

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 Negative

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.

Negative: This sample is negative for the four pathogenic variants tested in the SMPD1 gene. If this is an asymptomatic individual of Ashkenazi Jewish descent, his/her risk of being a carrier of Niemann-Pick disease type A has been reduced from 1 in 90 to approximately 1 in 900.

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	23-304-101594	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Niemann-Pick Type A (SMPD1), Allele 1	23-304-101594	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Niemann-Pick Type A (SMPD1), Allele 2	23-304-101594	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Niemann-Pick Type A (SMPD1), Interp	23-304-101594	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

Unless otherwise indicated, testing performed at: