ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

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 fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR CITRIN DEFICIENCY (*SLC25A13*) 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)

☐ Chinese
☐ Japanese
☐ Korean
☐ Taiwanese
☐ Vietnamese
☐ African-American
☐ Ashkenazi Jewish
☐ Caucasian
☐ Hispanic
☐ Middle Eastern
☐ Native American
☐ Asian, other: _____
☐ Other: _____

Did the patient have an abnormal newborn screen? ☐ No ☐ Yes ☐ Unknown

Does the patient have symptoms? ☐ No ☐ Yes (check all that apply and describe) ☐ Unknown

Age of onset: _____

☐ Carbohydrate aversion
☐ Coma
☐ Failure to thrive
☐ Growth retardation
☐ Hemolytic anemia
☐ Hypoproteinemia
☐ Intrahepatic cholestasis
☐ Liver fibrosis
☐ Seizures
☐ Neuropsychiatric symptoms; describe: _____
☐ Other symptom(s): _____

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

Has the patient undergone previous DNA testing? ☐ No ☐ Yes ☐ Unknown

If yes, describe the genes, disorder, methodology, and results: _____

_____

Is there any relevant family history? ☐ No ☐ Yes ☐ Unknown

If yes, attach a pedigree or specify the relative's relationship to the patient: _____

Has DNA testing been for *SLC25A13* 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.)

Check the test you intend to order.

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

☐ **2001961 Familial Mutation, Targeted Sequencing:** Tests for a *SLC25A13* mutation previously identified in a family member; a copy of the relative's lab result is REQUIRED.

Master Label

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