

**THIS IS NOT A TEST REQUEST FORM.**  
**The information below is required to perform Primary Carnitine Deficiency (SLC22A5) testing.**  
**Please fill out this form and submit it with the test request form or electronic packing list.**

**PATIENT HISTORY FOR PRIMARY CARNITINE DEFICIENCY (SLC22A5) TESTING**

Patient's Name \_\_\_\_\_ Date of Birth \_\_\_\_/\_\_\_\_/\_\_\_\_ Gender  F  M

Physician \_\_\_\_\_ Physician Phone (\_\_\_\_) \_\_\_\_\_ Practice Specialty \_\_\_\_\_

Genetic Counselor \_\_\_\_\_ Counselor Phone (\_\_\_\_) \_\_\_\_\_

**Patient's Ethnicity** (check all that apply)

- African American     Ashkenazi Jewish     Asian     Caucasian  
 Hispanic     Middle Eastern     Native American     Other \_\_\_\_\_

**Did the patient have an abnormal NEWBORN SCREEN?**     No     Yes     Unknown

**Did the patient's CHILD have an abnormal newborn screen?**     No     Yes     N/A

If yes, describe result \_\_\_\_\_

**Does the patient have SYMPTOMS?**     No     Yes     Unknown

If yes, check all that apply

- Hypoglycemia     Cardiomyopathy     Hypotonia     Other \_\_\_\_\_

**LABORATORY FINDINGS**

Plasma carnitine (**without supplements**):

- Free:     Normal     Low     High     Unknown     Not performed  
Total:     Normal     Low     High     Unknown     Not performed

URINE carnitine (**without supplements**):

- Free:     Normal     Low     High     Unknown     Not performed  
Total:     Normal     Low     High     Unknown     Not performed

Measured carnitine transport activity in fibroblast (% of control) \_\_\_\_\_

**Does the patient have a FAMILY HISTORY OF PRIMARY CARNITINE DEFICIENCY?**     No     Yes

If yes, attach a PEDIGREE or specify the **RELATIONSHIP** of family member(s) to the patient. \_\_\_\_\_

Is the relative  a healthy carrier     affected with Primary Carnitine Deficiency?

What are the *SLC22A5* mutations in the family member? \_\_\_\_\_

**Circle the test below you intend to order.**

**2004203 Primary Carnitine Deficiency (SLC22A5) Sequencing and Deletion/Duplication:** Clinical sensitivity may be as high as 95%.

**0051682 Primary Carnitine Deficiency (SLC22A5) Sequencing:** Clinical sensitivity is ~80%.

**2004199 Primary Carnitine Deficiency (SLC22A5) Deletion/Duplication:** Clinical sensitivity may be as high as 10-15%.

**2001961 Familial Mutation, Targeted Sequencing:** Tests for a *SLC22A5* mutation previously identified in a family member. A copy of the relative's DNA lab report is REQUIRED for this test.

**0080068 Carnitine, Free & Total, Plasma:** Initial test for individuals with symptoms or abnormal newborn screen.

**0080512 Carnitine Transport, Fibroblasts:** Measures carnitine transport activity in fibroblasts.

**For questions, contact an ARUP genetic counselor at (800) 242-2787 ext. 2141**

Master Label