ARUP LABORATORIES

A nonprofit enterprise of the University of Utah and its Department of Pathology

500 Chipeta Way
Salt Lake City, UT 84108-1221
phone: 801-583-2787 | toll free: 800-242-2787
fax: 801-584-5249 | aruplab.com

THIS IS NOT A TEST REQUEST FORM. Please complete and submit with the test request form or electronic packing list.

PATIENT HISTORY FOR *ATP7A* GENETIC 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): _____

Suspected Diagnosis: ☐ Menkes disease ☐ Occipital horn syndrome ☐ X-linked distal motor neuropathy

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

Age of onset: _____

☐ Hypotonia	☐ Muscle weakness	☐ Abnormal hair
☐ Intellectual disability	☐ Occipital horn	☐ Bladder diverticula
☐ Jowly appearance	☐ Seizures	☐ Failure to thrive
☐ Motor neuropathy	☐ Skin laxity	

☐ Other symptom(s): _____

Laboratory Findings

☐ Serum copper concentration	☐ Normal	☐ Abnormal (result: _____)	☐ Not performed	☐ Unknown
☐ Serum ceruloplasmin concentration	☐ Normal	☐ Abnormal (result: _____)	☐ Not performed	☐ Unknown
☐ Plasma catecholamine concentrations	☐ Normal	☐ Abnormal (result: _____)	☐ Not performed	☐ Unknown
☐ HVA: VMA ratio (urine)	☐ Normal	☐ Abnormal (result: _____)	☐ Not performed	☐ Unknown
☐ Nerve conduction tests	☐ Normal	☐ Abnormal (result: _____)	☐ Not performed	☐ Unknown

Is there any relevant family history of *ATP7A*-Related Disorder? ☐ No ☐ Yes ☐ Unknown

If yes, attach a pedigree or specify the relative's relationship to the patient: _____

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.

☐ **2008471 *ATP7A*-Related Copper Transport Disorders (*ATP7A*) Sequencing and Deletion/Duplication:** Sequencing of the *ATP7A* coding regions and deletion/duplication analysis for large gene rearrangements. Clinical sensitivity for Menkes disease and occipital horn syndrome is approximately 95%.

☐ **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

☐ **2001961 Familial Mutation, Targeted Sequencing:** Targeted sequencing for an *ATP7A* variant 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.