MAYO CLINIC LABORATORIES

1-800-533-1710

SMNDX

Spinal Muscular Atrophy Diagnostic Assay, Deletion/ Duplication Analysis, Varies

Patient ID	Patient Name		Birth Date	Gender	Age
SA00101240	TESTINGRNV, BILLING		1945-12-04	M	72
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00101240	SA00101240	CLIENT, CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		08 Dec 2017 08:00			

SMA Diagnostic by Del/Dup

Result Summary

MCR

POSITIVE

Result

MCR

Homozygous deletions of SMN1 exon 7 were detected.
Three copies of SMN2 were detected.

Interpretation

[1] MCR

This result indicates that this individual is affected with Spinal Muscular Atrophy (SMA). Disease severity is associated with SMN2 copy number. Since homozygous deletions of SMN1 were detected, genetic testing of at risk family members could be considered.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

Laboratory developed test (LDT) for SMN1 exon 7 and SMN2 exon 7 copy number 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 SMNDX) 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

Blood Spot

Released By

MCR

LINDA HASADSRI

Received: 09 Dec 2017 15:55 **Reported:** 12 Dec 2017 11:26

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