Policy	MM-047
Effective Date	**09/01/2024**
Reviewed/Revised Date	04/27/2026
Next Review Date	04/27/2027
Origination Date	10/01/2026
Originated Department	Clinical Operations

General Policy on Genetic Testing

Audience
Providers, Members, Brokers, MHC

Purpose

Medical policies provide general support for applying Mountain Health Co-Op member policy document coverage decisions, and the member-specific benefit plan document must be referenced. The terms of the member-specific Policy document may differ from the standard benefit plan based on this medical policy. If there is a conflict between a member-specific policy document and the Mountain Health Co-Op medical policy, the document supersedes this policy. Any person(s) applying this medical policy must identify member eligibility, the member-specific policy document, and related policies or guidelines before applying this medical policy, including the existence of any state or federal guidance. Mountain Health Co-Op medical policies are designed for informational purposes only and are not an authorization, explanation of benefits, or contract. Receipt of benefits is subject to the satisfaction of all terms and conditions of the member-specific policy document coverage. Mountain Health Co-Op reserves the sole discretionary right to modify all policies and guidelines at any time

Definition

Genetic testing involves identifying specific changes to the DNA, RNA, genes, or other chromosomes in a person's body. The results of a genetic test can confirm or rule out a suspected genetic condition causing a disease or illness. These changes can be inherited or arise spontaneously (somatic).

Several types of genetic testing can be performed. These include:

· Molecular genetic tests (or gene tests) study single genes or short lengths of DNA to identify variations or mutations that lead to a genetic disorder.

· Chromosomal genetic tests analyze whole chromosomes or long lengths of DNA to see if

significant genetic changes, such as an extra copy of a chromosome, cause a genetic condition.
· Biochemical genetic tests study the amount or activity level of proteins; abnormalities in either can indicate changes to the DNA that result in a genetic disorder.
Genetic testing may be performed for several different purposes, including but not limited to:
· **Diagnostic**: This identifies or rules out a specific genetic or chromosomal condition. Genetic testing is often 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.
· **Prognostic**: To determine or refine estimates of the disease's natural history or recurrence in patients already diagnosed with the disease. To predict natural disease course, e.g., aggressiveness, recurrence, and risk of death. This type of testing may use gene expression of affected tissue to predict the course of disease.
· **Predictive**: These tests are used in pre-symptomatic individuals 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. Predictive testing can identify mutations that increase a person's risk of developing disorders with a genetic basis, such as certain types of cancer. Pre-symptomatic testing can determine whether a person will develop a genetic disorder, such as
hereditary hemochromatosis (an iron overload disorder), before any signs or symptoms appear. Predictive and pre-symptomatic testing results can provide information about a person’s risk of developing a specific disorder and help make decisions about medical care.
· **Therapeutic**: To determine that a particular therapeutic intervention is potentially effective (or ineffective) for an individual patient. To determine the probability of favorable or adverse response to medications. To detect genetic variants that alter the risk of treatment, adverse events, drug metabolism, drug effectiveness, etc., e.g., cytochrome p450 testing). To detect genetic mutations that adversely affect response to environmental exposures that are ordinarily tolerated, such as G6PD deficiency, genetic disorders of immune function, and amino
acidopathies.
· **Newborn screening**: This is used just after birth to identify genetic disorders that can be treated early in life. Millions of babies are tested each year in the United States. All states currently test infants for phenylketonuria (a genetic disorder that causes intellectual disability if left untreated) and congenital hypothyroidism (a disorder of the thyroid gland). Most states also test for other genetic disorders.
· **Carrier testing**: Carrier testing is 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.
· **Prenatal testing**: This type of testing detects changes in a fetus's genes or chromosomes before birth. It 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. However, it cannot identify all possible inherited disorders and birth defects.

