LABORATORY REPORT

Redcliffe labs

Patient NAME	**: Mrs Dummy**		
DOB/Age/Gender	: 50 Y/Female	Report STATUS	: Final Report
Patient ID / UHID	: XXX	Barcode NO	: XXX
Referred BY	: Dr. XXX	Sample Type	: Whole blood EDTA
Sample Collected	: Oct 12, 2024, 08:19 PM.	Report Date	: Nov 05, 2024, 04:14 PM.

Spinal Cerebral Ataxia (SCA) Type- 17

Specimen : EDTA Whole Blood

Marker tested :- The CAG/CAA repeat expansion in the TBP (TATA box binding protein)

Method:- PCR/fragment Analysis the CAG.CAA repeat expansion in the TBP gene for diagnostic confirmation of SCA 17 is determined by PCR by Fragment Analysis

Result :

CAG Repeats	Interpetations
36 on Allele 1 and 38 on Allele 2	Not Detected - Diagnosis of SCA17 excluded

Interpretation :

CAG/CAA Repeats in the TBP gene and corresponding clinical interpretation:-

Ataxia	Gene	Repeat	Number of CAG/CAA Repeats		
			Normal	Reduced Penetrance	Full Penetrance
SCA17	TBP	(CAG)3 (CAA)3 (CAG)n CAACAGCAA (CAG)N CAACAG	25 - 42	43 - 48	49 - 66

Clinical Background :
1. Autosomal Dominant Cerebellar Ataxia (ADCA) is heterogeneous group of neurodegenerative disorders with variable expression and phenotypic overlap.
2. Spinocerebellar ataxia type 17 (SCA17) is an autosomal dominant (AD) neurodegenerative disorder characterized by ataxia, dementia, involuntary movemnet like chorea and dystonia and parkinsonism.
3. The age of onset of SCA17 ranges between 3-75 years. Threre is an inverse correlation between age of onset and repeat size.
4. SCA17 is caused by expansion of CAG/CAA repeats in the TBP gene which is present on chr6q27.
5. Individual with CAG.CAA repeats size in the range of 25-42 are negative for SCA17 and those with repeats greater than 48 have high probability of having SCA17.
6. Diagnosis is based on the clinical picture, familial history and ultimately on genetic testing. Differential diagnosis is broad and includes other types of SCA which may have similar features.
7. Genetic counseling is recommended in symptomatic patients or those with a family history of the disorder due to known SCA mutation, and pre-symptomatic testing should be discussed in adults.

Indications for SCA 17 DNA Test:-
1. Individuals with a family history of SCA17 who want to determine their risk.
2. To differentiate individuals with SCA17 from other ataxias.

Limitation of the assay:-
1. Presence of PCR inhibitors in the sample may prevent DNA amplification. Paradoxical results may arise due selection of inappropriate specimens and contamination during specimen collection.

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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2. Genetic Counseling is recommended for the patient and family members.

References:-

1. Choubtum et al., 2015, BMC Neurology, 15:166
2. Sequeiros et al., 2010, European Journal of Human Genetics, 18:1173–6
3. Nanda et al., 2007 Vol.22, No.3 p.436
4. Srivastava et al., 2013, 333:e65-e108
5. Gao et al., 2010, European Journal of Human Genetics, 16:215-222.

*** End Of Report ***

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