

MEDICAL POLICY

POLICY TITLE	GENERAL APPROACH TO GENETIC TESTING
POLICY NUMBER	MP 2.326

CLINICAL BENEFIT	<input checked="" type="checkbox"/> MINIMIZE SAFETY RISK OR CONCERN. <input checked="" type="checkbox"/> MINIMIZE HARMFUL OR INEFFECTIVE INTERVENTIONS. <input type="checkbox"/> ASSURE APPROPRIATE LEVEL OF CARE. <input type="checkbox"/> ASSURE APPROPRIATE DURATION OF SERVICE FOR INTERVENTIONS. <input checked="" type="checkbox"/> ASSURE THAT RECOMMENDED MEDICAL PREREQUISITES HAVE BEEN MET. <input type="checkbox"/> ASSURE APPROPRIATE SITE OF TREATMENT OR SERVICE.
Effective Date:	11/1/2025

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I. POLICY

This policy applies only if there is not a specific Medical Policy that outlines criteria for testing. If a specific policy does exist, then the criteria for medical necessity in that policy supersede the guidelines in this policy. If there is a disagreement about the medical necessity for a test under a specific policy, alternative tests must meet criteria of any applicable specific policies as well.

Genetic testing may be considered **medically necessary** for a genetic or heritable disorder when the following are met.

For ALL genetic testing, the condition being tested for must have either:

- Reduced life expectancy; **or**
- At least moderate to severe morbidity

In addition, genetic testing classified in one of the categories below may be considered **medically necessary** when all criteria are met for each category, as outlined in the *Medically Necessary Criteria* section below.

- A. Testing affected (symptomatic) individual
 1. Diagnostic
 2. Prognostic
 3. Therapeutic
- B. Testing of DNA from cancer cells of an affected individual to benefit the individual
 1. Diagnostic
 2. Prognostic
 3. Therapeutic (testing to predict treatment response)
- C. Testing an asymptomatic individual to determine future risk of disease
- D. Testing an individual to benefit a family member, no benefit for individual being tested.

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Genetic testing that does not meet the criteria for a specific category is considered **investigational** or **not medically necessary**.

Genetic testing is considered **not medically necessary** when:

- Testing is not considered standard of care, such as when the clinical diagnosis can be made without the use of a genetic test
- Testing is not clinically appropriate for the patient's condition, for example, when it would not change diagnosis and/or management. Other situations where testing is not clinically appropriate include, but are not limited to:
 - Testing is performed entirely for non-medical (e.g., social) reasons
 - Testing is not expected to provide a definitive diagnosis that would obviate the need for further testing.
- Testing is performed primarily for the convenience of the patient, physician, or other health care provider.
- Testing would result in outcomes that are equivalent to outcomes using an alternative strategy, and the genetic test is more costly.

Genetic testing is considered **investigational** when there is insufficient evidence to support a conclusion concerning the health outcomes or benefits associated with this procedure.

“At-home” or “direct-to-consumer” genetic testing is considered **investigational** as there is insufficient evidence to support a conclusion concerning the health outcomes or benefits associated with this procedure.

Medically Necessary Criteria

A. Affected (Symptomatic) Individual

1. Diagnostic Testing for an Affected (Symptomatic) Individual

Diagnostic testing is completed to confirm or exclude genetic or heritable variants in a symptomatic person. This refers to a molecular diagnosis supported by the presence of a known pathologic variant. For the purposes of genetic testing, a symptomatic person is defined as a person with a clinical phenotype that is correlated with a known pathologic variant.

Diagnostic testing of an affected (symptomatic) individual's germline DNA to benefit the individual (excluding reproductive testing) may be considered **medically necessary** when the following are **also** met:

- An association of the marker with the disorder has been established; **and**
- Symptoms of the disease are present; **and**
- A definitive diagnosis cannot be made based on history, physical examination, pedigree analysis, and standard diagnostic studies/tests; **and**
- The clinical utility of identifying the variant has been established as evidenced by the following:
 - Leads to changes in clinical management of the condition that improve outcomes; **or**
 - Eliminates the need for further clinical workup or invasive testing; **or**

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- Leads to discontinuation of interventions that are unnecessary and/or ineffective.

