

Genetic Testing and Genetic Counseling for Diagnosis and Monitoring-Non-Cancer Indications



Medical Coverage Policy

Original Effective Date: 09/01/1995
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Change Summary: Updated Description, Coverage Determination, Provider Claims Codes, Medical Terms, References

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Disclaimer

State and federal law, as well as contract language, including definitions and specific inclusions/exclusions, take precedence over clinical policy and must be considered first in determining eligibility for coverage. Coverage may also differ for our Medicare and/or Medicaid members based on any applicable Centers for Medicare & Medicaid Services (CMS) coverage statements including National Coverage Determinations (NCD), Local Medical Review Policies (LMRP), and/or Local Coverage Determinations. See the CMS web site at <http://www.cms.hhs.gov/>. The member's health plan benefits, in effect on the date services are rendered, must be used. Clinical policy is not intended to preempt the judgment of the reviewing Medical Director or dictate to providers how to practice medicine. Providers are expected to exercise their medical judgment in rendering the most appropriate care. Clinical technology is constantly evolving, and we reserve the right to review and update this policy periodically. No part of this publication may be reproduced, stored in a retrieval system, or transmitted, in any shape or form or by any means, electronic, mechanical, photocopying, or otherwise, without permission from Humana Inc.

Description

Genetic tests are laboratory studies of human deoxyribonucleic acid (DNA), chromosomes, genes or gene products to diagnose the presence of a genetic variation associated with a high risk of having or transmitting a specific genetic disorder.

This policy addresses genetic testing and genetic counseling for the diagnosis and monitoring for non-cancer indications only. For cancer indications, refer to Humana Medical Coverage Policy, [Genetic Testing and Genetic Counseling for the Diagnosis and Monitoring for Cancer](#). For testing regarding disease risk, refer to Humana Medical Coverage Policy, [Genetic Testing and Genetic Counseling for Disease Risk](#).

Genetic testing can be utilized for the purpose of diagnosing and monitoring non-cancer indications, such as Factor V Leiden and Long QT Syndrome. This type of testing is indicated for individuals who exhibit disease symptoms and

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testing is necessary to diagnose or rule out a known or suspected genetic disorder. This testing can also be used to monitor prognosis of a disease or response to treatment.

Genetic counseling is performed by a genetics expert who provides education and guidance to individuals and families who have a genetic disease or who are at risk for such a disease. A genetics counselor uses a detailed family history and results of genetic tests to help their patients make informed decisions. Genetic counseling is recommended before and after testing.

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

Coverage Determination

Humana members **MAY** be eligible under the Plan for **genetic testing and counseling** (performed with a physician or certified genetic counselor pre- and post-test) **for the diagnosis and monitoring of non-cancer indications** when **ALL** of the following are met:

- Availability of a clinically valid test, based on published peer-reviewed medical literature; **AND**
- Results of genetic testing must impact treatment or management for a covered member; **AND**
- For testing panels including but not limited to multiple genes or multiple conditions, and in cases where a tiered approach/method is clinically available, testing would be covered **ONLY** for the number of genes or tests deemed medically necessary to establish a diagnosis; **AND**
- For diagnostic purposes, allowed once during lifetime per disease

Note: In general, genetic testing for diagnosis and monitoring for noncancer indications for a particular disease should be performed once per lifetime; however, there are rare instances in which testing should be performed more than once in a lifetime (e.g., previous testing methodology is inaccurate or a new discovery has added relevant mutations for a disease).

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Criteria for Specific Genetic Tests

Alpha-1 Antitrypsin Deficiency (AATD)

Humana members **MAY** be eligible under the Plan for **genetic testing for AATD** for **ANY** of the following indications:

- Newborns with prolonged jaundice or nonspecific signs of liver disease; **OR**
- Patients with **ANY** of the following indications **AND** have abnormally low (<120mg/dL) or borderline (90-140mg/dL) alpha-1 antitrypsin (AAT) levels:
 - Anti-proteinase 3 positive vasculitis (C-ANCA-positive vasculitis); **OR**
 - Chronic liver disease; **OR**
 - Necrotizing panniculitis; **OR**
 - Chronic respiratory diseases (including but may not be limited to chronic obstructive pulmonary disease, bronchial asthma, or bronchiectasis).

