**Lab Dept:** Chemistry

**Test Name:** ALPHA-1-ANTITRYPsin PHEnOTYPE

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### 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

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### 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

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### 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
Collection: Routine venipuncture

Special Processing: Lab Staff: Centrifuge specimen, remove serum aliquot into a screw-capped round bottom plastic vial. Store and ship at refrigerated temperatures. Forward promptly.

Specimen stable refrigerated (preferred), ambient or frozen for 28 days.

Patient Preparation: None

Sample Rejection: Specimens other than serum; gross lipemia; mislabeled or unlabeled specimens

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 Clinic Laboratories (June 2023)

Updates: 06/2023: Testing resumed at reference lab, added specimen stability.