2. Prognostic Testing for an Affected (Symptomatic) Individual

Prognostic testing is completed to determine or refine estimates of disease natural history or recurrence in patients already diagnosed with disease in order to predict natural disease course, (e.g., aggressiveness, recurrence, risk of death). This type of testing may use gene expression of affected tissue to predict the course of disease (e.g., testing breast cancer tissue with Oncotype DX).

Prognostic testing of an affected (symptomatic) individual's germline DNA to benefit the individual (excluding reproductive testing) may be considered **medically necessary** when the following are **also** met:

- An association of the marker with the natural history of the disease has been established; **and**
- Clinical utility of identifying the variant has been established as evidence by the following:
 - Provides incremental prognostic information above that of standard testing; **and**
 - Reclassifies patients into clinically relevant prognostic categories for which there are different treatment strategies; **and**
 - Reclassification leads to changes in management that improve outcomes.

3. Therapeutic Testing for an Affected (Symptomatic) Individual

Therapeutic testing for an affected (symptomatic) individual is completed to determine that a particular therapeutic intervention is effective (or ineffective) for an individual patient. To determine the probability of favorable or adverse response to medications. To detect genetic variants that alter risk of treatment response, adverse events, drug metabolism, drug effectiveness, etc. (e.g., cytochrome p450 testing). To detect genetic variants that adversely affect response to exposures in the environment that are ordinarily tolerated, such as *G6PD* deficiency, genetic disorders of immune function, and aminoacidopathies.

Therapeutic testing for an affected (symptomatic) individual's germline DNA to benefit the individual (excluding reproductive testing) may be considered **medically necessary** when the following are **also** met:

- Genetic testing identifies variants of a phenotype/metabolic state that relate to different pharmacokinetics, drug efficacy, or adverse drug reactions; **and**
- Clinical utility of identifying the variant has been established as evidenced by the following:
 - Leads to initiation of effective medication(s); **or**
 - Leads to discontinuation of medications that are ineffective or harmful OR
 - Leads to clinical meaningful change in dosing of medication that is likely to improve outcomes.

B. Testing of DNA from Cancer Cells of an Affected Individual to Benefit the Individual

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The specified categories of testing listed below for an affected (symptomatic) individual's DNA from cancer cells to benefit the individual may be considered **medically necessary** when the following criteria are **also** met:

1. Diagnostic testing - To determine the origin of a cancer or to determine a clinically relevant subgroup that a cancer falls into:
 - Genetic testing is completed to establish the cell origin of a cancer when the origin is uncertain following standard work-up; **and**
 - Clinical utility of identifying the variant has been established as evidenced by the following:
 - Establishes the necessity to start effective treatment; **or**
 - Establishes the necessity to discontinue ineffective or harmful treatment
2. Prognostic testing - To determine the risk of progression, recurrence, or mortality for a cancer that is already diagnosed.
 - An association of the marker with the natural history of the disease has been established; **and**
 - Clinical utility of identifying the variant has been established as evidenced by the following:
 - Provides incremental prognostic information above that of standard testing; **and**
 - Reclassifies patients into clinically relevant prognostic categories for which there are different treatment strategies; **and**
 - Reclassification leads to changes in management that improve outcomes.
3. Therapeutic (testing to predict treatment response) - To determine the likelihood that a patient will respond to a targeted cancer therapy that is based on the presence or absence of a specific variant.
 - Association between a variant and treatment response to a particular drug has been established; **and**
 - Clinical utility has been established as evidenced by the following:
 - The patient is a candidate for targeted drug therapy associated with a specific variant; **and**
 - There is a clinically meaningful improvement in outcomes when targeted therapy is given for the condition.

C. Testing an Asymptomatic Individual to Determine Future Risk of Disease

This testing is completed to detect genetic variants associated with disorders that appear after birth, usually later in life. The testing is intended for individuals with a family history of a genetic disorder, but who themselves have no features of the disorder at the time of testing, in order to determine their risk for developing the disorder.