Alzheimer's Disease

Humana members **MAY** be eligible under the Plan for **genetic testing for Alzheimer's disease** when **ALL** of the following are present:

- Patient exhibits signs and/or symptoms of dementia; **AND**
- Patient is less than 50 years of age; **AND**
- Testing for ApoE genotype*
- Confirmed family history of at least two close blood relatives (first or second degree) with early-onset AD; **AND**

*Athena Diagnostics offers the following genetic tests for AD:

- ADmark® APP DNA Sequencing/Duplication Test, ADmark® PS-1 Sequencing Test and ADmark® PS-2 Sequencing Test—**These tests are used to predict risk of AD and are covered when the above criteria are met. Refer to the Humana CI, [Genetic Testing and Counseling for Disease Risk](#).**
- ADmark® Phospho-Tau/Ab42 CSF Analysis & Interpretation (Symptomatic) and ADmark® ApoE Genotype Analysis & Interpretation (Symptomatic)—**These tests are used to confirm diagnosis, not to determine risk.**

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Angelman Syndrome

Humana members **MAY** be eligible under the Plan for **genetic testing for Angelman Syndrome** when **ALL** of the following are present::

- Behavioral uniqueness, including any combination of frequent laughter/smiling; apparent happy demeanor; excitability, often with hand-flapping movements; hypermotoric behavior; short attention span; **AND**
- Delayed attainment of developmental milestones without loss of skills; **AND**
- Evidence of developmental delay by six to twelve months of age, eventually classified as severe; **AND**
- Movement or balance disorder, usually ataxia of gait and/or tremulous movement of the limbs; **AND**
- Normal metabolic, hematologic, and chemical laboratory profiles; **AND**
- Normal prenatal and birth history, normal head circumference at birth, no major birth defects; **AND**
- Speech impairment, with minimal to no use of words; receptive language skills and nonverbal communication skills are higher than expressive language skills; **AND**
- Structurally normal brain by MRI or CT, although mild cortical atrophy or dysmyelination may be observed.

Celiac Disease

Humana members **MAY** be eligible under the Plan for **genetic testing for Celiac disease** for **ANY** of the following indications:

- Differential diagnosis of an individual with celiac disease-like symptoms; **OR**
- Symptomatic individuals who have not responded to a gluten-free diet; **OR**
- Symptomatic individuals with borderline or ambiguous celiac-associated antibody results or small-bowel biopsy results

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Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL)

Humana members **MAY** be eligible under the Plan for **genetic testing for CADASIL** when **ALL** of the following are present:

- MRI findings including diffuse white matter lesions, subcortical infarcts, and/or subcortical lacunar lesions; **AND**
- Family history of stroke or dementia; **AND**
- Presence of at least one of the following symptoms:
 - Cognitive deficits; **OR**
 - Epilepsy; **OR**
 - History of migraine with aura; **OR**
 - Ischemic episodes; **OR**
 - Mood disorders; **OR**
 - Progressive dementia

Charcot-Marie-Tooth Neuropathy

Humana members **MAY** be eligible under the Plan for **genetic testing for Charcot-Marie-Tooth Neuropathy** in symptomatic patients with **ALL** of the following present:

- Acquired causes of peripheral neuropathy have been reasonably ruled out; **AND**
- Evidence of demyelinating neuropathy by EMG/NCS; **AND**
- High index of suspicion for CMT based on clinical findings. (Suggestive clinical findings may include, but are not limited to, the following: progressive peripheral motor and sensory neuropathy, distal muscle weakness and atrophy, slow nerve conduction velocity, palpably enlarged nerves, and onset 5-25 years)

Chromosome 22q11.2 Deletion Syndrome (DiGeorge Syndrome, Velocardiofacial Syndrome, Shprintzen Syndrome, Conotruncal Anomaly Face Syndrome, Cayler Cardiofacial Syndrome)

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Humana members **MAY** be eligible under the Plan to **confirm diagnosis of suspected Chromosome 22q11.2 Deletion Syndrome**.

Chromosome Analysis and Karyotyping

Humana members **MAY** be eligible under the Plan for **chromosome analysis or karyotyping** for **ANY** of the following:

- Ambiguous or abnormal genitalia; **OR**
- Amenorrhea or delay in secondary sexual characteristics; **OR**
- Failure to thrive or short stature; **OR**
- Infertility or repeat miscarriage, as follows:
 - Karyotype testing for couples with recurrent pregnancy loss, defined as two or more consecutive spontaneous abortions; **OR**
 - Karyotype testing of tissue when a couple with recurrent pregnancy loss experiences a subsequent spontaneous abortion; **OR**
 - Y-chromosome microdeletion testing in males with nonobstructive azoospermia or severe oligospermia (see Humana Medical Coverage Policy, Infertility Evaluation and Treatment); **OR**
- Mental retardation, developmental delay, or autism; **OR**
- Multiple congenital anomalies or birth defect; **OR**
- Suspected chromosomal disorder, such as Down syndrome, Turner syndrome or Klinefelter syndrome [see Humana Medical Coverage Policy, Diagnosis and Treatment of Pervasive Development Disorders (PDD)].

