

Patient ID SA00101234	Patient Name TESTINGRNV, BILLING	Birth Date 2000-07-09	Gender M	Age 17
Order Number SA00101234	Client Order Number SA00101234	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 08 Dec 2017 10:00		

SMA Carrier by Del/Dup

Result Summary

POSITIVE CARRIER

MCR

Result

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

MCR

Interpretation

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.

1 MCR

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 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 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

Blood Spot

MCR

Released By

LINDA HASADSRI

MCR

Received: 09 Dec 2017 15:54

Reported: 12 Dec 2017 11:24

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