MAYO CLINIC LABORATORIES

Test Definition: AGU20

Acylglycines, Quantitative, Random, Urine

Overview

Useful For

Diagnosis and monitoring for patients affected with one of the following inborn errors of metabolism:

Fatty Acid Oxidation Disorders:
-Glutaric acidemia type II
-Medium-chain 3-ketoacyl-coenzyme A (CoA) thiolase (MCKAT) deficiency
-Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
-Short chain acyl-CoA dehydrogenase (SCAD) deficiency

Organic Acidurias:
-2-Methyl-3-hydroxybutyryl-CoA dehydrogenase (2M3HBD) deficiency
-2-Methylbutyryl-CoA dehydrogenase deficiency
-3-Methylcrotonyl-CoA carboxylase deficiency
-3-Methylglutaconyl-CoA-hydratase deficiency
-Aminoacylase 1 deficiency
-Beta-ketothiolase deficiency
-Ethylmalonic encephalopathy
-Glutaryl-CoA dehydrogenase deficiency
-Isobutyryl-CoA dehydrogenase deficiency
-Isovaleryl-CoA dehydrogenase deficiency
-Multiple carboxylase deficiency
-Propionic acidemia

Testing Algorithm

For more information see Newborn Screen Follow-up for Elevated C5-OH.

Special Instructions

- Biochemical Genetics Patient Information
- Newborn Screen Follow-up for Elevated C5-OH

Highlights

This test provides a quantitative report of abnormal levels of acylglycines in urine, identified via gas chromatography mass spectrometry.

Method Name

Gas Chromatography Mass Spectrometry (GC-MS)

NY State Available

Yes

Specimen

Specimen Type
Urine

Additional Testing Requirements
Diagnostic specificity of inborn errors of metabolism via urine acylglycine testing is available only for selected inborn errors of metabolism; it is recommended that urine organic acids (OAU / Organic Acids Screen, Random, Urine) be ordered and assessed simultaneously due to the limited number of metabolites included in this urine acylglycine test.

Necessary Information
1. Patient's age and sex are required.
2. Include family history, clinical condition (asymptomatic or acute episode), diet, and drug therapy information

Specimen Required
Supplies: Urine Tubes, 10 mL (T068)
Container/Tube: Plastic, 10-mL urine tube
Specimen Volume: 10 mL
Pediatric: If insufficient collection volume, submit as much specimen as possible in a single container; the laboratory will determine if volume is sufficient for testing.
Collection Instructions:
1. Collect a random urine specimen.
2. No preservative.

Forms
1. Biochemical Genetics Patient Information (T602)
2. If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

Specimen Minimum Volume
4 mL

Reject Due To
All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Urine	Frozen (preferred)	416 days	
	Refrigerated	9 days	

Clinical & Interpretive

Clinical Information

Acylglycines are glycine conjugates of acyl-coenzyme A species, which occur as normal intermediates of amino acid and fatty acid metabolism. In abnormal concentrations, acylglycines are biochemical markers of selected inborn errors of metabolism. Analysis of acylglycines is useful for the diagnosis and monitoring for specific fatty acid oxidation disorders and organic acidurias; however, it is recommended to use this testing in conjunction with urine organic acids and plasma acylcarnitines testing in order to establish a diagnosis.

In particular, acylglycine analysis is more sensitive and specific for the identification of asymptomatic patients and those who may experience mild or intermittent biochemical phenotypes that could be missed by organic acid analysis alone.

Reference Values

n-Acetylglycine:< or =3.50 mg/g Creatinine
n-Propionylglycine:< or =2.25 mg/g Creatinine
Isobutyrylglycine:< or =3.00 mg/g Creatinine
Ethylmalonic acid:< or =25.00 mg/g Creatinine
n-Butyrylglycine:< or =2.50 mg/g Creatinine
2-Methylsuccinic acid:< or =9.00 mg/g Creatinine
2-Methylbutyrylglycine:< or =2.00 mg/g Creatinine
Isovalerylglycine:< or =8.00 mg/g Creatinine
Glutaric acid:< or =8.00 mg/g Creatinine
3-Methylcrotonylglycine:< or =2.25 mg/g Creatinine
n-Tiglylglycine:< or =9.00 mg/g Creatinine
3-Methylglutaconic acid:< or =25.00 mg/g Creatinine
n-Hexanoylglycine:< or =2.00 mg/g Creatinine
n-Octanoylglycine:< or =2.00 mg/g Creatinine
3-Phenylpropionylglycine:< or =2.00 mg/g Creatinine
trans-Cinnamoylglycine:< or =5.50 mg/g Creatinine
Suberylglycine:< or =5.00 mg/g Creatinine
Dodecanedioic acid:< or =0.50 mg/g Creatinine
Tetradecanedioic acid:< or =0.50 mg/g Creatinine
Hexadecanedioic acid:< or =0.50 mg/g Creatinine

