DRAFT Medical Coverage Policy | Genetic Testing Services

EFFECTIVE DATE: 06|01|2026
POLICY LAST REVIEWED: 02|18|2026

OVERVIEW

Genetic tests can perform a host of functions, such as providing a guided intervention in both symptomatic or asymptomatic people, identifying people at risk for future disorders, predicting the prognosis of a diagnosed disease, and predicting the appropriate treatment response.

This policy indicates genetic testing services:

- for which prior authorization is required for Medicare Advantage Plans or recommended for Commercial Products via the online authorization tool, or
- that are not medically necessary, or
- not covered, or
- covered

For information regarding Proprietary Laboratory Analyses Codes (PLA) codes and Multianalyte Assays with Algorithmic Analyses (MAAA) codes, please see the Proprietary Laboratory Analyses (PLA) and Multianalyte Assays with Algorithmic Analyses (MAAA) Policy.

MEDICAL CRITERIA

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

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

NOTE REGARDING PANEL TESTING: Panel tests are subject to additional criteria. Please refer to the Policy Statement and Prior Authorization sections below for specific information regarding panel testing before utilizing the medical necessity criteria set forth below.

The following general criteria is used in the online authorization tool depending on the category of screening, when separate criteria is not identified for the specific test being requested.

Carrier screening (preconception or prenatal testing) for genetic diseases is considered medically when the following criteria are met:

- One or both individuals have a first- or second-degree relative (see definitions below) who is affected;
- One individual is known to be a carrier;
- One or both individuals are members of a population known to have a carrier rate that exceeds a threshold considered appropriate for testing for a particular condition; AND
- Previous carrier screening or individual targeted gene testing for the gene variant(s) of interest has not been performed.

First-degree relatives include a biological parent, brother, sister, or child.
Second-degree relatives include a biologic grandparent, aunt, uncle, niece, nephew, grandchildren, and half-sibling.

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Genetic screening or testing for genetic or hereditary conditions is considered medically necessary when the diagnostic test of the individual's germline will benefit the individual and one of the following criteria is met:

- To confirm a suspected diagnosis in a patient with signs and/or symptoms of the condition
- To identify a causative etiology for a clinical syndrome, for which there are multiple possible underlying conditions
- Testing an asymptomatic individual to determine future risk of disease

Genetic testing for cancer is considered medically necessary when one of the following criteria is met:

- Testing an asymptomatic patient to determine future risk of cancer
- Therapeutic testing of cancer cells from an affected individual to benefit the individual by directing targeted treatment based on specific somatic variants.

PRIOR AUTHORIZATION

For those tests in which prior authorization is indicated in the attached code grid, prior authorization is required for Medicare Advantage Plans and recommended for Commercial Products.

Requests for authorization of genetic testing should be submitted via the BCBSRI online prior authorization tool, which is available to BCBSRI-participating providers. All other providers may fax a prior authorization request to Utilization Management at (401) 272-8885.

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

Panel testing: prior authorization is required for each component and/or gene/gene variant of panel testing 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.

Effective 10/1/2025, for Fully-Funded Commercial Products only, prior authorization requests may not be needed when the requesting physician is a BCBSRI Contracted Primary Care Provider. See below for a list of specialties. Prior authorization continues to be needed for all other Commercial Products, including Self-Funded and Medicare Advantage Plans.

The following specialties, that are credentialed as a primary care provider, are included in this exemption:

- Internal Medicine
- Pediatric Medicine
- Family Practice
- Obstetrics and Gynecology
- Doctor of Osteopathic Medicine
- NP (Nurse Practitioner)/PCP (Primary Care Physician or Provider)
- PA (Physician Assistant)

POLICY STATEMENT

Medicare Advantage Plans and Commercial Products

For services in which prior authorization is indicated, genetic testing may be considered medically necessary when the criteria in the online authorization tool and/or BCBSRI's Policy has been met. Please see Related Policies below for additional policies indicating criteria and coverage requirements for certain genetic testing.

Genetic testing services are not covered for Medicare Advantage Plans and not medically necessary for Commercial Products when:

- there is insufficient clinical evidence or strength of recommendation,
- results would not reasonably be used in management of a patient,
- services are unlikely to impact therapeutic decision-making in the clinical management of the patient.

