Genetic testing: epilepsy, neurodegenerative, and neuromuscular disorders

These services may or may not be covered by your HealthPartners plan. Please see your plan documents for your specific coverage information. If there is a difference between this general information and your plan documents, your plan documents will be used to determine your coverage.

Administrative Process

Prior authorization is required for the following services:

- Comprehensive Neuromuscular Disorders Panels
- Comprehensive Ataxia Panels
- Testing for the following conditions:
  - Rett Syndrome
  - Epilepsy
  - CADASIL
  - Alzheimer Disease
  - Facioscapulohumeral Muscular Dystrophy (FSHD)
  - Inherited Peripheral Neuropathy (Charcot-Marie-Tooth and Hereditary Neuropathy with Liability to Pressure Palsies) multigene panel
  - Limb-Girdle Muscular Dystrophies (LGMD)
  - Hereditary Dystonia
  - Parkinson Disease
  - Hereditary Spastic Paraplegia
  - Congenital Myasthenic Syndrome
  - Myotonia Congenita
  - Hypokalemic Periodic Paralysis
- Genetic testing for other Covered Epilepsy, Neuromuscular, and Neurodegenerative Disorders
- Testing that is associated with a procedure code listed in “Box A”, below.

Prior authorization is not required for the following services:

- Spinal Muscular Atrophy
- Amyotrophic Lateral Sclerosis (ALS) multigene panel
- Duchenne and Becker Muscular Dystrophy
- Friedreich’s Ataxia
- Huntington Disease (HD) *HTT* repeat analysis
- Inherited Peripheral Neuropathy (Charcot-Marie-Tooth and Hereditary Neuropathy with Liability to Pressure Palsies) *PMP22* Sequencing and/or Deletion/Duplication Analysis (81324, 81325)
- Myotonic Dystrophy testing

Tests that require prior authorization will be reviewed for medical necessity of the testing as a whole. That is, a single coverage decision will apply to all of the tests, services, and/or procedure codes associated with the genetic test, whether they are requested/billed together or separately.

Box A: Genetic testing procedure codes that require prior authorization
Molecular pathology procedures, Tier 2 or unlisted (CPT 81400-81408, 81479)
Unlisted multianalyte assays (CPT 81599)
Any other listed or unlisted laboratory/pathology CPT code when it is used in association with a genetic test.

Policy Reference Table

If available, codes are listed below for informational purposes only, and do not guarantee member coverage or provider reimbursement. The list may not be all-inclusive.

Coverage Criteria Sections	Example Tests; Labs	Common CPT Codes	Common ICD Codes
Comprehensive Neuromuscular Disorders Panel			

Comprehensive Neuromuscular Disorders Panel	Comprehensive Neuromuscular Panel (PreventionGenetics, part of Exact Sciences)	81161, 81404, 81405, 81406, 81479	G12, G13, G23-G26, G31, G32, G36, G37
	Comprehensive Neuromuscular Disorders Panel (Invitae)		
	Neuromuscular Disorders Panel (GeneDx)		
Comprehensive Ataxia Panel			
Comprehensive Ataxia Panel	Genomic Unity Ataxia Repeat Expansion Analysis (Variantyx, Inc.)	0216U	G11.1, G11.19, G11.8, G11.9, Z82.0
	Genomic Unity Comprehensive Ataxia Analysis (Variantyx, Inc.)	0217U	
	Ataxia Xpanded Panel (GeneDx)	81185, 81189, 81286, 81403, 81404, 81479	
	Ataxia Panel (Blueprint Genetics)		
Spinal Muscular Atrophy			
SMN1 Sequencing and/or Deletion/Duplication Analysis	Spinal Muscular Atrophy (SMA), Diagnostic (Quest Diagnostics)	81329	G12, Z84.81
	SMN1 Sequencing Analysis (Fulgent Genetics)	81336, 81405	
	Genomic Unity SMN1/2 Analysis (Variantyx Inc.)	0236U	
SMN2 Deletion/Duplication Analysis	SMN2 Deletion/Duplication (GeneDx)	81401	
Rett Syndrome			
MECP2 Sequencing and/or Deletion/Duplication Analysis	MECP2 Full Gene Sequencing and Deletion/Duplication (Invitae)	81302, 81304	F70-F79, F80, F81, F82, F84, F88, F89, Z13.4, Z82.79, Z84
	MECP2 Gene Sequencing & Del/Dup (GeneDx)		
	Genomic Unity MECP2 Analysis (Variantyx, Inc.)	0234U	
Epilepsy			
Epilepsy Multigene Panel	Comprehensive Epilepsy Panel (Blueprint Genetics)	81185, 81189, 81302, 81406, 81419, 81479	G40.001- G40.919
	Comprehensive Epilepsy Panel (GeneDx)		
	Clinical Epilepsy NGS Panel (LabCorp)		
	EpilepsyNext (Ambry Genetics)		
	Epilepsy Panel (Invitae)		
CADASIL			
NOTCH3 Sequencing and/or Deletion/Duplication Analysis	NOTCH3 Full Gene Sequencing and Deletion/Duplication (Invitae)	81406, 81479	I67.850, F02.80, F02.81
Alzheimer Disease			
PSEN1, *PSEN2*, and *APP* Sequencing and/or Deletion/Duplication Analysis	PSEN1 Full Gene Sequencing and Deletion/Duplication (Invitae)	81405, 81479	F03, G30, G31.1, R41.0, R41.81, Z13.858, Z82.0, Z84.81
	Alzheimer's Disease, Familial via the PSEN2 Gene (PreventionGenetics, part of Exact Sciences)	81406, 81479	
	APP Full Gene Sequencing and Deletion/Duplication (Invitae)	81406, 81479	

