MAYO CLINIC LABORATORIES

1-800-533-1710

SCAP

Spinocerebellar Ataxia Repeat Expansion Panel, Varies

Patient ID	Patient Name		Birth Date	Gender	Age
SA00147430	TESTINGNRV, REPORTVALIDATION		2010-03-23	M	11
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00147430	SA00147430	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		23 Aug 2021 08:00			

Spinocerebellar Ataxia Panel

Result Summary

MCR

NEGATIVE

Result

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Spinocerebellar Ataxia Type 1 (ATXN1 gene):
Number of CAG repeats: 24 (Normal) and 30 (Normal)
Final result: Normal

Spinocerebellar Ataxia Type 2 (ATXN2 gene):
Number of CAG repeats: 18 (Normal) and 22 (Normal)
Final result: Normal

Spinocerebellar Ataxia Type 3 (ATXN3 gene):
Number of CAG repeats: 18 (Normal) and 29 (Normal)
Final result: Normal

Spinocerebellar Ataxia Type 6 (CACNA1A gene):
Number of CAG repeats: 11 (Normal) and 12 (Normal)
Final result: Normal

Spinocerebellar Ataxia Type 7 (ATXN7 gene):
Number of CAG repeats: 9 (Normal) and 10 (Normal)
Final result: Normal

Interpretation

MCR

This result suggests this individual is very unlikely to have spinocerebellar ataxia (SCA). This result does not rule out the diagnosis of other inherited spinocerebellar ataxias that have overlapping clinical features with SCA types 1–3, 6, and 7.

If testing was performed for the indication of ALS, additional testing for disorders with overlapping phenotypes (SBULB/ Spinobulbar Musc Atrophy, Kennedy's and/or NMPAN/Motor Neuron Disease Panel and/or C9ORF/C9orf72, Molecular Analysis) could be considered.

A genetic consultation may be of benefit.

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 ±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

1 MCR

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, Zhiyv, Ph.D.

Received: 23 Aug 2021 13:32 **Reported:** 07 Sep 2021 10:08

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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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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