Alpha-1 Antitrypsin Phenotyping

Synonyms	AATP
Clinical Indication	Further investigation of low serum alpha-1 antitrypsin levels. Alpha-1 antitrypsin levels are measured and samples may be referred for phenotyping if the concentration is below the age related reference range or borderline low (At the discretion of the Clinical Biochemist). If low alpha-1 antitrypsin has been picked up as an incidental finding during protein electrophoresis testing then a comment will be added to the report asking for the requesting clinician to discuss with the patient before phenotyping is carried out. Patients with borderline low alpha-1 antitrypsin are probably carriers and may not require further investigation
Part of Profile / See Also	
Request Form	Combined Pathology manual Blood form or ICE request
Availability / Frequency of Analysis	Analysed by Protein Reference Unit, St George's Hospital (9745), if specific criteria met.
Turnaround Time	1 month
Patient Preparation	None required
Sample Requirements	
Specimen Type	Serum
Volume	2 ml
Container	 Yellow top (SST) tube Or  Paediatric lithium-heparin (orange top – Sarstedt tube)
Reference Range & Units	The report will identify the alleles present. The most common normal phenotype is M with most Caucasians being homozygous MM. Alpha-1-antitrypsin deficiency is usually associated with the Z phenotype (homozygous ZZ) but SS and SZ are also associated with decreased alpha-1-antitrypsin levels.
Interferences	
Interpretation & Clinical Decision Value (if applicable)	A sample for genotyping (5mL pink-top EDTA) may be requested as a follow up confirmation for deficiency alleles identified by phenotyping. A full interpretive comment is provided by the referral laboratory along with a recommendation for family studies if indicated.

References

Test code	AATP
Lab Handling	Aliquot 500ul and store in referrals rack at 4C. Sent daily by courier to St George's Hospital, London.

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