Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

[redacted]

Adrenoleukodystrophy, X-Linked (ABCD1) Sequencing and Deletion/Duplication

ARUP test code 2011906

ABCD1 Seq, Del Dup Specimen	Whole Blood
ABCD1 Seq, Del/Dup Interp	Negative

TEST PERFORMED - 2011906
TEST DESCRIPTION - Adrenoleukodystrophy, X-Linked (ABCD1) Sequencing and Deletion/Duplication
INDICATION FOR TEST - Confirm Diagnosis

RESULT
No pathogenic variants were detected in the ABCD1 gene.

INTERPRETATION
No pathogenic variants were detected in the ABCD1 gene by sequencing of all coding regions and intron-exon boundaries or by deletion/duplication analysis. This result decreases the likelihood that this individual is a symptomatic carrier of X-linked adrenoleukodystrophy. Please refer to the background information included in this report for the clinical sensitivity and limitations of this test.

RECOMMENDATIONS
Medical screening and management should rely on clinical and biochemical findings. Genetic consultation is recommended.

COMMENTS
Reference Sequence: GenBank # NM_000033.3 (ABCD1)
Nucleotide numbering begins at the "A" of the ATG initiation codon.
Likely benign and benign variants are not reported.

This result has been reviewed and approved by [redacted]

H=High, L=Low, *=Abnormal, C=Critical

Background Information: Adrenoleukodystrophy, X-Linked (ABCD1) Sequencing and Deletion/Duplication
Characteristics: X-linked adrenoleukodystrophy (X-ALD) is a peroxisomal disorder that affects the adrenal cortex and white matter of the nervous system. Clinical presentation is extremely variable, even within the same family, and can include a childhood cerebral form with rapid progression of neurologic disturbances, an adrenomyeloneuropathy form with neurological disturbances developing over decades, and primary adrenocortical insufficiency (Addison disease). Presentation in female carriers is variable.
Incidence: About 1 in 20,000.
Inheritance: X-linked.
Cause: Pathogenic ABCD1 mutations.
Clinical Sensitivity: 96 percent.
Methodology: Bidirectional sequencing of all coding regions and intron-exon boundaries of the ABCD1 gene; primers are specifically selected to target the functional ABCD1 gene. Multiplex Ligation-dependent Probe Amplification (MLPA) to detect large ABCD1 deletions/duplications.
Analytical Sensitivity and Specificity: 99 percent.
Limitations: Exons 7-10 are not evaluated by deletion/duplication analysis. Diagnostic errors can occur due to rare sequence variations. Regulatory region mutations and deep intronic mutations will not be detected. The breakpoints of large deletions/duplications will not be determined. Mutations in genes other than ABCD1 will not be detected.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
ABCD1 Seq, Del Dup Specimen	20-045-401904	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
ABCD1 Seq, Del/Dup Interp	20-045-401904	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical