

Preimplantation Genetic Testing- Monogenic Disorder Workflow Options

Preimplantation Genetic Testing- Monogenic Disorder (PGT-M) is genetic screening technique used to select embryos free of the genetic variation(s) that have been confirmed(carrier/affected) in prospective parents.

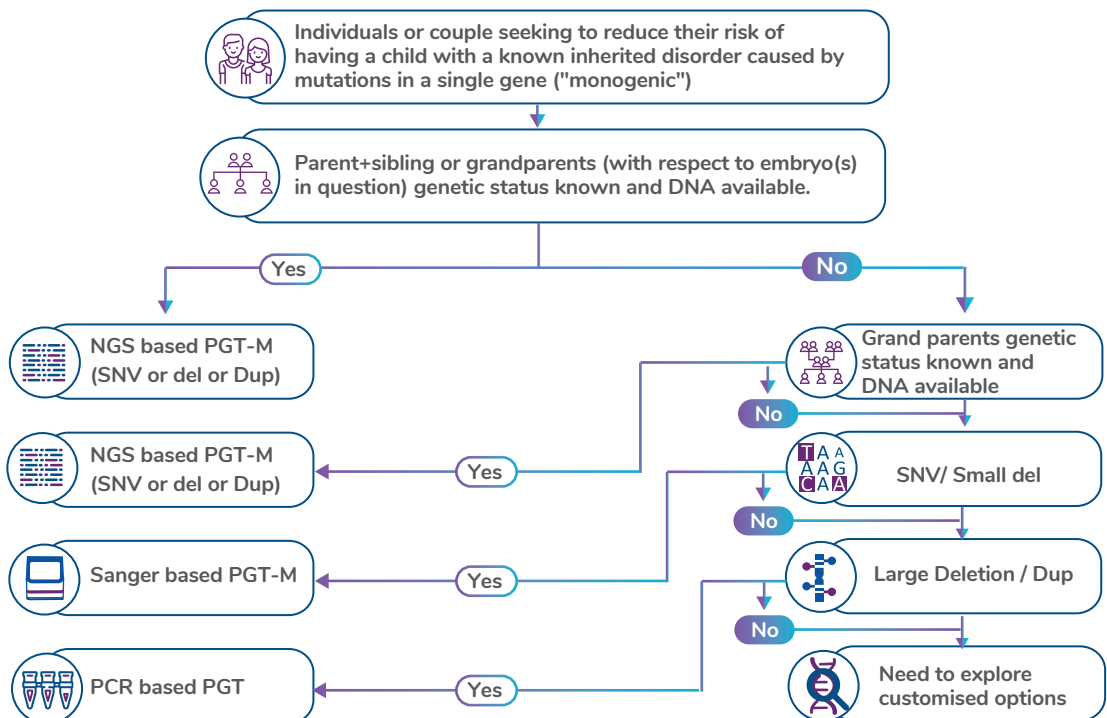
Who is PGT-M testing for

- 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

Benefits of PGT-M

- It can test for most single gene disorders (Autosomal/X-linked dominant and recessive)
- It allows the clinician to select embryos that do not carry the single gene disorder for implantation
- It helps prevent the passing on of single gene disorders to the next generation
- Whole gene deletions and genes with pseudogenes can also be tested along with missense and small indels variants

PGT-M Treatment Flow



Claria PGT-M workflow



Genetic assessment

- The patient's family and genetic history are assessed
- Genetic testing is done to identify the variants in the couple and affected child (if any)



Pre-PGT-M work-up

- PCR primers are designed for known mutations(s) obtained from the genetic assessment
- Whole Genome Amplification(WGA) is carried out on the couples blood using the primer to check for mutations
- PGT-M, haplomap is created using the SNP information 2Mb upstream and downstream from the region of variant



In Vitro Fertilisation*

Embryos obtained from IVF of the patient are incubated



Embryo biopsy*

A small sample of cells is taken from the embryos on Day 3 (Blastomeres) or Day 5 (Trophectoderm)



Sample collection and transportation

An embryo biopsy is collected using the kit provided by MedGenome and is shipped to the MedGenome lab in Bangalore in dry ice.



PGT-M analysis carried out

Embryo biopsy is analysed for the presence of mutations



Report generation and interpretation

Our highly qualified scientific team will help interpret the results and make recommendations on embryo transfer

PGT-M can be combined with Pre-Implantation Genetic Screening (PGS) to offer screening for Chromosomal Aneuploidies along with mutations.

*Carried out at the IVF center



Embryo transfer*

Recommended embryos are transferred



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