Medical Coverage Policy | Preimplantation Genetic Testing

EFFECTIVE DATE: 01|01|2024
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OVERVIEW

Preimplantation genetic testing involves the analysis of biopsied cells as part of an assisted reproductive procedure. It is generally considered to be divided into two categories: preimplantation genetic *diagnosis* and preimplantation genetic *screening*.

Preimplantation genetic *diagnosis* is used to detect a specific inherited disorder in conjunction with in vitro fertilization (IVF) and aims to prevent the birth of affected children to couples at high-risk of transmitting a disorder. Preimplantation genetic *screening* may also involve testing for potential genetic abnormalities in conjunction with IVF for couples without a specific, known inherited disorder.

MEDICAL CRITERIA

Medicare Advantage Plans and Commercial Products

Preimplantation genetic *diagnosis* may be considered medically necessary as an adjunct to in vitro fertilization (IVF) when one of the criteria listed below are met:

For evaluation of an embryo at an identified elevated risk of a genetic disorder based on genetic inheritance such as when:

- Both parents are known carriers of a single-gene autosomal recessive disorder; or
- One parent is a known carrier of a single-gene autosomal recessive disorder, and the parents have an offspring who has been diagnosed with that recessive disorder; or
- One parent is a known carrier of a single-gene autosomal dominant disorder; or
- One parent is a known carrier of a single X-linked disorder, or

For evaluation of an embryo at an identified elevated risk of structural chromosomal abnormality such as for a:

- Parent with balanced or unbalanced chromosomal translocation.

PRIOR AUTHORIZATION

Medicare Advantage Plans and Commercial Products

Prior authorization is required for preimplantation genetic *diagnosis* for Medicare Advantage Plans and is recommended for Commercial Products via the online portal for participating providers.

Note: Laboratories are not allowed to obtain clinical authorization or participate in the authorization process on behalf of the ordering physician. Only the ordering physician shall be involved in the authorization, appeal or other administrative processes related to prior authorization/medical necessity.

In no circumstance shall a laboratory or a physician/provider use a representative of a laboratory or anyone with a relationship to a laboratory and/or a third party to obtain authorization on behalf of the ordering physician, to facilitate any portion of the authorization process or any subsequent appeal of a claim where the authorization process was not followed and/or a denial for clinical appropriateness was issued, including any element of the preparation of necessary documentation of clinical appropriateness. If a laboratory or a third party is found to be supporting any portion of the authorization process, BCBSRI will deem the action a violation of this policy and severe action will be taken up to and including termination from the BCBSRI provider network. If a laboratory provides a laboratory service that has not been authorized, the service will be denied as the financial liability of the participating laboratory and may not be billed to the member.

POLICY STATEMENT

Medicare Advantage Plans and Commercial Products

Preimplantation genetic *diagnosis* may be considered medically necessary as an adjunct to in vitro fertilization (IVF) when the medical criteria above has been met.

Medicare Advantage Plans

Preimplantation genetic *diagnosis* as an adjunct to IVF is not covered in individuals or couples who are undergoing IVF in all situations other than those specified above in the Medical Criteria section.

Preimplantation genetic *screening*, alone or as an adjunct to IVF in patients or couples who are undergoing IVF, is not covered in all situations as the evidence is insufficient to determine that the technology results in an improvement in the net health outcome.

Commercial Products

Preimplantation genetic *diagnosis* as an adjunct to IVF is considered not medically necessary in individuals or couples who are undergoing IVF in all situations other than those specified above in the Medical Criteria section.

Preimplantation genetic *screening*, alone or as an adjunct to IVF in patients or couples who are undergoing IVF, is not medically necessary in all situations as evidence is insufficient to determine that the technology results in an improvement in the net health outcome.

Some genetic testing services are not covered and a contract exclusion for any self-funded group that has excluded the expanded coverage of biomarker testing related to the state mandate, R.I.G.L**. §27-19-81** described in the Biomarker Testing Mandate policy. For these groups, a list of which genetic testing services are covered with prior authorization, are not medically necessary or are not covered because they are a contract exclusion can be found in the Coding section of the Genetic Testing Services or Proprietary Laboratory Analyses policies. Please refer to the appropriate Benefit Booklet to determine whether the member's plan has customized benefit coverage. Please refer to the list of Related Policies for more information.

COVERAGE

Benefits may vary between groups and contracts. Please refer to the appropriate Benefit Booklet, Evidence of Coverage or Subscriber Agreement for applicable laboratory and not medically necessary/not covered benefits/coverage.

