

Spinocerebellar Ataxia Genetic Testing

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Spinocerebellar ataxia (SCA) genetic testing is addressed by this guideline.

Procedures addressed

The inclusion of any procedure code in this table does not imply that the code is under management or requires prior authorization. Refer to the specific Health Plan's procedure code list for management requirements.

Procedures addressed by this guideline	Procedure codes
ATXN1 gene analysis, evaluation to detect abnormal (eg,expanded) allele	81178
ATXN2 gene analysis, evaluation to detect abnormal (eg,expanded) allele	81179
ATXN3 gene analysis, evaluation to detect abnormal (eg,expanded) allele	81180
ATXN7 gene analysis, evaluation to detect abnormal (eg,expanded) allele	81181
ATXN8 gene analysis, evaluation to detect abnormal (eg, expanded) alleles	81182
ATXN10 gene analysis, evaluation to detect abnormal (eg, expanded) alleles	81183
CACNA1A gene analysis; evaluation to detect abnormal (eg, expanded) alleles	81184
CACNA1A gene analysis; full gene sequence	81185
CACNA1A gene analysis; known familial variant	81186
Genomic Unity CACNA1A Analysis	0231U
PPP2R2B gene analysis, evaluation to detect abnormal (eg, expanded) alleles	81343
SCA multigene panel	81479

Spinocerebellar Ataxia

Procedures addressed by this guideline	Procedure codes
TBP gene analysis, evaluation to detect abnormal (eg, expanded) alleles	81344

Criteria

Requests for spinocerebellar ataxia (SCA) testing are reviewed using the following criteria.

Known Familial Mutation Analysis

- Genetic Counseling:
 - Pre- and post-test genetic counseling by an appropriate provider (as deemed by the Health Plan policy), AND
- Previous Genetic Testing:
 - No previous genetic testing that would detect the familial mutation, AND
- Presymptomatic Testing for Asymptomatic Individuals:
 - Member is 18 years of age or older, and
 - Known disease-causing mutation in SCA gene identified in 1st or 2nd degree relative(s), OR
- Diagnostic Testing for Symptomatic Individuals:
 - Known disease-causing mutation in SCA gene identified in 1st or 2nd degree relative(s), AND
- Rendering laboratory is a qualified provider of service per the Health Plan policy

Single Gene Testing

- Genetic Counseling:
 - Pre- and post-test genetic counseling by an appropriate provider (as deemed by the Health Plan policy), AND
- Previous Genetic Testing:
 - No previous testing of requested gene(s), and
 - No mutation identified by previous analysis, if performed, and
 - No known familial mutation in a gene known to cause ataxia, AND
- Diagnostic Testing for Symptomatic Individuals:
 - Individual has been diagnosed with cerebellar ataxia, and
 - Medical history points to the specific subtype of SCA requested (e.g. age of onset, distinguishing features present, etc), AND

- Documentation from ordering provider indicating how test results will be used to directly impact medical care for the individual (e.g. change in surveillance or treatment plan), AND
- The member does not have a known underlying cause for their ataxia (e.g. alcoholism, vitamin deficiencies, multiple sclerosis, vascular disease, tumors, known mutation, etc), AND
- Family history is consistent with an autosomal dominant inheritance pattern (including simplex cases), AND
- Rendering laboratory is a qualified provider of service per the Health Plan policy

Multigene Panel Testing

- Genetic counseling:
 - Pre- and post-test genetic counseling by an appropriate provider (as deemed by the Health Plan policy), AND
- Previous Genetic Testing:
 - No previous testing of requested genes, and
 - No mutation identified by previous analysis, if performed, and
 - No known familial mutation in a gene known to cause ataxia, AND
- Diagnostic Testing for Symptomatic Individuals:
 - Individual has been diagnosed with cerebellar ataxia, regardless of age of onset, AND
- Documentation from ordering provider indicating how test results will be used to directly impact medical care for the individual (e.g. change in surveillance or treatment plan), AND
- The member does not have a known underlying cause for their ataxia (e.g. alcoholism, vitamin deficiencies, multiple sclerosis, vascular disease, tumors, known mutation, etc), AND
- Family history is consistent with an autosomal dominant inheritance pattern (including simplex cases), AND
- Medical history does not point to a specific genetic diagnosis for which a more focused test or panel would be appropriate, AND
- Rendering laboratory is a qualified provider of service per the Health Plan policy

