Genetic Testing

Description

Genetic tests are medical tests that detect changes in human deoxyribonucleic acid (DNA), chromosomes, genes or gene products, such as proteins. DNA for genetic testing may be extracted from a variety of samples including blood, saliva, buccal (cheek) smear, fresh or frozen tissues, formalin-fixed, paraffin-embedded tissues as well as prenatal specimens obtained by chorionic villus sampling or amniocentesis. Most laboratories prefer blood samples, although buccal swabs and saliva samples also may be an option for certain types of genetic testing.

Genetic testing may be used for a variety of purposes:

- Carrier screening is used to identify individuals who carry one copy of a gene mutation that, when present in two copies, causes a genetic disorder. For
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information regarding carrier screening, please refer to Genetic Testing for Carrier Screening or Genetic Testing for Cystic Fibrosis Medical Coverage Policies.

- Diagnostic testing is utilized to identify or rule out a suspected genetic condition in an individual who exhibits signs and symptoms of the disorder. For information regarding diagnostic genetic testing, please refer to the following medical coverage policies:
  - BCR-ABL Testing
  - Comparative Genomic Hybridization
  - Genetic Testing for Alzheimer Disease
  - Genetic Testing for Angelman and Prader Willi Syndrome
  - Genetic Testing for Cardiac Conditions
  - Genetic Testing for Cystic Fibrosis
  - Genetic Testing for Diagnosis and Monitoring of Noncancer Indications
  - Genetic Testing for Ehlers-Danlos Syndrome
  - Genetic Testing for Hereditary Hemochromatosis
  - Genetic Testing for Inherited Thrombophilias
  - Genetic Testing for Marfan Syndrome
  - Genetic Testing for Muscular Dystrophy and Spinal Muscular Atrophy
  - Genetic Testing for Spinocerebellar Ataxia
  - JAK2 Testing
  - Whole Genome/Exome Sequencing

- Direct-to-consumer (DTC) genetic tests, also known as at-home genetic testing, are sold directly to individuals via the Internet, television, print advertisements or other marketing materials without the involvement of a doctor or other healthcare professional. After the individual places an order, a test kit is mailed to the individual who collects a sample typically by swabbing the inside of the cheek and placing the swab inside a container to return the sample by mail to the lab. Results may be provided within weeks via a website, mail or telephone. (Refer to Coverage Limitations section)

- Pharmacogenomic testing may be applied to guide treatment or assess response to medication. For information regarding pharmacogenomic tests, please refer to the following medical coverage policies:

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- **Pharmacogenomics – Cytochrome P450 Polymorphisms and VKORC1**
- **Pharmacogenomics (Pharmacogenetics) – Noncancer Indications**
- **Pharmacogenomics and Companion Diagnostics**

- Predictive testing may be used for individuals who do not exhibit signs or symptoms of a disorder but may be at increased risk for developing the disorder due to family history. There are two types of predictive testing: presymptomatic (ie, development of symptoms is certain in the presence of a gene mutation [eg, hereditary hemochromatosis, Huntington disease]) or predispositional (ie, development of symptoms is likely but not certain in the presence of a gene mutation [eg, breast cancer]). For information regarding **predictive genetic testing**, please refer to the following medical coverage policies:
  - **Genetic Testing for Alzheimer Disease**
  - **Genetic Testing for Breast and Ovarian Cancer Susceptibility**
  - **Genetic Testing for Cancer Susceptibility**
  - **Genetic Testing for Colorectal Cancer Susceptibility**
  - **Genetic Testing for Disease Risk**
  - **Genetic Testing for Hereditary Hemochromatosis**
  - **Genetic Testing for Inherited Thrombophilias**
  - **Genetic Testing for Muscular Dystrophy and Spinal Muscular Atrophy**

- Preimplantation testing is performed to identify genetic defects in embryos created through in vitro fertilization. For information regarding **preimplantation genetic testing**, please refer to **Preimplantation Genetic Testing** Medical Coverage Policy.

- Prenatal testing is offered during pregnancy to identify genetic disorders in fetuses. For information regarding **prenatal genetic testing**, please refer to **Prenatal Diagnostic Genetic Testing** Medical Coverage Policy.

Humana recognizes that the field of genetic testing is rapidly changing and that other tests may become available.
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**Coverage Determination**

Any state mandates for genetic testing take precedence over this clinical policy.

Genetic testing may be excluded by contract. Please consult the member’s individual contract regarding Plan coverage.

**General Criteria for Genetic Tests**

The General Criteria for Genetic Tests may be applied if specific criteria for a genetic test are not available on any medical coverage policy.