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There is not enough research to show that genetic panels can lead to better health outcomes for patients. When there is not enough research to show that a gene and/or gene variant alone in a genetic panel test may be useful for guiding patient management and to improve net health outcomes, then the entire genetic panel test is considered not covered for Medicare Advantage Plans and not medically necessary for Commercial Products.

For coverage of any panel test filed with a specific individual CPT code, please refer to the code grid in this policy and/or the Proprietary Laboratory Analyses (PLA) and Multianalyte Assays with Algorithmic Analyses (MAAA) policy.

For some genetic tests, medical necessity, and coverage of the test, is determined by the diagnosis code submitted with the claim. Please refer to the codes on the attached grid and the information in the Comments column for diagnosis coding or for a Related Policy if applicable.

Laboratories are not allowed to obtain clinical authorization or participate in the authorization process on behalf of the ordering physician. Only the ordering physician shall be involved in the authorization, appeal or other administrative processes related to prior authorization/medical necessity.

In no circumstance shall a laboratory or a physician/provider use a representative of a laboratory or anyone with a relationship to a laboratory and/or a third party to obtain authorization on behalf of the ordering physician, to facilitate any portion of the authorization process or any subsequent appeal of a claim where the authorization process was not followed and/or a denial for clinical appropriateness was issued, including any element of the preparation of necessary documentation of clinical appropriateness. If a laboratory or a third party 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 laboratory provides a laboratory service that has not been authorized, the service will be denied as the financial liability of the participating laboratory and may not be billed to the member."

Commercial Products
Some genetic testing services are not covered and a contract exclusion for any self-funded group that has excluded the expanded coverage of biomarker testing related to the state mandate, R.I.G.L**. §27-19-81** described in the Biomarker Testing Mandate policy. For these groups, a list of which genetic testing services are covered with prior authorization, are not medically necessary or are not covered because they are a contract exclusion can be found in the Coding section of the Genetic Testing Services and Proprietary Laboratory Analyses policies. Please refer to the appropriate Benefit Booklet to determine whether the member's plan has customized benefit coverage. Please refer to the list of Related Policies for more information.

COVERAGE

Medicare Advantage Plans and Commercial Products
Benefits may vary between groups/contracts. Please refer to the Benefit Booklet, Evidence of Coverage, or Subscriber Agreement for applicable laboratory testing benefits/coverage and not medically necessary/not covered benefits/coverage.

BACKGROUND

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.

Genomic Sequencing Procedures and Other Molecular Multianalyte Assays

Genomic sequencing procedures (GSPs) and other molecular multianalyte assays are DNA and/or 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 they may assay the exome or genome. The technology typically used for genomic sequencing is massively parallel sequencing (MPS), (eg, next-generation sequencing [NGS]) although other technologies may be employed. 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. The definition of a panel will not include panels that report on gene expression profiling, which generally do not directly evaluate genetic variants.

New genetic technology, such as NGS and chromosomal microarray, has led to the ability to examine many genes simultaneously. This in turn has resulted in a proliferation of genetic panels. Panels using next-generation technology are currently widely available, covering a broad range of conditions related to inherited disorders, cancer, and reproductive testing. These panels are intuitively attractive to use in clinical care because they can analyze multiple genes more quickly and may lead to greater efficiency in the workup of genetic disorders. It is also possible that newer technology can be performed more cheaply than direct sequencing, although this may not be true in all cases.

Newer sequencing techniques were initially associated with higher error rates than direct sequencing. While there are limited published data directly comparing the accuracy of NGS with direct sequencing, several publications have reported that the concordance between NGS and Sanger sequencing is greater than 99% for cancer susceptibility testing, inherited disorders, and hereditary hearing loss. Another potential pitfall is the easy availability of a multitude of genetic information, much of which has uncertain clinical consequences. Variants of uncertain significance are found commonly and in greater numbers with NGS than with direct sequencing.

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 variant 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 variants 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 variants are discovered and added to existing panels.

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 the 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 has reported that the analytic validity of panels approaches that of direct sequencing. The

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clinical validity and clinical utility of panels are condition-specific. The clinical validity of panels will reflect the clinical validity of the underlying individual variants. 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.

