

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 ORNITHINE TRANSCARBAMYLASE DEFICIENCY (OTC) TESTING

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: _____

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

- Coma Protein aversion
 Cyclical vomiting Reye-like syndrome
 Encephalopathy Seizures
 Hyperammonemia Other symptom(s): _____
 Lethargy

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 Unknown

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

Is there any relevant family history? No Yes 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 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 Transcarbonylase Deficiency (OTC) Sequencing and Deletion/Duplication:** Sequencing and deletion/duplication analysis of the OTC coding regions and intron/exon boundaries. Clinical sensitivity approaches 90%.
- 2004901 Ornithine Transcarbonylase Deficiency (OTC) Sequencing:** Sequencing of the OTC coding regions and intron/exon boundaries. Clinical sensitivity approximately 80%.
- 2001961 Familial Mutation, Targeted Sequencing:** Tests for a mutation previously identified in a family member; a copy of relative's lab result is REQUIRED.

Master Label

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