· **Preimplantation testing**: Also called preimplantation genetic diagnosis (PGD), is a specialized technique that can reduce the risk of having a child with a particular genetic or chromosomal disorder. It detects genetic changes in embryos created using assisted reproductive techniques such as in-vitro fertilization. In-vitro fertilization involves removing egg cells from a woman's ovaries and fertilizing them with sperm cells outside the body. A few cells are taken from these
embryos to perform preimplantation testing and test for specific genetic changes. Only embryos without these changes are implanted in the uterus to initiate a pregnancy.

· **Forensic testing**: Forensic testing uses DNA sequences to identify individuals for legal purposes. Unlike the tests described above, forensic testing is not used to detect gene mutations associated with disease. This type of testing can identify crime or catastrophe victims, rule out or implicate a crime suspect, or establish biological relationships between people (for example, paternity).

Policy/Procedure

Mountain Health Co-Op covers genetic tests and considers them medically necessary when all the following General Guidelines are met.

General Guidelines for Determining Genetic Testing Coverage:

1. **For Diagnostic Testing:**
   - **1.1** The member displays clinical features or is at direct risk of inheriting the mutation in question; and
   - **1.2** The test results will be used to develop a clinically useful approach or course of treatment or to cease unnecessary monitoring or treatments for the individual being tested. Clinically useful test results allow providers to do at least one of the following:
     - **a)** Inform interventions that could prevent or delay disease onset;
     - **b)** Detect disease at an earlier stage when treatment is more effective;
     - **c)** Manage the treatable progression of an established disease;
     - **d)** Treat current symptoms significantly affecting a member's health
     - **e)** Guide decision-making for the member's current or planned pregnancy; an
   - **1.3** The genetic disorder could not be diagnosed through the completion of conventional diagnostic studies, pedigree analysis, and genetic counseling consistent with the community standards and
   - **1.4** The member has not previously undergone genetic testing for the disorder unless significant changes in testing technology or treatments indicate that test results or outcomes may change due to repeat testing and
   - **1.5** Technical and clinical performance of the genetic test is supported by published peer-reviewed medical literature.
2. **For Prognostic Testing:**
   - **2.1** An association of the marker with the natural history of the disease has been established, and
   - **2.2** Clinical efficacy of identifying the mutation has been established, and:

a) Provides incremental prognostic information above that of standard testing and
b) Reclassifies patients into clinically relevant prognostic categories for which there are different treatment strategies and
c) Reclassification leads to changes in medical management that improve clinical outcomes.

3. **For Therapeutic Testing**
   3.1 Genetic testing identifies variants of a phenotype/metabolic state that relate to different pharmacokinetics, drug efficacy, or adverse drug reactions and
   3.2 Clinical efficacy of identifying the mutation has been established, and:
      a) Leads to initiation of effective medication(s) or other treatments; or
      b) Leads to discontinuation of medications or other treatments that are ineffective or harmful; or
      c) Leads to clinically meaningful change in the dosing of medications that is likely to improve outcomes.
4. **For Carrier Screening:**
   4.1 Single gene testing for carrier status of heritable diseases is COVERED when documentation in the medical records indicates that the individual meets either a OR b below:
      a) The test is ordered by a board-certified medical geneticist or genetic counselor or provider with specialty knowledge aligned with the testing being requested, not employed by or contracted with the commercial laboratory performing the testing, and medical records document a detailed family history/pedigree and pre-test genetic counseling; or
      b) For tests ordered by a provider other than a geneticist or genetic counselor, ALL of the following criteria must be met:
         i. Medical records document a detailed family history/pedigree and pre-test genetic counseling by ONE of the following:
            - A board-certified medical geneticist not employed by or contracted with the commercial laboratory performing the testing or
            - A board-certified genetic counselor not employed by or contracted with the commercial laboratory performing the testing or
            - A certified genetic clinical nurse or advanced practice nurse in genetics (APNG) is not employed by or contracted with the commercial laboratory performing the testing. Another qualified healthcare professional (e.g., physician, physician assistant, or nurse practitioner) with specialized training or certification in medical genetics is not employed by or contracted with the commercial laboratory performing the testing.

