Fact Sheet

From ReproductiveFacts.org

The Patient Education Website of the American Society for Reproductive Medicine

Preimplantation genetic testing

(including preimplantation genetic diagnosis and preimplantation genetic screening)

What is preimplantation genetic testing (PGT)?
Your embryos can be tested for abnormal chromosomes before they are transferred to the uterus. This is called preimplantation genetic testing, or PGT. It is done in a lab, using in vitro fertilization (IVF). To learn more about IVF, please see the ASRM fact sheet titled *In vitro fertilization (IVF)*. One or more cells from each embryo is sent for genetic testing. Genetically healthy embryos are transferred to the uterus, where they may attach to its lining and produce a pregnancy.

What genetic tests can be performed?
Cells from the embryo can be tested for a gene that may produce a specific disease. This will show if an embryo is normal (no disease) or affected (has the disease). It will also show if the embryo is carrying the disease but unaffected by it. An unaffected carrier is someone who can pass the disease to his or her children, but does not personally have any signs of the disease.

A karyotype is another kind of genetic test that can check the chromosomes. It is done during a pregnancy, either by a chorionic villus sampling or amniocentesis. This test shows if the fetus has more or fewer chromosomes than the usual 46. Having too many or too few chromosomes can cause disorders such as Down syndrome (Trisomy 21) or Trisomy 13. This test can also look for out-of-order chromosomes, called translocations, which can cause problems with growth or function.

The number of tests for genetic problems keeps growing every year.

What is preimplantation genetic diagnosis (PGD)?
PGD involves testing an embryo before it implants for a specific, known genetic disorder. PGD is used so that embryos unaffected by the disorder can be returned to the uterus.

What is preimplantation genetic screening (PGS)?
PGS involves testing an embryo for chromosomal abnormalities. Many embryos with chromosomal abnormalities will not lead to a pregnancy, or will result in a miscarriage. All women will have some eggs that are chromosomally abnormal. All men will have some sperm that are chromosomally abnormal. The percentage of embryos that are abnormal can be affected by many factors, including the age and health history of the parents.

How is genetic testing performed?
One or more cells are taken from an embryo and sent for genetic testing while the embryo is growing in the IVF laboratory. The testing can be done at different stages using different techniques. Techniques include fluorescent in situ hybridization (FISH), microarray, and single-nucleotide polymorphism (SNP).

Techniques are constantly evolving and changing to be more accurate, easier to run, and give more information. Newer techniques that include microarray and genome sequencing are more common methods of genetic testing.

Who should have PGD or PGS?
PGD is for someone who has or is a carrier of a known genetic disorder. PGS may be recommended for someone with recurrent miscarriage, someone who is older or whose ovaries do not work as well as expected, or someone with multiple failed fertility treatments. Some couples also can choose to do PGS for personal reasons.

Anyone interested in PGD or PGS should consult with a fertility specialist so that the procedure and any alternatives can be discussed.

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For more information on this and other reproductive health topics, visit www.ReproductiveFacts.org

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