PREIMPLANTATION GENETIC DIAGNOSIS (PGD)

TRANSLOCATIONS
AND OTHER CHROMOSOMAL REARRANGEMENTS

A PATIENT GUIDE

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PREIMPLANTATION GENETIC DIAGNOSIS (PGD) AT RGI

The Reproductive Genetics Institute (RGI) performs preimplantation genetic diagnosis (PGD) for the purpose of aiding individuals at risk for having a pregnancy or child with a genetic disease. There are several reasons why couples may choose to pursue PGD testing. These couples may already have had a child or pregnancy with a genetic condition or chromosome abnormality. Those who already have children with a genetic condition may also try to have a baby who is an immunological (HLA) match to their child and can therefore act as a bone marrow or stem cell donor. Some couples choose to pursue PGD because one of the partners carries a balanced translocation, which causes their pregnancies to be at high risk for miscarriage or abnormal outcome. Other couples may be at increased risk for Down syndrome and other chromosome abnormalities due to advanced maternal age. RGI can assist you and your family planning by offering genetic counseling regarding PGD and how it fits into the process of In Vitro Fertilization (IVF). This packet will assist you in understanding PGD and IVF for translocations and other chromosome rearrangements.

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.
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PGD 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 previously performed PGD for over 300 disorders.

In order to do PGD, 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 egg or embryo is tested at various points in development and either transferred back into the uterus or frozen for a later transfer (see pages 10-12). This way we can identify genetically abnormal embryos before transferring them. This is what makes PGD 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 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 Chicago. If you do not have a local specialist you desire to work with or prefer to travel to Chicago for treatments we can assist you in referring you to an affiliated IVF center/physician (see pages 17-18 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.
Send in chromosome reports to determine appropriate testing strategy based on your case

PGD Consult with Genetic Counselor at RGI

If your case requires a PGD set-up

Send requested blood samples consents, and payment to begin PGD set up

~2-4 weeks

PGD set-up complete

Begin IVF medications and stimulation process

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

Biopsy options

Day 1 Biopsy: Polar Body (not for chromosome rearrangement itself)

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

Day 5 Transfer (same day PGD results)

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

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

* 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. Reciprocal translocations typically require a PGD set-up. 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%).

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.

Inversions usually require a PGD set-up. 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).
There are two 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, both 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 FISH or aCGH. These two methods are described below:

**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 often 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 one’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. Due to the time required for this process, conversion is only possible if they embryos are in our lab in Chicago. Conversion may be required for some chromosome rearrangement cases.

It is recommended for FISH to be performed on Day 3 embryos (see pages 10-11).
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 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 testing does not require a set-up, so the IVF cycle can begin as soon as we have notified you that your case can be accurately performed by aCGH.

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 pages 10-11).
Some chromosomal rearrangements will require a PGD set-up. This typically includes any chromosomal rearrangement that will be tested by FISH analysis (see pages 7-8), other than Robertsonian translocations.

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 (in sodium heparin green top blood tubes)
3. Signed and witnessed/notarized PGD consent forms
4. Set-up payment (see page 16)

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 FISH probes. See pages 7-9 for more information about FISH.
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 aCGH cases but is optional for FISH cases.** After the eggs are fertilized, there are a few different testing 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 testing methods. The strategies for testing an embryo are:

**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 7). 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.
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 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 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. If additional chromosome testing is desired (see pages 7-8), there is a risk of chromosomal mosaicism (the presence of two different types of cells in the embryo). Mosaicism is not usually an issue when testing for a known chromosome rearrangement alone. **Day 3 biopsy is the recommended method for all cases performed by FISH.**

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.

Blastocyst (Day 5/6) Biopsy

After five to six days, 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 cases, 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 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 method for all cases performed by aCGH.**
EMBRYO TRANSFER AND CRYOPRESERVATION

Embryo Transfer

Once an embryo is predicted to be balanced or normal, 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 eggs and 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 7) 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 eggs or 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 some embryos may have a lower accuracy than others, and that this would be noted on your PGD results report.

Sample PGD results

<table>
<thead>
<tr>
<th>EMBRYO #</th>
<th>DIAGNOSIS</th>
<th>EMBRYO TRANSFER</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>UNBALANCED</td>
<td>NO</td>
</tr>
<tr>
<td>5</td>
<td>UNBALANCED</td>
<td>NO</td>
</tr>
<tr>
<td>6</td>
<td>UNBALANCED</td>
<td>NO</td>
</tr>
<tr>
<td>9</td>
<td>BALANCED/NORMAL</td>
<td>YES</td>
</tr>
<tr>
<td>10</td>
<td>BALANCED/NORMAL, abnormal for chromosome 21</td>
<td>NO</td>
</tr>
<tr>
<td>12</td>
<td>UNBALANCED</td>
<td>NO</td>
</tr>
<tr>
<td>13</td>
<td>NO RESULT</td>
<td>NO</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.
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 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 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 egg or 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 at RGI (where they have been performed on a clinical basis for over 25 years) or through a physician local to you. Please contact one of our genetic counselors if you have questions or need help coordinating testing.
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. If you are working with an RGI-affiliated physician in Chicago, the IVF fees will be paid to this physician’s office.

Insurance

Most insurance companies do not cover PGD. However, as a courtesy, RGI will attempt to verify your insurance benefits for PGD. 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, then RGI may not require any fees to be paid up front and will submit all costs to insurance after your 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 RGI does not currently accept all insurance plans for aCGH/microarray.

Please contact billing@reproductivegenetics.com with any questions about insurance billing.
**NEXT STEPS**

1. After reviewing this information packet, please contact one of our genetic counselors by calling (773) 472-4900 or emailing info@reproductivegenetics.com. They will be able to answer questions regarding our center and help you to start this process if you are interested in pursuing PGD. They will also be able to give you PGD cost information. Insurance inquiries can be directed to billing@reproductivegenetics.com. For information on IVF costs, please contact your local IVF physician or contact our center for a referral to our affiliate physician.

2. The genetic counselor 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 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 if a set-up is needed, the blood collection kits. Once we have all of the samples, the necessary notarized original consents and required payment, we will start your PGD set-up. 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. **Testing by aCGH/microarray does not usually require a set-up.**

You have the option of undergoing IVF with an affiliated physician at our center 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 like to pursue your IVF cycle at RGI in Chicago:**

   a) Your genetic counselor will put you in touch with one of our Reproductive Endocrinologists, to arrange a new patient consultation. This consult can be done over the phone or in person.

   b) The physician will send you a list of required pre-IVF laboratory tests. If you are not local to RGI, these tests can often be arranged through your local OBGYN or IVF center.

   c) The physician will review all pre-IVF laboratory test results and will select the medication protocol for ovarian stimulation.

   d) You must have a center to monitor you while on IVF medications. This monitoring can be done through our IVF center or through a center closer to your home, and will be coordinated by our affiliated IVF physician.

   e) You may arrive in Chicago before or during ovarian stimulation and complete the monitoring here. We need you and your male partner (or a frozen sperm sample) to be in Chicago on the day of egg retrieval. Your physician will discuss the specific length of time required for your stay (or the number of visits required if you plan to travel back and forth).
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 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 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 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 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 one of our genetic counselors to get information regarding the process, cost and to schedule a free consultation. You can reach us by phone (773) 472-4900 or by email at info@reproductivegenetics.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 cases done by aCGH/microarray, there is no waiting period so an IVF cycle can begin whenever your physician is ready. For cases done by FISH, there is often a set-up period and 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 Chicago is not usually necessary. Please see pages 17 and 18.

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 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:  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.