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Tracy I. George, MD, Chief Medical Officer

Patient Report

Patient Age/Sex: 37 years Male

Specimen Collected: 08-Mar-22 10:44

X-Medium Chain Acyl-CoA Dehydrogenase | **Received: 08-Mar-22 10:44** **Report/Verified: 10-Mar-22 13:56**

Procedure	Result	Units	Reference Interval
MCADPCR Specimen	Whole Blood		
MCAD Mutation A985G	Negative		
MCAD Mutation 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: Negative
T199C: Negative

This sample is negative for the ACADM variants, c.985A>G and c.199T>C. Persons affected with medium chain acyl-CoA dehydrogenase (MCAD) deficiency may have pathogenic variants not detected by this assay. If the patient has biochemical and/or clinical evidence of MCAD deficiency, plasma acylcarnitine profile testing and/or ACADM gene sequencing is recommended.

This result has been reviewed and approved by Sherin Shaaban, M.D., Ph.D.

Test Information

i1: Medium Chain Acyl-CoA Interpretation

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.

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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