

Preimplantation Genetic Diagnosis (PGD): A New Option For Patients With Recurrent Pregnancy Loss

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Losing a pregnancy can be emotionally devastating. Often there is no identifiable predisposing cause for these losses, and patients are often relegated to attempting pregnancy again and hoping for a better outcome. Occasionally, a woman can continue to miscarry repetitively. While a number of predisposing factors for recurrent pregnancy loss exist, at least half the time there is no identifiable cause. However, when the miscarriage is analyzed, a majority of these failed pregnancies are genetically abnormal, most commonly due to an abnormal number of chromosomes (the structures which contain the genetic material). A novel option for therapy in these patients is to screen embryos for the most common chromosomal abnormalities that cause miscarriage through a procedure called preimplantation genetic diagnosis, or PGD.

PGD is a state of the art procedure that combines the technology of in-vitro fertilization (IVF) with new molecular biology techniques. Following fertilization of an egg, a single cell is removed from an embryo in a procedure called an "embryo biopsy". Special probes are used to detect genetic abnormalities. Embryos found to be void of chromosomal abnormalities or specific genetic disorders are then transferred to the patient's uterus.

PGD was originally intended to screen embryos to avoid transfer of known genetic disorders. Through PGD patients with a family history of certain diseases or who carried traits for inheritable diseases such as cystic fibrosis, Tay Sachs, and others can now achieve pregnancies with little genetic risk to the fetus. More recently, the use of PGD has gained acceptance as an option for patients with a history of recurrent first-trimester miscarriages due to a chromosomal abnormality or unexplained loss. When an embryo has an abnormal number of chromosomes, the embryo may be unable to implant in the uterus, implant and miscarry, or result in a child with birth defects. By using PGD, we may directly determine some chromosomal abnormalities prior to implantation, potentially increasing the likelihood of a successful pregnancy and decreasing the risk of miscarriage.

When PGD is performed in women with recurrent pregnancy loss, it is important to understand that not all potential genetic abnormalities are assessed. Unlike traditional prenatal diagnostic techniques like amniocentesis, which evaluate all 23 chromosome pairs for numerical and structural abnormalities, analysis through PGD is more complex and limited. With PGD only one or two cells are available for analysis, and results must be obtained very rapidly in order to make decisions on which embryos to transfer. Originally only a handful of chromosomes could be analyzed through PGD, but with advancing technology we are now able to ascertain numerical abnormalities in as many as ten or more chromosomes at once. Nevertheless, if a woman achieves a pregnancy through PGD, she may still carry a fetus with a chromosome abnormality if that chromosome was not included in the screening. Hopefully as the technology evolves, we will be able to screen all the chromosomes for numerical abnormalities in the near future, enabling an even greater rate of success in this patient population.

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