PREIMPLANTATION GENETIC SCREENING (PGS)

ANEUPLOIDY

A PATIENT GUIDE

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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 PGS and IVF for common chromosomal problems (aneuploidy), which also applies to those couples who are interested in sex selection.

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

This informational packet will discuss the PGS options available to you for testing for aneuploidy (abnormal number of chromosomes), which also applies to those couples who are interested in sex selection.
IVF & PGS: SEQUENCE OF EVENTS OVERVIEW

IVF Consult/work-up with physician

PGS Consult with Genetic Counselor at RGI

Begin IVF medications and stimulation process

Day 0: Egg Retrieval and Fertilization (ICSI required for NGS and aCGH cases)

Biopsy options

Day 0/1 Biopsy: Polar Body

Day 3 Biopsy: Embryo is 4-8 cells (Blastomere)

Day 5/6 Biopsy: Embryo is >100 cells (Blastocyst)

Day 3 Transfer

Day 5 Transfer (same day PGS results)

Freeze embryos for transfer in a future frozen embryo transfer (FET) cycle (PGS results 1-2 weeks after biopsy)

* If rebiopsy is indicated
OVERVIEW OF CHROMOSOMAL ABNORMALITIES (ANEUPLOIDY)

Aneuploidy is a term used to describe a chromosome problem that is caused by an extra or missing chromosome. The most commonly known example of aneuploidy is Down syndrome, which is caused by an extra copy of chromosome 21.

Chromosomes are the structures in our cells that carry our genetic information (genes). Typically, we have 46 chromosomes in each of our cells. The chromosomes come 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 aneuploid (incorrect chromosome number).

Types of aneuploidy

Many aneuploidies will cause an embryo to fail implanting in the uterus. If an aneuploid embryo does implant, the pregnancy will usually end in miscarriage. However, there are five numerical chromosome abnormalities that can result in a baby that comes to term. These aneuploidies involve the following chromosomes:

**Chromosome 21** – An extra copy of this chromosome results in Trisomy 21, which is also known as Down syndrome. Most pregnancies with Down syndrome will miscarriage; however, 25% of these pregnancies will come to term. Individuals with Down syndrome are often born with heart defects, low muscle tone, gastrointestinal problems, and abnormalities with other organs, and have varying degrees of developmental delay and intellectual disability. They are at higher risk for thyroid problems, early-onset dementia, and leukemia.

**Chromosome 18** – An extra copy of this chromosome results in Trisomy 18. Most pregnancies with Trisomy 18 will miscarry; however, 5-10% of these pregnancies will come to term. Babies with Trisomy 18 are often born with birth defects in the heart, kidneys, intestines, and other organs, and have difficulty feeding and gaining weight. Over 90% of babies born with Trisomy 18 will not survive past the first year of life, and those who do survive typically have severe developmental delays and intellectual disability.

**Chromosome 13** – An extra copy of this chromosome results in Trisomy 13. Most pregnancies with Trisomy 13 will miscarry; however, approximately 5% of these pregnancies will come to term. Babies with Trisomy 13 are often born with heart defects, cleft lip and/or palate, extra fingers, hernias, and low muscle tone. They may have abnormalities with the eyes, skin, and limbs, and often have seizures and feeding problems. The majority of babies born with Trisomy 13 will not survive past the first year of life, and those who do survive typically have severe developmental delays and intellectual disability.

**Chromosomes X and Y** – Extra or missing copies of the X or Y chromosome are called sex chromosome abnormalities. Most sex chromosome abnormalities tend to cause infertility and may require hormone therapy. Individuals affected with sex chromosome abnormalities may have learning disabilities or behavioral problems, but typically do not have intellectual disability.
Although the sex chromosomes determine the sex of an individual, abnormalities involving extra or missing X and Y chromosomes do not usually cause problems related to gender.

Abnormalities involving chromosomes 16 and 22 are common causes of first trimester miscarriage. Abnormalities involving chromosomes 8, 9, 15, and 17 are more rare causes of first trimester miscarriage. All other chromosome abnormalities usually result in failed implantation of the embryo.

**Risk of aneuploidy**

Every couple will produce a proportion of embryos with chromosome abnormalities. This proportion increases as a woman gets older. The risk for aneuploidy is independent of pregnancy history, medical history, family history and ethnicity.

Since most embryos with chromosome abnormalities will fail to implant or will result in miscarriage, the frequency of aneuploidy in IVF embryos is much higher than the number of babies born with chromosome abnormalities. For example, a 40-year-old woman has approximately a 1 in 66 (1.5%) risk of having a live born child with a chromosome problem. At the time of IVF for a 40-year-old woman, approximately 60% of embryos will be abnormal, the majority of which would result in failed implantation or miscarriage.

**Why test for aneuploidy?**

Without PGS, embryos are selected for transfer based only on their morphology (physical appearance and development). It is important to note, however, that an embryo’s development is not always associated with its chromosomal content. Therefore, a well-developed embryo may be selected for transfer, but may not result in a pregnancy due to abnormal chromosomes. PGS can give your physician another way to select the best embryos for transfer, in order to maximize the chances of having a successful IVF cycle.

PGS for aneuploidy increases the chance of an embryo implanting, decreases the chance of miscarriage, and decreases the chance of having a child with a chromosome abnormality (which often causes birth defects and cognitive disabilities). Therefore, the purpose of PGS for aneuploidy is to increase the chance of having a successful pregnancy and a healthy baby.
PGS OPTIONS

There are several different methods available for PGS testing. Our genetic counselors can help you to determine which option is best for you. With all of our PGS testing methods, blood samples are not required and there is no waiting period.

The testing strategies involved in PGS for aneuploidy are NGS, aCGH or FISH. These three methods are described below:

**Next-Generation Sequencing (NGS)**

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.

