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

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.