PREIMPLANTATION GENETIC DIAGNOSIS (PGD)

TRANSLOCATIONS AND OTHER CHROMOSOMAL REARRANGEMENTS

A PATIENT GUIDE

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PREIMPLANTATION GENETIC DIAGNOSIS (PGD) 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 OVERVIEW

RGI offers PGD and PGS to families who are at increased risk of having children with genetic disorders for a variety of reasons. The purpose of PGD/PGS 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.

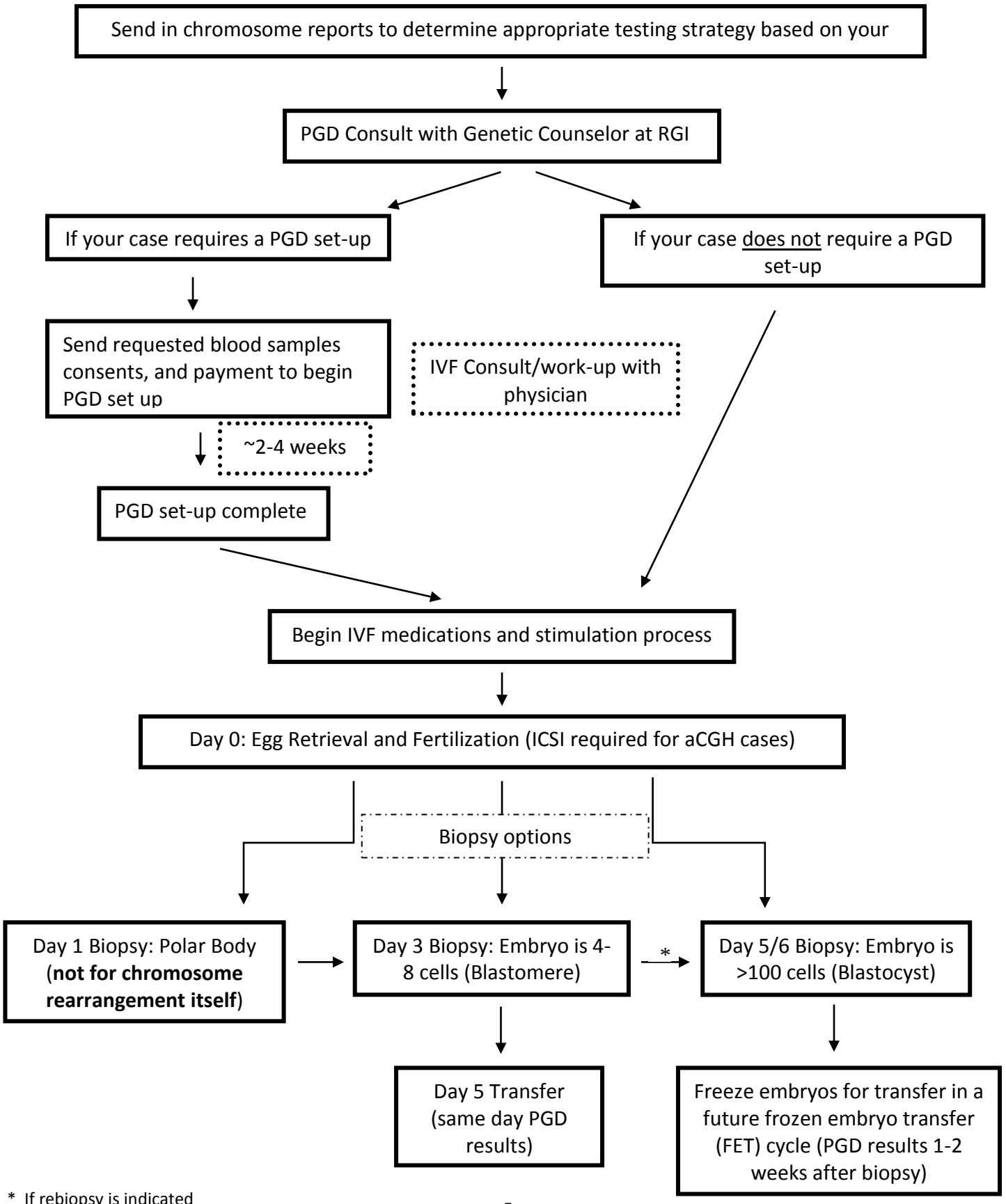
For over 20 years, we have helped many families and over 2,000 babies have been born who were unaffected for chromosomal and single gene disorders. We have previously performed PGD for over 300 disorders.

