LABORATORY REPORT

Redcliffe labs

Patient Name	**: Mrs Dummy**		
DOB/Age/Gender	: 15 Y/Female	Sample Collected	: May 21, 2024, 05:52 PM.
Patient ID / UHID	: XXX	Report Date	: Jun 03, 2024, 03:21 PM.
Referred By	: Dr. XXX	Barcode No	: XXX
Sample Type	: Whole blood EDTA	Report Status	: Final Report

Spino Cerebral Ataxia (SCA) Panel

INDICATIONS
Query diagnosis: ?SCA

RESULT :

Type of SCA	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	29	Not detected
SCA2	ATXN2	CAG	14-32	33-500	20	Not detected
SCA3	ATXN3	CAG	12-44	60-87	30	Not detected
SCA6	CACNA1A	CAG	≤18	20-33	11	Not detected
SCA7	ATXN7	CAG	7-27	≥34	12	Not detected
SCA10	ATXN10	ATTCT	10-22	≥280	12	Not detected
SCA12	PPP2R2B	CAG	7-32	51-78	14	Not detected
DRPLA	ATN1	CAG	6-35	≥48	17	Not detected

INTERPRETATION :
Repeat expansion not detected.

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 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.

Reviewed by
Imran Haider
Senior Scientific Officer
Onco-Genomics

Approved by
Dr. Himani Pandey
Postdoc-SGPGIMS Lucknow
Lab Head-Clinical Genomics

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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%.

REFERENCES:

1. Lian M, Limwongse C, Yoon CS, Lee CG, Law HY, Chong SS. Single-Tube Screen for Rapid Detection of Repeat Expansions in Seven Common Spinocerebellar Ataxias. Clin Chem. 2022 Jun 1;68(6):794-802.

2. Naito H, Takahashi T, Kamada M, Morino H, Yoshino H, Hattori N, Maruyama H, Kawakami H, Matsumoto M. First report of a Japanese family with spinocerebellar ataxia type 10: The second report from Asia after a report from China. PLoS One. 2017 May 19;12(5):e0177955.

3. Stevanin G, Dürr A, Brice A. Clinical and molecular advances in autosomal dominant cerebellar ataxias: from genotype to phenotype and physiopathology. Eur J Hum Genet. 2000 Jan;8(1):4-18.

*** End Of Report ***

Disclaimer: Method given in report are only indicative and can be changed depending upon type of machine and kit available at time of testing.

Not all tests at all locations are under NABL scope. Availability of tests under NABL scope varies from lab to lab.

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