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

Physician: Doctor, Example

Patient: Patient, Example

DOB	2/2/2020
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL) Sequencing and Deletion/Duplication

ARUP test code 2004212

VLCAD FGA Specimen	Whole Blood

VLCAD (ACADVL) Interpretation	Negative

TEST PERFORMED - 2004212
TEST DESCRIPTION - Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL) Sequencing and Deletion/Duplication
INDICATION FOR TEST - Confirm Diagnosis

RESULT
No pathogenic variants were detected in the ACADVL gene.

INTERPRETATION
No pathogenic variants were detected in the ACADVL gene by sequencing all coding regions and intron-exon boundaries or by deletion/duplication analysis. This result significantly decreases the likelihood that this individual is affected with, or a carrier of, very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency. Please refer to the background information included in this report for the clinical sensitivity and limitations of this test.

RECOMMENDATIONS
The diagnosis and management of VLCAD deficiency should rely on clinical symptoms and biochemical/functional assays. Genetic consultation is recommended.

COMMENTS
Reference Sequence: GenBank # NM_000018.2 (ACADVL)
Nucleotide numbering begins at the "A" of the ATG initiation codon.
Likely benign and benign variants are not reported.

This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION: Very Long-Chain Acyl-CoA Dehydrogenase Deficiency/VLCAD (ACADVL) Deletion/Duplication:

CHARACTERISTICS: Fatty acid beta-oxidation disorder leading to hypoketotic hypoglycemia, dicarboxylic aciduria, hepatic failure, Reye-like symptoms, cardiomyopathy, skeletal myopathy, and sudden death. Clinical presentation varies in severity and age of onset.
INCIDENCE: Approximately 1 in 40,000.
INHERITENCE: Autosomal recessive.
CAUSE: Pathogenic ACADVL gene mutations.
CLINICAL SENSITIVITY: May be as high as 95 percent.
METHODOLOGY: Bidirectional sequencing of the entire coding region and intron-exon boundaries of the ACADVL gene. Multiplex Ligation-dependent Probe Amplification (MLPA) to detect large ACADVL coding region deletions/duplications.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Regulatory region mutations and deep intronic mutations will not be detected; deletion/duplication breakpoints will not be determined. Deletions/duplications in exon 2 of ACADVL will not be detected.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
VLCAD FGA Specimen	20-043-401080	2/11/2020 12:18 00 PM	2/13/2020 9:38:00 AM	3/17/2020 10:26:00 AM
VLCAD (ACADVL) Interpretation	20-043-401080	2/11/2020 12:18 00 PM	2/13/2020 9:38:00 AM	3/17/2020 10:26:00 AM

END OF CHART

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