ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

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

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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? ☐ N ☐ Y ☐ Unknown

Has the patient's mother been evaluated for primary carnitine deficiency? ☐ N ☐ Y ☐ Unknown

Is the patient's mother on a vegan or vegetarian diet? ☐ N ☐ Y ☐ Unknown

If yes, describe:_____

Did the patient's child have an abnormal newborn screen? ☐ N ☐ Y ☐ N/A

If yes, describe result:_____

Does the patient have symptoms? ☐ N ☐ Y ☐ 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

Does the patient have a family history of carnitine deficiency? ☐ N ☐ Y

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* variants in the family member?_____

Check 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 (performed on plasma): Initial test for individuals with symptoms or abnormal newborn screen.

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

Master Label