

**MEDICAL ASSOCIATES HEALTH PLANS
HEALTH CARE SERVICES POLICY AND PROCEDURE MANUAL
POLICY NUMBER: PP 91**

POLICY TITLE: GENETIC TESTING

POLICY STATEMENT: A genetic test is the analysis of human DNA, RNA, chromosomes, proteins or metabolites in order to detect alterations related to an inheritable or acquired disease or disorder. Genetic tests are conducted for a number of purposes, including predicting disease risk, newborn screening, determining clinical management, identifying carriers, and establishing prenatal or clinical diagnoses or prognoses in individuals, families, or populations.

Types of Genetic Tests

- Diagnostic – symptomatic individual to confirm clinical diagnosis
- Carrier – family member, asymptomatic
- Predictive/Presymptomatic – at risk to develop disorder in future
- Predisposition – also at risk but not 100% penetrant (breast cancer)
- Prenatal – fetus at risk for genetic disorder
- Newborn Screening – testing for a panel of treatable biochemical disorders at birth

NOTE: Health insurer shall not release genetic information pertaining to an individual without prior written authorization of the individual. Written authorization shall be required for each disclosure and shall include the person to whom the disclosure is being made.

The following exceptions apply to the requirement:

- (a) Individuals participating in research settings, including individuals governed by the federal policy for the protection of human research subjects.
- (b) Tests conducted purely for research, tests for somatic as opposed to heritable mutations and testing for forensic purposes.
- (c) Newborn screening.
- (d) Paternity testing.
- (e) Criminal investigations.

POLICY:

MAHP covers genetic testing as being medically necessary to establish a molecular diagnosis of an inheritable disease or disorder when the member meets all of the criteria.

1. For genetic testing to be considered medically necessary by MAHP all of the following must be met:
 - The member displays clinical features or is at direct risk of inheriting the mutation in question; **and**
 - The result of the test will directly impact the treatment being delivered to the member; **and**
 - After history, physical examination, pedigree analysis, pre and post genetic counseling by a qualified and appropriately trained practitioner (can be primary care practitioner), and completion of conventional diagnostic studies, a definitive diagnosis remains uncertain.

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2. Genetic testing of children to confirm current symptomatology or predict the adult onset of disease is not considered medically necessary unless direct medical benefit would be lost by waiting until the child has reached adulthood. It is generally accepted that unless useful medical intervention can be offered to children as a result of the testing, formal testing should wait until the child is old enough to understand the consequences of testing and request it for him or herself.
3. If the member has already been diagnosed with the disease or disorder, MAHP would not provide coverage for the genetic testing.
4. There must be reasonable expectation based on family history, pedigree analysis, risk factors and/or symptomatology that a genetically inherited condition exists.
5. The genetic molecular testing must be conducted in a laboratory certified, at a minimum, under Clinical Laboratory Improvement Amendments (CLIA).
6. If appropriate, according to the recognized standards of care, there must be documentation that the patient has received pre-test counseling from a qualified professional and has given informed consent for the test as appropriate. There must also be a plan for providing post-test counseling.
7. The genotypes to be detected by the genetic test must be shown by scientific valid methods to be associated with the occurrence of the disease and analytical and clinical validity of the test must be established.

Cystic Fibrosis

Genetic molecular testing may be considered medically necessary for individuals presenting with symptoms of cystic fibrosis but have a negative sweat chloride test.

Long QT Syndrome

Genetic molecular testing for the diagnosis and management of Long QT Syndrome may be considered medically necessary for any of the following individuals:

- Those presenting with demonstrated prolonged QT intervals on resting EKG or Holter and in whom acquired cause has been ruled out.
- Those with a positive first-degree family history of sudden death or near-sudden death.
- Those with a first-degree relative who has a positive genetic test for Long QT Syndrome.

Tay Sachs Disease

Genetic molecular testing for the diagnosis and management of Tay Sachs Disease may be considered medically necessary for any of the following individuals:

- Those with known risk factors (Ashkenazi Jewish or French-Canadian heritage, family history, medical history) for Tay Sachs disease.
- For partners of Tay Sachs disease carriers as an aid in reproductive decision-making.
- For prenatal diagnosis when both parents are known to be Tay Sachs disease carriers.

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- For individuals suspected of having a variant form of Tay Sachs disease, such as adult- or –juvenile- onset or chronic Tay Sachs disease.
- For individuals suspected of having pseudodeficiency condition (asymptomatic non-Jewish individuals with low in vitro Hex-A activity).

Factor V Leiden Thrombophilia

Genetic molecular testing may be considered medically necessary for the diagnosis and management of factor V Leiden thrombophilia for patients presenting with any of the following:

- Age \leq 50, any history of unexplained venous thrombosis.
- Age \leq 50 with unexplained arterial thrombosis in the absence of other risk factors for atherosclerotic vascular disease.
- Venous thrombosis I unusual sites such as portal hepatic, mesenteric, and cerebral veins.
- Recurrent venous thrombosis
- Venous thrombosis and a strong family history of thrombotic disease.
- Venous thrombosis in pregnant women or women taking oral contraceptives.
- Asymptomatic first-degree relatives of individuals with proven symptomatic thrombophilia,
- Myocardial infarction in female smokers under age 50.
- Recurrent pregnancy loss (i.e., two or more consecutive pregnancy losses)

Conditions in which genetic molecular testing is considered **not medically necessary** include, but are not limited to:

- Alzheimer’s disease
- Alpha 1 antitrypsin deficiency
- Ataxia telangiectasis syndrome
- Hemophilia A and B
- Huntington’s chorea
- Myotonic dystrophy
- Sickle cell disease
- Prader-Willis syndrome
- Amyotrophic lateral sclerosis
- Left ventricular noncompaction cardiomyopathy.

References: Other Major Health Plan Policy Statements

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