

Preimplantation genetic diagnosis (PGD)/Preimplantation genetic screening (PGS), is a procedure carried out on embryos as part of some IVF cycles, before choosing which embryos to transfer into the mother's uterus. With PGD/PGS, cells from the embryo, called blastomeres, are removed, or biopsied, on the third, fifth, or sixth day after egg retrieval. Day 5 and day 6 biopsies are called trophectoderm biopsies. The biopsied cells from the embryos are dissolved in a lysis buffer, and transferred to a specialized genetics lab where DNA from the cells can be analyzed for genetic abnormalities, or for sex determination. Embryo biopsy is a highly skilled procedure performed by our embryology team. The entire process of embryo biopsy, transport to the genetics lab, and testing by the genetics lab takes two days. Once we have the report from the genetics lab, we can determine which embryo(s) to transfer back into the mother's uterus. PGD is especially useful for patients with inherited genetic disorders, such as cystic fibrosis, hereditary breast and ovarian cancer, sickle cell anemia, muscular dystrophy, and many others. The role for PGS is evolving. This technology allows for exclusion of embryos with disorders such as trisomies (e.g. Down's syndrome) and other embryos that have no potential for implantation and delivery (e.g. triploidies, monosomies, etc.), and it also can be used for sex selection. It may prove to be useful in improving pregnancy rates and decreasing multiple pregnancy risks, by identifying embryos with normal chromosome structure.