EFFECTIVE DATE: 01|01|2017
POLICY LAST UPDATED: 12|19|2017

OVERVIEW

Genetic testing is a technique used to identify people at risk for a specific genetic disease, predict the possibility of future genetic disease, or to determine the risk for transmitting such a disease to their offspring. Testing may also be used as part of the process to identify, confirm, or predict the possibility of a specific medical condition and develop a treatment plan.

This policy indicates genetic testing services that

- require/recommend preauthorization via the Clear Coverage online tool
- not medically necessary
- not covered
- covered

For information regarding Proprietary Laboratory Analyses Codes, please see the Proprietary Laboratory Analyses Policy.

MEDICAL CRITERIA

Generally InterQual criteria is used to determine medical necessity for a majority of genetic testing, and is found in the Clear Coverage online authorization tool:

https://www.bcbsri.com/BCBSRIWeb/Login.do?redirectTo=/providers/preauth/preauthProviderOverview.jsp

The following criteria is used in the online authorization tool when separate criteria is not identified for the test being performed.

Genetic testing is considered medically necessary to establish a molecular diagnosis of an inheritable disease when all of the following are met:

- The member displays clinical features; and
- The genetic disorder is associated with a potentially significant disability or has a lethal natural history; and
- After history, physical examination, pedigree analysis, genetic counseling and completion of conventional diagnostic studies, a definitive diagnosis remains uncertain; and
- The results of the test will be used specifically for diagnosis; and
- The disease is treatable or preventable; and
- The result of the test will directly influence the treatment being delivered to the patient, including increasing the intensity of surveillance/treatment of that disease including family planning; and
- The providing laboratory must be approved by the U.S. Food and Drug Administration (FDA) or other governmental agencies; and
- Peer-reviewed literature is available that provides evidence for the indications and performance of the test or the indication for the test is in accordance with the guidelines of the American College of Medical Genetics.

Genetic screening is considered medically necessary when all of the following are met:

- To determine if the member is at direct risk of inheriting the mutation in question; and
- The genetic disorder is associated with a potentially significant disability or has a lethal natural history; and
- A specific mutation, or set of mutations, has been established in the scientific literature to be reliably associated with the disease; and
- The results of the test will be used specifically for diagnosis or the result of the test will directly influence the treatment being delivered to the patient, including increasing the intensity of surveillance/treatment of that disease or have an impact on family planning; and
- The providing laboratory must be approved by the FDA or other governmental agencies; and
- Peer-reviewed literature is available that provides evidence for the indications and performance of the test or the indication for the test is in accordance with the guidelines of the American College of Medical Genetics.

Carrier testing is considered medically necessary when all of the following are met:

- To determine if the member is at direct risk of transmitting the mutation in question to their offspring; and
- The genetic disorder is associated with a potentially significant disability or has a lethal natural history; and
- A specific mutation, or set of mutations, has been established in the scientific literature to be reliably associated with the disease; and
- The results of the test will have an impact on family planning; and
- The providing laboratory must be approved by the FDA or other governmental agencies; and
- Peer reviewed literature is available that provides evidence for the indications and performance of the test or the indication for the test is in accordance with the guidelines of the American College of Medical Genetics.

PRIOR AUTHORIZATION

Prior authorization is required for BlueCHiP for Medicare and recommended for Commercial Products.

Prior authorization is required for each component of panel testing when it is not a next generation sequencing panel and when the panel is represented by multiple CPT codes. Each individual CPT code must be entered into and processed through the online authorization tool independently.

Requests for genetic testing should be obtained via the BCBSRI online preauthorization tool, which is available only to BCBSRI-participating providers. All other providers need to fax a preauthorization request to Utilization Management at (401) 272-8885.

If a genetic test or family is not found in the online authorization tool, please fax a request to our Utilization Management Dept at (401) 272-8885.

POLICY STATEMENT

BlueCHiP for Medicare and Commercial Products

Genetic testing is considered medically necessary when the criteria in the online authorization tool has been met.

