PREIMPLANTATION GENETIC DIAGNOSIS (PGD)

SINGLE GENE DISORDERS

A PATIENT GUIDE

Reproductive Genetic Innovations, LLC
2910 MacArthur Boulevard
Northbrook, Illinois 60062
Phone: (847) 400-1515
Fax: (847) 400-1516
Email: info@rgipgd.com
www.rgipgd.com

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REPRODUCTIVE GENETIC INNOVATIONS, LLC (RGI)

Directors:

Dr. Svetlana Rechitsky, PhD	President, Laboratory Director
Dr. Anver Kuliev, MD, PhD	Research Director
Dr. Joe Leigh Simpson, MD, FACMG	Clinical PGD Director
Dr. Lee P. Shulman, MD, FACMG	Medical Director
Dr. Jeffrey Dungan, MD, FACMG	Medical Director

Certified Genetic Counselors:

Divya Shah, MS, LCGC
Agnes Machaj, MS, GC
Savanie Maithripala, MS, GC

PREIMPLANTATION GENETIC DIAGNOSIS/SCREENING (PGD/PGS) AT RGI

Reproductive Genetic Innovations, LLC (RGI) performs preimplantation genetic diagnosis (PGD) and preimplantation genetic screening (PGS) for the purpose of aiding individuals at risk for genetic diseases and disorders before birth. There are a number of reasons couples choose to pursue PGD and/or PGS testing. These couples may already have had a child with a genetic condition or have had a previous pregnancy that was chromosomally abnormal. Those who already have children with a genetic condition may also try to have a baby who is an HLA match to their child and can therefore act as a bone marrow or stem cell donor. Some choose to pursue PGD because one of the partners carries a balanced translocation which places their pregnancies at a high risk for miscarriage or abnormal outcome. Other couples will choose PGS due to an increased risk for Down syndrome and other chromosome abnormalities due to advancing maternal age. RGI can assist you in your family planning by offering genetic counseling regarding PGD/PGS and how it fits into the process of In Vitro Fertilization (IVF). This packet will assist you in understanding PGD and IVF for single gene conditions.

Please keep in mind that there are parts of this packet that may not apply to you depending on your reasons for pursuing PGD. If you have questions about the process, please feel free to contact a genetic counselor.

IN THIS PACKET…

PGD/PGS OVERVIEW

RGI offers PGD to families who are at increased risk of having children with genetic disorders for a variety of reasons. The purpose of PGD is to reduce the chance of having an abnormal pregnancy and therefore save families from the stressful decisions that come with receiving a prenatal diagnosis.

For over 20 years, we have helped many families and over 2,000 babies have been born who were unaffected for chromosomal and single gene disorders. We have performed PGD for over 300 single gene disorders, and are able to do PGD for most genetic disorders that have an identified associated gene or mutation identified in the family. We can also test for HLA status, on its own or in addition to a single gene disorder.

In order to do PGD/PGS, In Vitro Fertilization (IVF) is required to obtain the eggs and embryos that are used for the testing itself. IVF is an assisted reproductive technology (ART) procedure that involves fertilizing the egg outside of the body in a controlled setting. Depending on the testing method, the embryo is tested at various points in development and either transferred back into the uterus or frozen for a later transfer (see pages 11-13). This way we can identify genetically abnormal embryos before transferring them. This is what makes PGD/PGS such a valuable alternative for couples and families who are at risk for a genetic condition. Prenatal diagnosis by chorionic villus sampling (CVS) or amniocentesis is still encouraged to be performed during pregnancy, in order to confirm the PGD/PGS results (see page 15).

If you do not live in proximity to our laboratory, it is possible for us to work with your local Reproductive Endocrinologist for your cycle in order to reduce or completely eliminate your need to travel to the Chicagoland area. If you do not have a local specialist you desire to work with or prefer to travel to the Chicagoland area for treatments we can assist you in referring you to an affiliated IVF center/physician (see page 17 for more details).

Please note:
It may be necessary to obtain DNA samples (blood or cheek swabs) and/or genetic test reports from your relatives in order to provide the most accurate diagnosis. In certain circumstances, PGD may not be feasible without DNA samples or specific medical reports. This will depend on the particular disorder or gene mutation in the family.

This information packet will discuss the options available to you for testing for single gene disorders.

