

Patient ID SA00117549	Patient Name TESTINGRNV, SAMPLEREPORT	Birth Date 2018-12-01	Gender F	Age 8 W
Order Number SA00117549	Client Order Number SA00117549	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 31 Jan 2019 11:00		

C4 Acylcarnitine, QN, U

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15.67 mmol/mol Cr


MCR

Reference Value
<3.00

determine the presence of ethylmalonic acid (see: Oglesbee et al Genet Med 2007; 9: 108–116).

Please contact the Biochemical Genetics consultant on call (1–800–533–1710) if you have any questions.

ADDITIONAL INFORMATION

Flow Injection Analysis-Tandem Mass Spectrometry (FIA-MS/MS)

Reviewed By

Amy Breitlow

MCR

Received: 01 Feb 2019 12:33

Reported: 01 Feb 2019 12:47

C4 Interpretation

1 MCR

In this urine sample, the excretion of C4 acylcarnitine (a mixture of butyryl- and isobutyrylcarnitine) was elevated, the C4/C3 ratio was normal. In our experience, this result is likely secondary to a biochemical diagnosis of short-chain acyl-CoA dehydrogenase (SCAD) deficiency, a condition of uncertain clinical significance (see: Jethva et al Mol Genet Metab 2008; 95: 195–200). We recommend urine organic acid or acylglycine analysis to

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