

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's Name _____ Date of Birth ____/____/____ Gender F M
 Physician _____ Physician Phone (____) _____ Practice Specialty _____
 Genetic Counselor _____ Counselor Phone # (____) _____

PATIENT ETHNICITY (check all that apply)

African American Ashkenazi Jewish Asian Caucasian
 Hispanic Middle Eastern Native American Other _____

SYMPTOMS of OTC No Yes Unknown

If yes, check all that apply

Hyperammonemia Cyclical vomiting Seizures
 Lethargy Coma Reye-like syndrome
 Encephalopathy Protein aversion 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 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%.

2004901 Ornithine Transcarbamylase Deficiency (OTC) Sequencing: sequencing of the *OTC* coding regions and intron/exon boundaries. Clinical sensitivity approximately 80%.

2004892 Ornithine Transcarbamylase Deficiency (OTC) Deletion/Duplication: Deletion/duplication analysis of the *OTC* coding regions. Clinical sensitivity approximately 10%.

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