
Lab Dept: Chemistry

Test Name: ALPHA-1-ANTITRYPSIN DEFICIENCY PROFILE

General Information

Lab Order Codes: AATP

Synonyms: A1A Genotype, serum; AAT Genotype, serum; Alpha-1-Antitrypsin Proteotype S/Z

CPT Codes: 82103 – Alpha-1-antitrypsin; total
82542 – Column chromatography/Mass spectrophotometry, stable isotope dilution, multiple analytes, quantitative, single stationary and mobile phase
82104 – Alpha-1-antitrypsin; phenotype (if appropriate)

Test Includes: Alpha-1-Antitrypsin Proteotype S/Z, serum alpha-1-antitrypsin and phenotype if appropriate. An interpretive report will be provided.
Note: DO NOT order this test and a separate Phenotype test.
See [Mayo's A1AT Testing algorithm](#) for more information.

Logistics

Test Indications: Useful for confirmation of clinical diagnosis of Alpha-1-Antitrypsin deficiency and determination of the specific allelic variant. Genotyping also provides some insight as to the possible course of disease.

Lab Testing Sections: Chemistry - Sendouts

Referred to: Mayo Medical Laboratories (MML Test: A1ALC)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 5 - 7 days, performed Monday and Wednesday

Special Instructions: N/A

Specimen

Specimen Type: Blood

Container: SST (Marble, gold or red)

Draw Volume:	4 mL (Minimum: 0.6 mL) blood
Processed Volume:	1.25 mL (Minimum: 0.2 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.
Patient Preparation:	None
Sample Rejection:	Mislabeled or unlabeled specimens; lipemic specimens

Interpretive

Reference Range: **Alpha-1-Antitrypsin Proteotype:** Negative for S and Z phenotype (Non S, Non Z)

Alpha-1-Antitrypsin: 100 – 190 mg/dL

Alpha-1-Antitrypsin Phenotyping (when indicated): 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.

Critical Values: N/A

Limitations: This assay will not detect 5% of the known mutations that cause alpha-1-antitrypsin (A1A) deficiency. Therefore, the absence of a detectable mutation does not rule out the possibility that an individual is a carrier of, or affected with, this disease.

Test results should be interpreted in the context of Alpha-1-Antitrypsin quantitation, clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information provided to us is inaccurate or incomplete.

Rare mutations exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Errors in interpretation may occur if patients have had a recent blood transfusion or are on A1A replacement therapy.

Methodology: Liquid Chromatography- Tandem Mass Spectrophotometry (LC-MS/MS) Nephelometry, and Isoelectric Focusing (when indicated)

References:

[Mayo Medical Laboratories Web Page](#) (August 2016)

Updates:

3/1/2007: Test expanded to become a genotype profile. Previously listed as a Phenotype test. Phenotyping will now only occur if necessary for a complete evaluation.

3/1/2011: EDTA draw level previously listed as 3 mL.

10/8/2013: CPT update 2013.

8/2/15: Addition of Proteotype S/Z, CPT update.

8/2/16: Updated volume, CPT update, method update