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
SA00167173	TESTINGRNV, SMNCS		2005-06-06	F	18
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00167173	SA00167173	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		13 Mar 2024 00:00			

SMA Carrier by Del/Dup

Result Summary

MCR

NEGATIVE FOR SMN1 DELETION (SEE INTERPRETATION)

Result

MCR

Two copies of SMN1 exon 7 were detected.
Three copies of SMN2 were detected.

The g.27134T>G polymorphism is absent.

Interpretation

(1) MCR

This result indicates a reduced carrier risk for Spinal Muscular Atrophy (SMA). Please see the table below for residual risk based on ancestry and absence of SMN1 g.27134T>G (1). Individuals who carry two copies of the SMN1 gene on one chromosome and zero copies of SMN1 on their other chromosome (i.e. 2+0 carriers) are at risk to have an affected child when their partner is also an SMA carrier. Other alterations within the SMN1 gene, such as point mutations, are not detected by this assay.

Patient Ancestry	Pre-test Carrier Frequency	Residual Risk of (2+0) SMA Carrier Status
Ashkenazi Jewish	1 in 41.1	1 in 580
Asian	1 in 53	1 in 701.8
African	1 in 66	1 in 395.7
Latino	1 in 117	1 in 1,762
European	1 in 35	1 in 769.3

The calculations noted in the chart are based on known population carrier frequencies and assume no family history of SMA. We are unable to provide a revised risk assessment for ancestries other than those listed as there is insufficient information available about the SMA carrier frequency for other populations.

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.

Additional Information

MCR

REFERENCES

1. Genet Med. 2014 Feb;16(2):149–156. PMID 23788250

Specimen

MCR

WB Whole Blood

Released By

MCR

LINDA HASADSRI

Received: 13 Mar 2024 13:48 **Reported:** 14 Mar 2024 12:19

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

MAYO CLINIC LABORATORIES

Patient ID	Patient Name		Birth Date	Sex	Age
SA00167173	TESTINGRNV, SMNCS		2005-06-06	F	18
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00167173	SA00167173	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		13 Mar 2024 00:00			

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