Genetic and Genomic Outpatient Testing Guideline (0112-GL-DEPT-0022-HS)

All (one exception below) genetic testing requires prior authorization (PA)*.

EXCEPTION: Prenatal screening for Cystic Fibrosis does not require a PA.

Background

Genetics refers to the study of genes and their role in inheritance – the way certain traits or conditions are passed down from one generation to another. Genetics involves scientific studies of the inheritance of variable characteristics. Genes (units of heredity) carry the instructions for making proteins, which direct the activities of cells and functions of the body.

Genes influence traits such as hair and eye color as well as health and disease development. Genetics determines much (but not all) of a person’s health status; environmental differences also play a part. A genome is defined as all the genetic material in the chromosomes of a particular organism. Genomics is a relatively new term describing the study of multiple genes from the same person, including interactions of those genes with each other and the person’s environment. Genomics involves the scientific study of complex diseases such as heart disease, asthma, diabetes and cancer. Genomics is offering new possibilities for therapies and treatment of some diseases, as well as new diagnostic methods. The major tools and methods related to genomics studies are bioinformatics, genetic analysis, measurement of gene expression, and determination of gene function.

Genetic tests are most often used to identify mutations in single genes which cause rare disorders. A genetic test should be able to identify a person with the disorder or at risk for a disorder with high sensitivity and specificity. In order to evaluate the efficacy of the genetic tests, certain criteria should be met.

1. A test must have moderate to high clinical utility in order to be considered an added value to clinical patient management. In other words, how useful is the result of a genetic test in regards to care management for the patient. The test should have a clear result that will impact clinical outcomes and management. There should be well designed and conducted studies supporting clinical utility.

2. A test must have moderate to high clinical validity in order to be considered an added value to clinical patient management. In other words, the result of the genetic test accurately identifies or predicts a patient’s clinical status. The genetic variation being tested for correlates with the clinical status. Because many tests are performed by single proprietary labs, this is difficult to monitor. Often the studies are biased, limited in number and not performed by outside entities.

Genetic testing should be ordered by a physician, pediatrician, and/or certified genetic counselor qualified to interpret the testing results. Appropriate documentation of patient consent should be obtained. Consultations with qualified genetic counselors and physicians should be part of the treatment plan in order for the patient to receive the appropriate consent and interpretation of the genetic testing.

Types of Genetic Tests:

- **Diagnostic testing:** Used to identify or rule out a specific genetic or chromosomal condition. In many cases, genetic testing is used to confirm a diagnosis when a particular condition is suspected based on physical signs and symptoms. Diagnostic testing can be performed before birth or at any time during a person’s life, but is not available for all genes or all genetic conditions. The results of a diagnostic test can influence a person’s choices about health care and the management of the disorder.
• **Predictive and presymptomatic testing:** Used to detect 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. The results of the testing can provide information about a person’s risk of developing a specific disorder and help with making decisions about medical care.

• **Carrier testing:** Used to identify people who carry 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. If both parents are tested, the test can provide information about a couple’s risk of having a child with a genetic condition. More recently, some companies are offering a panethnic carrier test for common mutations in dozens of disorders.

  “The optimal time for determination of risk, clarification of carrier status and discussion of the availability of prenatal testing is before pregnancy.”

• **Prenatal testing:** Used to detect changes in a fetus’ genes or chromosomes before birth. This type of testing is offered during pregnancy if there is an increased risk that the baby will have a genetic or chromosomal disorder. In some cases, prenatal testing can lessen a couple’s uncertainty or help them make decisions about a pregnancy. It cannot identify all possible inherited disorders and birth defects.

• **Preimplantation testing:** A specialized technique that can virtually eliminate the risk of having a child with a particular genetic or chromosomal disorder. It is used to detect genetic changes in embryos created using assisted reproductive techniques such as in-vitro fertilization. Only embryos without certain genetic changes are implanted in the uterus to initiate a pregnancy.

• **Newborn screening:** Used just after birth to identify genetic disorders that can be treated early in life.

• **Pharmacogenomic testing:** Used to detect genetic variations which influence a person’s response to drugs. Various gene changes correlate with a medication’s efficacy or toxicity. These tests may be useful in preventing adverse medication reactions or identifying persons which may benefit from a specific drug.

• **Cologuard:** Stool DNA sample testing used to screen for colorectal cancer.

**Guideline Criteria**

I. **Diagnostic Testing:** (Member is symptomatic)

1. Physical exam and routine testing have been performed and a diagnosis has not been established.
2. The member is at risk for a genetic disorder based on family history or physical features.
3. The provider must be able to support that the member will have improved clinical outcomes based on the diagnosis and subsequent intervention or treatment.
4. The treatment would need to be supported as different than the current or expected treatment plan based on the clinical outcome.
5. The availability of information must be supported as useful for decision making for the treatment of the member.
6. This would end a diagnostic odyssey for the development of a treatment plan.

II. **Predictive and Presymptomatic testing:** (Member is asymptomatic, non-carrier status)
Member is asymptomatic, has susceptibility to disease or requires a specific disease prognosis (i.e., cystic fibrosis, breast cancer, hemochromatosis, familial dysautotonia, Huntington’s Chorea, celiac disease).

_Only Genetic Counselors or Geneticists may order testing for the presence of hereditary predictive and/or presymptomatic conditions._

1. Member _has risk assessment/susceptibility to a disease_ based on an association to a marker, family history or other indicator. There needs to be a consideration of the number of people who manifest this mutation.
2. Member _requires a specific disease diagnosis_ in which the prognosis or specific benchmarks of the disorder are established.
   a) The provider must support the early identification for a screen positive individual to identify a disorder for which there is an intervention or treatment supporting the plan.
   b) The provision of the information is useful for clinical decision making which creates a significant improved health outcome for the member.
   c) The prevention or early detection strategies must lead to improved health outcomes.
   d) The provider must support that the result will support improved health outcomes citing expected specific changes.
   e) The provider must describe how the test results will assist in a change in patient management.

