MAYO CLINIC LABORATORIES

Test Definition: CARN

Carnitine, Plasma

Overview

Useful For

Evaluation of patients with a clinical suspicion of a wide range of conditions including organic acidemias, fatty acid oxidation disorders, and primary carnitine deficiency using plasma specimens

Highlights

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

Additional testing is required to distinguish between primary and secondary deficiencies of carnitine.

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

Plasma

Necessary Information

Patient's age is required.

Specimen Required

Supplies: Sarstedt Aliquot Tube, 5 mL (T914)
Collection Container/Tube:
Preferred: Green top (sodium heparin)
Acceptable: Lavender top (EDTA), green top (lithium heparin)
Submission Container/Tube: Plastic vial
Specimen Volume: 0.5 mL
Collection Instructions: Centrifuge and aliquot plasma into a plastic vial

Forms

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

Specimen Minimum Volume

0.2 mL

Reject Due To

Gross hemolysis	OK
Gross lipemia	OK
Gross icterus	OK

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Plasma	Frozen (preferred)	60 days	
	Refrigerated	21 days	
	Ambient	7 days	

Clinical & 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 is a biochemical screening test 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 1000 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

	Total carnitine	Free carnitine	Acylcarnitine (AC)	AC/FC Ratio

	(TC)	(FC)		
Age Group	**Range***	**Range***	**Range***	**Range**
< or =1 day	23-68	12-36	7-37	0.4-1.7
2-7 days	17-41	10-21	3-24	0.2-1.4
8-31 days	19-59	12-46	4-15	0.1-0.7
32 days-12 months	38-68	27-49	7-19	0.2-0.5
13 months-6 years	35-84	24-63	4-28	0.1-0.8
7-10 years	28-83	22-66	3-32	0.1-0.9
11-17 years	34-77	22-65	4-29	0.1-0.9
> or =18 years	34-78	25-54	5-30	0.1-0.8

*Values expressed as nmol/mL

Schmidt-Sommerfeld E, Werner E, Penn D: Carnitine plasma concentrations in 353 metabolically healthy children. Eur J Pediatr. 1988;147:356-360

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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;18;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: Adam MP, Ardinger HH, Pagon RA, et al, eds. GeneReviews [Internet]. University of Washington, Seattle; 2012. Updated November 3, 2016. Accessed November 27, 2023. Available at www.ncbi.nlm.nih.gov/books/NBK84551/

5. Longo N., Frigeni M., Pasquali M. Carnitine transport and fatty acid oxidation. Biochim. Biophys. Acta. 2016;1863:2422-2435

6. Almannai M, Alfadhel M, El-Hattab AW: Carnitine inborn errors of metabolism. Molecules. 2019;24(18):3251

Performance

Method Description

Free and total carnitines 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 detects carnitine and d3-carnitine precursors and transmits them to a collision cell within the mass spectrometer where they are fragmented. Specific fragments derived from the carnitine and internal standard are monitored in the second mass spectrometer.(Stevens RD, Hillman SL, Worthy S, Sanders D, Millington DS. Assay for free and total carnitine in human plasma using tandem mass spectrometry. Clin Chem. 2000;46(5):727-729; 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 through Friday

Report Available
3 to 5 days

Specimen Retention Time
1 month

Performing Laboratory Location
Rochester

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
82379

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
CARN	Carnitine, P	97182-0

Result ID	Test Result Name	Result LOINC® Value

32050	Total	14288-5
32051	Free (FC)	14286-9
32052	Acylcarnitine (AC)	14282-8
21032	AC/FC Ratio	30193-7
21549	Interpretation	59462-2