



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: 09/01/2023 12:10 Received: 09/01/2023 12:10

Test Name Result Flag Ref-Ranges Units Site

Spinal Muscular Atrophy Carrier Screening

Result Summary SEE BELOW MMRL

RESULT: POSITIVE CARRIER

Result SEE BELOW MMRL

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

Interpretation SEE BELOW MMRL

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. 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, SMN2 exon 7 copy number and SMN1 rs143838139 (g.27134T>G) detection 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

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



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: 09/01/2023 12:10 Received: 09/01/2023 12:10

Table with 6 columns: Test Name, Result, Flag, Ref-Ranges, Units, Site. Contains test details, classification, and specimen information.

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

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

Reported Date: 2023.09.28 13:42 SMNCS

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