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 16).

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 pages 18 and 19 for more details).

This information packet will discuss the options available to you for testing for chromosomal rearrangements, such as translocations, inversions, deletions, and insertions.

IVF & PGD: SEQUENCE OF EVENTS OVERVIEW



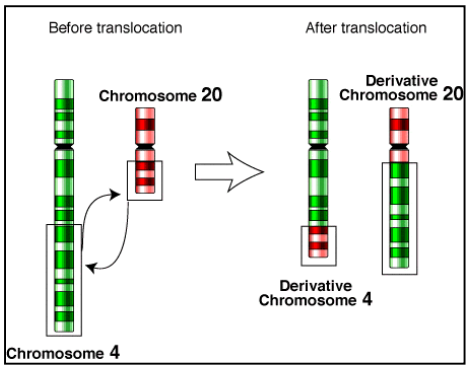
* If rebiopsy is indicated

OVERVIEW OF CHROMOSOME REARRANGEMENTS

Chromosome rearrangements usually do not cause any abnormalities as long as the rearrangement is **balanced** (having no extra or missing genetic information). However, an individual who is balanced for a rearrangement is at increased risk for producing eggs or sperm that are **unbalanced** (having extra or missing genetic information). Unbalanced embryos will usually result in failed implantation or miscarriage. It is also possible for a baby to be born with an unbalanced chromosome rearrangement, which is often associated with birth defects and/or mental retardation.

The most common types of chromosome rearrangements are translocations and inversions:

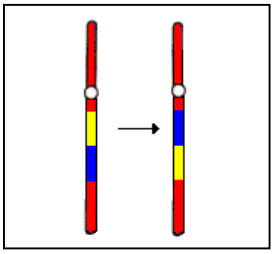
Translocations



The majority of chromosome rearrangements tested by RGI are translocations. A translocation occurs when two pieces of different chromosomes break off and switch places, reattaching themselves to the wrong chromosomes.

The two main types of translocations are called **reciprocal** and **Robertsonian**. Individuals with a balanced reciprocal translocation have the normal 46 chromosomes, with two chromosomes having exchanged genetic material. Individuals with a balanced Robertsonian translocation have 45 chromosomes; however, the material that has been lost does not actually contain any genes (which is why these individuals are still referred to as "balanced"). Robertsonian translocations usually do not require a PGD set-up and are typically associated with a greater number of healthy embryos (~30-45%) compared to reciprocal translocations (20-30%). Most translocation cases do not require a PGD set-up.

Inversions



Inversions are structural rearrangements of genetic material within a chromosome. An inversion occurs when a chromosomal segment breaks in two places, rotates 180 degrees, and reinserts itself into the same chromosome in reversed orientation.

Since inversions are rare, it is difficult to estimate the percentage of healthy embryos that will be determined through PGD. It should be noted that some inversions are common in the general population and do not result in an increased risk of fertility problems or unbalanced embryos.

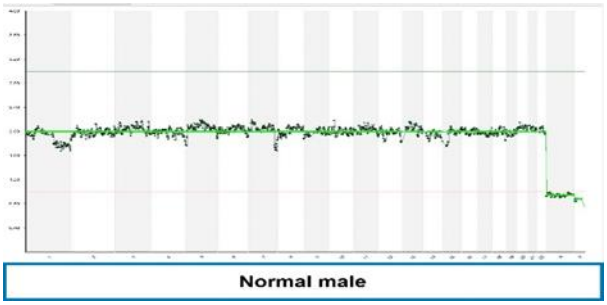
Other types of chromosome rearrangements include **deletions**, **duplications**, and **insertions**. PGD is often feasible to reduce the chance of passing on these types of rearrangements. One of our genetic counselors will discuss which types of testing are available after reviewing your chromosome report (karyotype).

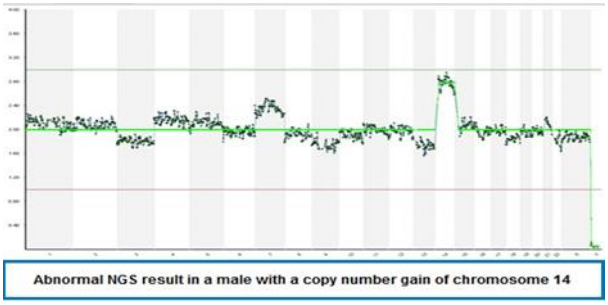
PGD FOR CHROMOSOME REARRANGEMENTS

There are three different methods available for PGD testing. Each chromosome rearrangement report is reviewed by our laboratory staff to determine which method is the best option for your case. In some cases, only one option may be recommended, while in other cases, several options may be available to you.

