

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: ██████████
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 101 mg/dL (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 **Heterozygous** *

Alpha-1-Antitrypsin Z Allele **Heterozygous** *

Alpha-1-Antitrypsin Interpretation

See Note

SZ Compound Heterozygote/Protein concentration >90 mg/dL: This sample has a serum AAT protein concentration in the normal range (101 mg/dl). One copy of both the S and Z deficiency alleles were detected by genotyping. Twenty to fifty percent of individuals with the SZ genotype develop emphysema. The risk for pulmonary complications is higher in SZ compound heterozygotes who are smokers or who are exposed to occupational or environmental pollutants. Liver disorders may occur at an increased frequency in adults. However, this patient has a protein concentration in the normal range; serum AAT is an acute phase protein and subject to variation. Given the SZ genotype and normal AAT at present, the risk for clinical manifestations in this patient is unclear. This individuals reproductive partner and family members should be offered AAT testing. Genetic consultation is recommended.

This result has been reviewed and approved by ██████████, M.D.

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

Unless otherwise indicated, testing performed at:

**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 immunoturbidimetric 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	21-169-135900	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha-1-Antitrypsin Genotype Specimen	21-169-135900	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha-1-Antitrypsin S Allele	21-169-135900	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha-1-Antitrypsin Z Allele	21-169-135900	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha-1-Antitrypsin Interpretation	21-169-135900	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha-1-Antitrypsin Phenotype	21-169-135900	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

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Tracy I. George, MD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 21-169-135900
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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