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	[redacted]
Sex:	[redacted]
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Mucolipidosis Type IV (MCOLN1), 2 Variants

ARUP test code 0051448

Mucolipidosis IV (MCOLN1), Specimen	Whole Blood
Mucolipidosis IV (MCOLN1), Allele 1	g.511_6943del *
Mucolipidosis IV (MCOLN1), Allele 2	Negative
Mucolipidosis IV (MCOLN1), Interp	See Note

Indication for testing: Carrier screening or diagnostic testing for mucolipidosis type IV.

Positive: One pathogenic variant, g.511_6943del (6.4 kb deletion), was detected in the MCOLN1 gene; therefore, this individual is at least a carrier of mucolipidosis type IV. 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]

BACKGROUND INFORMATION: Mucolipidosis Type IV (MCOLN1), 2 Variants:
CHARACTERISTICS: Mucolipidosis type IV is characterized by early onset of severe psychomotor delay and progressive visual impairment due to corneal clouding and retinal degeneration. Affected individuals may occasionally learn to say a few words or walk independently. While most affected individuals remain neurologically static until age 30, about 15 percent will display neurological degeneration.
INCIDENCE: 1 in 63,000 Ashkenazi Jewish individuals.
INHERITANCE: Autosomal recessive.
CAUSE: MCOLN1 pathogenic variants.
VARIANTS TESTED: g.511_6943del and c.406-2A>G.
CLINICAL SENSITIVITY: 95 percent in Ashkenazi Jewish individuals, 6 to 10 percent in other ethnicities.
METHODOLOGY: Polymerase chain reaction (PCR) and fluorescence monitoring.
ANALYTICAL SENSITIVITY AND SPECIFICITY: Greater than 99 percent.
LIMITATIONS: Variants other than g.511_6943del and c.406-2A>G 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.

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

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Mucolipidosis IV (MCOLN1), Specimen	21-349-113240	12/15/2021 2:25 00 PM	12/15/2021 2:26:34 PM	12/16/2021 8:40 00 AM
Mucolipidosis IV (MCOLN1), Allele 1	21-349-113240	12/15/2021 2:25 00 PM	12/15/2021 2:26:34 PM	12/16/2021 8:40 00 AM
Mucolipidosis IV (MCOLN1), Allele 2	21-349-113240	12/15/2021 2:25 00 PM	12/15/2021 2:26:34 PM	12/16/2021 8:40 00 AM
Mucolipidosis IV (MCOLN1), Interp	21-349-113240	12/15/2021 2:25 00 PM	12/15/2021 2:26:34 PM	12/16/2021 8:40 00 AM

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