

# MEDICAL POLICY

Medical Policy Title	Prenatal Genetic Testing
Policy Number	4.01.03
Current Effective Date	May 15, 2026
Next Review Date	January 2027

Our medical policies are guides to evaluate technologies or services for medical necessity. Criteria are established through the assessment of evidence based, peer-reviewed scientific literature, and national professional guidelines. Federal and state law(s), regulatory mandates and the member's subscriber contract language are considered first in the determination of a covered service.

(Link to [Product Disclaimer](#))

**This policy does NOT address neoplastic disorders.**

## POLICY STATEMENT(S)

- I. Genetic testing related to pregnancy management is considered **medically appropriate** for **ANY** of the following indications:
  - A. Preconception and prenatal parental carrier testing with supportive documentation of **EITHER** the following conditions:
    1. Suspected genetic etiology of pregnancy losses in a patient with a history of multiple (two (2) or more) spontaneous abortions, prior stillbirth or infant death; **or**
    2. Strong family history or relevant ethnicity of genetic disorders or the likelihood that the parents are carrier(s) for genetic disorders (e.g., Tay Sachs);
  - B. Prenatal in utero diagnostic testing (e.g., amniocentesis or chorionic villus sampling [CVS]) for **ALL** of the following conditions:
    1. Advanced maternal age, defined as 35 years of age or older at the estimated date of delivery;
    2. Abnormal quadruple maternal serum multiple marker screening, including alpha fetoprotein (MSAFP), human chorionic gonadotropin (hCG), unconjugated estriol (uE3), and dimeric inhibin A (DIA or INH-A);
    3. Abnormal fetal ultrasound examination revealing signs proven to be associated with fetal abnormality;
    4. Previous pregnancy that resulted in the birth of a child with chromosomal (e.g., Down's syndrome) or genetic abnormality or major malformation;
    5. Parental or family (first or second degree relative) history of a known genetic or chromosomal abnormality;
    6. History of multiple (three or more) spontaneous abortions in either partner (e.g., male with another female partner or female with another male partner);

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7. Suspected risk for a specific detectable fetal disorder based upon the maternal medical history (e.g., maternal metabolic disorders such as type 1 diabetes, Phenylketonuria);
  8. Previous child or first or second degree relative with a neural tubal defect;
  9. Fetal sex determination in pregnancies at risk for an X-linked hereditary disorder;
  10. Known exposure to a teratogen, described as any drug or agent producing abnormal fetal development (e.g., anticonvulsants, ionizing radiation, alcohol, lithium, isotretinoin/retinA), or to viral infections; **and**
  11. Consanguinity;
- C. Preimplantation genetic diagnosis for evaluation of an embryo, as an adjunct to an assisted reproductive procedure, for **ANY** of the following conditions:
1. Preimplantation genetic testing-monogenic (PGT-M) for partners who are known carriers of a potentially lethal or disabling genetic mutation with limited treatment options and who meet the following criteria:
    - a. Testing is supported by published peer-reviewed literature that demonstrates an improvement in health outcomes;

**and**

    - b. Both partners are known carriers of a single autosomal recessive gene; **or**
    - c. One partner is a known carrier of a single gene autosomal dominant disorder; **or**
    - d. One partner is a known carrier of a single X-linked disorder;
  2. Preimplantation genetic testing- aneuploidy (PGT-A) for partners with an elevated risk of chromosomal abnormality and prior parental history of offspring with aneuploidy;
  3. Preimplantation genetic testing-structural rearrangements (PGT-SR) for partners with an elevated risk of a potentially lethal or disabling genetic translocation with limited treatment options and one partner with a balanced or unbalanced chromosomal translocation.
- Please note: PGT-M performed to determine the human leukocyte antigen (HLA) or other marker status of an embryo as a potential future stem cell donor is considered a donor search. Coverage of PGT-M for this purpose is contract-dependent and, as most contracts exclude donor searches, likely to be **ineligible for coverage**.
- II. Embryo Biopsy (CPT 89290 and 89291) is considered **medically necessary** when criteria for preimplantation genetic testing and in vitro fertilization are met.
- III. Genetic testing for inheritable diseases in individuals seeking preconception or prenatal care is considered **medically appropriate** when **ALL** of the following criteria are met:
- A. There is reasonable expectation, based on family history, pedigree analysis, risk factors, and/or signs or symptoms that a genetically inherited condition exists;

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- B. The testing method is considered a proven method for the identification of a genetically linked disease;
  - C. The test results will influence decisions concerning disease treatment or prevention.
- IV. Targeted preconception or prenatal carrier screening for **ANY** of the following is considered **medically appropriate**:
- A. Spinal Muscular Atrophy (SMA);
  - B. Cystic Fibrosis;
  - C. Fragile X or fragile X-related disorders for **ANY** of the following indications:
    - 1. In women with a family history of fragile X-related disorders;
    - 2. Intellectual disability suggestive of fragile X syndrome; **or**
    - 3. In women with primary ovarian insufficiency (POI) or premature ovarian failure (POF) who have **BOTH**:
      - a. a personal or family history of ovarian failure; **and**
      - b. an elevated FSH level before age 40 years without a known cause (see policy guideline XII);
  - D. Individuals of Eastern and Central European Jewish (Ashkenazi) descent, for **ANY** of the following:
    - 1. Tay–Sachs disease (TSD);
    - 2. Canavan disease;
    - 3. Cystic fibrosis; and
    - 4. Familial dysautonomia;
  - E. Individuals of French-Canadian, or Cajun descent for:
    - 1. Tay–Sachs disease (TSD).
- V. Non-targeted preconception or prenatal carrier screening panels for autosomal recessive and X-linked genetic disorders may be considered **medically necessary** as an alternative to testing of individual genes (e.g., SMN1 gene and CFTR gene) for individuals who:
- A. Are pregnant or are considering pregnancy; **and**
  - B. Are at any risk level when **ALL** the following criteria are met:
    - 1. The natural history of each disease is well understood and there is reasonable likelihood that the disease is one with high morbidity or early mortality in the homozygous or compound homozygous state;
    - 2. Other biochemical or clinical tests to confirm carrier status either are not available, provide an indeterminate result, or are less accurate than genetic testing;

