Overview

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

Profile Information

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Reporting Name</th>
<th>Available Separately</th>
<th>Always Performed</th>
</tr>
</thead>
<tbody>
<tr>
<td>A1AP2</td>
<td>Alpha-1-Antitrypsin Phenotype</td>
<td>No</td>
<td>Yes</td>
</tr>
<tr>
<td>AATP</td>
<td>Alpha-1-Antitrypsin, S</td>
<td>Yes, (Order AAT)</td>
<td>Yes</td>
</tr>
</tbody>
</table>

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
Container/Tube:
Preferred: Red top
Acceptable: Serum gel

Specimen Volume: 1.25 mL

Forms
If not ordering electronically, complete, print, and send 1 of the following forms with the specimen:
- [General Request](#) (T239)
- [Gastroenterology and Hepatology Client Test Request](#) (T728)
Test Definition: A1APP
Alpha-1-Antitrypsin Phenotype

Specimen Minimum Volume
0.5 mL

Reject Due To

<table>
<thead>
<tr>
<th>Specimen Condition</th>
<th>Acceptance</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gross hemolysis</td>
<td>OK</td>
</tr>
<tr>
<td>Gross lipemia</td>
<td>Reject</td>
</tr>
<tr>
<td>Gross icterus</td>
<td>OK</td>
</tr>
</tbody>
</table>

Specimen Stability Information

<table>
<thead>
<tr>
<th>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
<th>Special Container</th>
</tr>
</thead>
<tbody>
<tr>
<td>Serum</td>
<td>Refrigerated (preferred)</td>
<td>28 days</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Ambient</td>
<td>28 days</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Frozen</td>
<td>28 days</td>
<td></td>
</tr>
</tbody>
</table>

Clinical and 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 Caucasians 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**


**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) and Time(s) Test Performed**

Monday through Friday; 9 a.m.

**Analytic Time**

2 days

**Maximum Laboratory Time**

6 days

**Performing Laboratory Location**

Rochester

**Fees and 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 Regional Manager. For assistance, contact Customer Service.
Test Definition: A1APP
Alpha-1-Antitrypsin Phenotype

Test Classification
This test has been cleared, approved or is exempt by the U.S. 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

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Test Order Name</th>
<th>Order LOINC Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>A1APP</td>
<td>Alpha-1-Antitrypsin Phenotype</td>
<td>32769-2</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Result ID</th>
<th>Test Result Name</th>
<th>Result LOINC Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>AATP</td>
<td>Alpha-1-Antitrypsin, S</td>
<td>6771-0</td>
</tr>
<tr>
<td>8166</td>
<td>Alpha-1-Antitrypsin Phenotype</td>
<td>32769-2</td>
</tr>
</tbody>
</table>