Testing an asymptomatic individual to determine future risk of disease may be considered **medically necessary** when the following criteria are **also** met:

- An association of the marker with future disorder has been established; **and**
- Clinical utility has been established as evidenced by the following:
 - There is a presymptomatic phase for this disorder in which interventions/surveillance are available; **and**

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- Interventions in the presymptomatic phase are likely to improve outcomes:
 - Prevent/delay onset of disease; **or**
 - Detect disease at an earlier stage for which treatment is more effective; **or**
 - Discontinuation of interventions that are ineffective or unnecessary.

D. Testing an individual to benefit a family member with no benefit for individual being tested.

For the following category in which the benefit of testing is for another individual, the definition of medical necessity may not apply. When an individual is tested to benefit a family member, and there is no benefit for the individual being tested, eligibility for coverage is dependent on individual plan benefit language. Individual plans may differ as to whether benefit structure allows testing of an individual to benefit an unaffected family member.

Because of these concerns, the following criteria are considered to be criteria for clinical utility of testing and not for medical necessity.

For testing of an affected individual's germline DNA to a benefit family member(s) the following criteria must be met:

- An association of the genetic variant with clinical disease has been established; **and**
- Family members are available who may be at risk for the disorder; **and**
- The individual tested has a clinical diagnosis of the condition (or represents the family member who is most likely to harbor the pathogenic variant), but genetic testing has not been performed; **and**
- There is a presymptomatic phase for the disorder in which interventions are available; **and**
- Interventions in the presymptomatic phase are likely to improve outcomes in one of the following ways:
 - Prevent/delay onset of disease
 - Detect disease at an earlier stage for which treatment is more effective
 - Discontinuation of interventions that are ineffective or unneeded

POLICY GUIDELINES

This policy does not include cytogenetic testing (karyotyping), biochemical testing, or molecular testing for infectious disease.

This policy does not address reproductive genetic testing. See separate policies.

Testing should be cleared or approved by the U.S. Food and Drug Administration as an in vitro diagnostic test (LDT) or performed in a Clinical Laboratory Improvement Amendment compliant or approved laboratory and validated as a laboratory developed test.

Frequency of Testing

In the absence of specific information regarding advances in the knowledge of variant characteristics for a particular disorder, the current literature indicates that genetic tests for inherited disease need only be performed **once per lifetime** of the patient.

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Genetics Nomenclature Update

The Human Genome Variation Society nomenclature is used to report information on variants found in DNA and serves as an international standard in DNA diagnostics. It is being implemented for genetic testing medical evidence review updates starting in 2017 (see Table PG1). The Society's nomenclature is recommended by the Human Variome Project, the Human Genome Organization, and by the Human Genome Variation Society itself.

The American College of Medical Genetics and Genomics and the Association for Molecular Pathology standards and guidelines for interpretation of sequence variants represent expert opinion from both organizations, in addition to the College of American Pathologists. These recommendations primarily apply to genetic tests used in clinical laboratories, including genotyping, single genes, panels, exomes, and genomes. Table PG2 shows the recommended standard terminology—"pathogenic," "likely pathogenic," "uncertain significance," "likely benign," and "benign"—to describe variants identified that cause Mendelian disorders.

Table PG1. Nomenclature to Report on Variants Found in DNA

Previous	Updated	Definition
Mutation	Diseased-Assoc.Variant	Disease-associated change in the DNA sequence.
	Variant	Change in DNA sequence
	Familial Variant	Disease-associated variant identified in a proband for use in subsequent targeted genetic testing in first-degree relatives.

Table PG2. ACMG-AMP Standards and Guidelines for Variant Classification

Variant Classification	Definition
Pathogenic	Disease-causing change in the DNA sequence
Likely Pathogenic	Likely disease-causing change in the DNA sequence
Variant of uncertain significance	Change in DNA sequence with uncertain effects on disease
Likely benign	Likely benign change in the DNA sequence
Benign	Benign change in the DNA sequence

ACMG: American College of Medical Genetics and Genomics; AMP: Association of Molecular Pathology.