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3. The genetic test has adequate clinical validity to guide clinical decision-making and residual risk is understood;
  4. An association of the markers with the disorders has been established;
  5. If testing is performed by a panel, the panel meets the minimum number of recommended gene variants but does not exceed the maximum, as determined by The American College of Obstetricians and Gynecologists (ACOG) (see Policy Guideline XIII);
  6. Testing includes CFTR and SMNI genes; **and**
  7. Previous carrier screening has not been performed.
- VI. Genetic testing of products of conception for evaluation of two (2) or more consecutive pregnancy losses (recurrent pregnancy loss) is considered **medically appropriate**.
- VII. Non-targeted preconception or prenatal carrier screening panels greater than 22 genes are considered **not medically necessary**.
- VIII. Preconception or prenatal carrier screening for inherited disorders without family history or other risk factors is considered **not medically necessary** as part of routine care.

### RELATED POLICIES

#### Corporate Medical Policy

2.02.03 Genetic Testing for Inherited Disorders

2.02.17 Genetic Testing for Cystic Fibrosis

2.02.25 Non-invasive Prenatal Testing

2.02.42 Chromosomal Microarray (CMA) Analysis for Prenatal Evaluation and Evaluation of Patients with Developmental Delay/Intellectual Disability or Autism Spectrum Disorders

2.02.46 Whole Exome and Whole Genome Sequencing for Diagnosis of Genetic Disorders

4.01.05 Assisted Reproductive Technologies

### POLICY GUIDELINE(S)

- I. The Health Plan and its employees adhere to all State and Federal laws concerning the confidentiality of genetic testing and the results of genetic testing. All records, findings and results of any genetic test performed on any person shall be deemed confidential and shall not be disclosed without the written informed consent of the person to whom such genetic test relates. This information shall not be released to any person or organization not specifically authorized by the individual subject of the test or in compliance with applicable law.
- II. Genetic testing is appropriate only when performed by a qualified laboratory certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) and offered in a setting with adequately trained health care professionals who are qualified to provide appropriate pre- and post-test counseling.

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- III. Genetic testing is contract dependent. Coverage only applies to members with a valid contract; coverage is not provided for family members without a valid contract.
- IV. Supporting documentation required:
  - A. The following factors will be considered when determining the medical appropriateness of a genetic test:
  - B. There must be reasonable expectation based on family history, pedigree analysis, risk factors, and/or symptomatology that a genetically inherited condition exists. Autosomal recessive disorders may be present without a family history.
  - C. The genotypes to be detected by a genetic test must be shown by scientifically valid methods to be associated with the occurrence of the disease, and the analytical and clinical validity of the test must be established.
  - D. The clinical utility of the test must be established (e.g., test results will influence decisions concerning disease treatment or prevention).
  - E. Genetic testing should be performed for management or treatment of the patient and not only for knowledge purposes. Documentation should demonstrate how test results will impact treatment or medical management.
  - F. When there is family history or phenotype suggestive of a specific syndrome, results of targeted testing for the mutation associated with the syndrome should be documented prior to any panel testing. If targeted testing has not been performed, rationale as to why panel testing is medically necessary should be documented.
- V. Preconception and prenatal parental carrier testing should not be completed more than once per lifetime.
- VI. If genetic etiology for spontaneous abortions is suspected, documentation of karyotyping results for both partners are required for non-targeted/multi-gene panel testing requests.
- VII. A first-degree relative is a blood relative who shares approximately 50% of an individual's genes (parents, full siblings, and children). A second-degree relative is a blood relative who shares approximately 25% of an individual's genes (grandparents, grandchildren, aunts, uncles, nephews, nieces and half-siblings). A third-degree relative is a blood relative who shares approximately 12.5% of an individual's genes (great-grandparents, great-grandchildren, great-aunts, great-uncles, first cousin, and grand-niece or nephew.)
- VIII. All preconception, prenatal and PGD testing must be rendered in a setting with health care professionals who are adequately trained to provide the appropriate pre- and post-test counseling and testing must be performed by a laboratory qualified to perform the testing.
- IX. If the genetic test is being done for knowledge only, and that knowledge will not alter management or treatment of the patient or family member, then the testing is not medically appropriate.

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- X. Coverage of preconception and prenatal genetic testing and counseling and PGD are contract dependent.
- XI. Benefits for genetic testing by PGD are provided in accordance with the member's subscriber contract and the above medical criteria for genetic testing, dependent upon the member's infertility benefit. PGD will not be covered beyond the number of covered in vitro fertilization (IVF) cycles (Refer to Corporate Medical Policy #4.01.05 Assisted Reproductive Technologies-In Vitro Fertilization).
- XII. An elevated FSH level is defined as FSH greater than 40mIU/ml with an estradiol less than 50pg/ml, confirmed on two separate occasions at least one month apart, in women over 40 with greater than or equal to six (6) months of amenorrhea or irregular cycles with no other known cause.
- XIII. Non-targeted carrier screening applies to persons of any risk, including average risk. Any panel billed with CPT code 81443 for non-targeted carrier screening must include the CFTR and SMN1 genes. Non-targeted carrier screening panels should include the minimum number of genes but not exceed the maximum number of genes recommended by ACOG (2-22 conditions). A list of 22 conditions deemed reasonable to include in a carrier screening panel were published by ACOG in Committee Opinion No. 690: Carrier Screening in the Age of Genomic Medicine. These conditions are summarized below:

