



## Preimplantation Genetic Testing at ONE Fertility: PGT-M and PGT-SR

**Preimplantation Genetic Testing (PGT)** – involves the biopsy and testing of cells from embryos created through IVF prior to transfer into the uterus (ie before implantation).

**PGT-M** is Preimplantation Genetic Testing for Monogenic/Single Gene known defect (also known as PGD). It involves the screening of embryos for a specific **known** gene defect. Disorders such as Cystic Fibrosis, Fragile X Syndrome, Huntington’s Disease and Sickle Cell Anemia are only a few disorders that can be identified through PGT-M. **PGT-SR** is Preimplantation Genetic Testing for Chromosome Structural Rearrangements. Examples include balanced reciprocal translocations, Robertsonian translocations, inversions, and other complex chromosome rearrangements.

### **What is the Purpose of PGT-M and PGT-SR?**

Without **PGT** embryos are selected for transfer based on their physical appearance and development observed under the microscope. It is important to note that the appearance of the embryo does not tell us if the embryo is genetically normal. **PGT-M and PGT-SR** allows us to identify abnormal embryos and therefore can give your physician additional information to help **SELECT** the normal embryo for transfer to maximize the chance of having a successful pregnancy and a baby.

### **Who may benefit most from PGT-M**

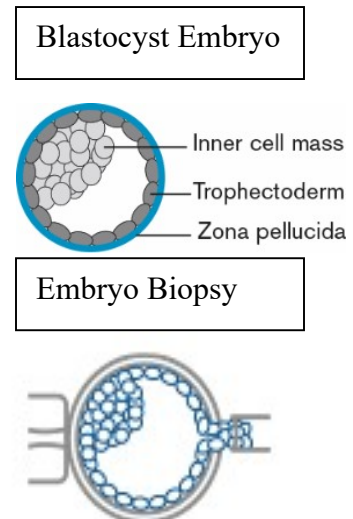
- Carriers of a known genetic defects
- Individuals who have previously had a pregnancy or child with an inherited genetic disease

### **Who may benefit most from PGT-SR**

- Individuals with chromosomal rearrangements (ie translocations)

### **How is the testing performed?**

Five to six days after egg retrieval, well developed embryos (called blastocysts) will have over 100 cells. A few cells are carefully removed from the trophectoderm which is the part of the embryo that will form the placenta. These cells are sent for genetic analysis to an outside lab and the embryos are frozen for future use. Embryos may not qualify for testing if they do not reach the blastocyst stage or if it is not technically possible to perform the biopsy due to insufficient quality of the outer layer of cells. Embryos for biopsy must be large enough for biopsy and have a trophectoderm quality grading of A or B. Some embryos may therefore not reach a developmental stage for testing. Even biopsied embryos are occasionally not suitable for analysis by the lab. You will need to give us direction as to your wishes for the management of embryos that are not suitable for biopsy.



### **Results – What to expect?**

The percentage of healthy embryos generally varies from person to person. Unfortunately, sometimes there are no healthy embryos for transfer. This technology gives us the best chance of transferring a normal embryo. However, transferring a normal embryo does not ensure the pregnancy will occur. It does decrease the risk of pregnancy loss, though a loss could still occur.

### **Are there any risks?**

There is no guarantee that biopsied embryos will survive. The risk of loss of the embryo is low. In addition there remains a small risk that the results will not be accurate. For this reason, screening for genetic disorders in pregnancy is still recommended.

## Sequence of Events

Discuss IVF and PGTM/SR with your Physician



### **Enroll for PGT with outside lab**

(ONE Fertility will complete an enrollment form and send it to outside lab along with your relevant test results)



### **Genetic Counseling**

A genetic counselor will phone you. Additional testing may be required of you or other family members.



### **Test Preparation for PGT-M only**

The PGT Lab will design a unique test for you. This can take some time depending on the test required. It is very important **NOT TO START IVF** until your unique test is created.



### **Prepare for IVF**

1. Do the testing that was ordered by your Physician (when applicable)
2. Attend IVF teaching
3. Sign IVF and PGT consents



**Call ONE Fertility on DAY 1 of your period the MONTH BEFORE IVF** and speak to an IVF nurse to:

1. Make sure your unique test is ready for PGT-M only
2. Review Estrace priming instructions
3. Arrange a follow up visit after your IVF

*Call 905-634-4440 ext 4. Messages left on weekends/holidays will be returned on next business day*



Begin Estrace priming as directed  
Book IVF Stimulation start date as directed by the IVF nurse



Begin IVF Stimulation



Egg Retrieval and Fertilization



Biopsy of the Embryo(s) - Day 5/6 of embryo development  
Embryos for biopsy must be expanded enough for biopsy and have a trophectoderm quality grading of B or better.



Sample sent for biopsy to outside lab



Embryos frozen for transfer in a future frozen embryo transfer (FET)



Visit with your Physician where you will review the outcome of your IVF treatment including PGT results, plan your next steps and sign appropriate consents



*Genetic Counselling - This step may be required or optional depending on your outcome. Your Physician will advise you.*



Frozen embryo transfer with appropriate embryo