## Lab Dept: Chemistry

### Test Name: ALPHA-1-ANTITRYPsin PHENOTYPE

#### General Information

**Lab Order Codes:**

A1AP

**Synonyms:**

A1A Phenotype, serum; AAT Phenotype, serum; Pi Typing; Protease Inhibitor Allo Typing

**CPT Codes:**

82103 - Alpha-1-antitrypsin; total

82104 - Alpha-1-antitrypsin; phenotype

**Test Includes:**

Alpha-1-Antitrypsin, serum reported in mg/dL and phenotype.

**NOTE:** DO NOT order with Alpha-1-Antitrypsin Proteotype Deficiency Panel. For more information on this testing, see Mayo's Testing Algorithm

#### Logistics

**Test Indications:**

Useful for identification of homozygous and heterozygous phenotypes of alpha-1-antitrypsin deficiency.

**Lab Testing Sections:**

Chemistry - Sendouts

**Referred to:**

Mayo Medical Laboratories (MML Test: A1APP)

**Phone Numbers:**

MIN Lab: 612-813-6280

STP Lab: 651-220-6550

**Test Availability:**

Daily, 24 hours

**Turnaround Time:**

2 - 6 days, performed Monday - Friday

**Special Instructions:**

N/A

#### Specimen

**Specimen Type:**

Blood

**Container:**

SST (Marble, gold or red)

**Draw Volume:**

4 mL (Minimum: 1.5 mL) blood

**Processed Volume:**

1.25 mL (Minimum: 0.5 mL) serum
<table>
<thead>
<tr>
<th><strong>Collection:</strong></th>
<th>Routine venipuncture</th>
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</thead>
<tbody>
<tr>
<td><strong>Special Processing:</strong></td>
<td>Lab Staff: Centrifuge specimen, remove serum aliquot into a screw-capped round bottom plastic vial. Store and ship at refrigerated temperatures. Forward promptly.</td>
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<tr>
<td><strong>Patient Preparation:</strong></td>
<td>None</td>
</tr>
<tr>
<td><strong>Sample Rejection:</strong></td>
<td>Specimens other than serum; gross lipemia; mislabeled or unlabeled specimens</td>
</tr>
</tbody>
</table>

### Interpretive

**Reference Range:**  
**Alpha-1-Antitrypsin, Serum:** 100 - 190 mg/dL

**Alpha-1-Antitrypsin Phenotyping:** The interpretive report will identify the alleles present. For rare alleles, the report will indicate whether or not they have been associated with reduced quantitative levels of alpha-1-antitrypsin.

Most normal individuals have the M phenotype (M, M1 or M2). Over 99% of M phenotypes are genotypically MM. In the absence of family studies, the phenotype (M) and quantitative level can be used to infer the genotype (MM). The most common alleles associated with a quantitative deficiency are Z and S. The reports for the rare alleles will indicate whether or not they have been associated with reduced quantitative levels.

Interpretation: There are >40 Alpha-1-antitrypsin (A1A) phenotypes (most of these are associated with normal quantitative levels of protein). The most common normal phenotype is M (M, M1 or M2), and >90% of Caucasians are homozygous M (MM) genotype.

A1A deficiency is usually associated with the Z phenotype (ZZ genotype), but genotypes such as SS and SZ are also associated with decreased A1A levels.

**Critical Values:** N/A

**Limitations:**  
This assay identifies the phenotype of the circulating alpha-1-antitrypsin (A1A) protein. If the patient is already on replacement therapy, the phenotype will detect patient and replacement protein.

If two bands are seen, such as an M band and a Z band, it is reported as MZ (eg, heterozygous).

If one band is seen, such as the Z band and the quantitative level is consistent with a homozygote, the phenotype is assumed to be homozygous and is reported as ZZ.

**Methodology:** Isoelectric Focusing and Nephelometry
References:  

Mayo Medical Laboratories Web Page (August 2016)