III. _Carrier Testing in relation to Reproductive Planning:_

A. Carrier testing is optimally performed prior to conception.

B. Family History based carrier testing:

   Genetic testing of the parents or prospective parents to determine carrier status of inherited disorders is considered medically necessary when one of the following criteria is met:

   1. An affected child is identified with either an autosomal recessive disorder, an x-linked disorder, or an inherited disorder with incomplete penetrance and/or variable expression and genetic testing is performed to determine the pattern of inheritance and to guide subsequent reproductive decisions; OR
   2. One or both parents or prospective parent(s) have another first or a second degree relative who is affected, or the first degree relative has an affected child, with either an autosomal recessive disorder, an x-linked disorder, or an inherited disorder with incomplete penetrance and/or variable expression and genetic testing is performed to determine the pattern of inheritance and to guide subsequent reproductive decisions; OR
   3. The parent or prospective parent is at high risk for a genetic disorder with a late onset presentation, and genetic testing is performed to determine genetic status and to guide subsequent reproductive decisions.
   4. Testing is accompanied by genetic counseling.

C. Ethnicity-based carrier screening:

   The parents or prospective parents are members of an ethnic group with a high risk of a specific genetic disorder with an autosomal recessive pattern of inheritance with an allele frequency of at least 1:100 in that ethnic group, including but not limited to Tay-Sach's disease, Canavan disease, mucolipidosis IV, Nieman Pick Disease Type A, Fanconi anemia group C, Glycogen storage 1A, Bloom syndrome, or Familial Dysautonomia in the Ashkenazi Jewish population as well as cystic fibrosis in any ethnic group when pregnancy is being planned; AND ALL of the following criteria regarding the genetic condition and test are met:
1. A specific mutation, or set of mutations, has been established in the scientific literature to be reliably associated with the disease, including both a clear association between the genotype and phenotype as well as an allele detection rate of at least 60%.

2. The genetic disorder is associated with a potentially severe disability or has a lethal natural history.

3. A biochemical or other test is identified but the results are indeterminate, or the genetic disorder cannot be identified through biochemical or other testing.

4. Testing is accompanied by genetic counseling.

Panethnic carrier testing:
- Since many of the disorders tested have an allele frequency of >1/100 and some conditions are not severe or lethal, this testing may be considered in certain situations.
- The test panel must contain only conditions where the allele frequency is at least 1 in 100;
- The genetic disorders on the panel are associated with potentially severe disabilities or lethality;
- Testing is accompanied by genetic counseling.

IV. Preimplantation testing:

*Only* Genetic Counselors or Family Practice physicians providing OB services or Obstetrics/Gynecologists or Certified Nurse Midwives (CNM) may order preimplantation genetic testing.

Preimplantation genetic testing is covered for certain genetic diseases if there is an increased risk (known family history) that an offspring will have a genetic or chromosomal disorder (cystic fibrosis, sickle cell anemia). Please refer to the Preimplantation Genetic Diagnosis Guideline.

V. Newborn Screening: Newborn screening for genetic disorders as mandated by state guidelines is covered.

VI. Pharmacogenomic Testing:

Member requires a predictive treatment response based on pharmacogenomic reactions based on specific marker (i.e. Genotype) with metabolic state (i.e. Phenotype).

1. The provider must support the improved health outcomes based on the drug selection and dosage for the specific disease process.
2. The provider must support the drug efficacy.
3. The genetic testing utilized in the field of pharmacogenomics must meet the same standards as those listed for genetic testing (i.e. high clinical validity, clinical utility and analytical validity).

VII. Cologuard: Cologuard for colorectal cancer (CRC) screening is covered following the CT (Virtual) Colonoscopy & Cologuard Guideline.

VIII. General Guidelines:

- Genetic testing for a specific disease is only covered once in a person's lifetime. Coverage will be extended if additional tests are developed that expand the ability to find mutations in patients who have been previously tested, and if the test is considered a proven method.
- Genetic testing of members is excluded from coverage under Physicians Plus' benefit plans if the testing is performed primarily for the medical management of other family members who are not covered under a Physicians Plus' benefit plan. In these circumstances, the family
members who are not covered by Physicians Plus should contact their insurance carrier to ask about coverage of genetic testing.

- In some instances, more specific criteria are required for coverage of a genetic test. Physicians Plus utilizes Hayes Genetic Testing Evaluation Reports. When these reports are available, a Hayes rating of A or B is required for coverage. Hayes ratings of C will be considered based on further review by the Medical Director. For genetic tests associated with cancer, Physicians Plus may use the National Comprehensive Cancer Network guidelines to determine coverage.
- Additional expenses for banking of genetic material (i.e. DNA Banking) are not covered.

*Prior authorization does not guarantee payment. Coverage of services is based on member eligibility and member's benefits per the medical certificate of coverage at the time services are rendered.

References:


Burke, W., MD., PhD., Genetic Testing, Genomic Medicine, New England Journal of Medicine, Volume 347, Number 23, December 5, 2002.


Up-to-Date review. Basic principles of genetic testing. Last literature review June 2013; This topic last updated: April 20, 2012

Up-to-Date review. Genetic counseling and testing. Last literature review June 2013; This topic last updated: June 20,2013


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