Condition	Carrier Frequency in General Population	Carrier Frequency in Specific Ethnic Groups
$\alpha$ -thalassemia	Unknown	African (particularly sub-Saharan): 1 in 3 Mediterranean: 1 in 30 Southeast Asian and Middle Eastern: 1 in 20
$\beta$ -thalassemia	Unknown	African American: <1 in 8 Ashkenazi Jewish: Varied Asian: 1 in 20 Mediterranean: 1 in 7
Bloom syndrome	<1 in 500	Ashkenazi Jewish: 1 in 100
Canavan disease	<1 in 150	Ashkenazi Jewish: 1 in 41
Cystic fibrosis	Unknown	African American: 1 in 61 Asian: 1 in 94 Ashkenazi Jewish: 1 in 24 Caucasian: 1 in 25 Hispanic: 1 in 58
Familial dysautonomia	<1 in 500	Ashkenazi Jewish: 1 in 31
Familial hyperinsulinism	<1 in 150	Ashkenazi Jewish: 1 in 52
Fanconi anemia C	<1 in 790	Ashkenazi Jewish: 1 in 89

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Fragile X syndrome	1 in 259	
Galactosemia	1 in 87	Ashkenazi Jewish: 1 in 127
Gaucher disease	<1 in 100	Ashkenazi Jewish: 1 in 15
Glycogen storage disease type 1A	<1 in 150	Ashkenazi Jewish: 1 in 71
Joubert syndrome	<1 in 500	Ashkenazi Jewish: 1 in 92
Medium-chain acyl-CoA dehydrogenase deficiency	Unknown	Caucasian: 1 in 50
Maple syrup urine disease types 1A and 1B	1 in 240	Ashkenazi Jewish: 1 in 81 (type 1B) Mennonite: 1 in 10 (type 1A-BCKDHA p.Y438N)
Mucopolipidosis IV	<1 in 500	Ashkenazi Jewish: 1 in 96
Niemann-Pick disease type A	<1 in 500	Ashkenazi Jewish: 1 in 90
Phenylketonuria	Unknown	Caucasian: 1 in 50 Irish: 1 in 34
Sickle cell anemia	Unknown	African American: 1 in 10
Smith-Lemli-Opitz syndrome	Unknown	Caucasian: 1 in 70
Spinal muscular atrophy	Unknown	African American: 1 in 66 Asian: 1 in 53 Ashkenazi Jewish: 1 in 41 Caucasian: 1 in 35 Hispanic: 1 in 117
Tay-Sachs disease	1 in 300	Ashkenazi Jewish: 1 in 30

### DESCRIPTION

Genetic disease is defined as a morbid disorder that is caused by an abnormality in human genetic material. Genetic defects find their most varied expression in disruptions of the intricate chemistry that underlies human structure and metabolism. These manifestations range from such well-known conditions as Down's syndrome and phenylketonuria (PKU) to very rare conditions. Certain ethnic groups are at increased risk for specific genetic disorders (e.g., sickle cell anemia, thalassemia, Tay-Sachs disease, Canavan's disease). Major birth defects are apparent in 2-3% of live births with chromosome abnormalities occurring in about 0.5% of all live births.

Preconception and prenatal genetic counseling entails screening prospective parents on the basis of diagnostic tests and family studies. Testing can be performed by various techniques; such as entire gene sequencing, single-strand conformation analysis, conformation sensitive gel electrophoresis assay for truncated protein, and specific mutation assay. Each technique has its own strengths and weaknesses, depending on the disease/condition being tested. In addition to studying chromosomes

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or genes, genetic testing in a broader sense includes biochemical tests for the presence or absence of key proteins that signal aberrant genes.

Preconception, or carrier, genetic counseling and testing is conducted before conception occurs, through analysis of family and parental history and, if indicated, parental testing. It is intended to estimate the risk of a fetus having a genetic defect to assist with reproductive decision making. Individuals who are identified for increased risk may choose in-vitro fertilization with preimplantation genetic testing, the use of donor gametes, adoption, or may pursue no action at all.

Preconception or prenatal genetic screening can be targeted risk-based carrier screening, or through non-targeted panel tests. Targeted risk-based carrier screening identifies single genes for individuals or their partners who are known to be at a higher risk of having or carrying specific X-linked or autosomal recessive disease, and therefore, are at an increased risk of having offspring with that condition. Non-targeted panel tests are done regardless of an individual's risk and can include any number of genes.

Prenatal genetic counseling and testing is conducted after conception with the intent of identifying parental or fetal genetic defects through analysis of family and parental history and if indicated, parental or fetal testing (e.g., amniocentesis, chorionic villus sampling, fetal ultrasound, maternal multiple marker serum sampling, periumbilical blood sampling/cordocentesis, or placental biopsy).

Preimplantation genetic testing evaluates embryos, as an adjunct to in-vitro fertilization. There are different tests, with their own indications, as well as limitations- preimplantation genetic testing-monogenic (PGT-M), preimplantation genetic testing-structural rearrangements (PGT-SR), and preimplantation genetic testing-aneuploidy (PGT-A). Genetic testing is done via embryo biopsy, prior to transferring the embryo to the uterus and can be conducted at 3 different stages after fertilization, 1) from the oocyte via the use of polar bodies, 2) single cell blastomere from embryos when they are composed of 6 to 8 cells at the cleavage stage, or 3) using a group of cells from the trophectoderm during the blastocyst stage (the most common methodology used today, done from 5 to 6 days after insemination). The trophectoderm portion of the embryo develops into the placenta, and this technique avoids disrupting the inner cell mass which develops into the fetus. A disadvantage is that not all embryos develop to the blastocyst phase invitro and, when they do, there is a short time before embryo transfer needs to take place. This form of biopsy has been combined with a form of cryopreservation known as vitrification, to allow time for test results to be obtained before transfer of the embryo takes place. The biopsied materials can be analyzed utilizing different methods (e.g., fluorescent in situ hybridization, array comparative genome hybridization testing, single nucleotide variant microarrays, quantitative polymerase chain reaction, next generation sequencing).

