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

Physician: Doctor, Example

Patient: Patient, Example

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

Medium Chain Acyl-CoA Dehydrogenase (ACADM) 2 Mutations

ARUP test code 0051205

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

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

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

Result
A985G: Homozygous
T199C: Negative

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

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

Characteristics: Limited mitochondrial fatty acid beta-oxidation leading to hypoglycemia, lethargy, seizures, hypoketotic dicarboxylic 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 Detected: ACADM mutations c.985A>G (p.K304E) and c.199T>C (p.Y42H).
Methodology: Polymerase chain reaction and fluorescence monitoring.
Analytical Sensitivity and Specificity: 99 percent.
Limitations: Rare diagnostic errors can occur due to primer site mutations. ACADM mutations other than c.985A>G and c.199T>C will not be detected.

This test is performed pursuant to an agreement with Roche Molecular Systems, Inc.

This result has been reviewed and approved by Rong Mao, M.D.

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 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.

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

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
MCADPCR Specimen	21-088-105072	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
MCAD Mutation A985G	21-088-105072	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
MCAD Mutation T199C	21-088-105072	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Medium Chain Acyl-CoA Interpretation	21-088-105072	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