Patient NAME	:		
DOB/Age/Gender	:	Report STATUS	:
Patient ID / UHID	:	Barcode NO	:
Referred BY	:	Sample Type	:
Sample Collected	: Jan 10, 2026, 02:36 PM.	Report Date	: Jan 30, 2026, 09:24 AM.

#Spino Cerebral Ataxia (SCA) Panel

INDICATIONS :

Query diagnosis: ? Spinocerebellar Ataxia (SCA)

RESULT :

Type of SCA and DRPLA	Gene Involved	Nucleotide repeats	Nucleotide repeat Numbers (Normal)	Nucleotide repeat Numbers (Abnormal)	Nucleotide repeat Numbers (Patient)	Expansion (Detected/Not Detected)
SCA1	ATXN1	CAG	6-38	41-83	**65**	**Detected**
SCA2	ATXN2	CAG	14-32	33-500	21	Not detected
SCA3	ATXN3	CAG	12-44	60-87	26	Not detected
SCA6	CACNA1A	CAG	≤18	20-33	13	Not detected
SCA7	ATXN7	CAG	7-27	≥34	11	Not detected
SCA10	ATXN10	ATTCT	10-22	>280	16	Not detected
SCA12	PPP2R2B	CAG	7-32	51-78	16	Not detected
DRPLA	ATN1	CAG	6-35	≥48	20	Not detected

INTERPRETATION :

Repeat expansion detected in SCA1.

RECOMMENDATION :

- These results must be interpreted in the context of this individuals' clinical profile.
- Genetic counseling is recommended.

TEST METHODOLOGY AND TEST SUMMARY :

Methodology: Triplet repeat-primed polymerase chain reaction (PCR) followed by size analysis using capillary electrophoresis.

Summary: The autosomal dominant Spinocerebellar Ataxias and Dentatorubral-pallidoluysian atrophy (DRPLA) are a heterogenous group of neurodegenerative disorders with variable expression and phenotypic overlap. An accurate diagnosis

Approved by
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Postdoc-SGPGIMS Lucknow
Lab Head-Clinical Genomics

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relies on detection of a mutation in a specific causative gene. In this assay we screen 7 common SCA types and DRPLA for the presence of nucleotide repeat expansions. ATXN1, ATXN2, ATXN3, CACNA1A, ATXN7, PPP2R2B and ATN1 genes are screened for presence of expansion in CAG repeats pertaining to SCA1, SCA2, SCA3, SCA6, SCA7, SCA12 and DRPLA respectively. ATXN10 gene is screened for presence of expansion in ATTCT repeats pertaining to SCA10.

LIMITATIONS :

- Other neurodegenerative disorders will not be detected.
- Diagnostic errors can occur due to rare sequence variations.
- Although all precautions are taken during Molecular Genetic testing the currently available data indicate that the technical error rate for all types of Molecular DNA analysis is approximately 1%.

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NOTE- **This test is processed at Redcliffe's partnered lab.

*** End Of Report ***

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