MAYO CLINIC LABORATORIES

Test Definition: C5OHU

C5-OH Acylcarnitine, QN, U

Overview

Useful For

Evaluation of patients with an abnormal newborn screen showing elevations of 3-hydroxyisovaleryl-/2-methyl-3-hydroxybutyryl-carnitine (C5-OH)

Genetics Test Information

Evaluation of patients with an abnormal newborn screen showing elevations of 3-hydroxyisovaleryl-/2-methyl-3-hydroxy acylcarnitine (C5-OH).

Highlights

Elevated 3-hydroxyisovaleryl-/2-methyl-3-hydroxy acylcarnitine (C5-OH in plasma or newborn screening blood spots is due to one of several biochemical genetic diagnoses: 3-methylcrotonylglycinuria, 3-hydroxy 3-methylglutaryl-(HMG)-CoA lyase deficiency, beta-ketothiolase deficiency, 2-methyl 3-hydroxybutyryl-CoA dehydrogenase deficiency, 3-methylglutaconic aciduria, biotinidase deficiency or holocarboxylase deficiency.
Urine C5OH is useful in differentiating patients with 3-methylcrotonylglycinuria and with 3-methylglutaconic aciduria as they typically excrete larger amounts of C5-OH in urine compared to patients with the other diagnoses.

Method Name

Flow Injection Analysis-Tandem Mass Spectrometry (FIA-MS/MS)

NY State Available

Yes

Specimen

Specimen Type

Urine

Ordering Guidance

This second-tier test is used specifically to evaluate a newborn screening elevation of 3-hydroxyisovaleryl-/2-methyl-3-hydroxy acylcarnitine (C5-OH) and **must not** be ordered with either C4U / C4 Acylcarnitine, Quantitative, Random, Urine or C5DCU / C5-DC Acylcarnitine, Quantitative, Random, Urine.
For general screening for metabolic disorders, see OAU / Organic Acids Screen, Random, Urine; ACRN / Acylcarnitines, Quantitative, Plasma; and AAQP / Amino Acids, Quantitative, Plasma.

Necessary Information

Include patient's age, family history, clinical condition (asymptomatic or acute episode), diet, and drug therapy information.

Specimen Required

Patient Preparation: If clinically feasible, discontinue L-carnitine supplementation at least 72 hours before specimen collection.

Supplies: Urine Tubes, 10 mL (T068)
Collection Container/Tube: Clean, plastic urine collection container
Submission Container/Tube: Plastic, 10-mL urine tube
Specimen Volume: 5 mL
Collection Instructions:
1. Collect a random urine specimen.
2. Freeze specimen immediately.

Forms

If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

Reject Due To

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

Specimen Minimum Volume

1 mL

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Urine	Frozen (preferred)	7 days	
	Refrigerated		

Clinical & Interpretive

Clinical Information

The differential diagnosis of an isolated elevation of 3-hydroxyisovaleryl-/2-methyl-3-hydroxy acylcarnitine (C5-OH) in plasma or (newborn screening) blood spots includes the following disorders:
-3-Methylcrotonyl-CoA carboxylase deficiency (common name: 3-methylcrotonylglycinuria), either infantile or maternal
-3-Hydroxy 3-methylglutaryl-(HMG)-CoA lyase deficiency
-Beta-ketothiolase deficiency
-2-Methyl 3-hydroxybutyryl-CoA dehydrogenase deficiency
-3-methylglutaconic aciduria type I
-Biotinidase deficiency
-Holocarboxylase deficiency
Confirmatory and diagnostic testing are necessary to differentiate these clinical entities. This test can be used to differentiate patients with 3-methylcrotonylglycinuria and with 3-methylglutaconic aciduria as they typically excrete larger amounts of C5-OH in urine compared to patients with the other diagnoses.
The American College of Medical Genetics and Genomics (ACMG) newborn screening work group published diagnostic algorithms for the follow-up of infants who had positive newborn screening results. For more information, see the Practice Resources: ACT Sheets and Algorithms at http://www.acmg.net.

Reference Values

<2.93 millimoles/mole creatinine

Interpretation

Preliminary data showed that an elevated excretion in urine and concentration in plasma of 3-hydroxyisovaleryl-/2-methyl-3-hydroxy acylcarnitine (C5-OH) can be the only biochemical abnormalities in patients with 3-methylcrotonylglycinuria.

Cautions

The results of urine acylcarnitines are typically not informative when the patient is receiving L-carnitine supplements.

Clinical Reference

Wolfe LA, Finegold DN, Vockley J, et al: Potential misdiagnosis of 3-methylcrotonyl-coenzyme A carboxylase deficiency associated with absent or trace urinary 3-methylcrotonylglycine. Pediatrics. 2007 Nov;120(5):e1335-1340

Performance

Method Description

Acylcarnitines, including 3-hydroxy isovalerylcarnitine, are determined in urine by flow injection analysis tandem mass spectrometry using acetyl-d3-carnitine, propionyl-d3-carnitine, butyryl-d3-carnitine, octanoyl-d3-carnitine, dodecanoyl-d3-carnitine, and palmitoyl-d3-carnitine as internal standards. The supernatant is evaporated and the residue treated with n-butanolic hydrochloric acid yielding the acylcarnitines for analysis as their n-butyl esters.(Tortorelli S, Hahn SH, Cowan TM, Brewster TG, Rinaldo P, Matern D: The urinary excretion of glutarylcarnitine is an informative tool in the biochemical diagnosis of glutaric acidemia type I. Mol Genet Metab. 2005 Feb;84(2):137-143)

PDF Report

No

Specimen Retention Time

1 month

Performing Laboratory Location

Rochester

Fees & Codes

Test Classification

This test was developed, and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

82017

LOINC® Information

Test Definition: C5OHU

C5-OH Acylcarnitine, QN, U

Test ID	Test Order Name	Order LOINC Value
C5OHU	C5-OH Acylcarnitine, QN, U	50091-8

Result ID	Test Result Name	Result LOINC Value
88830	C5-OH Acylcarnitine, QN, U	50091-8
28125	C5-OH Interpretation	59462-2
34469	Reviewed By	18771-6