aneuploidies (PGT-A)
Preimplantation Genetic Testing- Monogenic Disorders (PGT-M)
Preimplantation Genetic Testing- Structural Rearrangements (PGT-SR)

Maximise the success of your IVF procedures.
And help transform your patients’ life.
Claria
From MedGenome

MedGenome is driven to enable 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 PreImplantation Genetic Testing-A,M,SR.

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.

Why is screening an embryo before implantation critical?

1. One-in-two human preimplantation embryos from IVF (in vitro fertilized) are chromosomally abnormal.
2. Even up to 40% of morphologically normal embryos harbour aneuploidies.
3. 73% pregnancy rate with PGT-A vs 36% without.
4. Reduces number of IVF cycles the patient has to undergo.
5. Improves the overall success rate of the IVF Center.

What is Preimplantation Genetic Testing- Aneuploidies (PGT-A)?

Preimplantation Genetic Testing- Aneuploidies (PGT-A) is a test that examines the chromosomal material of an IVF embryo before implantation. It involves removing one or more cells from an IVF embryo to test for chromosome number and check for any numerical chromosomal abnormalities (Aneuploidy). This screening method facilitates the selective implantation of embryos that have the normal number of chromosomes (Euploid Embryos).

Why PGT-A?

1. Leads to greater implantation rates and improved IVF outcomes.
2. Reduces the number of IVF cycles required to achieve a successful pregnancy.
3. Increases success rate for single embryo transfer.
4. Reduces the likelihood of miscarriage due to Aneuploidies.
5. Increases reproductive success rates in women above 35 years.

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Who should be offered PGT-A?

- Couples undergoing IVF
- Patients at any age that have repeated implantation failure or recurrent pregnancy loss while undergoing IVF
- Women over 35 years old undergoing IVF
- Couples with recurrent miscarriages
- Positive history of chromosomal abnormalities in the family
- Diagnosed carriers of chromosomal aberrations

When should a biopsy be done?

A biopsy can be done on Day 3 (Blastomere) or Day 5 (Trophectoderm).

A biopsy can be done on Day 3 (Blastomere) However, on Day 5, a frozen embryo transfer is possible, which is why Day 5 biopsy is recommended.

Why is Day 5 biopsy preferred for PGT-A?*

- At this stage, there are sufficient number of cells from which DNA can be isolated thus ensuring success of the test
- Mosaicism of Aneuploidies can be detected at this stage
- Vitrification (rapid-freezing) of embryos after biopsy also allows the clinician to determine the optimum conditions for implantation

Why Claria PGT is better?

- Sequencing based PGT-A lead to higher resolution and detects segmental deletions and duplications
- Single biopsy for PGT-A, M and SR
- CAP accredited and AABA proficiency testing passed
- We provide end-to-end support: From result data interpretation to phenotype correlation and genetic counselling
- Robust sequencing technology that provides sensitive and replicable results.

Claria PGS is carried out using advanced Next-Generation Sequencing (NGS) technology.

What are the advantages of NGS based techniques?

- Rapid and convenient
- Screening of all 23 pairs of chromosomes for abnormalities in one test
- Able to detect greater than 4Mb gains and losses in chromosomes
- Higher resolution - 1Mb areas are analysed to provide data with high confidence
- High sensitivity in detecting Aneuploidy (100 % sensitivity)
- High specificity and accuracy (99.98% specificity)
- Lower chances of test failure with NGS

Externally validated results

MedGenome’s PGT-A test has shown 100% accuracy in Aneuploidy detection, when external validation was done using proficiency testing (PT) samples by the American Association of Bioanalysts.

MedGenome’s PGT-M has shown 100% accuracy in mutation detection when externally validation was done using proficiency testing (PT) samples by QualiGene Ltd.

