

Neurology

Movement Disorders

Phenotype-Directed Testing Services for Hereditary Ataxias

Spinocerebellar Ataxia

DRPLA

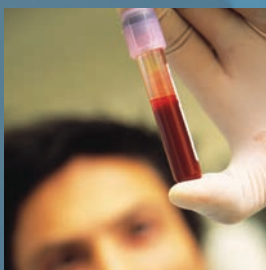
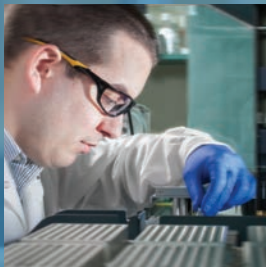
Episodic Ataxia

Friedreich's Ataxia

Ataxia-Telangiectasia (ATM)

Ataxia with Vitamin E Deficiency

Oculomotor Apraxia-Ataxia



Ataxia Testing that Impacts Quality of Life.



Phenotype-Directed Testing Streamlines the Path to Diagnosis

Providing a conclusive diagnosis for a patient presenting with ataxia can be life-changing. Many types of acquired or non-genetic forms of ataxia can be treated and resolved. In the cases of hereditary ataxia, establishing a molecular cause can provide both you and your patient with a basis for planning support services that can extend mobility and quality of life. It can also eliminate the need for further testing.

Need to Know

At Athena Diagnostics®, we understand how a definitive diagnosis can bring confidence to life-altering decisions.

“What’s causing my symptoms?”

“What treatments can help me lead a more normal life?”

“What are the chances I will pass this on to my children?”

A Range of Possibilities

Symptoms of ataxia can present almost identically yet yield widely variable prognoses. Many hereditary ataxias have overlapping phenotypes that are challenging to differentiate clinically.

The differential diagnosis of hereditary ataxia can include acquired, non-genetic causes such as¹:

- Alcoholism
- Vitamin deficiencies
- Multiple sclerosis
- Vascular disease
- Primary or metastatic tumors
- Paraneoplastic diseases associated with occult carcinoma of the ovary, breast, or lung

Genetic Testing for Ataxia Optimizes Efficiency, Economy, and Certainty

Only molecular genetic testing can provide a conclusive diagnosis, ending the diagnostic odyssey that is often frustrating, time-consuming, and expensive. Ataxia need not be a diagnosis by exclusion.

At Athena Diagnostics, we understand that you and your patients want an efficient, economical way to obtain definitive answers. Our phenotype-directed genetic testing approach streamlines the diagnostic process for hereditary ataxia.

A Unique Approach

Guided by the clinical workup, including family history and phenotype, ataxia genetic testing from Athena Diagnostics is organized by prevalence — starting with the genes most likely to cause disease — progressing to the next levels of testing only as needed to achieve a positive result. Backed by clinical guidelines and peer-published diagnostic algorithms, this unique approach to genetic testing allocates only the resources necessary to detect the causative mutation, avoiding over-testing and unnecessary services and expense — reducing time to diagnosis and overall cost.

The More You Know, The More You Can Do.

Illuminating Answers

At Athena Diagnostics, we know that finding the answer — identifying the ataxia-causing mutation and establishing the cause of disease — can empower confidence in treatments, lifestyle modifications, and personal family planning decisions. Our testing for ataxia is designed with the physician, patient, and payer in mind — for efficiency, economy, and certainty.

Patient presents with imbalance, progressive gait and limb incoordination, dysarthria, and eye disturbances¹

Exclude non-genetic causes: alcoholism, vitamin deficiencies, multiple sclerosis, vascular disease, primary or metastatic tumors, paraneoplastic diseases

Establish family history of ataxia

Consider patient phenotype

Test for highly prevalent disease-causing genes

Test for less prevalent disease-causing genes

Continue testing as necessary to detect the causative mutation



Rely on Athena Diagnostics, the Leader in Genetic Testing for Neurological Disorders.

