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Spinocerebellar Ataxia Type 10

Synonym: SCA10

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Summary

Clinical characteristics

Spinocerebellar ataxia type 10 (SCA10) is characterized by slowly progressive cerebellar ataxia that usually starts as poor balance and unsteady gait, followed by upper-limb ataxia, scanning dysarthria, and dysphagia. Abnormal tracking eye movements are common. Recurrent seizures after the onset of gait ataxia have been reported with variable frequencies among different families. Some individuals have cognitive dysfunction, behavioral disturbances, mood disorders, mild pyramidal signs, and peripheral neuropathy. Age of onset ranges from 12 to 48 years.

Diagnosis/testing

Diagnosis of SCA10 is established in a proband by identification of a heterozygous ATTCT pentanucleotide-repeat expansion in *ATXN10*. Affected individuals have expanded alleles with up to 4,500 ATTCT pentanucleotide repeats; intermediate alleles (280 to 850 repeats) may show reduced penetrance.

Management

Treatment of manifestations: Treatment is primarily focused on control of seizures, as uncontrolled seizures may lead to potentially fatal status epilepticus. Conventional anticonvulsants such as levetiracetam, phenytoin, carbamazepine, and valproic acid achieve reasonable control, although occasional breakthrough seizures may occur. Treatment measures for ataxia: canes / walkers / mobilized chairs; standard home modifications; exercise and physical therapy; and weight control to avoid difficulty with ambulation and mobility. For dysphagia: percutaneous placement of a gastrostomy tube for both prevention of aspiration and maintenance of nutritional intake; vitamin supplementation. For dysarthria: speech therapy and speech/communication assistive devices. Weighted utensils and dressing hooks for upper-limb coordination issues. Mild tranquilizers may be helpful for those with anxiety.

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Surveillance: Clinical neurology evaluation every four to six months; video esophagrams to evaluate those with dysphagia.

Agents/circumstances to avoid: Alcohol and drugs that are known to adversely affect cerebellar functions; falls, which may compromise motor function; activities that are potentially dangerous to individuals with ataxia or epilepsy.

Genetic counseling

SCA10 is inherited in an autosomal dominant manner. Offspring of an affected individual have a 50% chance of inheriting the repeat expansion. The risk of developing the SCA10 phenotype in individuals with expanded alleles in the intermediate range (280-850) is uncertain because of the apparently reduced penetrance. Anticipation has been observed in some families with paternal (but not maternal) transmission of the pentanucleotide repeat expansion. Prenatal testing for pregnancies at increased risk is possible if the diagnosis has been established by molecular genetic testing in an affected family member.

Diagnosis

Suggestive Findings

Spinocerebellar ataxia type 10 (SCA10) **should be suspected** in individuals with the following findings:

- Slowly progressive cerebellar ataxia starting as poor balance and unsteady gait
- Scanning dysarthria, dysphagia, and upper-limb ataxia following the gait ataxia
- Family history consistent with autosomal dominant inheritance and Native American or East Asian ancestry
- Generalized motor seizures and/or complex partial seizures

Note: Ataxia and its impact on quality of life is mild compared to SCA3 [Cruz et al 2019].

Other suggestive findings:

- **Brain MRI examination.** White matter atrophy exclusively in the cerebellum with extensive gray matter degeneration in the cerebellum, brain stem, thalamus, and putamen. Degeneration of thalamic gray matter and white matter in the cerebellar lobule VI is associated with epilepsy [Hernandez-Castillo et al 2019].
- **EEG.** Evidence of cortical dysfunction with or without focal epileptiform discharges on interictal electroencephalography in some affected individuals
- **Neurophysiology.** Polyneuropathy

Establishing the Diagnosis

The diagnosis of SCA10 **is established** in a proband by identification of a heterozygous ATTCT repeat expansion in *ATXN10* by molecular genetic testing (see Table 1).

