MAYO CLINIC LABORATORIES

Test Definition: A1APP

Alpha-1-Antitrypsin Phenotype, Serum

Overview

Useful For

Identification of homozygous and heterozygous phenotypes of the alpha-1-antitrypsin deficiency

Profile Information

Test Id	Reporting Name	Available Separately	Always Performed
A1AP2	Alpha-1-Antitrypsin Phenotype	No	Yes
AATP	Alpha-1-Antitrypsin, S	Yes, (Order AAT)	Yes

Testing Algorithm

See Alpha-1-Antitrypsin-A Comprehensive Testing Algorithm in Special Instructions.

Special Instructions

- Alpha 1 Antitrypsin-A Comprehensive Testing Algorithm

Method Name

A1AP2: Isoelectric Focusing

AATP: Nephelometry

NY State Available

Yes

Specimen

Specimen Type

Serum

Specimen Required

Collection Container/Tube:

Preferred: Red top

Acceptable: Serum gel

Submission Container/Tube: Plastic vial

Specimen Volume: 1.25 mL

Collection Instructions: Centrifuge and aliquot serum into a plastic vial.

Forms

If not ordering electronically, complete, print, and send 1 of the following with the specimen:

-Gastroenterology and Hepatology Test Request (T728)

-General Request (T239)

Specimen Minimum Volume

0.5 mL

Reject Due To

Gross hemolysis	OK
Gross lipemia	Reject
Gross icterus	OK

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Serum	Ambient	28 days	
	Refrigerated (preferred)	28 days	
	Frozen	28 days	

Clinical & Interpretive

Clinical Information

Alpha-1-antitrypsin (A1A) is the most abundant serum protease inhibitor and inhibits trypsin and elastin, as well as several other proteases. The release of proteolytic enzymes from plasma onto organ surfaces and into tissue spaces results in tissue damage unless inhibitors are present. Congenital deficiency of A1A is associated with the development of emphysema at an unusually early age and with an increased incidence of neonatal hepatitis, usually progressing to cirrhosis.

Most normal individuals have the M phenotype (M, M1, or M2). Over 99% of M phenotypes are genetically 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.

See Alpha-1-Antitrypsin-A Comprehensive Testing Algorithm in Special Instructions.

Reference Values

ALPHA-1-ANTITRYPSIN

100-190 mg/dL

ALPHA-1-ANTITRYPSIN PHENOTYPE

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.

Interpretation

There are greater than 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 greater than 90% of individuals of European descent are genetically homozygous M (MM).

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

Cautions

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

If 1 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.

Clinical Reference

1. Morse JO: Alpha-1-antitrypsin deficiency. N Engl J Med 1978;299:1045-1048;1099-1105
2. Donato LJ, Jenkins SM, Smith C, et al: Reference and interpretive ranges for alpha(1)-antitrypsin quantitation by phenotype in adult and pediatric populations. Am J Clin Pathol 2012 Sep;138(3):398-405

Performance

Method Description

Phenotyping is done by isoelectric focusing in agarose gels.(Package insert: Hydragel 18 A1AT Isofocusing. Sebia. 2013 July)

Nephelometry.(Instruction manual: Siemens Nephelometer II. Siemens, Inc., Newark, DE)

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available

2 to 6 days

Performing Laboratory Location

Rochester

Fees & Codes

Fees

- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

Test Classification

This test has been cleared, approved, or is exempt by the US Food and Drug Administration and is used per manufacturer's instructions. Performance characteristics were verified by Mayo Clinic in a manner consistent with CLIA requirements.

CPT Code Information

82103
82104

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
A1APP	Alpha-1-Antitrypsin Phenotype	32769-2

Result ID	Test Result Name	Result LOINC® Value
AATP	Alpha-1-Antitrypsin, S	6771-0
8166	Alpha-1-Antitrypsin Phenotype	32769-2