ARUP LABORATORIES
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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	2/20/1989
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Alpha-1-Antitrypsin (SERPINA1) Enzyme Concentration and 2 Mutations with Reflex to Alpha-1-Antitrypsin Phenotype

ARUP test code 0051256

Alpha-1-Antitrypsin	82 mg/dL L (Ref Interval: 90-200) To convert to umol/L, multiply mg/dL by 0.185
Alpha-1-Antitrypsin Genotype Specimen	Whole Blood
Alpha-1-Antitrypsin S Allele	Negative
Alpha-1-Antitrypsin Z Allele	Heterozygous *
Alpha-1-Antitrypsin Interpretation	See Note Indication for testing: Carrier screening or diagnostic testing for alpha-1-antitrypsin (AAT) deficiency. Z Heterozygote/ Protein concentration <90 mg/dL: This sample has a reduced serum AAT protein concentration and one copy of the Z deficiency allele was detected by genotyping. The S deficiency allele was not identified. AAT phenotyping was performed by isoelectric focusing electrophoresis to rule out the possibility of a concurrent rare deficiency allele not detected by the genotyping assay. Phenotyping demonstrated the Pi MZ phenotype; the M allele is associated with normal serum concentrations of AAT while the Z allele produces a deficiency variant. In combination, these findings are most consistent with carrier status for AAT deficiency. Nonsmokers do not appear to be at increased risk for lung disease. Smokers with this phenotype have an increased rate of loss of lung elasticity but rarely develop clinical disease. Caution in interpretation is advised if the patient has been transfused in the previous 21 days. This individual's reproductive partner and other family members should be offered AAT testing. Genetic consultation is recommended. This result has been reviewed and approved by [redacted]

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

	BACKGROUND INFORMATION: A1A (SERPINA1) Enzyme Concentration and 2 Mutations with Reflex to A1A Phenotype CHARACTERISTICS of Alpha-1-Antitrypsin (AAT) Deficiency: Coughing, wheezing, bronchiectasis, chronic obstructive pulmonary disease, emphysema, and cirrhosis. INCIDENCE: 1 in 3000 to 5000 North American individuals. INHERITANCE: Autosomal recessive. CAUSE: Two pathogenic mutations in the SERPINA1 gene on opposite chromosomes. CLINICAL SENSITIVITY: 95 percent. MUTATIONS TESTED: S allele (c.791A>T) and Z allele (c.1024G>A). METHODS: Genotyping performed by polymerase chain reaction (PCR) and fluorescence monitoring; AAT protein concentration measured using immunoturbidmetric assay; phenotyping performed by isoelectric focusing electrophoresis. Genotyping and AAT serum protein concentration determination are performed on all specimens. Protein phenotyping is only performed on specimens that have AAT protein concentrations of less than 90 mg/dL and are not homozygous or compound heterozygous for the S or Z deficiency alleles by genotyping. ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent. LIMITATIONS: SERPINA1 mutations, other than the S (c.791A>T) and Z (c.1024G>A) alleles, will not be detected. Diagnostic errors can occur due to rare sequence variations. This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes. Counseling and informed consent are recommended for genetic testing. Consent forms are available online.
Alpha-1-Antitrypsin Phenotype	M1Z

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Alpha-1-Antitrypsin	24-052-122238	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha-1-Antitrypsin Genotype Specimen	24-052-122238	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha-1-Antitrypsin S Allele	24-052-122238	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha-1-Antitrypsin Z Allele	24-052-122238	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha-1-Antitrypsin Interpretation	24-052-122238	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha-1-Antitrypsin Phenotype	24-052-122238	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

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