Genetic testing, using panels of genes, including Next Generation Sequencing, is considered not medically necessary when the focus of the testing is on an individual component of the panel and there is an appropriate test available for the individual component. Individual components of a panel may be submitted for review and may be considered medically necessary when criteria is met.

Some genetic testing services for the screening or diagnosis of genetic disorders are considered not medically necessary as the evidence is insufficient to determine the effects of the technology on health outcomes. See Coding Section for details.

Laboratories are not allowed to obtain clinical authorization on behalf of the ordering physician. In no circumstance shall a physician/provider use a representative of a laboratory or anyone with a relationship to a laboratory, to facilitate any portion of the authorization process, including any element of the preparation of necessary documentation of clinical appropriateness. If a laboratory is found to be supporting any portion of the authorization process, BCBSRI will deem the action a violation of this policy and severe action will be taken up to and including termination from the BCBSRI provider network. If a physician/provider provides a laboratory service that has not been authorized, the service will be denied as the financial liability of the laboratory and may not be billed to the member.

COVERAGE

BlueCHiP for Medicare and Commercial Products

Benefits may vary between groups/contracts. Please refer to the Benefit Booklet, Evidence of Coverage, or Subscriber Agreement for applicable genetic testing coverage/benefits.

BACKGROUND

Commercially available genetic tests can guide intervention in symptomatic or asymptomatic people, identify people at risk for future disorders, predict the prognosis of diagnosed diseases, and predict treatment response.

Molecular Pathology

Molecular pathology procedures are medical laboratory procedures involving the analyses of nucleic acid (ie, DNA, RNA) to detect variants in genes that may be indicative of germline (eg, constitutional disorders) or somatic (eg, neoplasia) conditions, or to test for histocompatibility antigens (eg, HLA). Code selection is typically based on the specific gene(s) that is being analyzed. Genes are described using Human Genome Organization (HUGO) approved gene names.

Next Generation Sequencing

Genomic sequencing procedures (GSPs) and other molecular multianalyte assays GSPs are DNA and RNA sequence analysis methods that simultaneously assay multiple genes or genetic regions relevant to a clinical situation. They may target specific combinations of genes or genetic material, or assay the exome or genome. The technology used for genetic sequencing is commonly referred to as next generation sequencing (NGS) or massively parallel sequencing (MPS). GSPs are performed on nucleic acids from germline or neoplastic samples.

MultiAnalyte Assays with Algorithmic Analyses

Multianalyte Assays with Algorithmic Analyses (MAAAs) are procedures that utilize multiple results derived from panels of analyses of various types, including molecular pathology assays, fluorescent in situ hybridization assays, and non-nucleic acid based assays (eg, proteins, polypeptides, lipids, carbohydrates). Algorithmic analysis using the results of these assays as well as other patient information (if used) is then performed and typically reported as a numeric score(s) or as a probability. MAAAs are typically unique to a single clinical laboratory or manufacturer.

A genetic panel is defined as a test that simultaneously evaluates multiple genes, as opposed to sequential testing of individual genes. This includes panels performed by next-generation sequencing (NGS), massive parallel sequencing, and chromosomal microarray analysis (CMA) testing. The definition of a panel will not include panels that report on gene expression profiling, which generally do not directly evaluate genetic mutations.

Genetic panels using next-generation technology or chromosomal microarray analysis are available for many clinical conditions. The major advantage of panels is the ability to analyze many genes simultaneously, potentially improving the breadth and efficiency of genetic workup. A potential disadvantage of panels is that they provide a large of amount of ancillary information whose significance may be uncertain. Limited published evidence reports that the analytic validity of panels approaches that of direct sequencing. The clinical validity and clinical utility of panels are condition-specific. The clinical validity of panels will reflect the clinical validity of the underlying individual mutations. The clinical utility of panels will depend on the context in which they are used, ie, whether the advantages of panel testing outweigh the disadvantages for the specific condition under consideration.