Allele sizes

- **Normal alleles.** 10-32 ATTCT repeats; standard nomenclature c.1430+54822_54826ATTCT(10_32) [Matsuura et al 2000, Wang et al 2010]
  - 82% of unaffected individuals are compound heterozygotes for ATTCT repeat sizes in this range.
  - 18% of unaffected individuals are homozygous for ATTCT repeat sizes in this range.
- **Mutable normal alleles.** None identified
- **Reduced-penetrance alleles.** Further investigation is needed to determine what range of expanded allele sizes between 33 and 850 ATTCT repeats results in reduced penetrance. Alleles between 400 and 760

found in Brazilian individuals with SCA10 were reported as full-penetrance but are likely to have reduced penetrance [Alonso et al 2006].
    - 280 ATTCT repeats; standard nomenclature c.1430+54822_54826ATTCT(280). Identified in an individual with ataxia whose asymptomatic mother has the same size expansion, probably representing reduced penetrance [Matsuura et al 2006].
    - Alleles of 360 and 370 ATTCT repeats; standard nomenclature c.1430+54822_54826ATTCT(360_370). May be intermediate alleles with reduced or no penetrance [Alonso et al 2006].
    - Overlap of full- and reduced-penetrance alleles in 800-850 ATTCT repeat range needs to be clarified by further studies. 850 ATTCT repeats; standard nomenclature c.1430+54822_54826ATTCT(850). May be intermediate alleles with reduced penetrance [Raskin et al 2007].
    - An individual with Sioux ancestry without a family history of ataxia or seizure had an expansion allele of 1,400 pure ATTCT repeats; standard nomenclature c.1430+54822_54826ATTCT(1400). The individual developed pure ataxia at age 83 years. This could be classified as a reduced-penetrance allele [Bushara et al 2013, McFarland et al 2015].
- **Full-penetrance alleles.** 800 to 4,500 ATTCT repeats; standard nomenclature c.1430+54822_54826ATTCT(800_4500). The lower end of the full-penetrance allele range of 800 is not well defined; overlap with reduced-penetrance alleles exists.
- **Alleles of questionable significance.** Alleles between 33 and 280 ATTCT repeats have not been observed but could in some individuals show reduced penetrance.

Molecular genetic testing approaches can include **single-gene testing** or use of a **multigene panel**.

Single-gene testing. Perform targeted analysis for a heterozygous *ATXN10* allele with more than 33 ATTCT repeats. PCR and Southern blot analysis may be performed sequentially or concurrently.

- Analysis by PCR detects normal alleles. The presence of compound heterozygous *ATXN10* alleles excludes the diagnosis of SCA10.
- If PCR analysis shows only one allele, an alternate PCR test – the ATTCT-repeat-primed PCR [Matsuura & Ashizawa 2002] – can detect presence or absence of large numbers of repeats of reduced-penetrance or full-penetrance *ATXN10* alleles, but it cannot determine the size of the repeat tract. The clear absence of large numbers of repeats excludes the diagnosis of SCA10.
- Southern blot analysis of genomic DNA is necessary to determine the size of expanded alleles and to differentiate reduced-penetrance from full-penetrance alleles [Matsuura & Ashizawa 2002, Cagnoli et al 2004]. Long-range PCR may be a potentially useful clinical test in the future to distinguish between these two categories of alleles [Matsuura et al 2006, Kurosaki et al 2008].

A multigene panel that includes *ATXN10* ATTCT-repeat analysis and other genes of interest (see Differential Diagnosis) may also be considered. Note: (1) The genes included in the panel and the diagnostic sensitivity of the testing used for each gene vary by laboratory and are likely to change over time. (2) Some multigene panels may include genes not associated with the condition discussed in this *GeneReview*. (3) In some laboratories, panel options may include a custom laboratory-designed panel and/or custom phenotype-focused exome analysis that includes genes specified by the clinician. (4) Methods used in a panel may include sequence analysis, deletion/duplication analysis, and/or other non-sequencing-based tests. (5) Exome sequencing and genome sequencing do not readily detect *ATXN10* repeat expansions.

For an introduction to multigene panels click here. More detailed information for clinicians ordering genetic tests can be found here.

Table 1. Molecular Genetic Testing Used in Spinocerebellar Ataxia Type 10

Gene [1]	Method	Proportion of Pathogenic Variants [2] Identified by Method
ATXN10	Targeted analysis for ATTCT pentanucleotide repeat [3]	100% [4]

1. See Table A. Genes and Databases for chromosome locus and protein.
2. See Molecular Genetics for information on variants detected in this gene.
3. Southern blot analysis of both genomic DNA and amplicons from ATTCT-repeat primed PCR has been shown to be a reliable method to detect and characterize the ATTCT repeat insertion (see **Single-gene testing**).
4. Insertion of a variable number of ATTCT repeats in *ATXN10* is the mutational mechanism in all families with SCA10 examined to date [Matsuura & Ashizawa 2002, Cagnoli et al 2004, McFarland et al 2013, McFarland et al 2014].

