Spinocerebellar Ataxia and Related Disorders Panel

DISORDER ALSO KNOWN AS
SCA, Hereditary Ataxia, Spinocerebellar Degeneration

PANEL GENE LIST
AARS2, ABCD1, AFG3L2, ANO10, APTX, ATM, CACNA1A, CACNA1G*, CLN5*, CYP27A1, CYP7B1, C10ORF2*, DNMT1, EEF2, ELOVL4, ELOVL5, FGF14, GNB4, GRID2, GRM1, HEXA, HEXB*, ITPR1*, KCNA1*, KCNC3, KCND3, KIAA0196, KIF5A, MME, NOTCH3*, PDYN, PEX7*, PMM2*, PNKP, PNPLA6, POLG, POLR3A, PRKCG, PRNP, RRM2B*, SACS, SETX, SLC20A2, SPG7, SPG11, SPTBN2, STUB1, SYNE1, SYT14, TDP1, TGM6, TMEM240**, TPP1*, TTBK2, TTPA, ZFYVE26*

* Sequence analysis only of theAARS2, CACNA1G, CLN5, C10ORF2, HEXB, ITPR1, KCNA1, NOTCH3, PEX7, PMM2, RRM2B, and TPP1genes

** Only whole gene deletions or duplications may be detected for TMEM240 gene

CLINICAL FEATURES
The Spinocerebellar Ataxias (SCAs) are a clinically and genetically heterogeneous group of neurodegenerative disorders characterized by progressive incoordination of movements. Clinical features include uncoordinated gait and hand movements, dysarthria, dysphagia, nystagmus and are usually associated with atrophy of the cerebullum.[1] In addition to ataxia, the disorders evaluated on this panel can also present with oculomotor apraxia and/or spastic paraplegia. Oculomotor apraxia (abnormal horizontal eye movement) may also be present with myoclonus, dystonia, and/or neuropathy.[2,3] Spastic paraplegia, characterized by lower limb spasticity and muscle weakness, can also be seen with decreased vibration sense, optic atrophy, hyperreflexia, and neuropathy.[4] Also included on this panel are episodic ataxias that often present as periods of unsteady gait with or without myokymia, which may result in permanent ataxia late in disease course.[1] Although considerable phenotypic overlap exists between the SCAs and related disorders, distinguishing features have been noted in most subtypes.[1,2,4,5] The SCAs are typically classified based on mode of inheritance, age of onset and clinical features. Age of onset ranges from the 3rd to 4th decades of life, although some disorders can present as early as childhood.[5] Treatment is primarily supportive for most forms of inherited SCA.[1] The prevalence of autosomal dominant and autosomal recessive cerebellar ataxia, including individuals with polynucleotide repeat disorders, is estimated to be approximately 1-5:100,000 and 3:100,000 individuals in the general population, respectively.[1,6]

INHERITANCE PATTERN/GENETICS
Ataxias can be either genetic or acquired in nature. Acquired ataxia may be the result of alcoholism, vitamin deficiency, multiple sclerosis, vascular disease, metastatic cancer, or paraneoplastic diseases.[1,7] Genetic forms may display autosomal dominant, autosomal recessive, X-linked, or mitochondrial inheritance.[1,5] The hereditary ataxias, especially adult onset forms, can be difficult to differentiate by clinical evaluation alone due to similarities in early manifestations and overlapping ages of onset.[1] Often, molecular genetic testing can assist in diagnosis, when combined with family history, physical examination and neuroimaging. Approximately 50% of ataxia is attributed to pathogenic repeat expansions in a small subset of genes, an additional 20% is attributed to pathogenic sequence based changes, in a larger number of genes and the remaining causes are acquired or undetermined.[8,9] The Spinocerebellar Ataxia and Related Disorders Panel at GeneDx includes sequencing and deletion/duplication analysis of genes associated with autosomal dominant and recessive disorders, excluding repeat expansion related disorders. The genes on this panel mostly code for ion channels transporters, mitochondrial and cell membrane proteins, and proteins involved in DNA repair and RNA/protein processing.10 The complete list of genes, inheritances, and associated disorders is included in the table below.