Other considerations

- Gene panels that are specific to SCA will be considered for medical necessity according to the criteria outlined in this guideline. Test methodology should be appropriate to the disease-causing mutations that are commonly reported for the disorder in question (e.g., sequencing-only panels will not detect triplet repeat or large deletion/duplication mutations).

Note:

*The panel code(s) listed here may not be all-inclusive. For further discussion of what is considered an appropriate panel code, please refer to the guideline *Laboratory Billing and Reimbursement*.

Billing and Reimbursement

This section outlines the billing requirements for tests addressed in this guideline. These requirements will be enforced during the case review process whenever appropriate. Examples of requirements may include specific coding scenarios, limits on allowable test combinations or frequency and/or information that must be provided on a claim for automated processing. Any claims submitted without the necessary information to allow for automated processing (e.g. ICD code, place of service, etc.) will not be reimbursable as billed. Any claim may require submission of medical records for post service review.

Gene panels that are specific to SCA will be eligible for reimbursement according to the criteria outlined in this guideline.

Any individual gene or multi-gene panel is only reimbursable once per lifetime.

When otherwise reimbursable, the following limitations apply:

- When a panel is being performed, it is only reimbursable when billed with a single, appropriate panel procedure code (e.g., 81479*).
- When use of a panel code is not possible, each billed component procedure will be assessed independently.
- In general, only a limited number of panel components that are most likely to explain the member's presentation will be reimbursable. The remaining panel components will not be reimbursable.
- When the test is billed with multiple stacked procedure codes, only the following genes may be considered for reimbursement:
 - ATXN1 (SCA1)
 - ATXN2 (SCA2)
 - ATXN3 (SCA3)
 - CACNA1A (SCA6)
 - ATXN7 (SCA7)
 - TBP (SCA17)

Note:

*The panel code(s) listed here may not be all-inclusive. For further discussion of what is considered an appropriate panel code, please refer to the guideline *Genetic Testing by Multigene Panels*.

For general coding requirements, please refer to the guideline *Laboratory Procedure Code Requirements*.

What is spinocerebellar ataxia?

Spinocerebellar ataxias (SCA) are a group of autosomal dominant ataxias that have a range of phenotypes. There are various subtypes of SCA, which are denoted by numbers (e.g. SCA1, SCA3, etc.)¹

Prevalence

The prevalence of specific subtypes of SCA vary by region, often because of founder effects.²

Symptoms

Although the specific phenotype of each subtype varies, most individuals with SCA have “progressive adult-onset gait ataxia (often with hand dysmetria) and dysarthria associated with cerebellar atrophy on brain imaging.”¹ The age of onset for the different subtypes also overlaps, which it makes it difficult to distinguish between subtypes based on clinical phenotype only.^{1,2} See the table below for the various subtypes of SCA and the associated clinical features.

Cause

SCAs are caused by mutations in one of numerous genes. See the table below for the various subtypes of SCA and the associated genes.

Inheritance

SCAs are autosomal dominant disorders. Anticipation is observed in some of the SCAs. This means that as the disease passes through generations, the severity can increase and the age of onset can decrease.

Autosomal dominant inheritance

In autosomal dominant inheritance, individuals have 2 copies of the gene and only one mutation is required to cause disease. When a parent has a mutation, each offspring has a 50% risk of inheriting the mutation. Males and females are equally likely to be affected.

