

Medical Policy Preimplantation Genetic Testing

Policy Number: 044

	Commercial* and Qualified Health Plans	Mass General Brigham ACO	Medicare Advantage	OneCare	Senior Care Options (SCO)
Authorization required	X		Х	Х	X
No notification or					
authorization					
Not payable		Х			

^{*}Not all commercial plans cover this service, please check plan's benefit package to verify coverage.

Overview

The purpose of this document is to describe the guidelines Mass General Brigham Health Plan utilizes to determine medical necessity for preimplantation genetic testing (PGT) which includes preimplantation genetic diagnosis for single gene defects (PGT-M) and translocations (PGT-SR), and preimplantation genetic screening for aneuploidy (PGT-A).

Coverage Guidelines

Mass General Brigham Health Plan covers medically necessary preimplantation genetic diagnosis (PGT-M) (and associated assisted reproductive services, e.g., in vitro fertilization, intracytoplasmic sperm injection) for a debilitating, genetically defined and genetically predictable disease with early onset mortality or morbidity when there is no known treatment for the condition, or the available interventions are either inadequately effective or significantly burdensome. Mass General Brigham Health Plan does not cover PGT-A.

This process requires two authorizations, the first from Mass General Brigham Health Plan utilization management (UM). If approval is obtained, a second authorization must be obtained by EviCore.

The specialist and/or the primary care provider are responsible for providing all necessary clinical information including medical history of patient and partner or child, where appropriate. It is expected that the member/couple are counselled regarding the testing alternatives to PGT-M (e.g., amniocentesis, chorionic villous sampling), potential risks of PGT-M (embryo arrest, diagnostic uncertainly and unknown long-term effects of PGT-M), and that traditional prenatal diagnostic testing may still be recommended after successful PGT-M and pregnancy.

Authorization of PGT-M or PGT-SR is limited to the following criteria:

Preimplantation Genetic Testing (PGT)

- 1. Mass General Brigham Health Plan covers medically necessary PGT-SR to test for unbalanced chromosome rearrangements when one of the genetic parents is known to have an unbalanced translocation, a balanced reciprocal or Robertsonian translocation, or a microdeletion/duplication or other structural chromosomal abnormality associated with the birth of an affected child. Mass General Brigham Health Plan may require laboratory documentation of the genetic tests.
- 2. Mass General Brigham Health Plan covers medically necessary PGT-M to detect evidence of any of the following genetic disorders in an embryo when:



- a. Both genetic parents are known carriers of a single gene autosomal recessive disorder, or one of the genetic parents is a known carrier and they have a child who has been diagnosed with the disorder such as, but **not limited to** the following:
 - i. Canavan disease
 - ii. Cystic Fibrosis
 - iii. Epidermolysis Bullosa Simplex (autosomal recessive type)
 - iv. Familial dysautonomia
 - v. Fanconi's Anemia
 - vi. Gaucher Disease
 - vii. Hurler Syndrome
 - viii. Methylmalonic acidemia
 - ix. Propionic academia
 - x. Sickle Cell Anemia
 - xi. Spinal Muscular Atrophy Type I
 - xii. Spinocerebellar Ataxia (autosomal recessive type)
 - xiii. Tay-Sachs Disease
 - xiv. Thalassemia Syndromes
- b. One genetic parent is a known carrier of a single gene autosomal dominant disorder such as, but **not limited to**, the following:
 - i. Epidermolysis Bullosa (autosomal dominant type)
 - ii. Huntington's Disease
 - iii. Myotonic Dystrophy
 - iv. Neurofibromatosis Type I AND II
 - v. Retinoblastoma
 - vi. Spinocerebellar Ataxia (autosomal dominant type)
 - vii. Tuberous sclerosis
- c. The genetic female parent is a known carrier of a single gene X-linked recessive disorder such as, but **not limited to** the following:
 - i. Adrenoleukodystrophy
 - ii. Alport Syndrome
 - iii. Becker muscular dystrophy
 - iv. Fabry disease
 - v. Choroideremia



- vi. Duchenne muscular dystrophy
- vii. Fragile X syndrome
- viii. Hemophilia A & B
- ix. Hunter Syndrome
- x. Incontinentia pigmenti
- xi. Lesch-Nyhan Syndrome
- xii. X-linked intellectual disability
- 3. Members with ovaries at least 40 years of age must demonstrate adequate ovarian reserve evidenced by menstrual history and results from any of the following:
 - a. Clomiphene Citrate Challenge Test (CCCT) within the past 6 months by showing a Day 3 FSH level < 15 mIU/ml, Day 3 Estradiol Level < 80 pg/mL, and Day 10 FSH level < 15 mIU/ml; or
 - b. A CCCT within the parameters above performed within the past 12 months, and a Day 3 FSH level < 15 mIU/ml and Day 3 Estradiol Level < 80 pg/mL performed within the past 6 months; or
 - c. AMH level > 1.0 mg/mL or antral follicle count > 6 within the past 12 months, and Day 3 FSH <15 mlU/mL within the past 6 months.

