ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

A nonprofit enterprise of the University of Utah and its Department of Pathology

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THIS IS NOT A TEST REQUEST FORM.
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 Name _____ Date of Birth _____ Sex ☐ F ☐ M

Physician _____ Physician Phone _____

Practice Specialty _____ Physician Fax _____

Genetic Counselor _____ Counselor Phone _____

Patient's Ethnicity (check all that apply)

☐ African-American ☐ Asian ☐ Hispanic ☐ Native American

☐ Ashkenazi Jewish ☐ Caucasian ☐ Middle Eastern ☐ Other: _____

Did the patient have an abnormal newborn screen? ☐ No ☐ Yes ☐ Unknown

Has the patient's mother been evaluated for primary carnitine deficiency? ☐ No ☐ Yes ☐ Unknown

Is the patient's mother on a vegan or vegetarian diet? ☐ No ☐ Yes ☐ Unknown

If yes, describe: _____

Did the patient's child have an abnormal newborn screen? ☐ No ☐ Yes ☐ Unknown

If yes, describe: _____

Does the patient have symptoms? ☐ No ☐ Yes (check all that apply) ☐ N/A

☐ Hypoglycemia ☐ Cardiomyopathy ☐ Hypotonia ☐ Other symptom(s): _____

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

Is there any relevant family history? ☐ No ☐ Yes ☐ Unknown

If yes, attach a pedigree or specify the relative's relationship to the patient. _____

The relative is: ☐ a healthy carrier ☐ affected with Primary Carnitine Deficiency

Has DNA testing been performed for the family member(s)? ☐ No ☐ Yes ☐ Unknown

If yes, attach a copy of the relative's DNA laboratory result. (REQUIRED for familial mutation testing)

Identify the *SLC22A5* variants in the family member: _____

Check the test 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%.

☐ **0080068 Carnitine, Free & Total (performed on plasma):** Initial test for individuals with symptoms or abnormal newborn screen.

☐ **2001961 Familial Mutation, Targeted Sequencing.** Tests for a mutation previously identified in a family member; a copy of relative's lab result is REQUIRED.

Master Label

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