Diagnosis

Molecular genetic testing can be used to establish a specific diagnosis, which aids in understanding the prognosis and risk assessment for family members.¹

Management

Treatment of ataxia is largely supportive, and includes the use of canes and walkers for ambulation, speech therapy, and other assistive devices.¹

SCA subtype	Gene Associated	Clinical Features
SCA1	ATXN1	Progressive cerebellar ataxia, dysarthria, deterioration of bulbar functions, pyramidal signs, peripheral neuropathy ^{2,3}
SCA2	ATXN2	Progressive ataxia and dysarthria, nystagmus, slow saccadic eye movements, peripheral neuropathy, decreased DTRs, dementia ^{2,4}
SCA3	ATXN3	Gait problems, speech difficulties, clumsiness, visual blurring, diplopia, hyperreflexia, progressive ataxia, nystagmus, dysarthria, pyramidal and extrapyramidal signs; lid retraction, nystagmus, decreased saccade velocity; amyotrophy fasciculations, sensory loss ^{2,5}
SCA4	16q22.1	Sensory axonal neuropathy, deafness; may be allelic with 16q22-linked SCA ²
SCA5	SPTBN2	Early onset, slow course ²
SCA6	CACNA1A	Progressive cerebellar ataxia, dysarthria, nystagmus, sometimes episodic ataxia, very slow progression ^{2,6}

SCA subtype	Gene Associated	Clinical Features
SCA7	ATXN7	Progressive cerebellar ataxia, dysarthria, dysphagia, cone-rod and retinal dystrophy with progressive central visual loss resulting in blindness ^{2,7}
SCA8	ATXN8	Principally cerebellar ataxia, slowly progressing ataxia, scanning dysarthria, truncal instability, hyperactive tendon reflexes, decreased vibration sense; rarely, cognitive impairment ^{2,8}
SCA10	ATXN10	Progressive cerebellar ataxia, scanning dysarthria, dysphagia, upper-limb ataxia, generalized motor seizures and/or complex partial seizures, most families are of Native American background ^{2,9}
SCA11	TTBK2	Progressive cerebellar ataxia, abnormal eye signs (jerky pursuit, horizontal and vertical nystagmus), mild, remain ambulatory ^{2,10}
SCA12	PPP2R2B	Slowly progressive ataxia; action tremor in the 30s; hyperreflexia; subtle Parkinsonism possible; cognitive/psychiatric disorders including dementia ²

SCA subtype	Gene Associated	Clinical Features
SCA13	KCNC3	Ranges from progressive childhood-onset cerebellar ataxia, cerebellar dysarthria, occasional seizures to adult-onset progressive ataxia, mild intellectual disability, short stature ^{2,11}
SCA14	PRKCG	Progressive cerebellar ataxia, dysarthria, nystagmus, axial myoclonus, cognitive impairment, tremor, sensory loss, Parkinsonian features including rigidity and tremor ^{2,12}
SCA15	ITPR1	Progressive gait and limb ataxia, ataxic dysarthria, titubation, upper limb postural tremor, mild hyperreflexia, gaze-evoked nystagmus, and impaired vestibuloocular reflex gain ^{2,13}
SCA16	SCA16	Head tremor; reported in one Japanese family ²
SCA17	TBP	Ataxia, dementia, mental deterioration; occasional chorea, dystonia, myoclonus, epilepsy; Purkinje cell loss, intranuclear inclusions with expanded polyglutamine ^{2,14}

SCA subtype	Gene Associated	Clinical Features
SCA18	7q22-q32	Ataxia with early sensory/ motor neuropathy, nystagmus, dysarthria, decreased tendon reflexes, muscle weakness, atrophy, fasciculations, Babinski responses ²
SCA19/22	KCND3	Slowly progressive, rare cognitive impairment, myoclonus, hyperreflexia ²
SCA20	11q12.2-11q12.3	Progressive ataxia, dysarthria, palatal tremor (myoclonus), and/or abnormal phonation clinically resembling spasmodic adductor dysphonia, hyperreflexia, bradykinesia; calcification of the dentate nucleus. ^{2,15}
SCA21	TMEM240	Mild cognitive impairment ²
SCA23	PDYN	Dysarthria, abnormal eye movements, reduced vibration and position sense; reported in one Dutch family; neuropathology ²
SCA25	SCA25	Sensory neuropathy; reported in one French family ²
SCA26	EEF2	Dysarthria, irregular visual pursuits; reported in one Norwegian-American family; MRI: cerebellar atrophy ²

