

Case ID : 00000	Sample Type : EDTA PERIPHERAL BLOOD
Name : Ms. DUMMY	Date & Time Collected : 00-Oct-2023 05:00 PM
Sex/Age : XX,Y	Date & Time Received : 00-Oct-2023 01:10 PM
Bill. Loc. : NA	Date & Time Reported : 00-Oct-2023 06:11 PM
Ref. By : Dr.	
Indication :	

Report on Genetic screening of Seven Common Spinocerebellar Ataxias Type

Specimen Description: Sample quality is optimum for the test. DNA conc.: 44.6 ng/μl

TEST DESCRIPTION:

The autosomal Dominant Spinocerebellar Ataxias (ADCA) is a heterogeneous 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 for the presence of nucleotide repeat expansions. ATXN1, ATXN2, ATXN3, CACNA1A, ATXN7 and PPP2R2B genes are screened for presence of expansion in CAG repeats pertaining to SCA1, SCA2, SCA3, SCA6, SCA7 and SCA12 respectively. ATXN10 gene is screened for presence of expansion in ATTCT repeats pertaining to SCA10.

METHODOLOGY:

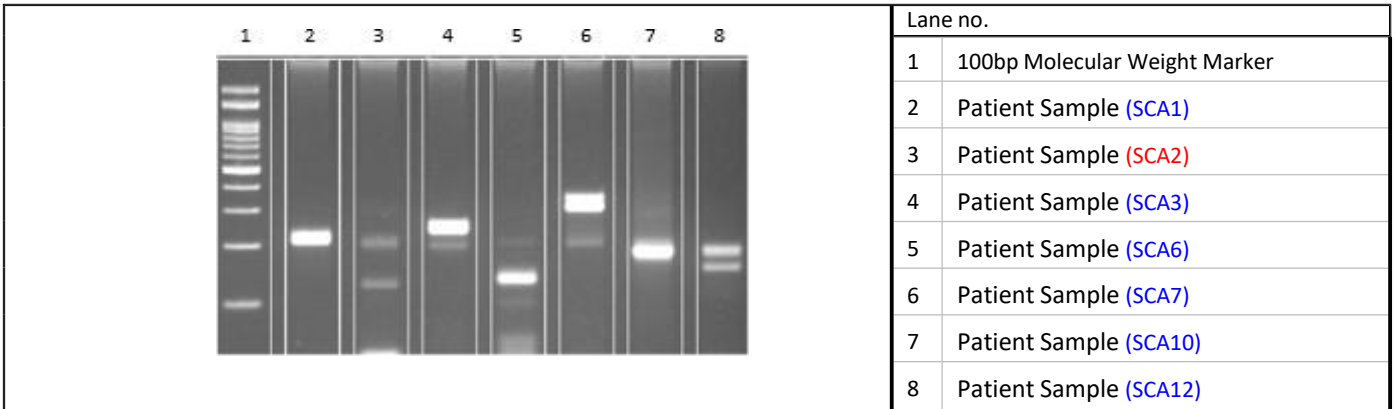
Polymerase Chain Reaction.

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	6-39	41-83	31	Not detected
SCA2	ATXN2	CAG	14-32	33-77	50	Detected
SCA3	ATXN3	CAG	12-40	51-86	29	Not detected
SCA6	CACNA1A	CAG	4-18	20-41	11	Not detected
SCA7	ATXN7	CAG	4-27	37-200	23	Not detected
SCA10	ATXN10	ATTCT	10-22	>280	13	Not detected
SCA12	PPP2R2B	CAG	7-32	55-93	23	Not detected

Case ID : 000000
 Name : Ms. DUMMY
 Sex/Age : XX,Y
 Bill. Loc. : NA
 Ref. By : NA

Sample Type : EDTA PERIPHERAL BLOOD
 Date & Time Collected : 00-Oct-2023 12:00 AM
 Date & Time Received : 00-Oct-2023 01:10 PM
 Date & Time Reported : 00-Oct-2023 06:11 PM



INTERPRETATION:

Repeat expansion detected in ATXN2 gene (consistent with diagnosis of Spino Cerebellar Ataxia type 2). Clinical correlation is advised

REFERENCE:

Stevanin G, Du"rr A, Brice A: Clinical and molecular advances in autosomal-dominant cerebellar ataxias: from genotype to phenotype and physiopathology. Eur J Hum Genet 2000, 8:4-18

..... 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.

Dr. Himani Pandey
 Lab Head - Genomics
 Post-Doc. Fellowship
 (Medical Genetics), SGPGIMS

Terms and Conditions of Reporting

1. The presented findings in the Reports are intended solely for informational and interpretational purposes by the referring physician or other qualified medical professionals possessing a comprehensive understanding of reporting units, reference ranges, and technological limitations. The laboratory shall not be held liable for any interpretation or misinterpretation of the results, nor for any consequential or incidental damages arising from such interpretation.
2. It is to be presumed that the tests performed pertain to the specimen/sample attributed to the Customer's name or identification. It is presumed that the verification particulars have been cleared out by the customer or his/her representation at the point of generation of said specimen / sample. It is hereby clarified that the reports furnished are restricted solely to the given specimen only.
3. It is to be noted that variations in results may occur between different laboratories and over time, even for the same parameter for the same Customer. The assays are performed and conducted in accordance with standard procedures, and the reported outcomes are contingent on the specific individual assay methods and equipment(s) used, as well as the quality of the received specimen.
4. This report shall not be deemed valid or admissible for any medico-legal purposes.
5. The Customers assume full responsibility for apprising the Company of any factors that may impact the test finding. These factors, among others, includes dietary intake, alcohol, or medication / drug(s) consumption, or fasting. This list of factors is only representative and not exhaustive.

DISCLAIMER

This is a sample report provided for demonstration purposes only and does not represent an actual patient report. Test results, reference ranges, methodologies, instrumentation, and report formats may vary depending on the laboratory performing the test. The format and representation shown are indicative of reports generated by the National Reference Laboratory of Redcliffe Labs, Noida. This sample report should not be used for medical interpretation, diagnosis, or treatment decisions.