

# Preimplantation Genetic Screening / Diagnosis

## “Why Consider Genetic Testing”

**Q: I am 40 years old, recently married, and my husband has severe male factor infertility with a very low sperm count under 2 million. My gynecologist suggested that we should see a Reproductive Endocrinologist (RE) for In Vitro Fertilization (IVF) because of my age and the low sperm count. He also mentioned the possibility of doing genetic testing on our embryos before putting them back into my uterus in order to significantly reduce our chances for miscarriage and certain birth defects, such as Down’s syndrome. Can you give me more information on the genetic testing?**

**A:** Your age and his severe male factor problem are major obstacles to a successful pregnancy. Unfortunately, for women ages 40-44, 50 -100% of their eggs may be genetically abnormal, causing infertility but also significantly increasing the risks for miscarriages and genetic birth defects. If you are going to use your eggs (instead of donor egg), IVF is your best option so your egg quality as well as embryo quality can be evaluated and then a few of the healthiest appearing embryos can be transferred to your uterus. Furthermore, assessing many of the chromosomes of each normally developing embryo, for *structural abnormalities or abnormal numbers of chromosomes (Preimplantation Genetic Screening, or PGS)*, allows the RE to be even more selective and transfer only genetically normal appearing embryos. PGS can reduce the risk of miscarriage as well as the risk of many genetic birth defects. In addition, PGS may slightly increase pregnancy success rates, but only when you end up with more embryos than you plan to transfer back to the uterus.

This technology was first developed in the early 1990s. The technique involves the microscopic removal of generally a single cell from a day 3 developing embryo. Most normal embryos on day 3 have 5-8 cells, so removal of one cell usually does not disrupt the embryo. The chromosomes chosen for testing (usually Chromosomes # 13 14 15 16 17 18 21 22 X and Y) account for over 90% of the genetic miscarriages and birth defects. For example, Down’s syndrome is caused by an extra # 21 chromosome (Trisomy 21). Aneuploidy (any abnormal number of chromosomes....missing or extra) increases dramatically as women age. If PGS is not performed, any transferred embryos demonstrating aneuploidy will either not implant in the uterus, or result in a miscarriage or birth defect---no good outcome. PGS should not be looked at as a substitute for CVS- Chorionic villi sampling or for amniocentesis, but it markedly reduces the chances for genetic birth defects.

The most common situations for recommending PGS include:

- Women age 39 or older (although some women 35-38 ask for the procedure)
- Severe male factor (especially when testicular biopsy is needed to obtain sperm)
- Miscarriages (2 or more genetic or unexplained losses)
- IVF failures ( 2 or more failures, despite normal appearing quality embryos)

In contrast, there are genetic diseases secondary to '*single gene disorders*' that result from mutations affecting individual genes on a chromosome. **Preimplantation Genetic Diagnosis, or PGD**, involves assessing the chromosomes for these single gene abnormalities.

The most common indications for recommending PGD include:

- Previous birth of a child with a single gene disorder (examples - Cystic Fibrosis, Tay Sachs, Muscular Dystrophy, Hemophilia, or Sickle cell to name a few)
- Both partners are 'carriers' for a single gene disorder, based on screening tests and therefore at risk for passing on inherited genetic disease to their offspring

Two newer technologies, called **MicroArray** and **Comparative Genomic Hybridization (CGH)** allow for the assessment of all 23 chromosome pairs instead of the 9 or 10 chromosomes currently tested.

At Fertility Centers of Illinois (FCI), we use 2 excellent genetics labs for our PGS/PGD testing. For more information, you can go to our FCI website [www.FCIconline.com](http://www.FCIconline.com) or to the websites of our genetics labs:

- Reprogenetics (PGS/PGD and CGH) [www.reprogenetics.com](http://www.reprogenetics.com)
- Center for Preimplantation Genetics (PGS/PGD and MicroArray) [www.PGDcenter.com](http://www.PGDcenter.com)

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