

## The Efficiency of Alpha1-antitrypsin Deficiency Detection by Isoelectric Focusing Phenotypes in Relation to Serum Protein Concentrations in COPD Patients

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Alpha 1- antitrypsin (AAT) deficiency is one of the most hereditary disorders in the world, but there is misdiagnosis for this deficiency. There are about 100 different AAT variants cannot be easily detected by the known methods such as genotyping which reveals only the most common deficiency alleles Z and S. These underscore the need of phenotyping to determine both common and rare deficiency variants. The traditional method used in hospitals for diagnosis is the quantification of serum AAT level. Our work is an attempt to evaluate serum AAT concentration in comparison to phenotyping. 300 Chronic obstructive pulmonary disease (COPD) patients were subjected to the determination of Pi protein phenotypes by isoelectric focusing electrophoresis (IEF) and serum AAT concentrations using nephelometry. The patients were classified according to their AAT levels into 3 groups, less than 89 mg/dl (deficiency, group1), 90-140 mg/dl (intermediate, group2) and more than 141 mg/dl (normal, group3). Phenotypes of the deficient group were MZ, ZZ, SZ and SS, while the majority of phenotypes in the intermediate and normal groups were MM with the presence of some at risk variants MS, MZ, SS. Where, the rare deficiency variants FS and FM phenotypes were observed in the intermediate group. The presence of some deficiency alleles in the intermediate and normal groups which have relatively high AAT levels reflects that serum AAT concentration was not sufficient to be used for the detection of AAT deficiency indicating the importance of using other methods such as phenotyping.

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**Keywords:** Isoelectric focusing; AAT; COPD; Phenotype

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