

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 Ashkenazi Jewish Asian Caucasian
 Hispanic Middle Eastern Native American 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? _____

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 (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

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