

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



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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. <u>Biochemical Genetics Patient Information</u> (T602)
- 2. <u>If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request</u> (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



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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



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- 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§ionid=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 <u>Test Prices</u> for detailed fee information.
- Clients without access to Test Prices can contact <u>Customer Service</u> 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact <u>Customer Service</u>.

Test Classification

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



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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 |