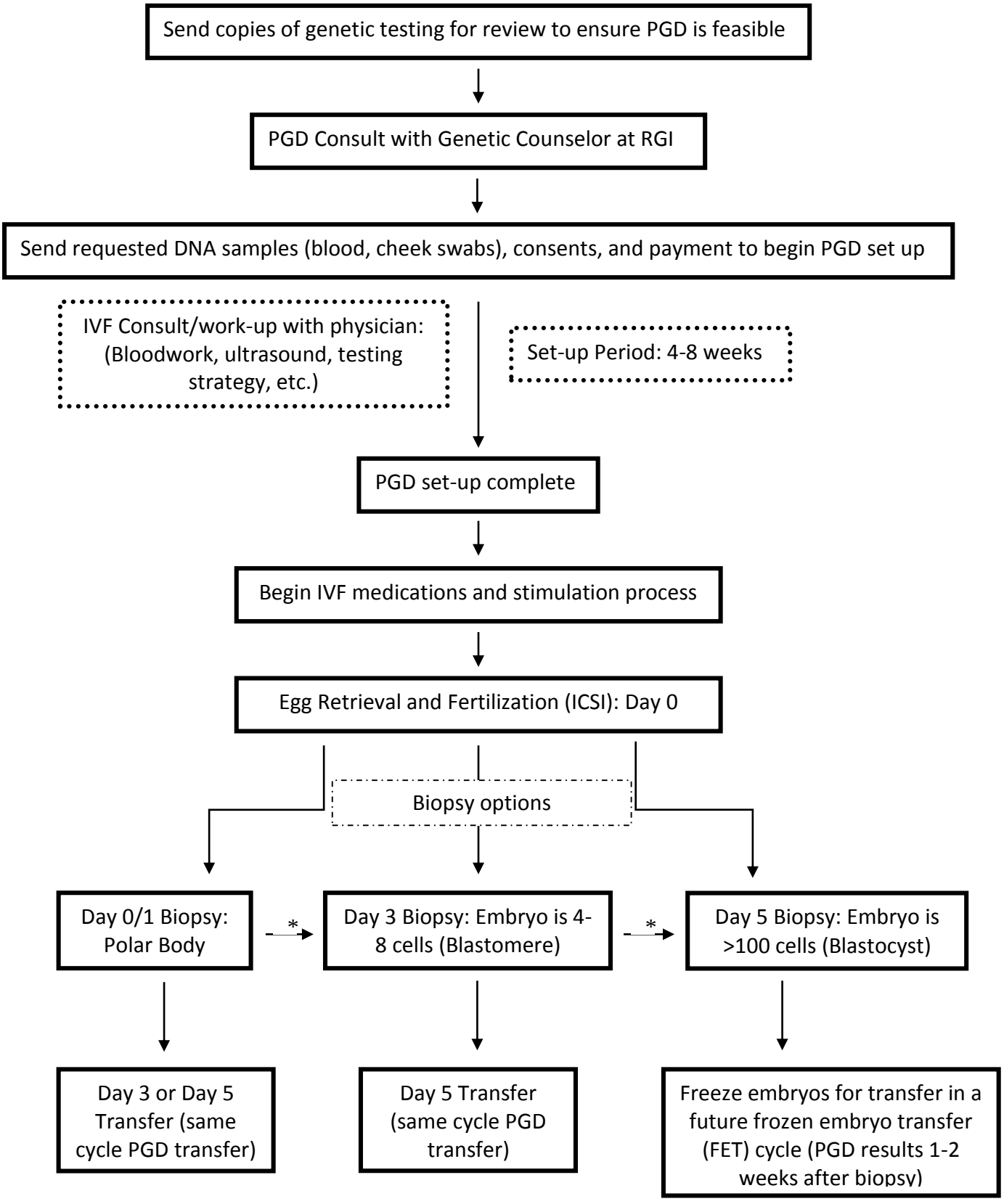
PGD FOR SINGLE GENE DISORDERS

There are a few different methods available for PGD testing, which are selected on a case-by-case basis. Our genetic counselors can help you to determine which method of analysis is the best option for you.

In order to diagnose an embryo, we test the biopsied cell(s) for a known genetic mutation in the family. We also test the embryos for linked markers that are inherited along with the gene and act as a form of "DNA fingerprinting". The testing strategy involved in PGD for single gene disorders is **Polymerase Chain Reaction (PCR).** PCR is a technique that is used to produce large amounts of specific DNA sequences from a small amount of DNA so that further analysis can be performed. The process begins with a sample of an individual's DNA. The DNA is then split into two strands and these are used as templates to create two copies. These copies are then split to make two copies each. This process is repeated over and over and can amplify the original DNA sequence up to a billion times. This is a valuable technique because of the small amount of DNA we obtain from polar body or embryo biopsy. This allows us to rapidly multiply the DNA and make a diagnosis. Linkage analysis is used in conjunction with PCR to increase the accuracy of the testing (see pages 6-7).

RGI has performed over 4,000 PGD cycles for nearly 400 single gene disorders. These cycles have resulted in pregnancies and deliveries confirmed to be unaffected by chorionic villus sampling (CVS), amniocentesis, or genetic testing following delivery.

