



Pre-implantation genetic **screening** (PGS) is the process of obtaining a few cells from a developing embryo following *in vitro* fertilization (IVF) in an attempt to determine if there are any extra or missing chromosomes (called aneuploidy). Embryos that appear to not have aneuploidy are then either selected for transfer to the uterus for implantation or can be frozen for future use. PGS is an available option to all patients or couples undergoing IVF.

Pre-implantation genetic **diagnosis** (PGD) refers specifically to testing an embryo for a *known* genetic difference in the family, such as a single gene disorder (like [cystic fibrosis](#)) or a chromosome rearrangement (such as a [chromosome translocation](#)). Unlike PGS, PGD is not an option for all patients or couples who complete IVF, as not all couples have a specific concern where PGD would be helpful.

### HOW IT WORKS

PGS and PGD involve the use of assisted reproductive technologies (ART) and can be performed as part of the IVF process. In brief, the following steps get performed:

1. A woman takes medications initially to suppress and then stimulate the ovaries in attempt to make multiple eggs grow
2. Eggs are retrieved with a transvaginal needle as part of a procedure
3. Eggs are fertilized with the male's sperm
4. Embryos develop in the laboratory over the course of roughly 5-6 days
5. On day 5-6 of embryo growth (also referred to as the blastocyst stage) a few cells are removed from the outer layer of the embryo and sent to a laboratory that will test for overall chromosome number (PGS) and/or the specific genetic difference in question (PGD).

Following removal of these cells for PGS/PGD, the embryos are immediately frozen using a process called vitrification. When the results of PGS and/or PGD are available, the embryo(s) desired for transfer can be thawed and placed into the uterus. If additional embryos remain, they can be frozen for future use.

### REASONS TO PURSUE

Chromosome abnormalities, such as aneuploidy, are a common reason for embryo implantation failure and miscarriage. In addition, the chance for aneuploidy in an embryo increases as a woman's age increases. A woman might choose to pursue PGS if she is



anticipated to be over the age of 35 at the time of a future delivery, given her risk for aneuploidy is higher. Some couples would choose to undergo PGS to lower their chance to have a pregnancy with a chromosome condition. Other couples have had a history of [multiple miscarriages](#), and use PGS in an attempt to maximize their chance for a successful pregnancy.

Patients might elect to do PGD if a parent has a diagnosis of a single gene disorder that could be directly passed on to their children. Parents who are healthy but are carriers of an [autosomal recessive](#) condition with a 25% chance of having an affected child might also choose to pursue PGD. Additionally, a parent that was found to be a carrier of a [chromosome translocation](#) may choose to complete PGD to reduce the odds of a chromosome imbalance in a pregnancy.

It is possible to pursue both PGS and PGD on an embryo, when indicated.

### **LIMITATIONS**

PGS and PGD are unable to test for all types of genetic abnormalities in an embryo. While PGS and PGD are thought to be accurate, there have been both false-positive and false-negative results. For this reason, [prenatal screening](#) and [diagnostic](#) testing options are still offered in pregnancies that have had PGD and/or PGS.

### **OTHER FACTORS**

Using IVF with PGS and/or PGD does not guarantee a healthy pregnancy. The IVF process alone can be time-intensive as well as emotionally and financially demanding. PGS and PGD are both extra steps in the IVF process and consequently, tend to increase the overall cost of IVF. It is also possible that insurance will not cover the cost of such testing.

Click [here](#) to learn more about scheduling a genetic counseling appointment for pregnancy-related questions.

Click [here](#) to learn more about scheduling a genetic counseling appointment for infertility or preconception questions.

Click [here](#) to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.



## Preimplantation Genetic Screening/Diagnosis