It is recommended for NGS to be performed on Day 5/6 embryos (see page 11).
Array Comparative Genomic Hybridization (aCGH)/Microarray (RGI-Complete™)

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). 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). 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.

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 chromosomes 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 chromosomes from one another, however we cannot test all 24 chromosomes using this method. There are three options for testing by FISH analysis:

5 Chromosome Panel: 13, 18, 21, X, Y

This panel is designed to reduce the risk of having a pregnancy with a chromosome abnormality that could result in a live birth (please see pages 6-7 for descriptions of the chromosomes tested by this option). We can use the number of X and Y chromosomes to determine the sex of an embryo.

5 Chromosome Panel: 13, 16, 18, 21, 22

This panel looks at some chromosome abnormalities that are associated with genetic syndromes, as well as chromosome abnormalities that are the most common causes of early miscarriage (please see pages 6-7 for descriptions of the chromosomes tested by this option). This panel will not provide information about the sex of an embryo.

7-11 Chromosome Panel: 13, 16, 18, 21, 22, X, Y + 8, 9, 15, 17

This panel is designed to reduce the risk of having a pregnancy with a chromosome abnormality that could result in a live birth, as well as reduce the risk of miscarriage and failed implantation (please see pages 6-7 for descriptions of the chromosomes tested by this option). We primarily attempt to test embryos for the more common abnormalities, and probes for the less common abnormalities are added if possible.

It is recommended for FISH to be performed on Polar Bodies and/or Day 3 embryos (see page 11-12).
BIOPSY AND TESTING STRATEGIES

Once you have had a consult with a genetic counselor, 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.

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 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.**

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 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. Day 3 biopsy is the recommended method for all cases performed by FISH. 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 whether testing is performed by FISH or aCGH). 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.
PGS 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 tends to range anywhere from 20-80% will depend on the age of the woman (or the age of the egg donor if one is being used). 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 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, some cycles result in having no healthy and developing embryos to transfer.**

If you are pursuing PGS for the purposes of sex selection, it is expected that 50% of embryos will be male and 50% will be female. **However, some cycles result in having no healthy, developing embryos of the desired sex to transfer.**

**Not all of your embryos may have a conclusive diagnosis following PGS.** 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 some chromosomes in some embryos may have a lower accuracy than others, and that this would be noted on your PGS results report.

**Sample PGS results (24-chromosome microarray)**

<table>
<thead>
<tr>
<th>Embryo #</th>
<th>aCGH Results</th>
<th>Diagnosis</th>
<th>Embryo Transfer</th>
<th>Comments</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>46,XY</td>
<td>NORMAL Male</td>
<td>YES</td>
<td></td>
</tr>
<tr>
<td>5</td>
<td>47,XX,+13</td>
<td>ABNORMAL</td>
<td>NO</td>
<td></td>
</tr>
<tr>
<td>9</td>
<td>46,XX</td>
<td>NORMAL Female</td>
<td>YES</td>
<td></td>
</tr>
<tr>
<td>10</td>
<td>Complex abnormalities</td>
<td>ABNORMAL</td>
<td>NO</td>
<td></td>
</tr>
<tr>
<td>12</td>
<td>46,XX</td>
<td>NORMAL Female</td>
<td>YES</td>
<td></td>
</tr>
<tr>
<td>13</td>
<td>FA</td>
<td>FA</td>
<td>NO</td>
<td>Rebiopsy recommended</td>
</tr>
</tbody>
</table>

**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.
The purpose of PGS is to significantly reduce the risk of having a pregnancy affected by a genetic disorder; however, it is not perfect. **The accuracy of PGS for aneuploidy ranges from 90-98% depending on the testing strategy used.** The accuracies quoted by our laboratory are listed below:

- NGS testing – Day 5: 95-98%
- Microarray testing - Day 5: 95-98%
- Microarray testing - Day 3 (not recommended): 90%
- FISH - Day 1 and Day 3: 90-95%
- FISH – Day 1 or Day 3 or Day 5: 90%

It is important to know that PGS does not test for all genetic conditions; it can only test for conditions that are caused by a full extra or missing chromosome. PGS does not test for any causes of birth defects or mental retardation 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 PGS is not perfect (due to a limited amount of DNA to test), we recommend that patients undergo prenatal diagnosis following PGS for confirmation. Prenatal diagnosis overcomes the challenges of 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. As well, prenatal testing can visualize the full chromosome set (called a karyotype), which PGS 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 trans-cervically (using a catheter through the cervix) or trans-abdominally (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. Ultrasound guidance is used throughout the procedure.

**Amniocentesis**

Amniocentesis is typically performed after 15 or 16 weeks gestation. It is performed trans-abdominally, 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. 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.
Please contact a genetic counselor for updated cost information.

**All fees must be received prior to your egg retrieval.**

All fees 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 PGS, if possible, following your consultation with a genetic counselor. Your insurance company will usually request a letter of medical necessity describing the PGS 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 PGS. 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 this process if you are interested in pursuing PGD/PGS. 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 one of our affiliate physicians.

2. The genetic counseling coordinator will work with you to schedule an appointment for a consultation with one of our genetic counselors. This consultation can be done over the phone or in person, and typically lasts approximately 20-45 minutes. During the consultation, the genetic counselor will review the PGS options, procedures, timeline, and cost, as well as limitations of PGS testing. 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/PGS. All paperwork and payment must be received prior to testing.

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: There is no waiting period for PGS for aneuploidy. Therefore, your specific timeline will be dictated by your IVF cycle.

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 page 17.

Q: How long has RGI been doing PGD/PGS?
A: RGI has been performing PGD/PGS 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 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.