Coverage of any medical intervention discussed in a WellFirst Health medical policy is subject to the limitations and exclusions outlined in the member's benefit certificate or policy and to applicable state and/or federal laws.

Genetic Testing for Neurologic Disorders MP9497

Covered Service: Yes

Prior Authorization Required: Yes

Additional Information: Genetic testing is covered for a WellFirst Health member if the test results provide a direct medical benefit or guides reproductive decision-making for the WellFirst Health member. See Genetic Testing MP9012 for additional information.

Pre- and post-test genetic counseling is required for any individual undergoing genetic testing for neurological disorders.

A first-degree relative is defined as an individual's parents, full siblings, and children.

A second-degree relative is defined as an individual's grandparents, grandchildren, aunts, uncles, nephews, nieces and half-siblings.

*Axonal neuropathy indicates etiology is related to diabetes, toxic medications, or thyroid disease.

Reproductive carrier screening (prenatal testing) does not require prior authorization and is addressed per MP9477

WellFirst Health Medical Policy:

1.0 **Genetic Testing** for hereditary neurologic disorders **requires** prior authorization through the Health Services Division and is considered medically necessary and must meet **ALL** of the following criteria:

1.1 The member displays clinical features, or is at direct risk of inheriting the mutation in question (pre-symptomatic); **AND;**

1.2 The result of the test will directly impact the treatment being delivered to the member; **AND;**

1.3 After history, physical examination, pedigree analysis, genetic counseling, and completion of conventional diagnostic studies a definitive diagnosis remains

uncertain or identification of a genetic mutation will guide reproductive decision making.

2.0 Please reference the following link for specific criteria requirements for:

2.1 **Spinal Muscular Atrophy** - Genetic Testing for Reproductive Carrier Screening and Prenatal Care MP9477

3.0 **Alzheimer Disease (Early Onset) APP, PSEN1, PSEN2** genetic testing **requires** prior authorization through the Health Services Division and is considered medically necessary for the diagnosis or screening for Alzheimer disease if **1 or more** of the following are met:

3.1 Early-onset familial Alzheimer disease, suspected, as indicated by **ALL** of the following:

3.1.1 Dementia diagnosed in patient 60 years or younger and has **1 or more** of the following:

3.1.1.1 Documented mutation of PSEN1, PSEN2, or APP gene in relative; **OR;**

3.1.1.2 Family history of dementia; **OR;**

3.1.1.3 Family history of dementia is unable to be determined (e.g. adoption)

3.1.2 Reversible causes of dementia have been excluded by clinical examination, neuroimaging, and laboratory testing.

3.2 Predictive testing for at-risk asymptomatic adult if **BOTH** of the following are met:

3.2.1 Disease causing mutation in PSEN1, PSEN2, or APP gene has been identified in affected first-degree or second-degree relative; **AND;**

3.2.2 Testing is preceded by baseline neurologic examination.

4.0 **Ataxia – Telangiectasia ATM** genetic testing **requires** prior authorization through the Health Services Division and is considered medically necessary when **BOTH** of the following is present:

4.1 Acquired causes of ataxia have been ruled out; **AND;**

4.2 Diagnosis or screening for ataxia-telangiectasia, as indicated by **1 or more** of the following:

4.2.1 ATM protein or ATM protein kinase activity equivocal or indeterminate; **OR;**

4.2.2 Carrier testing for patient with family history of ataxia-telangiectasia; **OR;**

4.2.3 Confirmed diagnosis, and need to establish disease-causing mutation; **OR;**

5.0 **CADASIL (Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy) - NOTCH3** gene testing **requires prior authorization** through the Health Services Division and is considered medically necessary for **1 or more** of the following indications:

5.1 Asymptomatic individual 18 years or older, when disease-causing mutation has been confirmed in an affected relative

5.2 Asymptomatic at-risk individuals who have a 1st degree relative with a clinical diagnosis of CADASIL but who are deceased or otherwise unable to be tested

5.3 Prenatal diagnosis, when disease-causing mutation has been confirmed in affected parent or in affected relative of at-risk parent

5.4 Confirmation of diagnosis in patient with clinical features and neuroimaging findings suggestive of CADASIL, irrespective of age

Charcot-Marie-Tooth Hereditary Neuropathy

6.0 **Charcot-Marie-Tooth (CMT) Hereditary Neuropathy** – Gene testing for the diagnosis or screening for CMT **requires prior authorization** through the Health Services Division and may include genes as described below:

