Selecting Healthy Embryos with Preimplantation Genetic Diagnosis (PGD) and Preimplantation Genetic Screening (PGS)

Genetic abnormalities, whether the result of an inherited condition or DNA damage in the egg or sperm, can prevent couples from being able to have and raise a healthy child. Fortunately, a genetic testing procedure known as preimplantation genetic diagnosis (PGD) or preimplantation genetic screening (PGS) offers such couples a chance to become biological parents with minimal risk of passing on a serious genetic disorder.

Preimplantation Genetic Diagnosis (PGD) and Preimplantation Genetic Screening (PGS) require just one extra step in the IVF treatment process. Once the eggs have been fertilized and embryonic development begins, an embryo biopsy is performed. Up to five cells from each embryo on day five of development are removed in our laboratory, and then we send the biopsy to our reference genetics laboratory where genetic specialists can determine which embryos carry a genetic anomaly and which are normal. Only the normal, healthy embryos are transferred to the uterus or cryopreserved (vitrified) for later use.

Preimplantation genetic screening (PGS) is done to screen all 23 pairs of chromosomes found in each cell. This evaluation is to check for aneuploidy - abnormal numbers of chromosomes, such as conditions like Down syndrome also known as Trisomy 21. Embryos demonstrating aneuploidy (any missing or extra chromosomes) will result in miscarriage or various birth defects. These aneuploid abnormal embryos are therefore never transferred back into the woman's uterus. PGS is an optional screening procedure for those couples with advanced age, or those who have failed two or more IVF cycles or severe male factor infertility or those who have suffered multiple miscarriages without a known cause.

Pre-implantation genetic screening (PGS) also allows Dr. Jacobs to identify many serious genetic abnormalities in our patients' embryos before they are inserted in the uterus. Genetic diagnosis detects many abnormalities in embryos, and also allows parents to choose the gender, if they prefer. Gender selection also allows patients to avoid some sex-linked genetic abnormalities involving the X chromosome. This selection process has more than 99 percent accuracy and is completely safe for embryos.

Aneuploidy (abnormal chromosome numbers) such as Down syndrome (Trisomy 21) is the most common reason for couples of advanced age to have PGS performed. Genetic testing
also makes gender selection possible for couples carrying X-linked recessive genes, such as Hemophilia, or those simply interested in gender selection (family balancing).

**Preimplantation genetic diagnosis (PGD)** in contrast is not a general screening procedure. It is recommended for couples who know they have the potential to pass on a serious genetic condition like Tay-Sachs or sickle cell anemia. PGD is recommended when both genetic parents have a known genetic mutation, such as cystic fibrosis (CF) -- they are referred to as carriers. To find out if you are a ‘carrier’ for a genetic mutation you can undergo pre-conception genetic screening with a blood test from Counsyl. ([www.Counsyl.com](http://www.Counsyl.com)) PGD is performed on embryos to determine if there are two mutations (one from each parent), which will result in the child having the actual genetic disease.

PGD and PGS are well-established genetic testing procedures that dramatically reduce the risks of having a child with a known hereditary disorder such as cystic fibrosis (CF), as well as reducing certain birth defects like Down syndrome and miscarriages by not transferring genetically abnormal embryos. However, further testing once the pregnancy has been established is still recommended, just to be sure. A chorionic villi sampling (CVS), performed at about 9 to 12 weeks, or an amniocentesis, performed after at least week 14, can confirm that no apparent genetic abnormalities are present.

The newest genetic technologies, called MicroArray and **Comparative Genomic Hybridization (CGH)** and Next Generation Sequencing (NGS) have improved pregnancy outcomes.

At Fertility Centers of Illinois (FCI), after we do the embryo biopsy, we use several excellent genetics labs for our PGS/PGD testing. For more information, you can go to my website ([www.IVFChicago.com](http://www.IVFChicago.com)) or the FCI website ([www.fcionline.com](http://www.fcionline.com)).

**L A J**

**Laurence A. Jacobs M.D.**

Laurence.Jacobs@integramed.com

---

**IVF CHICAGO.COM**

*Your Miracle. Our Mission.*

Revised 3-10-15