ARUP LABORATORIES
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Jonathan R. Genzen, MD, PhD, Chief Medical Officer

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	Unknown
Gender:	Male
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

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

Positive: One copy of the pathogenic exon 55 deletion (p.R2478_D2512del) in the NEB gene was detected; therefore, this individual is at least a carrier of NEB-related nemaline myopathy. Genetic counseling is recommended. This individual's reproductive partner should be offered screening for the disorder. At-risk family members should be offered testing to determine carrier status for the identified variant.

This result has been reviewed and approved by [redacted]

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

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
NEB-Related Nemaline Myopathy, Specimen	23-062-104390	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NEB-Related Nemaline Myopathy, Allele 1	23-062-104390	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NEB-Related Nemaline Myopathy, Allele 2	23-062-104390	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NEB-Related Nemaline Myopathy, Interp	23-062-104390	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