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

Physician: Doctor, Example

Patient: Patient, Example

DOB	6/17/1945
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Familial Transthyretin Amyloidosis (TTR) Sequencing

ARUP test code 2014035

TTR FGS Spec	Whole Blood
TTR FGS Interpretation	Negative

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TEST PERFORMED - 2014035
TEST DESCRIPTION - Familial Transthyretin Amyloidosis (TTR)
Sequencing
INDICATION FOR TEST - Not Provided

RESULT
No pathogenic variants were detected in the TTR gene.

INTERPRETATION
No pathogenic variants were detected in the TTR gene by
sequencing all coding regions and intron-exon boundaries. This
result significantly decreases the probability of, but does not
exclude, a diagnosis of Familial Transthyretin Amyloidosis.
Please refer to the background information included in this
report for the clinical sensitivity and limitations of this test.

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

COMMENTS
Reference Sequence: GenBank # NM_000371.3 (TTR)
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 Steven Steinberg,
Ph.D.
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H=High, L=Low, *=Abnormal, C=Critical

BACKGROUND INFORMATION: Familial Transthyretin Amyloidosis (TTR) Sequencing
CHARACTERISTICS: Familial Transthyretin Amyloidosis is caused by pathogenic variants of the TTR gene resulting in abnormal amyloid accumulation in various tissues and is generally categorized into three phenotypes: 1) familial amyloid polyneuropathy, a slowly progressive sensorimotor and autonomic neuropathy; 2) familial amyloid cardiomyopathy, a restrictive cardiomyopathy with cardiomegaly, conduction block, angina, congestive heart failure and aortic dissection/dilatation; and 3) leptomeningeal amyloidosis, primarily affecting the CNS, causing dementia, visual impairment, seizures, ataxia, psychosis, hemorrhage, and hydrocephalus. TTR variants can also be associated with benign familial euthyroid hyperthyroxinemia.
INCIDENCE: 1 in 568 individuals from Northern Portugal; 1 in 100,000 individuals of Northern European ancestry.
INHERITANCE: Autosomal dominant.
PENETRANCE: Incomplete.
CAUSE: Pathogenic TTR gene variants.
CLINICAL SENSITIVITY: 99 percent for Familial TTR Amyloidosis.
METHODOLOGY: Bidirectional sequencing of all coding regions and intron-exon boundaries of the TTR gene.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Regulatory region variants, deep intronic variants and large deletions/duplications in TTR 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
TTR FGS Spec	19-173-400529	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
TTR FGS Interpretation	19-173-400529	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