

## Genetic Counseling and Testing

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### Definitions

Carrier testing	<p>Used to identify people who carry one copy of a gene mutation that, when present in two copies, causes a genetic disorder.</p> <p>The testing is offered to individuals who have a family history of a genetic disorder and to people in certain ethnic groups with an increased risk of specific genetic conditions.</p> <p>If both parents are tested, the test can provide information about a couple's risk of having a child with a genetic condition.</p>
Clinical utility	<p>The value of a test, including risks and benefits, for the purposes of confirmation of diagnosis in a symptomatic patient, presymptomatic analysis, treatment, family planning and prenatal testing (when relevant).</p>
Cytogenetic studies	<p>Evaluation of chromosomes, their structure and inheritance as applied to the practice of medical genetics.</p>
Genetic counseling	<p>The process of explaining medical and scientific information about an inherited condition or birth defect to an individual or family.</p> <p>The goal of genetic counseling is for families and individuals to understand the information, participate in decision-making about their medical care, and be able to manage the associated problems in a way that is best for them and their families. Genetic counselors are health care professionals who have completed training in an accredited Masters Degree program and have passed the certification examination administered by the American Board of Genetic Counseling. Genetic Counselors are trained to:</p> <ol style="list-style-type: none"> <li>1. Review family histories and medical records.</li> <li>2. Discuss genetic conditions and how they are inherited.</li> <li>3. Explain inheritance patterns.</li> <li>4. Perform genetic risk assessments.</li> <li>5. Review available testing options.</li> <li>6. Discuss disease management, treatment and surveillance options.</li> <li>7. Explore the impact of genetic disorders on both affected and unaffected family members and assist families and individuals as they adjust to the diagnosis.</li> </ol> <p>Most genetic counselors work in conjunction with a medical geneticist or as part of a department, program, or institution. They play a crucial role in health care delivery, particularly in the areas of prenatal diagnosis and cancer genetics.</p>

Genetic testing (aka gene tests)	<p>Refers to the analysis of human DNA, RNA, genes, chromosomes, gene products, enzymes, or metabolites to detect inheritable and/or acquired alterations that cause, or are likely to cause, a particular disorder or condition. The testing may also provide a diagnosis and/or a probability of a specific disease onset prior to symptom occurrence and it can identify if a person is the carrier of a specific gene that could be passed on to children. While gene tests are promising; limitations include the following:</p> <ol style="list-style-type: none"> <li>1. The knowledge of a mutation does not guarantee disease development, nor can the severity of the disease be predicted in the mutation carrier (e.g., some individuals with cystic fibrosis have mild symptoms while others develop debilitating lung disease and pancreatitis).</li> <li>2. Not all disease-causing mutations can be detected; therefore, while a positive result can be informative, a negative result may be inconclusive.</li> <li>3. Many diseases are the result of an interaction between genes and environment, and the way these interactions cause disease is not clearly understood.</li> </ol>
First-degree relative	An individual's parents, siblings, and children.
Second-degree relative	An individual's grandparents, aunts, uncles, half-siblings, nieces, nephews, and grandchildren.
Mutation	A change from the normal gene sequence of which underlies a detrimental clinical presentation associated with disease.
Prenatal testing	<p>Detects changes in a fetus's genes or chromosomes before birth.</p> <p>The testing is offered during pregnancy if there is an increased risk that the baby will have a genetic or chromosomal disorder.</p>
Predictive and presymptomatic types of testing	<p>Detects gene mutations associated with disorders that appear after birth, often later in life. These tests can be helpful to people who have a family member with a genetic disorder, but who have no features of the disorder themselves at the time of testing.</p> <ol style="list-style-type: none"> <li>1. Predictive testing can identify mutations that increase a person's risk of developing disorders with a genetic basis (e.g., certain types of cancer).</li> <li>2. Presymptomatic testing can determine whether a person will develop a genetic disorder before signs or symptoms appear.</li> </ol> <p>The results of predictive and presymptomatic testing can provide information about a person's risk of developing a specific disorder and help with making decisions about medical care.</p>

## Related Medical Guidelines

[AlloMap® Molecular Expression Testing for Post-Heart-Transplant Rejection](#)

[Analysis of KRAS Status](#)

[BCR-ABL1 Genetic Mutation Testing in Chronic Myelogenous Leukemia and Acute Lymphoblastic Leukemia](#)

[BRAF Mutation Analysis](#)

[BRCA 1 and 2 Genetic Testing \(Sequence Analysis/Rearrangement\)](#)

[Cardiac Ion Channel Genetic Testing](#)

[Carrier Screening for Parents or Prospective Parents](#)

[Chromosomal Microarray Analysis \(CMA\)](#)

[Factor V Leiden Mutation Analysis](#)

[Gene Expression Profiling](#)

[Gene Expression Profiling and Biomarker Testing for Breast Cancer](#)

[Gene Expression Profiling for Multiple Myeloma](#)

[Gene Expression Profiling of Melanomas](#)

[Genetic Analysis of PIK3CA Status in Tumor Cells](#)

[Genetic Testing for Alzheimer's Disease](#)

[Genetic Testing for Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy \(CADASIL\) Syndrome](#)

[Genetic Testing for Colorectal Cancer/Lynch Syndrome](#)

[Genetic Testing for Cystic Fibrosis](#)

[Genetic Testing for Frontotemporal Dementia \(FTD\)](#)

[Genetic Testing for Hereditary Cardiomyopathy](#)

[Genetic Testing for Hereditary Hemochromatosis](#)

[Genetic Testing for Hereditary Pancreatitis](#)

[Genetic Testing for Peripheral Neuropathies](#)

[Genetic Testing for PTEN Hamartoma Tumor Syndrome](#)

[Genetic Testing for RET Proto-Oncogene Germline Mutations](#)

[Genetic Testing for Statin Induced Myopathy](#)

[Genetic Testing for TP53 Mutations \(Li-Fraumeni Syndrome\)](#)

[Noninvasive Prenatal Testing \(NIPT\) for Fetal Aneuploidy](#)

## Guidelines\*

### A. Genetic Counseling

Members are eligible for pre- and post-test genetic counseling by a physician or a licensed, or certified, genetic counselor when recommended for EmblemHealth-covered tests.

1. Counseling unrelated to pregnancy — when provided in conjunction with medically necessary genetic testing.
2. Counseling in connection with pregnancy — when medically necessary for evaluation of any of the following:
  - Couples who are closely related genetically (consanguinity, incest).
  - Familial cancer disorders.
  - Individuals from high-risk ethnic groups for specific genetic disorders (e.g., African, Southeast Asian and Mediterranean descent [hemoglobinopathies, sickle cell disease], Ashkenazi eastern European Jews [Tay-Sachs disease]).
  - Cystic fibrosis
  - Fragile X syndrome
  - Spinal muscular atrophy
  - Infertility cases where either parent has a known chromosomal abnormality.
  - Individuals with primary amenorrhea, azoospermia, abnormal sexual development or failure of secondary sexual characteristic development.
  - Mother of known or presumed carrier of an X-linked recessive disorder.
  - Known carriers of autosomal recessive disorder (one or both parents).
  - Parents of a child born with a genetic disorder, birth defect, inborn error of metabolism or chromosome abnormality.
  - Parents of a child with mental retardation, autism, developmental delays or learning disabilities.
  - Pregnant women whose pregnancy may be at increased risk for complications or birth defects, as evidenced by prenatal ultrasound or an abnormal multiple marker screening test, maternal

serum alpha-fetoprotein (AFP) test, test for sickle cell anemia or tests for other genetic abnormalities.

- Pregnant women  $\geq 35$  years of age at delivery.
- Previous unexplained stillbirth or repeated 1st trimester miscarriages ( $\geq 3$ )

## B. Genetic Testing

Members are eligible for genetic testing when there has been a genetic evaluation (complete history, physical exam, conventional diagnostic studies, pedigree analysis and genetic counseling). In addition, all of the following criteria must be met for the test to be considered medically necessary:

1. Test result must impact medical management of a current pregnancy and/or disease state (e.g. surgery, change in surveillance, chemotherapy, hormonal manipulation, etc.).
2. The member displays clinical features of (the specific mutation in question) or is presymptomatic, but with a clear risk of inheritance (e.g., belongs to a high-risk group based on personal and/or family history of a genetic mutation).
3. The test has proven validity in the medical community for the identification of a specific genetically linked inheritable disease (e.g., the observations must be able to be independently replicated and subject to peer review). (*See Limitations/Exclusions*)
4. Definitive diagnosis not established despite comprehensive workup.

## Limitations/Exclusions

1. Coverage consideration may be given for testing when the following are applicable (the provider is expected to provide substantiating documentation to accompany the request):
  - Absence of medical necessity endorsement from a professional medical organization.
    - i. Endorsement examples — Consensus documents, position papers, clinical practice guidelines, etc.
    - ii. Medical organization examples — American College of Medical Genetics (ACMG), American Congress of Obstetricians and Gynecologists (ACOG), American Society of Clinical Oncology (ASCO), National Comprehensive Cancer Network (NCCN).
  - When clinical validity has not been definitively determined due to the paucity of data (e.g., rare disorder).

*(Note: The ACMG view is that at least two peer-reviewed publications should provide very strong evidence of the involvement of the particular gene in the development of the disease, or in some cases, only a single publication with an extensive data set; however, Data must be sufficient to determine, with some level of confidence, the sensitivity and specificity of the test to be offered)*
2. Genetic testing is not covered for a clinically affected individual for the purposes of family planning (e.g., disease risk assessment of other family members) when the treatment and surveillance of the member will not be affected, or in any other circumstance when it will not directly affect the diagnosis or treatment of the member.
3. Genetic testing is not considered medically necessary when the plan has determined that there is insufficient evidence of therapeutic value (e.g., whole exome/genome sequencing).
4. Home testing (e.g., direct-to-consumer; aka home-testing kits) or self referral testing (e.g., genetic tests ordered by members via telephone or Internet) is not considered medically necessary, as there is no evidence of efficacy efficacy of these tests to impact medical management of a current pregnancy and/or disease state. This includes all FDA approved tests listed at

<https://www.fda.gov/MedicalDevices/ProductsandMedicalProcedures/InVitroDiagnostics/ucm624726.htm>. These tests include large panels of individual tests, some of which may be covered for other conditions described in this policy. The finding of a positive value on direct to consumer test does not create a medical necessity beyond the presence of known risk factors.

**Note: ACMG cautions that risks of direct-to-consumer tests include misinterpretation of results, testing that is inaccurate or not clinically valid, lack of follow-up care, misinformation, and other adverse consequences**

## Revision History

10/13/2017 — removed Fragile X restrictions

## Procedure Codes

96040	Medical genetics and genetic counseling services, each 30_ minutes face-to-face with patient/family
S0265	Genetic counseling, under physician supervision, each 15 minutes

## References

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U.S. Food & Drug Administration. Direct-to-Consumer Tests. Nov. 2018.

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\* Genetic information obtained during the course of utilization and care management activities is considered protected health information (PHI). Genetic information may be used to determine medical appropriateness of services and will not be used for determination of coverage, benefit or other underwriting practices.