MAYO CLINIC LABORATORIES

1-800-533-1710

SMNCS

Spinal Muscular Atrophy Carrier Screening, Deletion/Duplication Analysis, Varies

Patient ID	Patient Name		Birth Date	Sex	Age
SA00167174	TESTINGRNV, SMNCS		2005-06-06	F	18
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00167174	SA00167174	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		13 Mar 2024 00:00			

SMA Carrier by Del/Dup

Result Summary

MCR

POSITIVE CARRIER

Result

MCR

A heterozygous deletion of SMN1 exon 7 was detected. Two copies of SMN2 were detected.

Interpretation

[1] MCR

This result indicates that this individual is a carrier for Spinal Muscular Atrophy (SMA). This interpretation assumes that this individual is not affected with SMA.

Since a deletion in SMN1 was identified, genetic testing of at risk family members could be considered. If appropriate, genetic testing should be offered to this individual's reproductive partner to further clarify the risk of having a child affected with SMA. Disease severity is also associated with SMN2 copy number.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

Laboratory developed test (LDT) for SMN1 exon 7, SMN2 exon 7 copy number and SMN1 rs143838139 (g.27134T>G) detection by droplet digital PCR. Mutation nomenclature is based on the following GenBank Accession number(s) (build GRCh37 (hg19)): NM_022874.

See www.mayocliniclabs.com (Test ID SMNCS) for additional information about this test.

CAUTIONS:

CLINICAL CORRELATIONS

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

If testing was performed because of a family history of Spinal Muscular Atrophy, it is often useful to first test an affected family member.

TECHNICAL LIMITATIONS

Point mutations are undetectable by this assay. Nor can the assay discriminate between two copies of SMN1 on the same chromosome versus two copies on separate chromosomes. 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.

Specimen

MCR

WB Whole Blood

Released By

MCR

LINDA HASADSRI

Received: 13 Mar 2024 13:49 **Reported:** 14 Mar 2024 12:29

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.

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