Comparative Genome Hybridization (CGH) Microarray Testing

Humana members **MAY** be eligible under the Plan for **CHG microarray testing** as an adjunct to conventional karyotype testing for **ANY** of the following:

- For the evaluation of chromosomal imbalances in miscarried fetuses and stillbirths; **OR**

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- For the evaluation of chromosomal imbalances in patients suspected of having a genetic syndrome, including, congenital anomalies, dysmorphic features, developmental delays, mental retardation [see Humana Medical Coverage Policy, [Diagnosis and Treatment of Pervasive Development Disorders \(PDD\)](#)]

Cystic Fibrosis (CF)

Humana members **MAY** be eligible under the Plan for **genetic testing to diagnose Cystic Fibrosis** when **BOTH** of the following are present:

- Patient has at least one characteristic of clinical feature, **or** family history of CF, **or** a positive neonatal screen; **AND**
- A positive sweat chloride on at least two occasions, **or** the presence of 2 CF mutations, **or** a positive nasal transmembrane potential

Duchenne or Becker Muscular Dystrophy

Humana members **MAY** be eligible under the Plan for **genetic testing to confirm a diagnosis of Duchenne or Becker muscular dystrophy** in individuals with **EITHER** of the following:

- Clinical findings suggestive of Duchenne or Becker muscular dystrophy. Clinical findings include, but may not be limited to, progressive symmetric muscle weakness, calf pseudohypertrophy, or cardiomyopathy; **OR**
- Greatly elevated serum CK values (serum CK concentration ≥ 5 times normal; normal CK values are <160 IU/L).

Factor V Leiden

Humana members **MAY** be eligible under the Plan for **genetic testing for Factor V Leiden** for **ANY** of the following indications:

- Have venous thrombosis in unusual sites (such as hepatic, mesenteric, and cerebral veins); **OR**
- Individuals must be <50 years with any venous thrombosis; **OR**
- Myocardial infarction in female smokers under age 50; **OR**

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- Recurrent unexplained fetal loss; **OR**
- Recurrent venous thrombosis; **OR**
- Relatives of individuals with venous thrombosis under age 50; **OR**
- Venous thrombosis and a strong family history of thrombotic disease; **OR**
- Venous thrombosis in pregnant women or taking oral contraceptives; **OR**
- Women considering oral contraceptives who have a personal or family history of venous thrombosis.

Fragile X Syndrome

Humana members **MAY** be eligible under the Plan for **genetic testing for fragile X syndrome**, when **ANY** of the following are present:

- Late-onset intention tremor or cerebellar ataxia of unknown origin; **OR**
- Premature ovarian failure; **OR**
- Mental retardation, developmental delay or autism [see Humana Medical Coverage Policy, Diagnosis and Treatment of Pervasive Development Disorders (PDD).]

Friedreich Ataxia

Humana members **MAY** be eligible under the Plan for **genetic testing for Friedreich Ataxia** to confirm diagnosis in symptomatic patients.

HFE-Associated Hemochromatosis

Humana members **MAY** be eligible under the Plan for **genetic testing for HFE-associated hemochromatosis** in symptomatic patients who have serum transferrin saturation greater than or equal to 45%. Symptoms of hemochromatosis include liver function abnormalities, weakness, lethargy, skin hyperpigmentation, diabetes mellitus, severe joint pain, impotence in males, and electrocardiographic abnormalities.

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Huntington Disease

Humana members **MAY** be eligible under the Plan for **genetic testing (CAG repeat length) for Huntington disease** in symptomatic individuals. Symptoms of Huntington disease include psychiatric and emotional changes, dementia, and movement disorder.

Long QT Syndrome (LQTS) (Jervell and Lang-Nielson Syndrome/Romano-Ward Syndrome)

Humana members **MAY** be eligible under the Plan for **genetic testing for long QT syndrome** for a member who has been confirmed to have prolonged QT interval on electrocardiogram (ECG or EKG) or Holter monitor, and an acquired cause has been ruled out (such as heart failure, bradycardia, electrolyte imbalances, or certain medications).

