ARUP LABORATORIES

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform *SLC25A13* genetic testing.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR CITRIN DEFICIENCY (*SLC25A13*) TESTING

Patient's Name _____ Date of Birth_____ /_____/_____Gender [ ] F [ ] M

Physician _____ Physician Phone (_____)_____Practice Specialty_____

Genetic Counselor _____ Counselor Phone (_____)_____

Patient's Ethnicity (check all that apply)

[ ] Chinese [ ] Japanese [ ] Korean [ ] Taiwanese
[ ] Vietnamese [ ] Asian, other _____

[ ] African American [ ] Ashkenazi Jewish [ ] Caucasian [ ] Hispanic
[ ] Middle Eastern [ ] Native American [ ] Other_____

Did the patient have an abnormal NEWBORN SCREEN? [ ] No [ ] Yes [ ] Unknown

Does the patient have SYMPTOMS? [ ] No [ ] Yes (age of onset: _____) [ ] Unknown

If yes, check all that apply

[ ] Intrahepatic cholestasis [ ] Carbohydrate aversion [ ] Hypoproteinemia
[ ] Coma [ ] Seizures [ ] Growth retardation
[ ] Liver fibrosis [ ] Failure to thrive [ ] Hemolytic anemia
[ ] Neuropsychiatric symptoms, describe _____
[ ] Other _____

LABORATORY FINDINGS

Ammonia level	[ ] Normal	[ ] Abnormal (result: _____)	[ ] Not performed	[ ] Unknown
Plasma amino acids	[ ] Normal	[ ] Abnormal (result: _____)	[ ] Not performed	[ ] Unknown
Urine organic acids	[ ] Normal	[ ] Abnormal (result: _____)	[ ] Not performed	[ ] Unknown

Pancreatic secretory trypsin inhibitor (PSTI) [ ] Normal [ ] Abnormal (result: _____) [ ] Not performed [ ] Unknown

Is there a FAMILY HISTORY OF CITRIN DEFICIENCY? [ ] No [ ] Yes [ ] Unknown

If yes, describe relationship(s) to the patient _____

Has DNA testing for *SLC25A13* been performed for these family member(s)? [ ] No [ ] Yes [ ] Unknown
If yes, please attach a copy of the laboratory result. (REQUIRED for familial mutation testing.)

DESCRIPTION OF CITRIN DEFICIENCY (*SLC25A13*) MOLECULAR TESTS

2006261 Citrin Deficiency (*SLC25A13*) Sequencing: Sequencing of the *ASS1* coding regions and intron/exon boundaries. Clinical sensitivity is approximately 95 percent.

2001961 Familial Mutation, Targeted Sequencing: Targeted sequencing for an *ASS1* mutation previously identified in a family member.

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

Master Label