PGT-M targets specific genetic mutations associated with a known carrier of a single gene-disorder, autosomal dominant, autosomal recessive, or X-linked condition. When either partner is a known carrier of a genetic defect, this testing can deselect embryos that are harboring the defective gene. The most common single-gene defects include cystic fibrosis,  $\beta$ -thalassemia, muscular dystrophy, Huntington disease, hemophilia, and fragile X disease. It can also be used to determine embryos for potential stem cell donor identification, through assessment of human leukocyte antigen and other markers. This test is limited in that it does not test for all single gene disorders at once and is unable

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to detect new disease-causing variants and therefore, chorionic villus sampling (CVS) or amniocentesis should be conducted to confirm results.

PGT-SR can identify chromosomal translocations, inversions, deletions, and insertions. These tests cannot differentiate between an embryo that has a normal karyotype and an embryo that carries a balanced form of the familial chromosome rearrangement. PGT-SR utilizes only a few cells from the trophoctoderm; therefore, CVS or amniocentesis should be conducted to confirm the results.

PGT-A is a broader test that screens for whole chromosome abnormalities in all chromosomes, with the goal of increasing live birth rates and decreasing early pregnancy failure rates, as aneuploidy 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, due to the nondisjunction of chromosomes during maternal meiosis, therefore, the technique has been explored to deselect aneuploid oocytes in older women. PGT-A tests for all 22 pairs of autosomes as well as the sex chromosomes.

### SUPPORTIVE LITERATURE

The genotypes to be detected by a genetic test must be shown by scientifically valid methods to be associated with a high positive predictive value of the occurrence of a disease. Analytical sensitivity and specificity of a genetic test must be of such a level that the test results can and will be used in making treatment decisions. Information on the risks and benefits of genetic testing must be presented fully and objectively without coercion to persons contemplating genetic testing.

Because fetal gene mutations can be either inherited from a parent or acquired by exposure to environmental stresses such as radiation or toxins, in utero testing of an at-risk fetus offers partners an additional opportunity to make informed choices regarding reproductive options.

Targeting at-risk populations has historically been done through identifying ethnic groups that may be isolated or have cultural norms and customs that make them more likely to reproduce with someone of the same ancestry, placing them at a higher risk for transmitting a genetic disorder to their offspring. Marketing and public awareness campaigns by laboratories and advocacy organizations are promoting non-targeted panel screening, arguing that individuals may self-identify with a specific race/ethnicity that is far different from their ancestry defined by genetics, and therefore, would make them unaware of the risk of their fetus being affected by a genetic condition.

Recent research on non-targeted screening panels highlights their growing role in reproductive care. Studies show that non-targeted screening panels identify a high proportion of carriers and detects at-risk couples who might otherwise be missed, influencing clinical decisions. Research also underscores its potential to improve mental and infant health outcomes by enabling early intervention. With adequate genetic counseling, carrier screening panels can inform reproductive choices, and observational studies have shown that a majority of couples would consider intervention that depends on the severity of the condition. Therefore, non-targeted carrier screening panels for severe recessive and X-linked genetic disorders can have a significant clinical impact. The evidence is sufficient to determine that the technology results in an improvement in the net health outcome.

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The available literature for preimplantation genetic testing for those undergoing IVF includes observational studies and systematic reviews. Data suggests 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.

The available literature for preimplantation genetic screening of individuals that have no identified elevated risk of a genetic disorder undergoing IVF includes randomized controlled trials and meta-analyses. Studies using the FISH screening method have found lower or similar ongoing pregnancy and live birth rates compared with IVF without preimplantation genetic screening. Findings on newer methods of screening are mixed. There are few studies that directly compare laboratory tests for evaluating aneuploidy. Meta-analyses have found higher implantation rates than with standard care, but most studies utilized patients with good prognosis, and the improvements in other outcomes such as cost effectiveness, the role and effect of cryopreservation, time to pregnancy and utility for specific subgroups are inconsistent. Well-designed RCTs evaluating preimplantation genetic screening in various target populations (e.g., women of advanced maternal age, prior implantation failure, women with recurring pregnancy loss) are needed before conclusions can be drawn as to whether the technology improves the net health outcome.

### PROFESSIONAL GUIDELINE(S)

The American College of Obstetricians and Gynecologists (ACOG) Practice Bulletin No. 88 (2007), Invasive Prenatal Testing for Aneuploidy (Down Syndrome), issued Level A recommendations. Among the recommendations was that women found to have increased risk of aneuploidy with first-trimester screening should be offered genetic counseling and the option of CVS or second-trimester amniocentesis.

In 2001, the American College of Obstetricians and Gynecologists and the American College of Medical Genetics introduced guidelines for prenatal and preconception carrier screening for cystic fibrosis (CF). The guidelines recommended that screening for CF be performed as part of routine obstetric practice for all patients. Given that CF screening has been a routine part of reproductive care for women since 2001, it is prudent to determine if the patient has been previously screened, before ordering CF screening that may be redundant. If a patient has been screened previously, CF screening results should be documented but the test should not be repeated.

In July 2014, reaffirmed 2025, ACOG published committee opinion number 605 which states, "If a woman has a personal or family history of ovarian failure or an elevated follicle-stimulating hormone (FSH) level before age 40 years without a known cause, fragile X premutation carrier testing should be offered." The guideline also states, "if gonadotropins are elevated into the menopausal range (typically, basal FSH levels will be greater than 30-40mIU/ml, depending on the laboratory used), a repeat FSH measurement is indicated in one month. If the result indicates that FSH is elevated, a diagnosis of primary ovarian insufficiency can be established."

The American Society of Reproductive Medicine (ASRM), European Society of Human Reproduction and Embryology (ESHRE) and Internal Menopause Society (IMS) developed a guideline on premature ovarian insufficiency (2025) that defined POI as disordered menstrual cycles (spontaneous amenorrhea or irregular menstrual cycles) for at least four months, and an elevated FSH of greater

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than 25 IU/l. FSH assessment should be repeated after four to six weeks if there is diagnostic uncertainty. FSH testing for the diagnosis of POI does not have to be timed to a specific day of the menstrual cycle.