6.1 **Charcot-Marie-Tooth Hereditary Neuropathy Type 1 - EGR2, FBLN5, LITAF, MPZ, NEFL and PMP22** gene testing individually or as part of a panel **requires prior authorization** through the Health Services Division and is considered medically necessary as indicated by **1 or more** of the following:

6.1.1 High clinical suspicion after acquired causes of peripheral neuropathy have been excluded by standard diagnostic evaluation as indicated by **1 or more** of the following:

6.1.1.1 Clinical findings suggestive of Charcot-Marie-Tooth hereditary neuropathy as indicated by **1 or more** of the following:

6.1.1.1.1. Diminished tendon reflexes

6.1.1.1.2. Distal muscle atrophy

6.1.1.1.3. Distal sensory loss

6.1.1.1.4. Foot drop

6.1.1.1.5. High-arched feet (pes cavus deformity)

6.1.1.1.6. Palpably enlarged nerves (e.g. ulnar nerve at olecranon groove, greater auricular nerve along lateral aspect of neck)

6.1.1.1.7. Progressive weakness of hands and feet

6.1.1.1.8. Weak ankle dorsiflexion

6.1.1.2 Family history suggestive of autosomal dominant inheritance, and electromyography and nerve conduction velocity studies consistent with demyelinating neuropathy

6.1.1.3 Family history negative or unavailable, and electromyography and nerve conduction velocity studies consistent with demyelinating neuropathy

6.2 **Charcot-Marie-Tooth Hereditary Neuropathy Type 2 - HSPB1, MFN2, and MPZ** gene testing individually or as part of a panel **requires prior authorization** through the Health Services Division and is considered medically necessary as indicated by **1 or more** of the following:

6.2.1 High clinical suspicion after acquired causes of peripheral neuropathy have been excluded by standard diagnostic evaluation as indicated by **1 or more** of the following:

6.2.1.1 Clinical findings suggestive of Charcot-Marie-Tooth hereditary neuropathy as indicated by **1 or more** of the following:

6.2.1.1.1. Diminished tendon reflexes

6.2.1.1.2. Distal muscle atrophy

6.2.1.1.3. Distal sensory loss

6.2.1.1.4. Foot drop

6.2.1.1.5. High-arched feet (pes cavus deformity)

6.2.1.1.6. Progressive weakness of hands and feet

6.2.1.1.7. Weak ankle dorsiflexion

6.2.1.2 Family history suggestive of autosomal dominant inheritance, and electromyography and nerve conduction velocity studies consistent with axonal neuropathy

6.2.1.3 Family history suggestive of autosomal recessive inheritance, and electromyography and nerve conduction velocity studies consistent with axonal neuropathy

6.2.1.4 Family history negative or unavailable, and electromyography and nerve conduction velocity studies consistent with axonal neuropathy

6.2.2 Carrier testing for **1 or more** of the following:

6.2.2.1 Adult patient with family history of autosomal recessive Charcot-Marie-Tooth hereditary neuropathy

6.2.2.2 Reproductive partner of autosomal recessive gene mutation carrier

6.3 **Charcot-Marie-Tooth Hereditary Neuropathy Type 4 - FGD4, GDAP1, NDRG1, PRX, SBF2, and SH3TC2** gene testing individually or as part of a panel **requires prior authorization** through the Health Services Division and is considered medically necessary as indicated by **1 or more** of the following:

6.3.1 High clinical suspicion after acquired causes of peripheral neuropathy have been excluded by standard diagnostic evaluation as indicated by **1 or more** of the following:

6.3.1.1 Clinical findings suggestive of Charcot-Marie-Tooth hereditary neuropathy as indicated by **1 or more** of the following:

6.3.1.1.1. Diminished tendon reflexes

6.3.1.1.2. Distal muscle atrophy

6.3.1.1.3. Distal sensory loss

6.3.1.1.4. Foot drop

6.3.1.1.5. High-arched feet (pes cavus deformity)

6.3.1.1.6. Progressive weakness of hands and feet

6.3.1.1.7. Weak ankle dorsiflexion

6.3.1.2 Family history suggestive of autosomal recessive inheritance, and electromyography and nerve conduction

velocity studies consistent with axonal or demyelinating neuropathy

6.3.1.3 Family history negative or unavailable, and electromyography and nerve conduction velocity studies consistent with axonal or demyelinating neuropathy