In order to diagnose an embryo, we test the biopsied cell(s) for a known chromosome rearrangement in the family. The testing strategies involved in PGD for chromosome rearrangements are **NGS, aCGH, or FISH.** These three methods are described below:

Next-Generation Sequencing (NGS)


Normal male

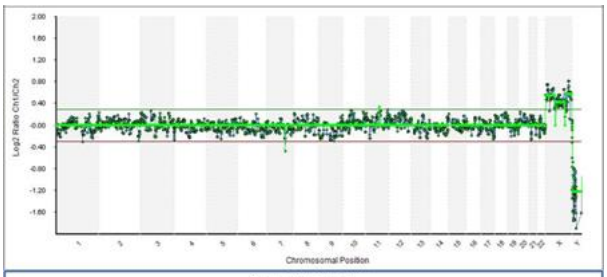

Abnormal NGS result in a male with a copy number gain of chromosome 14

This test looks at the number of all 24 chromosomes (pairs 1-22, X and Y) through a method called Next-Generation Sequencing (NGS). Sequencing is determining the order of DNA bases, the "letters" that make up our genetic code. The human genome is made up of over 3 billion of these genetic letters. NGS is a method in which specific fragments of DNA from the embryo are compared to a control sample. This DNA is amplified (multiplied) and the concentration is standardized to the control sample. The next step is DNA sequencing of these fragments. In this step, we sequentially analyze the copy number, i.e. the number of times we see a fragment. This copy number is compared to the control sample which enables us to see if there is missing or extra genetic material. This testing method can typically only distinguish between whole extra or missing chromosomes but may sometimes detect smaller pieces of duplicated or deleted chromosomal material. Depending on the case, this method may be used to detect the presence of an unbalanced translocation.

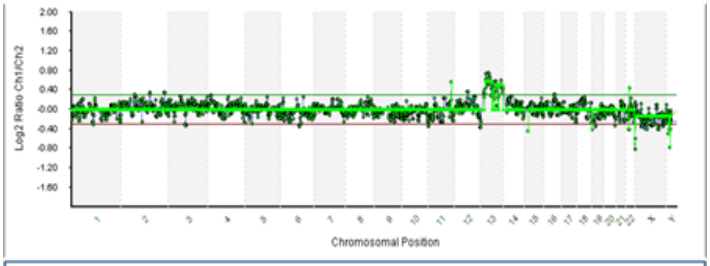
NGS cannot distinguish between embryos that are balanced (have a translocation in a balanced form like one of the parents) and embryos that are normal (have no translocation).

It is recommended for NGS to be performed on Day 5/6 embryos (see page 11).

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



Normal female



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

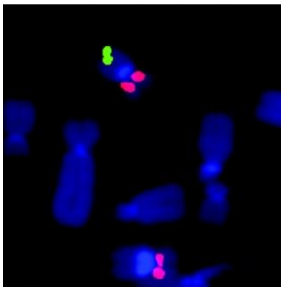
This test looks at the number of all 24 chromosomes (pairs 1-22, X and Y) through a method called array comparative genomic hybridization (aCGH). Through this method, DNA from the embryo is bound (hybridized) to a control sample. The control sample is labeled in red probes and the embryo's sample is labeled in green. When the control sample combines with the embryo's sample, the red and green should combine to make yellow. When we look through the probes, we can see if there is missing or extra information by the colors that we see at certain points (i.e. if we see a red color, we know there isn't enough green in that area, i.e. something is missing in the embryo's DNA sample). Depending on the case, this method may be used to detect the presence of an unbalanced translocation.

Microarray cannot distinguish between embryos that are balanced (have a translocation in a balanced form like one of the parents) and embryos that are normal (have no translocation).

It is recommended for aCGH to be performed on Day 5/6 embryos (see page 11).

Fluorescent In-Situ Hybridization (FISH)

The FISH testing method looks for the presence of certain chromosome regions by using specific probes, which are designed to look for unique genetic sequences. If the probe sees this sequence, it will attach and then light up (fluoresce). We can use multiple probes that are each labeled with a different color so that we can distinguish different chromosome regions from one another.



FISH testing occasionally requires a set-up, which would mean obtaining a blood sample from the person who carries the rearrangement. The set-up typically takes approximately 2-4 weeks to complete. If a set-up is required for your particular case, the IVF medications cannot be started until we have notified you that it is complete.

