

# GENETIC TESTING: PREIMPLANTATION GENETIC TESTING

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## OVERVIEW

Preimplantation genetic testing involves analysis of biopsied cells from an embryo as a part of an assisted reproductive procedure. Preimplantation genetic testing for monogenic disorders ([PGT-M](#)) and preimplantation genetic testing for structural rearrangements ([PGT-SR](#)) are 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 an increased risk of transmitting either a gene mutation(s) or an unbalanced structural chromosomal rearrangement that can be typically targeted in this context. Preimplantation genetic testing for aneuploidy ([PGT-A](#)) is used to screen for potential chromosomal or subchromosomal abnormalities (e.g., chromosomal aneuploidy) in conjunction with IVF for couples; in this case testing is untargeted.

Genetic counseling is highly encouraged for patients considering and undergoing in vitro fertilization. Genetic counseling should be performed by an individual with experience and expertise in genetic medicine and genetic testing methods, such as a genetic counselor, medical geneticist, or advanced practice practitioner specializing in genetics.

All patients who undergo [PGT-M](#) or [PGT-SR](#) should be offered diagnostic testing via chorionic villus sampling (CVS) or amniocentesis for confirmation of results.

All patients who undergo [PGT-A](#) should be offered traditional diagnostic testing or screening for aneuploidy in accordance with recommendations for all pregnant patients.

## POLICY REFERENCE TABLE

The tests and associated laboratories and CPT codes contained within this document serve only as examples to help users navigate claims and corresponding coverage criteria; as such, they are not

comprehensive and are not a guarantee of coverage or non-coverage. Please see the [Concert Genetics Platform](#) for a comprehensive list of registered tests.

<a href="#">Coverage Criteria Sections</a>	Example Tests (Labs)	Common CPT Codes	Common ICD Codes	<a href="#">Ref</a>
<a href="#">Preimplantation Genetic Testing for Aneuploidy (PGT-A)</a>	Spectrum - 24-chromosome Preimplantation Genetic Testing for Aneuploidy (PGT-A) (Natera)	81229, 81479, 89290, 89291	N97.0, N97.9, Z31	2, 3, 4
	SMART PGT-A (Preimplantation Genetic Testing - Aneuploidy) (Igenomix)	0254U		
<a href="#">Preimplantation Genetic Testing for Monogenic Disorders (PGT-M)</a>	Spectrum PGT-M (Natera)	0396U	N97.0, N97.9, Z14.8, Z31	1, 2
	PGT-M (Cooper Genomics)	89290, 89291, 81479		
<a href="#">Preimplantation Genetic Testing for Structural Rearrangements (PGT-SR)</a>	Spectrum - Preimplantation Genetic Testing for Structural Rearrangements (PGT-SR) (Natera)	81228, 81229, 81479, 89290, 89291	N97.0, N97.9, Z14.8, Z31	2

## OTHER RELATED POLICIES

This policy document provides coverage criteria for preimplantation genetic testing. Please refer to:

- **Genetic Testing: Prenatal and Preconception Carrier Screening** for coverage criteria related to carrier screening.
- **Genetic Testing: Prenatal Diagnosis (via amniocentesis, CVS, or PUBS) and Pregnancy Loss** for coverage related to diagnostic genetic testing during pregnancy or for a pregnancy loss.
- **Genetic Testing: Noninvasive Prenatal Screening (NIPS)** for coverage criteria related to prenatal cell-free DNA screening tests.

- **Genetic Testing: Multisystem Inherited Disorders, Intellectual Disability, and Developmental Delay** for coverage criteria related to diagnostic genetic testing in the postnatal period.
- **Genetic Testing: General Approach to Genetic and Molecular Testing** for coverage criteria related to preimplantation genetic testing that is not specifically discussed in this or another non-general policy.

[back to top](#)

## COVERAGE CRITERIA

### PREIMPLANTATION GENETIC TESTING FOR ANEUPLOIDY (PGT-A)

- I. Preimplantation genetic testing for aneuploidy ([PGT-A](#)) (81229, 81479, 89290, 89291, 0254U) is considered **investigational**.

[back to top](#)

### PREIMPLANTATION GENETIC TESTING FOR MONOGENIC DISORDERS (PGT-M)

- I. Preimplantation genetic testing for monogenic disorders ([PGT-M](#)) (89290, 89291, 81479) may be considered **medically necessary** when:
  - A. The embryo is at an elevated risk of a genetic disorder due to one of the following:
    1. Both biological parents are known carriers for the same autosomal recessive disorder, **OR**
    2. One biological parent is a known carrier of an autosomal dominant disorder, **OR**
    3. One biological parent is a known carrier of an X-linked recessive disorder.

- II. Preimplantation genetic testing for monogenic disorders ([PGT-M](#)) (89290, 89291, 81479) is considered **investigational** for all other indications.

[back to top](#)

## PREIMPLANTATION GENETIC TESTING FOR STRUCTURAL REARRANGEMENTS (PGT-SR)

- I. Preimplantation genetic testing for structural rearrangements ([PGT-SR](#)) (81228, 81479, 81229, 89290, 89291) may be considered **medically necessary** when:
  - A. The embryo is at an elevated risk of a genetic disorder because one biological parent has a chromosomal rearrangement.
- II. Preimplantation genetic testing for structural rearrangements ([PGT-SR](#)) (81228, 81229, 81479, 89290, 89291) is considered **investigational** for all other indications.

