ARUP LABORATORIES
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Jonathan R. Genzen, MD, PhD, Chief Medical Officer

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	Unknown
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Niemann-Pick Type A (SMPD1), 4 Variants

ARUP test code 0051458

Niemann-Pick Type A (SMPD1), Specimen	Whole Blood
Niemann-Pick Type A (SMPD1), Allele 1	c.911T>C *
Niemann-Pick Type A (SMPD1), Allele 2	Negative
Niemann-Pick Type A (SMPD1), Interp	See Note

Indication for testing: Carrier screening or diagnostic testing for Niemann-Pick disease, type A.

Positive: One pathogenic variant, p.L304P (c.911T>C), was detected in the SMPD1 gene; therefore, this individual is at least a carrier of Niemann-Pick disease type A. At-risk family members should be offered testing to determine carrier status for the identified variant. This individual's reproductive partner should be offered screening for the disorder. Genetic counseling is recommended.

This result has been reviewed and approved by [redacted]

H=High, L=Low, *=Abnormal, C=Critical

BACKGROUND INFORMATION: Niemann-Pick Type A (SMPD1), 4 Variants
CHARACTERISTICS: Niemann-Pick type A is a lysosomal storage disease causing hepatosplenomegaly, delayed physical and mental development, hypotonia, rigidity, intellectual disability, and death typically by age 3.
INCIDENCE: 1 in 32,000 Ashkenazi Jewish individuals.
INHERITANCE: Autosomal recessive.
CAUSE: SMPD1 pathogenic variants.
VARIANTS TESTED: p.L304P (c.911T>C), p.F333Sfs (c.996delC), p.R498L (c.1493G>T), and p.R610del (c.1829_1831delGCC).
CLINICAL SENSITIVITY: 90 percent in Ashkenazi Jewish individuals, varies by ethnicity in non-Ashkenazi Jewish individuals.
METHODOLOGY: Polymerase chain reaction (PCR) and fluorescence monitoring.
ANALYTICAL SENSITIVITY AND SPECIFICITY: Greater than 99 percent.
LIMITATIONS: Variants other than those tested will not be detected. Diagnostic errors can occur due to rare sequence variations.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Niemann-Pick Type A (SMPD1), Specimen	23-062-106358	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Niemann-Pick Type A (SMPD1), Allele 1	23-062-106358	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Niemann-Pick Type A (SMPD1), Allele 2	23-062-106358	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Niemann-Pick Type A (SMPD1), Interp	23-062-106358	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical