ARUP LABORATORIES

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 complete and submit with the test request form or electronic packing list.

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

Patient Name: _____ **Date of Birth:** _____ **Sex:** ☐ Female ☐ Male

Ordering Provider: _____ **Provider's Phone:** _____

Practice Specialty: _____ **Provider's Fax:** _____

Genetic Counselor: _____ **Counselor Phone:** _____

Patient's Ethnicity/Ancestry (check all that apply)

☐ African American/Black ☐ Asian ☐ Hispanic ☐ White ☐ Other: _____

List country of origin (if known): _____

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 the relative's lab result is REQUIRED.

Master Label

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