IVF & PGD: SEQUENCE OF EVENTS OVERVIEW



* If rebiopsy is indicated

PGD SET-UP

What is needed for RGI to begin the PGD set-up?

The following items are required before RGI can proceed with your PGD set-up:

1. Genetic reports on your family
2. DNA samples from you and your family, as requested (blood and/or cheek swabs)
    - Please note this may involve your parents, your partner's parents, or other extended family as needed.
3. Signed notarized PGD consent forms.
4. Set-up payment (see page 15).

Once these items are received, the PGD set-up will take approximately 4-8 weeks to complete. **You cannot start your IVF medications until you have been notified that your PGD set-up is complete.**

How is the PGD set-up performed?

The PGD set-up involves confirmation of the disease-causing mutations in the family, as well as **linkage analysis**.

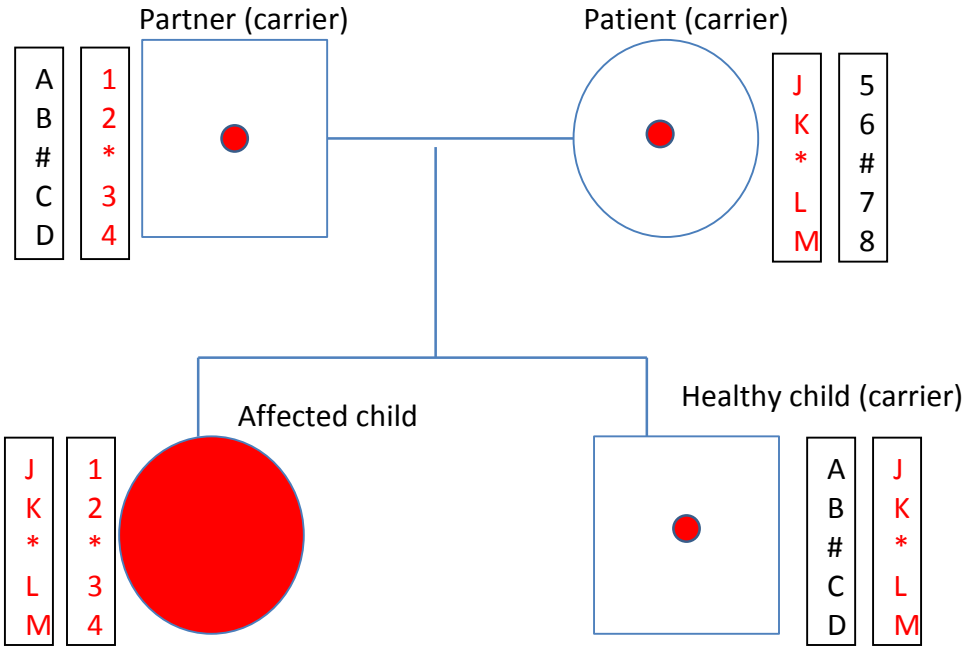
Linkage analysis is a method of determining the likelihood of two or more genes to be passed down together. If genes are considered to be "linked", the genes are together in the same chromosome region and will have a high probability of being passed down together. When we look at genes within the chromosome, they are arranged in a row. The closer together two genes are (i.e. the smaller the distance between them), the more likely they will travel as a unit and be inherited together. Linkage is established by analyzing the DNA of multiple family members whose genetic status is known.

This process improves the accuracy of PGD because we can look for the inheritance of **linked markers** (unique points in the DNA that are associated with a particular gene being studied) in addition to the mutation causing the disease. The use of linked markers essentially provides a backup system for the detection of **allele drop out (ADO**; the risk of one of the two gene copies not showing up during analysis, which is very common when working with such a small amount of DNA). ADO is a common cause of misdiagnosis or inconclusive results; therefore, the use of linked markers greatly reduces the chance of misdiagnosis or inconclusive results. The number of informative linked markers we use will vary per genetic condition and family, and cannot be determined until we are done with the PGD set-up process. The greater the number of unique linked markers, the higher the accuracy of the PGD testing.

The following page provides an illustration of the use of linked markers for PGD set-up.

LINKAGE ANALYSIS

(Example: Autosomal Recessive Disorder)



Circles represent females; squares represent males. A dot inside a circle or square represents an individual who is a carrier of a genetic disorder. A solid red circle or square represents an individual who is affected with a genetic disorder.

The two narrow rectangles next to a particular circle or square represent the two gene copies that individual possesses. A "*" symbol represents a genetic mutation; a "#" symbol represents a normal sequence (i.e. no genetic mutation). The numbers and letters represent the unique genetic sequences (**linked markers**) present at that location in the chromosome, surrounding the affected site (mutation) within the chromosome.

