Patient Name	Centre
Age/Gender	OP/IP No/UHID
MaxID/Lab ID	Collection Date/Time
Ref By	Reporting Date/Time

SIN No:B2B5152471

TEST REQUESTED

SCA comprehensive profile

METHOD USED

PCR, Fragment Analysis

RESULTS

		CAG repeat size (Allele 1)	CAG repeat size (Allele 2)	COMMENT
SCA-1/ATXN1	**Not Detected**	29	31	The results indicate that the patient is not affected with SCA1.
SCA-2/ATXN2	**Detected**	22	42	The results indicate that the patient is affected with SCA2.
SCA-3/ATXN3	**Not Detected**	22	26	The results indicate that the patient is not affected with SCA3.
SCA-6/CACNA1A	**Not Detected**	12	12	The results indicate that the patient is not affected with SCA6.
SCA-7/ATXN7	**Not Detected**	10	10	The results indicate that the patient is not affected with SCA7.
SCA-12/PP2R2B	**Not Detected**	10	10	The results indicate that the patient is not affected with SCA12.

INTERPRETATION

Reference ranges for oligonucleotide repeat sizes at the main SCA loci

Ataxia	Normal Repeat size	Uncertain Repeat Size/Gray Area	Reduced Penetrance	Full Penetrance	References (PMID)
SCA-1/ATXN1	6-38	-	-	≥39	11973625
SCA-2/ATXN2	14-31	32-34	-	≥35	10668721, 11502947, 11689490, 17923635
SCA-3/ATXN3	11-44	45-60	-	≥61	15457499, 11409435, 11708990,
SCA-6/CACNA1A	4-18	-	19	≥20	15362569, 1167360, 11403042
SCA-7/ATXN7	4-19	28-33	34-35	≥36	15349877, 15747371,
SCA-12/PP2R2B	4-32	40-45	-	≥51	16138911, 10581021, 11198281, 10581021

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NOTE

1. This is an in-house developed assay.
2. Results should be interpreted in context of clinical findings.
3. This assay detects oligonucleotide repeat sizes at the main SCA loci.
4. Presence of PCR inhibitors if any, might lead to amplification failure and indeterminate results.
5. Genetic counselling is recommended.

COMMENTS

Ataxia is a symptom, not a specific disease or diagnosis. All types of spinocerebellar ataxia are characterized by a progressive incoordination of walking. In addition, they are often associated with poor coordination of hand movements, eye movements, and speech. The genetic change that causes SCA types 1, 2, 3, 6, 7 and 12 is called a CAG repeat expansion. Most types of SCA are inherited in an autosomal dominant pattern.

Spinocerebellar ataxia type 1 (SCA1) is characterized by progressive cerebellar ataxia, dysarthria, and eventual deterioration of bulbar functions. SCA1 is inherited in an autosomal dominant manner. Anticipation has been observed in SCA1; expansions are more likely to occur when the pathogenic ATXN1 allele is paternally transmitted, and contractions are more typical of maternal transmissions. Results showing homozygosity for normal alleles of the same size (homoallelism) should be confirmed by an appropriate method (eg, triplet primed-PCR or Southern blotting), especially in early-onset cases (below the age of 26 years).

Spinocerebellar ataxia type 2 (SCA2) is characterized by progressive cerebellar ataxia, including nystagmus, slow saccadic eye movements, and in some individuals, ophthalmoparesis or parkinsonism. SCA2 is inherited in an autosomal dominant manner. Interruption of a CAG expanded allele by a CAA repeat does not mitigate the pathogenicity of the repeat size because both CAG and CAA code for glutamine; however, the interruption may enhance the meiotic stability of the repeat.

Spinocerebellar ataxia type 3 (SCA3), also known as Machado-Joseph disease (MJD), is characterized by progressive cerebellar ataxia and variable findings including pyramidal signs, a dystonic-rigid extrapyramidal syndrome, significant peripheral amyotrophy and generalized areflexia, progressive external ophthalmoplegia, action-induced facial and lingual fasciculations, and bulging eyes. SCA3 is inherited in an autosomal dominant manner.

Spinocerebellar ataxia type 6 (SCA6) is characterized by adult-onset, slowly progressive cerebellar ataxia, dysarthria, and nystagmus. Initial symptoms are gait unsteadiness, stumbling, and imbalance in approximately 90% of individuals; the remainder present with dysarthria. SCA6 is inherited in an autosomal dominant manner.

Spinocerebellar ataxia type 7 (SCA7) comprises a phenotypic spectrum ranging from adolescent- or adult-onset progressive cerebellar ataxia and cone-rod retinal dystrophy to infantile or early-childhood onset with multiorgan failure, an accelerated course, and early death.

Spinocerebellar ataxia type 12 (SCA12) is a very rare subtype of type I autosomal dominant cerebellar ataxia (ADCA type I; see this term). It is characterized by the presence of action tremor associated with relatively mild cerebellar ataxia. Associated pyramidal and extrapyramidal signs and dementia have been reported (orpha.net).

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