Exclusions

- 1. PGT as an adjunct to infertility services for members who do not meet the *Preimplantation Genetic Testing* clinical coverage criteria above.
- 2. PGT is not covered for members who are not expected to be fertile due to:
 - a. age-related decline in fertility (including members with uterus/ovaries ≥44 years of age, and members with uterus/ovaries who have had FSH ≥15 mIU/mI at any time after their 40th birthday)
 - b. voluntary sterilization procedures unless the procedure has been reversed and criteria are met under "Individuals with a Sterilization Reversal" in the policy "Assisted Reproductive Services."
- 3. PGT for:
 - a. Screening for an euploidy (PGT-A) including in the setting of recurrent miscarriage, repeated failed implantation during IVF, or advanced maternal age.
 - b. Carrier testing to determine embryo's carrier status.
 - c. Human Leukocyte antigen (HLA) typing of an embryo to identify a future suitable stem cell, tissue, or organ transplantation donor.
 - d. Translocations which will always produce an abnormal gamete such as 45XX (21;21) & 45XY(21;21).
 - e. Gender selection in the absence of a documented X-linked disorder.
 - f. Selecting non-medical traits.
 - g. Selecting against predisposition to disease when there is no single known genetic or chromosomal defect that definitively causes the disease.
 - h. Late onset/adult-onset disorders that are not listed in criteria above.



i. Genetic conditions contributed to by donor egg and sperm.

Medicare Variation

Mass General Brigham Health Plan uses guidance from the Centers for Medicare and Medicaid Services (CMS) for medical necessity determinations for its Medicare Advantage plan members. National Coverage Determinations (NCDs), Local Coverage Determinations (LCDs), Local Coverage Articles (LCAs), and documentation included in the Medicare manuals are the basis for medical necessity determinations. When there is no guidance from CMS for the requested service, Mass General Brigham Health Plan's medical policies are used for medical necessity determinations. At the time of Mass General Brigham Health Plan's most recent policy review, CMS had no NCDs/LCDs for preimplantation genetic testing.

MassHealth Variation

Mass General Brigham Health Plan uses guidance from MassHealth for medical necessity determinations for its Mass General Brigham ACO members. When there is no guidance from MassHealth for a requested service, Mass General Brigham Health Plan's medical policies are used for medical necessity determinations. As of Mass General Brigham Health Plan's most recent policy review MassHealth did not consider preimplantation genetic testing payable.

OneCare and SCO Variation

Mass General Brigham Health Plan uses guidance from CMS for medical necessity determinations for its OneCare and SCO plan members. NCDs, LCDs, LCAs, and documentation included in the Medicare manuals are the basis for medical necessity determinations. When there is no guidance from CMS for the requested service, Mass General Brigham Health Plan uses medical necessity guidelines from MassHealth. When there is no guidance from CMS or from MassHealth, Mass General Brigham Health Plan's medical policies are used for medical necessity determinations.

Definitions

<u>Autosomal Dominant</u>: Autosomal dominant is one of several ways that a trait or disorder can be passed down through families. If a disease is autosomal dominant, it means you only need to get the abnormal gene from one parent in order for you to inherit the disease. One of the parents may often have the disease.

<u>Autosomal Recessive</u>: A disorder characterized by two mutated copies of the gene must be present in each cell in order for the disease or trait to develop. Affected persons usually have two unaffected parents who each carry a single copy of the mutated gene and they are known as carriers.

<u>Preimplantation Genetic Testing</u>: A test involving an embryo that has been created using assisted reproductive technology such as in-vitro fertilization. After the eggs are removed the eggs are fertilized. Those eggs which are successfully fertilized are developed into blastocyst. Five to ten cells are removed from the growing blastocyst in order to test for the specific genetic condition in question.

<u>X-linked Dominant Disorders</u>: Caused by mutations in the gene on the X chromosome. Females are more frequently affected than males and the chances of passing on an X linked dominant disorder differ between men and women.