Athena Diagnostics Phenotype-Directed Testing for Hereditary Ataxias

Autosomal Dominant Family History							
Phenotype	Ataxia with Autosomal Dominance Inheritance	Retinal Degeneration	Pyramidal, Extra-pyramidal, or Peripheral Neuropathy			Chorea, Dementia, Myoclonus, Seizures	Pure Cerebellar Ataxia
Phenotype Characterization	Slowly Progressive, often associated with cerebellar atrophy as seen from brain imaging studies	Retinal Degeneration, Ophthalmoplegia, Pyramidal Signs	Dementia, Nystagmus, Slow Saccades, Pyramidal Signs, Neuropathy	Dementia, Slow Saccades, Hyporeflexia, Amyotrophy, Neuropathy, Myoclonus	Nystagmus, Diplopia, Ophthalmoplegia, Red-Lid Retraction, Parkinsonism, Spasticity Neuropathy	Chorea, Seizures, Dementia, Myoclonus. DRPLA is often confused with Huntington Disease.	Downbeat Positioning Nystagmus, Sometimes Episodic Ataxia at Onset, Double Vision, Pyramidal Signs, Deep Sensory Loss, Migraine
Age at Onset	Early Childhood to Elderly	0–76	4–74	6–67	5–65	10–59	19–77



STEP 1

Test	6900, Ataxia, Complete Dominant Evaluation	6901, Ataxia, Common Repeat Expansion Evaluation	677, SCA7 (ATXN7) Repeat Expansion Test	371, SCA1 (ATXN1) Repeat Expansion Test	672, SCA2 (ATXN2) Repeat Expansion Test	105, SCA3 (MJD/ATXN3) Repeat Expansion Test	401, DRPLA (ATN1) Repeat Expansion Test	373, SCA6 (CACNA1A) Repeat Expansion Test
Genes Tested	26 Genes Tested	8 Genes Tested	1 Gene Tested	1 Gene Tested	1 Gene Tested	1 Gene Tested	1 Gene Tested	1 Gene Tested
Gene Prevalence*	Includes all genes known to cause AD SCA, per OMIM	50-60%/AD SCA	5%/SCA	6%/SCA	15%/SCA	21%/SCA	Occurs mostly in those with Asian origin	15%/SCA



STEP 2

Test	6903, Ataxia, Supplemental Dominant Evaluation							
Genes Tested		16 Genes Tested						
Gene Prevalence*		This step completes testing of all known AD SCA genes, per OMIM						

With an intuitive selection of tests—conveniently arranged by family history and patient phenotype—ataxias with the highest prevalence are tested first. Subsequent testing can progress to the next levels of gene prevalence on an as-needed basis until the ataxia-causing mutation is found. Test organization and content is derived from expert guidelines, publications, and consensus statements on the diagnosis of hereditary ataxias.^{1,2,3,4}

	Autosomal Recessive Family History					No Family History
Episodic Ataxia	Ataxia with Recessive Inheritance	None, or: Normal MRI, Peripheral Sensory Neuropathy	Oculomotor Apraxia, Low Albumin, High Cholesterol	Low Vitamin E Levels	High Alpha-Fetoprotein	No Associated Features
Recurrent Ataxia, Giddiness, Vertigo			Cerebellar Ataxia, Sensorimotor Neuropathy, Nystagmus, Extrapyrimalidal Signs, Mild Cognitive Impairment	Progressive Gait and Limb Ataxia, Dysarthria, Absent Deep Tendon Reflexes, Sensory Loss, Pyramidal Weakness	Typical High Serum Alpha Fetoprotein, Cerebellar Ataxia, Ocular Apraxia, Telangiectasias, Immune Defects, Predisposition to Malignancy	
Up to 60	2-55	2-55	1-30	2-52	1-4	