6.3.2 Carrier testing for **1 or more** of the following:

6.3.2.1 Adult patient with family history of autosomal recessive Charcot-Marie-Tooth hereditary neuropathy

6.3.2.2 Reproductive partner of autosomal recessive gene mutation carrier

6.4 **Charcot-Marie-Tooth Hereditary Neuropathy Type X - AIFM1, GJB1, PDK3, and PRPS1** gene testing individually or as part of a panel **requires prior authorization** through the Health Services Division and is considered medically necessary as indicated by **1 or more** of the following:

6.4.1 High clinical suspicion after acquired causes of peripheral neuropathy have been excluded by standard diagnostic evaluation as indicated by **1 or more** of the following:

6.4.1.1 Clinical findings suggestive of Charcot-Marie-Tooth hereditary neuropathy as indicated by **1 or more** of the following:

6.4.1.1.1. Diminished tendon reflexes

6.4.1.1.2. Distal muscle atrophy

6.4.1.1.3. Distal sensory loss

6.4.1.1.4. Foot drop

6.4.1.1.5. High-arched feet (pes cavus deformity)

6.4.1.1.6. Progressive weakness of hands and feet

6.4.1.1.7. Weak ankle dorsiflexion

6.4.1.2 Family history suggestive of X-linked inheritance

6.4.1.3 Family history negative or unavailable, and electromyography and nerve conduction velocity studies consistent with axonal neuropathy

6.4.1.4 Family history negative or unavailable, and electromyography and nerve conduction velocity studies consistent with demyelinating neuropathy

6.4.2 Carrier testing for at-risk female relative of affected male with X linked mutation

6.4.3 Prenatal diagnosis, when disease-causing mutation in Charcot-Marie-Tooth hereditary neuropathy gene has been identified in affected relative

7.0 **Familial Dysautonomia IKBKAP** genetic testing **requires** prior authorization through the Health Services Division and is considered medically necessary for the diagnosis or screening for familial dysautonomia, as indicated by **1 or more** of the following:

7.1 Carrier testing for **1 or more** of the following:

7.1.1 Individual of Ashkenazi Jewish ancestry and of reproductive age;

7.1.2 Individual with first- or second-degree family history of familial dysautonomia.

7.1.3 Reproductive partner of ELP1 gene mutation carrier

7.2 Clinical findings suggestive of familial dysautonomia, including **ALL** of the following:

7.2.1 Absence of axon flare response after intradermal histamine injection; **AND**

7.2.2 Absence of overflow tears with emotional crying; **AND**

7.2.3 Decreased or absent deep tendon reflexes; **AND**

7.2.4 Decreased taste and absence of fungiform papillae of tongue on visual inspection; **AND**

7.2.5 Hypotonia in infancy; **AND**

7.2.6 Pupillary hypersensitivity to parasympathomimetic agents (e.g. topical methacholine or pilocarpine).

8.0 **Friedreich Ataxia (FXN)** gene testing **requires prior authorization** through the Health Services Division and is considered medically necessary for the diagnosis or carrier testing for Friedreich ataxia when **1 or more** of the following are present:

8.1 Carrier testing in at-risk relative when disease-causing FXN gene mutation has been identified in family; **OR;**

8.2 Carrier testing in reproductive partner of known FXN mutation carrier; **OR;**

8.3 Confirmation of diagnosis in patient with clinical findings suggestive of Friedreich ataxia (e.g. fatigue, aggressive scoliosis, loss of coordination in arms and legs).

9.0 **Myotonic Dystrophy, Type 1 (DMPK)** gene testing **requires prior authorization** through the Health Services Division and is considered medically necessary when **ANY** of the following criteria is met:

9.1 Confirmation of diagnosis when clinical findings and family history are suggestive of myotonic dystrophy type 1

9.2 Predictive testing of individuals when DMPK expansion has been identified in affected relative

9.3 Carrier screening when the individual to be tested is asymptomatic with family history of myotonic dystrophy, type 1

9.4 Prenatal diagnosis by amniocentesis if polyhydramnios or decreased fetal activity is detected in third trimester

9.5 Prenatal diagnosis when DMPK expansion has been identified in affected parent

10.0 **Myotonic Dystrophy, Type 2 CNBP** gene testing **requires prior authorization** through the Health Services Division and is considered medically necessary when **1 or more** of the following criteria is met:

10.1 Asymptomatic individual 18 years or older with family history of myotonic dystrophy type 2

10.2 Carrier screening when the individual to be tested is asymptomatic with family history of myotonic dystrophy, type 2

10.3 Confirmation of diagnosis when clinical findings and family history are suggestive of myotonic dystrophy type 2

11.0 **Muscular Dystrophies (Duchene and Becker) - DMD** gene testing **requires prior authorization** through the Health Services Division and is considered medically necessary when **ANY** one of the following criteria is met:

11.1 Carrier screening when the individual to be tested is an asymptomatic female with family history of Duchenne muscular dystrophy, Becker muscular dystrophy, or DMD-associated dilated cardiomyopathy

11.2 Patient with clinical findings consistent with muscular dystrophy, elevated serum creatinine kinase and family history

11.3 Patient with confirmed diagnosis of muscular dystrophy, to establish disease-causing mutation

11.4 Prenatal diagnosis in fetus with 46,XY karyotype, when disease-causing mutation in DMD gene has been identified in carrier mother or if linkage has been established suggesting mother is carrier

12.0 **Nemaline Myopathy - ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, MYO18B, MYPN, NEB, TNNT1, TPM2 and TPM3** gene testing individually or as part of a panel **requires prior authorization** through the Health Services Division and is considered medically necessary for diagnosis or screening for nemaline myopathy when **1 or more** of the following criteria are met:

12.1 Carrier testing for **1 or more** of the following:

12.1.1 For NEB gene mutations: individual of Ashkenazi Jewish ancestry and of reproductive age

12.1.2 For TNNT1 gene mutations: individual of Old Order Amish ancestry and of reproductive age

12.1.3 Individual with family history of nemaline myopathy

12.2 Clinical findings are suggestive of nemaline myopathy and equivocal findings on muscle biopsy

12.3 Need to establish disease-causing mutation in patient with confirmed diagnosis

12.4 Screening of parent of affected individual with ACTA1, KBTBD13, TPM2 or TPM3 gene mutation and no known family history.

13.0 **Seizure Disorders, Hereditary - SCN1A** gene testing **requires prior authorization** through the Health Services Division and is considered medically necessary for the diagnosis of SCN1A-related seizure disorder or screening of at-risk relatives, as indicated by **either** of the following:

13.1 Confirmation of SCN1A-related seizure disorder in individual with clinical suspicion of **1 or more** of the following:

13.1.1 Dravet syndrome

13.1.2 Intractable childhood epilepsy with generalized tonic-clonic seizures

13.1.3 Severe infantile multifocal epilepsy

13.2 Evaluation of asymptomatic parent of proband with pathogenic SCN1A mutation and no known history of other affected family members.

14.0 **Spinocerebellar Ataxia (SCA) – ATXN1, ATXN2, ATXN3, ATXN7, and CACNA1A** gene testing individually or as part of a panel **requires prior authorization** through the Health Services Division and is considered medically necessary for the following:

14.1 Diagnosis of spinocerebellar ataxia, as indicated by **1 or more** of the following:

14.1.1 ATXN1, ATXN2, ATXN3, ATXN7 and CACNA1A gene and gene panel testing for confirmation of diagnosis in patient with clinical findings suggestive of spinocerebellar ataxia

14.1.2 Prenatal diagnosis, for family in which disease-causing mutation in spinocerebellar ataxia gene has been identified.

14.1.3 Comprehensive multigene panel testing, as indicated by **ALL** of the following:

14.1.3.1 Confirmation of diagnosis in patient with clinical findings suggestive of spinocerebellar ataxia; **AND**

14.1.3.2 Targeted single gene or limited panel testing to include ATXN1, ATXN2, ATXN3, ATXN7, and CACNA1A mutations is negative

15.0 **Spinal Muscular Atrophy - SMN1 and SMN2** gene testing **requires prior authorization** through the Health Services Division and is considered medically necessary to confirm or establish diagnosis in individual suspected of having spinal muscular atrophy, in addition to other indications as mentioned in Reproductive Carrier Screening and Prenatal Care MP9477.