Genetic Counseling

Genetic counseling is primarily aimed at patients who are at risk for inherited disorders, and experts recommend formal genetic counseling in most cases when genetic testing for an inherited condition is considered. The interpretation of the results of genetic tests and the understanding of risk factors can be very difficult and complex. Therefore, genetic counseling will assist individuals in understanding the possible benefits and harms of genetic testing, including the possible impact of the information on the individual's family. Genetic counseling may alter the utilization of genetic testing substantially and may reduce inappropriate testing. Genetic counseling should be performed by an individual with experience and expertise in genetic medicine and genetic testing methods.

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Cross-References:

See related policies at the end of this document

II. PRODUCT VARIATIONS

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This policy is only applicable to certain programs and products administered by Capital Blue Cross and subject to benefit variations as discussed in Section VI. Please see additional information below.

FEP PPO: Refer to FEP Medical Policy Manual. The FEP Medical Policy manual can be found at: <https://www.fepblue.org/benefit-plans/medical-policies-and-utilization-management-guidelines/medical-policies>

III. DESCRIPTION/BACKGROUND

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There are numerous commercially available genetic tests, including those used to guide intervention in symptomatic or asymptomatic individuals, to identify individuals at risk for future disorders, to predict the prognosis of diagnosed disease, and to predict treatment response. This policy offers a framework for evaluating the utility of genetic tests, by classifying the types of genetic tests into clinically relevant categories and developing criteria that can be used for evaluating tests in each category.

The purpose of this policy is to provide assistance in evaluating the utility of genetic tests. In providing a framework for evaluating genetic tests, this policy will not attempt to determine the clinical utility of genetic testing for specific disorders. Rather, it provides guidelines that can be applied to a wide range of different tests.

IV. RATIONALE

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SUMMARY OF EVIDENCE

This conceptual framework addresses genetic testing in non-reproductive settings. Genetic testing in reproductive settings is addressed separately. For categories of genetic testing for which the benefit of testing is the individual, criteria for medical necessity apply. When the benefit of testing is not for the individual, but for a family member, medical necessity criteria may not apply, and the criteria are developed for clinical utility.

V. DEFINITIONS

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ANALYTIC VALIDITY of a genetic test defines its ability to accurately and reliably measure the genotype of interest.

ARRAY COMPARATIVE GENOMIC HYBRIDIZATION (chromosomal microarray analysis [CMA], microarray-based comparative genomic hybridization, array CGH, a-CGH, aCGH, or virtual karyotype) is a technique to detect genomic copy number variations at a higher resolution level than chromosome-based comparative genomic hybridization (CGH).

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CARRIER TESTING A carrier of a genetic disorder has one 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 individual has one normal and one 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 the carrier may remain unaffected because of the sex-limited nature of the disease. Carrier testing may be offered to individuals: 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.

CHROMOSOME is one of the threadlike “packages” of genes and other DNA in the nucleus of a cell.

COPY-NUMBER VARIATIONS are alterations of the DNA of a genome that results in the cell having an abnormal number of copies of one or more sections of the DNA. CNVs correspond to relatively large regions of the genome that have been deleted (fewer than the normal number) or amplified (more than the normal number) on certain chromosomes. For example, the chromosome that normally has sections in order as A-B-C-D might instead have sections A-B-C-C-D (a duplication of "C") or A-B-D (a deletion of "C").

CLINICAL VALIDITY of a genetic test defines its ability to detect or predict the associated disorder (phenotype).

DIRECT-TO-CONSUMER GENETIC TESTING refers to genetic tests that are marketed directly to consumers via television, print advertisements, or the Internet. This form of testing, which is also known as at-home genetic testing, provides access to a person’s genetic information without necessarily involving a doctor or insurance company in the process.

DNA a large nucleic acid molecule, found principally in the chromosomes of the nucleus of a cell, that is the carrier of genetic information.

FIRST-DEGREE RELATIVE refers to a parent, sibling, or child.

GENE is the basic unit of heredity, made of DNA, the code for a specific protein.

