# Preimplantation Genetic Testing-Monogenic [PGT-M]

Preimplantation genetic testing is performed on embryos created through in-vitro fertilization (IVF). An embryo is created after an egg is fertilized by a sperm. After fertilization, embryos develop from a single cell into a cluster of around 200 cells. In order to perform the genetic analysis, a few cells are biopsied from the embryo and sent to a genetic testing laboratory. The goal of PGT-M is to select and implant embryos that do not have the genetic condition.

**Mono = one  genic = gene**

### Understanding Monogenic Conditions

Genes are like instruction manuals for our bodies. They provide important information on how we grow and develop. We inherit two copies of each gene, one from our mother or egg donor, and the other from our father or sperm donor. When a gene has a change in its code, it may not work properly and can lead to health problems. Monogenic conditions are caused by changes in a specific gene.

### Cystic Fibrosis (CF) as an Example

Cystic Fibrosis (CF) is an example of a monogenic condition. CF affects the lungs and digestive system because a gene called CFTR cannot carry out its intended function of regulating the movement of substances in and out of cells. If both copies of the CFTR gene have a change in their code, the person will have cystic fibrosis. If only one copy has a change, the person is a carrier of the condition. If neither copy has a change, there is no risk of cystic fibrosis.

### What does it mean to be a carrier?

A carrier is someone who does not have the condition but carries one copy of a gene with a change. They can pass this gene to their children. It's important to note that being a carrier is generally not a health concern unless the partner or donor is also a carrier for the same condition. When both members of a reproductive pair are carriers, there is a 1 in 4 chance with each pregnancy to have the condition. This is called autosomal recessive inheritance.

### How can PGT-M be helpful?

PGT-M is a test that screens embryos for specific genetic conditions. It allows the selection of embryos that do not have the condition to be transferred during IVF. However, it's important to know that PGT-M cannot test for all genetic conditions. To test for a specific condition, the exact genetic information needs to be known, so that a unique test can be created for the embryos. This process takes time and may require involving other family members to design a personalized test for the family.

PGT-M is also considered a screen, so while unlikely, there can be false negatives and false positives. Therefore, testing in pregnancy or after a baby is born can still be considered.

For more information, please speak to your healthcare provider.