In March 2017, the ACOG Committee on Genetics (Committee Opinion 691) recommended that information about genetic carrier screening should be provided to every pregnant woman. Carrier screening and counseling ideally should be performed before pregnancy. The committee noted that it is important to obtain the family history of the patient and, if possible, her partner as a screening tool for inherited risk. Carrier screening for a particular condition generally should be performed only once in a person's lifetime and the results should be documented in the patient's health record. The recommendations also indicated that screening for spinal muscular atrophy should be offered to all women who are considering pregnancy or are currently pregnant, as well as fragile X permutation carrier screening for women with a family history of fragile X-related disorders or intellectual disability suggestive of fragile X syndrome.

ACOG Committee on Genetics also released Committee Opinion 690 in March 2017 which states that ethnic-specific, pan ethnic, and expanded carrier screening (ECS) are acceptable strategies for pre-pregnancy and prenatal carrier screening. Given the multitude of conditions that can be included in expanded carrier screening panels, the disorders selected for inclusion should meet several of the following consensus-determined criteria: have a carrier frequency of 1 in 100 or greater, have a well-defined phenotype, have a detrimental effects on quality of life, cause cognitive or physical impairment, require surgical or medical intervention, or have an onset early in life. Additionally, screened conditions should be able to be diagnosed prenatally and may afford opportunities for antenatal intervention to improve prenatal outcomes, changes to delivery management to optimize newborn and infant outcomes, and education of the parents about special care needs after birth.

The ACMG consensus group made the following recommendations:

- Replacing the term "expanded carrier screening" with "carrier screening" as no precise definition for "expanded" exists.
- Establishing a tier-based system of carrier screening, to enhance communication and precision while advancing equity in carrier screening.
- Carrier screening paradigms should be ethnic and population neutral and more inclusive of diverse populations to promote equity and inclusion.
- Offering Tier 3 carrier screening to all pregnant patients and those planning a pregnancy.
- Male partners of pregnant women and those planning a pregnancy may be offered Tier 3 carrier screening for autosomal recessive conditions when carrier screening is performed simultaneously with their female partner.
- Consider offering Tier 4 screening when a pregnancy stems from a known or possible consanguineous relationship (second cousins or closer) or when family or personal medical history warrants.

The ACMG does not recommend:

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- Offering Tier 1 and/or Tier 2 screening, because these do not provide equitable evaluation of all racial/ethnic groups.
- Routine offering of Tier 4 panels.

In 2023, the National Society of Genetic Counselors released a practice guideline (Sagaser 2023) to address expanded carrier screening for reproductive risk assessment. The guideline recommends that ECS be made available for all individuals considering reproduction and all pregnant reproductive pairs, as ECS presents an ethnicity-based carrier screening alternative which does not rely on race-based medicine. The final decision to pursue carrier screening should be directed by shared decision-making, which considers specific features of patients as well as their preferences and values. As a preconceptional reproductive risk assessment tool, ECS is superior compared to ethnicity-based carrier screening in that it both identifies more carriers of autosomal recessive (AR) and X-linked (XL) conditions as well as eliminates a single race-based medical practice. ECS should be offered to all who are currently pregnant, considering pregnancy, or might otherwise biologically contribute to pregnancy.

Certain autosomal recessive disease conditions are more prevalent in individuals of Eastern European Jewish (Ashkenazi) descent. ACOG has recommended that individuals of Eastern European Jewish ancestry be offered carrier screening for Tay–Sachs disease (TSD), Canavan disease, and cystic fibrosis as part of routine obstetric care. They recently recommended additional carrier screening for familial dysautonomia. All of these tests have a high sensitivity in the Jewish population. The prevalence of these disorders in non-Jewish populations, except for TSD and cystic fibrosis, is unknown. The sensitivity of these carrier tests in non-Jewish populations has not been established. The mutations may be different and more diverse. Consequently, when only one partner is Jewish, it is difficult to assess the risk of having an affected offspring. Therefore, carrier screening of the non-Jewish partner is of limited value.

ACOG has also recommended that women with a specific family history of hereditary disorders (e.g., fragile X) or other clinical features (e.g., unexplained mental retardation or developmental delay, autism, or premature ovarian insufficiency) be candidates for genetic counseling carrier screening for that particular disorder or syndrome.

In 2020, ACOG reaffirmed its 2016 recommendation that chromosomal microarray analysis of fetal tissue, such as amniotic fluid, placenta, or products of conception in the evaluation of intrauterine fetal death or still birth when further cytogenetic analysis is desired because of the test's increased likelihood of obtaining results and improved detection of causative abnormalities.

ACOG issued Committee Opinion #799 in 2020 stating that "the clinical utility of [PGT-M] and [PGT-SR] is firmly established; however, the best use of [PGT-A] remains to be determined. Currently, in concordance with ASRM, there is insufficient evidence to recommend the routine use of PGT-A in all infertile women. Future research is necessary to establish the overall clinical utility for [PGT-A], the subset of patients that may benefit from [PGT-A], the clinical significance of mosaicism, and the residual risk for aneuploidy in [PGT-A] screened embryos."

NCCN Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic guidelines (V.2.2026) states "Although the use of prenatal diagnosis or PGT is relatively well established for severe

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hereditary disorders with very high penetrance and/or early onset (e.g., Fanconi anemia), its use in conditions associated with lower penetrance and/or later onset (e.g., hereditary breast or ovarian cancer syndrome) remains somewhat controversial from both an ethical and regulatory standpoint....Successful births have been reported with the use of PGT and IVF in carriers of a BRCA1/2 pathogenic or likely pathogenic variant, but data in the published literature are still very limited. In addition, data pertaining to long-term safety or outcomes of PGT and assisted reproduction in carriers of a pathogenic or likely pathogenic variant are not yet available."

In addition, NCCN Genetic/Familial High-Risk Assessment: Breast, Ovarian and Pancreatic guidelines (V.2.2026) recommends that for individuals of Ashkenazi Jewish descent, complete gene panel analysis including specific AJ founder mutations should be considered based on family history; testing limited to AJ founder testing may be appropriate for families segregating known mutations, or in population screening in which a negative test is followed by more complete testing depending on personal and/or family history.

