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.

PEROXISOMAL DISORDERS PATIENT HISTORY FORM

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): _____

Did the patient have an abnormal newborn screen? ☐ No ☐ Yes ☐ Unknown

If yes, please provide details: _____

Suspected Diagnosis: ☐ Zellweger spectrum disorder ☐ Rhizomelic chondrodysplasia punctata (RCDP)

☐ Other: _____

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

☐ Growth failure/feeding difficulties
☐ Hearing loss
☐ Visual impairment
☐ Liver dysfunction
☐ Renal insufficiency
☐ Adrenal insufficiency
☐ Skeletal abnormalities
☐ Seizures
☐ Hypotonia
☐ Brain MRI, abnormal findings: _____
☐ Other symptom(s): _____

Laboratory findings:

VLCFA levels (plasma)	☐ Normal	☐ Abnormal	☐ Not Performed
C26:0-lysophosphatidylcholine (LPC) (NBS/plasma)	☐ Normal	☐ Abnormal	☐ Not Performed
Plasmalogens (erythrocytes)	☐ Normal	☐ Abnormal	☐ Not Performed
Phytanic/pristanic acid levels (plasma)	☐ Normal	☐ Abnormal	☐ Not Performed
Pipecolic acid levels (plasma/urine)	☐ Normal	☐ Abnormal	☐ Not Performed
Bile acid intermediates (plasma/urine)	☐ Normal	☐ Abnormal	☐ Not Performed

Has the patient had an allogenic bone marrow or umbilical cord blood transplant? ☐ No ☐ Yes ☐ Unknown

Has the patient undergone previous DNA testing? ☐ No ☐ Yes ☐ Unknown

If yes, describe the test(s) and results: _____

Is there any relevant family history of peroxisomal disorders? ☐ No ☐ Yes ☐ Unknown

If yes, attach a pedigree or specify the relative's relationship to the patient. List their symptoms and age of onset: _____

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.

☐ **3002700 Peroxisomal Disorders Panel, Sequencing:** Recommended test to evaluate a suspected peroxisomal disorder

☐ **2001961 Familial Mutation, Targeted Sequencing:** Tests for a sequence variant previously identified in a family member; a copy of a relative's lab result is REQUIRED.

Master Label

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