FISH testing can test ONLY for the known chromosome rearrangement, or can also test for up to seven additional chromosomes (chromosome numbers 13, 16, 18, 21, 22, X and Y). The additional chromosomes that can be tested for are not related to the familial rearrangement; however, spontaneous abnormalities causing extra or missing chromosomes can happen in the eggs or sperm of anyone. They are not related to an individual's personal health or family history. When there are extra or missing copies of these chromosomes, specific syndromes that cause medical complications can occur. For example, an extra copy of chromosome 21 is the cause of Down syndrome. An extra copy of chromosome 13 or 18 usually results in miscarriage but can result in the birth of a baby with Trisomy 13 or Trisomy 18, which are syndromes that are fatal in infancy. 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. Extra copies of chromosome 16 or 22 are common causes of early miscarriage.

In most cases, FISH testing cannot distinguish between embryos that are balanced (have a translocation in a balanced form like one of the parents) and embryos that are normal (have no translocation). However, an additional procedure called **conversion** can distinguish between these two groups of embryos. Conversion is successful approximately 70% of the time in which it is attempted; if it is not successful, then we cannot distinguish between balanced and normal embryos. We can still, however, distinguish between balanced/normal and unbalanced embryos. **Conversion is a highly specialized technique which requires RGI to send one of our embryologists to your IVF center to perform the procedure.** Conversion may be required for some chromosome rearrangement cases.

It is recommended for FISH to be performed on Day 3 embryos (see pages 12).

PGD SET-UP

Some chromosomal rearrangements will require a PGD set-up.

If a set-up is needed for my case, 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. Chromosome report from the individual who has the rearrangement
2. A blood sample from the individual who has the rearrangement
3. Signed and notarized PGD consent forms
4. Set-up payment (see page 17)

Once these items are received, the PGD set-up will take approximately 2-4 weeks to complete. Please note that some complex rearrangements may have a longer set-up period. **You cannot start your IVF medications until you have been notified that your PGD set-up is complete.**

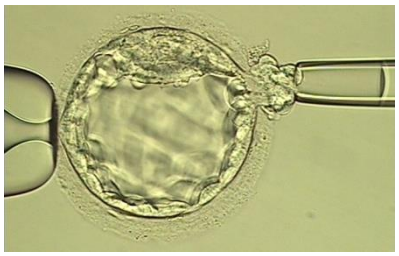
How is the PGD set-up performed?

The PGD set-up involves confirmation of the known chromosomal rearrangement and its detection using NGS, aCGH or FISH probes. See pages 7-9 for more information about these testing methods.

BIOPSY AND TESTING STRATEGIES

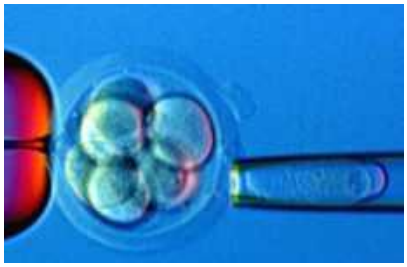
Once you have had a consult with a genetic counselor and your PGD set-up (if required) is complete, then you are free to start the IVF medications. Each physician has a slightly different process so please consult with your physician to learn about his/her recommendations for you. The medications are 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 NGS and aCGH cases but is optional for FISH cases.

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 aCGH and NGS 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. **Day 5/6 biopsy is the recommended methods for all cases performed by aCGH or NGS.**

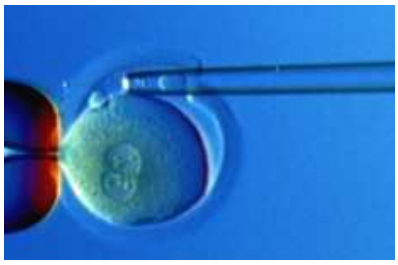
Blastomere (Day 3) Biopsy:



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. **Day 3 biopsy is the recommended method for all cases performed by FISH**. **If additional chromosome testing is desired (see page 9), there is a high risk of chromosomal mosaicism (the presence of two different types of cells in the embryo) which may lead to inaccurate test results. Mosaicism is not usually an issue when testing for a known chromosome rearrangement alone.**

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. **Polar bodies cannot be used to test for a chromosomal rearrangement.** However, if the rearrangement will be tested by FISH, it may be possible to test the polar bodies for additional maternally-derived chromosome abnormalities which are unrelated to the rearrangement (see page 9). Sometimes we are unable to get a conclusive result from this testing and we need to re-test for the additional chromosomes at a later stage.