[back to top](#)

## DEFINITIONS

1. **Preimplantation genetic testing for monogenic disorders (PGT-M)** and **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).
2. **Preimplantation genetic testing for aneuploidy (PGT-A)** is used to screen for chromosomal aneuploidy in conjunction with IVF for couples.

[back to top](#)

## BACKGROUND AND RATIONALE

### Preimplantation Genetic Testing for Aneuploidy (PGT-A)

### *American Society of Reproductive Medicine*

The American Society for Reproductive Medicine issued an opinion on the use of preimplantation genetic testing (PGS) for aneuploidy (2018) which concluded, "Large, prospective, well-controlled studies evaluating the combination of multiple approaches (genomics, time-lapse imaging, transcriptomics, proteomics, metabolomics, etc.) for enhanced embryo selection applicable in a more inclusive IVF population are needed to determine not only the effectiveness, but also the safety and potential risks of these technologies. PGT-A will likely be part of a future multidimensional approach to embryo screening and selection. At present, however, there is insufficient evidence to recommend the routine use of blastocyst biopsy with aneuploidy testing in all infertile patients." (p. 34)

This position was reaffirmed in a 2020 committee opinion regarding clinical management of mosaic results from preimplantation genetic testing for aneuploidy of blastocysts, stating, "It should be recognized that this document does not endorse nor does it suggest that PGT-A is appropriate for all cases of IVF." (p. 253)

### *American College of Obstetricians and Gynecologists (ACOG)*

The American College of Obstetricians and Gynecologists issued Committee Opinion No. 799 (2020, reaffirmed 2023) regarding Preimplantation Genetic Testing. The recommendations include the following:

"The clinical utility of preimplantation genetic testing-monogenic and preimplantation genetic testing-structural rearrangements is firmly established; however, the best use of preimplantation genetic testing-aneuploidy remains to be determined." (p. e133)

## **Preimplantation Genetic Testing for Monogenic Disorders (PGT-M)**

### *American Society for Reproductive Medicine*

The American Society for Reproductive Medicine published an opinion on the use of preimplantation genetic diagnosis (PGD) for serious adult-onset conditions (2013). The statement includes the following:

- "Preimplantation genetic diagnosis (PGD) for adult-onset conditions is ethically justifiable when the conditions are serious and when there are no known interventions for the conditions or the available interventions are either inadequately effective or significantly burdensome."

- “For conditions that are less serious or of lower penetrance, PGD for adult[-]onset conditions is ethically acceptable as a matter of reproductive liberty. It should be discouraged, however, if the risks of PGD are found to be more than merely speculative.”

The opinion also stated that physicians and patients should be aware that much remains unknown about the long-term effects of embryo biopsy on the developing fetus and that experienced genetic counselors should be involved in the decision process. (p. 54)

#### *American College of Obstetricians and Gynecologists (ACOG)*

The American College of Obstetricians and Gynecologists issued Committee Opinion No. 799 (2020, reaffirmed 2023) regarding Preimplantation Genetic Testing. The recommendations include the following:

"Preimplantation genetic testing comprises a group of genetic assays used to evaluate embryos before transfer to the uterus. Preimplantation genetic testing-monogenic (known as PGT-M) is targeted to single gene disorders. Preimplantation genetic testing-monogenic uses only a few cells from the early embryo, usually at the blastocyst stage, and misdiagnosis is possible but rare with modern techniques. Confirmation of preimplantation genetic testing-monogenic results with chorionic villus sampling (CVS) or amniocentesis should be offered." (p. 133)

#### **Preimplantation Genetic Testing for Structural Rearrangements (PGT-SR)**

##### *American College of Obstetricians and Gynecologists (ACOG)*

The American College of Obstetricians and Gynecologists issued Committee Opinion No. 799 (2020, reaffirmed 2023) regarding Preimplantation Genetic Testing. The recommendations include the following: "To detect structural chromosomal abnormalities such as translocations, preimplantation genetic testing-structural rearrangements (known as PGT-SR) is used. Confirmation of preimplantation genetic testing-structural rearrangements results with CVS or amniocentesis should be offered." (p. 133)

[back to top](#)

## REFERENCES

1. Ethics Committee of American Society for Reproductive Medicine. Use of preimplantation genetic diagnosis for serious adult onset conditions: a committee opinion. *Fertil Steril.* 2013;100(1):54-57. doi:10.1016/j.fertnstert.2013.02.043
2. Preimplantation Genetic Testing: ACOG Committee Opinion, Number 799. *Obstet Gynecol.* 2020 (reaffirmed 2023);135(3):e133-e137. doi:10.1097/AOG.0000000000003714
3. Practice Committees of the American Society for Reproductive Medicine and the Society for Assisted Reproductive Technology. Electronic address: ASRM@asrm.org; Practice Committees of the American Society for Reproductive Medicine and the Society for Assisted Reproductive Technology. The use of preimplantation genetic testing for aneuploidy (PGT-A): a committee opinion. *Fertil Steril.* 2018;109(3):429-436. doi:10.1016/j.fertnstert.2018.01.002
4. Practice Committee and Genetic Counseling Professional Group (GCPG) of the American Society for Reproductive Medicine. Electronic address: asrm@asrm.org. Clinical management of mosaic results from preimplantation genetic testing for aneuploidy (PGT-A) of blastocysts: a committee opinion. *Fertil Steril.* 2020;114(2):246-254. doi:10.1016/j.fertnstert.2020.05.014

[back to top](#)