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

PATIENT REPORT

Patient Age/Sex: 32 years Female

Specimen Collected: 5/6/2025 08:12 MDT

Medium Chain Acyl-CoA Dehydrogenase PCR | **Received: 5/6/2025 08:12 MDT** | **Report/Verified: 5/7/2025 15:50 MDT**

Procedure	Result	Units	Reference Interval
MCAD_PCR Specimen	Whole Blood		
MCAD A985G	**Homozygous** *		
MCAD T199C	Negative		
Medium Chain Acyl-CoA Interpretation	See Note f1 i1		

Result Footnote

f1: Medium Chain Acyl-CoA Interpretation

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]

Test Information

i1: Medium Chain Acyl-CoA Interpretation

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

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

Test Information

i1: Medium Chain Acyl-CoA Interpretation

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

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