During the PGD set-up process, the linked markers are determined for each individual in the family. They are then compared between healthy and affected individuals to determine which linked markers are inherited along with the healthy gene copy, and which linked markers are inherited along with the affected (mutated) gene copy. Therefore, if the mutation does not amplify when we are analyzing a cell from an egg or embryo (allele drop out or ADO, see previous page), knowing which linked markers are associated with the affected copy and which linked markers are associated with the healthy copy will allow us to make an accurate diagnosis of the embryo.

Please be aware that the above description is for the ideal situation. In other families, there may be no affected child, or the child may be deceased. If this situation exists, DNA samples and/or genetic testing for the parents of you or your partner may be required. **It is essential that you and your partner facilitate any additional testing or sample collection required for other family members. RGI's genetic counselors are happy to refer your relatives to a genetics service local to them.**

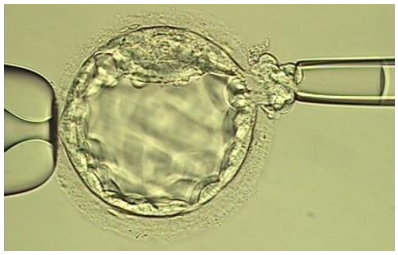
BIOPSY AND TESTING STRATEGIES

Once you have had a consult with a genetic counselor and have completed the set-up process, then you are free to start the IVF medications. Each physician has a slightly different process so please consult with your physician to learn about his/her recommendations for you. The medications are intended to regulate and stimulate the ovaries to produce many follicles, which contain eggs. After the egg retrieval procedure, the collected eggs will be fertilized. The embryologist may use a method called Intracytoplasmic Sperm Injection (ICSI) which consists of a single sperm being inserted into the egg. This helps to reduce the risk of contamination from other sperm and lets us know that we are only looking at the genetic information of the egg and the sperm that created that particular embryo. ICSI is required for all PGD cases.

An embryo biopsy is required to allow PGD/PGS testing, as the biopsied material is used for the testing.

After the eggs are fertilized, there are a few different biopsy options that can be completed at different stages in embryo development. The specific strategy for your case will depend on your preferences, as well as the recommendations of your physician and the equipment available at your IVF center. Sometimes, we are unable to get conclusive results and may need to utilize a combination of biopsy methods. The available strategies for biopsy of an embryo are:

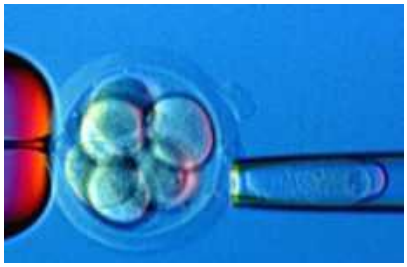
Blastocyst (Day 5/6) Biopsy



Five to six days after egg retrieval, well-developed embryos (called blastocysts) will have over 100 cells. At this point in development, several cells can be removed from the outer layer of the embryo (called the trophectoderm), which will eventually become the placenta. Once the trophectoderm cells are removed, an embryo typically needs to be frozen to avoid degradation of the embryo while genetic testing occurs. At this stage, the embryo consists of both paternal and maternal chromosomes. For PGD/PGS testing, this type of biopsy allows for multiple cells to be studied at the same time, improving the chances of a conclusive result. Once genetic testing is complete (approximately 7-10 business days later), a frozen embryo transfer (FET) can be performed during a future cycle with any unaffected embryos.

Please note that the most accurate method of PGS for aneuploidy is aCGH or NGS (24-chromosomes) on Day 5/6 blastocyst/trophectoderm samples. Please see page 15 for more information about the accuracy of PGD/PGS testing.

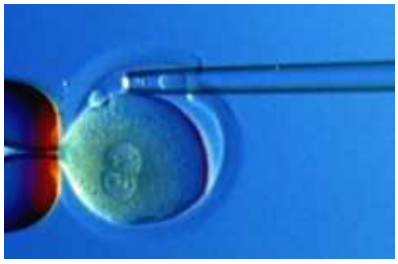
Blastomere (Day 3) Biopsy:



Three days after egg retrieval, the embryo is approximately four to eight cells in size. At this point we can remove/biopsy one cell and perform genetic testing while the embryo continues to grow and develop in the laboratory. At this point in development, the cells have not differentiated into different tissue types, and removing one cell has not been associated with an increased risk for birth defects or mental retardation. The embryo will usually compensate for the removed cell and continue to divide. At this stage, the embryo consists of both paternal and maternal chromosomes. **However, Day 3 testing has a very high risk of chromosomal mosaicism (the presence of two different types of cells in the embryo) which may lead to inaccurate test results.**

If the embryo is biopsied on Day 3, results are typically available in time for a Day 5 transfer. If cells need to be re-biopsied on Day 5 or if there are extra unused embryos after a transfer, the embryos can be cryopreserved (frozen) for use in a future cycle.

