



## Enrolling a Patient for Preimplantation Genetic Diagnosis (PGD) Single Gene Disease

This protocol is for PGD. PGS (24-chromosome microarray CGH) is a different protocol.

### IVF-Reproductive Center Steps

- Register the Couple:** Complete our basic demographic intake information form for the couple. This acts as a physician's order for us to enroll them into the program.
- DNA Diagnostic Report:** You should have a DNA diagnostic report(s) in the patient's notes. For example, a cystic fibrosis mutation report defining the gene mutation(s). [If you do not have one, we can assist you in obtaining it from the university/commercial laboratory that determined the molecular basis of the inherited disorder for which the couple is at risk]. We use this data to custom design the molecular probes necessary for their single-cell PGD testing.

***What does the Genesis Genetics Institute do next, once you have referred a case for PGD?***

### Genesis Genetics Institute Steps

- Collect and Review Genetic Information**
  - We contact the patient, introduce Genesis Genetics to them, and explain that we are working with your IVF team to assist them in building a healthy family.
  - Our Certified Genetic Counselors then take a Genetic History, review the existing DNA information, and answer any initial questions.
  - The patient pays up front for the PGD probes to be designed.
- Obtain DNA Samples**
  - Based on the family pedigree our Counselors obtain, we send a buccal swab kit to the family members necessary to construct the DNA probes for the PGD testing. There is no need for you (the IVF clinic) to draw blood - we handle collecting DNA for you.
  - We mail the couple a pack of information about Preimplantation Genetics Diagnosis, including a DVD for them to watch together, as part of the Informed Consent process.
- Design, Construct and Optimize Probes**
  - EACH and EVERY PGD case is customised for each couple. We start immediately after receiving the necessary cheek swab samples and payment for this several week process.
- Notification that the Test is Ready**
  - The genetic counsellor assigned to the patient contacts the couple and you, the IVF Center, once the probes are constructed, optimized and certified for the couple's PGD case. It is important you wait for this before starting the couple's treatment.
- Our Laboratory then waits until you are ready to send us the embryo biopsy samples**
- Send Your Center a Biopsy Kit**
  - When you tell us an approximate egg retrieval date, we schedule a FedEx delivery of the biopsy kit to your laboratory. The buffers are stored in your freezer until needed on the day of biopsy.
- Arrange Courier Transport**
  - We arrange courier transportation of the biopsy samples for you. You do not have to complete any forms or pay for any shipping. *Genesis Genetics handles all of this for you.*



## “Genesis-24” Preimplantation Genetic Screening (PGS) 24 chromosomes in 24 hours

This protocol is for PGS (24-chromosome microarray CGH). Single Gene PGD is a different protocol.

### IVF-Reproductive Center Steps

- Register the Couple:** Complete our basic demographic intake information form for the couple. This acts as a physician’s order for us to enroll them into the program.
- Report Documents:** We do NOT need any “chromosome (karyotype) reports” for PGS, unless the case involves a chromosome translocation. If it does, then please send that formal cytogenetic report to us. No probes or preliminary testing are required for translocation patients so you can enroll them at your convenience. Normally our office contacts the patient directly via email with our PGS Informed Consent and financial information. However, if you would like to collect the consent forms yourself please let us know and we are happy to accommodate. We MUST have the signed Informed Consent document before we can process the samples.
- Timing:** Once you know an approximate date for egg retrieval, let us know. You can do this electronically *via* the Genesis Genetics portal. [unlike PGD where gene probes need to be designed and optimized, in PGS you can order 24-chromosome testing at any time, even just before retrieval including for translocation cases].

***What does the Genesis Genetics Institute do next, once you have referred a case for PGS?***

### Genesis Genetics Institute Steps

- Send Your Center a Biopsy Kit:** When you tell us an approximate egg retrieval date, we schedule a FedEx delivery of the biopsy kit to your laboratory. The buffers are stored in your freezer until needed on the day of biopsy.
- Arrange Courier Transport:** We arrange courier transportation of the biopsy samples for you. You do not have to complete any forms or pay for any shipping. *Genesis Genetics handles all of this for you.*
- PGS using Microarray CGH:** Genesis Genetics performs 24-chromosome analysis in 24 hours from receipt of the sample, providing the data in time for the embryo transfer. The timing is exactly like single-gene PGD. We can also perform expedited testing in 24 hours on blastocyst biopsies (Day 5) in time for a fresh transfer on Day 6.
  - We can test for both single gene mutations (PGD) and 24-chromosomes (PGS) in the same sample [same polar body or blastomere or trophoctoderm]. It is not necessary to take two biopsies. However, please remember that if single-gene PGD is required too, we first have to build the probes.