16.0 **Amyotrophic Lateral Sclerosis (ALS) Familial** – **(***C9* orf *72, SOD1, TARDBP, FUS, FIG4, ANG* gene testing individually or as part of a panel **requires prior authorization** through the Health Services Division and is considered medically necessary for the following:

16.1 For assessing risk among family members of an affected relative with an established genetic cause

16.2 If the member displays evidence of ALS or is diagnosed with ALS when familial cause is suspected

16.3 Juvenile-onset ALS genetic testing the following genes are also appropriate SPG11, SETX, and ALS2

16.4 Asymptomatic at-risk individuals 18 years of age or older with a family history of ALS and the proband (initial affected) individual is deceased or unavailable for testing.

17.0 **Non-Covered Tests**: The following tests are considered experimental and investigational and therefore are not medically necessary:

17.1 Alzheimer Disease (Late Onset) – APOE Genotyping

17.2 Autism Spectrum Disorders – Multi-Gene Panels

17.3 Familial Frontotemporal Dementia - C9orf72, GRN and MAPT Genes

17.4 Parkinson Disease - ATP13A2, GBA, LRRK2, PARK7, PINK1, PRKN, SNCA, and VPS35 Genes

17.5 Epilepsy/Seizure Disorders (Hereditary) – Multi-Gene Panels

17.6 Sensory-Motor Neuropathy – Multi-Gene Panels

17.7 Oculopharyngeal Muscular Dystrophy (OPMD) – PABPN1

18.0 All other indications not listed above are considered experimental and investigational and therefore are not medically necessary.

CPT/HCPCS Codes Related to MP9497

* The list of codes (and their descriptors, if any) is provided for informational purposes only and may not be all inclusive or current. Listing of a code in this medical policy does not imply that the service described by the code is a covered or non-covered service. Benefit coverage for any service is determined by the member's policy of health coverage with WellFirst Health. Inclusion of a code above does not imply any right to reimbursement or guarantee claim payment. Other medical policies may also apply.

CPT Code	Description
0023U	AR (androgen receptor) (eg, spinal and bulbar muscular atrophy, Kennedy disease, X chromosome inactivation), full sequence analysis, including small sequence changes in exonic and intronic regions, deletions, duplications, short tandem repeat (STR) expansions, mobile element insertions, and variants in non-uniquely mappable regions

CPT Code	Description
0063U	Neurology (autism), 32 amines by LC-MS/MS, using plasma, algorithm reported as metabolic signature associated with autism spectrum disorder (e.g. NeuroPointDX)
0136U	ATM (ataxia telangiectasia mutated) (e.g., ataxia telangiectasia) mRNA sequence analysis
0139U	Neurology (autism spectrum disorder [ASD]), quantitative measurements of 6 central carbon metabolites (ie, a-ketoglutarate, alanine, lactate, phenylalanine, pyruvate, and succinate), LC-MS/MS, plasma, algorithmic analysis with result reported as negative or positive (with metabolic subtypes of ASD) (e.g. NPDX ASD Energy Metabolism)
0170U	Neurology (autism spectrum disorder [ASD]), RNA, next-generation sequencing, saliva, algorithmic analysis, and results reported as predictive probability of ASD diagnosis
0206U	Neurology (Alzheimer disease); cell aggregation using morphometric imaging and protein kinase C-epsilon (PKCe) concentration in response to amylospheroid treatment by ELISA, cultured skin fibroblasts, each reported as positive or negative for Alzheimer disease
0207U	Neurology (Alzheimer disease); quantitative imaging of phosphorylated ERK1 and ERK2 in response to bradykinin treatment by in situ immunofluorescence, using cultured skin fibroblasts, reported as a probability index for Alzheimer disease (List separately in addition to code for primary procedure)
0216U	Neurology (inherited ataxias), genomic DNA sequence analysis of 12 common genes including small sequence changes, deletions, duplications, short tandem repeat gene expansions, and variants in non-uniquely mappable regions, blood or saliva, identification and categorization of genetic variants
0217U	Neurology (inherited ataxias), genomic DNA sequence analysis of 51 genes including small sequence changes, deletions, duplications, short tandem repeat gene expansions, and variants in non-uniquely mappable regions, blood or saliva, identification and categorization of genetic variants
0218U	Neurology (muscular dystrophy), DMD gene sequence analysis, including small sequence changes, deletions, duplications, and variants