6920, Episodic Ataxia Evaluation	6910, Ataxia, Complete Recessive Evaluation	349, Friedreich's Ataxia (FXN) Evaluation	6912, Oculomotor Apraxia-Ataxia Advanced Sequencing Evaluation	283, Ataxia with Vitamin E Deficiency (AVED) TTPA DNA Sequencing Test	353, Ataxia-Telangiectasia (ATM) Evaluation	6930, Ataxia, Comprehensive Evaluation	6901, Ataxia, Common Repeat Expansion Evaluation	349, Friedreich's Ataxia (FXN) Evaluation
4 Genes Tested	18 Genes Tested	1 Gene Tested	2 Genes Tested	1 Gene Tested	1 Gene Tested	43 Genes Tested	8 Genes Tested	1 Gene Tested
	Includes all genes known to cause AR SCA, per OMIM	Most Common AR Ataxia, 1-2:50,000	Unknown Prevalence	Rare Prevalence	1:40,000 to 1:100,000	Includes all genes known to cause AD and AR SCA, per OMIM	50-60%/AD SCA	Most Common AR Ataxia, 1-2:50,000

		6911, Ataxia, Supplemental Recessive Evaluation					6903, Ataxia, Supplemental Dominant Evaluation	6911, Ataxia, Supplemental Recessive Evaluation
		17 Genes Tested					16 Genes Tested	17 Genes Tested
		Includes remaining genes known to cause AR SCA, per OMIM						Includes remaining genes known to cause AR SCA, per OMIM

- = Athena Diagnostics Ataxia Evaluations
- = Athena Diagnostics Single Gene Tests

* Gene Prevalence: Global. Can vary by ethnic background.
Abbreviations: SCA, Spinocerebellar Ataxia; AD, Autosomal Dominant; AR, Autosomal Recessive; OMIM, Online Mendelian Inheritance in Man.

Genes Tested for Hereditary Ataxia Disorders: Spinocerebellar Ataxia Evaluations from Athena Diagnostics

Evaluation	Gene	Disease
349 Friedreich's Ataxia	<i>FXN</i> ##	Friedreich's Ataxia
353 Ataxia-Telangiectasia	<i>ATM</i> #	Ataxia-Telangiectasia
6900 Ataxia, Complete Dominant Evaluation	<i>ATXN1*</i>	Spinocerebellar Ataxia 1
	<i>ATXN2*</i>	Spinocerebellar Ataxia 2
	<i>ATXN3*</i>	Spinocerebellar Ataxia 3
	<i>CACNA1A*</i>	Spinocerebellar Ataxia 6
	<i>ATXN7*</i>	Spinocerebellar Ataxia 7
	<i>TBP*</i>	Spinocerebellar Ataxia 17
	<i>ATXN8OS*</i>	Spinocerebellar Ataxia 8
	<i>ATXN10*</i>	Spinocerebellar Ataxia 10
	<i>PPP2R2B*</i>	Spinocerebellar Ataxia 12
	<i>ATN1*</i>	Dentatorubral-Pallidoluysian Atrophy (DRPLA)
	<i>AFG3L2</i>	Spinocerebellar Ataxia 28
	<i>KCNC3</i>	Spinocerebellar Ataxia 13
	<i>PRKCG</i>	Spinocerebellar Ataxia 14
	<i>SPTBN2</i>	Spinocerebellar Ataxia 5
	<i>EEF2</i>	Spinocerebellar Ataxia 26
	<i>FGF14</i>	Spinocerebellar Ataxia 27
	<i>ITPR1</i>	Spinocerebellar Ataxia 29
	<i>KCND3</i>	Spinocerebellar Ataxia 19
	<i>PDYN</i>	Spinocerebellar Ataxia 23
	<i>TGM6</i>	Spinocerebellar Ataxia 35
	<i>TTBK2</i>	Spinocerebellar Ataxia 11
<i>VAMP1</i>	Spastic Ataxia 1	
<i>KCNA1</i>	Episodic Ataxia, Type 1	
<i>CACNB4</i>	Episodic Ataxia, Type 5	
<i>SLC1A3</i>	Episodic Ataxia, Type 6	
<i>CACNA1A</i>	Episodic Ataxia, Type 2	
6901 Ataxia, Common Repeat Expansion Evaluation	<i>ATXN1*</i>	Spinocerebellar Ataxia 1
	<i>ATXN2*</i>	Spinocerebellar Ataxia 2
	<i>ATXN3*</i>	Spinocerebellar Ataxia 3
	<i>CACNA1A*</i>	Spinocerebellar Ataxia 6
	<i>ATXN7*</i>	Spinocerebellar Ataxia 7
	<i>TBP*</i>	Spinocerebellar Ataxia 17
	<i>ATXN8OS*</i>	Spinocerebellar Ataxia 8
<i>ATXN10*</i>	Spinocerebellar Ataxia 10	
6903 Ataxia, Supplemental Dominant Evaluation	<i>AFG3L2</i>	Spinocerebellar Ataxia 28
	<i>KCNC3</i>	Spinocerebellar Ataxia 13
	<i>PRKCG</i>	Spinocerebellar Ataxia 14
	<i>SPTBN2</i>	Spinocerebellar Ataxia 5
	<i>EEF2</i>	Spinocerebellar Ataxia 26
	<i>FGF14</i>	Spinocerebellar Ataxia 27
	<i>ITPR1</i>	Spinocerebellar Ataxia 29
	<i>KCND3</i>	Spinocerebellar Ataxia 19
	<i>PDYN</i>	Spinocerebellar Ataxia 23
	<i>TGM6</i>	Spinocerebellar Ataxia 35
	<i>TTBK2</i>	Spinocerebellar Ataxia 11
	<i>VAMP1</i>	Spastic Ataxia 1
	<i>KCNA1</i>	Episodic Ataxia, Type 1
	<i>CACNB4</i>	Episodic Ataxia, Type 5
	<i>SLC1A3</i>	Episodic Ataxia, Type 6
	<i>CACNA1A</i>	Episodic Ataxia, Type 2

Evaluation	Gene	Disease
6910 Ataxia, Complete Recessive Evaluation	<i>FXN</i> ##	Friedreich's Ataxia
	<i>APTX</i>	Ataxia with Oculomotor Apraxia, Type 1, or Early Onset with Oculomotor Apraxia and Hypoalbuminemia
	<i>ATM</i> #	Ataxia-Telangiectasia
	<i>SETX</i>	Ataxia with Oculomotor Apraxia, Type 2
	<i>TTPA</i>	Ataxia with Vitamin E Deficiency
	<i>ADCK3</i>	Coenzyme Q10 Deficiency, Primary, 4
	<i>AFG3L2</i>	Spastic Ataxia 5
	<i>ANO10</i>	Spinocerebellar Ataxia, AR 10
	<i>FLVCR1</i>	Ataxia, Posterior Column, with Retinitis Pigmentosa
	<i>GRM1</i>	Spinocerebellar Ataxia, AR 13
	<i>MRE11A</i>	Ataxia-Telangiectasia-Like Disorder
	<i>MTPAP</i>	Spastic Ataxia 4
	<i>SACS</i>	Spastic Ataxia, Charlevoix-Saguenay Type
	<i>SYNE1</i>	Spinocerebellar Ataxia, AR 8
	<i>SYT14</i>	Spinocerebellar Ataxia, AR 11
	<i>TDP1</i>	Spinocerebellar Ataxia, AR, with Axonal Neuropathy
	<i>SIL1</i>	Marinesco-Sjogren Syndrome
<i>POLG</i>	Mitochondrial Recessive Ataxia Syndrome	
6911 Ataxia, Supplemental Recessive Evaluation	<i>APTX</i>	Ataxia with Oculomotor Apraxia, Type 1, or Early Onset with Oculomotor Apraxia and Hypoalbuminemia
	<i>ATM</i> #	Ataxia-Telangiectasia
	<i>SETX</i>	Ataxia with Oculomotor Apraxia, Type 2
	<i>TTPA</i>	Ataxia with Vitamin E Deficiency
	<i>ADCK3</i>	Coenzyme Q10 Deficiency, Primary, 4
	<i>AFG3L2</i>	Spastic Ataxia 5
	<i>ANO10</i>	Spinocerebellar Ataxia, AR 10
	<i>FLVCR1</i>	Ataxia, Posterior Column, with Retinitis Pigmentosa
	<i>GRM1</i>	Spinocerebellar Ataxia, AR 13
	<i>MRE11A</i>	Ataxia-Telangiectasia-Like Disorder
	<i>MTPAP</i>	Spastic Ataxia 4
	<i>SACS</i>	Spastic Ataxia, Charlevoix-Saguenay Type
<i>SYNE1</i>	Spinocerebellar Ataxia, AR 8	
<i>SYT14</i>	Spinocerebellar Ataxia, AR 11	
<i>TDP1</i>	Spinocerebellar Ataxia, AR, with Axonal Neuropathy	
<i>SIL1</i>	Marinesco-Sjogren Syndrome	
<i>POLG</i>	Mitochondrial Recessive Ataxia Syndrome	
6912 Oculomotor Apraxia-Ataxia	<i>APTX</i>	Ataxia with Oculomotor Apraxia, Type 1
	<i>SETX</i>	Ataxia with Oculomotor Apraxia, Type 2
6920 Episodic Ataxia	<i>CACNB4</i>	Episodic Ataxia, Type 5
	<i>KCNA1</i>	Episodic Ataxia, Type 1
	<i>SLC1A3</i>	Episodic Ataxia, Type 6
	<i>CACNA1A</i>	Episodic Ataxia, Type 2

Genes are analyzed using next generation sequencing unless otherwise noted. * Repeat expansion analysis, # Sequencing and duplication/deletion, ## Sequencing and repeat expansion

Evaluation	Gene	Disease
6930 Ataxia, Comprehensive Evaluation	ATXN1*	Spinocerebellar Ataxia 1
	ATXN2*	Spinocerebellar Ataxia 2
	ATXN3*	Spinocerebellar Ataxia 3
	CACNA1A*	Spinocerebellar Ataxia 6
	ATXN7*	Spinocerebellar Ataxia 7
	TBP*	Spinocerebellar Ataxia 17
	ATXN80S*	Spinocerebellar Ataxia 8
	ATXN10*	Spinocerebellar Ataxia 10
	PPP2R2B*	Spinocerebellar Ataxia 12
	ATN1*	Dentatorubral-Pallidoluysian Atrophy (DRPLA)
	AFG3L2	Spinocerebellar Ataxia 28
	KCNC3	Spinocerebellar Ataxia 13
	PRKCG	Spinocerebellar Ataxia 14
	SPTBN2	Spinocerebellar Ataxia 5
	EEF2	Spinocerebellar Ataxia 26
	FGF14	Spinocerebellar Ataxia 27
	ITPR1	Spinocerebellar Ataxia 29
	KCND3	Spinocerebellar Ataxia 19
	PDYN	Spinocerebellar Ataxia 23
	TGM6	Spinocerebellar Ataxia 35
	TTBK2	Spinocerebellar Ataxia 11
	VAMP1	Spastic Ataxia 1
	KCNA1	Episodic Ataxia, Type 1
	CACNB4	Episodic Ataxia, Type 5
	SLC1A3	Episodic Ataxia, Type 6
	CACNA1A	Episodic Ataxia, Type 2
	FXN **	Friedreich's Ataxia
	APTX	Ataxia with Oculomotor Apraxia, Type 1, or Early Onset with Oculomotor Apraxia and Hypoalbuminemia
	ATM *	Ataxia-Telangiectasia
	SETX	Ataxia with Oculomotor Apraxia, Type 2
	TPPA	Ataxia with Vitamin E Deficiency
	ADCK3	Coenzyme Q10 Deficiency, Primary, 4
	AFG3L2	Spastic Ataxia 5
	ANO10	Spinocerebellar Ataxia, AR 10
	FLVCR1	Ataxia, Posterior Column, with Retinitis Pigmentosa
	GRM1	Spinocerebellar Ataxia, AR 13
	MRE11A	Ataxia-Telangiectasia-Like Disorder
	MTPAP	Spastic Ataxia 4
	SACS	Spastic Ataxia, Charlevoix-Saguenay Type
	SYNE1	Spinocerebellar Ataxia, AR 8
	SYT14	Spinocerebellar Ataxia, AR 11
	TDP1	Spinocerebellar Ataxia, AR, with Axonal Neuropathy
	SIL1	Marinesco-Sjogren Syndrome
	POLG	Mitochondrial Recessive Ataxia Syndrome

Athena Insight™ Goes Beyond the Results.

As a powerful bioinformatic service, Athena Insight goes beyond the results. Over 14,000 pathogenicity assessments on 11,000 unique variants have been successfully performed to date. Pre- or post-testing, we encourage clinicians to confer with one of our board-certified geneticists, researchers, or doctors.



Carol A. Hoffman, Ph.D., M.S., LGC
Genetic Counselor

Our scientists and geneticists are some of the industry's top-tier thought leaders, clinicians and scholars. Many have lectured around the world, sharing their observations and discoveries on variant investigation and interpretation with colleagues in government and industry. Our high-touch team will discuss an individual's genetic disposition, variant investigation findings, or recent discoveries that affect you and your patient.

With the commitment of the Athena Insight team behind you, you can see and do more for your patients. Genetic counselors are available at 1-800-394-4493 or Genetics@AthenaDiagnostics.com.

Each variant identified is plotted within a 7-category scale that assesses its relative likelihood of pathogenicity.

Result reports are clear and concise, with a pathogenicity assessment and complete sourced synopsis that clinicians can review with patients.

SETX DNA Sequencing Test

This test identified a variant of unknown clinical significance (VUS) in the SETX gene.

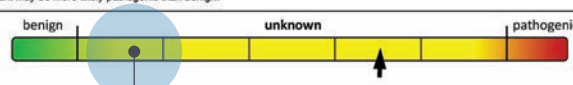
INTERPRETIVE RESULTS TABLE						
VUS	Gene/Test	Technical Result	Mutation Type	Inheritance	Clinical Relevance	Pub Med ID
	SETX	c.820A>G; p.Met274Val	Heterozygous Missense	Autosomal	Unknown	23566282

No other abnormal variants were detected.

Comments: The variant detected in this analysis has not been definitively demonstrated to be associated with ataxia-oculomotor apraxia-2 (AOA2). This result does not confirm the diagnosis since the variant detected is of unknown clinical significance.

Athena Insight pathogenicity assessment

This test detected a DNA sequence variant of unknown clinical significance (SETX c.820 A>G), but the following data indicate that this variant may be more likely pathogenic than benign:



Variant:	SETX c.820 A>G (p.Met274Val)
Segregation Analysis:	Inconclusive
Co-occurrence:	Not enough information
General pop. freq.:	0.000 (0 in 13006 chromosomes)
Amino Acid Conservation:	Highly conserved across species
Grantham Score:	21 [0-215] (conservative difference)
SIFT:	Predicted NOT Tolerated
PolyPhen-2.2.2 (HumVar):	Probably Damaging
Protein Domain:	N-terminal domain (putative protein interaction domain)
dbSNP Reference:	None

- Published research does not provide sufficient evidence for classification of this variant as pathogenic or benign.
- Analysis of this variant's possible segregation with disease was inconclusive. Within the only affected family known to have the variant, a total of two affected individuals were genotyped, and both of these individuals have the variant. Consistent with pathogenicity, the variant was found in combination with a known recessive disease mutation (in the same gene) in both of the affected individuals. Also within this family, a total of 2 unaffected family members were genotyped, and of these, one individual does not have the variant and one unaffected individual has the variant in the heterozygous state.
- There is not enough information regarding co-occurrence with known disease variants among index cases to be useful in characterizing this variant.
- Both SIFT and PolyPhen-2.2.2 (HumVar) predict that this variant is pathogenic.

References:
Datta, N, et al. (2013) Int J Neurosci 123(9):670-3. (PMID: 23566282)
Exome Sequencing Project, <http://evs.gs.washington.edu/EVS/>

