ARUP LABORATORIES

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform *ATP7A* genetic testing.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR *ATP7A* GENETIC TESTING

Patient Name _____ Date of Birth_____ /_____/_____Gender [ ] F [ ] M

Physician _____ Physician Phone (_____)_____Practice Specialty_____

Genetic Counselor _____ Counselor Phone (_____)_____

Patient's Ethnicity (check all that apply)

[ ] African American [ ] Ashkenazi Jewish [ ] Asian [ ] Caucasian
[ ] Hispanic [ ] Middle Eastern [ ] Native American [ ] Other_____

SUSPECTED DIAGNOSIS: [ ] Menkes disease [ ] Occipital horn syndrome [ ] X-linked distal motor neuropathy

Does the patient have SYMPTOMS? [ ] No [ ] Yes (age of onset: _____) [ ] Unknown

If yes, check all that apply

[ ] Hypotonia [ ] Abnormal hair [ ] Bladder diverticula
[ ] Seizures [ ] Jowly appearance [ ] Intellectual disability
[ ] Failure to thrive [ ] Skin laxity [ ] Muscle weakness
[ ] Occipital horn [ ] Motor Neuropathy [ ] Other _____

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 A FAMILY HISTORY OF *ATP7A*-RELATED DISORDER? [ ] No [ ] Yes [ ] Unknown

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

Has DNA testing for *ATP7A* 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).

DESCRIPTION OF *ATP7A*-RELATED MOLECULAR TESTS

2007872 *ATP7A*-Related Copper Transport Disorders (*ATP7A*), Sequencing: Sequencing of the *ATP7A* coding regions and intron/exon boundaries. Clinical sensitivity for Menkes disease and occipital horn syndrome is approximately 80 percent.

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 percent.

2008443 *ATP7A*-Related Copper Transporter Disorders *(ATP7A)* Deletion/Duplication: For patients with negative *ATP7A* sequencing results. Clinical sensitivity for Menkes disease and occipital horn syndrome is approximately 15 percent.

2001961 Familial Mutation, Targeted Sequencing: Targeted sequencing for an *ATP7A* mutation previously identified in a family member; copy of relative's lab result is REQUIRED.

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

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