

Patient ID SA00147431	Patient Name TESTINGRNA, REPORTVALIDATION	Birth Date 2012-06-23	Gender F	Age 9
Order Number SA00147431	Client Order Number SA00147431	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 23 Aug 2021 08:00		

Spinocerebellar Ataxia Panel

Result Summary

MCR

REPEAT EXPANSION DETECTED

Result

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Spinocerebellar Ataxia Type 1 (ATXN1 gene):

Number of CAG repeats: 22 (Normal) and 26 (Normal)

Final result: Normal

Spinocerebellar Ataxia Type 2 (ATXN2 gene):

Number of CAG repeats: 18 (Normal) and 23 (Normal)

Final result: Normal

Spinocerebellar Ataxia Type 3 (ATXN3 gene):

Number of CAG repeats: 63 (Expanded) and 28 (Normal)

Final result: Expanded

Spinocerebellar Ataxia Type 6 (CACNA1A gene):

Number of CAG repeats: 11 (Normal) and 13 (Normal)

Final result: Normal

Spinocerebellar Ataxia Type 7 (ATXN7 gene):

Number of CAG repeats: 8 (Normal) and 11 (Normal)

Final result: Normal

Interpretation

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This result is consistent with a diagnosis of spinocerebellar ataxia type 3 (1).

Identification of a mutation has important implications for family members. Testing of at risk individuals is possible, but it is recommended that predictive testing be performed in conjunction with appropriate pre- and post-test counseling.

A genetic consultation may be of benefit.

REFERENCES

1. GeneReviews: www.ncbi.nlm.nih.gov/books/NBK1196

Specimen

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WB Whole Blood

Method

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A PCR-based assay was utilized to detect CAG repeat expansions within the ATXN1, ATXN2, ATXN3, CACNA1A, and ATXN7 genes and CAT trinucleotides that interrupt the CAG repeat tract within the ATXN1 gene. We estimate that the number of CAG repeats is correct to within $\pm 5\%$. ATXN1 (SCA1): Normal: <36 , Normal with CAT interruptions: $36-43$, Intermediate without CAT interruptions: $36-37$, Uncertain Significance: 38 , Expanded: >38 ; ATXN2 (SCA2): Normal: <32 , Uncertain Significance: 31 homozygous and 32 , Reduced Penetrance: $33-34$, Expanded: >34 ; ATXN3 (SCA3): Normal: <45 , Intermediate: $45-59$, Expanded: >59 ; CACNA1A (SCA6): Normal: <19 , Intermediate: 19 , Expanded: >19 ; ATXN7 (SCA7): Normal: <19 , Uncertain Significance: $19-27$, Intermediate: $28-33$, Reduced Penetrance: $34-36$, Expanded: >36 .

Disclaimer

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Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Bone Marrow transplants from allogenic donors will interfere with testing. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

Released By

MCR

Niu, Zhiy, Ph.D.

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Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292



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Test
Environment
ETBM Template

Laboratory Notes

- 1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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