Newborn Screen Follow-up for X-Linked Adrenoleukodystrophy

Elevated lysophosphatidylcholines C24:0, C26:0

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

Elevated VLCFA

XALDZ / X-Linked Adrenoleukodystrophy, Full Gene Analysis

Normal VLCFA

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

No mutation or VUS identified

Absence of suspicion of a peroxisomal disorder

VUS

ABCD1 deletion/ duplication

Not XALD

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

Red blood cell plasmalogens

BAIPD / Bile Acids for Peroxisomal Disorders, Serum

Consider:
PDP / Peroxisomal Disorder Panel, by Next-Generation Sequencing

Refer to Genetics Specialist**

VLCFA=very long chain fatty acids
XALD=X-Linked Adrenoleukodystrophy
VUS=variant of uncertain significance

* 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