Advantages of PGS by NGS vs. other screening approaches

<table>
<thead>
<tr>
<th>Fluorescent In Situ Hybridisation (FISH)</th>
<th>Array Comparative Genomic Hybridisation (aCGH)</th>
<th>Single Nucleotide Polymorphism (SNP) Microarray</th>
<th>NGS</th>
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</thead>
<tbody>
<tr>
<td>A largely manual process, highly skill/ operator dependant</td>
<td>Requires control DNA for each sample to provide a comparison. A prolonged hybridisation step</td>
<td>SNP array analysis of DNA, extracted from a cell population, cannot indicate the mosaicism within the sample.</td>
<td>NGS detects partial chromosomal gains and losses more accurately</td>
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<td>Levels of mosaicism not detected.</td>
<td>Low level of mosaicism not detected</td>
<td>NGS detects Aneuploidy and segmental imbalances at the same time.</td>
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<tr>
<td>Highly expensive</td>
<td>Relatively expensive.</td>
<td>Relatively expensive.</td>
<td>NGS provides more accurate detection of mosaicism of the Trophectoderm cells from blastocyst biopsy.</td>
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<tr>
<td>Highly expensive</td>
<td>Relatively expensive.</td>
<td>Relatively expensive.</td>
<td>NGS offers reduced costs and enhanced precision. It allows parallel analysis for multiple embryos for a single patient.</td>
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What can Claria PGT-A detect?

- Turner Syndrome
- Klinefelter Syndrome
- Down Syndrome
- Edwards Syndrome
- Patau Syndrome
- Other trisomies and monosomies which could increase the risk of implantation failure and miscarriage
- Segmental Gain and Losses (>4Mb) in chromosomes which can lead to abnormalities in the embryo

Claria PGT workflow

Case study

36 years old woman realizes dream of motherhood through PGS technology

Patient information

Mrs Swamy (36), (name changed) a house wife and her husband have been wanting a baby since they got married in 2006. Unfortunately, each time she got pregnant, it wouldn’t last more than 6-7 weeks. They tried to conceive through Intra Uterine Insemination (IUI) process 7 times and once through In Vitro Fertilisation (IVF) in UK and India respectively.

Previous genetic testing

On investigation it was found that the miscarriages occurred due to aneuploidies in the foetus. While there was no family history of chromosomal abnormalities, advanced maternal age was considered as one of the contributing factors.

Doctor Recommendation

She was recommended Intra Cytoplasmic Sperm Injection (ICSI) along with Pre-implantation Genetic Testing- Aneuploidy (PGT-A) for the Embryos that developed.

Genetic testing at MedGenome

In the next IVF cycle 11 embryos were screened using PGT-A at MedGenome Labs. The report provided by the company recommended the best embryos for transfer.

Implications of the test

After a 12 year long struggle the couples dream of becoming parents became a reality as they were blessed with a baby.

Summary

PGT-A is an advanced genetic testing technique, which screens IVF embryos for numerical chromosomal defects (known as Aneuploidies) prior to implantation. This allowed the clinician to choose normal (known as Euploid) embryos for transfer. By using PGS the chances of having a successful IVF pregnancy increases from 40% to 70%.
Preimplantation Genetic Testing-Monogenic (PGT-M)

What is PGT-M

PGT-M is a diagnostic procedure to test the material collected from an embryo for the presence of mutations carried by one or both parents. This is carried out when one or both genetic parents has a known genetic abnormality. In PGT-M every test is prepared on a case-by-case basis.

Possible candidates for PGT-M

- Carriers of X-linked genetic disorders
- Carriers of single gene disorders
- Couples who have a child/children affected by a single gene disorder
- Couples who have a family history of a single gene disorder

What are the benefits of PGT-M?

- PGT-M can test for most single gene disorders
- PGT-M allows the clinician to select embryos that do not carry the single gene disorder being tested for the implantation
- By using PGT-M, the single gene disorder can be prevented from being passed on from one generation to the next

How to order the Embryo Biopsy Kit

Contact Customer Support
MedGenome Labs Pvt. Ltd., Bangalore
Ph 91-80-67154990/91
(At least 2 days in advance)

- Clearly indicate number of kits required

- Kits will be transported to the provided address at room temperature

- Ensure the contents of the kit are intact before proceeding with the biopsy. Refer to the instructions on the kit.

For further queries or clarification, contact customer care
Claria from MedGenome offers the complete range of Reproductive Testing solutions

Non-Invasive Prenatal Screening Test

Carrier Screening Test

Cytogenetic Testing

Prenatal Diagnostics

Pre-implantation Genetic Screening/ Diagnosis

POC/ Recurrent Pregnancy Loss Testing

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