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.

PATIENT HISTORY FOR ORNITHINE TRANSCARBAMYLASE DEFICIENCY (OTC) TESTING

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): _____

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

☐ Coma ☐ Hyperammonemia ☐ Reye-like syndrome

☐ Cyclical vomiting ☐ Lethargy ☐ Seizures

☐ Encephalopathy ☐ Protein aversion

☐ Other symptom(s): _____

Laboratory Findings

Plasma amino acids ..... ☐ Normal ☐ Abnormal (result: _____) ☐ Not performed ☐ Unknown

Orotic acid ..... ☐ Normal ☐ Abnormal (result: _____) ☐ Not performed ☐ Unknown

Ammonia level ..... ☐ Normal ☐ Abnormal (result: _____) ☐ Not performed ☐ Unknown

Has the patient undergone previous DNA testing? ..... ☐ No ☐ Yes (check all that apply) ☐ Unknown

If yes, describe the test(s) and results: _____

_____

Is there any relevant family history?..... ☐ No ☐ Yes (check all that apply) ☐ Unknown

If yes, attach a pedigree or specify the relative's relationship to the patient. List their symptoms and age of onset: _____

_____

_____

Has DNA testing been performed for the family member(s)? ..... ☐ No ☐ Yes (check all that apply) ☐ 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.

☐ **2004896 Ornithine Transcarbamylase Deficiency (OTC) Sequencing and Deletion/Duplication:** Sequencing and deletion/duplication analysis of the OTC coding regions and intron/exon boundaries. Clinical sensitivity approaches 90%.

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

☐ **3003144 Deletion/Duplication Analysis by MLPA:** Tests for large deletion/duplication previously identified in a family member; a copy of a relative's lab report is REQUIRED.

Master Label

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