Clinical Characteristics

Clinical Description

The clinical findings of spinocerebellar ataxia type 10 (SCA10) are relatively homogeneous. Ataxia causes progressive disability, and seizures may become life threatening if status epilepticus emerges. Reported age of onset ranges from 12 to 83 years [Matsuura et al 1999, Zu et al 1999, Rasmussen et al 2001, Teive et al 2004, Bushara et al 2013]. To date, more than 300 individuals have been identified with SCA10. The following description of the phenotypic features associated with this condition is based on these reports.

Ataxia. The central feature of the phenotype is slowly progressive cerebellar ataxia that usually starts as poor balance and unsteady gait.

The gait ataxia gradually worsens, leading to an increasing number of falls and necessitating use of a cane, walker, and eventually wheelchair. In the advanced stage, the affected individual is unable to stand or sit without support.

Upper-limb coordination begins to deteriorate within a few years after the onset of gait ataxia. Handwriting and other fine motor tasks, such as buttoning, are the first to be impaired, followed by increasing difficulties in daily activities such as feeding, dressing, and personal hygiene.

Scanning dysarthria, a type of slurred speech typically seen in cerebellar ataxia, appears within a few years after the onset of gait ataxia. Scanning speech is the result of impaired coordination of the movements of the vocal cords, tongue, palate, cheeks, and lips. Impaired coordination of the diaphragm and other respiratory muscles contributes to the speech impairment.

Poor coordination of tongue, throat, and mouth muscles causes dysphagia in later stages of the disease, often leading to life-threatening aspiration pneumonia.

Most individuals develop abnormal tracking eye movements: fragmented ocular pursuit, ocular dysmetria, and occasionally ocular flutter. Impaired ocular movements are attributable to cerebellar dysfunction. Some individuals with relatively severe ataxia show coarse gaze-induced nystagmus. Saccade velocity is normal.

Ataxia may be induced by small amounts of alcohol [Teive et al 2011c], by treatment with glucocorticosteroids [Moro et al 2013], or during pregnancy and puerperium [Teive et al 2011a].

Intention tremor was identified only in women from one of the 16 families with SCA10 reported in a Brazilian series [Domingues et al 2019].

Seizures. In most individuals, seizures are noted after the onset of gait ataxia.

Recurrent seizures have been reported in 20%-100% of affected individuals [Matsuura et al 1999, Zu et al 1999, Rasmussen et al 2001]. Generalized motor seizures are most common, but complex partial seizures occur. An attack of complex partial seizures may occasionally be followed by a generalized motor seizure, suggesting secondary generalization of focal seizure activity. Seizure characteristics do not appear to change with age.

Without treatment, generalized motor seizures may occur daily and complex partial seizures may occur up to several times a day. Poorly treated seizures may result in life-threatening status epilepticus and/or death [Grewal et al 2002].

Seizures were found to occur in six of 91 Brazilians (6.6%) with SCA10 from the Parana/Santa Catarina region, and those with seizures had earlier age at onset [Domingues et al 2019]; this is in contrast to a higher incidence of epilepsy in individuals of Mexican ancestry (60%) [Alonso et al 2006] and Brazilians from other regions (64.7%) [de Castilhos et al 2014].

Other. While overt progressive dementia is not observed, some individuals with SCA10 exhibit mild cognitive dysfunction (IQ ~70) as well as mood disorders.

Mild pyramidal signs (either hyperreflexia, Babinski sign, or both), behavioral disturbances (including psychosis, paranoid schizophrenia), dystonia, parkinsonism, peripheral neuropathy, central auditory processing, and sleep disorders have been variably seen [Rasmussen et al 2001, Gatto et al 2007, Wexler & Fogel 2011, Trikamji et al 2015, Zeigelboim et al 2015, Moro et al 2017, Schüle et al 2017, London et al 2018, Nascimento et al 2019].

Extraneural abnormalities including hepatic failure, anemia, and/or thrombocytopenia have been recorded in one family [Rasmussen et al 2001].

Low IQ, behavioral disturbances, and extraneural abnormalities have not been found in Brazilians with SCA10, although mild or equivocal pyramidal tract signs and rare sensory polyneuropathy were noted [Teive et al 2004, Alonso et al 2006]. Overall, Brazilians with SCA10 show a milder neurologic phenotype with fewer extracerebellar features than individuals of Mexican ancestry.