         ii. The individual is currently pregnant or contemplating pregnancy AND is at high risk of being a carrier of a specific genetic disorder based on family history.
      c) Examples include, but are not limited to:

i. One parent is a known carrier of a clinically significant X-linked recessive or autosomal recessive disease (e.g., hemophilia, cystic fibrosis, Duchenne muscular dystrophy, sickle cell anemia, or Tay Sachs disease).
ii. A child of the individual(s) has been identified with an autosomal recessive or X-linked disorder.
iii. One or both parents have a close blood relative (e.g., first or second-degree relative) who is affected by a specific genetic disorder, or the first-degree relative has an affected child with an autosomal recessive or X-linked disorder.
iv. There is a maternal history of two or more fetal losses.
v. The test results will affect reproductive choices

5. **For Pre-implantation Testing:**

5.1 Preimplantation genetic diagnosis (PGD) testing may be considered medically necessary as an adjunct to in vitro fertilization (IVF)(when infertility services are a covered benefit by the plan) in couples not known to be infertile who meet the following criteria (must meet ALL):

a) The natural history of the disorder is well understood, and there is a reasonable and

b) The disease being tested is one with high morbidity/mortality in the homozygous or compound heterozygous state and

c) The member/couple has undergone formal genetic counseling, with a certified genetic counselor or clinical medical geneticist not directly employed or contracted with the lab performing the test and

d) The member has a > 5% chance of live birth per cycle of IVF with or without ICSI and

e) For evaluation of an embryo at an identified elevated risk of a genetic disorder such as when (Must Meet i or ii):

i. Any **ONE** of the following:
- Both partners are known carriers of a single-gene autosomal recessive disorder.
- One partner is a known carrier of a single-gene autosomal recessive disorder, and the partners have one offspring that has been diagnosed with that recessive disorder.
- One partner is a known carrier of a single-gene autosomal dominant disorder;
- One partner is a known carrier of a single X-linked disorder or

ii. For evaluation of an embryo at an identified elevated risk of structural chromosomal abnormality such as:
- Parent with balanced or unbalanced chromosomal translocation.

f) Testing is limited to targeted testing (i.e., known parental mutations).

5.2 Preimplantation genetic diagnosis as an adjunct to IVF is considered investigational in patients/couples who are undergoing IVF in all situations other than those specified above. There is insufficient evidence to support a conclusion concerning this procedure's health outcomes or benefits.

6. **Genetic Testing is NOT Covered in the following circumstances:**

6.1 The test is performed without symptoms or high-risk factors for a genetic disease.

6.2 Knowledge of genetic status will not affect treatment decisions, surveillance, reproductive decisions, and/or health outcomes of the individual being tested.

6.3 The test is obtained without an order from a licensed healthcare professional, including direct-to-consumer testing (mail order, online ordering).

6.4 Tests for the member or family members are performed solely for the purposes of genetic counseling, family planning, or health screening.

6.5 Tests for research to find a rare or new gene not previously identified or of unclear clinical significance.

6.6 The test is performed as a general screening tool, other than newborn screening, per state mandates.

6.7 The testing is performed to screen for nonmedical traits (e.g., eye color, hair color).

6.8 The testing is performed solely to determine the sex of a child.

6.9 The testing is performed solely to determine the paternity of a child.

6.10 Genetic testing is considered unnecessary when performed entirely for nonmedical reasons (e.g., a general interest in genetic test results).

6.11 Tests of a member's germline DNA are done to benefit family member(s) rather than to benefit the member being tested unless a family member is also a plan member.

6.12 Forensic genetic testing or testing required as part of legal proceedings

6.13 **Note**: Separate Medical Policies may apply to some specific genetic tests and panels. For additional genetic testing policies, see the Genetics Category Section of the Coverage Website.

