

Overview

Useful For

Evaluation of patients with a clinical suspicion of a wide range of conditions including organic acidemias and fatty acid oxidation disorders

Monitoring carnitine treatment

Highlights

Carnitine levels are disturbed in primary disorders of the carnitine cycle, or secondary disturbances of carnitine metabolism due to other biochemical disorders.

While plasma or serum carnitine is reduced in primary carnitine deficiency, urine carnitine may be normal or elevated.

Dietary intake (meat, carnitine supplementation) may cause increased carnitine values.

Abnormal results are accompanied by detailed interpretation including recommendations for follow-up testing.

Method Name

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

NY State Available

Yes

Specimen

Specimen Type

Urine

Specimen Required

Supplies: Urine Tubes, 10 mL (T068)

Container/Tube: Plastic, 10-mL urine tube

Specimen Volume: 1.5 mL

Collection Instructions: Collect a random urine specimen.

Forms

If not ordering electronically, complete, print, and send an [Inborn Errors of Metabolism 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) | 365 days | |
| | Refrigerated | 72 hours | |

Clinical and Interpretive**Clinical Information**

Carnitine and its esters are required for normal energy metabolism and serve 4 primary functions:

- Importing long-chain fatty acids into the mitochondria
- Exporting naturally occurring short-chain acyl-CoA groups from the mitochondria
- Maintaining the ratio of free CoA to esterified CoA
- Removing potentially toxic acyl-CoA groups from the cells and tissues

Evaluation of carnitine in serum, plasma, and urine screens patients for suspected primary disorders of the carnitine cycle, or secondary disturbances in carnitine levels as a result of organic acidemias and fatty acid oxidation disorders. In the latter, acyl-CoA groups accumulate and are excreted into the urine and bile as carnitine derivatives, resulting in a secondary carnitine deficiency. More than 100 such primary and secondary disorders have been described. Collectively, their incidence is approximately 1 in 1,000 live births. Primary carnitine deficiency has an incidence of approximately 1 in 21,000 live births based on Minnesota newborn screening data.

Other conditions that could cause an abnormal carnitine level include neuromuscular diseases, gastrointestinal disorders, familial cardiomyopathy, renal tubulopathies and chronic renal failure (dialysis), and prolonged treatment with steroids, antibiotics (pivalic acid), anticonvulsants (valproic acid), and total parenteral nutrition.

Follow-up testing is required to differentiate primary and secondary carnitine deficiencies and to elucidate the exact cause.

Reference Values

FREE

77-214 nmol/mg of creatinine

TOTAL

180-412 nmol/mg of creatinine

RATIO

Acyl to free: 0.7-3.4

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 a phone number to reach one of the laboratory directors in case the referring physician has additional questions.

Cautions

Increased values may be obtained after carnitine supplementation or meat consumption.

Clinical Reference

1. Magoulas PL, El-Hattab AW: Systemic primary carnitine deficiency: an overview of clinical manifestations, diagnosis, and management. *Orphanet J Rare Dis* 2012;7:68
2. Longo N, Amat di San Filippo C, Pasquali M: Disorders of carnitine transport and the carnitine cycle. *Am J Med Genet C Semin Med Genet* 2006;142C(2):77-85
3. Zammit VA, Ramsay RR, Bonomini M, Arduini A: Carnitine, mitochondrial function and therapy. *Adv Drug Deliv Rev* 2009;61(14):1353-1362
4. El-Hattab AW: Systemic Primary Carnitine Deficiency. In *GeneReviews* Edited by MP Adam, HH Ardinger, RA Pagon, et al. University of Washington, Seattle; 1993-2019. 2012 Mar 15 (Updated 2016 Nov 3) Accessed 3/22/2019. Available at www.ncbi.nlm.nih.gov/books/NBK84551/

Performance**Method Description**

Free and total carnitine are measured by tandem mass spectrometry (MS/MS) stable isotope dilution analysis. Hydrolysis enables measurement of total carnitine, and esterified carnitine (acylcarnitine) is calculated as the difference between the total and free carnitine. Quantification is enabled using deuterium labeled carnitine (d3-carnitine) added as internal standard. A selected reaction monitoring experiment is performed by MS/MS. The first mass spectrometer (Q1) detects carnitine and d3-carnitine precursors, and transmits them to a collision cell (Q2) within the mass spectrometer where they are fragmented. Specific fragments derived from the carnitine and internal standard are monitored in the second mass spectrometer (Q3). (Stevens RD, Hillman SL, Worthy S, et al: Assay for free and total carnitine in human plasma using tandem mass spectrometry. *Clin Chem* 2000;46:727-729)

PDF Report

No

Day(s) and Time(s) Test Performed

Wednesday; 8 a.m.

Analytic Time

7 days

Maximum Laboratory Time

9 days

Specimen Retention Time

1 month

Performing Laboratory Location

Rochester

Fees and 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 Regional Manager. 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. This test has not been cleared or approved by the U.S. Food and Drug Administration.

CPT Code Information

82379

LOINC® Information

| Test ID | Test Order Name | Order LOINC Value |
|---------|-----------------|-------------------|
| CARNU | Carnitine, U | In Process |

| Result ID | Test Result Name | Result LOINC Value |
|-----------|------------------|--------------------|
| 27121 | Total | 17866-5 |
| 27122 | Free (FC) | 17867-3 |
| 15789 | AC/FC Ratio | 43576-8 |
| 21550 | Interpretation | 59462-2 |