TEST METHODS
Genomic DNA is extracted from the submitted specimen. For skin punch biopsies, fibroblasts are cultured and used for DNA extraction. The DNA is enriched for the complete coding regions and splice site junctions of the

genes on this panel using a proprietary targeted capture system developed by GeneDx for next-generation sequencing with CNV calling (NGS-CNV). The enriched targets are simultaneously sequenced with paired-end reads on an Illumina platform. Bi-directional sequence reads are assembled and aligned to reference sequences based on NCBI RefSeq transcripts and human genome build GRCh37/UCSC hg19. After gene specific filtering, data are analyzed to identify sequence variants and most deletions and duplications involving coding exons at the exon-level; however, technical limitations and inherent sequence properties effectively reduce this resolution for some genes. For the *AARS2, CACNA1G, CLN5, C10ORF2, HEXB, ITPR1, KCNA1, NOTCH3, PEX7, PMM2, RRM2B, and TPP1* genes, sequencing but not deletion/duplication analysis are performed. For the *TMEM240* gene, only whole gene CNVs are reported. Alternative sequencing or copy number detection methods are used to analyze regions with inadequate sequence or copy number data by NGS. Sequence variants are reported according to the Human Genome Variation Society (HGVS) guidelines. Copy number variants are reported based on the probe coordinates, the coordinates of the exons involved, or precise breakpoints when known. Reportable variants include pathogenic variants, likely pathogenic variants, and variants of uncertain significance. Likely benign and benign variants, if present, are not routinely reported but are available upon request. Available evidence for variant classification may change over time and the reported variant(s) may be re-classified according to the AMP/ACMG guidelines for variant classification (Richards et al. 2015), which may lead to re-issuing a revised report.

The technical sensitivity of sequencing is estimated to be >99% at detecting single nucleotide events, but less for deletions greater than 20 base pairs, insertions or rearrangements greater than 10 base pairs, or low-level mosaicism. The copy number assessment methods used with this test identify most deletions and duplications involving coding exons but are less reliable for detecting copy number variants of less than 500 base pairs. Assessment of copy number events also depends on the inherent sequence properties of the targeted regions, including shared homology and exon size. Mosaicism detection is limited and balanced chromosome aberrations cannot be identified.

TEST SENSITIVITY

In a clinically heterogeneous cohort of individuals with ataxia, Nemeth et al. (2013) identified a pathogenic single nucleotide variant in 8.3% of individuals with adult onset ataxia and 40% of individuals with childhood onset ataxia, who previously had a negative result for a panel of common polynucleotide repeat expansion disorders.

Spinocerebellar Ataxia and Related Disorders Panel

Gene	Disease Associations	Inheritance	Diagnostic Yield in Selected Population(s)
*AARS2***	Leukoencephalopathy with ovarian failure	AR	Rare[1,10]
ABCD1	Adrenoleukodystrophy	XR	Rare in ataxia[34]
AFG3L2	SCA28	AD	~1.5% of AD cerebellar ataxia[11]
ANO10	SCA10	AR	Common in Mexican and Brazilian populations[12]
APTX	AOA1	AR	4-9% of AR ataxia[13]
ATM	Ataxia-telangiectasia	AD/AR	1:40,000-1:100,000 in US, Common cause of child onset cerebellar ataxia[14]
*CACNA1A**	Episodic ataxia type 2 (EA2); familial hemiplegic migraine (FHM); spinocerebellar ataxia type 6 (SCA6)	AD	EA2 is rare form of ataxia[15-17]