CPT Code	Description
	in non-uniquely mappable regions, blood or saliva, identification and characterization of genetic variants
0230U	AR (androgen receptor) (eg, spinal and bulbar muscular atrophy, Kennedy disease, X chromosome inactivation), full sequence analysis, including small sequence changes in exonic and intronic regions, deletions, duplications, short tandem repeat (STR) expansions, mobile element insertions, and variants in non-uniquely mappable regions
0231U	CACNA1A (calcium voltage-gated channel subunit alpha 1A) (eg, spinocerebellar ataxia), full gene analysis, including small sequence changes in exonic and intronic regions, deletions, duplications, short tandem repeat (STR) gene expansions, mobile element insertions, and variants in non-uniquely mappable regions
0232U	CSTB (cystatin B) (eg, progressive myoclonic epilepsy type 1A, Unverricht-Lundborg disease), full gene analysis, including small sequence changes in exonic and intronic regions, deletions, duplications, short tandem repeat (STR) expansions, mobile element insertions, and variants in non-uniquely mappable regions
0233U	FXN (frataxin) (eg, Friedreich ataxia), gene analysis, including small sequence changes in exonic and intronic regions, deletions, duplications, short tandem repeat (STR) expansions, mobile element insertions, and variants in non-uniquely mappable regions
0234U	MECP2 (methyl CpG binding protein 2) (eg, Rett syndrome), full gene analysis, including small sequence changes in exonic and intronic regions, deletions, duplications, mobile element insertions, and variants in non-uniquely mappable regions
0235U	PTEN (phosphatase and tensin homolog) (eg, Cowden syndrome, PTEN hamartoma tumor syndrome), full gene analysis, including small sequence changes in exonic and intronic regions, deletions, duplications, mobile element insertions, and variants in non-uniquely mappable regions
0236U	SMN1 (survival of motor neuron 1, telomeric) and SMN2 (survival of motor neuron 2, centromeric) (eg, spinal muscular atrophy) full gene analysis, including small sequence changes in exonic and intronic regions, duplications and deletions, and mobile element insertions

CPT Code	Description
81161	DMD (dystrophin) (e.g., Duchenne/Becker muscular dystrophy) deletion analysis, and duplication analysis, if performed
81177	ATN1 (atrophin 1) (eg, dentatorubral-pallidoluysian atrophy) gene analysis, evaluation to detect abnormal (eg, expanded) alleles
81178	ATXN1 (ataxin 1) (e.g., spinocerebellar ataxia) gene analysis, evaluation to detect abnormal (e.g., expanded) alleles
81179	ATXN2 (ataxin 2) (e.g., spinocerebellar ataxia) gene analysis, evaluation to detect abnormal (e.g., expanded) alleles
81180	ATXN3 (ataxin 3) (e.g., spinocerebellar ataxia, Machado-Joseph disease) gene analysis, evaluation to detect abnormal (e.g., expanded) alleles
81181	ATXN7 (ataxin 7) (e.g., spinocerebellar ataxia) gene analysis, evaluation to detect abnormal (e.g., expanded) alleles
81182	ATXN8OS (ATXN8 opposite strand [non-protein coding]) (e.g., spinocerebellar ataxia) gene analysis, evaluation to detect abnormal (e.g., expanded) alleles
81183	ATXN10 (ataxin 10) (e.g., spinocerebellar ataxia) gene analysis, evaluation to detect abnormal (e.g., expanded) alleles
81184	CACNA1A (calcium voltage-gated channel subunit alpha1 A) (e.g., spinocerebellar ataxia) gene analysis; evaluation to detect abnormal (e.g., expanded) alleles
81185	CACNA1A (calcium voltage-gated channel subunit alpha1 A) (e.g., spinocerebellar ataxia) gene analysis; full gene sequence
81186	CACNA1A (calcium voltage-gated channel subunit alpha1 A) (e.g., spinocerebellar ataxia) gene analysis; known familial variant
81187	CALR (calreticulin) (e.g., myeloproliferative disorders), gene analysis, common variants in exon 9
81188	CSTB (cystatin B) (e.g., Unverricht-Lundborg disease) gene analysis; evaluation to detect abnormal (e.g., expanded) alleles
81189	CSTB (cystatin B) (e.g., Unverricht-Lundborg disease) gene analysis; full gene sequence

