Warde Medical Laboratory

LABORATORY REPORT

Example Client, XYZ123
1234 Warde Road
Ann Arbor MI 48108

EXAMPLE, REPORT W
WX0000003826 F 12/05/1988 34 Y

Non-invasive Prenatal Testing (NIPT)

Collected: 08/29/2023 15:30 Received: 08/29/2023 15:30

Test Name	Result	Flag	Ref-Ranges	Units	Site

Spinal Muscular Atrophy Carrier Screening

Result Summary — SEE BELOW — MMRL

RESULT: NEGATIVE FOR SMN1 DELETION (SEE INTERPRETATION)

Result — SEE BELOW — MMRL

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

The g.27134T>G polymorphism is absent.

Interpretation — SEE BELOW — MMRL

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

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by droplet digital PCR. Mutation nomenclature is based on the following GenBank Accession number(s) (build GRCh37 (hg19)):

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.

TEST CLASSIFICATION
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.

Additional Information	SEE BELOW		MMRL

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

Specimen	WB Whole Blood		MMRL
Source	.		MMRL
Released by	SEE BELOW		MMRL

RESULT: Linda Hasadsri, M.D., Ph.D.

Test Performed by:
Mayo Clinic Laboratories - Rochester Main Campus
200 First Street SW, Rochester, MN 55905
Lab Director: William G. Morice M.D. Ph.D.; CLIA# 24D0404292

LAB: L - LOW, H - HIGH, AB - ABNORMAL, C - CRITICAL, . - NOT TESTED

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Performing Site:
MMRL: MAYO MEDICAL REFERENCE LAB 3050 Superior Drive NW Rochester MN 55901

Reported Date: 2023.09.28 13:40 SMNCS

LAB: L - LOW, H - HIGH, AB - ABNORMAL, C - CRITICAL, . - NOT TESTED

F229000054
WX0000003826
Printed D&T: 09/28/23 13:41

Ordered By: KAJAL SITWALA, MD, PhD
WX00000000002353

Kajal V. Sitwala, MD, PhD - Medical Director
Form: MM RL1