ARUP LABORATORIES
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Tracy I. George, MD, Chief Medical Officer

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	12/31/1752
Gender:	Unknown
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Primary Carnitine Deficiency (SLC22A5) Sequencing and Deletion/Duplication

ARUP test code 2004203

PCD FGA Specimen	Whole Blood
Primary Carnitine Deficiency Interpretat	Negative

TEST PERFORMED - 2004203
TEST DESCRIPTION - Primary Carnitine Deficiency (SLC22A5) Sequencing and Deletion/Duplication
INDICATION FOR TEST - Not Provided

RESULT
No pathogenic variants were detected in the SLC22A5 gene.

INTERPRETATION
No pathogenic variants were detected in the SLC22A5 gene by sequencing the coding region and intron/exon boundaries or by deletion/duplication analysis. This reduces the chance that the individual is affected with or a carrier of primary carnitine deficiency. Please refer to the background information included in this report for the clinical sensitivity and limitations of this test.

RECOMMENDATIONS
Medical screening and management of this individual, including initiation of dietary carnitine supplementation should rely on clinical and biochemical findings. Since this test does not detect all pathogenic SLC22A5 variants (e.g., deep intronic or regulatory region variants) measurement of carnitine transport activity in fibroblasts should be considered if clinical symptoms are present.

COMMENTS
Reference Sequence: GenBank # NM_003060.3 (SLC22A5)
Nucleotide numbering begins at the "A" of the ATG initiation codon.
Likely benign and benign variants are not included in this report.

This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION: Primary Carnitine Deficiency (SLC22A5) Sequencing and Deletion/Duplication

CHARACTERISTICS: Hypoketotic hypoglycemia during periods of fasting, hepatomegaly, Reye syndrome, sudden infant death, developmental delay, cardiac and/or skeletal myopathy, hypotonia and enlarged heart.
INCIDENCE: 1 in 40,000 for European Caucasian and Japanese, lower in other populations.
INHERITENCE: Autosomal recessive.
CAUSE: Pathogenic SLC22A5 gene mutations.
CLINICAL SENSITIVITY: May be as high as 95 percent.
METHODS: Bidirectional sequencing of the entire coding region and intron-exon boundaries of SLC22A5 gene; Multiplex Ligation-dependent Probe Amplification (MLPA) to detect large SLC22A5 coding region deletions/duplications.
ANALYTICAL SENSITIVITY: Greater than 99 percent.
LIMITATIONS: Mutations in genes other than SLC22A5 will not be detected; deletion/duplication breakpoints will not be determined; deep intronic mutations and promoter mutations in the SLC22A5 gene will not be detected. Mutations within the primer/probe regions could affect the analytical sensitivity of this assay. 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
PCD FGA Specimen	20-056-112505	2/25/2020 1:11:00 PM	2/25/2020 1:18:11 PM	2/28/2020 10:49:00 AM
Primary Carnitine Deficiency Interpretat	20-056-112505	2/25/2020 1:11:00 PM	2/25/2020 1:18:11 PM	2/28/2020 10:49:00 AM

END OF CHART

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