	Alzheimer's Disease, Familial, Panel (PreventionGenetics, part of Exact Sciences)	81405, 81406, 81479	
	Hereditary Alzheimer's Disease Panel (Invitae)		
APOE Variant Analysis for Alzheimer's Disease	APOE Single Gene Test (Blueprint Genetics)	81401, 81479, S3852	
Amyotrophic Lateral Sclerosis (ALS)			
Amyotrophic Lateral Sclerosis (ALS) Multigene Panel	Amyotrophic Lateral Sclerosis (ALS) Panel (PreventionGenetics, part of Exact Sciences)	81179, 81403, 81404, 81405, 81406, 81407, 81479, S3800	G12.21
	Amyotrophic Lateral Sclerosis Panel (Invitae)		
Duchenne and Becker Muscular Dystrophy			
Diagnostic DMD Sequencing and/or Deletion/Duplication Analysis	Dystrophinopathies Test (Invitae)	81161	G71.01, R62.59, Z84.81
	Duchenne/Becker MD (DMD) Gene Sequencing (GeneDx)	81408	
	Genomic Unity DMD Gene Analysis (Variantyx)	0218U	
Facioscapulohumeral Muscular Dystrophy (FSHD)			
D4Z4 or Haplotype Analysis, and/or *SMCHD1* and *DNMT3B* Sequencing and/or Deletion/Duplication Analysis or Multigene Panel	*FSHD1* Southern Blot Test (Quest Diagnostics)	81404	G71.02, Z84.81
	Facioscapulohumeral Muscular Dystrophy 2 via the SMCHD1 Gene (PreventionGenetics, part of Exact Sciences)	81479	
	DNMT3B Full Gene Sequencing And Deletion/Duplication (Invitae)		
	FSHD-(FSHD1 & FSHD2) Detection of Abnormal Alleles with Interpretation (University of Iowa Hospitals and Clinics - Department of Pathology)	81404, 81479	
Friedreich's Ataxia			
FXN Repeat Analysis and/or Sequencing Analysis	Friedreich Ataxia (FXN) Repeat Expansion Test (Athena Diagnostics)	81284, 81285	G11, Z84.81
	Friedreich Ataxia (FXN) DNA Sequencing Test (Athena Diagnostics)	81286, 81404	
	Genomic Unity FXN Analysis (Variantyx Inc)	0233U	
Huntington Disease (HD)			
HTT Repeat Analysis	Huntington Disease (HTT) Genetic Testing (Repeat Expansion) (LabCorp)	81271, 81274	G10, Z84.81
Inherited Peripheral Neuropathy (Charcot-Marie-Tooth and Hereditary Neuropathy with Liability to Pressure Palsies)			
PMP22 Sequencing and/or Deletion/Duplication Analysis or Multigene Panel	Deletion/Duplication (PMP22) (GeneDx)	81324	G60.0, G60.8, G60.9
	PMP22 DNA Sequencing Test (Quest Diagnostics)	81325	

