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

Physician: Doctor, Example

Patient: Patient, Example

DOB [redacted]
Sex: [redacted]
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 01/01/2017 12:34

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

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

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]

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	22-040-113753	2/9/2022 2:00:00 PM	2/9/2022 2:30:41 PM	2/9/2022 2:32:00 PM
Niemann-Pick Type A (SMPD1), Allele 1	22-040-113753	2/9/2022 2:00:00 PM	2/9/2022 2:30:41 PM	2/9/2022 2:32:00 PM
Niemann-Pick Type A (SMPD1), Allele 2	22-040-113753	2/9/2022 2:00:00 PM	2/9/2022 2:30:41 PM	2/9/2022 2:32:00 PM
Niemann-Pick Type A (SMPD1), Interp	22-040-113753	2/9/2022 2:00:00 PM	2/9/2022 2:30:41 PM	2/9/2022 2:32:00 PM

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