Quest Diagnostics

Report Status: Final

Patient Information	Specimen Information	Client Information
DOB: **AGE:** Gender: Fasting: Phone: Patient ID:	Specimen: Requisition: Lab Ref #: Collected: Received: Reported:	REQUEST A TEST - PW N HEALTH 7027 MILL RD STE 201 BRECKSVILLE, OH 44141-1852

Test Name	In Range	Out Of Range	Reference Range	Lab
CARNITINE				EZ
CARNITINE, TOTAL	42		30-70 umol/L	
CARNITINE, FREE	31		23-59 umol/L	
CARNITINE ESTERS	12		4-15 umol/L	
ESTERIFIED/FREE RATIO:	0.38		0.12-0.39	

This serum carnitine profile is normal. Clinical correlation is recommended.

Interpretation reviewed by: Denise Salazar, Ph.D., FACMG.-
IF YOU HAVE ANY QUESTIONS REGARDING THESE RESULTS, PLEASE CONTACT THE QUEST DIAGNOSTICS BIOCHEMICAL GENETICS LABORATORY AT 1-800-642-4657 ext 4817 or ext 4423 AND ASK TO SPEAK WITH THE LABORATORY DIRECTOR ON CALL. FOR GENERAL QUESTIONS ABOUT QUEST DIAGNOSTICS GENETIC TESTING, PLEASE CALL THE GENE INFO LINE AT 1-866-GENE-INFO.-

Carnitine, an amino acid synthesized in animal tissues from lysine and methionine by an iron-ascorbate dependent pathway, functions as a carrier of fatty acids across cell membranes. Serum carnitine analysis is useful in the diagnosis and monitoring of patients with carnitine deficiency (either primary or secondary). Primary systemic carnitine deficiency (CDSP), also known as carnitine uptake defect (CUD) is an autosomal recessive disorder that affects carnitine uptake by cells and tissues through a defect in the plasma membrane carnitine transporter (OCTN2, encoded by the SLC22A5 gene located on chromosome 5q31.1). The incidence of CDSP is approximately 1 in 50,000. The clinical presentation and age of onset of CDSP can vary, but typical findings include hypoketotic hypoglycemia, hepatomegaly, cardiomyopathy, skeletal myopathy, and weakness. If diagnosed early, treatment with carnitine can reverse many of the clinical symptoms. Secondary carnitine deficiency occurs in some disease states, in patients on carnitine-poor diets, or secondary to medications including valproic acid. It is also found in a number of metabolic disorders affecting fatty acid and organic acid metabolism. In these disorders, carnitine complexes with the accumulated substrate of the blocked metabolic step, and the resulting acylcarnitines are excreted in the urine, leading to a depletion of carnitine in the patient. Elevated free carnitine can occur in CPT-1 deficiency or secondary to dietary carnitine supplementation, while increased esterified carnitine may indicate an underlying metabolic defect. This test is not intended to diagnose these disorders. All results should be interpreted in the context of clinical findings, relevant history, and other laboratory data.

This test was developed and its analytical performance characteristics have been determined by Quest Diagnostics Nichols Institute San Juan Capistrano. It has not been cleared or approved by FDA. This assay has been validated

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pursuant to the CLIA regulations and is used for clinical purposes.

PERFORMING SITE:

EZ QUEST DIAGNOSTICS/NICHOLS SJC, 33608 ORTEGA HWY, SAN JUAN CAPISTRANO, CA 92675-2042 Laboratory Director: IRINA MARAMICA,MD,PHD,MBA, CLIA: 05D0643352