Polar Body Biopsy:



As eggs grow they divide and create byproducts called polar bodies. These polar bodies have no known function and are not part of the developing embryo. Polar bodies are useful because they contain genetic material that is discarded from the egg. Since we know the genetic information that is originally present in the egg, examining the discarded material allows us to determine what genetic information remains in the egg and will ultimately be the maternal genetic contribution to the embryo. These polar bodies can be removed and tested on Day 0 or Day 1 after egg retrieval and fertilization (the specific strategy for polar body biopsy depends on your specific testing plan). Sometimes we are unable to get a conclusive result from this testing and we need to re-test at a later stage.

EMBRYO TRANSFER AND CRYOPRESERVATION

Fresh Embryo Transfer

Once an embryo is predicted to be normal for the mutations(s) tested, the embryo will be recommended for transfer. Your IVF physician will work with you to determine the number of embryos to be transferred. If your case is performed by a Day 3 biopsy, then your embryo transfer will typically be on Day 5.

Embryo transfer is typically a brief procedure that is performed by inserting a catheter preloaded with embryos into the uterus under ultrasound guidance. It is associated with minor discomfort (described as similar to a pap smear). Your IVF physician will discuss this process and post-procedure instructions with you in more detail.

Embryo Cryopreservation/Frozen Embryo Transfer (FET)

If your embryos are biopsied on Day 5/6 (blastocyst/trophectoderm biopsy), the embryos will be frozen after biopsy for a future FET. Embryos that are unused after a fresh transfer may also be frozen (depending on their development) for a future FET if the first transfer is unsuccessful or if you decide you want to become pregnant again in the future. Please ask your IVF physician about the risks associated with freezing and thawing embryos.

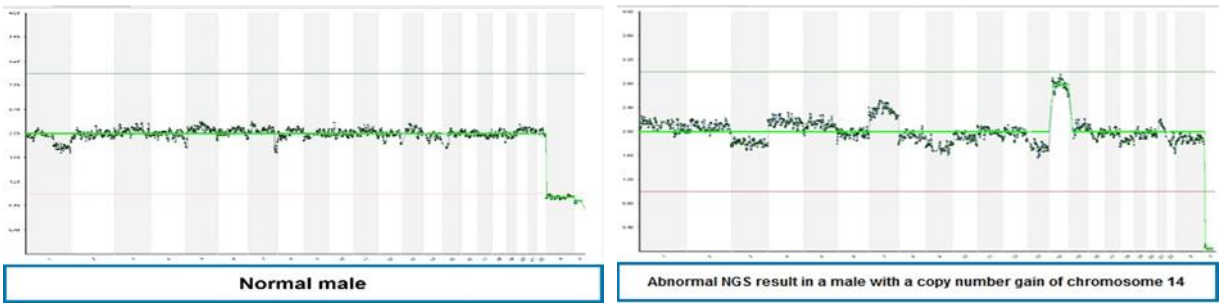
ADDITIONAL PGS TESTING

(ANEUPLOIDY/ CHROMOSOME ABNORMALITIES)

Chromosomes are the structures in our cells that carry our genes. Typically, we have 46 chromosomes in each of our cells. The chromosomes are in pairs (23 pairs in total); one copy of each chromosome is inherited from the egg, and the other copy is inherited from the sperm. Cells/embryos with 46 chromosomes are called euploid (correct chromosome number). If an egg or sperm is missing a chromosome or has an extra chromosome, this situation is referred to as **aneuploidy** (incorrect chromosome number). The majority of aneuploid embryos will fail to implant or will result in an early miscarriage; however, babies can be born with aneuploidies such as Down syndrome (Trisomy 21) or Trisomy 13/18. Extra or missing X and Y chromosomes are associated with miscarriage or milder syndromes, and can also provide information about the sex of an embryo.

The three aneuploidy testing options below can help to increase the chances of having a healthy baby:

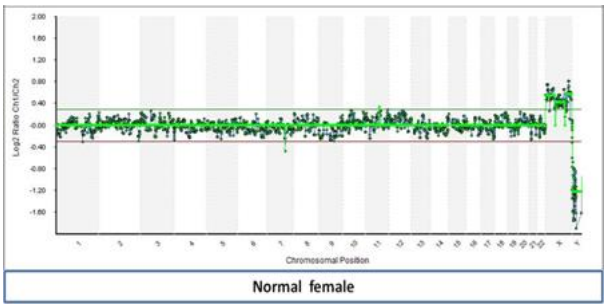
Next-Generation Sequencing (NGS)