	Charcot-Marie Tooth (CMT) - Comprehensive Panel (PreventionGenetics, part of Exact Sciences)	81448	
	Charcot-Marie-Tooth Disease NGS Panel (HNL Lab Medicine)		
Limb-Girdle Muscular Dystrophies (LGMD)			
Limb Girdle Muscular Dystrophy Multigene Panel	Limb-Girdle Muscular Dystrophy Panel (GeneDx)	81405, 81406, 81408, 81479	G71.0, Z13.71, Z82.0, Z84.81
	Limb-Girdle Muscular Dystrophy Panel (Invitae)		
Myotonic Dystrophy			
DMPK and/or *CNBP* (ZNF9) Repeat Analysis	Myotonic Dystrophy 1 (DMPK) Genetic Testing (Repeat Expansion) (LabCorp)	81234, 81239, 81401, 81404, S3853	G71.11, Z84.81
	Myotonic Dystrophy 2 (ZNF9 / CNBP) Genetic Testing (Repeat Expansion) (LabCorp)	81187, S3853	
Hereditary Dystonia			
Hereditary Dystonia Multigene Panel	Dystonia Panel (GeneDx)	81404, 81405, 81406, 81407, 81408, 81479	G24.1, G24.9
	Dystonia Panel (PreventionGenetics, part of Exact Sciences)		
	Dystonia Comprehensive Panel (Invitae)		
Parkinson Disease			
Parkinson Disease Multigene Panel	Parkinson Disease Panel (BluePrint Genetics)	81479	G20
	Parkinson Disease Panel (GeneDx)		
	Invitae Parkinson Disease and Parkinsonism Panel (Invitae)		
Hereditary Spastic Paraplegia			
Hereditary Spastic Paraplegia Multigene Panel	Spastic Paraplegia Panel (Blueprint Genetics)	81448	G11.4, G82.2
	Hereditary Spastic Paraplegia Comprehensive Panel (Invitae)		
Congenital Myasthenic Syndrome			
Congenital Myasthenic Syndromes Multigene Panel	Congenital Myasthenic Syndrome Panel (PreventionGenetics, part of Exact Sciences)	81406, 81407, 81479	G70.2
	Congenital Myasthenic Syndrome Panel (Invitae)		
Myotonia Congenita			
CLCN1 Sequencing and/or Deletion/Duplication Analysis	Myotonia Congenita via the *CLCN1* gene (PreventionGenetics, part of Exact Sciences)	81406, 81479	G71.12
	CLCN1 Full Gene Sequencing and Deletion/Duplication (Invitae)		
Hypokalemic Periodic Paralysis			
CACNA1S and *SCN4A* Sequencing and/or	*CACNA1S* Full Gene Sequencing and/or Deletion/Duplication (Invitae)	81406, 81479	E87.6, G72.3

Deletion/Duplication Analysis, or Periodic Paralysis Multigene Panel	*SCN4A* Full Gene Sequencing and/or Deletion/Duplication (Invitae)		
Other Covered Epilepsy, Neurodegenerative, and Neuromuscular Disorders			
Other Covered Epilepsy, Neuromuscular, and Neurodegenerative Disorders	See list below	81400, 81401, 81402, 81403, 81404, 81405, 81406, 81407, 81408, 81479	

Coverage

Comprehensive Neuromuscular Disorders Panel

1. Comprehensive neuromuscular panel analysis to establish a genetic diagnosis for a neuromuscular disorder is considered **medically necessary** when:
   - A. The member meets either of the following:
     - i. The member is a neonate and displays at least one of the following:
       - a) Respiratory insufficiency, with sudden episodic apnea and cyanosis, **or**
       - b) Joint contractures (e.g., arthrogryposis multiplex congenita), **or**
       - c) Stridor, **or**
       - d) Feeding difficulties, **or**
       - e) Poor suck/cry, **or**
       - f) Choking spells, **or**
       - g) Facial, bulbar, or generalized weakness, **or**
     - ii. The member is any age and displays at least one of the following:
       - a) Abnormal muscle fatigability/weakness, **or**
       - b) Delayed motor milestones, **or**
       - c) Eyelid ptosis or extraocular muscle weakness, **or**
       - d) Facial and bulbar weakness with nasal speech and difficulties in coughing and swallowing, **or**
       - e) Spinal deformity or muscle atrophy, **or**
       - f) Abnormal electromyography (EMG) testing showing a defect in neuromuscular transmission, **or**
       - g) Elevated serum creatine kinase levels, **and**
   - B. The member meets one of the following:
     - i. The member's presentation is not consistent with a neuromuscular disorder for which targeted or single-gene analysis (e.g., *SMN1, DMD, PMP22*) is appropriate, **or**
     - ii. The member underwent targeted or single-gene analysis for a neuromuscular disorder (e.g., *SMN1, DMD, PMP22*) and the results were non-diagnostic.
2. Comprehensive neuromuscular panel analysis to establish a genetic diagnosis for a neuromuscular disorder is considered **investigational** for all other indications.

Back to top

Comprehensive Ataxia Panel

1. Comprehensive ataxia panel analysis to establish a genetic diagnosis of an ataxia is considered **medically necessary** when:
   - A. The member displays one or more of the following:
     - i. Poorly coordinated gait and finger/hand movements, **or**
     - ii. Weakness of the eye muscles (ophthalmoplegia), **or**
     - iii. Dysarthria, **or**
     - iv. Eye movement abnormalities (nystagmus, abnormal saccade movements), **and**
   - B. Non-genetic causes of ataxia have been ruled out (e.g., alcoholism, vitamin deficiencies, multiple sclerosis, vascular disease, primary or metastatic tumors, and paraneoplastic disease associated with occult carcinoma of the ovary, breast, or lung, and spinal muscular atrophy).
2. Comprehensive ataxia panel analysis to establish a genetic diagnosis of an ataxia is considered **investigational** for all other indications.

