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|--|--------------------------------------|--------------------------------------|--------------|------------|
| Patient ID SA00050147 | Patient Name TESTINGRNV, ACYLGABN | Birth Date 2012-01-09 | Gender M | Age 9 M |
| Order Number SA00050147 | Client Order Number SA00050147 | Ordering Physician Client, Client | Report Notes | |
| Account Information C7028846 DLMP Rochester | | Collected 29 Oct 2012 00:00 | | |

Acylglycines, QN, U

Ethylmalonic Acid



High

156.20 mg/g Cr

MCR

Reference Value
0.5–20.2

Suberylglycine

10.10 mg/g Cr

MCR

Reference Value
0.00–11.0

2-Methylsuccinic acid

12.30 mg/g Cr

MCR

Reference Value
0.4–13.8

trans-Cinnamoylglycine

12.30 mg/g Cr

MCR

Reference Value
0.2–14.7

Glutaric acid

7.80 mg/g Cr

MCR

Reference Value
0.6–15.2

Dodecanedioic acid

0.55 mg/g Cr

MCR

Reference Value
0.00–1.10

Isobutyrylglycine

9.90 mg/g Cr

MCR

Reference Value
0.00–11.0

Tetradecanedioic acid

0.22 mg/g Cr

MCR

Reference Value
0.00–1.00

n-Butyrylglycine

1.85 mg/g Cr

MCR

Reference Value
0.10–2.10

Hexadecanedioic acid

0.33 mg/g Cr

MCR

Reference Value
0.00–1.00

2-Methylbutyrylglycine

5.66 mg/g Cr

MCR

Reference Value
0.3–7.5

Interpretation

MCR

In this sample, the excretion of ethylmalonic acid (EMA) was elevated. The differential diagnosis of EMA aciduria includes short-chain acyl-CoA dehydrogenase (SCAD) deficiency, polymorphic variants of the SCAD gene, and mitochondrial respiratory chain defects. SCAD deficiency is a condition of uncertain clinical significance and has been diagnosed in some patients during the first weeks of life with muscle tone abnormalities, hypoglycemia and vomiting. More frequently, however, EMA aciduria is identified in early childhood in patients presenting with muscle hypotonia and developmental delay. If clinically indicated consider plasma acylcarnitines, lactate, pyruvate, and molecular genetic analysis of the ACADS gene.

ADDITIONAL INFORMATION

Gas Chromatography-Mass Spectrometry (GC-MS) Stable Isotope Dilution Analysis

Isovalerylglycine

12.30 mg/g Cr

MCR

Reference Value
0.3–14.3

n-Hexanoylglycine

1.55 mg/g Cr

MCR

Reference Value
0.20–1.90

n-Octanoylglycine

2.00 mg/g Cr

MCR

Reference Value
0.1–2.1

3-Phenylpropionylglycine

0.99 mg/g Cr

MCR

Reference Value
0.00–1.10

Performing Site Legend

| Code | Laboratory | Address |
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| MCR | Mayo Clinic Dept. of Lab Med and Pathology | 200 First Street SW, Rochester, MN 55905 |



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Reviewed By

MCR

JANICE HELGESON

Received: 30 Oct 2012 15:06

Reported: 31 Oct 2012 10:19

QA Environment

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| Code | Laboratory | Address |
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| MCR | Mayo Clinic Dept. of Lab Med and Pathology | 200 First Street SW, Rochester, MN 55905 |