MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,ACYLGABN	SA00050147	9M	M	SA00050147
Ordering Phys				**DOB** 01/09/2012
Client Order # SA00050147	**Account Information**			**Report Notes**
Collected 10/29/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 12/14/2012 11:03				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Acylglycines, QN, U			REPORTED 10/31/2012 10:19		
Ethylmalonic Acid	H	156.20	mg/g Cr	0.5-20.2	MCR
2-Methylsuccinic acid		12.30	mg/g Cr	0.4-13.8	MCR
Glutaric acid		7.80	mg/g Cr	0.6-15.2	MCR
Isobutyrylglycine		9.90	mg/g Cr	0.00-11.0	MCR
n-Butyrylglycine		1.85	mg/g Cr	0.10-2.10	MCR
2-Methylbutyrylglycine		5.66	mg/g Cr	0.3-7.5	MCR
Isovalerylglycine		12.30	mg/g Cr	0.3-14.3	MCR
n-Hexanoylglycine		1.55	mg/g Cr	0.20-1.90	MCR
n-Octanoylglycine		2.00	mg/g Cr	0.1-2.1	MCR
3-Phenylpropionylglycine		0.99	mg/g Cr	0.00-1.10	MCR
Suberylglycine		10.10	mg/g Cr	0.00-11.0	MCR
trans-Cinnamoylglycine		12.30	mg/g Cr	0.2-14.7	MCR
Dodecanedioic acid		0.55	mg/g Cr	0.00-1.10	MCR
Tetradecanedioic acid		0.22	mg/g Cr	0.00-1.00	MCR
Hexadecanedioic acid		0.33	mg/g Cr	0.00-1.00	MCR
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.
Gas Chromatography-Mass Spectrometry (GC-MS) Stable Isotope Dilution Analysis

Reviewed By	JANICE HELGESON	MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

Patient Name	Collection Date and Time	Report Status
TESTINGRNV,ACYLGABN	10/29/2012	Final
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* Report times for Mayo performed tests are CST/CDT