7. Clinical Rationale

7.1 The National Institutes of Health's (NIH) National Human Genome Research Institute (NHGRI) defines genetic testing as the analysis of human DNA, RNA, chromosomes, proteins, and specific metabolites to detect disease-related genotypes, mutations, phenotypes, or karyotypes for clinical purposes. Clinical purposes may include:

- **a)** Diagnostic testing to confirm an individual is affected with a specific inherited disease (e.g., Duchenne or Becker muscular dystrophy)
- **b)** Predictive testing to identify gene variants that increase an individual's risk of developing an inherited disease (e.g., Huntington's disease)
- **c)** Carrier testing to determine whether individuals carry a mutation for a disorder that could be passed on to their offspring (e.g., cystic fibrosis or Tay-Sachs disease)
- **d)** Prognostic testing to predict how a disease might progress (e.g., breast cancer genomics assays to predict recurrence in primary breast cancer)
- **e)** Prenatal/pre-implantation genetic testing to identify potentially serious genetic conditions in a fetus before birth
- **f)** Newborn screening is used to detect various inherited conditions in newborns that require prompt intervention to prevent long-term health consequences (e.g., phenylketonuria, cystic fibrosis, hyperbilirubinemia).

7.2 Genetic tests function in two environments: the laboratory and the clinic. Genetic tests are evaluated based primarily on analytical validity, clinical validity, and clinical utility. These characteristics evaluate the performance of a genetic test from the viewpoint of both the laboratory and the clinicalperspectives. Analytical validity

evaluates the test's ability to do what it intends; clinical validity evaluates the test result's link to a relevant clinical outcome; and clinical utility evaluates the result's link toeffective clinical treatment and management options.

7.3 **Analytical Validity.** Analytical validity is defined as the ability of a test to detect or measure the analyte it is intended to detect or measure. This characteristic is critical for all clinical laboratory testing, not only genetic testing, as it provides information about the test's ability to perform reliably at its most basiclevel. It is relevant to how well a test performs in a laboratory.

7.4 **Clinical Validity.** The clinical validity of a genetic test is its ability to accurately diagnose or predict the risk of a particular clinical outcome. A genetic test's clinical validity relies on an established connection between the DNA variant being tested for and a specific health outcome.

7.5 Clinical validity measures a test's performance in a clinical rather than laboratory setting. Many measures are used to assess clinical validity, but the two of key importance are clinical sensitivity and positive predictive value. Genetic tests can be either diagnostic or predictive; therefore, the measures used to assess the clinical validity of a genetic test must consider this. For the purposes of a genetic test, positive predictive value can be defined as the probability that a person with a positive test result (i.e., the DNA variant tested for is present) either has or will develop the disease the test is designed to detect. Positive predictive value is the test measure most commonly used by physicians to gauge the usefulness of a testto the clinical management of patients. Determining the positive predictive value of a predictive genetic test may be difficult because many different DNA variants and environmental modifiers may affect the development of a disease. In other words, a DNA variant may have a known association with a specific health outcome, but it may not always be causal. Clinical sensitivity may be defined as the probability that the test detects people who have or will develop a disease.

7.6 **Clinical Utility.** Clinical utility considers the impact and usefulness of the test results to the individual and family and primarily considers the implications that the test results have for health outcomes (for example, is treatment or preventive care available for the disease). It also includes the test's utility more broadly for society and can encompass considerations of the psychological, social, and economic consequences of testing.

<u>Policy Issues</u>

7.7 These three characteristics of genetic tests—analytical validity, clinical validity, and clinical utility—are tied to public policy issues. Specifically, these characteristics are relevant to (1) the federal regulation of genetic tests and (2) coverage decisions by payers.

7.8 Oversight of Genetic Tests. Genetic tests are regulated by the Food and Drug Administration (FDA) and the Centers for Medicare & Medicaid Services (CMS) through the Clinical Laboratory Improvement Amendments (CLIA). The FDA regulates genetic tests manufactured by industry and sold for clinical diagnostic use. These test kits usually come prepackaged with all the reagents and instructions that a laboratory needs to perform the test and are considered products by the FDA. FDA requires manufacturers of the kits to ensure that the test detects what the manufacturer says it will in the intended patient population. Concerning the

characteristics of a genetic test, this process requires manufacturers to prove that their test is clinically valid. Depending on the perceived risk associated with the intended use promoted by the manufacturer, the manufacturer must determine that the genetic test is safe and effective or that it is substantially equivalent to something already on the market with the same intended use.

