

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

Physician: Doctor, Example

**Patient: Patient, Example**

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

**NEB-Related Nemaline Myopathy, 1 Variant**

ARUP test code 2013745

NEB-Related Nemaline Myopathy, Specimen whole Blood

NEB-Related Nemaline Myopathy, Allele 1 **exon 55 del \***

NEB-Related Nemaline Myopathy, Allele 2 Negative

NEB-Related Nemaline Myopathy, Interp

See Note

BACKGROUND INFORMATION: NEB-Related Nemaline Myopathy, 1 Variant  
CHARACTERISTICS: NEB-related nemaline myopathy typically presents within the first year of life with hypotonia, feeding difficulties and muscle weakness of the face, neck, arms and legs. Muscle weakness is static or progresses very slowly, but lifespan is not usually decreased.  
INCIDENCE: 1 in 47,000 in Ashkenazi Jewish individuals.  
INHERITANCE: Autosomal recessive.  
CAUSE: NEB pathogenic variants.  
VARIANT TESTED: Exon 55 del (p.R2478\_D2512del).  
CLINICAL SENSITIVITY: 99 percent in Ashkenazi Jewish individuals; unknown in other ethnicities.  
METHODOLOGY: Polymerase chain reaction (PCR) and fluorescence monitoring.  
ANALYTICAL SENSITIVITY AND SPECIFICITY: Greater than 99 percent.  
LIMITATIONS: Variants other than exon 55 del will not be detected. Diagnostic errors can occur due to rare sequence variations.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

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

VERIFIED/REPORTED DATES				
Procedure	Accession	Collected	Received	Verified/Reported
NEB-Related Nemaline Myopathy, Specimen	21-218-117069	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NEB-Related Nemaline Myopathy, Allele 1	21-218-117069	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NEB-Related Nemaline Myopathy, Allele 2	21-218-117069	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NEB-Related Nemaline Myopathy, Interp	21-218-117069	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:

ARUP LABORATORIES | 800-522-2787 | aruplab.com  
500 Chipeta Way, Salt Lake City, UT 84108-1221  
Tracy I. George, MD, Laboratory Director

Patient: Patient, Example  
ARUP Accession: 21-218-117069  
Patient Identifiers: 01234567890ABCD, 012345  
Visit Number (FIN): 01234567890ABCD  
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