

**CLINICAL MEDICAL POLICY**

<table>
<thead>
<tr>
<th>Policy Name:</th>
<th>Testing for Genetic Disease</th>
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<td>Policy Number:</td>
<td>MP-010-MD-DE</td>
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<td>Approved By:</td>
<td>Medical Management</td>
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<td>Provider Notice Date:</td>
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<td>Products:</td>
<td>Highmark Health Options</td>
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<td>Application:</td>
<td>All participating hospitals and providers</td>
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**Disclaimer**

*Highmark Health Options medical payment and prior-authorization policy is intended to serve only as a general reference resource regarding payment and coverage for the services described. This policy does not constitute medical advice and is not intended to govern or otherwise influence medical decisions.*

**POLICY STATEMENT**

Highmark Health Options provides coverage for medically necessary genetic testing under the laboratory services of the medical surgical benefit of the Company’s Medicaid products to establish a molecular diagnosis of an inheritable disease. This policy is not inclusive of all known genetic tests.

This policy is designed to address medical guidelines that are appropriate for the majority of individuals with a particular disease, illness, or condition. Each person's unique clinical circumstances warrants individual consideration, based on review of applicable medical records.

The qualifications of the policy will meet the standards of the National Committee for Quality Assurance (NCQA) and the Delaware Department of Health and Social Services (DHSS) and all applicable state and federal regulations.

**DEFINITIONS**

**Biochemical Genetic Test** – Diverse spectrum of laboratory analysis of biomolecules (metabolites, enzyme activities and functional assays) in serum or tissue to detect inborn errors of metabolism,
genotype or mutations for clinical purposes (e.g. predict risk of disease, identify carriers and establish prenatal or clinical diagnoses or prognosis).

**Genetic testing** – Genetic testing requires the analysis of human chromosomes, DNA (deoxyribonucleic acid), RNA (ribonucleic acid), genes or gene products in order to detect or predict risk of inherited or non-inherited genetic variants related to disease, identify carriers, establish prenatal and clinical diagnosis or prognosis.

**Carrier Testing** – Carrier testing is used to determine whether they possess one copy of a gene mutation that, when present in two copies, causes a genetic disorder. This type of 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.

**Genetic Counseling** – The process in which a specially trained professional evaluates family history, medical records, and genetic test results, in the risk assessment of an individual for genetic disease, understanding the limitations and risks of genetic testing.

**Genetic Screening** – Genetic screening issued to identify individuals who do not currently exhibit signs or symptoms but may have an increased risk of developing or transmitting a specific genetic disorder.

**Genetic Screening Panels** – These screening panels are genetic tests that are performed for multiple conditions such as the Ashkenazi Jewish Panel.

**Diagnostic/Confirmatory Testing in Symptomatic Individuals** – Genetic testing that is performed to rule out, identify, or confirm a suspected genetic disorder in an affected individual.

**Direct Risk** – This is when there is documentation in the family history of a disorder that involves an autosomal dominant inheritance which has been demonstrated in either the mother or the father or evidence of a disorder inherited in an autosomal recessive or X-linked recessive manner with supporting documentation suggestive of family history of a suspected disorder.

**Family** –
- First degree relatives are defined as the parents, brothers, sisters, or children of an individual member.
- Second degree relatives are those people with whom one quarter of the member’s genes is shared (e.g., grandparent, grand child, uncle, aunt, nephew, niece or half-sibling).
- Third degree relatives are those people with whom one eighth of a member’s genes is shared (e.g., cousin, great grandparent, great aunt, or great uncle).

