ARUP LABORATORIES
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Tracy I. George, MD, Chief Medical Officer

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	[redacted]
Gender:	Male
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	30 mg/dL L To convert to umol/L, multiply mg/dL by 0.185	(Ref Interval: 90-200)
Alpha-1-Antitrypsin Genotype Specimen	Whole Blood	
Alpha-1-Antitrypsin S Allele	Negative	
Alpha-1-Antitrypsin Z Allele	Homozygous *	
Alpha-1-Antitrypsin Interpretation	See Note Indication for testing: Carrier screening or diagnostic testing for alpha-1-antitrypsin (AAT) deficiency. Z Homozygote/ Protein concentration <90 mg/dL: This sample has a greatly reduced serum AAT protein concentration and two copies of the Z deficiency allele were detected by genotyping. The S mild deficiency allele was not identified. This individual is predicted to be affected with classic AAT deficiency and has an increased risk of neonatal jaundice, adult cirrhosis, and early emphysema. 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] M.D., Ph.D.	

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 PCR followed by fluorescent probe melting analysis; 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	Not Applicable

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Alpha-1-Antitrypsin	21-168-107400	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha-1-Antitrypsin Genotype Specimen	21-168-107400	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha-1-Antitrypsin S Allele	21-168-107400	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha-1-Antitrypsin Z Allele	21-168-107400	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha-1-Antitrypsin Interpretation	21-168-107400	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha-1-Antitrypsin Phenotype	21-168-107400	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

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