The intended use for these panels is variable, for example, for the diagnosis of hereditary disorders, a clinical diagnosis may be already established, and genetic testing is performed to determine whether this is a hereditary condition, and/or to determine the specific mutation present. In other cases, there is a clinical syndrome (phenotype) with a broad number of potential diagnoses, and genetic testing is used to make a specific diagnosis. For cancer panels, there are also different intended uses. Some panels may be intended to determine whether a known cancer is part of a hereditary cancer syndrome. Other panels may include somatic mutations in a tumor biopsy specimen that may help identify a cancer type or subtype and/or help select best treatment.

There is no standardization to the makeup of genetic panels. Panel composition is variable, and different commercial products for the same condition may test a different set of genes. The makeup of the panels is determined by the specific lab that developed the test. In addition, the composition of any individual panel is likely to change over time, as new mutations are discovered and added to existing panels.

Definitions

Genetic Testing

Genetic testing involves the analysis of chromosomes, DNA, RNA, genes, or gene products to detect inherited (germline) or noninherited (somatic) genetic variants related to disease or health.

Carrier Testing

A carrier of a genetic disorder has 1 abnormal allele for a disorder. When associated with an autosomal recessive or X-linked disorder, carriers of the causative mutation are typically unaffected. When associated with an autosomal dominant disorder, the person has 1 normal and 1 mutated copy of the gene and may be affected with the disorder, may be unaffected but at high risk of developing the disease later in life, or may remain unaffected because of the sex-limited nature of the disease.

Carrier testing may be offered to people: (a) who have family members with a genetic condition; (b) who have family members who are identified carriers; and (c) who are members of ethnic or racial groups known to have a higher carrier rate for a particular condition.

Germline Mutations

Germline mutations are present in the DNA of every cell of the body, from the moment of conception. These include cells in the gonads (testes or ova) and could, therefore, be passed on to offspring.

Somatic Mutations

Somatic variations occur with the passage of time and are restricted to a specific cell or cells derived from it. If these variants are limited to cells that are not in the gonads, they will not be passed on to offspring.

Pharmacogenomics

Pharmacogenomics studies how a person's genetic makeup affects his or her body's response to drugs.

Limitations of Genetic Testing

- The testing methods may not detect all mutations that may occur in a gene

- Genetic testing may identify variants of unknown clinical significance
- Genetic testing may not necessarily determine the clinical outcome
- Different genes can cause the same disease (genetic heterogeneity)
- A mutation in a gene may cause different phenotypes (phenotypic heterogeneity)
- Some disease-causing genes may not yet be identified
- Genetic testing is subject to laboratory error

There are several tests with a lack of demonstrated clinical utility based on extremely limited published data and/or insufficient evidence demonstrating the clinical validity of the test. In these cases, the evidence is insufficient to determine the effect of the technologies on health outcomes and are therefore considered not medically necessary.

CODING

See the attached grid for BlueCHiP for Medicare and Commercial Products coverage of Genetic Testing Codes.

Grid of GT codes Coverage for Policy

RELATED POLICIES

Gene Expression Profiling and Protein Biomarkers for Prostate Cancer Management
Genetic and Protein Biomarkers for the Diagnosis and Cancer Risk Assessment of Prostate Cancer
Genetic Testing for Mental Health Conditions
Human Leukocyte Antigen (HLA) Testing Mandate
In Vitro Chemoresistance and Chemosensitivity Assays
Molecular Markers in Fine Needle Aspirates of the Thyroid
Multimarker Serum Testing Related to Ovarian Cancer
Newborn Metabolic, Endocrine and Hemoglobinopathy, and Newborn Hearing Loss Screening Programs Mandate
PathfinderTG® Molecular Testing
Preventive Services for BlueCHiP for Medicare
Preventive Services for Commercial Members
Proprietary Lab Analyses (PLA) Codes
Proteogenomic Testing for Patients with Cancer

PUBLISHED

Provider Update, February 2018
Provider Update, February 2017
Provider Update, November 2015
Provider Update, September 2013
Provider Update, February 2013
Provider Update, January 2012
Provider Update, August 2011

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