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 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