

Patient ID SA00117612	Patient Name TESTINGRNV, SAMPLEREPORT	Birth Date 1982-06-29	Gender F	Age 36	
Order Number SA00117612	Client Order Number SA00117612	Ordering Physician CLIENT, CLIENT	Report Notes		
Account Information C7028846 DLMP Rochester		Collected 06 Feb 2019 06:00			

HGEM, BS

2-OH Glutaric acid

19 nmol/mL

MCR

 Reference Value
≤25

3-OH Glutaric acid

1.50 nmol/mL

MCR

 Reference Value
≤1.5

Glutaric acid

0.80 nmol/mL

MCR

 Reference Value
≤1.5

Methylsuccinic acid

<0.45 nmol/mL

MCR

 Reference Value
≤0.45

Ethylmalonic acid

 15 nmol/mL

MCR

 Reference Value
≤3.5

High

Interpretation (HGEM)

ⓘ MCR

POSITIVE In this sample, the concentration of ethylmalonic acid (EMA) was increased within the range observed in patients with short-chain acyl-CoA dehydrogenase (SCAD) deficiency (two pathogenic mutations: ≥10 nmol/mL; see Wolfe L et al. Short-Chain Acyl-CoA Dehydrogenase Deficiency. GeneReviews [Internet]. Available from: <http://www.ncbi.nlm.nih.gov/books/NBK63582/>). The differential diagnosis for this finding also includes ethylmalonic encephalopathy. We recommend urine organic acids screen (OAU/Organic Acids Scrn, U) and plasma acylcarnitine (ACRN/Acylcarnitines, Quantitative, P) analyses. Molecular confirmation is available upon request.

ADDITIONAL INFORMATION

Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)

Reviewed By

MCR

Amy Breitlow

Received: 07 Feb 2019 06:47

Reported: 07 Feb 2019 06:56

Laboratory Notes

- ⓘ 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