SCA subtype	Gene Associated	Clinical Features
SCA27	FGF14	Early-onset tremor; dyskinesia, cognitive deficits; reported in one Dutch family ²
SCA28	AFG3L2	Young-adult onset, progressive gait and limb ataxia resulting in coordination and balance problems, dysarthria, ptosis, nystagmus, and ophthalmoparesis, increased tendon reflexes; reported in two Italian families ^{2,16}
SCA29	ITPR1	Learning deficits, infant-onset hypotonia, motor delays ^{2,17}
SCA30	4q34.3-q35.1	Hyperreflexia ²
SCA31	BEAN1	Normal sensation ²
SCA35	TGM6	Hyperreflexia, Babinski responses; spasmodic torticollis ²
SCA36	NOP56	Late-onset, slowly progressive cerebellar syndrome typically associated with sensorineural hearing loss, muscle atrophy and denervation, especially of the tongue, as well as pyramidal signs, muscle fasciculations, hyperreflexia ²

SCA subtype	Gene Associated	Clinical Features
SCA37	DAB1	Adult onset, abnormal vertical eye movements, dysarthria, dysmetria, dysphagia ^{1,18}
SCA38	ELOVL5	Adult onset, axonal neuropathy ¹
SCA40	CCDC88C	Adult onset, brisk reflexes, spasticity ¹
SCA41	TRPC3	Adult onset, uncomplicated ataxia ¹
SCA42	CACNA1G	Mild pyramidal signs, saccadic pursuit ¹

Survival

The SCAs are a group of progressive disorders with a range of phenotypes. Specific symptoms and a genetically determined diagnosis can assist with determining predicted survival and prognosis.

Test Information

Testing for SCA may include known familial mutation analysis, repeat expansion analysis, next generation sequencing, deletion/duplication analysis, and/or multigene panel testing. Test methods vary by gene of interest.

Known Familial Mutation Analysis

Analysis for known familial mutations is typically performed by nucleotide repeat expansion analysis. Some mutations may require Sanger sequencing or deletion/duplication analysis.

Known familial mutation analysis is performed when a causative mutation has been identified in a close relative of the individual requesting testing.

Repeat Expansion Analysis

Several of the SCAs are caused by repeat expansions. Testing for these conditions is performed by expansion analysis to identify the number of repeats. Expansion analysis

can be performed for diagnostic testing, presymptomatic testing, as well as prenatal testing.

Next Generation Sequencing Assay

Next generation sequencing (NGS), which is also sometimes called massively parallel sequencing, was developed in 2005 to allow larger scale and more efficient gene sequencing. NGS relies on sequencing many copies of small pieces of DNA simultaneously and using bioinformatics to assemble the sequence. Sequence analysis detects single nucleotide substitutions and small (several nucleotide) deletions and insertions. Regions analyzed typically include the coding sequence and intron/exon boundaries. Promoter regions and intronic sequences may also be sequenced if disease-causing mutations are known to occur in these regions of a gene.

Deletion and Duplication Analysis

Analysis for deletions and duplications can be performed using a variety of technical platforms including exon array, Multiplex ligation-dependent probe amplification (MLPA), and NGS data analysis. These assays detect gains and losses too large to be identified through standard sequence analysis, often single or multiple exons or whole genes.