Myotonic Dystrophy

Humana members **MAY** be eligible under the Plan for genetic testing to confirm a diagnosis of **myotonic dystrophy**, in those who exhibit characteristics of the disease as follows:

- Adults, with some combination of muscle weakness (especially of the distal leg, hand, neck, and face), myotonia, posterior subcapsular cataracts; **OR**
- Neonates, with some combination of hypotonia, facial muscle weakness, generalized weakness, positional malformations including club foot, respiratory insufficiency

Niemann-Pick Disease

Humana members **MAY** be eligible under the Plan for genetic testing for **Niemann-Pick disease** to confirm diagnosis after biochemical testing for acid sphingomyelinase activity as follows:

- Suspected **Niemann-Pick Disease Type A** in patients presenting with hepatosplenomegaly, developmental delay, evidence of interstitial lung disease on chest radiograph, cherry-red macula; **OR**

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- Suspected **Niemann-Pick Disease Type B** in patients presenting with hepatosplenomegaly, interstitial lung disease, hyperlipidemia, thrombocytopenia; **OR**
- Suspected **Niemann-Pick Disease Type C** in patients presenting with fetal ascites or neonatal liver disease, infantile hypotonia, vertical supranuclear gaze palsy, progressive ataxia, dysarthria, dystonia.

Prader-Willi Syndrome (PWS)

Humana members **MAY** be eligible under the Plan for **genetic testing for PWS** when the following are met:

- Birth - 2 years of age:
 - Hypotonia with poor suck; **OR**
- 2 years - 6 years of age:
 - Hypotonia with history of poor suck; **AND**
 - Global developmental delay; **OR**
- 6 years - 12 years of age:
 - Excessive eating with obesity if food intake isn't controlled; **AND**
 - Global developmental delay; **AND**
 - Hypotonia with history of poor suck (hypotonia often persists); **OR**
- 13 years through adulthood:
 - Cognitive impairment; usually mild mental retardation; **AND**
 - Excessive eating with central obesity if uncontrolled; **AND**
 - Hypothalamic hypogonadism and/or typical behavior problems (such as temper tantrums or obsessive-compulsive features).

Primary Dystonia (DYT1)

Humana members **MAY** be eligible under the Plan for **genetic testing for Primary Dystonia (DYT1)** when **EITHER** of the following indications are present:

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- Patients with primary dystonia with onset at less than 30 years of age; **OR**
- Patients with primary dystonia with onset at 30 years of age or older with a relative who developed primary dystonia at less than 30 years of age.

Prothrombin (Factor II) G20210A Thrombophilia

Humana members **MAY** be eligible under the Plan for **genetic testing for Prothrombin (Factor II) G20210A Thrombophilia** as follows:

- Female, **ANY** of the following:
 - Myocardial infarction in a smoker under age 50 years; **OR**
 - Recurrent unexplained fetal loss; **OR**
 - Venous thrombosis during pregnancy or while taking oral contraceptives; **OR**
 - Personal or family history of venous thrombosis and considering oral contraceptives
- Male or female, **ANY** of the following:
 - Any venous thrombosis under the age of 50 years; **OR**
 - Recurrent venous thrombosis; **OR**
 - Relatives with venous thrombosis under age 50 years; **OR**
 - Venous thrombosis and a strong family history of thrombotic disease; **OR**
 - Venous thrombosis in unusual sites (such as hepatic, mesenteric, and cerebral veins).

Spinal Muscular Atrophy (SMA)

Humana members **MAY** be eligible under the Plan for **genetic testing to diagnose Spinal Muscular Atrophy (SMA)** in those presenting with hypotonia and muscle weakness.

Coverage Limitations

Humana members **MAY NOT** be eligible under the Plan for **genetic testing and genetic counseling for the diagnosis and monitoring of noncancer indications** for any other indications other than those listed above, including

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diagnosing **hypertrophic cardiomyopathy, methylenetetrahydrofolate reductase (MTHFR) and narcolepsy**. This technology is considered experimental/ investigational or **NOT** medically necessary as it is not identified as widely used and generally accepted for any other proposed use as reported in nationally recognized peer-reviewed medical literature published in the English language.

Self Testing Home Kits

Humana members **MAY NOT** be eligible under the Plan for **self-testing home kits** due to potential risks associated with genetic testing such as inappropriate testing, misinterpretation of results, inaccurate or not clinically valid testing, lack of follow-up care and other adverse consequences.

General Population Screening

Humana members **MAY NOT** be eligible under the Plan for general population **genetic testing for diagnosis and monitoring of non-cancer indications**. This technology is considered experimental/ investigational or **NOT** medically necessary if it is not utilized in accordance with nationally recognized standards of medical practice and/or identified as safe, widely used and generally accepted as effective for any other proposed use as reported in nationally recognized peer-reviewed medical literature published in the English language.

