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

Physician: Doctor, Example

Patient: Patient, Example

DOB	9/3/1974
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Cystic Fibrosis (CFTR) Sequencing (Temporary Referral as of 12/07/20)

ARUP test code 0051110

CF CFTR Specimen	Whole Blood
Cystic Fibrosis (CFTR) Sequencing	Negative

TEST PERFORMED - 0051110
TEST DESCRIPTION - Cystic Fibrosis (CFTR) Sequencing
INDICATION FOR TEST - Confirm Diagnosis

RESULT
No pathogenic variants were detected in the CFTR gene.

INTERPRETATION
No pathogenic variants were detected in the CFTR gene using bidirectional sequencing of the coding regions and intron-exon boundaries. Sequencing detects 97 percent of CFTR variants, thus, the risk for cystic fibrosis (CF) or CF carrier status is reduced.

RECOMMENDATIONS
Medical management should rely on clinical findings and family history. According to information provided to ARUP, this individual has pancreatitis. Variants in the CTRC, SPINK1, and PRSS1 genes are also associated with hereditary pancreatitis. If strong clinical concern for hereditary pancreatitis remains, sequencing of the SPINK1 and CTRC genes, and full gene analysis of the PRSS1 gene (ARUP test codes 2002012, 2010703, and 3001768, respectively should be considered. Genetic consultation is indicated.

COMMENTS
Reference Sequence: GenBank # NM_000492.3 (CFTR)
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: Cystic Fibrosis (CFTR) Sequencing

CHARACTERISTICS: Chronic sino-pulmonary disease, gastrointestinal malabsorption/pancreatic insufficiency, and obstructive azoospermia. Findings are often limited to a single organ system such as isolated pancreatitis, bilateral absence of the vas deferens, nasal polyposis, or bronchiectasis in non-classic cystic fibrosis (CF).
INCIDENCE OF CLASSIC CF: 1 in 3,000 Caucasians or Ashkenazi Jewish, 1 in 8,000 Hispanics, 1 in 15,000 African Americans, 1 in 32,000 Asians.
INCIDENCE OF NONCLASSIC CF: Unknown.
INHERITANCE: Autosomal recessive.
PENETRANCE: High for severe mutations, variable for mild/moderate mutations.
CAUSE OF CLASSIC CF: Two deleterious CFTR mutations on opposite chromosomes.
CAUSE OF NONCLASSIC CF: Typically one severe and one mild/moderate CFTR mutations on opposite chromosomes.
CLINICAL SENSITIVITY: 97 percent.
METHODOLOGY: Bidirectional sequencing of the entire CFTR coding region, intron-exon boundaries, and two deep intronic mutations.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Regulatory region mutations, large gene deletions/duplications and some deep intronic mutations 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
CF CFTR Specimen	20-318-401249	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Cystic Fibrosis (CFTR) Sequencing	20-318-401249	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