

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform OTC deficiency testing.
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** ____/____/____ **Gender** F M
Physician _____ **Physician Phone** (____) _____ **Practice Specialty** _____
Genetic Counselor _____ **Counselor Phone #** (____) _____

PATIENT ETHNICITY (check all that apply)

- | | | | |
|---|---|--|--------------------------------------|
| <input type="checkbox"/> African American | <input type="checkbox"/> Ashkenazi Jewish | <input type="checkbox"/> Asian | <input type="checkbox"/> Caucasian |
| <input type="checkbox"/> Hispanic | <input type="checkbox"/> Middle Eastern | <input type="checkbox"/> Native American | <input type="checkbox"/> Other _____ |

SYMPTOMS of OTC No Yes Unknown

If yes, check all that apply

- | | | |
|---|--|---|
| <input type="checkbox"/> Hyperammonemia | <input type="checkbox"/> Cyclical vomiting | <input type="checkbox"/> Seizures |
| <input type="checkbox"/> Lethargy | <input type="checkbox"/> Coma | <input type="checkbox"/> Reye-like syndrome |
| <input type="checkbox"/> Encephalopathy | <input type="checkbox"/> Protein aversion | <input type="checkbox"/> Other _____ |

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

FAMILY HISTORY OF OTC DEFICIENCY? No Yes Unknown

If yes, describe relationship(s) to the patient _____

Has DNA testing for OTC been performed for these family member(s)? No Yes Unknown

If yes, please attach a copy of the laboratory result (REQUIRED for familial mutation testing)

Circle the OTC test you intend to order.

2004896 Ornithine Transcarbamyase 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 Transcarbamyase Deficiency (OTC) Sequencing: sequencing of the *OTC* coding regions and intron/exon boundaries. Clinical sensitivity approximately 80%.

2001961 Familial Mutation, Targeted Sequencing: Targeted sequencing for an *OTC* mutation previously identified in a family member.

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

Master Label