The American Society of Reproductive Medicine (2012) recommends karyotypic analysis of products of conception which may be useful in the setting of ongoing therapy for recurrent pregnancy loss.

In a 2024 Technical Standard update on laboratory testing for preconception/prenatal carrier screening, ACMG established the criteria for the design and validation of carrier screening tests, defined the scope and limitations of such tests, established the guidelines for interpreting and reporting test results, and recommended appropriate follow-up testing as applicable.

The American Society for Reproductive Medicine (ASRM) released a committee opinion regarding PGT-A use in 2018 stating that, "the value of PGT-A as a universal screening test for all IVF patients has yet to be determined." ASRM notes that existing studies have important limitations regarding the timing of randomization, that the randomized trials were performed in experienced centers, and questions remain regarding the appropriate patient selections and testing platforms. "Large, prospective, well-controlled studies evaluating the combination of multiple approaches 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."

In 2024, ASRM released a follow up to the 2018 committee opinion regarding PGT-A use stating that, "The value of PGT-A as a routine screening test for all patients undergoing in vitro fertilization has not been demonstrated. Although some earlier single-center studies reported higher live-birth rates after PGT-A in favorable-prognosis patients, recent multicenter, randomized control trials in women with available blastocysts concluded that the overall pregnancy outcomes via frozen embryo transfer were similar between PGT-A and conventional in vitro fertilization. The value of PGT-A to lower the risk of clinical miscarriage is also unclear, although these studies have important limitations. Other important considerations about PGT-A that must be addressed by further research include cost-effectiveness, use of mosaic embryos, false-positive results, risk of embryo damage, the role and effect of cryopreservation, time to pregnancy, utility in specific subgroups (such as RPL, prior implantation failure, advanced maternal age, and so on), use of sex selection, and total reproductive potential per intervention. Large, prospective, well-controlled studies evaluating the combination of multiple approaches (genomics, time-lapse imaging, transcriptomics, proteomics, metabolomics, artificial intelligence, and so on) for enhanced embryo selection applicable in a more inclusive patient

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population are needed to determine not only the effectiveness, but also the safety and potential risks of these technologies.”

### REGULATORY STATUS

Clinical laboratories may develop and validate tests in-house and market them as a laboratory service. Laboratory-developed tests must meet the general regulatory standards of the Clinical Laboratory Improvement Amendments (CLIA).

Laboratories that offer laboratory-developed tests must be licensed by CLIA for high-complexity testing. More information is available at: [Clinical Laboratory Improvement Amendments \(CLIA\) | FDA](#) [accessed 2025 Nov 18].

### CODE(S)

- Codes may not be covered under all circumstances.
- Code list may not be all inclusive (AMA and CMS code updates may occur more frequently than policy updates).
- (E/I)=Experimental/Investigational
- (NMN)=Not medically necessary/appropriate

### CPT Codes

Code	Description
59000	Amniocentesis; diagnostic
59012	Cordocentesis (intrauterine), any method
59015	Chorionic villus sampling, any method
76945	Ultrasonic guidance for chorionic villus sampling, imaging supervision and interpretation
76946	Ultrasonic guidance for amniocentesis, imaging supervision and interpretation
81171	AFF2 (ALF transcription elongation factor 2 [FMR2]) (e.g., fragile X intellectual disability 2 [FRAXE]) gene analysis; evaluation to detect abnormal (e.g., expanded) alleles
81172	AFF2 (ALF transcription elongation factor 2 [FMR2]) (e.g., fragile X intellectual disability 2 [FRAXE]) gene analysis; characterization of alleles (e.g., expanded size and methylation status)
81173	AR (androgen receptor) (e.g., spinal and bulbar muscular atrophy, Kennedy disease, X chromosome inactivation) gene analysis; full gene sequence
81174	AR (androgen receptor) (e.g., spinal and bulbar muscular atrophy, Kennedy disease, X chromosome inactivation) gene analysis; known familial variant

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Code	Description
81200	ASPA (aspartoacylase) (e.g., Canavan disease) gene analysis, common variants (e.g., E285A, Y231X)
81204	AR (androgen receptor) (e.g., spinal and bulbar muscular atrophy, Kennedy disease, X chromosome inactivation) gene analysis; characterization of alleles (e.g., expanded size and methylation status)
81228	Cytogenomic (genome-wide) analysis for constitutional chromosomal abnormalities; interrogation of genomic regions for copy number variants, comparative genomic hybridization [CGH] microarray analysis
81229	Cytogenomic (genome-wide) analysis for constitutional chromosomal abnormalities; interrogation of genomic regions for copy number and single nucleotide polymorphism (SNP) variants, comparative genomic hybridization [CGH] microarray analysis
81243	FMR1 (fragile X messenger ribonucleoprotein 1) (e.g., fragile X syndrome, X-linked intellectual disability [XLID]) gene analysis; evaluation to detect abnormal (e.g., expanded) alleles
81244	FMR1 (fragile X messenger ribonucleoprotein 1) (e.g., fragile X syndrome, X-linked intellectual disability [XLID]) gene analysis; characterization of alleles (e.g., expanded size and promoter methylation status)
81255	HEXA (hexosaminidase A [alpha polypeptide]) (e.g., Tay-Sachs disease) gene analysis, common variants (e.g., 1278insTATC, 1421+1G>C, G269S)
81260	IKBKAP (inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase complex-associated protein) (e.g., familial dysautonomia) gene analysis, common variants (e.g., 2507+6T>C, R696P)
81265	Comparative analysis using Short Tandem Repeat (STR) markers; patient and comparative specimen (e.g., pre-transplant recipient and donor germline testing, post-transplant non-hematopoietic recipient germline [e.g., buccal swab or other germline tissue sample] and donor testing, twin zygosity testing, or maternal cell contamination of fetal cells)
81266	Comparative analysis using Short Tandem Repeat (STR) markers; each additional specimen (e.g., additional cord blood donor, additional fetal samples from different cultures, or additional zygosity in multiple birth pregnancies) (List separately in addition to code for primary procedure)
81329	SMN1 (survival of motor neuron 1, telomeric) (e.g., spinal muscular atrophy) gene analysis; dosage/deletion analysis (e.g., carrier testing), includes SMN2 (survival of motor neuron 2, centromeric) analysis, if performed

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Code	Description
81336	SMN1 (survival of motor neuron 1, telomeric) (e.g., spinal muscular atrophy) gene analysis; full gene sequence
81337	SMN1 (survival of motor neuron 1, telomeric) (e.g., spinal muscular atrophy) gene analysis; known familial sequence variant(s)
81349	Cytogenomic (genome-wide) analysis for constitutional chromosomal abnormalities; interrogation of genomic regions for copy number and loss-of-heterozygosity variants, low-pass sequencing analysis
81400	Molecular pathology procedure level 1 (e.g., identification of single germline variant [eg, SNP] by techniques such as restriction enzyme digestion or melt curve analysis)
81401	Molecular pathology procedure level 2 (e.g., 2-10 SNPs, 1 methylated variant, or 1 somatic variant [typically using nonsequencing target variant analysis], or detection of a dynamic mutation disorder/triplet repeat)
81402	Molecular pathology procedure level 3 (e.g., >10 SNPs, 2-10 methylated variants, or 2-10 somatic variants [typically using non-sequencing target variant analysis], immunoglobulin and T-cell receptor gene rearrangements, duplication/deletion variants of 1 exon, loss of heterozygosity [LOH], uniparental disomy [UPD])
81403	Molecular pathology procedure level 4 (e.g., analysis of single exon by DNA sequence analysis, analysis of >10 amplicons using multiplex PCR in 2 or more independent reactions, mutation scanning or duplication/deletion variants of 2-5 exons)
81404	Molecular pathology procedure level 5 (e.g., analysis of 2-5 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 6-10 exons, or characterization of a dynamic mutation disorder/triplet repeat by Southern blot analysis)
81405	Molecular pathology procedure level 6 (e.g., analysis of 6-10 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 11-25 exons, regionally targeted cytogenomic array analysis)
81406	Molecular pathology procedure level 7 (e.g., analysis of 11-25 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 26-50 exons)
81407	Molecular pathology procedure level 8 (e.g., analysis of 26-50 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of >50 exons, sequence analysis of multiple genes on one platform)
81408	Molecular pathology procedure level 9 (e.g., analysis of >50 exons in a single gene by DNA sequence analysis)

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Code	Description
81412	Ashkenazi Jewish associated disorders (e.g., Bloom syndrome, Canavan disease, cystic fibrosis, familial dysautonomia, Fanconi anemia group C, Gaucher disease, Tay-Sachs disease), genomic sequence analysis panel, must include sequencing of at least 9 genes, including ASPA, BLM, CFTR, FANCC, GBA, HEXA, IKBKAP, MCOLN1, and SMPD1
81443	Genetic testing for severe inherited conditions (e.g., cystic fibrosis, Ashkenazi Jewish-associated disorders [e.g., Bloom syndrome, Canavan disease, Fanconi Anemia type C, mucopolidosis type VI, Gaucher disease, Tay-Sachs disease], beta hemoglobinopathies, phenylketonuria, galactosemia), genomic sequence analysis panel, must include sequencing of at least 15 genes (e.g., ACADM, ARSA, ASPA, ATP7B, BCKDHA, BCKDHB, BLM, CFTR, DHCR7, FANCC, G6PC, GAA, GALT, GBA, GBE1, HBB, HEXA, IKBKAP, MCOLN1, PAH)
81479	Unlisted molecular pathology procedure
81510	Fetal congenital abnormalities, biochemical assays of three analytes (AFP, uE3, hCG [any form]), utilizing maternal serum, algorithm reported as a risk score
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
0236U	SMN1 (survival of motor neuron 1, telomeric) and SMN2 (survival of motor neuron 2, centromeric) (e.g., spinal muscular atrophy) full gene analysis, including small sequence changes in exonic and intronic regions, duplications, deletions, and mobile element insertions (Genomic Unity SMN1/2 Analysis, Variantyx Inc)
0252U	Fetal aneuploidy short tandem-repeat comparative analysis, fetal DNA from products of conception, reported as normal (euploidy), monosomy, trisomy, or partial deletion/duplication, mosaicism, and segmental aneuploidy (e.g., POC, Igenomix USA)
0254U	Reproductive medicine (preimplantation genetic assessment), analysis of 24 chromosomes using embryonic DNA genomic sequence analysis for aneuploidy, and a mitochondrial DNA score in euploid embryos, results reported as normal (euploidy), monosomy, trisomy, or partial deletion/duplication, mosaicism, and segmental aneuploidy, per embryo tested (e.g., SMART PGT-A, Igenomix USA)
0341U	Fetal aneuploidy DNA sequencing comparative analysis, fetal DNA from products of conception, reported as normal (euploidy), monosomy, trisomy, or partial deletion/duplication, mosaicism, and segmental aneuploid

