

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: ██████████
Gender: Female
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 Negative

NEB-Related Nemaline Myopathy, Allele 2 Negative

NEB-Related Nemaline Myopathy, Interp See Note

This result has been reviewed and approved by ██████████ M.D.

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	20-338-105886	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NEB-Related Nemaline Myopathy, Allele 1	20-338-105886	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NEB-Related Nemaline Myopathy, Allele 2	20-338-105886	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NEB-Related Nemaline Myopathy, Interp	20-338-105886	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: 20-338-105886
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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