*CACNA1G**	SCA42	AD	Recurrent variant identified in specific populations[35]
*CLN5***	Adult-onset AR ataxia associated with neuronal ceroid-lipofuscinosis 5	AR	Rare[1,10]
CYP27A1	Cerebrotendinous xanthomatosis	AR	Rare in ataxia[1,10]; Treatment options available[36]
CYP7B1	Spastic paraplegia 5A	AR	Rare cause of sensory ataxia[37]
*C100RF2***	Perrault syndrome 5; Progressive external ophthalmoplegia with mitochondrial DNA deletions	AR	Rare[1,10]
DNMT1	Cerebellar ataxia, deafness, and narcolepsy; hereditary sensory neuropathy type IE	AD	Rare[1,18]
EEF2	SCA26	AD	Rare[1,10,38]
ELOVL4	SCA34	AD	Rare[1,10,19]
ELOVL5	SCA28	AD	Rare[1,20]
FGF14	SCA27	AD	Rare[21,22]
GNB4	Charcot-Marie-Tooth disease	AD	Rare in ataxia[1,10,39]
GRID2	SCA18	AD/AR	Rarely AD[40]
GRM1	SCA44; SCAR13	AR/AR	Rare[23,24]
HEXA	Tay-Sachs disease	AR	Rare in ataxia[25]
*HEXB***	Sandhoff disease	AR	Rare in ataxia[26]
*ITPR1***	SCA15; SCA16;SCA29;Gillespie syndrome	AD/AR	2-3% of familial ataxia in Australianpopulations[1,10,27]
*KCNA1***	Episodic ataxiatype 1 (EA1)	AD	Rare[1,10]
KCNC3	SCA13	AD	Rare[1,10]
KCND3	SCA19;SCA22	AD	Rare[1,10]
KIAA0196	Spastic paraplegia 8	AD	Rare[10]
KIF5A	Spastic paraplegia 10	AD	Rare in ataxia[41]
MME	SCA43	AD/AR	Rare[42]
*NOTCH3***	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1 (CADASIL)	AD	Rare in ataxia[43]
PDYN	SCA23	AD	Rare[1,10]
*PMM2***	Peroxisome biogenesis disorder (REFSUM)	AR	Rare in ataxia[44]
PNKP	Congenital disorder of glycosylationtype Ia	AR	Rare in ataxia[28]
PNPLA6	AOA4	AR	Rare[1,10]
POLG	Spastic paraplegia 39; Boucher-Neuhäuser syndrome	AR	Rare in ataxia[1,30]
POLR3A	POLG-related disorders	AR/AD	~3% of spastic ataxia[45]

PRKCG	Spastic ataxia	AR	Rare[1,10]
PRNP	SCA14	AD	Gait ataxia present in >50% of prionopathies[46]
*RRM2B***	Progressive external ophthalmoplegia with mitochondrial DNA deletions	AD/AR	Rare in ataxia[1,10]
SACS	Autosomal recessive spastic ataxia of Charlevoix-S aguenay (ARSACS)	AR	Rare[1,10]
SETX	AOA2	AR	Rare[1,10]
SLC20A2	Basal ganglia calcification, idiopathic	AD	Rare[10]
SPG7	Spastic paraplegia 7	AD/AR	Rare in ataxia1,[10,31]
SPG11	SPG11 related neurodegenerative disorders	AR	Rare in ataxia[10]
SPTBN2	SCA5 (Lincoln's Ataxia); SCAR14	AD/AR	Rare[1,10]
STUB1	SCAR16	AR	Rare[1,10]
SYNE1	SYNE1-related autosomal recessive cerebellar ataxia	AR	5-6% of adult onset AR ataxias[32]
SYT14	SCA11	AD	Rare[47]
TDP1	SCA with axonal neuropathy	AR	Rare[48]
TGM6	SCA35	AD	Rare[1,10]
*TMEM240***	SCA21	AD	Rare[1,10,33]
*TPP1***	SCAR7; Late-infantile neuronal ceroid-lipofuscinosis 2 (CLN2)	AR	Rare inataxia[1,10]
TTBK2	SCA11	AD	Rare[1,10]
TTPA	Ataxia with vitamin E deficiency (AVED)	AR	Rare[1,10]
ZFYVE26	Spastic paraplegia 15	AR	Rare[10]

***The CAG repeat expansion in *CACNA1A* that causes SCA6 is not detectable by this test.**
****Does not include deletion/duplication testing of *AARS2, CACNA1G, CLN5, C10ORF2, HEXB, ITPR1, KCNA1, NOTCH3, PEX7, PMM2, RRM2B, and TPP1.***
*****Only whole gene deletions or duplications of be detected.**

Abbreviations:
AD – Autosomal Dominant
AR – Autosomal Recessive
XR – X-linked recessive
SCA – Spinocerebellar Ataxia
AOA – Ataxia with Oculomotor Apraxia
SCAR – Autosomal Recessive Spinocerebellar Ataxia

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