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

Patient Report

Client: ARUP Physician Services
321 TESTING ANSR EXTRACT
Salt Lake City, NY 84108-
USA

Provider: 68912 -arup,arup

Patient: CERT, SAMPLE4
DOB:
Gender: Female
Patient Identifiers: 598781
Visit Number (FIN): 622853
Client Supplied ID:

Specimen Collected: 14-Jul-21 13:44

Fatty Acid Oxidation Disorders | **Received: 14-Jul-21 13:48** **Report/Verified: 14-Jul-21 17:17**

Procedure	Result	Units	Reference Interval
Fatty Acid Oxidation Disorders Specimen	Whole Blood		
Fatty Acid Oxidation Disorders Interp	Uncertain [f1 i1]		

Result Footnote

f1: Fatty Acid Oxidation Disorders Interp

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

Test Information

i1: Fatty Acid Oxidation Disorders Interp

BACKGROUND INFORMATION: Fatty Acid Oxidation Disorders Panel, Sequencing

CHARACTERISTICS: Fatty acid oxidation disorders can present with hypoketotic hypoglycemia, lethargy, episodic emesis, seizures, dicarboxylic aciduria, hepatomegaly, hepatic failure, cardiomyopathy, Reye-like symptoms, skeletal myopathy, myalgia, exercise intolerance, coma, and sudden death. Clinical presentation varies in severity and age of onset.

INCIDENCE: Approximately 1 in 5,000 to 1 in 10,000 births.

CAUSE: Pathogenic germline variants in genes associated with fatty acid oxidation disorders.

INHERITANCE: Mostly autosomal recessive; rarely autosomal dominant or X-linked.

CLINICAL SENSITIVITY: May be as high as 96 percent.

GENES TESTED: ACAD9, ACADM, ACADS, ACADVL, ACAT1, CPT1A, CPT2, ECHS1, ETFA, ETFB, ETFDH, FLAD1, HADH, HADHA, HADHB, HMGCL, HMGCS2, HSD17B10, LPIN1*, MLYCD, SLC22A5, SLC25A20, SLC52A1, SLC52A2, SLC52A3

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab

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Test Information

i1: Fatty Acid Oxidation Disorders Interp

*One or more exons are not covered by sequencing for the indicated gene; see limitations section below.

METHODOLOGY: Capture of all coding exons and exon-intron junctions of the targeted genes, followed by massively parallel sequencing. Sanger sequencing was performed to fill in regions of low coverage and confirm reported variants as necessary. Human genome build 19 (Hg 19) was used for data analysis.

ANALYTICAL SENSITIVITY/SPECIFICITY: The analytical sensitivity of this test is approximately 99 percent for single nucleotide variants (SNVs) and greater than 93 percent for insertions/duplications/deletions from 1-10 base pairs in size. Variants greater than 10 base pairs may be detected, but the analytical sensitivity may be reduced.

LIMITATIONS: A negative result does not exclude a diagnosis of a fatty acid oxidation disorder. This test only detects variants within the coding regions and intron-exon boundaries of the targeted genes. Regulatory region variants and deep intronic variants will not be identified. Deletions/duplications/insertions of any size may not be detected by massively parallel sequencing. Diagnostic errors can occur due to rare sequence variations. In some cases, variants may not be identified due to technical limitations in the presence of pseudogenes, repetitive, or homologous regions. This assay may not detect low-level mosaic or somatic variants associated with disease. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation. Noncoding transcripts were not analyzed.

The following regions are not sequenced due to technical limitations of the assay:
LPIN1(NM_001349200) exon 13
LPIN1(NM_001349201) exon 12