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

Physician: Doctor, Example

Patient: Patient, Example

DOB	Unknown
Gender:	Unknown
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

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

ARUP test code 2011906

ABCD1 Seq, Del Dup Specimen	Whole Blood	
ABCD1 Seq, Del/Dup Interp	**Positive**	*

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

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.
TEST PERFORMED - 2011906
TEST DESCRIPTION - Adrenoleukodystrophy, X-Linked (ABCD1) Sequencing and Deletion/Duplication
INDICATION FOR TEST - Confirm Diagnosis

RESULT
One pathogenic variant was detected in the ABCD1 gene.

DNA VARIANT
Classification: Pathogenic
Gene: ABCD1
Nucleic Acid Change: c.1621_1628delTACATCCC; Hemizygous
Amino Acid Alteration: p.Tyr541AlafsTer12

INTERPRETATION
One pathogenic variant, c.1621_1628delTACATCCC; p.Tyr541AlafsTer12, was detected in the ABCD1 gene by sequencing. This result is consistent with a diagnosis of X-linked adrenoleukodystrophy in this individual. Disease phenotype is variable and age dependent. All of this individual's daughters will be carriers, but none of his sons will inherit the pathogenic variant. No pathogenic variants were detected by deletion/duplication analysis.

Evidence for variant classification: The ABCD1 c.1621_1628delTACATCCC; p.Tyr541AlafsTer12 variant, to our knowledge, is not reported in the medical literature or gene specific databases. This variant is also absent from general population databases (1000 Genomes Project, Exome Variant Server, and Genome Aggregation Database), indicating it is not a common polymorphism. This variant causes a frameshift by deleting 8 nucleotides, so it is predicted to result in a truncated protein or mRNA subject to nonsense-mediated decay. Based on available information, this variant is considered to be pathogenic.

RECOMMENDATIONS
Genetic consultation is indicated, including a discussion of medical screening and management. At-risk family members should be offered testing for the identified variant (Familial Mutation, Targeted Sequencing, ARUP test code 2001961).

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 included in this report.

This result has been reviewed and approved by [redacted]

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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	20-329-111189	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
ABCD1 Seq, Del/Dup Interp	20-329-111189	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