What is the accuracy of the testing?

Preimplantation testing is not 100% accurate. There is a risk of misdiagnosis with each sample tested, which could be due to human error, inherent technological limitations, and the possibility of mosaicism (more than one cell line) in the embryo. Please ask one of our genetic counselors about the benefits and limitations of testing different sample types.

Does PGD replace prenatal testing?

No, PGD does not replace prenatal testing, such as chorionic villus sampling (CVS) or amniocentesis. PGD is a research-based test allowing for a similar diagnosis to those available by prenatal testing. However, prenatal testing is still recommended, as this is currently the standard-of-care. Our genetic counselors can discuss what prenatal testing options are available to you.

What is the cost?

Please contact one of our genetic counselors for updated pricing information.

FREQUENTLY ASKED QUESTIONS

What are the limitations of PGD?

As with prenatal testing, PGD is aimed at reducing your chances of having a child with a genetic disease or chromosome problem; however, it does not test for all birth defects. Every couple, regardless of their ethnic background and family history, has a 3-5% risk for birth defects with each pregnancy. With PGD, this risk is reduced but not eliminated.

Will my insurance cover the costs of PGD?

Most insurance carriers will not cover the cost of PGD, however, some do. As insurance policies vary, it is always to your benefit to check with your insurance carrier. It is your responsibility to contact your insurance carrier regarding your coverage.

How do I get more information about PGD?

To receive additional information, please contact our office at (773) 472-4900. You can also send an email request to rgiworld@gmail.com.

How do I get started?

Please contact one of our genetic counselors for an initial consultation at (773) 472-4900. They will provide the necessary information to begin the process for your family.

REPRODUCTIVE GENETICS INSTITUTE

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Chicago, Illinois 60657

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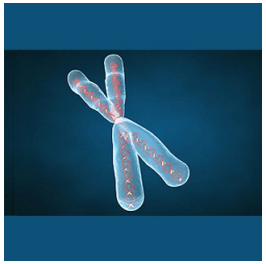
REPRODUCTIVE GENETICS INSTITUTE

24 Chromosome Testing



BY MICROARRAY TECHNOLOGY
(array CGH)

What is preimplantation genetic diagnosis?

By definition, preimplantation genetic diagnosis (PGD) is the diagnosis of a genetic condition prior to achievement of a pregnancy. PGD requires the use of In Vitro Fertilization (IVF). PGD was first performed in the early 1990's as a way for couples to prevent a pregnancy with a genetic disease. Currently, we are able to perform PGD for many genetic conditions including single gene disorders and chromosomal abnormalities. At RGI, we have 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.

What is IVF?

IVF (in vitro fertilization) refers to a process by which the eggs are retrieved from the ovaries of a woman before they are released, and fertilization of the eggs occurs in the laboratory. The resultant embryos are placed back into the woman's uterus several days later. An IVF cycle includes the woman taking injectable medications to stimulate the ovaries to produce more than one egg at a time. This is a fairly complicated process which requires a number of physician visits and monitoring. IVF is required when an individual chooses to have PGD testing performed.

What are chromosomes?

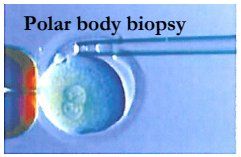
Chromosomes are the structures in our cells that carry our genetic information or genes. Our genes determine our physical makeup, growth and development. Normally, we have 46 chromosomes in each cell. Our children inherit 23 chromosomes from each parent, in the egg and sperm. A woman's risk for having an abnormal number of chromosomes in her eggs increases with her age. PGD testing can detect chromosomal abnormalities found during pregnancy, including Down syndrome (Trisomy 21), Trisomy 13 and Trisomy 18, as well as abnormalities found in any other chromosome.

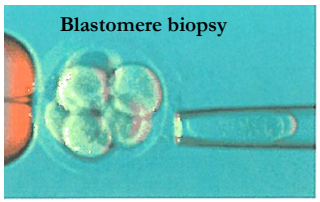
How can PGD help my family and me?

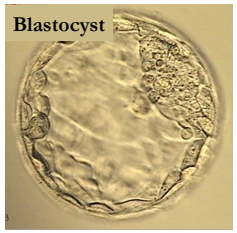
Preimplantation genetic testing is currently the only way to determine if the egg or embryo contains an abnormal number of chromosomes prior to pregnancy. This may not only contribute to the prevention of the birth of children with common chromosomal abnormalities, but also to the efficiency of IVF. PGD for chromosome problems can increase the chance of an embryo implanting, decrease the chance of miscarriage and decrease the likelihood of having a pregnancy with a chromosome problem (like Down syndrome). In essence, PGD can give your physician another way to determine which are the best embryos for transfer.

How is the testing performed?

There are three basic sample types that can be tested – polar bodies (from fertilized eggs), blastomeres (from the 3-day cleavage stage embryo), and trophectoderm (from the 5-6 day blastocyst). Each type has its advantages and its limitations. Polar body testing focuses on the maternal contribution and is an earlier method of testing, which in some instances may have a higher accuracy. Both blastomere and blastocyst testing provide information on both maternal and paternal genetic contributions. Blastomere testing allows for embryo transfer during the same cycle, but may not be as accurate as polar body or blastocyst testing. Blastocyst testing may require embryo freezing and transfer at a later date.

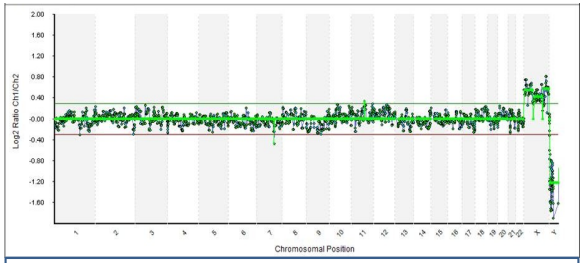




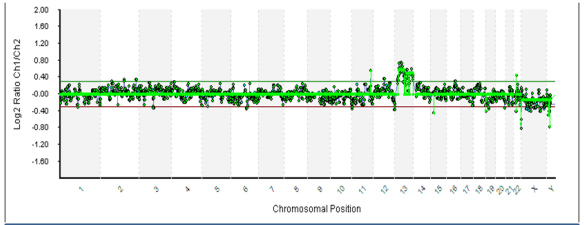


What is array-CGH analysis?

Array-CGH allows the laboratory to determine if the correct number of each chromosome is present in the egg or embryo. This technology simultaneously tests for all 24 chromosomes (1-22, X and Y). Chromosome abnormalities are associated with failed implantation, miscarriage, or live births with multiple anomalies. With array-CGH, the amount of DNA present for each chromosome is compared to that of a normal standard, enabling us to detect monosomies (missing chromosomes), trisomies (extra chromosomes), and other abnormalities. Array-CGH typically does not require additional blood tests or a set-up period, and therefore there is no wait time or penalty for cancellations.



Normal female



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

When do I get results?

Timing of results depends on the type of biopsy performed. In cases where the polar bodies (Day 0/1) or blastomeres (Day 3) are being studied, results are available in time for a day 5 transfer. If trophectoderm (Day 5) is being analyzed, results are not usually ready in time for a transfer in the same cycle. Therefore, embryos may need to be frozen and thawed at a later date for transfer.