



Preimplantation Genetic Screening (PGS) ©2006

Pre-implantation Genetic Screening (PGS), also known as PGS or preimplantation genetic diagnosis, is a technology that allows for genetic disease testing on embryos. PGS, first performed on human embryos in the late 1980's, requires that one or two cells, or blastomeres, be removed from an embryo and analyzed for genetic or chromosomal defects.

PGS is recommended for patients who have a history of heritable genetic disease, unexplained recurrent pregnancy loss or several failed IVF cycles, and for women of advanced maternal age. Individuals who are carriers of a genetic disease or chromosome translocation are at increased risk to have a child with a genetic or chromosomal disorder. In addition, women who are 35 years of age or older are at increased risk to have a child with a chromosome abnormality. In these instances, PGS can be used to help reduce the risk for the known genetic or chromosomal disorder in the family. There may be other reasons to perform PGS as determined by your physician.

In order to have PGS, a couple must consent to have in-vitro fertilization (IVF). A fact sheet on IVF is also available. Once the egg is fertilized in the laboratory, one or two cells are biopsied from the embryo, fixed on a slide and transferred to a laboratory for the genetic analysis. The reproductive endocrinologist with whom you are working will discuss the risks, benefits, limitations and procedures involved with IVF, embryo biopsy and cell fixing. The laboratory will then perform the requested genetic testing, usually with results in 1-2 days. Embryos that test normal will be used for transfer. Embryos that are abnormal will be discarded.

It is important to remember that this testing is being done on only one or two cells. The amount of DNA available for testing is therefore very limited and testing for only one type of genetic disorder is possible per embryo. For example, testing for abnormal chromosomes cannot be combined with testing for a specific genetic disorder, such as cystic fibrosis (CF). In addition, because the testing is only done on one or two cells from a single embryo, there is a possibility that the cells tested do not accurately represent the status of the entire embryo. Studies have shown that PGS is usually about 85% accurate, with the potential for both false-positive or false-negative results. This is why "diagnosis" in PGD was changed to "screening".

Because of the risk for an incorrect PGS diagnosis, prenatal diagnosis by amniocentesis or chorionic villus sampling testing is *strongly* recommended. PGS is only a screen to increase the chances of carrying a non-affected child. This means you will be less likely to have to face a decision regarding terminating an affected pregnancy. In many instances PGS allows a couple to both reduce the risk for a specified genetic or chromosomal disorder and be biologically related to their offspring. Alternatives to PGS would be testing by CVS or amniocentesis after pregnancy is achieved or no testing at all.

Finally, there is a 3% general population risk for birth defects and mental retardation that cannot be reduced even if PGS and subsequent prenatal testing is completed. This risk is addressed by certain routine screening tests in pregnancy and by comprehensive ultrasound, but can never be completely removed because some conditions cannot be diagnosed prenatally.