Back to top

Spinal Muscular Atrophy

SMN1 Sequencing and/or Deletion/Duplication Analysis

1. *SMN1* sequencing and/or deletion/duplication analysis to establish or confirm a diagnosis of spinal muscular atrophy (SMA) is considered **medically necessary** when:

A. The member has a positive newborn screen for SMA, **or**
B. The member has any of the following:
i. History of motor difficulties, especially with loss of skills, **or**
ii. Muscle weakness, especially proximal muscles, **or**
iii. Hypotonia, **or**
iv. Areflexia/hyporeflexia, **or**
v. Tongue fasciculations, **or**
vi. Hand tremor, **or**
vii. Recurrent lower respiratory tract infections or severe bronchiolitis in the first few months of life, **or**
viii. Evidence of motor unit disease on electromyogram.

2. *SMN1* sequencing and/or deletion/duplication analysis to establish or confirm a diagnosis of spinal muscular atrophy (SMA) is considered **investigational** for all other indications.

Back to top

SMN2 Deletion/Duplication Analysis

1. *SMN2* deletion/duplication analysis is considered **medically necessary** when:
A. The member has a diagnosis of spinal muscular atrophy.
2. *SMN2* deletion/duplication analysis is considered **investigational** for all other indications.

Back to top

Rett Syndrome

MECP2 Sequencing and/or Deletion/Duplication Analysis

1. *MECP2* sequencing and/or deletion/duplication analysis to establish or confirm a diagnosis of Rett syndrome is considered **medically necessary** when:
A. The member experienced a period of developmental regression (range: ages 1-4 years) followed by recovery or stabilization (range: ages 2-10 years), **and**
B. The member has at least one of the following:
i. Partial or complete loss of acquired purposeful hand skills, **or**
ii. Partial or complete loss of acquired spoken language or language skill (e.g., babble), **or**
iii. Gait abnormalities: impaired (dyspraxic) or absence of ability, **or**
iv. Stereotypic hand movements including hand wringing/squeezing, clapping/tapping, mouthing, and washing/rubbing automatisms, **and**
C. The member does **not** have either of the following:
i. Brain injury secondary to peri- or postnatal trauma, neurometabolic disease, or severe infection that causes neurological problems, **or**
ii. Grossly abnormal psychomotor development in the first six months of life, with early milestones not being met.
2. *MECP2* sequencing and/or deletion/duplication analysis to establish or confirm a diagnosis of Rett syndrome is considered **investigational** for all other indications.

Back to top

Epilepsy

Epilepsy Multigene Panel

1. The use of an epilepsy multigene panel is considered **medically necessary** when:
A. The member has a history of unexplained epilepsy (i.e., seizures not caused by acquired etiology such as trauma, infection, structural brain abnormality, and/or stroke).
2. The use of an epilepsy multigene panel is considered **investigational** for all other indications.

Back to top

CADASIL

NOTCH3 Sequencing and/or Deletion/Duplication Analysis

1. *NOTCH3* sequencing and/or deletion/duplication analysis to establish or confirm a diagnosis of CADASIL (cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy) is considered **medically necessary** when:
A. The member has a family history of stroke and/or vascular dementia consistent with an autosomal dominant pattern of inheritance, **or**
B. The member has at least one of the following clinical features of CADASIL:
i. Transient ischemic attacks and ischemic stroke, **or**
ii. Cognitive impairment, manifesting initially with executive dysfunction, with a concurrent stepwise deterioration due to recurrent strokes to vascular dementia, **or**
iii. Migraine with aura (mean age of onset of 30 years), **or**
iv. Psychiatric disturbances, most frequently mood disturbances and apathy, **or**
C. The member has at least one of the following brain imaging findings of CADASIL:

i. Symmetric and progressive white matter hyperintensities, often involving the anterior temporal lobes and external capsules, **or**
ii. Lacunes of presumed vascular origin, **or**
iii. Recent subcortical infarcts, **or**
iv. Dilated perivascular spaces, sometimes referred to as subcortical lacunar lesions, **or**
v. Brain atrophy, **or**
vi. Cerebral microbleeds.

2. *NOTCH3* sequencing and/or deletion/duplication analysis to establish or confirm a diagnosis of CADASIL (cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy) is considered **investigational** for all other indications.

