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

ARUP test code 0051256

<table>
<thead>
<tr>
<th>Test</th>
<th>Result</th>
<th>Reference Interval</th>
</tr>
</thead>
<tbody>
<tr>
<td>Alpha-1-Antitrypsin</td>
<td>124 mg/dL</td>
<td>90-200 (Ref Interval)</td>
</tr>
</tbody>
</table>

To convert to umol/L, multiply mg/dL by 0.185

<table>
<thead>
<tr>
<th>Genotype Specimen</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Whole Blood</td>
<td></td>
</tr>
</tbody>
</table>

**Alpha-1-Antitrypsin S Allele**

- Heterozygous

**Alpha-1-Antitrypsin Z Allele**

- Negative

**Alpha-1-Antitrypsin Interpretation**

- See Note

Indication for testing: Carrier screening or diagnostic testing for alpha-1-antitrypsin (AAT) deficiency.

S Heterozygote/ Protein concentration >=90 mg/dL: This sample has a serum AAT protein concentration in the normal range, but one copy of the S mild deficiency allele was detected by genotyping. The Z deficiency allele was not identified. In combination, these results predict this individual is a carrier of AAT deficiency. Individuals heterozygous for the S deficiency allele are not at risk for development of hepatic or pulmonary disease related to AAT deficiency. Because this genotyping assay cannot exclude the presence of a concurrent rare deficiency allele, AAT phenotyping by isoelectric focusing electrophoresis is recommended if the patient is in acute phase. 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 Rong Mao, M.D.
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.

Counseling and informed consent are recommended for genetic
testing. Consent forms are available online at www.aruplab.com.

See Compliance Statement C: www.aruplab.com/CS

Alpha-1-Antitrypsin Phenotype Not Applicable

<table>
<thead>
<tr>
<th>Procedure</th>
<th>Accession</th>
<th>Collected</th>
<th>Received</th>
<th>Verified/Reported</th>
</tr>
</thead>
<tbody>
<tr>
<td>Alpha-1-Antitrypsin</td>
<td>18-222-129955</td>
<td>8/10/2018 10:07:00 AM</td>
<td>8/12/2018 11:59:56 PM</td>
<td>8/12/2018 3:49:00 PM</td>
</tr>
<tr>
<td>Alpha-1-Antitrypsin Genotype Specimen</td>
<td>18-222-129955</td>
<td>8/10/2018 10:07:00 AM</td>
<td>8/12/2018 11:59:56 PM</td>
<td>8/13/2018 7:18:00 PM</td>
</tr>
<tr>
<td>Alpha-1-Antitrypsin S Allele</td>
<td>18-222-129955</td>
<td>8/10/2018 10:07:00 AM</td>
<td>8/12/2018 11:59:56 PM</td>
<td>8/13/2018 7:18:00 PM</td>
</tr>
<tr>
<td>Alpha-1-Antitrypsin Z Allele</td>
<td>18-222-129955</td>
<td>8/10/2018 10:07:00 AM</td>
<td>8/12/2018 11:59:56 PM</td>
<td>8/13/2018 7:18:00 PM</td>
</tr>
<tr>
<td>Alpha-1-Antitrypsin Interpretation</td>
<td>18-222-129955</td>
<td>8/10/2018 10:07:00 AM</td>
<td>8/12/2018 11:59:56 PM</td>
<td>8/13/2018 7:18:00 PM</td>
</tr>
<tr>
<td>Alpha-1-Antitrypsin Phenotype</td>
<td>18-222-129955</td>
<td>8/10/2018 10:07:00 AM</td>
<td>8/12/2018 11:59:56 PM</td>
<td>8/12/2018 3:49:00 PM</td>
</tr>
</tbody>
</table>

END OF CHART