Whole Genome Sequencing/Genome Wide Arrays

Humana members **MAY NOT** be eligible under the Plan for **whole genome sequencing or genomewide arrays** for any indication. This technology is considered experimental/ investigational or **NOT** medically necessary if it is not utilized in accordance with nationally recognized standards of medical practice and/or identified as safe, widely used and generally accepted as effective for any other proposed use as reported in nationally recognized peer-reviewed medical literature published in the English language.

Background

Deoxyribonucleic acid, or DNA, is the hereditary material in nearly all organisms, including humans. DNA contains the biological instructions, or blueprints, for how all living organisms develop and function. A gene is a segment of DNA and a chromosome houses many genes. Every human cell contains 46 chromosomes (23 pairs). If chromosomes or a piece of a chromosome is missing or duplicated,

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then there will be missing or extra genes which may result in problems in that person's health or development.

You can learn more about specific diseases from the following sites:

- American Cancer Society - <http://www.cancer.org>
- American College of Gastroenterology - <http://www.gi.org>
- American College of Medical Genetics - <http://www.acmg.net>
- American Diabetes Association - <http://www.diabetes.org>
- American Gastroenterological Association - <http://www.gastro.org>
- American Heart Association - <http://www.americanheart.org>
- American Lung Association - <http://www.lungusa.org>
- Gene Tests - <http://www.genetests.org>
- Lab Tests Online® - <http://www.labtestsonline.org>
- National Cancer Institute - <http://www.cancer.gov>
- National Comprehensive Cancer Network (NCCN) - <http://www.nccn.org>
- National Institutes of Health (NIH) - <http://www.nih.gov>
- National Library of Medicine - <http://www.nlm.nih.gov>
- National Organization for Rare Diseases - <http://www.rarediseases.org>

Medical Alternatives

To make the best health decision for your individual needs, consult your physician.

Humana may offer a disease management program for this disease. **Call the number on your member identification card to ask about our programs to help you manage your care.**

Provider Claims Codes

All provider claims codes surrounding this topic may not be included in the following table:

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CPT® Codes	Description	Comments
83890	Molecular diagnostics; molecular isolation or extraction	
83891	Molecular diagnostics; isolation or extraction of highly purified nucleic acid	Not covered for Pervasive Development Disorder
83892	Molecular diagnostics; enzymatic digestion	
83893	Molecular diagnostics; dot/slot blot production	
83894	Molecular diagnostics; separation by gel electrophoresis (e.g., agarose, polyacrylamide)	Not covered for Pervasive Development Disorder
83896	Molecular diagnostics; nucleic acid probe, each	
83897	Molecular diagnostics; nucleic acid transfer (e.g., Southern, Northern)	
83898	Molecular diagnostics; amplification, target, each nucleic acid sequence	Not covered for Pervasive Development Disorder
83900	Molecular diagnostics; amplification, target, multiplex, first two nucleic acid sequences	
83901	Molecular diagnostics; amplification, target, multiplex, each additional nucleic acid sequence beyond 2	
83902	Molecular diagnostics; reverse transcription	
83903	Molecular diagnostics; mutation scanning, by physical properties (e.g., Single strand conformational polymorphisms (SSCP), heteroduplex, denaturing gradient gel electrophoresis(DGGE) RNA'ase A) single segment, each	
83904	Molecular diagnostics; mutation identification by sequencing, single segment, each segment	
83905	Molecular diagnostics; mutation identification by allele specific transcription, single segment, each segment	

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83906	Molecular diagnostics; mutation identification by allele specific translation, single segment, each segment	
83907	Molecular diagnostics; lysis of cells prior to nucleic acid extraction (e.g., stool specimens, paraffin embedded tissue)	
83908	Molecular diagnostics; amplification, signal, each nucleic acid sequence	
83909	Molecular diagnostics; separation and identification by high resolution technique (e.g., capillary electrophoresis)	
83912	Molecular diagnostics; interpretation and report	Not covered for Pervasive Development Disorder
83913	Molecular diagnostics; RNA stabilization	
83914	Mutation identification by enzymatic ligation or primer extension, single segment, each segment (e.g., Oligonucleotide ligation assay (OLA), single base chain extension (SBCE) or allele-specific primer extension (ASPE))	
88230	Tissue culture for non-neoplastic disorder; lymphocytic	
88248	Chromosome analysis for breakage syndromes; baseline breakage, score 50-100 cells, count 20 cells, 2 karyotypes	Refer also to Medical Coverage Policy, Diagnosis & Treatment of Pervasive Development Disorders
88261	Chromosome analysis; count 5 cells, 1 karyotype, with banding	Refer also to Medical Coverage Policy, Diagnosis & Treatment of Pervasive Development Disorders

