MAYO CLINIC LABORATORIES

1-800-533-1710

# SMNCS

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

| Patient ID | Patient Name | | Birth Date | Gender | Age |
|---|---|---|---|---|---|
| SA00101235 | TESTINGRNV, BILLING | | 2000-10-09 | M | 17 |
| **Order Number** | **Client Order Number** | **Ordering Physician** | **Report Notes** | | |
| SA00101235 | SA00101235 | CLIENT, CLIENT | | | |
| **Account Information** | | **Collected** | | | |
| C7028846 DLMP Rochester | | 08 Dec 2017 10:00 | | | |

## 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.

### Interpretation

1 MCR

This result decreases the likelihood that this individual is a carrier for Spinal Muscular Atrophy (SMA). Carrier status cannot be completely excluded as this test cannot differentiate one copy of SMN1 exon 7 on each chromosome (1/1) from two copies of SMN1 on one chromosome and none on the other chromosome (2/0). Other alterations within SMN1 are also undetectable by this test.

An individual's residual risk for SMA carrier status varies by ancestry and SMN1 copies (1).

Adjusted Risk

Ancestry Prior Risk 2 SMN1 Copies 3 SMN1 Copies

European 1 in 35 1 in 632 1 in 3500

Ashkenazi Jewish 1 in 41 1 in 350 1 in 4000

Asian 1 in 53 1 in 628 1 in 5000

African 1 in 66 1 in 121 1 in 3000

Latino 1 in 117 1 in 1061 1 in 11000

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

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.

### Additional Information

MCR

### REFERENCES

1. J Med Genet. 2009; 46: 641–644 (PMID: 19625283)

### Specimen

MCR

Blood Spot



**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 | Gender | Age |
|---|---|---|---|---|---|
| SA00101235 | TESTINGRNV, BILLING | | 2000-10-09 | M | 17 |
| **Order Number** | **Client Order Number** | **Ordering Physician** | **Report Notes** | | |
| SA00101235 | SA00101235 | CLIENT, CLIENT | | | |
| **Account Information** | | **Collected** | | | |
| C7028846 DLMP Rochester | | 08 Dec 2017 10:00 | | | |

**Released By** MCR

LINDA HASADSRI

**Received:** 09 Dec 2017 15:54 **Reported:** 12 Dec 2017 11:21



## 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 |