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

GENETIC TESTING involves the analysis of chromosomes, DNA (deoxyribonucleic acid), RNA (ribonucleic acid), genes, or gene products to detect inherited (germline) or non-inherited (somatic) genetic variants related to disease or health.

GENOTYPE is the specific genetic makeup of an individual, usually in the form of DNA.

IN VITRO DIAGNOSTICS tests on samples such as blood or tissue that can detect diseases or other conditions and can be used to monitor a person’s overall health to help cure, treat, or prevent diseases.

KARYOTYPE is the chromosomal complement of an individual, including the number of chromosomes and any abnormalities

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A LABORATORY DEVELOPED TEST (LDT) is a type of in vitro diagnostic test that is designed, manufactured, and used within a single laboratory.

MICROARRAY is a tool for analyzing gene expression that consists of a small membrane or glass slide containing samples of many genes arranged in a regular pattern. Each spot on an array is associated with a particular gene. Each color in an array represents either healthy (control) or diseased (sample) tissue. Depending on the type of array used, the location and intensity of a color will indicate whether the gene, or variant, is present in either the control and/or sample DNA. It will also provide an estimate of the expression level of the gene(s) in the sample and control DNA.

MITOCHONDRIA are intracellular organelles that are responsible for energy production and cellular respiration.

MITOCHONDRIAL DISEASE refers to one of hundreds of congenital illnesses that result from variants in mitochondrial DNA. As a result, the mitochondria are unable to completely burn food and oxygen in order to generate energy.

VARIANT is a permanent structural alteration in DNA.

PHARMACOGENOMICS The study of how an individual's genetic makeup affects the body's response to drugs.

PHENOTYPE is the physical characteristics of an organism or the presence of a disease that may or may not be genetic.

RNA is a molecule similar to DNA. Unlike DNA, RNA is single-stranded. RNA delivers DNA's genetic message to the cytoplasm of a cell where proteins are made.

SECOND-DEGREE RELATIVE refers to an aunt, uncle, niece, nephew, or grandparent.

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

THIRD-DEGREE RELATIVE refers to a great aunt/uncle, first cousin, or great grandmother/grandfather.

VARIABLE EXPRESSION refers to variation in the manner in which a trait is manifested. When there is variable expressivity, the trait may vary in clinical expression from mild to severe.

VI. DISCLAIMER

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Capital Blue Cross' medical policies are used to determine coverage for specific medical technologies, procedures, equipment, and services. These medical policies do not constitute medical advice and are subject to change as required by law or applicable clinical evidence from independent treatment guidelines. Treating providers are solely responsible for medical advice and treatment of members. These policies are not a guarantee of coverage or payment. Payment of claims is subject to a determination regarding the member's benefit program and eligibility on the date of service, and a determination that the services are medically necessary and appropriate. Final processing of a claim is based upon the terms of contract that applies to the

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members' benefit program, including benefit limitations and exclusions. If a provider or a member has a question concerning this medical policy, please contact Capital Blue Cross' Provider Services or Member Services.

VII. CODING INFORMATION

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Note: This list of codes may not be all-inclusive, and codes are subject to change at any time. The identification of a code in this section does not denote coverage as coverage is determined by the terms of member benefit information. In addition, not all covered services are eligible for separate reimbursement.

Procedure Codes that may be associated with this policy:

Proprietary Laboratory Analyses (PLA) Codes*:

0029U, 0035U, 0084U, 0154U, 0155U, 0172U, 0177U, 0180U, 0181U, 0182U, 0183U, 0184U, 0185U, 0186U, 0187U, 0188U, 0189U, 0190U, 0191U, 0192U, 0193U, 0194U, 0195U, 0196U, 0197U, 0199U, 0200U, 0201U, 0216U, 0217U, 0230U, 0231U, 0232U, 0233U, 0234U, 0236U, 0258U, 0266U, 0268U, 0269U, 0270U, 0271U, 0272U, 0273U, 0274U, 0276U, 0277U, 0278U, 0282U, 0342U, 0478U, 0481U, 0584U

Tier 1 Molecular Pathology Procedure Codes*:

