ARUP LABORATORIES | **aruplab.com**
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: 801-583-2787, toll free: 800-522-2787
Julio Delgado, MD, MS, Chief Medical Officer

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	2/24/2020
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL) Sequencing

ARUP test code 2002001

VLCAD FGS Specimen	Whole Blood

VLCAD (ACADVL) Sequencing	Negative

TEST PERFORMED - 2002001
TEST DESCRIPTION - Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL) Sequencing
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. 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. If clinical suspicion for VLCAD deficiency remains high, consideration should be given to ACADVL Deletion/Duplication analysis (ARUP test code 2004208).

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 Steven Steinberg, Ph.D.

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

BACKGROUND INFORMATION: Very Long-Chain Acyl-CoA Dehydrogenas Deficiency (ACADVL) Sequencing

CHARACTERISTICS: Fatty acid beta-oxidation disorder leading to hypoketotic hypoglycemia, hepatic failure, Reye-likesymptoms, cardiomyopathy, skeletal myopathy and sudden death. Clinical presentation varies in severity and age of onset.
INCIDENCE: Approximately 1 in 40,000.
INHERITANCE: Autosomal recessive.
CAUSE: Deleterious ACADVL gene mutations.
CLINICAL SENSITIVITY: 80-90 percent.
METHODOLOGY: Bidirectional sequencing of the entire ACADVL coding region and intron-exon boundaries.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Regulatory region mutations, deep intronic mutations, and large deletions/duplications 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 FGS Specimen	20-064-400757	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
VLCAD (ACADVL) Sequencing	20-064-400757	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