Genotype-Phenotype Correlations

A comparison of clinical data and genotypes in individuals with SCA10 revealed an inverse correlation between expansion size and age of onset ($p = 0.018$) [Matsuura et al 2000]. The number of repeats ranged from 800 to 4,500 and age of onset from 11 to 48 years. The correlation coefficient (r^2) was 0.34, suggesting that the ATTCT expansion size can explain only about one third of the variation in age of onset and implying the existence of other determinants of age of onset. A later study of Brazilians with SCA10 showed a similar inverse correlation with $r^2=0.532$ and $p<0.01$ [Teive et al 2004].

The presence of the *ATXN10* ATCCT interruption motif is associated with a higher prevalence of epileptic seizures [McFarland et al 2014].

Though not as yet assessed quantitatively, the severity of the disease in individuals with SCA10 does not appear to correlate with expansion size. Longitudinal clinical data are needed to examine whether repeat size correlates with disease progression.

Penetrance

Penetrance is usually complete. However, apparent reduced penetrance has been reported [Alonso et al 2006, Matsuura et al 2006, Raskin et al 2007].

Anticipation

Anticipation is usually associated with progressively larger ATTCT repeat expansions in successive generations. The expanded repeat alleles are mostly unstable with paternal transmission but remarkably stable with maternal transmission [Grewal et al 2002]. However, some paternal transmissions have shown intergenerational contraction of the expanded repeat allele, in spite of the clinically observed anticipation [Matsuura et al 2004].

Anticipation was first noted in one large family with SCA10 [Zu et al 1999]; less marked anticipation was observed in another, larger family [Matsuura et al 1999]. Severe early-childhood onset has been reported and juvenile onset has also been observed [Zu et al 1999, Rasmussen et al 2001, Matsuura et al 2006]. In small families, anticipation may be variable and difficult to evaluate [Rasmussen et al 2001]. Anticipation has been suggested in Brazilian families with SCA10; further studies are needed to confirm this observation [Teive et al 2004].

Interrupted repeat expansions show anticipation but are accompanied by a paradoxic contraction in intergenerational repeat size [McFarland et al 2013].

Prevalence

The exact prevalence of SCA10 is unknown.

In a cohort of families from Mexico who had inherited ataxia, SCA10 was determined to be the second most common inherited ataxia, after SCA2 [Rasmussen et al 2000]. In the Brazilian states of Santa Catarina and Parana, SCA10 is the second most common SCA (after SCA3/MJD) [Teive et al 2011b]; however, in other regions of Brazil the relative prevalence of SCA10 among all SCAs is lower [Cintra et al 2014, de Castilhos et al 2014].

SCA10 has also been identified in Asian populations including Japanese [Naito et al 2017] and Chinese [Wang et al 2015].

Genetically Related (Allelic) Disorders

No other phenotypes with the pentanucleotide expansion of *ATXN10* have been reported.

Differential Diagnosis

Significant overlap exists in the clinical presentation of the SCAs (see Hereditary Ataxia Overview). All are characterized by ataxia, and some by other neurologic signs. Clinical presentation may vary even among affected members of the same family. SCA type cannot generally be determined by clinical or neuroimaging studies of single individuals.

Although the combination of "pure" cerebellar ataxia (lacking other motor or cranial nerve involvement) and seizures is typical for SCA10 and has seldom been seen in other autosomal dominant cerebellar ataxias, it is possible that in some families, SCA10 could be a pure cerebellar ataxia without seizures.

See Table 2 for notable clinical features of selected hereditary disorders in the differential diagnosis of SCA10.

Table 2. Hereditary Ataxia Disorders of Interest in the Differential Diagnosis of Spinocerebellar Ataxia Type 10

Gene	Disorder	MOI	Clinical Features
ATN1	DRPLA	AD	• Seizures • Conspicuous neurologic signs (e.g., extrapyramidal signs) not seen in SCA10
ATXN1	SCA1	AD	Pyramidal signs are more robust than in SCA10.

Table 2. continued from previous page.

