ALPHA 1-ANTITRYPSIN PHENOTYPE, SERUM

London Health Sciences Centre
St JOSEPH'S HEALTH CARE LONDON
Pathology and Laboratory Medicine

Orderable - A1ATP

Turnaround Time: 2 weeks

Laboratory:
Core Lab

Requisition:
GENERAL LABORATORY REQUISITION

Method of Analysis:
Isoelectric focusing

Test Schedule:
Referred out Monday-Thursday

Referred Out Location:
In-Common Laboratories

Alternate Name(s):

Alpha-1 Antitrypsin Phenotype
Alpha-1-Antitrypsin Phenotype
Alpha1 Antitrypsin Phenotype
Alpha 1 Antitrypsin Phenotype
A1AT Phenotyping

Specimen:

Adult	Pediatric
5 mL Gold top Vacutainer tube	0-2 years: 2 x 0.5 mL Red or Gold top Microtainer 2-10 years: 3 mL Red Vacutainer top

Collection Information:

The preferred volume of serum is 1 mL, but the minimum volume of serum is 0.1 mL.

Reference Ranges:

Refer to interpretive comments below.

Interpretive Comments:

90% of normal individuals have the MM phenotype, with normal quantitative AAT levels. Many phenotypic patterns have been described, including deficiency states with F, S, Z, or other alleles. As a general estimation, compared to M allele of 100% of normal A-1-Antitrypsin protein, the S allele produces approximately 60% and the Z allele 20%. For example, an MS phenotype would have about 80% of normal A-1-Antitrypsin protein level, a 50% contribution from the M allele and 30% from the S allele. A ZZ phenotype would have about 20% of normal levels, a 10% contribution from each z gene. The F allele has normal A-1-Antitrypsin levels, but the kinetics of elastase inhibition is not as efficient as an M allele product; F alleles should be considered fuctionally mildly deficient. Other variants are identifiable by phenotypic

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analysis. These include CM, DP, EM GM, IS, LM, M1M2, M3M3, MP, MT, XX, MY, and M1N. I, P, T and null alleles are considered deleterious. C, D, E, G, L, M1, M2, M3, X and Y alleles are generally considered normal variants. The MZ-Pratt phenotype is a normal variant; care should be taken to avoid confusion with the deficient MZ phenotype.

Comments:

This test is available exclusively to LHSC/SJHC physicians.

All specimens submitted for A1AT phenotyping will have a serum A1AT level measured at LHSC as a screening test. Those specimens with an A1AT result of ≤1.50 g/L will be referred out for phenotyping automatically as long as phenotyping has been ordered and phenotyping/proteotyping has not been done in the past. If the A1AT level is >1.50 g/L, the sample will not be referred out for phenotyping without biochemist approval.

Storage and Shipment:

Store and ship serum frozen.