Elevated lysophosphatidylcholines C24:0, C26:0

POX / Fatty Acid Profile, Peroxisomal (C22-C26), Serum

Elevated very long chain fatty acids (VLCFA)

XALDZ / X-Linked Adrenoleukodystrophy, Full Gene Analysis, Varies

Genotype consistent with XALD (male) or XALD carrier status (female)

Variant of uncertain significance (VUS)

No mutation or VUS identified

Clinical suspicion* of a Zellweger spectrum disorder (ZSD) present

Absence of suspicion of a peroxisomal disorder

ABCD1 deletion/duplication

Red blood cell plasmalogens

BAIPD / Bile Acids for Peroxisomal Disorders, Serum

Consider:

LPGD / Lysosomal, Peroxisomal, Glycogen, and Neuronal Ceroid Lipofuscinosis Panels, Next-Generation Sequencing, Varies

Normal VLCFA

Not X-linked adrenoleukodystrophy (XALD)

No mutation or VUS identified

Absence of suspicion of a peroxisomal disorder

ABCD1 deletion/duplication

Refer to Genetics Specialist**

Variant of uncertain significance (VUS)

Elevated very long chain fatty acids (VLCFA)

XALDZ / X-Linked Adrenoleukodystrophy, Full Gene Analysis, Varies

Genotype consistent with XALD (male) or XALD carrier status (female)

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* Evaluate for presence of: dysmorphic features, hypotonia, vision abnormalities, hearing loss, renal cysts, abnormal coagulation studies and liver function tests

** Consider familial testing, especially for mother and male siblings