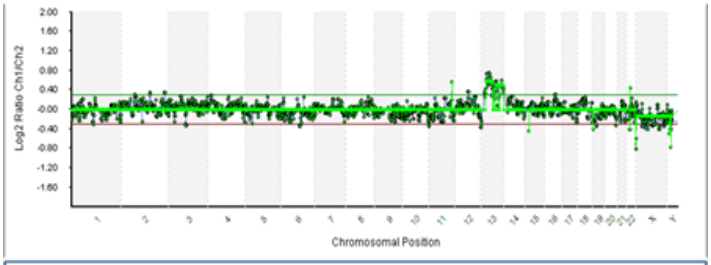
This test option looks at the number of all 24 chromosomes (pairs 1-22, X and Y) through a method called Next-Generation Sequencing (NGS). NGS is a method in which the genetic sequence of specific fragments of DNA from the embryo are compared to a control sample, which enables us to see if there is missing or extra genetic material. This testing method would provide information about the copy number of all chromosomes; this will increase your chances of a successful implantation, reduce your chances of miscarriage after transfer, and decrease the chance of having a child with a chromosomal abnormality. This testing can also provide information about the sex of the embryo. The accuracy of this testing is approximately 95-98% on Day 5/6 trophectoderm samples.

NGS is recommended for Day 5/6 trophectoderm samples only.

Array Comparative Genomic Hybridization (aCGH)/Microarray (RGI-Complete™)



Normal female



Abnormal microarray result in a male with a copy number gain of chromosome 13.

This test looks at the number of all 24 chromosomes (pairs 1-22, X and Y) through a method called array comparative genome hybridization (aCGH). This process compares the embryo's genetic material to a control sample, which enables us to see if there is missing or extra genetic material. This testing method would provide information about the copy number of all chromosomes; this will increase your chances of a successful implantation, reduce your chances of miscarriage after transfer, and decrease the chance of having a child with a chromosomal abnormality. This testing can also provide information about the sex of the embryo. The accuracy of this testing is approximately 95-98% on Day 5/6 trophectoderm samples.

aCGH is recommended for Day 5/6 trophectoderm samples only.

NGS and aCGH are not recommended on Day 3 blastomere samples, due to a high risk of chromosomal mosaicism (meaning that only a subset of cells in an embryo may have a chromosome problem) and failed amplification (inconclusive results). Currently, the most accurate strategy for chromosome testing is by NGS or aCGH on Day 5/6 trophectoderm samples.

5 chromosome testing

This option would test for chromosomes 13, 18, 21, X and Y (not all 24 chromosomes) using the same technology as the single gene testing (**PCR**) by looking for unique sequences (linked markers) associated with each chromosome, in order to count the number of each of these chromosomes within an embryo sample. This testing will test for only the most common chromosome abnormalities associated with chromosomal syndromes that could result in a live birth. This test can still provide information about the sex of the embryo. The accuracy of this testing is approximately 90%. Please note that this testing option cannot be performed on polar bodies.

PGD RESULTS: WHAT TO EXPECT

The number of embryos produced will depend on several factors, and can vary greatly from one person to another. Your results may also be different from one cycle to another.

The percentage of healthy embryos will depend on the inheritance pattern of the disease being tested. Your genetic counselor will review the expected percentages during your consultation. Only healthy embryos will be recommended for transfer by our laboratory.

A greater number of embryos available for testing will increase the chance of having a healthy embryo to transfer into the woman's uterus. **However, it is extremely important to remember that generalized statistics do not always hold up in small sample sizes.** Therefore, it is very possible to see a higher or lower number of healthy embryos than predicted. Healthy embryos must also be developing to be considered for transfer. **Some cycles may result in having no healthy and developing embryos to transfer.**

Not all of your embryos may have a conclusive diagnosis following PGD. Due to the limited amount of DNA that is available for testing, it is not uncommon to have some embryos without a conclusive result. Embryos without results will not be recommended for transfer, but may be able to be re-biopsied (depending on embryo development and your IVF center's capabilities) for further testing. Depending on the stage of the embryo, re-biopsied embryos will usually need to be frozen.

The accuracy of the results is described on the next page. Please note that the accuracy of your test results may vary from embryo to embryo.

Sample PGD results

Embryo #	Predicted Embryo Genotype	Embryo Transfer
1	NORMAL	YES
5	AFFECTED	NO
6	NO RESULT	N/A - RE-biopsy Recommended
9	CARRIER	YES
13	CARRIER	YES*

*Reduced accuracy. Transfer per patient consent only.

Timing of results

If you are having a Day 5 embryo transfer, then results will be available on the day of your scheduled transfer. It is very likely that you may already be at your IVF center when the results become available. If your embryos will be frozen after biopsy, then results will be available within 1-2 weeks from the time our laboratory receives the samples for testing.