Back to top

Alzheimer Disease

PSEN1, *PSEN2*, and *APP* Sequencing and/or Deletion/Duplication Analysis

1. *PSEN1*, *PSEN2*, and/or *APP* sequencing and/or deletion/duplication analysis to establish a diagnosis or determine future risk to develop early-onset Alzheimer disease (diagnosed before age 65 years) is considered **medically necessary** when:
   A. The member is 18 years of age or older, **and**
   B. The member is asymptomatic*, **and**
      i. Has a family history of dementia that is consistent with an autosomal dominant pattern of inheritance, **and**
         a) Has at least one relative with a history of early-onset Alzheimer disease (diagnosed before age 65 years), **or**
   C. The member is symptomatic with dementia, **and**
      i. Was diagnosed with dementia at 65 years of age or younger, **and**
         a) Has a close relative diagnosed with dementia, **or**
         b) Has an unknown family history (e.g., adoption), **or**
      ii. Was diagnosed with dementia at 66 years of age or older, **and**
         a) Has a family history of dementia that is consistent with an autosomal dominant pattern of inheritance, **and**
         b) Has at least one close relative who was diagnosed with dementia at 65 years of age or younger.
2. Genetic testing for Alzheimer's disease via other genes is considered **investigational.****
3. *PSEN1*, *PSEN2*, and/or *APP* sequencing and/or deletion/duplication analysis to establish the diagnosis or determine future risk to develop early-onset Alzheimer disease (diagnosed before age 65 years) is considered **investigational** for all other indications.

* Predictive testing should only be performed in the setting and context of thorough pre- and post-test counseling
**Please see clinical guidelines "*APOE* Variant Analysis for Alzheimer's Disease" for coverage criteria for *APOE* testing

Back to top

APOE Variant Analysis for Alzheimer's Disease

1. *APOE* variant analysis is considered **medically necessary** when:
   A. The member has a diagnosis of Alzheimer's disease, **and**
   B. The member is being evaluated for suitability of treatment with monoclonal antibodies directed against aggregated forms of beta amyloid (such as Leqembi or Kisunla).
2. *APOE* variant analysis is considered **investigational** for all other indications

Back to top

Amyotrophic Lateral Sclerosis (ALS)

Amyotrophic Lateral Sclerosis (ALS) Multigene Panel

1. Multigene panel analysis to establish a genetic etiology of amyotrophic lateral sclerosis (ALS) is considered **medically necessary** when:
   A. The member displays all of the following:
      i. Evidence of lower motor neuron (LMN) degeneration, **and**
      ii. Evidence of upper motor neuron (UMN) degeneration, **and**
      iii. Progressive spread of symptoms, **and**
      iv. No evidence of other disease processes that could explain the LMN and UMN degeneration, **and**
   B. The panel includes, at a minimum, the following genes: *C9orf72*, *SOD1*, *FUS*, and *TARDBP*.
2. Multigene panel analysis to establish a genetic etiology of amyotrophic lateral sclerosis (ALS) is considered **investigational** for all other indications.

Back to top

Duchenne and Becker Muscular Dystrophy

Diagnostic *DMD* Sequencing and/or Deletion/Duplication Analysis

1. *DMD* sequencing and/or deletion/duplication analysis to establish or confirm a diagnosis of Duchenne muscular dystrophy (DMD) or Becker muscular dystrophy (BMD) is considered **medically necessary** when:
   - A. The member has all of the following clinical characteristics of DMD:
     - i. Progressive symmetric muscular weakness - proximal greater than distal, often with calf hypertrophy (enlargement), **and**
     - ii. Symptoms presenting before age five years, **and**
     - iii. Wheelchair dependency before age 13 years, **and**
     - iv. Elevated serum creatine kinase concentration, typically more than 10 times the normal levels, **or**
   - B. The member has all of the following clinical characteristics of BMD:
     - i. Elevated serum creatine kinase concentration, typically more than 5 times the normal levels, **and**
       - a) At least one of the following:
         - (a) Progressive symmetric muscle weakness (proximal more so than distal) often with calf hypertrophy (weakness of quadriceps femoris in some cases the only sign), **or**
         - (b) Activity-induced cramping, **or**
         - (c) Flexion contractures of the elbows, **or**
         - (d) Wheelchair dependency after age 16 years, **or**
         - (e) Preservation of neck flexor muscle strength, **or**
   - C. The member is asymptomatic (male or female), **and**
     - i. Has a biological sibling with a clinical diagnosis of Duchenne or Becker muscular dystrophy, **or**
     - ii. Has a biological mother that is an obligate carrier for Duchenne or Becker muscular dystrophy, **or**
   - D. The member is an asymptomatic female, **and**
     - i. Has a first or second-degree relative with a clinical diagnosis of Duchenne or Becker muscular dystrophy.
2. *DMD* sequencing and/or deletion/duplication analysis to establish a diagnosis of Duchenne muscular dystrophy (DMD) or Becker muscular dystrophy (BMD) is considered **investigational** for all other indications.