Gene	Disorder	MOI	Clinical Features
ATXN2	SCA2	AD	Slow saccadic eye movements (not seen in SCA10)
ATXN3	SCA3	AD	More extensive extrapyramidal signs & involvement of lower motor neurons than in SCA10
ATXN7	SCA7	AD	Retinopathy w/macular degeneration (not seen in SCA10)
FMR1	FXTAS (See *FMR1*-Related Disorders.)	XL	Onset of ataxia is later (age >50 yrs) than that assoc w/SCA10 full-penetrance alleles, but may be earlier than that assoc w/SCA10 reduced-penetrance alleles.
FXN	Friedreich ataxia	AR	Sensorispinal ataxia
ITPR1	SCA15 (OMIM 606658)	AD	Head tremor (not seen in SCA10)
KCNC3	SCA13	AD	• Seizures may accompany relatively "pure" cerebellar ataxia. • Identified primarily in persons of Filipino & French ancestry
PPP2R2B	SCA12 (OMIM 604326)	AD	Head tremor (not seen in SCA10)
PRKCG	SCA14	AD	• Seizures may accompany relatively "pure" cerebellar ataxia. • Axial myoclonus • Identified primarily in persons of European & Japanese ancestry (most persons w/SCA10 are of Latin American / Amerindian ancestry)
TBP	SCA17	AD	• Seizures • Conspicuous neurologic signs (e.g., extrapyramidal signs; not seen in SCA10)

AD = autosomal dominant; AR = autosomal recessive; DRPLA = dentatorubral-pallidoluysian atrophy; FXTAS = fragile X-associated tremor/ataxia syndrome; MOI = mode of inheritance; SCA = spinocerebellar ataxia; XL = X-linked

SCA4. Nerve conduction velocity studies indicate the presence of polyneuropathy in some individuals with SCA10; however, unlike those with SCA4, they have few signs or symptoms [Flanigan et al 1996]. The molecular basis of SCA4 is unknown (OMIM 600223).

Neurocysticercosis. Because neurocysticercosis is one of the most common causes of seizures in Mexican Americans, it needs to be considered in individuals who do not have a strong family history of seizures. The MRI and CT findings of neurocysticercosis consist of either solid or cystic lesions associated with calcification and surrounding edema.

Alcoholic cerebellar ataxia and seizures. Chronic alcoholic cerebellar ataxia may be progressive and accompany cerebellar and cerebral atrophy on neuroimaging studies. Alcohol-related seizure disorders may also mimic clinical features of SCA10.

Management

Evaluations Following Initial Diagnosis

To establish the extent of disease and needs in an individual diagnosed with spinocerebellar ataxia type 10 (SCA10), the evaluations summarized in Table 3 (if not performed as part of the evaluation that led to the diagnosis) are recommended.

Table 3. Recommended Evaluations Following Initial Diagnosis in Individuals with SCA10

System/Concern	Evaluation	Comment
Neurologic	Brain MRI examination	The extent of cerebellar atrophy on serial MRI studies may be useful for documenting progression of disease.
	EEG	To evaluate for seizures
	Nerve conduction studies	In individuals w/clinical evidence of polyneuropathy
	Neuropsychological tests	In individuals w/problems in learning & social adaptation
	Speech pathology evaluation	In individuals w/: • Atypical dysarthria or communication problems • Frequent choking or severe dysphagia to assess aspiration risks
Other	Consultation w/clinical geneticist &/or genetic counselor	

Treatment of Manifestations

Table 4. Treatment of Manifestations in Individuals with SCA10

Manifestation/ Concern	Treatment	Considerations/Other
Seizures	Conventional anticonvulsants (e.g., levetiracetam, phenytoin, carbamazepine, valproic acid) achieve reasonable control.	• Uncontrolled seizures may lead to potentially fatal status epilepticus. • Occasional breakthrough seizures may occur.
Ataxia	• Canes & walkers • Modification of home w/grab bars, raised toilet seats, & ramps to accommodate motorized chairs	To prevent falling
	Intensive coordinative training	Not specifically studied in SCA10, but improves motor performance in those w/degenerative ataxias [Ilg et al 2009, Ilg et al 2010]
	Physical therapy	
	Weight control	To avoid difficulties w/ambulation & mobility
Dysphagia	Percutaneous placement of gastrostomy tube for severe dysphagia	For prevention of aspiration
	Vitamin supplementation	For maintenance of nutritional intake
Dysarthria	• Speech therapy • Communication devices (e.g., writing pads, computer-based devices)	
Upper-limb coordination issues	Weighted eating utensils & dressing hooks	To maintain sense of independence
Anxiety	Mild tranquilizers	May improve motor coordination

Surveillance

Table 5. Recommended Surveillance for Individuals with SCA10

System/Concern	Evaluation	Frequency
Neurologic	Clinical evaluation w/neurologist	Every 4-6 mos to identify early signs of potential complications & to adjust anticonvulsant treatments
	Video esophagrams can identify consistency of food least likely to trigger aspiration.	As needed in individuals w/dysphagia

Agents/Circumstances to Avoid

Alcohol and drugs known to adversely affect cerebellar functions should be avoided.