PGD ACCURACY & PRENATAL TESTING

The purpose of PGD/PGS is to reduce significantly the risk of a pregnancy affected by a genetic disorder; however, it is not perfect. **The accuracy of PGD for a single gene disorder may be no more than 95-98%, and may vary per embryo.**

Factors that affect the accuracy of PGD/PGS are primarily due to the extremely small amount of DNA available in a single cell. These factors include:

- **Allele drop out (ADO)**: one of the gene copies does not show up (amplify) during analysis. This can reflect failed amplification for a particular marker or failure of the DNA probe to locate its complementary sequence.
- **Failed amplification**: no information is available about a particular marker in a gene
- **Recombination**: crossover between the two copies (alleles) of a given gene
- **Contamination**: DNA from outside sources or from other embryos interfere with the results
- **Human error:** problems with labeling, sample misidentification, etc.

It is important to know that PGD/PGS does not test for all genetic conditions; it can only test for those genetic disorders for which we have created a PGD set-up, or genetic syndrome associated with chromosomal abnormalities detectable by PGS. PGD/PGS does not test for any causes of birth defects or mental retardation that are not associated with the identified disorder(s) being tested. Every pregnancy has a 3-5% risk of a birth defect, regardless of the method of conception, and many defects are not amenable to testing.

Since PGD/PGS is not perfect, we recommend that patients undergo prenatal diagnosis following PGD/PGS for confirmation. Testing should be done by an independent, outside clinical laboratory. RGI offers confirmation testing on a research basis only. Prenatal diagnosis overcomes the challenges of PGD/PGS testing because there is a much greater amount of DNA to test in a prenatal sample compared to a sample from an egg or embryo. The two methods of prenatal diagnosis testing are:

Chorionic Villus Sampling (CVS)

CVS is typically performed between 10-13 weeks gestation. It can be performed transcervically (using a catheter through the cervix) or transabdominally (using a needle through the abdomen), depending on the location of the placenta. A small piece of the placenta is removed for examination of the chromosomes and testing of the single gene disorder for which a family is at risk. Ultrasound guidance is used throughout the procedure.

Amniocentesis

Amniocentesis is usually performed after 16 weeks gestation. It is performed transabdominally, using a needle through the abdomen. A small amount of amniotic fluid surrounding the fetus is aspirated for examination of the chromosomes and testing of the single gene disorder for which a family is at risk. Ultrasound guidance is used throughout the procedure.

Prenatal testing is recommended but is **not required.** They are invasive tests that have a 1/200-1/1000 risk of miscarriage, depending on your physician. These procedures may be performed through a physician local to you. Please contact one of our genetic counselors if you have questions or need a referral.

PAYMENT

Please contact a genetic counselor for updated cost information.

The fee for the PGD set-up must be received in order for our laboratory to begin your PGD set-up. The remaining fees (PGD testing, biopsy, travel, shipping, etc.) must be received prior to your egg retrieval.

We accept Visa, MasterCard, American Express, Discover, personal check, or wire transfer.

All IVF fees will be paid to your IVF center.

Insurance

As a courtesy, RGI will attempt to verify your insurance benefits for PGD following your consultation with a genetic counselor. Your insurance company will usually request a letter of medical necessity describing the PGD procedures, which our genetic counselors will submit within approximately one week of the request. **It usually takes up to 30 days or more before a response is issued from an insurance company.**

If a written approval is received from your insurance company, prior to the start of services, then RGI may not require any fees to be paid up front and will submit all costs to insurance after your setup or cycle is complete. **If a written approval cannot be issued, then payment will be required up front for any services.** RGI can file a claim with your insurance company after all PGD procedures are complete and reimburse you accordingly.

Please note that most insurance plans do not cover PGD. Additionally, not all of our services can be submitted to insurance.

Please review our *Insurance Guide for RGI PGD Patients* packet for more detailed information or contact billing@rgipgd.com with any questions regarding insurance and billing.

Please note: If your case requires genetic testing for your relatives, additional costs may be incurred through the genetic testing laboratory performing the test. RGI cannot provide pricing information about tests performed by other laboratories. Whether insurance will cover these tests will vary and you will need to check with the specific testing laboratory for pricing and insurance information. We are happy to provide a letter of explanation for any physicians or laboratories seeking information about the required testing.

NEXT STEPS

1. **After reviewing this information packet, please contact our genetic counseling coordinator by calling (847) 400-1515 or emailing info@rgipgd.com.** Our coordinator will be able to answer questions regarding our center and help you to start the PGD process if you are interested in pursuing PGD. General insurance inquiries can be directed to billing@rgipgd.com, but please note that we cannot answer specific questions about your coverage until our billing department has verified your benefits following your consultation. For information on IVF costs, please contact your local IVF physician or contact our center for a referral to an affiliated physician.