<u>X-Linked Recessive</u>: Are caused by mutations in the genes on the X chromosome. Males are more frequently affected than females and the chances of passing on the disorder differ between men and women. Families with an X linked recessive disorder often have affected males but rarely affected females in each generation. A characteristic of X linked inheritance is that fathers cannot pass X-linked traits to their sons.

Codes



The following codes are included below for informational purposes only; inclusion of a code does not constitute or imply coverage.

Authorized CPT/HCPCS Codes	Code Description
89290	Biopsy, oocyte polar body or embryo blastomere, micro technique (for pre-implantation genetic diagnosis); less than or equal to 5 embryos
89291	Biopsy, oocyte polar body or embryo blastomere, micro technique (for pre-implantation genetic diagnosis); greater than 5 embryos

Summary of Evidence

PGT-M and PGT-SR are important techniques used in assisted reproduction to prevent transmission of genetic diseases and improve pregnancy outcomes. A retrospective study by Zou et al. (2024) of 53 PGT-M cycles reduced the chance of having an affected child. Clinical pregnancy rates were found to be nearly 75% with a live birth rate of nearly 90%. This highlights the effectiveness of PGT-M in helping couples at risk of genetic conditions. PGT-SR focuses on detecting chromosomal structural abnormalities, such as balanced translocations and inversions. A systematic review by lews et al. (2018) looked at data from 772 couples. While there was no significant difference in live birth rates between couples who used PGT-SR and IVF and those who conceived naturally, the utilization of PGT-SR and IVF significantly reduced the rate of pregnancy loss and shortened the average time from cycle start to positive serum HCG. Guidelines from the American College of Obstetricians and Gynecologists (ACOG, 2020) and ASRM recommend PGT-M and PGT-SR as valuable tools in reproductive medicine, especially when combined with proper genetic counseling. These technologies help select the healthiest embryos, improving the chances of successful pregnancy and reducing the risk of miscarriage and genetic disease. Overall, PGT-M and PGT-SR represent important advances in reproductive genetics, offering personalized approaches that improve embryo selection and pregnancy success for couples with genetic risks.

PGT-A is still-evolving tool in assisted reproductive technology. Although some studies demonstrate improved pregnancy and live birth rates with PGT-A, outcomes can vary depending on patient populations, making it unclear which groups benefit most consistently (Bhatt et al., 2021). Technical factors can affect results and raise concerns about false positives, which may lead to the discarding of potentially viable embryos (Cornelisse et al., 2020). Ethical issues also remain relevant, as PGT-A adds procedural complexity without guaranteeing success in every case (ACOG Committee Opinion, 2020). Thus, further research is essential before PGT-A can be universally recommended.

Related Policies

Assisted Reproductive Services/Infertility Services

Effective

January 2026: Ad hoc update. Updated prior authorization table and added variation for OneCare and SCO members. Fixed code disclaimer.

May 2025: Annual update. Added MassHealth variation. Clarified Medicare variation. Added one biological parent having an unbalanced translocation to conditions eligible for PGT-SR. Added Retinoblastoma to list of single gene autosomal dominant disorders. References updated.

March 2024: Annual update.

March 2023: Annual update. Medicare Advantage added to table. Subheading title changed to Preimplantation Genetic Testing (PGT). Exclusions updated. Medicare Advantage language added. Definitions updated. March 2022: Annual update. References updated.



March 2021: Annual update. References updated.

June 2020: Annual update. Under coverage guidelines added clarification statement regarding authorization requirements. Updated references.

March 2019: Annual update. Updated references.

August 2018: Ad hoc update. Added language under item 1 subheading Preimplantation Genetic Diagnosis to include (and associated assisted reproductive services; e.g. IVF, ICSI).

April 2018: Annual update.

July 2017: Annual update. Added the exclusion "PGD Services if the member or member's spouse are using illicit substances or abusing substances known to negatively interfere with fertility or fetal development (e.g., marijuana, opiates, cocaine, or alcohol)".

July 2016: Annual update.

July 2015: Annual update. Updated references.

August 2014: Annual update. Added language to Coverage Guidelines, "a debilitating genetic disease with early onset mortality or morbidity and when there is no known treatment for the condition, or the available interventions are either inadequately effective or significantly burdensome." Added exclusions: 3f selecting for non-medical traits and 3j Genetic conditions contributed to by donor egg and sperm.

June 2013: Annual update. Added specific genetic disorders.

June 2012: Effective date.

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