ARUP LABORATORIES

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 complete and submit with the test request form or electronic packing list.

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

Patient Name: _____ **Date of Birth:** _____ **Sex:** ☐ Female ☐ Male

Ordering Provider: _____ **Provider's Phone:** _____

Practice Specialty: _____ **Provider's Fax:** _____

Genetic Counselor: _____ **Counselor Phone:** _____

Patient's Ethnicity/Ancestry (check all that apply)

☐ African American/Black ☐ Asian ☐ Hispanic ☐ White ☐ Other: _____

List country of origin (if known): _____

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 up to 95%.

☐ **0080068 Carnitine, Free & Total (Includes Carnitine, Esterified):** 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.