**Predictive Testing** – Predictive testing is used to determine whether individuals who have a family history of a disease but no current symptoms have the gene alteration associated with the disease. Predictive genetic testing includes pre-symptomatic testing and predispositional testing.
PROCEDURES

1) When Highmark Health Options does not have a specific medical guideline for genetic testing, the following medical necessity criteria must be met:
   a) A complete history, physical examination, family history and pedigree analysis, laboratory, imaging and other diagnostic testing, and a specific medical differential diagnosis has been established; AND
   b) The results of the genetic testing will have a direct impact on the plan member’s care/treatment plan, including the determination of the intensity of surveillance or initiate new course of treatment of that disease or altering an existing therapy; AND
   c) The member is at direct risk of inheriting the genetic mutation (pre-symptomatic) as determined on review of family history and risk factors-(carrier identification); AND
   d) The genetic disorder is associated with the potential for significant disability or has a lethal natural history; OR
   e) The member displays clinical features as documented in the physical exam and conventional testing are inconclusive and a definitive diagnosis is uncertain (diagnostic); AND
   f) The member has not had like or similar genetic testing previously. This does not apply to requests for comprehensive genetic testing when targeted testing has been previously performed; AND
   g) The providing laboratory is approved by the FDA and/or other professional or governmental agencies; AND
   h) The specific mutation or set of mutations has been established in the scientific literature as a reliable test associated with the disease; AND
   i) Peer reviewed literature is available that provides evidence for the indications and performance of the testing this policy is to be used in situations in which there is an absence of a medical policy.

Note: When available, please reference the separate Highmark Health Options medical policies for specific genetic tests.

2) When services are not covered
   a) For conditions other than those listed above scientific evidence has not been established.
   b) Generally, genetic testing for a particular disease should be performed once per lifetime; however, there are rare circumstances in which testing may be performed more than once in a lifetime (e.g., previous testing methodology is inaccurate or a new discovery has added significant relevant mutations for a disease).
   c) Direct-to-consumer testing including but not limited to ‘in-home’ test kits or genetic tests order by plan member over the phone or internet.
   d) Genetic testing of children to predict adult onset diseases is considered not medically necessary unless test results will guide current decisions concerning prevention which would be lost by waiting until the member has reached adulthood.
   e) Genetic testing or gene mapping in the screening of the general population.
3) Post-payment Audit Statement  
The medical record must include documentation that reflects the medical necessity criteria and is subject to audit by Highmark Health Options at any time pursuant to the terms of your provider agreement.

4) Place of Service-Inpatient or Outpatient  
The place of service for all medically necessary genetic testing is outpatient.

5) Genetic Counseling  
Pre- and post-test genetic counseling is required to be performed by an independent (not employed by a genetic testing lab) genetic provider prior to genetic counseling for mutations. This service is necessary in order to inform persons being tested about the benefits and limitations of a specific genetic test for the specific patient. Genetic testing for mutation requires documentation of medical necessity from one of the following providers who has evaluated the member and intends to see the person after testing has been performed for counseling:

- Board Eligible or Board Certified Genetic Counselor
- Advanced Genetics Nurse
- Genetic Clinical Nurse
- Advanced Practice Nurse in Genetics
- Board Eligible or Board Certified Clinical Geneticist
- A physician with experience in cancer genetics
- A physician specializing in the care for the indication(s) for genetic testing

6) Governing Bodies Approval  
FDA  
a) The FDA has only regulated a relatively small number of genetic tests sold to laboratories as kits. In 2010, the FDA announced plans to expand regulation to all genetic tests, this expansion has yet to take place (as of April 19, 2016).

b) The majority of genetic testing are laboratory developed tests that do not require premarket approval by the FDA. These types of tests are regulated under the Clinical Laboratory Improvement Amendments (CLIA) Act of 1998. The regulations of the CLIA Amendments do not include validation of specific test but rather there is procedural compliance.

c) Additional information available at:  
http://www.fda.gov/MedicalDevices/DeviceRegulationandGuidance/Overview/default.htm

CODING REQUIREMENTS

As outlined in the most current version of the CPT manual.

REIMBURSEMENT

Participating facilities will be reimbursed per their Highmark Health Options contract.
POLICY SOURCE(S)


