



FERTILITY FACT SHEET

San Ramon / Orinda / San Jose

Preimplantation Genetic Screening and Diagnosis

Preimplantation genetic testing is a diagnostic test used in conjunction with in vitro fertilization (IVF) and is performed on embryos to assist detection of known genetic diseases or chromosomal abnormalities. When the testing is performed looking for a specific genetic disease that is *known* to be present in a family, the test is called Preimplantation Genetic *Diagnosis* (PGD). When the testing is performed looking for a number of *possible* genetic problems with chromosome count or structure that *might* be present, it is called Preimplantation Genetic *Screening* (PGS). PGD and PGS have been shown to significantly decrease the risk of conceiving a child with a genetic or chromosomal abnormality.

The first part of a PGD/PGS cycle is an IVF cycle:

- The eggs are harvested and fertilized with sperm using Intra-Cytoplasmic Sperm Injection (ICSI)
- At the 3rd day of culture, the embryos are hatched (an opening is created in the outer layer with a laser) and placed back in the incubator for another 2-3 days
- Embryos that reach the blastocyst at the 5th or 6th day stage are biopsied
- Several cells from the embryo are removed through the previously made opening and are sent to a specialty genetics lab for testing.
- The embryos are frozen after the biopsy is done.
- Once the genetics report is completed, normal embryos are transferred to the woman's uterus, typically in the next month in a new cycle
- In embryo transfer treatment cycle, estrogen and progesterone are used to prepare the uterine lining

Indications for Preimplantation Genetic Diagnosis

For couples with a known genetic risk, PGD is a major medical breakthrough. This includes identified single gene disorders and genetic translocations. Examples include patients who are known carriers of Cystic Fibrosis or Sickle Cell Anemia.

Indications for Preimplantation Genetic Screening (PGS)

PGS is now often considered for the following situations:

- A woman's age (typically > 38 years)
- Two or more miscarriages
- Two or more IVF cycles that have not been successful

Studies have consistently shown that PGS does decrease the risk of miscarriage (6-10%) and does reduce the risk of facing a decision for genetic termination. Performing PGS as a part of the IVF cycle increases the treatment cost by about \$5000. It is not clear if *routine* use of PGS in the general population is cost-effective.

Different types of genetic tests

For a complete description of the different types of tests of genetic tests and some basic information about genetic disorders, please refer to our web site: <http://www.rscbayarea.com/pgd>. The most common genetic test used at RSC is a SNP-microarray which can analyze single genes and all 24 chromosomes. The genetics reference lab that performs this test is Natera and their website is: www.natera.com.

Limitations of Preimplantation Genetic Testing

PGD and PGS DO NOT screen for all potential genetic diseases and cannot guarantee a healthy child.

Studies are showing no increase risk of birth defects, however there are no long term outcome studies yet of children born through PGS with blastocyst biopsy and freezing.

Possible risks:

- Risks associated with IVF: ovarian hyperstimulation syndrome, multiple pregnancy, increase risk of birth defects not related to the genetic test.
- Risk of harm to the embryo from the biopsy or from freezing.
- Possibility of a false result. This may mean the transfer of an abnormal embryo thought to be normal or it may mean discarding a normal embryo thought to be abnormal.
- There may be no normal embryos to transfer.

Alternatives to PGD and PGS

- Testing the fetus during pregnancy can diagnose whether or not the fetus has a genetic abnormality.
 - Chorionic Villus Sampling is a procedure where a biopsy of the placenta is performed at 10-12 weeks into the pregnancy.
 - Amniocentesis is a procedure where the amniotic fluid is removed for analysis. Amniocentesis is performed at 14-16 weeks.
 - If an abnormality is detected by CVS or amniocentesis, the option would be to consider an abortion.
- Donor eggs or donor sperm can be used to avoid the risk of disease.
- There is also the option not to do prenatal testing, not to do IVF with genetic testing of embryos and take the risk of giving birth to a child with a genetic or chromosomal disorder.

Please note, if you are interested in having PGS testing done in your IVF cycle you must let your physician know about your decision before you start the IVF cycle in order to prepare the genetics reference lab.