7.9 Most genetic tests, however, are performed not with test kits but as laboratory testing services (referred to as either laboratory-developed or "homebrew" tests), meaning that clinical laboratories perform the test in-house and make most or all of the reagents used in the tests. The FDA does not currently regulate laboratory-developed tests (LDTs) in the way that test kits are. Therefore, the clinical validity of the majority of genetic tests is not regulated.

7.10 The FDA does currently regulate certain components used in LDTs, known as Analyte Specific Reagents (ASRs), but only if the ASR is commercially available. If the ASR is made in-house by a laboratory performing the LDT, the FDA does not regulate the test. This type of test is sometimes referred to informally as a "homebrew-homebrew" test.

7.11 Any clinical laboratory test performed for health-related reasons on a human specimen with results returned to the patient must be performed in a CLIA-certified laboratory. CMS primarily administers CLIA with the Centers for Disease Control and Prevention (CDC) and the FDA. FDA determines the test's complexity category so the laboratories know which CLIA requirements they must follow. As previously noted, CLIA only regulates the analytical validity of a clinical laboratory test. It generally establishes requirements for laboratory processes, such as personnel training and quality control or quality assurance programs. CLIA requires laboratories to prove that their tests work properly, maintain the appropriate documentation, and show that laboratory professionals interpret tests with the appropriate training. Supporters of the CLIA regulatory process argue that regulation of the testing process gives laboratories optimal flexibility to modify tests as new information becomes available. Critics argue that CLIA does not go far enough to assure the accuracy of genetic tests since it only addresses analytical validity and not clinical validity.

7.12 Although the analytical validity of genetic tests is regulated by CMS through CLIA (P.L. 100- 578), as noted, the majority of genetic tests are not controlled based on (in any part) an assessment of their clinical validity. Given that most genetic tests are LDTs, advocates for increased regulation of genetic tests have expressed concern that the majority of genetic tests are not assured to be clinically valid and that, therefore, the results of the tests could be either misleading or not useful to the individual. This has also raised concerns about direct-to-consumer marketing of genetic tests—as most of these tests are also LDTs and not test kits—where the connection between a DNA variant and a clinical outcome (clinical validity) has not been clearly established. Because clinical validity is not part of the regulatory regime for LDTs currently, tests with unproven clinical validity can be marketed to consumers. Marketing of such tests to consumers directly may mislead consumers into believing that the advice given to them based on the results of such tests could improve their health status or outcomes when, in fact, there is no scientific basis—or inadequate evidence—underlying such an assertion. This issue was the subject

of a July 2006 hearing by the Senate Special Committee on Aging, as well as two reports by the U.S. Government Accountability Office (GAO), in 2006 and 2010.

7.13 Coverage of Genetic Tests. While insurers generally require that, where applicable, a test be approved by the Food and Drug Administration, they also want evidence that it is "medically necessary"; that is, evidence demonstrating that a test will affect a patient's health outcome positively. This requirement of evidence of improved health outcomes underscores the importance of patient participation in long-term research in genetic medicine. Particularly for genetic tests, data on health outcomes may take a long time to collect. Although payers are beginning to cover companion diagnostics and other genetic tests, they may require stringent evidence of improved health outcomes.

7.14 Clinical utility and clinical validity both figure prominently in coverage decisions by payers, by both private health insurers and public programs, and in particular, "clinical utility data are necessary for reimbursement decisions." There are many genomics-based tests where the evidence of clinical utility is limited, and therefore, "[a] critical challenge to genomic medicine is how we bridge the evidence gap necessary to pave the way for coverage and reimbursement of genetic tests." While a lack of such data can hinder or complicate coverage and reimbursement decisions, potentially leaving patients without coverage for these tests, the lack of data may also leave payers unable to evaluate the effectiveness of a test comprehensively.

