MAYO CLINIC LABORATORIES

1-800-533-1710

SMN1Z

SMN1 Gene, Full Gene Analysis

Patient ID	Patient Name		Birth Date	Gender	Age
SA00109657	TESTINGRNV, ATLAS		1993-05-09	F	25
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00109657	SA00109657	CLIENT, CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		17 Sep 2018 08:00			

Result Summary

MCR

Mutation Identified

Result

MCR

The following heterozygous alteration was identified:
Amino Acid change: p.Y272C (Tyr272Cys)
DNA change: c.815A>G (g.70241984)
Classification: PATHOGENIC

Interpretation

1 MCR

The c.815A>G (p. Y272C) alteration is a known pathogenic mutation.

This result indicates that this individual is at a minimum a carrier for Spinal Muscular Atrophy (SMA). We cannot rule out that this individual is affected with SMA as there are SMN1 gene alterations that are undetectable by this assay.

Since a heterozygous SMN1 gene variant was detected, genetic testing of at risk family members could be considered.

If applicable, additional genetic testing strategies, such as SMN1 copy number analysis (SMNDX, SMA Diagnostic by Del/Dup), may assist with identifying SMN1 gene deletions. Correlation of this result with family and clinical history, as well as other laboratory tests is recommended. Contact the Genomics Laboratory at 1–800–533–1710 if you have any questions.

A genetic consultation may be of benefit.

Unless reported or predicted to cause disease, alterations found deep in the intron or alterations that do not result in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are available upon request.

ADDITIONAL INFORMATION

Bi-directional sequence analysis was performed to test for the presence of mutations in all coding regions and intron/exon boundaries of the SMN1 gene (GenBank accession number NM_000344; build GRCh37 (hg19)). An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

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.

Multiple in-silico evaluation tools may have been used to assist in the interpretation of these results. Of note, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.

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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Specimen MCR

WB Whole Blood

Source MCR

Cord Blood

Released By MCR

Devin Oglesbee, Ph.D.

Received: 18 Sep 2018 08:31 **Reported:** 22 Sep 2018 10:40

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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