

|                                                       |                                                 |                                             |                    |                  |
|-------------------------------------------------------|-------------------------------------------------|---------------------------------------------|--------------------|------------------|
| Patient ID<br><b>SA00117614</b>                       | Patient Name<br><b>TESTINGRNV, SAMPLEREPORT</b> | Birth Date<br><b>1982-06-29</b>             | Gender<br><b>F</b> | Age<br><b>36</b> |
| Order Number<br><b>SA00117614</b>                     | Client Order Number<br><b>SA00117614</b>        | Ordering Physician<br><b>CLIENT, CLIENT</b> | Report Notes       |                  |
| Account Information<br><b>C7028846 DLMP Rochester</b> |                                                 | Collected<br><b>06 Feb 2019 06:00</b>       |                    |                  |

## HGEM, S

### 2-OH Glutaric acid

4.00 nmol/mL

MCR

Reference Value  
≤4.5

### 3-OH Glutaric acid

0.50 nmol/mL

MCR

Reference Value  
≤0.7

### Glutaric acid

0.40 nmol/mL

MCR

Reference Value  
≤0.8

### Methylsuccinic acid

&lt;0.21 nmol/mL

MCR

Reference Value  
≤0.3

### Ethylmalonic acid

 20 nmol/mL

High

MCR

Reference Value  
≤1.5

### Interpretation (HGEMS)

1 MCR

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: ≥6 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 06:57

Reported: 07 Feb 2019 07:04

### 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 |
|------|--------------------------------------------------|------------------------------------------|-----------------------------|------------------|
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