7.15 Payers, both private and public, have implemented approaches to covering genomic technologies concomitant with the collection of clinical utility data. The Patient Protection and Affordable Care Act of 2010 (ACA, P.L. 111-148) in some cases requires and in some, allows private health insurers, Medicare, and Medicaid to cover clinical preventive services (as specified in the law) and outlines cost-sharing requirements in some cases for these services. However, the ACA provisions, in some cases, tie coverage of clinical preventive services to determinations by the U.S. Preventive Services Task Force (USPSTF, located in the Agency for Healthcare Research and Quality [AHRQ]), and these determinations are based on the quality of the evidence available to support a given clinical preventive service.

The Genetic Test Result

7.16 Genetic tests can provide information about inherited genetic variations, that is, the individual's genes inherited from their mother and father, and about acquired genetic variations, such as those that cause some tumors. Acquired variations are not inherited but acquired in DNA due to replication errors or exposure to mutagenic chemicals and radiation (e.g., UV rays). In contrast to most other medical tests, genetic tests can be performed on material from a body and may continue to provide information after the individual has died as a result of the stability of the DNA molecule.

7.17 DNA-based testing of inherited genetic variations differs from other medical testing in several ways. These test results can have exceptionally long-range predictive powers over an individual's lifespan; they can predict disease or increased risk for disease in the absence of clinical signs or symptoms; they can reveal the sharing of genetic variants within families at precise and calculable rates;

and, at least theoretically, they have the potential to generate a unique identifier profile for individuals.

7.18 Genetic changes to inherited genes can be acquired throughout a person's life (acquired genetic variation). Tests performed for acquired genetic variations that occur with a disease have implications only for individuals with the disease and not the genetic constitution of a family member. Tests for acquired genetic variations are also usually diagnostic rather than predictive since they are generally performed after the presentation of symptoms.

7.19 Types of genetic tests include sequencing the entire genome, exome, or targeted regions of the exome, panel testing, sequencing a specific gene or exon within a gene, analyzing duplication/deletion variants, evaluating single nucleotide polymorphisms (SNPs), evaluating chromosomes by karyotype or microarray, and evaluating gene rearrangements, deletions, duplications, or fusions. Common molecular techniques include karyotyping, microarray, polymerase chain reaction (PCR) sequencing, melt curve analysis, restriction digestion, fluorescent in situ hybridization (FISH), and chromogenic in situ hybridization (CISH).

7.20 Choi et al. (2009) examined how new genetic technology, such as next-generation sequencing and chromosomal microarray, has led to the ability to examine many genes simultaneously, resulting in a proliferation of genetic panels. Panels using next-generation technology are intuitively attractive in clinical care because they can screen for numerous variants within a single gene or multiple genes quickly, which may lead to greater efficiency in the work-up of genetic disorders. One potential challenge of gene panel testing is the identification of genetic variants of unknown significance and variants for which clinical management is uncertain and may lead to unnecessary follow-up testing and procedures. This testing may also identify genetic abnormalities for which there are no approved therapies or may not be actionable.

<u>Federal Regulation</u>

7.21 According to the NHGRI several federal agencies regulate genetic tests, including, the Food and Drug Administration (FDA), the Centers for Medicare and Medicaid Services (CMS), and the Federal Trade Commission (FTC). Genetic and Genomic tests, like other types of diagnostic tests, can be evaluated and regulated on the following three criteria, analytical validity, clinical validity and clinical utility.

7.22 CMS implements regulations to control the analytical validity of clinical genetic tests, but there is no federal oversight of the clinical validity of most genetic tests. Since clinical genomics is a relatively new field, frameworks to evaluate the clinical utility of genetic tests are still being developed. CMS regulates clinical laboratories, including clinical genetic testing laboratories, through its CLIA program. CLIA refers to the "Clinical Laboratory Improvement Amendments" of 1988, which established a certification process laboratories must pass to conduct clinical testing legally. CLIA aims to determine clinical testing quality, including verification of the procedures used and the qualifications of the technicians processing the tests. It also comprises proficiency testing for some tests.