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88262	Chromosome analysis; count 15-20 cells 2 karyotypes, with banding	Refer also to Medical Coverage Policy, Diagnosis & Treatment of Pervasive Development Disorders
88263	Chromosome analysis; count 45 cells for mosaicism, 2 karyotypes, with banding	Refer also to Medical Coverage Policy, Diagnosis & Treatment of Pervasive Development Disorders
88264	Chromosome analysis; analyze 20-25 cells	Refer also to Medical Coverage Policy, Diagnosis & Treatment of Pervasive Development Disorders
88267	Chromosomal analysis, amniotic fluid or chorionic villus, count 15 cells, 1 karyotype, with banding	
88269	Chromosomal analysis, in situ for amniotic fluid cells, count cells from 6-12 colonies, 1 karyotype, with banding	
88271	Molecular cytogenetics; DNA probe, each (e.g., FISH)	
88272	Molecular cytogenetics; chromosomal in situ hybridization, analyze 3-5 cells (e.g., For derivatives and markers)	
88273	Molecular cytogenetics; chromosomal in situ hybridization, analyze 10-30 cells (e.g., For microdeletions)	

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88274	Molecular cytogenetics; interphase in situ hybridization, analyze 25-99 cells	
88275	Molecular cytogenetics; interphase in situ hybridization, analyze 100-300 cells	
88291	Cytogenetics and molecular cytogenetics, interpretation and report	
88299	Unlisted cytogenetic study	
88360	Morphometric analysis, tumor immunohistochemistry (e.g., Her-2/neu, estrogen receptor/progesterone receptor), quantitative or semiquantitative, each antibody	
88384	Array-based evaluation of multiple molecular probes; 11 through 50 probes	
88385	Array-based evaluation of multiple molecular probes; 51 through 250 probes	
88386	Array-based evaluation of multiple molecular probes; 251 through 500 probes	
HCPCS® Codes	Description	Comments
S3800	Genetic testing for amyotrophic lateral sclerosis (ALS)	
S3835	Complete gene sequencing for cystic fibrosis genetic testing.	
S3837	Complete gene sequence analysis for hemochromatosis genetic testing	
S3843	DNA analysis of the F5 gene for susceptibility to factor V Leiden thrombophilia	
S3849	Genetic testing for Niemann-Pick Disease	
S3852	DNA analysis for APOE epsilon 4 allele for susceptibility to Alzheimer's disease	
S3853	Genetic testing for myotonic muscular dystrophy	
S3855	Genetic testing for detection of mutations in presenilin-1 gene	
S3860	Genetic testing, comprehensive cardiac ion channel analysis, for variants in 5 major cardiac ion channel genes for individuals with high index of suspicion for familial Long QT syndrome or related disorders.	
S3861	Genetic testing, sodium channel, voltage gated, type v, alpha subunit (scn5a) and variants for suspected brugada syndrome	

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S3862	Genetic testing, family-specific ion channel analysis, for blood relatives of individuals (index case) who have previously tested positive for a genetic variant or a cardiac ion channel syndrome using either one of the above test configurations or confirmed results from another laboratory	
S3865	Comprehensive gene sequence analysis for hypertrophic cardiomyopathy.	Not covered
S3866	Genetic analysis for a specific gene mutation for hypertrophic cardiomyopathy (HCM) in an individual with a known HCMO mutation in the family	Not covered
S3870	Comparative Genomic Hybridization (CGH) microarray testing for developmental delay, autism spectrum disorder and/or mental retardation	

Medical Terms

Acid Sphingomyelinase - A glycoprotein that functions as a lysosomal enzyme, the deficiency of which causes Niemann–Pick disease type A.

Alpha-1 Antitrypsin Deficiency (AATD) - Inherited metabolic disorder that results in low or no production of the protein alpha-1 antitrypsin and affects the lungs and/or liver.

Amenorrhea - Absence of a woman's monthly period not related to menopause.

Angelman Syndrome - Genetic developmental disorder characterized by mental retardation, hyperactivity, and unprovoked laughter.

Anomaly - Deviation from the norm.

Apolipoprotein E Genotype (APOE) - Gene that when mutated is associated with an individual's predisposition to Alzheimer's disease.

Ascites - An excessive accumulation of fluid in the abdominal cavity.

Ataxia - Jerky or uncoordinated movements.