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 variant are typically unaffected. When associated with an autosomal dominant disorder, the person has 1 normal copy of the gene and 1 mutated copy of the gene; such a person 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 Variants

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

Somatic Variants

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 variants that may occur in a gene
- Genetic testing may identify variants of uncertain significance
- Genetic testing may not necessarily determine the clinical outcome
- Different genes can cause the same disease (genetic heterogeneity)
- A variant 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 Medicare Advantage Plans and Commercial Products coverage of Genetic Testing Codes and indication of which codes may be covered, medically necessary if criteria are met, not medically necessary or not covered.

Genetic Testing Codes and Coverage Effective 6/1/2026

RELATED POLICIES

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Assays of Genetic Expression in Tumor Tissue as a Technique to Determine Prognosis in Patients with Breast Cancer
Biomarker Testing in Risk Assessment and Management of Cardiovascular Disease
Biomarker Testing Mandate
Bone Turnover Markers for Diagnosis and Management of Osteoporosis and Diseases Associated with High Bone Turnover
CA-125
Carcinoembryonic Antigen (CEA) Testing
Circulating Tumor DNA and Circulating Tumor Cells for Cancer Management (Liquid Biopsy)
Envisia for Idiopathic Pulmonary Fibrosis
Evaluation of Biomarkers for Alzheimer Disease
Fecal Calprotectin Testing
Gene Expression Profile Testing and Circulating Tumor DNA Testing for Predicting Recurrence in Colon Cancer
Gene Expression Profiling and Protein Biomarkers for Prostate Cancer Management
Gene Expression Profiling for Cutaneous Melanoma
Genetic and Protein Biomarkers for the Diagnosis and Cancer Risk Assessment of Prostate Cancer
Genetic Testing for Diagnosis and Management of Mental Health Conditions
Genetic Testing for Epilepsy
Genetic Testing for Inherited Thrombophilia
Genetic Testing for Mitochondrial Disorders
Genomic Sequence Analysis Panels in the Treatment of Hematolymphoid Diseases
Genomic Sequence Analysis Panels in the Treatment of Solid Organ Neoplasms
Homocysteine Testing in the Screening and Diagnosis and Management of Cardiovascular Disease
Human Leukocyte Antigen (HLA) Testing Mandate
Immune Cell Function Assay
In Vitro Chemoresistance and Chemosensitivity Assays
Laboratory Testing Investigational Services
Laboratory Tests Post Transplant and for Heart Failure
Lyme Disease Diagnosis and Treatment Mandate
Medicare Advantage Plans National and Local Coverage Determinations
Minimal Residual Disease Testing for Cancer
Molecular Markers in Fine Needle Aspiration of the Thyroid
Molecular Testing for the Management of Pancreatic Cysts, Barrett Esophagus, and Solid Pancreaticobiliary Lesions (Former Title: PathfinderTG Molecular Testing)
Molecular Testing in the Management of Pulmonary Nodules
Multicancer Early Detection Testing
Multimarker Serum Testing Related to Ovarian Cancer
Multitarget Polymerase Chain Reaction Testing for Diagnosis of Bacterial Vaginosis
Newborn Metabolic, Endocrine and Hemoglobinopathy, and Newborn Hearing Loss Screening Programs Mandate
Noninvasive Techniques for the Evaluation and Monitoring of Patients with Chronic Liver Disease
Nutrient/Nutritional Panel Testing
Preimplantation Genetic Testing
Preventive Services for Medicare Advantage Plans
Preventive Services for Commercial Members
Proprietary Laboratory Analyses (PLA) and Multianalyte Assays with Algorithmic Analyses (MAAA)
Proteogenomic Testing for Patients with Cancer
Proteomic Testing for Targeted Therapy in Non-Small Cell Lung Cancer
Urinary Biomarkers for Cancer Screening, Diagnosis and Surveillance
Vitamin-D Testing
Whole Exome and Whole Genome Sequencing for Diagnosis of Genetic Disorders

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Provider Update, January/February 2026
Provider Update, January/February/May/September/November 2025
Provider Update, March/July/September 2024
Provider Update, February/April/June/November 2023
Provider Update, March/August 2022
Provider Update, October 2021

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