MAYO CLINIC LABORATORIES

Test Definition: C5DCU

C5-DC Acylcarnitine, Quantitative, Random, Urine

Overview

Useful For

Evaluation of patients with an abnormal newborn screen showing elevations of glutarylcarnitine

Diagnosis of glutaric aciduria type 1 deficiency

Genetics Test Information

Elevated glutarylcarnitine (C5-DC) in plasma or newborn screening blood spots is due to glutaric aciduria type 1 (GA-1), also known as glutaric acidemia type 1.

Urine C5-DC is a biochemical marker of GA-1 that appears to be elevated even in low excretors, who are affected patients with normal levels of glutaric acid in urine.

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 glutarylcarnitine and **must not** be ordered with either C4U / C4 Acylcarnitine, Quantitative, Random, Urine or C5OHU / C5-OH 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.

Specimen Minimum Volume
1 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)	7 days	
	Refrigerated	24 hours	

Clinical & Interpretive

Clinical Information
An isolated elevation of glutarylcarnitine (C5-DC) in plasma or newborn screening blood spots is related to a diagnosis of glutaric aciduria type 1 (GA-1), also known as glutaric acidemia type 1. GA-1 is caused by a deficiency of glutaryl-CoA dehydrogenase. Diagnostic testing by acylcarnitine analysis, including the evaluation of C5DC in urine, is helpful to determine if a patient has GA1.(1) Urinary excretion of C5-DC is a specific biochemical marker of GA-1 that appears to be elevated even in low excretors, those patients who are affected but have normal levels of glutaric acid in urine.

GA-1 is characterized by bilateral striatal brain injury leading to dystonia, often a result of acute neurologic crises triggered by illness. Many affected individuals also have macrocephaly. Dietary treatment and aggressive interventions during time of illness are recommended to try to prevent or minimize neurologic injury, which is most likely to occur in infancy and early childhood. Prevalence is approximately 1 in 100,000 individuals.

The American College of Medical Genetics and Genomics newborn screening work group published diagnostic algorithms for the follow-up of infants who had a positive newborn screening result. For more information, see the Practice Resources: ACT Sheets and Algorithms at www.acmg.net.

Reference Values
<1.54 millimoles/mole creatinine

Interpretation

Elevated excretion of glutarylcarnitine is a specific biochemical marker of glutaric aciduria type 1 that is elevated in affected patients, apparently even in low excretors, ie, those affected individuals with normal levels of glutaric acid in urine.

Cautions

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

Clinical Reference

1. Miller MJ, Cusmano-Ozog K, Oglesbee D, Young S; ACMG Laboratory Quality Assurance Committee: Laboratory analysis of acylcarnitines, 2020 update: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2021;23(2):249-258

2. Kolker S, Christensen E, Leonar JV, et al. Diagnosis and management of glutaric aciduria type I-revised recommendations. J Inherit Metab Dis. 2011;34(3):677-694

3. Guenzel AJ, Hall P, Scott AI, et al. The low excretor phenotype of glutaric acidemia type I is a source of false negative newborn screening results and challenging diagnoses. JIMD Rep.2021;60(1)67–74

Performance

Method Description

Acylcarnitines, including glutarylcarnitine, 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, et al. The urinary excretion of glutarylcarnitine is an informative tool in the biochemical diagnosis of glutaric acidemia type I. Mol Genet Metab. 2005;84(2):137-143; Miller MJ, Cusmano-Ozog K, Oglesbee D, Young S; ACMG Laboratory Quality Assurance Committee. Laboratory analysis of acylcarnitines, 2020 update: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2021;23(2):249-258)

PDF Report

No

Day(s) Performed

Monday, Wednesday, Friday

Report Available

2 to 5 days

Specimen Retention Time

1 month

Performing Laboratory Location

Rochester

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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

82017

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
C5DCU	C5-DC Acylcarnitine, QN, U	54279-5

Result ID	Test Result Name	Result LOINC® Value
88831	C5-DC Acylcarnitine, QN, U	54279-5
28126	C5-DC Interpretation	59462-2
34470	Reviewed By	18771-6