Preimplantation Genetic Screening and Diagnosis

Preimplantation Genetic Diagnosis (PGD) and Preimplantation Genetic Screening (PGS) are used to detect genetic conditions, chromosome abnormalities, and fetal sex during assisted reproduction with in vitro fertilization (IVF). PGD refers to embryo testing that is performed when one or both parents have a known genetic abnormality. This includes single-gene mutations and chromosome rearrangements. PGS refers to screening an embryo for aneuploidy when both parents are chromosomally normal. Genetic testing is performed on cells from the developing embryo prior to implantation. Only those embryos not affected with a genetic condition are implanted. PGD may allow at-risk couples to avoid a pregnancy affected with a genetic condition. The Society for Assisted Reproductive Technology and the American Society for Reproductive Medicine have published joint practice committee opinions to address the safety, accuracy, and overall efficacy of PGD and PGS.¹,²

- This guideline does not include prenatal or preconception carrier screening. Please refer to Genetic Testing for Carrier Status for that purpose.
- This guideline does not include prenatal genetic testing. Please see Genetic Testing for Prenatal Screening and Diagnostic Testing for genetic testing done during pregnancy.

Terminology for preimplantation genetic testing has recently been updated, with terms created for various clinical testing indications:³

- PGT-M: testing performed when the embryo is at an increased risk for a monogenic disorder
- PGT-SR: testing performed when the embryo is at increased risk for a structural chromosome rearrangement
- PGT-A: testing performed to screen an embryo for aneuploidy when both parents are chromosomally normal

Guidelines and Evidence

The following section includes relevant guidelines and evidence pertaining to PGD and/or PGS.
Society for Assisted Reproductive Technology and American Society for Reproductive Medicine Practice Committee Opinion

In a joint practice committee opinion, The Society for Assisted Reproductive Technology and The American Society for Reproductive Medicine stated the following:

- “PGD is indicated for couples at risk for transmitting a specific genetic disease or abnormality to their offspring.”
- “Due to the risk for conceiving a child with a genetic disease or other abnormality, counseling for couples considering PGD is required…”
- Suggested key points of genetic counseling include IVF and embryo biopsy-related risks, natural history of the tested condition, other reproductive options, limitations of preimplantation testing, and prenatal follow-up options.

Criteria

Benefit Exclusion

The Health Plan does not cover any services related to assisted reproduction, including any genetic testing done for the purpose of preimplantation genetic screening or diagnosis.

References