Falls should be avoided because resulting injuries may greatly compromise motor function and the ability to perform activities of daily living.

Any activities that are potentially dangerous to individuals with ataxia or epilepsy should be avoided, depending on the severity of the manifestations.

Evaluation of Relatives at Risk

See Genetic Counseling for issues related to testing of at-risk relatives for genetic counseling purposes.

Pregnancy Management

At-risk individuals should be aware of the possibility of inducing ataxia during pregnancy or puerperium [Teive et al 2011a].

Epilepsy should be managed during pregnancy according to the American Academy of Neurology Practice Parameter Update: Management issues for women with epilepsy (an evidence-based review).

Therapies Under Investigation

Clinical trials of troriluzol (BHV-4157), a prodrug of riluzole, have included individuals with SCA10.

Search ClinicalTrials.gov in the US and EU Clinical Trials Register in Europe for information on clinical studies for a wide range of diseases and conditions.

Other

Although taltirelin hydrate is widely used for symptomatic treatment of ataxia in Japan, it has never been used for individuals with SCA10.

Tremor-controlling drugs, such as beta-blockers and primidone, are ineffective for cerebellar tremors.

Genetic Counseling

Genetic counseling is the process of providing individuals and families with information on the nature, mode(s) of inheritance, and implications of genetic disorders to help them make informed medical and personal decisions. The following section deals with genetic risk assessment and the use of family history and genetic testing to clarify genetic status for family members; it is not meant to address all personal, cultural, or ethical issues that may arise or to substitute for consultation with a genetics professional. —ED.

Mode of Inheritance

Spinocerebellar ataxia type 10 (SCA10) is inherited in an autosomal dominant manner.

Risk to Family Members

Parents of a proband

- To date, all reported individuals diagnosed with SCA10 inherited an expanded *ATXN10* allele from a heterozygous, usually affected parent.
- *De novo* expansion of the *ATXN10* repeat has not been reported.
- Recommendations for the evaluation of apparently asymptomatic parents of a proband include neurologic evaluation and molecular genetic testing.
- The family history may appear to be negative because of failure to recognize the disorder in family members, early death of the parent before the onset of symptoms, or late onset of the disease in the transmitting parent. Therefore, an apparently negative family history cannot be confirmed unless appropriate clinical evaluation and/or molecular genetic testing has been performed on the parents of the proband.

Sibs of a proband. The risk to the sibs of a proband depends on the genetic status of the proband's parents:

- If one of the parents of a proband is known to have a full-penetrance allele, the risk to the sibs is 50%.
  - In general, the age of onset and progression of SCA10 in sibs who inherit an expansion are variable and cannot be reliably predicted by the family history or molecular genetic testing.
  - The presence of the *ATXN10* ATCCT interruption motif is known to be associated with a higher prevalence of epileptic seizures.
  - Interrupted repeat expansions are also known to be associated with anticipation but are accompanied by a paradoxic contraction in intergenerational repeat size [McFarland et al 2013]. In paternal transmissions, expansions containing the ATCCT interruption motif contract, whereas ATCCT interruption motif-negative expansions enlarge. In maternal transmission, expansion sizes are relatively stable either in ATCCT interruption motif-positive or ATCCT interruption motif-negative expansions.
- If one of the parents of a proband is known to have a reduced-penetrance allele but is clinically unaffected, the risk to the sibs of a proband may be anywhere between zero and 50% (see Establishing the Diagnosis, **Reduced-penetrance alleles**).
- If an expanded *ATXN10* allele cannot be detected in the leukocyte DNA of either parent, the risk to sibs is low but greater than that of the general population because of the possibility of germline mosaicism (no instances of germline mosaicism have been reported, although it remains a possibility).

Offspring of a proband. Each child of an individual with SCA10 has a 50% chance of inheriting the expansion; see **Sibs of a proband** for specific risk issues related to transmission of the expansion.

Other family members. The risk to other family members depends on the genetic status of the proband's parents: if a parent has an ATTCT repeat expansion, the parent's family members are at risk.

Related Genetic Counseling Issues

Considerations in families with an apparent *de novo* pathogenic variant. In general, when neither parent of a proband with an autosomal dominant condition has the pathogenic variant identified in the proband or clinical evidence of the disorder, the pathogenic variant is likely *de novo*. However, non-medical explanations including alternate paternity or maternity (e.g., with assisted reproduction) and undisclosed adoption could also be explored.