Atelosteogenesis - Incomplete bone formation. A very rare inherited disorder of the skeleton characterized by disproportionate short stature, unusual facial appearance and multiple large joint dislocation with specific radiographic features.

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Atrophy - Wasting or decreasing in muscle mass.

Autism - A brain development disorder, observable in early childhood that significantly affects verbal and nonverbal communication and social interaction.

Azoospermia - Absence of live sperm in semen.

Boomerang Dysplasia - Very rare inherited disorder of the skeleton characterized by disproportionate short stature, unusual facial appearance and multiple large joint dislocation with specific radiographic features.

Bradycardia - Slow heart rate.

Bronchiectasis - Chronic inflammatory disease that affects the lungs.

Cataract - An eye disease that involves the clouding of the natural lens of the eye.

Celiac Disease - A disease of the small intestine in which a person is unable to tolerate wheat protein (gluten).

Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL) - Most common form of hereditary stroke disorder, caused by a gene mutation.

Charcot-Marie-Tooth Neuropathy - An inherited disorder of nerves characterized by loss of muscle tissue and touch sensation.

Chromosome - Cell-replicating genetic structures of the cells containing the cellular DNA that bears in its nucleotide sequence the linear array of genes.

Chromosome 22q11.2 Deletion Syndrome - A birth defect caused by an abnormality in chromosome 22 in which a small piece of the chromosome is missing. The syndrome affects the immune system. This syndrome is also referred to as DiGeorge Syndrome, Velocardiofacial Syndrome, Shprintzen Syndrome, Conotruncal Anomaly Face Syndrome, and Cayler Cardiofacial Syndrome.

Chromosome Complement - The whole set of chromosomes for the species. In humans, the chromosome complement (which is also called the karyotype) consists of 46 chromosomes.

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Cognitive - Refers to the thought processes which include thinking, learning, perception, awareness, and judgment.

Comparative Genomic Hybridization (CGH) - Technique that is used to detect chromosome gain or loss by hybridizing DNA from a target cell and a normal cell.

Congenital - Present at birth.

Corpus Callosum - Bundle of fibers connecting the two halves of the brain.

Cortical - Outer portion of an organ.

Cystic Fibrosis - An inherited disease characterized by a buildup of mucus in the lungs.

Cytoplasm - The contents of a cell except the nucleus.

Dementia - Progressive neurological condition which causes cognitive impairment.

Deoxyribonucleic Acid (DNA) - Molecule that encodes genetic information in the nucleus of cells. It determines the structure, function and behavior of the cell.

Developmental Delay - Significant lag in the development of a baby or child.

Down Syndrome - Congenital disorder caused by having an extra twenty-first chromosome; resulting in a flat face, short stature and mental retardation.

Dysarthria - A speech impairment.

Dysmorphic - Abnormality of the structure of part of the body.

Dysmyelination - Condition in which there is a much reduced amount of myelin in nervous tissue.

Dystonia - Painful involuntary muscle cramps or spasms.

Electrocardiogram - Test that records the electrical activity of the heart.

Enzyme - A protein which accelerates the rate of a chemical reaction.

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Factor V Leiden - Inherited blood clotting disorder.

First Degree Relative - Refers to the parents, siblings and children of an individual.

Fragile X Syndrome - Most common form of inherited mental retardation, characterized by developmental delay, variable levels of mental retardation, and behavioral and emotional difficulties.

Gait - Particular way of moving on foot.

Genes - Formed from DNA, carried on the chromosomes and are responsible for the inherited characteristics that distinguish one individual from another. Each human individual has an estimated 100,000 separate genes.

Genitourinary - Refers to the reproductive and urinary systems.

Genome - Complete set of genetic information contained in the DNA of an organism.

Glycoprotein - A protein coated with sugar.

Hematologic - Of or relating to blood.

Hemochromatosis - Hereditary disorder that causes your body tissues to absorb and store too much iron.

Hepatic - Referring to the liver.

Hepatosplenomegaly - Enlargement of the liver and spleen.

HFE-Associated Hemochromatosis - Inherited disorder characterized by iron overload.

Hirschsprung's Disease - Birth defect in which some nerve cells are missing in the large intestine.

Holter Monitor - Portable recorder worn to monitor heart rhythm.

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Huntington Disease - Inherited disorder that leads to an abnormal deterioration of the nervous system.

Hyperlipidemia - High blood cholesterol.

Hyperpigmentation - Abnormal darkening of the skin.

Hypertrophic Cardiomyopathy - A genetic disorder in which the heart muscle is so strong that it does not relax enough to fill with the heart with blood and so has reduced pumping ability.

