

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
Sex Assigned at Birth: Female Male Intersex **Gender Identity (optional):** Female Male _____
Ordering Provider: _____ **Provider's Phone:** _____
Practice Specialty: _____ **Provider's Fax:** _____
Genetic Counselor: _____ **Counselor's 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

<input type="checkbox"/> Growth failure/feeding difficulties	<input type="checkbox"/> Seizures
<input type="checkbox"/> Hearing loss	<input type="checkbox"/> Hypotonia
<input type="checkbox"/> Visual impairment	<input type="checkbox"/> Brain MRI, abnormal findings: _____
<input type="checkbox"/> Liver dysfunction	_____
<input type="checkbox"/> Renal insufficiency	_____
<input type="checkbox"/> Adrenal insufficiency	<input type="checkbox"/> Other symptom(s): _____
<input type="checkbox"/> Skeletal abnormalities	_____

Laboratory findings:

VLCFA levels (plasma)	<input type="checkbox"/> Normal	<input type="checkbox"/> Abnormal	<input type="checkbox"/> Not Performed
C26:0-lysophosphatidylcholine (LPC) (NBS/plasma)	<input type="checkbox"/> Normal	<input type="checkbox"/> Abnormal	<input type="checkbox"/> Not Performed
Plasmalogens (erythrocytes)	<input type="checkbox"/> Normal	<input type="checkbox"/> Abnormal	<input type="checkbox"/> Not Performed
Phytanic/pristanic acid levels (plasma)	<input type="checkbox"/> Normal	<input type="checkbox"/> Abnormal	<input type="checkbox"/> Not Performed
Pipecolic acid levels (plasma/urine)	<input type="checkbox"/> Normal	<input type="checkbox"/> Abnormal	<input type="checkbox"/> Not Performed
Bile acid intermediates (plasma/urine)	<input type="checkbox"/> Normal	<input type="checkbox"/> Abnormal	<input type="checkbox"/> Not Performed

Has the patient had an allogeneic 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 variant testing).

Master Label

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