Family planning

- The optimal time for determination of genetic risk and discussion of the availability of prenatal/preimplantation genetic testing is before pregnancy.
- It is appropriate to offer genetic counseling (including discussion of potential risks to offspring and reproductive options) to young adults who are affected or at risk.

Predictive testing (i.e., testing of asymptomatic at-risk individuals)

- Predictive testing for at-risk relatives is possible once an ATTCT repeat expansion in *ATXN10* has been identified in an affected family member.
- Potential consequences of such testing (including, but not limited to, socioeconomic changes and the need for long-term follow-up and evaluation arrangements for individuals with a positive test result) as well as the capabilities and limitations of predictive testing should be discussed in the context of formal genetic counseling prior to testing.

Predictive testing in minors (i.e., testing of asymptomatic at-risk individuals younger than age 18 years)

- For asymptomatic minors at risk for typically adult-onset conditions for which early treatment would have no beneficial effect on disease morbidity and mortality, predictive genetic testing is considered inappropriate, primarily because it negates the autonomy of the child with no compelling benefit. Further, concern exists regarding the potential unhealthy adverse effects that such information may have on family dynamics, the risk of discrimination and stigmatization in the future, and the anxiety that such information may cause.
- For more information, see the National Society of Genetic Counselors position statement on genetic testing of minors for adult-onset conditions and the American Academy of Pediatrics and American College of Medical Genetics and Genomics policy statement: ethical and policy issues in genetic testing and screening of children.

In a family with an established diagnosis of SCA10, it is appropriate to consider testing of symptomatic individuals regardless of age.

Prenatal Testing and Preimplantation Genetic Testing

Once an ATTCT repeat expansion in *ATXN10* been identified in an affected family member, prenatal testing for a pregnancy at 50% risk for SCA10 and preimplantation genetic testing are possible. Note: Age of onset, severity, and progression of SCA10 are variable and cannot be reliably predicted by the family history or prenatal molecular genetic testing.

Differences in perspective may exist among medical professionals and within families regarding the use of prenatal testing. While most centers would consider use of prenatal testing to be a personal decision, discussion of these issues may be helpful.

Resources

GeneReviews staff has selected the following disease-specific and/or umbrella support organizations and/or registries for the benefit of individuals with this disorder and their families. GeneReviews is not responsible for the information provided by other organizations. For information on selection criteria, click here.

- **National Ataxia Foundation**
  Phone: 763-553-0020
  Fax: 763-553-0167
  Email: naf@ataxia.org

www.ataxia.org

- **NCBI Genes and Disease**
  Spinocerebellar ataxia
- **Ataxia UK**
  United Kingdom
  Phone: 0800 995 6037; +44 (0) 20 7582 1444 (from abroad)
  Email: help@ataxia.org.uk
  www.ataxia.org.uk
- **euro-ATAXIA (European Federation of Hereditary Ataxias)**
  United Kingdom
  Email: lporter@ataxia.org.uk
  www.euroataxia.org
- **Spanish Ataxia Federation (FEDAES)**
  Spain
  Phone: 601 037 982
  Email: info@fedaes.org
  fedaes.org

Molecular Genetics

Information in the Molecular Genetics and OMIM tables may differ from that elsewhere in the GeneReview: tables may contain more recent information. —ED.

Table A. Spinocerebellar Ataxia Type 10: Genes and Databases

Gene	Chromosome Locus	Protein	Locus-Specific Databases	HGMD	ClinVar
ATXN10	22q13.31	Ataxin-10	ATXN10 database	ATXN10	ATXN10

Data are compiled from the following standard references: gene from HGNC; chromosome locus from OMIM; protein from UniProt. For a description of databases (Locus Specific, HGMD, ClinVar) to which links are provided, click here.

Table B. OMIM Entries for Spinocerebellar Ataxia Type 10 (View All in OMIM)

603516	SPINOCEREBELLAR ATAXIA 10; SCA10
611150	ATAXIN 10; ATXN10

Molecular Pathogenesis

ATXN10 is ubiquitously expressed. Strong expression is observed in brain, heart, muscle, kidney, and liver [Wakamiya et al 2006]. The normal physiologic function of ataxin-10 is largely unknown; however, ataxin-10 deficiency by small interfering RNA (siRNA) caused apoptosis of cerebellar neurons in primary cell culture [März et al 2004]. Based on protein interaction studies, ataxin-10 is thought to play a role in survival and differentiation of neurons or neuron-like cells [Andrali et al 2005, Waragai et al 2006].

