MAYO CLINIC LABORATORIES

Patient ID	Patient Name		Birth Date	Gender	Age
SA00117626	TESTING, ATLAS REPORT ABNORMAL		1994-06-12	M	24
Order Number: SA00117626	Client Order Number: SA00117626	Ordering Physician: CLIENT, CLIENT	Report Notes		
Account Information: C7028846 DLMP Rochester		Collected: 06 Feb 2019 08:30			

Cryopreserve for Biochem Studies

MCR

Fibroblasts successfully cryopreserved.

Received: 07 Feb 2019 10:55 **Reported:** 07 Feb 2019 12:25

Fatty Acid Ox Probe Assay, Fibro

Interpretation

1 MCR

POSITIVE
The acylcarnitine profiles revealed the following abnormalities (the concentrations provided correspond to the mean value for triplicate analysis):

Hexanoylcarnitine (C6):
0.055 nmol/mg protein (99th percentile of reference population: 0.093; N=128)

Octanoylcarnitine (C8):
0.127 nmol/mg protein (99th percentile of reference population: 0.098; N=128)

Decanoylcarnitine (C10):
0.067 nmol/mg protein (99th percentile of reference population: 0.147; N=128)

The score of CLIR post-analytical tool for Medium-Chain Acyl-CoA Dehydrogenase (MCAD) deficiency: 266 (informative >150; percentile rank in comparison to known cases: 87%, N=20)

INTERPRETATION

These results are consistent with MCAD deficiency. We recommend further confirmation by molecular genetic analysis of the MCAD gene. In addition, both parents, and if applicable, all siblings (of any age) should be tested (request urine acylglycines and plasma acylcarnitines). Please contact the Biochemical Genetics consultant or genetic counselor on call (Tel. 1–800–533–1710) if you have any questions.

ADDITIONAL INFORMATION

Fibroblasts Incubated with Enriched Medium Followed by Tandem Mass Spectrometry for Acylcarnitines.

Reviewed By

MCR

Daniel Kraft

Received: 07 Feb 2019 10:55 **Reported:** 07 Feb 2019 12:08

Fibroblast Culture

MCR

Fibroblasts successfully cultured.

Received: 07 Feb 2019 10:55 **Reported:** 07 Feb 2019 12:24

Laboratory Notes

1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292