Client: ARUP Example Report Only
500 Chipeta Way
Salt Lake City, UT 84108
UNITED STATES

Physician: arup, arup

Patient: EXAMPLE, NEGATIVE

DOB	
Gender:	Female
Patient Identifiers:	32594
Visit Number (FIN):	32903
Collection Date:	9/9/2021 09:20

Fatty Acid Oxidation Disorders Panel, Sequencing

ARUP test code 3001851

Fatty Acid Oxidation Disorders Specimen	Whole Blood
Fatty Acid Oxidation Disorders Interp	Negative

INDICATION FOR TESTING
Elevated C14:1, abnormal newborn screening (NBS).

RESULT
No pathogenic variants were detected in any of the genes tested.

INTERPRETATION
No pathogenic variants were identified by massively parallel sequencing of the coding regions and exon-intron boundaries of the genes tested. This result decreases the likelihood of, but does not exclude, a heritable form of a fatty acid oxidation disorder. Please refer to the background information included in this report for a list of the genes analyzed and limitations of this test.

RECOMMENDATIONS
The diagnosis and management of fatty acid oxidation disorders should rely on clinical symptoms and biochemical/functional assays. Genetic consultation is recommended.

COMMENTS
Likely benign and benign variants are not included in this report but are available upon request.

This result has been reviewed and approved by [redacted]

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.

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

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
*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

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Fatty Acid Oxidation Disorders Specimen	21-252-103097	9/9/2021 9:20:00 AM	9/9/2021 9:20:54 AM	9/9/2021 9:24:00 AM
Fatty Acid Oxidation Disorders Interp	21-252-103097	9/9/2021 9:20:00 AM	9/9/2021 9:20:54 AM	9/9/2021 9:24:00 AM

END OF CHART

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