ARUP LABORATORIES

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

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

PATIENT HISTORY FOR X-LINKED ADRENOLEUKODYSTROPHY (*ABCD1*) 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): _____

Does the patient have symptoms? .....☐ No ☐ Yes (check all that apply and describe)

Age of onset: _____

☐ Addison disease/adrenal insufficiency ☐ Cognitive impairment ☐ Seizures
☐ Ataxia ☐ Hearing loss ☐ Spasticity
☐ Behavioral disturbance ☐ Paralysis ☐ Visual impairment
☐ Abnormal brain MRI (describe): _____
☐ Other symptom(s): _____

Laboratory Findings

Newborn screen.....☐ Normal ☐ Abnormal (result: _____) ☐ Not performed ☐ Unknown
Very long chain fatty acids .....☐ Normal ☐ Abnormal (result: _____) ☐ Not performed ☐ Unknown
Beta-oxidation of C26:0 in fibroblasts .....☐ Normal ☐ Abnormal (result: _____) ☐ Not performed ☐ Unknown

Is there any relevant family history?.....☐ 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.

Biochemical Testing

☐ **2004250 Very Long-Chain and Branched-Chain Fatty Acids Profile:** Initial test to screen for disorders of peroxisomal biogenesis and/or function, including X-linked adrenoleukodystrophy.

Molecular Testing

☐ **2011906 Adrenoleukodystrophy, X-linked (*ABCD1*) Sequencing and Deletion/Duplication:** Sanger sequencing of *BCDE2* coding regions and MLPA for deletion/duplication analysis. Clinical sensitivity is~ 99%.

☐ **2011902 Adrenoleukodystrophy, X-linked (*ABCD1*) Sequencing:** Sanger sequencing of the *BCDE2* coding regions. Clinical sensitivity is ~93%.

☐ **2001961 Familial Mutation, Targeted Sequencing:** Targeted sequencing for an *BCDE2* mutation previously identified in a family member; a copy of relative's lab result is REQUIRED.

☐ **3003144 Deletion/Duplication Analysis by MLPA:** Large deletion/duplication analysis for a previously identified del/dup in a family member, a copy of a relative's lab report is REQUIRED.

Master Label

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