



# **Pre-Implantation Genetic Diagnosis:** *A Family Overview*

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# Preimplantation Genetic Diagnosis (PGD)

A family overview



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It's natural to have questions. We know our families come to us with varying levels of knowledge.

We will attempt to outline the basics of PGD, answer questions that are posed to us frequently and provide additional contact information so you can follow up personally with one of our Genetic Counselors.



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## What is PGD?

Testing technique that allows scientists to diagnose a genetic disease from a single cell of an embryo before transfer occurs

By performing this diagnostic test, we are able to tell couples who previously had a 25% - 50% chance of passing on a genetic disease if the embryo is affected by the disease... with about 98% accuracy.



## History of Genesis Genetics...

Genesis Genetics was founded by Dr. Mark Hughes. Dr. Hughes was the physician scientist who first pioneered PGD almost two decades ago.

Genesis Genetics is known world-wide for its expertise in performing PGD on some of the rarest diseases as well as for more frequently occurring mutations. This expertise is globally recognized and Genesis Genetics works with all the major IVF programs and universities in the USA and abroad.



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## How does the test work?

1. We review your mutation report and discuss the process with you.
2. You will be asked to use simple cheek swab kits so that we can build a custom test (or probe) for your family.
3. Once the probe is built, we optimize it and then let you know we are ready for you to begin IVF treatment.

Whether you have an infertility concern or not, you will undergo IVF. Once eggs are retrieved and fertilized, they are allowed to grow in an incubator. On Day 3 of being in the incubator, most embryos have grown to 6-10 cells.



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## How does the test work?

A highly trained embryologist then biopsies a single cell from the embryo. The embryo is put back into the incubator and the single cells are sent to our laboratory in Detroit.

Less than 24 hours after receiving the cells, our scientists have determined which embryos are affected, unaffected or carriers of the disease of concern. This information is shared with your IVF clinic. Your physician and you then decide which embryos to transfer.



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## How long does it take?

From the time we receive your DNA samples (the cheek swab kits) to the time we call to tell you to begin IVF treatment, you can expect the process to take 4-12 weeks.

Time varies because each test we build is custom made for the family. More complicated cases may take longer.

This part of the process is critical to success. While we want to make sure your test is completed in a timely manner, we also want to make sure an appropriate amount of time is spent optimizing it.



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## How much does it cost?

PGD at Genesis Genetics adds \$3,750 to your cost of IVF for your first cycle. This includes building and optimizing the custom probe, transporting the cells from your IVF clinic to Detroit, and testing as many cells as are retrieved from the embryos.

Additional cycles have a cost of \$2,750.



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## How reliable is PGD testing?

Couples with a 25% - 50% likelihood of passing down a genetic disease to their offspring reduce this risk to closer to 2%.



## How do we choose a clinic?

There are wonderful clinics who have extensive experience with IVF with PGD. It is very important that you select a clinic with this type of experience.

It is also critical that you have an experienced embryologist perform the embryo biopsy.

We can help you find a clinic in your area. There is no reason to travel away from home for this treatment.



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What next?

We would be pleased to share additional information with you by sending our Family DVD to you. This includes a conversation with Dr. Hughes as well as a segment from a program produced by the BBC which highlighted one of our families.



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## Von Hippel Lindau Syndrome

As you know....

VHL is inherited in an autosomal dominant fashion.

An affected individual has a 50% chance of passing the disease to each of their offspring.

The gene known to cause VHL is located on the short arm of chromosome 3.

We have performed PGD for a number of families affected with VHL.



## For the PGD handouts . . .

- DVD of BBC interview with Dr. Hughes
- Literature from Genesis Genetics

Send your request to: [office@vhl.org](mailto:office@vhl.org)  
with your postal mailing address

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# Teamwork for Health

Families and Health Care Professionals  
working together  
to improve diagnosis, treatment, and  
quality of life  
for individuals and families  
affected with von Hippel-Lindau

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## VHL Family Alliance