7.23 The FDA's mission is to protect public health by ensuring the safety, efficacy, and security of drugs, biological products, and medical devices. The agency considers genetic tests a special medical device; therefore, these diagnostic tools fall within

the FDA's regulatory purview. Until recent years, the FDA chose to apply "enforcement discretion" to the vast majority of genetic tests, which means they have the authority to regulate tests but choose not to. In the current regulatory landscape, whether the FDA regulates a test is determined by how it comes to market. For example, a test comes to market as a laboratory-developed test (LDT), where the test is developed and performed by a single laboratory and specimen samples are sent to that laboratory for testing. To date, the FDA has practiced enforcement discretion for LDTs. This means that LDTs are being used in the clinic without the FDA's assessment of their analytical and clinical validity. The FDA initially applied enforcement discretion on LDTs T because clinical genetic testing was not very widespread in the past - however, due to the rapid advances in next-generation sequencing (NGS) technology, the pervasiveness of clinical genetic testing today, the growth of direct-to-consumer (DTC) genomic testing, and FDA's mounting concern that unregulated tests pose a public health threat, FDA is modifying its approach.

7.24 The FTC's primary mission is to enforce Section 5 of the FTC Act, which prohibits unfair or deceptive trade practices. A company acts deceptively if it makes misleading material statements or omissions likely to mislead reasonable consumers. A practice is unjust if it causes or is likely to cause substantial injury to consumers that is neither reasonably avoidable by consumers nor outweighed by countervailing benefits to consumers or competition. The Commission has used its authority under Section 5 in cases where, for example, it has reason to believe that a business made false or misleading claims about its privacy or data security procedures or failed to employ reasonable security measures and, as a result, causes or is likely to cause substantial consumer injury.

7.25 Walker (2010) stated that according to an undercover investigation by the Government Accountability Office (GAO), home genetic tests often provide incomplete or misleading information to consumers. For the GAO investigation, investigators purchased 10 tests each from 4 different direct-to-consumer genetic test companies: 23andMe, deCode Genetics, Navigenics, and Pathway Genomics. Five saliva donors each sent 2 DNA samples to each company. In one sample, the donor used his or her real personal and medical information; for the second sample, they developed faux identifying and medical information. The results, according to the GAO, were far from precise. For example, a donor was told by a company that he had a "below average" risk of developing hypertension, but a second company rated his risk as "average", while a third company, using DNA from the same donor, said the sample revealed an "above average" risk for hypertension. Sometimes, the results conflicted with the donor's real medical condition. None of the genetic tests offered to consumers has undergone FDA pre-market review.

7.26 According to the American College of Medical Genetics (ACMG), an important issue in genetic testing is defining the scope of informed consent. The obligation to counsel and obtain consent is inherent in the clinician-patient and investigator-subject relationships. In the case of most genetic tests, the patient or subject should be informed that the test might yield information regarding a carrier or disease state that requires difficult choices regarding their current or future health, insurance coverage, career, marriage, or reproductive options. The objective of

informed consent is to preserve the individual's right to decide whether to have a genetic test. This right includes the right of refusal should the individual decide the potential harm (stigmatization or undesired choices) outweighs the potential benefits.

Applicable Codes

CPT Codes

All genetic testing codes and other laboratory testing codes apply to genetic/molecular testing. There are too numerous to list.

HCPCS Codes

All genetic testing codes and other laboratory testing codes apply to genetic/molecular testing. There are too numerous to list.

