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: 7 - 14 days, performed Monday and Thursday

Special Instructions: N/A

Specimen

Specimen Type: Blood

Container: SST (Marble, gold or red) tube
**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.

**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 December 2017

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.
8/2/2016: Updated volume, CPT update, method update