ARUP LABORATORIES

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform *ASS1*genetic testing.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR CITRULLINEMIA TYPE 1 (*ASS1*) SEQUENCING

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

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

If yes, check all that apply

[ ] Hyperammonemia [ ] Coma [ ] Increased intracranial pressure
[ ] Lethargy [ ] Seizures [ ] Developmental delay
[ ] Vomiting [ ] Failure to thrive [ ] Other _____

LABORATORY FINDINGS

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

FAMILY HISTORY OF CITRULLINEMIA TYPE I? [ ] No [ ] Yes [ ] Unknown

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

Has DNA testing for *ASS1* 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 *ASS1* Deficiency Molecular Tests

2007069 Citrullinemia Type I (*ASS1*) Sequencing: Sequencing of the *ASS1* coding regions and intron/exon boundaries. Clinical sensitivity approximately 96%.

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