

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

Patient ID SA00101238	Patient Name TESTINGRNV, BILLING	Birth Date 2002-12-07	Gender F	Age 15
Order Number SA00101238	Client Order Number SA00101238	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 08 Dec 2017 10:00		

SMA Diagnostic by Del/Dup

Result Summary

NEGATIVE

Result

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

Interpretation

This result decreases the likelihood that this individual has Spinal Muscular Atrophy (SMA). A diagnosis of SMA cannot be completely excluded as other alterations within SMN1 are undetectable by this test.

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.

MCR

MCR

1 MCR

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

Blood Spot

MCR

Released By

LINDA HASADSRI

MCR

Received: 09 Dec 2017 15:56

Reported: 12 Dec 2017 11:25

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