ARUP LABORATORIES

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)

[ ] 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