

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 11/18/1960
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

Indication for testing: Carrier screening or diagnostic testing for NEB-related nemaline myopathy.

Negative: This sample is negative for the pathogenic exon 55 deletion (p.R2478_D2512del) in the NEB gene. If this is an asymptomatic individual of Ashkenazi Jewish descent, his/her carrier risk is reduced from 1 in 92 to approximately 1 in 10,700.

This result has been reviewed and approved by [REDACTED]

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	23-348-400929	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NEB-Related Nemaline Myopathy, Allele 1	23-348-400929	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NEB-Related Nemaline Myopathy, Allele 2	23-348-400929	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NEB-Related Nemaline Myopathy, Interp	23-348-400929	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
Jonathan R. Genzen, MD, PhD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 23-348-400929
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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