ARUP LABORATORIES

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 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