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 phone number of key contacts who may provide these studies at Mayo Clinic or elsewhere; and a phone number to reach one of the laboratory directors in case the referring physician has additional questions.

Cautions

No significant cautionary statements

Clinical Reference

1. Rinaldo P, Hahn SH, Matern D. Inborn errors of amino acid, organic acid, and fatty acid metabolism. In: Burtis CA, Ashwood ER, Bruns DE eds. Tietz Textbook of Clinical Chemistry and Molecular Diagnostics. 4th ed. WB Saunders Company; 2005:2207-2247

2. Roe CR, Ding J. Mitochondrial fatty acid oxidation disorders. In: Valle D, Antonarakis S, Ballabio A, Beaudet A, Mitchell GA. eds. The Online Metabolic and Molecular Bases of Inherited Disease. McGraw-Hill; 2019. Accessed March 21, 2024. http://ommbid.mhmedical.com/content.aspx?bookid=2709&sectionid=225087274
3. Kolker S, Cazorla AG, Valayannopoulos V, et al. The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 1: the initial presentation. J Inherit Metab Dis. 2015;38(6):1041-1057. doi:10.1007/s10545-015-9839-3
4. Tuncel AT, Boy N, Morath MA, Horster F, Mutze U, Kolker S. Organic acidurias in adults: late complications and management. J Inherit Metab Dis. 2018;41(5):765-776. doi:10.1007/s10545-017-0135-2
5. Pasquali M, Longo N. Newborn screening and inborn errors of metabolism. In: Rifai N, Chiu, RWK, Young I, Burnham CD, Wittwer CT, eds. Tietz Textbook of Laboratory Medicine. 7th ed. Elsevier; 2023:chap 60

Performance

Method Description
Urine volumes equivalent to 0.25 to 0.50 mg of creatinine are spiked with the mixture of labeled internal standards, allowed to equilibrate, acidified, and then extracted. After evaporation, the dry residue is derivatized to butyl esters. Specimens are analyzed by capillary gas chromatography mass spectrometry selected ion monitoring using ammonia chemical ionization and a stable isotope dilution method.(Unpublished Mayo method)

PDF Report
No

Day(s) Performed
Monday, Thursday

Report Available
3 to 6 days

Specimen Retention Time
3 months

Performing Laboratory Location
Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees
- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

Test Classification
This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA

requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

82542

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
AGU20	Acylglycines, QN, U	50334-2

Result ID	Test Result Name	Result LOINC® Value
608931	Interpretation	53718-3
608910	n-Acetylglycine	99059-8
608911	n-Propionylglycine	13800-8
608912	Isobutyrylglycine	38360-4
608913	Ethylmalonic acid	13741-4
608914	n-Butyrylglycine	27892-9
608915	2-Methylsuccinic acid	13777-8
608916	2-Methylbutyrylglycine	27097-5
608917	Isovalerylglycine	13766-1
608918	Glutaric acid	13748-9
608919	3-Methylcrotonylglycine	13691-1
608920	n-Tiglylglycine	13816-4
608921	3-Methylglutaconic acid	13692-9
608922	n-Hexanoylglycine	13753-9
608923	n-Octanoylglycine	38367-9
608924	3-Phenylpropionylglycine	13793-5
608925	trans-Cinnamoylglycine	38417-2
608926	Suberylglycine	13811-5
608927	Dodecanedioic acid	13732-3
608928	Tetradecanedioic acid	50333-4
608929	Hexadecanedioic acid	50332-6
608930	Reviewed By	18771-6