BACKGROUND

Preimplantation genetic testing describes various adjuncts to an assisted reproductive procedure in which either maternal or embryonic DNA is sampled and genetically analyzed, thus permitting deselection of embryos harboring a genetic defect before implantation of an embryo into the uterus. The ability to identify preimplantation embryos with genetic defects before implantation provides an alternative to amniocentesis, chorionic villus sampling, and selective pregnancy termination of affected fetuses. Preimplantation genetic testing is generally categorized as either diagnostic (preimplantation genetic diagnosis) or screening (preimplantation genetic screening). Preimplantation genetic diagnosis is used to detect genetic evidence of a specific inherited disorder, in the oocyte or embryo, derived from mother or couple, respectively, that has a high risk of transmission. Preimplantation genetic screening is not used to detect a specific abnormality but instead uses similar techniques to identify a number of genetic abnormalities in the absence of a known heritable disorder. This terminology, however, is not used consistently (eg, some authors use preimplantation genetic diagnosis when testing for a number of possible abnormalities in the absence of a known disorder), following a terminology change from 'preimplantation genetic screening' to 'preimplantation genetic testing in 2017.

Biopsy

Biopsy for preimplantation genetic diagnosis can take place at 3 stages: the oocyte, cleavage stage embryo, or the blastocyst. In the earliest stage, both the first and second polar bodies are extruded from the oocyte as it completes the meiotic division after ovulation (first polar body) and fertilization (second polar body). This strategy thus focuses on maternal chromosomal abnormalities. If the mother is a known carrier of a genetic defect and genetic analysis of the polar body is normal, then it is assumed that the genetic defect was transferred to the oocyte during meiosis.

Biopsy of cleavage stage embryos or blastocysts can detect genetic abnormalities arising from either the maternal or paternal genetic material. Cleavage stage biopsy takes place after the first few cleavage divisions when the embryo is composed of 6 to 8 cells (ie, blastomeres). Sampling involves aspiration of 1 and sometimes 2 blastomeres from the embryo. Analysis of 2 cells may improve diagnosis but may also affect the implantation of the embryo. In addition, a potential disadvantage of testing at this phase is that mosaicism might be present. Mosaicism refers to genetic differences among the cells of the embryo that could result in an incorrect interpretation if the chromosomes of only a single cell are examined.

The third option is sampling the embryo at the blastocyst stage when there are about 100 cells. Blastocysts form 5 to 6 days after insemination. Three to 10 trophectoderm cells (outer layer of the blastocyst) are sampled. A disadvantage is that not all embryos develop to the blastocyst phase in vitro and, when they do, there is a short time before embryo transfer needs to take place. Blastocyst biopsy has been combined with embryonic vitrification to allow time for test results to be obtained before the embryo is transferred.

Analysis and Testing

The biopsied material can be analyzed in a variety of ways. Polymerase chain reaction or other amplification techniques can be used to amplify the harvested DNA with subsequent analysis for single genetic defects. This technique is most commonly used when the embryo is at risk for a specific genetic disorder such as Tay-Sachs disease or cystic fibrosis. Fluorescent in situ hybridization (FISH) is a technique that allows direct visualization of specific (but not all) chromosomes to determine the number or absence of chromosomes. This technique is most commonly used to screen for aneuploidy, sex determination, or to identify chromosomal translocations. Fluorescent in situ hybridization cannot be used to diagnose single genetic defect disorders. However, molecular techniques can be applied with FISH (eg, microdeletions, duplications) and, thus, single-gene defects can be recognized with this technique.

A more recent approach for preimplantation genetic screening is with comprehensive chromosome screening using techniques such as array comparative genome hybridization and next generation sequencing.

Embryo Classification

Three general categories of embryos have undergone preimplantation genetic testing, which is discussed in the following subsections.

Embryos at Risk for a Specific Inherited Single-Gene Defect

Inherited single-gene defects fall into 3 general categories: autosomal recessive, autosomal dominant, and X-linked. When either the mother or father is a known carrier of a genetic defect, embryos can undergo preimplantation genetic diagnosis to deselect embryos harboring the defective gene. Sex selection of a female embryo is another strategy when the mother is a known carrier of an X-linked disorder for which there is no specific molecular diagnosis. The most common example is female carriers of fragile X syndrome. In this scenario, preimplantation genetic diagnosis is used to deselect male embryos, half of which would be affected. Preimplantation genetic diagnosis could also be used to deselect affected male embryos. While there is a growing list of single-gene defects for which molecular diagnosis is possible, the most common indications include cystic fibrosis, β-thalassemia, muscular dystrophy, Huntington disease, hemophilia, and fragile X disease. It should be noted that when preimplantation genetic diagnosis is used to deselect affected embryos, the treated couple is not technically infertile but is undergoing an assisted reproductive procedure for the sole purpose of preimplantation genetic diagnosis. In this setting, preimplantation genetic diagnosis may be considered an alternative to selective termination of an established pregnancy after diagnosis by amniocentesis or chorionic villus sampling.