Back to top

Facioscapulohumeral Muscular Dystrophy (FSHD)

D4Z4 Haplotype Analysis, and/or *SMCHD1* and *DNMT3B* Sequencing and/or Deletion/Duplication Analysis or Multigene Panel

1. D4Z4 haplotype analysis, and/or *SMCHD1* and *DNMT3B* sequencing and/or deletion/duplication analysis or multigene panel analysis to establish or confirm a diagnosis of facioscapulohumeral muscular dystrophy is considered **medically necessary** when:
   - A. The member displays any of the following:
     - i. Weakness (which is often asymmetric) that predominantly involves the facial, scapular stabilizer, or foot dorsiflexor muscles without associated ocular or bulbar muscle weakness, **or**
     - ii. Progression of weakness after pregnancy, **or**
     - iii. Prior diagnosis of inflammatory myopathy that was refractory to immunosuppression.
2. D4Z4 haplotype analysis, and/or *SMCHD1* and *DNMT3B* sequencing and/or deletion/duplication analysis or multigene panel analysis to establish or confirm a diagnosis of facioscapulohumeral muscular dystrophy is considered **investigational** for all other indications.

Back to top

Friedreich's Ataxia

***FXN* Repeat Analysis and/or Sequencing Analysis**

1. *FXN* repeat analysis and/or sequencing analysis to establish or confirm a diagnosis of Friedreich's Ataxia is considered **medically necessary** when:
   - A. The member is symptomatic, **and**
     - i. The member has at least two of the following:
       - a) Progressive ataxia of the gait and limbs (e.g., cerebellar ataxia), **or**
       - b) Dysarthria, **or**
       - c) Decrease in/loss of position sense and/or vibration sense in lower limbs, **or**

d) Pyramidal weakness of the legs, **or**
e) Extensor plantar responses/Babinksi signs, **or**
f) Muscle weakness, **or**
g) Scoliosis, **or**
h) Pes cavus (flat feet), **or**
i) Hypertrophic nonobstructive cardiomyopathy, **or**
j) Glucose intolerance or diabetes mellitus, **or**
k) Optic atrophy and/or deafness, **and**

ii. Non-genetic causes of ataxia have been ruled out (e.g., alcoholism, vitamin deficiencies, multiple sclerosis, vascular disease, tumors), **or**

B. The member is asymptomatic*, **and**

i. Has a biological sibling with Friedreich's ataxia.

2. *FXN* repeat analysis and/or sequencing analysis to establish or confirm a diagnosis of Friedreich's Ataxia is considered **investigational** for all other indications.

* Predictive testing should only be performed in the setting and context of thorough pre- and post-test counseling

Back to top

Huntington's Disease

HTT Repeat Analysis

1. *HTT* repeat analysis to establish a diagnosis or for predictive testing of Huntington's disease (HD) is considered **medically necessary** when:

A. The member displays clinical features of Huntington's disease (i.e., progressive motor disability featuring chorea, where voluntary movement may also be affected), **or**
B. The member has a clinical diagnosis of Huntington's disease, **or**
C. The member is undergoing predictive testing*, **and**

i. The member is presymptomatic/asymptomatic, **and**
ii. The member is 18 years of age or older, **and**

a) The member has a close relative with CAG trinucleotide repeat expansion of 27 or more in *HTT*, **or**
b) The member has a first-degree relative with a clinical diagnosis of HD without prior genetic testing.

2. *HTT* repeat analysis to establish a diagnosis or for predictive testing of Huntington's disease (HD) is considered **investigational** for all other indications.

* Predictive testing should only be performed in the setting and context of thorough pre- and post-test counseling.

Back to top

Inherited Peripheral Neuropathies (examples: Charcot-Marie-Tooth Disease and Hereditary Neuropathy with Liability to Pressure Palsies)

PMP22 Sequencing and/or Deletion/Duplication Analysis or Multigene Panel

1. *PMP22* sequencing and/or deletion/duplication analysis or multigene panel analysis to establish a genetic diagnosis of an inherited peripheral neuropathy is considered **medically necessary** when:

A. The member displays one or more of the following:

i. Distal muscle weakness and atrophy, **or**
ii. Pes cavus foot deformity, **or**
iii. Weak ankle dorsiflexion, **or**
iv. Depressed tendon reflexes, **or**
v. Recurrent acute focal sensory and motor neuropathies mainly at entrapment sites, **or**
vi. Painless nerve palsy after minor trauma or compression, **or**
vii. Evidence on physical examination of previous nerve palsy such as focal weakness, atrophy, or sensory loss, **or**
viii. Complete spontaneous recovery from neuropathies.

2. *PMP22* sequencing and/or deletion/duplication analysis or multigene panel analysis to establish a genetic diagnosis of an inherited peripheral neuropathy is considered **investigational** for all other indications.