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Code	Description
0400U (NMN)	Obstetrics (expanded carrier screening), 145 genes by nextgeneration sequencing, fragment analysis and multiplex ligation dependent probe amplification, DNA, reported as carrier positive or negative (Genesys Carrier Panel, Genesys Diagnostics)
0552U	Reproductive medicine (preimplantation genetic assessment), analysis for known genetic disorders from trophectoderm biopsy, linkage analysis of disease-causing locus, and when possible, targeted mutation analysis for known familial variant, reported as low-risk or high-risk for familial genetic disorder (PGT-M, Igenomix®, Part of Vitrolife Group™, Igenomix®) (effective 07/01/25)
0553U	Reproductive medicine (preimplantation genetic assessment), analysis of 24 chromosomes using DNA genomic sequence analysis from embryonic trophectoderm for structural rearrangements, aneuploidy, and a mitochondrial DNA score, results reported as normal/balanced (euploidy/balanced), unbalanced structural rearrangement, monosomy, trisomy, segmental aneuploidy, or mosaic, per embryo tested (Smart PGT-A Plus, Igenomix®, Part of Vitrolife Group™, Thermo Fisher Scientific) (effective 07/01/25)
0554U	Reproductive medicine (preimplantation genetic assessment), analysis of 24 chromosomes using DNA genomic sequence analysis from trophectoderm biopsy for aneuploidy, ploidy, a mitochondrial DNA score, and embryo quality control, results reported as normal (euploidy), monosomy, trisomy, segmental aneuploidy, triploid, haploid, or mosaic, with quality control results reported as contamination detected or inconsistent cohort when applicable, per embryo tested (Smart PGT-SR, Igenomix®, Part of Vitrolife Group™, Thermo Fisher Scientific) (effective 07/01/25)
0555U	Reproductive medicine (preimplantation genetic assessment), analysis of 24 chromosomes using DNA genomic sequence analysis from embryonic trophectoderm for structural rearrangements, aneuploidy, ploidy, a mitochondrial DNA score, and embryo quality control, results reported as normal/balanced (euploidy/balanced), unbalanced structural rearrangement, monosomy, trisomy, segmental aneuploidy, triploid, haploid, or mosaic, with quality control results reported as contamination detected or inconsistent cohort when applicable, per embryo tested (Smart PGT-SR Plus, Igenomix®, Part of Vitrolife Group™, Thermo Fisher Scientific) (effective 07/01/25)
<b>The following genetic testing procedures are not specific to prenatal genetic testing:</b>	
88235	Tissue culture for non-neoplastic disorders; amniotic fluid or chorionic villus cells
88240-88291	Cytogenetic studies (code range)

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### HCPCS Codes

Code	Description
S0265	Genetic counseling under physician supervision, each 15 minutes

### ICD10 Codes

Code	Description
O09.511- O09.529	Supervision of elderly primigravida and multigravida (code range)
O09.611- O09.629	Supervision of young primigravida and multigravida (code range)
O09.70- O09.73	Supervision of high-risk pregnancy due to social problems (code range)
O09.811- O09.899	Supervision of other high-risk pregnancies (code range)
O26.20- O26.23	Pregnancy care for patient with recurrent pregnancy loss (code range)
O35.2xx0- O35.6xx9	Maternal care for (suspected) central nervous system malformation in fetus (code range)
O36.5110- O36.5199	Maternal care for known or suspected placental insufficiency (code range)
O36.5910- O36.5999	Maternal care for known or suspected poor fetal growth (code range)

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### SEARCH TERMS

Not Applicable

### CENTERS FOR MEDICARE AND MEDICAID SERVICES (CMS)

There is currently no National Coverage Determination (NCD) or Local Coverage Determination (LCD) for Preconception and Prenatal Genetic Testing and Counseling and Preimplantation Genetic Diagnosis.

However, please refer to the Medicare Managed Care Manual/Chapter 4: Benefits and Beneficiary Protections (Rev.121, Issued: 04-22-16)/Section 90 National and Local Coverage Determinations/Subsection 90.4.1 MAC with Exclusive Jurisdiction over a Medicare Item or Service:

In some instances, one Medicare A/B MAC processes all of the claims for a particular Medicare-covered item or service for all Medicare beneficiaries around the country. This generally occurs when there is only one provider of a particular item or service (for example, certain pathology and lab tests furnished by independent laboratories). In this situation, MA plans must follow the coverage policy reflected in an LCD issued by the A/B MAC that enrolled the provider and processes all the Medicare claims for that item or service. Available from: <https://www.cms.gov/Regulations-and-Guidance/Guidance/Manuals/Internet-Only-Manuals-IOMs-Items/CMS019326> [accessed 2025 Dec 08].

### PRODUCT DISCLAIMER

- Services are contract dependent; if a product does not cover a service, medical policy criteria do not apply.
- If a commercial product (including an Essential Plan or Child Health Plus product) covers a specific service, medical policy criteria apply to the benefit.
- If a Medicaid product covers a specific service, and there are no New York State Medicaid guidelines (eMedNY) criteria, medical policy criteria apply to the benefit.

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- If a Medicare product (including Medicare HMO-Dual Special Needs Program (DSNP) product) covers a specific service, and there is no national or local Medicare coverage decision for the service, medical policy criteria apply to the benefit.
- If a Medicare HMO-Dual Special Needs Program (DSNP) product DOES NOT cover a specific service, please refer to the Medicaid Product coverage line.

<b>POLICY HISTORY/REVISION</b>	
<b>Committee Approval Dates</b>	
10/18/01, 12/05/01, 01/16/03, 02/19/04, 12/16/04, 11/17/05, 09/21/06, 07/19/07, 06/19/08, 05/28/09, 05/27/10, 05/19/11, 03/15/12, 03/21/13, 06/19/14, 03/19/15, 03/17/16, 04/20/17, 03/15/18, 06/20/19, 06/18/20, 04/15/21, 10/20/22, 11/16/23, 11/21/24, 11/20/25, 01/22/26	
<b>Date</b>	<b>Summary of Changes</b>
01/22/26	<ul style="list-style-type: none"><li>• Off-cycle review; medically necessary criteria added for non-targeted carrier screening. Criteria for preimplantation genetic testing and in-vitro fertilization must be met for embryo biopsy. New policy guidelines on ACOG guidelines for non-targeted carrier screening including a table of 22 conditions.</li></ul>
11/20/25	<ul style="list-style-type: none"><li>• Annual review; new policy criteria for primary ovarian insufficiency (POI) and premature ovarian failure (POF). Reformatting of policy statements for ease of review.</li></ul>
01/01/25	<ul style="list-style-type: none"><li>• Summary of changes tracking implemented.</li></ul>
10/18/01	<ul style="list-style-type: none"><li>• Original effective date</li></ul>