Embryos at a Higher Risk of Translocations
Balanced translocations occur in 0.2% of the neonatal population but at a higher rate in infertile couples or those with recurrent spontaneous abortions. Preimplantation genetic diagnosis can be used to deselect embryos carrying the translocations, thus leading to an increase in fecundity or a decrease in the rate of spontaneous abortion.

Identification of Aneuploid Embryos
Implantation failure of fertilized embryos is common in assisted reproductive procedures; aneuploidy of embryos is thought to contribute to implantation failure and may also be the cause of recurrent spontaneous abortion. The prevalence of aneuploid oocytes increases in older women. These age-related aneuploidies are mainly due to nondisjunction of chromosomes during maternal meiosis. Therefore, preimplantation genetic screening has been explored as a technique to deselect aneuploid oocytes in older women and is also known as preimplantation genetic diagnosis for aneuploidy screening. Analysis of extruded polar bodies from the oocyte or no blastomeres at day 3 of embryo development using FISH was initially used to detect aneuploidy. A limitation of FISH is that analysis is restricted to a number of proteins. More recently, newer preimplantation genetic screening methods have been developed. These methods allow for all chromosomes' analysis with genetic platforms including array comparative genomic hybridization and single nucleotide variant chain reaction analysis. Moreover, in addition to older women, preimplantation genetic screening has been proposed for women with repeated implantation failures.

Definitions
Preimplantation genetic testing for aneuploidy (PGT-A) is used to screen for chromosomal aneuploidy in conjunction with IVF for couples.

Preimplantation genetic testing for monogenic disorders (PGT-M) are used to detect a specific single gene inherited disorder or chromosome rearrangement in conjunction with in vitro fertilization (IVF).

Preimplantation genetic testing for structural rearrangements (PGT-SR) are used to detect a specific single gene inherited disorder or chromosome rearrangement in conjunction with in vitro fertilization (IVF).

For individuals who have an identified elevated risk of a genetic disorder undergoing IVF who receive preimplantation genetic *diagnosis*, the evidence includes observational studies and systematic reviews. Relevant outcomes are health status measures and treatment-related morbidity. Data from observational studies and systematic reviews have suggested that preimplantation genetic diagnosis is associated with the birth of unaffected fetuses when performed for detection of single genetic defects and a decrease in spontaneous abortions for patients with structural chromosomal abnormalities. Moreover, preimplantation genetic diagnosis performed for single-gene defects does not appear to be associated with an increased risk of obstetric complications. The evidence is sufficient to determine that the technology results in an improvement in the net health outcome.

For individuals who have no identified elevated risk of a genetic disorder undergoing IVF who receive preimplantation genetic *screening*, the evidence includes randomized controlled trials (RCTs) and meta-analyses. Relevant outcomes are health status measures and treatment-related morbidity. Randomized controlled trials and meta-analyses of RCTs on initial preimplantation genetic screening methods (eg, fluorescent in situ hybridization) have found lower or similar ongoing pregnancy and live birth rates compared with IVF without preimplantation genetic screening. There are fewer RCTs on newer preimplantation genetic screening methods, and findings are mixed. Recent meta-analyses of newer methods have found some benefit in subgroups of patients (eg, advanced maternal age); however, the evidence is limited, and larger trials specific to these patient populations are needed. In a meta-analysis focused on safety outcomes, preimplantation genetic testing for aneuploidy was not associated with increased risk of adverse obstetric or neonatal outcomes compared with standard assisted reproductive techniques. Well-conducted RCTs evaluating preimplantation genetic screening in the various target populations (eg, women of advanced maternal age, women with recurrent pregnancy loss) are needed before conclusions can be drawn about the

impact on the net health benefit. The evidence is insufficient to determine that the technology results in an improvement in the net health outcome.

CODING

Medicare Advantage Plans and Commercial Products

The following CPT codes may be considered medically necessary for Medicare Advantage Plans and Commercial Products when medical criteria above are met:

89290 Biopsy, oocyte polar body or embryo blastomere, microtechnique (for pre-implantation genetic diagnosis); less than or equal to 5 embryos

89291 Biopsy, oocyte polar body or embryo blastomere, microtechnique (for pre-implantation genetic diagnosis); greater than 5 embryos

RELATED POLICIES

Biomarker Testing Mandate
Expanded Fertility Services
Infertility Services

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Provider Update, January/November 2026
Provider Update, November 2024
Provider Update, November 2023

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