



## Preimplantation Genetic Diagnosis (PGD) For Single Gene Disorders

### PRIOR TO YOUR IVF CYCLE:

1. **Genetic Counseling:** We require that you have a phone consultation with Dr. Mark Hughes (Genesis Genetics Institute, Detroit, MI, <http://www.genesisgenetics.org>), prior to undergoing PGD. To schedule a phone consultation, please contact Dr. Mark Hughes at (313) 544-4006.
2. **Genetic Reports and Patient Blood Samples:** Copies of previous genetic reports are required for each couple undergoing PGD and should be faxed to Dr. Mark Hughes at (313) 544-4006. For this purpose, we require blood samples from both of the prospective parents, and the affected child(ren) if applicable. The DNA will be extracted from the blood and the necessary tests will be performed to confirm the presence of a mutation and tailor the PGD test to the needs of the individual patient. This process requires 4-6 weeks or more. It is essential that Dr. Mark Hughes receives the genetic reports and blood samples before you commence your cycle. Dr. Mark Hughes will contact you as soon as the test is ready, after which you may begin your IVF/PGD cycle.  
  
*To schedule blood draws*, phone the IVF Coordinator's office at (503) 418-3700. Your credit card number will be needed to ship the samples.
3. **PGD Consent Forms:** We require that you sign our PGD consent form and Dr. Mark Hughes' PGD consent form. Please bring them back to our clinic as early as possible.
4. **ICSI for PGD of Single Gene Disorders:** We will utilize intracytoplasmic sperm injection (ICSI) in all cases for PGD for single gene disorders, regardless of whether or not there is any evidence of male factor infertility. The reason for this is to avoid the presence of surplus sperm (and their DNA) in the embryo cultures. These sperm could potentially lead to DNA contamination; therefore, lead to a misdiagnosis. ICSI avoids the presence of surplus sperm.

### SCHEDULING THE IVF CYCLE:

You will not be able to start your cycle until our preliminary analyses are complete and we have proven that the test is working properly. Results are usually available within 6-8 weeks. We will notify you as soon as these analyses are complete so that you can begin treatment. Your cycle start will be coordinated with our staff that can be reached at (503) 418-3700.

### AFTER START OF THE CYCLE:

1. **Biopsy Update:** On day-3 (3-days after egg retrieval), Dr. Nadia Ouhibi will review your embryo and biopsy reports. Please make sure to give us your phone number(s) to contact you that day.
2. **Shipping Expense:** The biopsies will be sent via Express Air courier to Dr. Mark Hughes' laboratory, in Detroit, MI, at patient expense.
3. **Results:** On day-5, the genetic results will be available to us from the laboratory. We will discuss the results with you prior to your embryo transfer time.

## Preimplantation Genetic Diagnosis (PGD) For Aneuploidy, Chromosome Translocation or X-Linked Disorders

### PRIOR TO YOUR IVF CYCLE:

#### Aneuploidy and X-Linked Disorders

1. **Genetic counseling:** We require that you have a consultation with one of our genetic counselors prior to undergoing PGD. To schedule a consult, phone 503-418-3700. If you are from out-of-town, you can arrange a consult with a genetic counselor referred by your physician.
2. **PGD Consent Form:** We require that you sign our consent form and return it to our clinic as early as possible.

#### Chromosome Translocations

1. **Genetic Counseling:** We require that you have a consultation with one of our genetic counselors prior to undergoing PGD. To schedule a consult, phone 503-418-3700. If you are from out-of-town, you can arrange a consult with a genetic counselor referred by your physician.
2. **PGD Consent Form:** We require that you sign our consent form and return it to our clinic as early as possible.
3. **Blood Draw:** For translocation carrier patients, the affected individual(s) and the partner will need 10 cc of peripheral blood to be drawn in a green-top (sodium heparin) tube in order to construct the probes specific to your translocation. To schedule blood draws, phone the IVF Coordinator's office at 503-418-3700.

### SCHEDULING THE IVF CYCLE:

You will not be able to start your cycle until our preliminary analyses and screening tests have been completed and we have proven that the probes work properly. For translocation carrier patients, the probes may take 6 to 8 weeks after the blood is collected. We will notify you as soon as these analyses are complete so that you can begin treatment. Your cycle start will be coordinated with our staff that can be reached at 503-418-3700.

### AFTER THE START OF THE CYCLE:

1. **Biopsy Update:** On day-3 (3-days after your egg retrieval), Dr. Nadia Ouhibi will review your embryo and biopsy reports between 11 a.m. and 12 o'clock Noon. Please make sure to give us your phone number(s) to contact you that day.
2. **Results:** On day-5, the genetic results will be available to us. We will discuss the results with you prior to your embryo transfer time.