Humana members may be eligible under the Plan for genetic testing when the following criteria are met:

- Individual has not previously received genetic testing for the disorder. **Note:** In general, genetic testing for a particular disorder should be performed once per lifetime; however, there are rare instances in which testing may be performed more than once in a lifetime (e.g., previous testing methodology is inaccurate, a new discovery has added significant relevant mutations for a disease, significant changes in technology or treatments indicate that test results or outcomes may change as a result of repeat testing); **AND**

- Laboratory or clinical tests to definitively diagnose the genetic disorder are unavailable or results are equivocal; **AND**

- Panels including, but may not limited to, multiple genes or multiple conditions, and in cases where a tiered approach/method is clinically available, may be covered **ONLY** for the number of genes or tests deemed medically necessary to establish a diagnosis; **AND**

- Results of genetic testing will directly impact and change clinical management of the individual being tested who is a covered member; **AND**

- Technical and clinical performance of the genetic test is supported by published peer-reviewed medical literature

**Physician interpretation and reporting for molecular pathology procedures is considered integral to the primary molecular pathology procedure/laboratory testing and not separately reimbursable.**
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**Coverage Limitations**

Humana members may **NOT** be eligible under the Plan for **genetic testing** for any indications other than those listed above including the following:

- An at-risk (unaffected) individual or affected individual when a family member has been tested for mutations and received a result of variant of unknown significance (VUS) (also known as unclassified variant, variant of uncertain significance); **OR**

- Direct-to-consumer (DTC) genetic testing

These are considered experimental/investigational as they are not identified as widely used and generally accepted for the proposed uses as reported in nationally recognized peer-reviewed medical literature published in the English language.

Humana members may **NOT** be eligible under the Plan for the following:

- General population screening; **OR**

- Genetic testing for children (ie, under 18 years old) for adult-onset conditions except when such testing impacts clinical management prior to adulthood

These are considered not medically necessary as defined in the member’s individual certificate. Please refer to the member’s individual certificate for the specific definition.

**Background**

Additional information about **genetic testing** may be found from the following websites:

- Genetics Home Reference
- National Library of Medicine

**Medical Alternatives**

Physician consultation is advised to make an informed decision based on an individual’s health needs.

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Provider Claims Codes

Please refer to the Medical Coverage Policy specific to the type of genetic testing in question for any associated CPT/HCPCS code(s).

Medical Terms

**Amniocentesis** – The sampling of amniotic fluid using a hollow needle inserted into the uterus, to screen for developmental abnormalities in a fetus.

**Amniotic Fluid** – A clear, slightly yellowish liquid that surrounds the unborn baby (fetus) during pregnancy. It is contained in the amniotic sac, which is a bag of fluid inside a woman's womb (uterus) where the unborn baby develops and grows.

**Chorionic Villus** – Microscopic, finger–like projections that emerge from the outer sac which surrounds the developing baby. Chorionic villi are of fetal origin and eventually form the placenta.

**Chorionic Villus Sampling (CVS)** – A prenatal test that can detect genetic and chromosomal abnormalities of a fetus; also known as chorionic villus biopsy.

**Chromosome** – A threadlike linear strand of deoxyribonucleic acid (DNA) and associated proteins in the nucleus of cells that carries the genes and transmits hereditary information.

**Deletion/Duplication Analysis** – Laboratory testing that identifies an absence of a segment of DNA (deletion) and/or the presence of an extra segment of DNA (duplication) in a coding region.

**Deoxyribonucleic Acid (DNA)** – The molecule that carries genetic information in all living systems.

**Embryo** – Unborn or unhatched offspring in the process of development.

**Fetus** – Unborn offspring of a mammal, in particular an unborn human baby more than eight weeks after conception.

**Formalin-Fixed Paraffin-Embedded** – A method for clinical sample preservation and archiving.

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Gene – Hereditary unit found in chromosomes that determine individual characteristics of an organism; it is the “blueprint” guide for cell function.

In Vitro Fertilization – A complex series of procedures used to treat fertility or genetic problems and assist with the conception of a child.

Mutation – Change of the DNA order (sequence) within a gene or chromosome of an organism resulting in the creation of a new character or trait not found in the parental type.

Unaffected – Referring to an individual who does not manifests symptoms of a particular condition.

Variant – Alteration in the normal order (sequence) of a gene.

Variant of Unknown Significance (VUS) – An alteration (variation) in a genetic sequence of which the association with disease risk is unknown. Also called unclassified variant, variant of uncertain significance.

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