What Is Reproductive Genetic Testing?

Genetic testing is the laboratory analysis of DNA, RNA or chromosomes. Testing also can involve analysis of proteins or metabolites that are the products of genes. Genetic results are used to predict risk of disease, screen newborns for disease, identify carriers of genetic disease, establish prenatal or clinical diagnoses or prognoses and direct clinical care.

Carrier testing is genetic testing to determine whether an individual carries one copy of an altered gene for a particular genetic condition. Carrier testing is done because of a family history of a genetic disorder. Some genetic conditions or diseases such as cystic fibrosis, sickle cell disease, and thalassemia are more prevalent in certain populations and carrier testing often is recommended to prospective parents. For these conditions, a person must have two copies of the genetic alteration to have the disease. Individuals who carry only one copy of the alteration are carriers and typically show no signs of the disease. When both parents are carriers, there is a one in four, or 25 percent, chance for each child to inherit the mutation from both parents and have the disease.

Preimplantation genetic diagnosis (PGD) is genetic testing on embryos produced through in vitro fertilization. Most commonly, one or two cells are removed from the embryo and tested for the presence of a particular genetic trait or alteration. The test results inform the selection of embryos for transfer to a woman's uterus to initiate a pregnancy.

Prenatal genetic testing (or prenatal genetic diagnosis) is genetic testing of fetal cells obtained through amniocentesis and chorionic villus sampling (CVS). Test results may be used to help parents prepare for the birth of an affected child or make a decision about terminating the pregnancy.

Prenatal screening involves tests and procedures used to assess fetal risk for health conditions including genetic disorders. It does not provide a definitive diagnosis of a genetic disease.

In Vitro Fertilization (IVF) is a process in which eggs are retrieved from a woman's ovaries and fertilized with sperm in the laboratory, and the resulting embryos are grown in a Petri dish and then transferred into a woman's uterus. Embryos that are not transferred may be discarded, frozen and stored for future use, donated to other couples or used for research.