Back to top

Limb-Girdle Muscular Dystrophy (LGMD)

Limb-girdle Muscular Dystrophy Multigene Panel

1. Multigene panel analysis to establish a diagnosis of limb-girdle muscular dystrophy is considered **medically necessary** when:

A. The member displays slowly progressive, symmetrical weakness, **and**
B. The member has any of the following features:

i. Limb-girdle pattern of weakness affecting proximal muscles of the arms and legs, **or**

ii. Scapuloperoneal weakness, **or**
iii. Distal weakness, **or**
iv. Elevated serum creatine kinase levels, **or**

C. The member is asymptomatic, **and**
i. The member has a close relative diagnosed with limb-girdle muscular dystrophy whose genetic status is unavailable.

2. Multigene panel analysis to establish a diagnosis of limb-girdle muscular dystrophy is considered **investigational** for all other indications.

Back to top

Myotonic Dystrophy

DMPK and/or *CNBP* (*ZNF9*) Repeat Analysis

1. *DMPK* repeat analysis and/or *CNBP* repeat analysis to establish a diagnosis of myotonic dystrophy is considered **medically necessary** when:
A. The member meets either of the following:
i. The member is a neonate with two or more of the following:
a) Hypotonia, **or**
b) Facial muscle weakness, **or**
c) Generalized weakness, **or**
d) Positional malformations, including clubfoot, **or**
e) Respiratory insufficiency, **or**
ii. The member is any age and displays any one of the following:
a) Muscle weakness, especially of the distal leg, hand, neck, and face, **or**
b) Myotonia, which often manifests as the inability to quickly release a hand grip (grip myotonia), **or**
c) Posterior subcapsular cataracts, **or**
d) Cardiac conduction defects or progressive cardiomyopathy, **or**
e) Insulin resistance, **or**
f) Hypogammaglobulinemia, **or**
B. The member is asymptomatic, **and**
i. The member is 18 years of age or older, **and**
ii. The member has a first-degree relative with myotonic dystrophy type 1 or 2.
2. *DMPK* repeat analysis and *CNBP* repeat analysis to establish a diagnosis of myotonic dystrophy is considered **investigational** for all other indications.

Back to top

Hereditary Dystonia

Hereditary Dystonia Multigene Panel

1. Multigene panel analysis to establish a genetic diagnosis of hereditary dystonia is considered **medically necessary** when:
A. The member has a clinical presentation consistent with dystonia or patterns of abnormal, repetitive, dystonic movements.
2. Multigene panel analysis to establish a genetic diagnosis of hereditary dystonia is considered **investigational** for all other indications.

Back to top

Parkinson Disease

Parkinson Disease Multigene Panel

1. Multigene panel testing to establish a genetic diagnosis of Parkinson disease is considered **medically necessary** when:
A. The member has a clinical diagnosis of Parkinson disease, **and**
B. The member has a family history of Parkinson disease.
2. Multigene panel testing to establish a genetic diagnosis of Parkinson disease is considered **investigational** for all other indications.

Back to top

Hereditary Spastic Paraplegia

Hereditary Spastic Paraplegia Multigene Panel

1. Multigene panel analysis to establish a genetic diagnosis of hereditary spastic paraplegia is considered **medically necessary** when:
A. The member has any of the following:
i. Lower-extremity spasticity especially in hamstrings, quadriceps, adductors, and gastrocnemius-soleus muscles, **or**
ii. Weakness especially in the iliopsoas, hamstring, and tibialis anterior, **or**
iii. Lower-extremity hyperreflexia and extensor plantar responses, **or**
iv. Mildly impaired vibration sensation in the distal lower extremities.

2. Multigene panel analysis to establish a genetic diagnosis of hereditary spastic paraplegia is considered **investigational** for all other indications.

Back to top

Congenital Myasthenic Syndromes

Congenital Myasthenic Syndromes Multigene Panel

1. Multigene panel analysis to establish a genetic diagnosis of congenital myasthenic syndromes is considered **medically necessary** when:
   - A. The member has any of the following:
     - i. Neonatal respiratory insufficiency, with sudden episodic apnea and cyanosis, **or**
     - ii. Neonatal joint contractures (e.g., arthrogryposis multiplex congenita), **or**
     - iii. Stridor, feeding difficulties, poor suck/cry, choking spells, eyelid ptosis, and/or facial, bulbar, or generalized weakness in neonates, **or**
     - iv. Abnormal muscle fatigability/weakness, **or**
     - v. Delayed motor milestones, **or**
     - vi. Eyelid ptosis or extraocular muscle weakness, **or**
     - vii. Facial and bulbar weakness with nasal speech and difficulties in coughing and swallowing, **or**
     - viii. Spinal deformity or muscle atrophy, **or**
     - ix. Abnormal electromyography (EMG) testing showing a defect in neuromuscular transmission.
2. Multigene panel analysis to establish a genetic diagnosis of congenital myasthenic syndromes is considered **investigational** for all other indications.

