ARUP LABORATORIES
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Jonathan R. Genzen, MD, PhD, Chief Medical Officer

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	[redacted]/1989
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Medium Chain Acyl-CoA Dehydrogenase (ACADM) 2 Variants

ARUP test code 3019336

MCAD_PCR Specimen	Whole Blood	
MCAD A985G	**Heterozygous**	*
MCAD T199C	**Heterozygous**	*
Medium Chain Acyl-CoA Interpretation	See Note	

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

Result
A985G: Heterozygous
T199C: Heterozygous

This sample is heterozygous for both a mild pathogenic variant, c.199T>C, and a severe pathogenic variant, c.985A>G in the ACADM gene. The c.199C>T variant is associated with some MCAD enzymatic activity; however, this genotype may result in an abnormal acylcarnitine profile, and this individual may be at risk for metabolic crisis. Genetic and dietary consultations are indicated. Family members should be offered targeted testing for the identified pathogenic variants. The patient's reproductive partner should be offered carrier testing for MCAD.

This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION: Medium Chain Acyl-CoA Dehydrogenase PCR
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.

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
MCAD_PCR Specimen	25-204-102499	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
MCAD A985G	25-204-102499	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
MCAD T199C	25-204-102499	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Medium Chain Acyl-CoA Interpretation	25-204-102499	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