

**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 CITRULLINEMIA TYPE 1 (ASS1) SEQUENCING**

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     Asian     Hispanic     Native American  
 Ashkenazi Jewish     Caucasian     Middle Eastern     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)

Age of onset: \_\_\_\_\_  Failure to thrive     Lethargy  
 Coma     Hyperammonemia     Seizures  
 Developmental delay     Increased intracranial pressure     Vomiting  
 Other symptom(s): \_\_\_\_\_

**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

Is there any relevant **family history** of Citrullinemia Type I?  No  Yes  Unknown

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

Has DNA testing been 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.**

- 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; a copy of relative's lab result is REQUIRED.

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

**Master Label**