Hypothalamic - Of or pertaining to the area of the brain that controls body temperature, hunger, and thirst.

Hypotonia - Decreased muscle tone.

Infarct - An area of dead tissue caused by a loss of blood supply.

Interstitial - Relating to or situated in the small, narrow spaces between tissues or parts of an organ.

Ischemic - Insufficient blood flow.

Jaundice - Yellow color of the skin due to an excess of bilirubin in the body.

Karyotype - To classify and array (the chromosome complement of an organism or a species) according to the arrangement, number, size, shape, or other characteristics of the chromosomes.

Klinefelter Syndrome - A genetic condition in which a male has extra X-chromosomes, thereby causing the development of female secondary sex characteristics.

Lacunar - Subtype of stroke that affects the deeper parts of the brain and involves the tiny perforating arteries.

Larsen Syndrome - Rare genetic disease characterized by cleft palate, flattened facies, multiple congenital joint dislocations, and deformities of the foot.

Lethargy - Lack of energy.

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Long QT Syndrome - Disorder of the heart's electrical system characterized by a delay in the time it takes for the electrical system to recharge after each heartbeat.

Lysosome - An organelle (a specialized cell) found in the cytoplasm of most cells.

Macula - A small spot.

Mesenteric - Referring to the tissue that supports the intestinal tract.

Methylenetetrahydrofolate Reductase (MTHFR) - The enzyme that reduces folic acid to its most active form, methyltetrahydrofolate.

Metachronous - Multiple separate occurrences.

Microarray - A laboratory technology to identify changes in genes or gene expression.

Microdeletion - A gene mutation in which a part of a chromosome or a sequence of DNA is missing.

Mutation - Change of the DNA 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.

Myocardial Infarction - Heart attack.

Myotonia - Difficulty relaxing muscles after contraction.

Myotonic Dystrophy - An inherited neuromuscular disorder characterized by progressive muscle weakness and wasting.

Necrotizing - Causing the death of a specific area of tissue.

Niemann-Pick Disease - An inherited disorder in which harmful amount of a fatty substance accumulates in different parts of the body.

Neonate - A newborn, typically up to four weeks of age.

Oligospermia - Low sperm count in semen.

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Panniculitis - Inflammation of subcutaneous fat.

Posterior Subcapsular Cataract - A cataract in the rear of the lens capsule.

Prader-Willi Syndrome - Genetic disorder characterized by mental retardation and obesity.

Predispositional - Any condition that tends to make an individual more prone to disease.

Presenilin-1 (PSEN 1) Gene - Gene that when mutated makes individuals more susceptible to Alzheimer's disease.

Presenilin-2 (PSEN 2) Gene - Gene that when mutated is associated with an individual's predisposition to Alzheimer's disease.

Prothrombin - A protein that is necessary for proper blood clotting.

Ribonucleic Acid (RNA) - Nucleic acid found in all living cells. It plays a role in transferring information from DNA to the protein forming system of the cell.

Second Degree Relative - Refers to the aunts, uncles, grandparents, grandchildren, nieces, nephews and half-siblings of an individual.

Spinal Muscular Atrophy - Refers to a number of different disorders, all having in common a genetic cause and the manifestation of weakness due to loss of the motor neurons of the spinal cord and brainstem.

Spondylocarpotarsal Synostosis - Rare, inherited syndrome characterized by short stature with short trunk, failure of normal spine segmentation resulting in block vertebrae and fusion of posterior elements, carpal and/or tarsal coalition, scoliosis, inward curvature of the spine, flat foot, decreased range of motion or dislocation of the elbow, kidney anomalies, and hearing loss.

Spontaneous Abortion - Miscarriage.

Subcortical - Relating to the portion of the brain immediately below the cerebral cortex.

Thrombocytopenia - Low platelet counts in the blood.

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Thrombophilia - A hereditary or acquired tendency to develop blood clots.

Thrombosis - Blood clot.

Transferrin - Protein that carries iron in the bloodstream.

Tremulous - Shaking or trembling.

Turner Syndrome - Occurs in females when one of the X-chromosomes is missing or damaged. The most common features of Turner syndrome are short stature and reduced or absent development of the ovaries. As adults, women with this disorder are typically infertile.

Vasculitis - Inflammation of a blood vessel.

Venous - Pertaining to veins.

White Matter - Tissue in the brain and spinal cord which is made up mostly of nerve fibers that carries electrical impulses from one part of the nervous system to another.

X-Chromosome - Sex chromosome that is carried by women.

Y-Chromosome - Sex chromosome that is carried by men.

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