Vendors

- **Personify**
- **HPS**

References

1. American College of Medical Genetics and Genomics (ACMG). "ACMG position statement onprenatal/preconception expanded carrier screening". June 2013. Accessed May 11, 2020. Available at: https://www.acmg.net/PDFLibrary/PrenatalPreconeption-Expanded-Carrier-Screening.pdf
2. Centers for Medicare and Medicaid Services (CMS). CMS Manual System. Pub 100-04 –Medicare Claims Processing. Transmittal 2639. Date: Jan. 25, 2013. SUBJECT: Healthcare Common Procedure Coding System (HCPCS) Codes Subject to and Excluded from Clinical Laboratory Improvement Amendments (CLIA) Edits. See http://www.cms.hhs.gov/CLIA/.
3. Choi, M, Scholl, UI, Ji, W, et al. Genetic diagnosis by whole exome capture and massively parallel DNA sequencing. Proceedings of the National Academy of Sciences of the United States of America. 2009 Nov 10;106(45):19096-101. PMID: 19861545
4. Congressional Research Service (CRS). (2015) "Genetic Testing: Background and Policy Issues" CRS Report RL33832. By Amanda K. Sarata. Available at: https://crsreports.congress.gov/product/pdf/RL/RL3383
5. Congressional Research Service (CRS) Report R43438, Regulation of Clinical Tests: In Vitro Diagnostic (IVD) Devices, Laboratory Developed Tests (LDTs), and Genetic Tests, by Amanda K. Sarata and Judith A. Johnson
6. Congressional Research Service (CRS) Report RL34376, Genetic Exceptionalism: Genetic Information and Public Policy, by Amanda K. Sarata.
7. Congressional Research Service (CRS) Report R41278, Public Health, Workforce, Quality, and Related Provisions in ACA: Summary and Timeline, coordinated by C. Stephen Redhead and Elayne J. Heisler.
8. Government Accountability Office (GAO) "Direct-To-Consumer Genetic Tests: Misleading Test Results Are Further Complicated by Deceptive Marketing and Other Questionable Practices," GAO-10-847T, July 22, 2010.
9. Government Accountability Office (GAO) "Nutrigenetic Testing: Tests Purchased from Four Web Sites Mislead Consumers," GAO-06-977T, July 27, 2006.
10. McCormack R.T. et al. "Co-development of Genome-Based Therapeutics and Companion Diagnostics," Journal of the American Medical Association, published online February 12, 2014
11. National Human Genome Research Institute (NHGRI), "Reimbursement Models to Promote Evidence Generation and Innovation for Genomic Tests," http://www.genome.gov/27552210.
12. Saidel ML, Ananth U, Rose D, Farrell C. Non-Invasive prenatal testing with rolling circle amplification: Real-world clinical experience in a non-molecular laboratory. J Clin Lab Anal. 2023 Mar;37(6):e24870. Doi: 10.1002/jcla.24870. Epub 2023 Mar 27. PMID: 36972484; PMCID: PMC10156098.

13. Senate Special Committee on Aging, “At Home DNA Tests: Marketing Scam or Medical Breakthrough?” http://www.aging.senate.gov/hearings/at-home-dna-tests-marketing-scam-or-medical-breakthrough.
14. Walker EP. Home genetic tests get 'F' from GAO. July 22, 2010. Medpage today. https://www.medpagetoday.com/Genetics/GeneticTesting/21326.

Review/Revision/Approval History	
Date	Description
06/01/2024	**New Policy**
04/27/2026	**Reviewed and approve by Mountain Health CO-OP Policy Committee**

Disclaimer

This document is for informational purposes only and should not be relied on in the diagnosis and care of individual patients. Medical and Coding/Reimbursement policies do not constitute medical advice, plan preauthorization, certification, an explanation of benefits, or a contract. Members should consult appropriate healthcare providers for medical advice, care, and treatment. Benefits and eligibility are determined before medical guidelines and payment guidelines are applied. Benefits are determined by the member’s benefit plan, effective when services are rendered.

The codes for treatments and procedures applicable to this policy are included for informational purposes. Including or excluding a procedure, diagnosis, or device code(s) does not constitute or imply member coverage or provider reimbursement policy. Please refer to the member's contract benefits in effect at the time of service to determine coverage or non-coverage of these services as they apply to an individual member.
Mountain Health Co-Op makes no representations and accepts no liability regarding the content of any external information cited or relied upon in this policy. Mountain Health Co-Op updates its Coverage Policies regularly and reserve
es the right to amend these policies and give notice per State and Federal requirements.