athena Insight

Genetic Test Menu for Ataxia from Athena Diagnostics

Test Code	Test Name	Whole Blood, Lavender Top Tube/Minimum Volume (mL)*	Turnaround Time
6900	Ataxia, Complete Dominant Evaluation; CPT Codes 81401(10), 81403(1), 81406(2), 81407(1), 81408(1), 81479(1)	8 mL	28–42 Days
6901	Ataxia, Common Repeat Expansion Evaluation; CPT Code 81401(8)	6 mL	28–42 Days
6903	Ataxia, Supplemental Dominant Evaluation; CPT Codes 81403(1), 81406(2), 81407(1), 81408(1), 81479(1)	6 mL	28–42 Days
6910	Ataxia, Complete Recessive Evaluation; CPT Codes 81401(1), 81404(2), 81405(2), 81406(3), 81408(1), 81479(1)	6 mL	28–42 Days
6911	Ataxia, Supplemental Recessive Evaluation; CPT Codes 81404(1), 81405(2), 81406(2), 81408(1), 81479(1)	6 mL	28–42 Days
6912	Oculomotor Apraxia-Ataxia Advanced Sequencing Evaluation; CPT Codes 81405(1), 81406(1)	6 mL	28–42 Days
6920	Episodic Ataxia Evaluation; CPT Code 81479(1)	6 mL	28–42 Days
6930	Ataxia, Comprehensive Evaluation; CPT Codes 81401(11), 81403(1), 81404(2), 81405(2), 81406(5), 81407(1), 81408(2), 81479(1)	8 mL	28–42 Days
119	Friedreich's Ataxia (<i>FXN</i>) Repeat Expansion Test; CPT Code 81401(1)	6 mL	28–42 Days
283	Ataxia with Vitamin E Deficiency (<i>AVED</i>) <i>TTPA</i> DNA Sequencing Test; CPT Code 81404(1)	6 mL	28–42 Days
285	SCA12 (<i>PPP2R2B</i>) Repeat Expansion Test; CPT Code 81401(1)	6 mL	28–42 Days
348	Friedreich's Ataxia (<i>FXN</i>) DNA Sequencing Test; CPT Code 81401(1)	6 mL	28–42 Days
349	Friedreich's Ataxia (<i>FXN</i>) Evaluation; CPT Codes 81401(1), 81404(1)	6 mL	28–42 Days
353	Ataxia-Telangiectasia (<i>ATM</i>) Evaluation; CPT Code 81408(2)	6 mL	28–42 Days
371	SCA1 (<i>ATXN1</i>) Repeat Expansion Test; CPT Code 81401(1)	6 mL	28–42 Days
373	SCA6 (<i>CACNA1A</i>) Repeat Expansion Test; CPT Code 81401(1)	6 mL	28–42 Days
384	SCA8 (<i>ATXN8OS</i>) Repeat Expansion Test; CPT Code 81401(1)	6 mL	28–42 Days
387	SCA10 (<i>ATXN10</i>) Repeat Expansion Test; CPT Code 81401(1)	6 mL	28–42 Days
388	SCA17 (<i>TBP</i>) Repeat Expansion Test; CPT Code 81401(1)	6 mL	28–42 Days
401	DRPLA (<i>ATN1</i>) Repeat Expansion Test; CPT Code 81401(1)	6 mL	28–42 Days
672	SCA2 (<i>ATXN2</i>) Repeat Expansion Test; CPT Code 81401(1)	6 mL	28–42 Days
677	SCA7 (<i>ATXN7</i>) Repeat Expansion Test; CPT Code 81401(1)	6 mL	28–42 Days

* Preferred volume is 8 mL

Client Services Representatives are available from 8:30 am to 6:30 pm Eastern Time (U.S.). Customers in the U.S. and Canada please call toll free **1-800-394-4493** or visit us online at AthenaDiagnostics.com.



References

1. Jayadev S, Bird TD. Hereditary ataxias: overview. *Genetics in Med*. 2013;15:673-683. 2. van de Warrenburg BPC, van Gaalen J, Boesch S, et al. EFNS/ENS Consensus on the diagnosis and management of chronic ataxias in adulthood. *Eur J Neurol*. 2014. Doi:10.1111/ene.12341. 3. Gasser T, Finsterer J, Baets J, et al. EFNS guidelines on the molecular diagnosis of ataxia and spastic paraplegias. *Eur J Neurol*. 2010;17:179-188. 4. Hereditary Ataxia Overview. Revision February 27, 2014. National Center for Biotechnology Information, National Institutes of Health. <http://www.ncbi.nlm.nih.gov/books/NBK1138>. Accessed March 19, 2015.

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