81105, 81106, 81107, 81108, 81109, 81110, 81111, 81112, 81120, 81121, 81168, 81171, 81172, 81173, 81174, 81175, 81176, 81177, 81178, 81179, 81180, 81181, 81182, 81183, 81184, 81185, 81186, 81187, 81188, 81189, 81190, 81200, 81204, 81205, 81209, 81220, 81221, 81222, 81223, 81224, 81233, 81234, 81236, 81237, 81238, 81239, 81247, 81248, 81249, 81250, 81251, 81255, 81260, 81261, 81262, 81263, 81264, 81265, 81266, 81267, 81268, 81271, 81272, 81273, 81274, 81278, 81283, 81284, 81285, 81286, 81289, 81290, 81302, 81303, 81304, 81305, 81306, 81309, 81312, 81314, 81315, 81316, 81320, 81329, 81330, 81331, 81333, 81334, 81336, 81337, 81340, 81341, 81342, 81343, 81344, 81347, 81348, 81350, 81357, 81360, 81361, 81362, 81363, 81364

Tier 2 Molecular Pathology Procedure Codes*:

81400, 81401, 81402, 81403, 81404, 81405, 81406, 81407, 81408, 81479

Genomic Sequencing Procedures and Other Molecular Multianalyte Assays*

81434, 81441, 81442

*If the specific analyte is listed in a specific code, the specific CPT code would be reported; however, if the specific analyte is not listed in the more specific CPT code, the unlisted code (81479) would be reported.

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ICD-10-CM Diagnosis Codes:

Diagnosis would depend on the condition for which the testing is being performed, if the test is being performed as screening or carrier testing, and any family history of the condition.

VIII. REFERENCES

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MP 2.326	01/01/2019 Administrative Update. CPT® PLA Codes: Expanded range to include 0080U- 0083U, effective 01/01/2019.
	05/15/2019 Consensus Review. Appendices removed. No change to policy statement. CPT® PLA Codes updated to include codes issued 07/01/2019: 0001U to 0104U*
	01/01/2020 Administrative Update. Updated PLA codes listed.
	03/11/2020 Administrative Update. Added new code 0169U. Effective 04/01/2020.
	05/11/2020 Administrative Update. New PLA codes added to policy, effective 07/01/2020.
	05/11/2020 Consensus Review. No change to policy statements. References added and summary of evidence reviewed.

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11/24/2020 Administrative Update. Added new 2021 codes to policy. 0233U-0224U, 81168, 81278, 81347 and 81348.
03/25/2021 Consensus Review. No change to policy statement. Revised listed PLA codes in coding section. Added NCCN statement, updated references.
08/31/2021 Administrative Update. Added new codes to policy: 0258U, 0266U, 0268U-0274U, 0276U-0278U, 0282U
11/16/2021 Administrative Update. Removed all references to appendix section.
01/27/2022 Administrative Update. Removed 0024U due to management by Avalon. Effective date 04/01/2022.
02/18/2022 Minor Review. Moved INV statement re: at-home testing from policy guidelines into policy statement. Removed 0007U, 0010U, 0223U, 0224U. Added 0055U, 0059U, 0079U, 0087U, 0120U, 0136U, 0216U-0218U, 0228U-0229U, 0287U, 0297U-0300U.
03/11/2022 Administrative Update. Added code 0315U, effective date 04/01/2022.
08/08/2022 Administrative Update. Formatting of coding tables. Effective date 09/01/2022
09/12/2022 Administrative Update. Added codes, deleted code 0012U, 0013U, 0014U & 0056U Effective date 10/01/2022.
10/28/2022 Administrative Update. Added 0172U. Effective date 12/01/2022.
12/01/2022 Administrative Update. Added new codes 0362U, & 81441. Effective 01/01/2023
04/19/2023 Minor Review. Added statement to introduction box regarding disagreement in medical necessity. Moved two statements from the background section into the policy guidelines. Many codes removed from the coding table as they are on more specific policies. 0232U added. 0061U not a genetic test so moving to MP 4.002. References and related policies updated.
10/25/2023 Administrative Update. Added 81434 and 81442. Effective 12/01/2023.
01/05/2024 Administrative Update. Removed codes 0002U, 0032U, 0045U, 0058U, 0059U, 0087U, 0120U, 0174U, 0294U, 0297U-0300U, 0315U, 0332U, and 0333U as they are being moved to MP 2.277. Eff date 06/01/2024.
04/10/2024 Consensus Review. References and related policies updated. Coding table updated
09/18/2024 Administrative Update. New code 0481U added, 0078U deleted effective 10/01/2024.
11/19/2024 Administrative Update. Removed NCCN statement.
07/22/2025 Administrative Update. Added 0478U. Eff date 10/01/2025.
07/23/2025 Consensus Review. Updated related policies list. Updated coding sheet.
09/09/2025 Administrative Update. Added 0584U Eff 10/01/2025.