There is evidence to suggest that expanded non-coding intron 9 ATTCT repeats do not interfere with the transcription and post-transcriptional processing of *ATXN10* [Wakamiya et al 2006]. Consequently, the level of processed mRNA from mutated *ATXN10* is unaltered.

Mechanism of disease causation. Data from in vitro and animal model systems support a RNA-gain-of-function hypothesis as the cause of SCA10. Expression of expanded AUUCU repeats results in RNA foci

containing AUUCU repeats [White et al 2010]. For a review on the cytotoxic effects of expanded RNA repeats, see Zhang & Ashizawa [2017], Nguyen et al [2019], and Rodriguez & Todd [2019].

Genetically altered mice heterozygous for ataxin-10 deficiency exhibit no disease phenotype, while homozygous deficiency of ataxin-10 is embryonically lethal [Wakamiya et al 2006]. In addition, an individual with a balanced translocation that disrupts *ATXN10* showed no phenotype, suggesting that haploinsufficiency is an unlikely mechanism for SCA10 [Keren et al 2010].

***ATXN10*-specific laboratory technical considerations.** SCA10 is caused by an intronic expansion of ATTCT repeats within *ATXN10*. ATTCT-repeat-primed PCR and Southern blot analysis have been used to reliably identify and size the pathogenic ATTCT repeat insertion (see Establishing the Diagnosis).

Expanded alleles are unstable in somatic tissues, appearing as "smeared" expanded alleles with multiple distinct expansion alleles on PCR and Southern blot analyses.

The expanded ATTCT repeat shows repeat-size instability when it is transmitted from generation to generation [Matsuura et al 2004]. The pattern of the instability depends on the sex of the transmitting parent:

- During paternal transmission, the expanded ATTCT repeats are highly unstable.
- During maternal transmission, the expanded ATTCT repeats undergo no change or changes of a smaller magnitude.

Table 6. Notable *ATXN10* Pathogenic Variants

Reference Sequences	Variant Classification	DNA Nucleotide Change	Predicted Protein Change	Comment [Reference]
NM_013236.2 NP_037368.1	**Benign**	c.1430+54822_54826ATTCT(10_32) [1, 2] (10 to 32 ATTCT repeats)	None	[Matsuura et al 2000, Matsuura et al 2006]
	Pathogenic	c.1430+54822_54826ATTCT(280) [3]	None	May be alleles w/reduced or no penetrance [Alonso et al 2006, Matsuura et al 2006, Raskin et al 2007]
		c.1430+54822_54826ATTCT(360_370 (360 to 370 ATTCT repeats)	None	
		c.1430+54822_54826ATTCT(400_760) (400 to 760 ATTCT repeats)	None	
		c.1430+54822_54826ATTCT(850) (850 ATTCT repeats)	None	
		c.1430+54822_54826ATTCT(800_4500) [4] (800 to 4500 ATTCT repeats)	None	Alleles w/full penetrance. The lower end of the fully penetrant allele range of 800 is not well defined; overlap w/reduced-penetrance alleles exists [Matsuura et al 2000].

Variants listed in the table have been provided by the authors. *GeneReviews* staff have not independently verified the classification of variants.
GeneReviews follows the standard naming conventions of the Human Genome Variation Society (varnomen.hgvs.org). See Quick Reference for an explanation of nomenclature.
1. Sequence analysis of *ATXN10* alleles ranging from 11 to 16 repeats showed tandem ATTCT repeats without interruptions [Matsuura et al 2000].
2. About 70% of large normal alleles (≥17 repeats), which comprise about 7% of normal alleles, have ATTGT-TTTCT or TTTCT interruptions at the second-to-last repeat [Matsuura et al 2006].
3. The sequence of one allele of 280 ATTCT repeats with apparent reduced penetrance showed a complex pattern of interruptions, including multiple repetitive ATGCT repeats at the 5' end of the expansion and ATTCTAT septanucleotide repeats at the 3' end.
4. Limited sequencing of fully expanded ATTCT repeat alleles showed interruptions by multiple ATTTTCTs and ATATTCTs or uninterrupted ATTCTs, depending on the family from which the mutated allele was obtained [Matsuura et al 2006].

Chapter Notes

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