Multi-Gene Testing Panels

The efficiency of NGS has led to an increasing number of large, multi-gene testing panels. NGS panels that test several genes at once are particularly well-suited to conditions caused by more than one gene or where there is considerable clinical overlap between conditions making it difficult to reliably narrow down likely causes. Additionally, tests should be chosen to maximize the likelihood of identifying mutations in the genes of interest, contribute to alterations in management for an individual, and/or minimize the chance of finding variants of uncertain clinical significance.

Guidelines and Evidence

European Federation of Neurological Sciences

The European Federation of Neurological Sciences (EFNS, 2014) stated the following with regard to testing for autosomal dominant cerebellar ataxia:¹⁹

- “In the case of a family history that is compatible with an autosomal dominant cerebellar ataxia, screening for SCA1, SCA2, SCA3, SCA6, SCA7, and SCA17 is recommended (Level B). In Asian patients, DRPLA should also be tested for.”
- “If mutation analysis is negative, we recommend contact with or referral to a specialized clinic for reviewing the phenotype and further genetic testing (good practice point)”

- “In the case of sporadic ataxia and independent from onset age, we recommend routine testing for SCA1, SCA2, SCA3, SCA6, and DRPLA (in Asian patients) (level B), the step one panel of the recessive ataxia workup, i.e. mutation analysis of the FRDA gene (level B), and biochemical testing that includes cholestanol, vitamin E, cholesterol, albumin, CK, and alpha-fetoprotein.”

Selected Relevant Publications

The diagnostic evaluations for ataxia may include assessments for acquired, other nongenetic, and genetic etiologies.^{2,20} Establishing the diagnosis of a hereditary ataxia may include demonstration of typical clinical signs on neurological examination and exclusion of acquired or other nongenetic causes. Additionally, a positive family history, documentation of a hereditary ataxia disease causing mutation, and/or the presence of a characteristic clinical phenotype of a specific hereditary ataxia may solidify the diagnosis.²

The results of additional evaluations, such as brain imaging, may increase the suspicion of a hereditary etiology. These additional studies may indicate that an ataxia is slowly progressive and long standing, which may signify early onset.²¹ Furthermore, findings on MR spectroscopy may indicate a hereditary etiology is more likely compared to an immune-mediated ataxia.²¹

The likelihood of a hereditary etiology is higher in those with early age of onset (81%) compared to late onset (55%) idiopathic ataxia.²¹ The presence of other clinical features also increases the likelihood of detecting a mutation.²¹ In those with a family history consistent with autosomal dominant inheritance, a mutation was detected in 50-60% with testing for SCA1, 2, 3, 6, 7, 8, 10, 12, 17 and dentatorubral-pallidoluysian atrophy (DRPLA).² In a simplex case with no known acquired cause, the likelihood an individual has SCA1, 2, 3, 6, 8, 17, or Friedreich ataxia (FRDA) is approximately 13%, and the likelihood of a mutation in a different hereditary ataxia gene is more rare.² Even in those with an unremarkable family history, genetic testing may aid in their medical evaluation and in genetic counseling.²

It was suggested that genetic testing for progressive ataxias should include evaluation of the genes for FRDA, SCA 1, 2, 3, 6, 7 (12, 17) and fragile X-associated tremor/ataxia syndrome.²⁰ Testing may proceed in a sequential fashion. For those with a family history suggestive of autosomal dominant inheritance, first round testing for the most common hereditary ataxias (SCA1, SCA2, SCA3, SCA6, and SCA7) followed by testing for less common hereditary etiologies which may be guided by ethnic background and/or specific clinical features.² Single-gene testing may be pursued if the clinical examination is consistent with a specific diagnosis or if a specific type is known in the family.²

Note:

This benefit/harm statement only applies to those jurisdictions that do not have Medicare guidance. Based upon the guidelines and evidence provided in the clinical policy, following EviCore's criteria for Spinocerebellar Ataxia testing will ensure that testing will be available to those members most likely to benefit from a genetic diagnosis. For those not meeting criteria, it ensures alternate diagnostic strategies are considered. However, it is possible that some members who have the condition, but have non-standard features, will not receive an immediate approval for testing.

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