CPT Code	Description
81190	CSTB (cystatin B) (e.g., Unverricht-Lundborg disease) gene analysis; known familial variant(s)
81204	AR (androgen receptor) (eg, spinal and bulbar muscular atrophy, Kennedy disease, X chromosome inactivation) gene analysis; characterization of alleles (eg, expanded size or methylation status)
81229	Cytogenomic constitutional (genome-wide) microarray analysis; interrogation of genomic regions for copy number and single nucleotide polymorphism (SNP) variants for chromosomal abnormalities
81234	DMPK (DM1 protein kinase) (e.g., myotonic dystrophy type 1) gene analysis; evaluation to detect abnormal (expanded) alleles
81239	DMPK (DM1 protein kinase) (e.g., myotonic dystrophy type 1) gene analysis; characterization of alleles (e.g., expanded size)
81242	FANCC (Fanconi anemia, complementation group C) (eg, Fanconi anemia, type C) gene analysis, common variant (eg, IVS4+4A>T)
81243	FMR1 (fragile X mental retardation 1) (e.g., fragile X mental retardation) gene analysis; evaluation to detect abnormal (e.g., expanded) alleles
81244	FMR1 (fragile X mental retardation 1) (e.g., fragile X mental retardation) gene analysis; characterization of alleles (e.g., expanded size and promoter methylation status)
81251	GBA (glucosidase, beta, acid) (eg, Gaucher disease) gene analysis, common variants (eg, N370S, 84GG, L444P, IVS2+1G>A)
81260	IKBKAP (inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase complex-associated protein) (e.g., familial dysautonomia) gene analysis, common variants (e.g., 2507+6T>C, R696P)
81271	HTT (huntingtin) (eg, Huntington disease) gene analysis; evaluation to detect abnormal (eg, expanded) alleles
81274	HTT (huntingtin) (eg, Huntington disease) gene analysis; characterization of alleles (eg, expanded size)
81284	FXN (frataxin) (e.g., Friedreich ataxia) gene analysis; evaluation to detect abnormal (expanded) alleles
81285	FXN (frataxin) (e.g., Friedreich ataxia) gene analysis; characterization of alleles (e.g., expanded size)

CPT Code	Description
81286	FXN (frataxin) (e.g., Friedreich ataxia) gene analysis; full gene sequence
81289	FXN (frataxin) (e.g., Friedreich ataxia) gene analysis; known familial variant(s)
81302	MECP2 (methyl CpG binding protein 2) (e.g., Rett syndrome) gene analysis; full sequence analysis
81303	MECP2 (methyl CpG binding protein 2) (e.g., Rett syndrome) gene analysis; known familial variant
81304	MECP2 (methyl CpG binding protein 2) (e.g., Rett syndrome) gene analysis; duplication/deletion variants
81312	PABPN1 (poly[A] binding protein nuclear 1) (eg, oculopharyngeal muscular dystrophy) gene analysis, evaluation to detect abnormal (eg, expanded) alleles
81321	PTEN (phosphatase and tensin homolog) (e.g., Cowden syndrome, PTEN hamartoma tumor syndrome) gene analysis; full sequence analysis
81322	PTEN (phosphatase and tensin homolog) (e.g., Cowden syndrome, PTEN hamartoma tumor syndrome) gene analysis; known familial variant
81323	PTEN (phosphatase and tensin homolog) (e.g., Cowden syndrome, PTEN hamartoma tumor syndrome) gene analysis; duplication/deletion variant
81324	PMP22 (peripheral myelin protein 22) (e.g., Charcot-Marie-Tooth, hereditary neuropathy with liability to pressure palsies) gene analysis; duplication/deletion analysis
81325	PMP22 (peripheral myelin protein 22) (e.g., Charcot-Marie-Tooth, hereditary neuropathy with liability to pressure palsies) gene analysis; full sequence analysis
81326	PMP22 (peripheral myelin protein 22) (e.g., Charcot-Marie-Tooth, hereditary neuropathy with liability to pressure palsies) gene analysis; known familial variant
81329	SMN1 (survival of motor neuron 1, telomeric) (e.g., spinal muscular atrophy) gene analysis; dosage/deletion analysis (e.g., carrier testing),

