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

Physician: arup, arup

Patient: Test, MCADPCR 2

DOB	
Sex:	Female
Patient Identifiers:	46924
Visit Number (FIN):	47262
Collection Date:	3/3/2023 11:21

Medium Chain Acyl-CoA Dehydrogenase (ACADM) 2 Mutations

ARUP test code 0051205

MCADPCR Specimen	Whole Blood
MCAD Mutation A985G	Homozygous *
MCAD Mutation T199C	Negative
Medium Chain Acyl-CoA Interpretation	See Note

Indication for testing: Carrier screening or diagnostic testing for MCAD deficiency.

Result
A985G: Homozygous
T199C: Negative

This sample is homozygous for the severe pathogenic variant c.985A>G in the ACADM gene; therefore, the patient is affected with medium-chain acyl-CoA dehydrogenase (MCAD) deficiency. Genetic and dietary consultations are indicated. Family members should be offered targeted testing for the identified pathogenic variant. The patient's reproductive partner should be offered carrier testing for MCAD.

This result has been reviewed and approved by [redacted]

BACKGROUND INFORMATION: Medium Chain Acyl-CoA Dehydrogenase (ACADM) 2 Mutations

CHARACTERISTICS: Limited mitochondrial fatty acid beta-oxidation leading to hypoglycemia, lethargy, seizures hypoketotic aciduria, vomiting, hepatomegaly, hepatic failure, encephalopathy, and sudden death. Manifestations often triggered by prolonged fasting or other metabolic stressors.
INCIDENCE: 1 in 15,000
INHERITANCE: Autosomal recessive.
CAUSE: Deleterious ACADM gene mutations.
CLINICAL SENSITIVITY: 75 percent for MCAD deficiency.
MUTATIONS TESTED: ACADM mutations c.985A>G (p.K329E, also known as K304E) and c.199T>C (p.Y67H, also known as Y42H).
METHODOLOGY: Polymerase chain reaction (PCR) and fluorescence monitoring.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. ACADM mutations other than c.985A>G and c.199T>C will not be detected.

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

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
MCADPCR Specimen	23-062-105721	3/3/2023 11:21:00 AM	3/3/2023 11:21:09 AM	4/13/2023 1:18:00 PM
MCAD Mutation A985G	23-062-105721	3/3/2023 11:21:00 AM	3/3/2023 11:21:09 AM	4/13/2023 1:18:00 PM
MCAD Mutation T199C	23-062-105721	3/3/2023 11:21:00 AM	3/3/2023 11:21:09 AM	4/13/2023 1:18:00 PM
Medium Chain Acyl-CoA Interpretation	23-062-105721	3/3/2023 11:21:00 AM	3/3/2023 11:21:09 AM	4/13/2023 1:18:00 PM

END OF CHART

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