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Julio C. Delgado, MD, MS, Director of Laboratories



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

Physician: Doctor, Example

## Patient: Patient, Example

| | |
|---|---|
| **DOB** | Unknown |
| **Gender:** | Unknown |
| **Patient Identifiers:** | 01234567890ABCD, 012345 |
| **Visit Number (FIN):** | 01234567890ABCD |
| **Collection Date:** | 00/00/0000 00:00 |

## Primary Carnitine Deficiency (SLC22A5) Sequencing

ARUP test code 0051682

| | |
|---|---|
| PCD FGS Specimen | Whole Blood |
| PCD Sequencing Interpretation | Negative |

TEST PERFORMED - 0051682
TEST DESCRIPTION - Primary Carnitine Deficiency (SLC22A5) Sequencing
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 of the coding region and intron/exon boundaries. 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 may not detect all pathogenic SLC22A5 variants (e.g., large deletions/duplications, deep intronic or regulatory region variants), measurement of carnitine transport activity in fibroblasts should be considered if 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 Rong Mao, M.D.

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

BACKGROUND INFORMATION: Primary Carnitine Deficiency (SLC22A5) Sequencing

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.
INHERITANCE: Autosomal recessive.
CAUSE: Deleterious SLC22A5 gene mutations causing a non-functional protein (OCTN2)
CLINICAL SENSITIVITY: Approximately 82 percent
METHODOLOGY: Bidirectional sequencing of the entire coding region and intron/exon boundaries of SLC22A5 gene.
ANALYTICAL SENSITIVITY: Greater than 99 percent
LIMITATIONS: Mutations in genes other than SLC22A5 will not be detected; large deletions, deep intronic mutations and promoter mutations in the SLC22A5 gene are not detected by this assay; analytical sensitivity may be compromised by rare primer site mutations. 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 FGS Specimen | 20-056-112501 | 00/00/0000 00:00 | 00/00/0000 00:00 | 00/00/0000 00:00 |
| PCD Sequencing Interpretation | 20-056-112501 | 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**