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Health care benefit programs issued or administered by Capital Blue Cross and/or its subsidiaries, Capital Advantage Insurance Company®, Capital Advantage Assurance, Company® and Keystone Health Plan® Central. Independent licensees of the Blue Cross BlueShield Association. Communications issued by Capital Blue Cross in its capacity as administrator of programs and provider relations for all companies.

Policies Related to Genetic Testing

Please note: Due to the rapid changes taking place in the genetic testing field this may not be a complete list of all Capital Blue Cross genetic testing policies. Please review the Capital Blue Cross policy website for additional policies which may have been added since the effective date of this policy.

G2022	Biomarker Testing for Autoimmune Rheumatic Disease
G2055	Prenatal Testing for Fetal Aneuploidy
G2100	In Vitro Chemoresistance and Chemosensitivity Assays
G2113	Oral Cancer Screening and Testing
G2123	Biomarker Testing for Multiple Sclerosis and Related Neurologic Diseases
G2125	Urinary Tumor Markers for Bladder Cancer
G2150	Biomarkers for Myocardial Infarction and Chronic Heart Failure
MP 2.050	Diagnostic Testing and Risk Assessment for Alzheimer's Disease (Biochemical and Genetic)
MP 2.211	Germline Genetic Testing for Hereditary Breast-Ovarian Cancer Syndrome and other High-Risk Cancers (BRCA1, BRCA2, PALB2)
MP 2.218	Pharmacogenomic and Metabolite Markers for Patients with Inflammatory Bowel Disease Treated with Thiopurines
MP 2.233	Genetic Testing for Cardiac Ion Channelopathies
MP 2.234	Cytochrome p450 Genotype Guided Treatment Strategy
MP 2.241	Molecular Analysis (Including Liquid Biopsy) for Targeted Therapy or Immunotherapy for Non-Small Cell Lung Cancer
MP 2.242	Genetic Testing for Developmental Delay/Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies
MP 2.245	Gene Expression-Based Assays for Cancers of Unknown Primary
MP 2.246	Genetic Testing for Familial Cutaneous Malignant Melanoma
MP 2.247	Genetic Testing of CADASIL Syndrome
MP 2.248	Genetic Testing for Predisposition to Inherited Hypertrophic Cardiomyopathy
MP 2.249	Use of Common Genetic Variants (Single Nucleotide Polymorphisms) to Predict Risk of Non-Familial Breast Cancer
MP 2.251	Genetic Testing for Alpha-1 Antitrypsin Deficiency
MP 2.253	Genetic Testing for Inherited Thrombophilia
MP 2.255	Genetic Testing for PTEN Hamartoma Tumor Syndrome
MP 2.257	Genetic Testing for Duchenne and Becker Muscular Dystrophy
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MEDICAL POLICY

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MP 2.261	Noninvasive Fetal RHD Genotyping Using Cell-Free Fetal DNA
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MP 2.267	Circulating Tumor DNA and Circulating Tumor Cells for Cancer Management (Liquid Biopsy)
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MP 2.274	Genetic Testing for Li-Fraumeni Syndrome
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MP 2.323	General Approach to Evaluating the Utility of Genetic Panels

MEDICAL POLICY

POLICY TITLE	GENERAL APPROACH TO GENETIC TESTING
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