Back to top

Myotonia Congenita

CLCN1 Sequencing and/or Deletion/Duplication Analysis

1. *CLCN1* sequencing and/or deletion/duplication analysis to establish a genetic diagnosis of myotonia congenita is considered **medically necessary** when:
   - A. The member has any of the following:
     - i. Episodes of muscle stiffness (myotonia) or cramps beginning in early childhood that are alleviated by brief exercise, **or**
     - ii. Myotonic contraction is elicited by percussion of muscles, **or**
     - iii. Electromyography (EMG) performed with needle electrodes discloses characteristic showers of spontaneous electrical activity (myotonic bursts).
2. *CLCN1* sequencing and/or deletion/duplication analysis to establish a genetic diagnosis of myotonia congenita is considered **investigational** for all other indications.

Back to top

Hypokalemic Periodic Paralysis

CACNA1S and *SCN4A* Sequencing and/or Deletion/Duplication Analysis, or Periodic Paralysis Multigene Panel

1. *CACNA1S* and *SCN4A* sequencing and/or deletion/duplication analysis, or Periodic Paralysis Multigene Panel to establish a genetic diagnosis of periodic paralysis is considered **medically necessary** when:
   - A. Alternative causes of hypokalemia have been excluded (e.g., renal, adrenal, thyroid dysfunction; renal tubular acidosis; diuretic and laxative abuse), **and**
   - B. The member has had two or more attacks of muscle weakness with documented serum potassium less than 3.5 mEq/L, **or**
   - C. The member has had one attack of muscle weakness, **and**
     - i. Has a close relative who has had one attack of muscle weakness with documented serum potassium less than 3.5 mEq/L, **or**
   - D. The member has three or more of the following features:
     - i. Onset of symptoms in the first or second decade, **or**
     - ii. Muscle weakness involving at least 1 limb lasting longer than two hours, **or**
     - iii. The presence of triggers (previous carbohydrate rich meal, symptom onset during rest after exercise, stress), **or**
     - iv. Improvement in symptoms with potassium intake, **or**
     - v. A family history of a clinical or genetic diagnosis of hypokalemic periodic paralysis in a close relative, **or**
     - vi. Positive long exercise test**.**
2. *CACNA1S* and *SCN4A* sequencing and/or deletion/duplication analysis, or Periodic Paralysis Multigene Panel to establish a genetic diagnosis of periodic paralysis is considered **investigational** for all other indications.

Back to top

Other Covered Epilepsy, Neuromuscular, and Neurodegenerative Disorders

1. Genetic testing to establish or confirm one of the following epilepsy, neuromuscular, and neurodegenerative conditions to guide management is considered **medically necessary** when the member demonstrates clinical features* consistent with the disorder (the list is not meant to be comprehensive, see 2. below):
    A. AADC deficiency
    B. Hereditary Transthyretin Amyloidosis
    C. X-linked Adrenoleukodystrophy
    D. L1 Syndrome
    E. SCN9A Neuropathic Pain Syndromes
    F. Cerebral Cavernous Malformation, Familial
    G. STAC3 Disorder
2. Genetic testing to establish or confirm the diagnosis of all other epilepsy, neurodegenerative, and neuromuscular disorders not specifically discussed within this or another medical policy will be evaluated by the criteria outlined in **General Approach to Genetic and Molecular Testing** (see policy for coverage criteria).

*Clinical features for a specific disorder may be outlined in resources such as GeneReviews, OMIM, National Library of Medicine, Genetics Home Reference, or other scholarly source.

Definitions

Close relatives include first, second, and third degree blood relatives on the same side of the family:

- **First-degree relatives** are parents, siblings, and children
- **Second-degree relatives** are grandparents, aunts, uncles, nieces, nephews, grandchildren, and half siblings
- **Third-degree relatives** are great grandparents, great aunts, great uncles, great grandchildren, and first cousins

Early-onset Alzheimer disease is defined as Alzheimer disease occurring in an individual under age 65

Childhood is the period of development until the 18th birthday.

Myotonia is defined as impaired relaxation of skeletal muscle after voluntary contraction.

Autosomal dominant inheritance patterns are generally characterized by the following traits*:

- There are individuals with the condition in multiple generations of a family
- Individuals who do not have the condition do not have children with the condition
- Individuals with the condition have a parent with the condition

*Factors such as incomplete penetrance (when not all individuals with a genetic variant develop symptoms) and variable expressivity (when symptoms/signs or severity of the condition vary from person to person) can complicate the identification of this pattern of inheritance.

Products

This information is for most, but not all, HealthPartners plans. Please read your plan documents to see if your plan has limits or will not cover some items. If there is a difference between this general information and your plan documents, your plan documents will be used to determine your coverage. These coverage criteria do not apply to Medicare Products. For more information regarding Medicare coverage criteria or for a copy of a Medicare coverage policy, contact Member Services at 952-883-7272 or 1-877-778-8384.

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