Laboratory Service Manual

Lab Dept: Urine/Stool

Test Name: ACYLGLYCINES, QUANTITATIVE, URINE

General Information

Lab Order Codes: ACYU

Synonyms: Glycine conjugates urine

CPT Codes: 82544 – Column chromatography, stable isotope dilution, multiple analytes, quantitative, single stationary and mobile phase

Test Includes: The following acylglycines reported in ug/mg Creatinine: Ethylmalonic Acid, 2-Methylsuccinic Acid, Glutaric Acid, Isobutyrylglycine, n-Butyrylglycine, 2-Methylbutyrylglycine, Isovalerylglycine, n-Hexanoylglycine, n-Octanoylglycine, 3-Phenylpropionylglycine, Suberylglycine, trans-Cinnamoylglycine, Dodecanedioic Acid (12:0), Tetradecanedioic Acid (14:0), Hexadecanedioic Acid (16:0).

Logistics

Test Indications: Useful for biochemical diagnosis of selected inborn errors of metabolism (see below) by quantitative determination of target urinary metabolites that are present in amounts below the detection limit of routine organic acid analysis. Acylglycine analysis is the method of choice, in urine, for the biochemical evaluation of asymptomatic patients affected with 1 of the following inborn errors of metabolism:

-Short chain acyl-CoA dehydrogenase (SCAD) deficiency
-Functional SCAD deficiency (G625A, C611T variants)
-Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
-Medium-chain 3-ketoacyl-CoA thiolase (MCKAT) deficiency
-Electron transfer flavoprotein (ETF) deficiency (Glutaric acidemia type 2)
-ETF: ubiquinone oxidoreductase (ETF-QO) deficiency
-(Glutaric acidemia type 2)
-Riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency
-Ethylmalonic encephalopathy
-2-Methylbutyryl-CoA dehydrogenase deficiency
-Isovaleryl-CoA dehydrogenase deficiency
-Glutaryl-CoA dehydrogenase deficiency

Lab Testing Sections: Urine/Stool - Sendouts

Referred to: Mayo Medical Laboratories (MML Test# 81249)

Phone Numbers:

Minneapolis: 612-813-6280

Children's Hospitals and Clinics of Minnesota

Saint Paul: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 5 – 14 days, test performed Monday, Wednesday, Friday

Special Instructions: Please include family history, clinical conditions (asymptomatic or acute episode), diet, and drug therapy information.

Specimen

Specimen Type: Urine, random

Container: Leak-proof urine container

Draw Volume: Entire specimen

Processed Volume: 10 mL (Minimum: 3 mL) urine

Collection: Routine urine collection

Special Processing: Lab Staff: Mix random urine sample well. Remove aliquot into a plastic, 13 mL urine tube. Store and ship at frozen temperatures. Forward promptly.

Patient Preparation: None

Sample Rejection: Specimens other than urine, warm specimens, mislabeled or unlabeled specimens

Interpretive

Reference Range:

Acylglycine:	**Range in mg/g Creatinine:**
Ethylmalonic Acid:	0.5 – 20.2
2-Methylsuccinic Acid	0.4 – 13.8
Glutaric Acid:	0.6 – 15.2
Isobutyrylglycine:	0.00 – 11.0
n-Butyrylglycine:	0.1 – 2.1
2-Methylbutyrylglycine:	0.3 – 7.5
Isovalerylglycine:	0.3 – 14.3

n-Hexanoylglycine:	0.2 – 1.9
n-Octanoylglycine:	0.1 – 2.1
3-Phenylpropionylglycine:	0.00 – 1.1
Suberylglycine:	0.00 – 11.0
trans-Cinnamoylglycine:	0.2 – 14.7
Dodecanedioic Acid (12:0):	0.0 – 1.1
Tetradecanedioic Acid (14:0):	0.00 – 1.0
Hexadecanedioic Acid (16:0):	0.00 – 1.0
Interpretation: When abnormal results are detected, a detailed interpretation is given, including an overview of the results and of their significance; a correlation to available clinical information; elements of differential diagnosis; recommendations for additional biochemical testing and in vitro confirmatory studies (enzyme assay, molecular analysis); name and number of key contacts who may provide these studies or answer questions.	

Critical Values: N/A

Limitations: Due to a limited number of metabolites included in the acylglycine analysis, it is recommended that an Organic Acid Screen, Urine also be performed concurrently.

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

References: Mayo Medical Laboratories November 2010

Updates: 6/23/2010: Updated recommended volume, previously listed as 5 mL.
11/30/2010: Units change, previously listed as mcg/mg Cr.