2. **The genetic counseling coordinator will request copies of your family's genetic test reports. Once all of the necessary reports are received, an appointment for a PGD consultation can be scheduled with a genetic counselor.** This consultation can be done over the phone or in-person, and typically lasts approximately 45-60 minutes. During the consultation, the genetic counselor will review the PGD procedure and timeline, as well as limitations of PGD testing and additional testing options. The genetic counselor will also ask questions about your family history and ethnicity, in order to determine if any additional tests are recommended.

3. After the consultation with a genetic counselor, you will be sent the necessary **paperwork and DNA collection kits required to begin the PGD process**. Once our laboratory has all of the required DNA samples, the necessary signed/notarized consent forms, and the initial payment, we will begin your PGD set-up.

   Please note: if it proves necessary to obtain DNA samples and/or genetic testing on your relatives, we can refer your relatives to a genetics provider local to them.

   Once started, the development of the set-up (based on mutation and/or linked markers, see pages 7-8) will take approximately 4-8 weeks to complete. Once you have been notified that your PGD set-up is complete, you will be free to **start your IVF medications**.

You have the option of undergoing IVF with an affiliated physician OR at another center local to you. If your selected IVF center cannot perform the required biopsies for your case, we may be able to send one of our experienced embryologists to your center to perform the biopsies for your case.

If you would prefer to undergo your IVF cycle at a center in your area:

a) Contact your preferred IVF center to determine if they are able to collaborate with our PGD laboratory.

b) If your entire IVF cycle will be completed in your area and you are working with an IVF center that is experienced in performing its own biopsies, an RGI embryologist will NOT be involved. **You will only need to make payment to RGI for the PGD/PGS services and possibly shipping of the samples**.

c) If your entire IVF cycle will be completed in your area and you are working with an IVF center that cannot perform their own biopsies, your case will require one of our experienced embryologists to travel to your area to perform the removal of the polar bodies and/or blastomeres and/or trophectoderm to bring back to our laboratory. **PGD/PGS costs, as well as the costs associated with the biopsy and embryologist's travel will apply.**

d) Please contact one of our genetic counselors when you have a written protocol of how your cycle is expected to be conducted or expected IVF timeline. **It is critical to inform our center about two specific timepoints:**

    1) When you are provided with a **stimulation start date**.

    2) When you have been instructed on when to administer the **hCG (trigger) shot** so that our lab can be prepared for your case.

FREQUENTLY ASKED QUESTIONS

Q: What is my first step?

A: Contact our genetic counseling coordinator to get information regarding the process, cost and to schedule a free consultation. You can reach our coordinator by phone (847) 400-1515 or by email at info@rgipgd.com.

Q: How long will it be between the time that I first contact you and the time that I'm having the eggs retrieved?

A: This is determined on a case-by-case basis, but is generally no sooner than three months.

Q: How do I choose an IVF center to work with?

A: If you are having trouble finding a center, please visit the Society for Assisted Reproductive Technology website (www.sart.org) to search for an IVF clinic in your area. We can help by letting you know which centers we have worked with previously. If you would like to work with a physician that we haven't worked with before, that is not a problem. We would just need to get some information about them so that we can set up a testing protocol and give you accurate information about the process.

Q: Do I have to travel?

A: Traveling is not usually necessary. Please see page 19.

Q: How long has RGI been doing PGD?

A: RGI has been performing PGD since it became available in 1990. We pioneered the polar body removal technology and are one of the most active centers offering PGD and PGS in the world. Our lab technicians are well-trained in all techniques involved.

Q: What is the pregnancy rate?

A: The pregnancy rate is dependent on several factors, including the woman's age and pre-IVF laboratory test results. Overall, the pregnancy rate associated with IVF is quoted as approximately 30-40% per IVF cycle. Please ask your IVF physician about the pregnancy rate quoted for your age and test results. It should be noted that several embryos are expected to be excluded from possible embryo transfer (i.e. embryos that are affected with the genetic disorder for which they are being tested). Therefore, it is important to remember that not all cycles will result in healthy embryos being available for transfer.

Q: Can you test for multiple single gene conditions on one cell?

A: Yes, this is often possible if there are multiple genetic mutations for which a family is at risk of passing on.

Q: Is there a risk to biopsying an egg or embryo?

A: RGI has followed up on most babies born after PGD through our laboratory. We have not seen an increased risk of birth defects or mental retardation following PGD, compared to the general population. There is, however, a small risk (typically <1%) that the biopsy will cause the egg or embryo to arrest, and therefore, not be useable for embryo transfer.