Preimplantation Genetic Diagnosis (PGD)

In order to meet medically necessary criteria for PGD, both A and B must be met:

A. There must be documentation confirming that PGD is medically necessary to detect a single gene disorder or chromosomal abnormality whose expression in the fetus or child would be expected to have a significant adverse medical impact and that detection in the pre-implantation period would directly affect reproductive decisions.

B. One of the following clinical circumstances must be documented:
   1. One genetic parent has a balanced, reciprocal translocation or Robertsonian translocation
   2. One genetic parent has a single gene autosomal dominant disorder
   3. Both genetic parents are known carriers of the same single gene autosomal recessive disorder
   4. The female genetic parent is a known carrier of a single gene X-linked recessive disorder

The procedure to obtain a cell sample from an embryo for PGD is covered when the above criteria for PGD are met. However, the procedures and services (such as IVF) required to create the embryos to be tested and the transfer of embryos to the uterus after testing, are covered only for members with advanced reproductive technology (ART) benefits and who meet medical necessity criteria for IVF (in vitro fertilization).

The following information was used in the development of this document and is provided as background only. It is not to be used as coverage criteria. Please only refer to the criteria listed above for coverage determinations.
Historically, couples at high risk of transmission of a genetic disorder have had limited reproductive options, forced after prenatal diagnosis to choose between either termination of affected pregnancies or acceptance of the emotional and financial burden of having a child with severe disability and early mortality. Preimplantation genetic diagnosis (PGD) was introduced to enhance efficiency in assisted conception. It is a technique for reducing the burden of genetic disease performed on couples who are at risk of a specific inherited disorder and used to identify genetic defects present in embryos created through in vitro fertilization (IVF) before transferring them to the uterus.

PGD is performed in conjunction with IVF and is offered to both fertile and infertile couples. Introduced in 1990 as an experimental procedure, PGD has now become an established clinical option in reproductive medicine (Handyside, Kontogianni et al. 1990; Verlinsky, Ginsberg et al. 1990). Because only unaffected embryos are transferred to the uterus for implantation, PGD can provide an alternative to current post conception diagnostic procedures such as amniocentesis or chorionic villus sampling which are sometimes followed by pregnancy termination when results are unfavorable (Verlinsky, Cohen et al. 2004). PGD techniques are now also being utilized for preimplantation genetic screening (PGS) with the intent to identify potential genetic abnormalities in conjunction with IVF for couples without specific known inherited disorders.

With single gene disorders and inherited chromosomal abnormalities being the main indicators for PGD, the technique is available for most known genetic mutations. With that said, PGD can be considered a rapidly evolving technique. Put simply, PGD requires egg extraction, IVF, cell biopsy, genetic analysis and embryo transfer (Handyside, Kontogianni et al. 1990). At present, there are three different procedures utilized for cell biopsy, each with its own advantages and disadvantages, including polar body biopsy, cleavage-stage embryo biopsy and blastocyst biopsy. Depending on the whether the characteristic being tested for is associated with chromosomes or DNA, the sample can be analyzed in one of three ways including polymerase chain reaction (PCR), fluorescence in situ hybridization (FISH) and comparative genomic hybridization with new technologies emerging rapidly. Regardless of the methods, the results are used by parents and providers to select which embryos are transferred back to the uterus with the ultimate goal of establishing an unaffected pregnancy.

The accuracy and reliability of PGD are key issues and exploring these matters requires consideration of the technical challenges and risks inherent in the genetic test itself and in the IVF procedure that it entails. Any PGD strategy has to deal with the detection and avoidance of misdiagnosis from the onset with the risk and outcome relating directly to the type of genetic disorder for which testing is performed. Although PGD has been suggested as an alternative for current post conception diagnostic procedures, the amount of DNA available for testing is limited. Due to this risk, prenatal diagnosis by amniocentesis or chronic villus sampling testing is strongly recommended upon established pregnancy to confirm genetic health.

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MPC Medical Policy Committee

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