EMBRYO TRANSFER AND CRYOPRESERVATION

Fresh Embryo Transfer

Once an embryo is predicted to be normal for the chromosomes 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.

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 type of chromosome rearrangement being tested for, and typically ranges from 20-45%. Keep in mind that the percentage of healthy embryos is expected to be lower if any additional chromosomes are being tested. If you are testing for a translocation, your genetic counselor will review the expected percentages during your consultation. For other chromosome rearrangements such as inversions or insertions, it is difficult to predict the expected percentages.

Only healthy embryos will be recommended for transfer by our laboratory. Healthy embryos are those which are balanced (have the rearrangement in the balanced form like one of the parents) or normal (no translocation at all). **Remember, we usually cannot distinguish between balanced or normal embryos.** Only FISH with conversion (see page 9) can distinguish between these two groups of embryos.

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 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. **Unfortunately, many cycles 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 page 16. Please note that some embryos may have a lower accuracy than others, and that this would be noted on your PGD results report.

Sample PGD results

EMBRYO #	DIAGNOSIS	EMBRYO TRANSFER
1	UNBALANCED	NO
5	UNBALANCED	NO
6	UNBALANCED	NO
9	BALANCED/NORMAL	YES
10	BALANCED/NORMAL, abnormal for chromosome 21	NO
12	UNBALANCED	NO
13	NO RESULT	NO

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 is to significantly reduce the risk of having a pregnancy affected by a genetic disorder; however, it is not perfect. **The accuracy of PGD for a chromosome rearrangement is approximately 95%.** If other chromosomes are being tested, the accuracy will typically range from 90-98% for these additional chromosomes, depending on the stage at which the embryo is biopsied and the testing method.

It is important to know that PGD does not test for all genetic conditions; it can only test for the identified chromosome rearrangement and possibly other additional chromosomes depending on the test being performed. PGD does not test for any causes of birth defects or intellectual disabilities that are not associated with abnormal chromosome number. Every pregnancy has a 3-5% risk of a birth defect, regardless of the method of conception.

Since PGD is not perfect (due to a limited amount of DNA to test), we recommend that patients undergo prenatal diagnosis following PGD for confirmation. Prenatal diagnosis overcomes the challenges of PGD testing because there is a much greater amount of DNA to test in a prenatal sample compared to a sample from an embryo. As well, prenatal testing can visualize the full chromosome set (called a karyotype), which PGD cannot do. The two common 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 by routine karyotyping or FISH analysis if needed. Ultrasound guidance is used throughout the procedure.

Amniocentesis

Amniocentesis is typically performed after 15 or 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 by routine karyotyping or FISH analysis if needed. 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 can be performed through a physician local to you. Please contact one of our genetic counselors if you have questions.

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.

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 a copy of your chromosomal rearrangement report(s). 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 talking with a genetic counselor, you will be sent the necessary consent form and other documents required for PGS. This consent form needs to be signed, notarized and returned (original copy, please, no faxed copies) along with the payment for the PGD. **All paperwork and payment must be received prior to testing.**

    a. If a set-up is required, the blood collection kit will be sent following your consultations. The requested blood samples, as well as the original notarized consents and payment are required to start your PGD set-up

    b. The development of the set-up will take approximately 2-4 weeks to complete. Once you have been notified that your PGD set-up is complete, you will be free to start your IVF medications.

    c. **PGD for chromosomal rearrangements typically does not require a set-up.**

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 a PGD/PGS 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 PGS 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. **PGS costs, as well as the costs associated with the biopsy and embryologist 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 time points:**

   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 and to schedule a free consultation. You can reach us 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, as it will depend on the particular chromosome rearrangement. For most chromosomal rearrangement cases, there is no waiting period so an IVF cycle can begin whenever your physician is ready. For cases requiring a PGD set-up, it may be a couple of months between your initial contact with us and your egg retrieval.

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

A: If you are having trouble finding a center, 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 to RGI is not usually necessary. Please see pages 18 and 19.

Q: How long has RGI been doing PGD?

A: RGI has been performing PGD/PGS since it became available in 1990. As the first PGD laboratory in the United States, we pioneered the polar body removal technology and continue to be 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 chromosome abnormalities). Therefore, it is important to remember that not all cycles will result in healthy embryos being available for transfer.

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

A: RGI has followed up on most babies born after PGD/PGS through our laboratory. We have not seen an increased risk of birth defects or intellectual disability following PGD/PGS, 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.