CPT Code	Description
	includes SMN2 (survival of motor neuron 2, centromeric) analysis, if performed
81336	SMN1 (survival of motor neuron 1, telomeric) (e.g., spinal muscular atrophy) gene analysis; full gene sequence
81337	SMN1 (survival of motor neuron 1, telomeric) (e.g., spinal muscular atrophy) gene analysis; known familial sequence variant(s)
81343	PPP2R2B (protein phosphatase 2 regulatory subunit Bbeta) (e.g., spinocerebellar ataxia) gene analysis, evaluation to detect abnormal (e.g., expanded) alleles
81344	TBP (TATA box binding protein) (e.g., spinocerebellar ataxia) gene analysis, evaluation to detect abnormal (e.g., expanded) alleles
81400	Molecular pathology procedure level 1
81401	Molecular pathology procedure level 2
81403	Molecular pathology procedure level 4
81404	Molecular pathology procedure level 5
81405	Molecular pathology procedure level 6
81406	Molecular pathology procedure level 7
81407	Molecular pathology procedure level 8
81408	Molecular pathology procedure level 9
81411	Aortic dysfunction or dilation (eg, Marfan syndrome, Loeys Dietz syndrome, Ehler Danlos syndrome type IV, arterial tortuosity syndrome); duplication/deletion analysis panel, must include analyses for TGFBR1, TGFBR2, MYH11, and COL3A1
81415	Exome (e.g., unexplained constitutional or heritable disorder or syndrome); sequence analysis
81416	Exome (e.g., unexplained constitutional or heritable disorder or syndrome); sequence analysis, each comparator exome (e.g., parents, siblings)

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CPT Code	Description
81417	Exome (e.g., unexplained constitutional or heritable disorder or syndrome); re-evaluation of previously obtained exome sequence (e.g., updated knowledge or unrelated condition/syndrome)
81419	Epilepsy genomic sequence analysis panel, must include analyses for ALDH7A1, CACNA1A, CDKL5, CHD2, GABRG2, GRIN2A, KCNQ2, MECP2, PCDH19, POLG, PRRT2, SCN1A, SCN1B, SCN2A, SCN8A, SLC2A1, SLC9A6, STXBP1, SYNGAP1, TCF4, TPP1, TSC1, TSC2, and ZEB2
81425	Genome (e.g., unexplained constitutional or heritable disorder or syndrome); sequence analysis
81426	Genome (e.g., unexplained constitutional or heritable disorder or syndrome); sequence analysis, each comparator genome (e.g., parents, siblings)
81427	Genome (e.g., unexplained constitutional or heritable disorder or syndrome); re-evaluation of previously obtained genome sequence (e.g., updated knowledge or unrelated condition/syndrome)
81440	Nuclear encoded mitochondrial genes (eg, neurologic or myopathic phenotypes), genomic sequence panel, must include analysis of at least 100 genes, including BCS1L, C10orf2, COQ2, COX10, DGUOK, MPV17, OPA1, PDSS2, POLG, POLG2, RRM2B, SCO1, SCO2, SLC25A4, SUCLA2, SUCLG1, TAZ, TK2, and TYMP
81448	Hereditary peripheral neuropathies (e.g., Charcot-Marie-Tooth, spastic paraplegia), genomic sequence analysis panel, must include sequencing of at least 5 peripheral neuropathy-related genes (e.g., BSCL2, GJB1, MFN2, MPZ, REEP1, SPAST, SPG11, SPTLC1)
81479	Unlisted molecular pathology procedure
88248	Chromosome analysis for breakage syndromes; baseline breakage, score 50-100 cells, count 20 cells, 2 karyotypes (e.g., for ataxia telangiectasia, Fanconi anemia, fragile X)
88271	Molecular cytogenetics; DNA probe, each (e.g., FISH)
88272	Molecular cytogenetics; chromosomal in situ hybridization, analyze 3-5 cells

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CPT Code	Description
88273	Molecular cytogenetics; chromosomal in situ hybridization, analyze 10-30 cells
88274	Molecular cytogenetics; interphase in situ hybridization, analyze 25-99 cells
88275	Molecular cytogenetics; interphase in situ hybridization, analyze 100-300 cells
S3800	Genetic testing for amyotrophic lateral sclerosis (ALS)
S3852	DNA analysis for APOE epsilon 4 allele for susceptibility to Alzheimer's disease
S3853	Genetic testing for myotonic muscular dystrophy

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