Summary and Conclusion
SSCP analysis of the 360bp PCR amplicons of Exon III for PiM1 variant showed the same mobility pattern for all samples except one. This sample (COPD Patient No. 176) had a sequence variation in the Exon III 360bp region.

These 360bp PCR products were further digested with BstE II enzyme to identify the normal PiM1 variant. All the samples digested were cleaved into 228, 83 and 49bp products indicating the PiM1 Val213 allele alone was present in the study population. No 311bp bands were seen in the reaction conditions, indicating the absence of PiM1 Ala213 allele among the COPD patients and normal controls. COPD patient sample No: 176 that showed a mobility shift on SSCP analysis, was also cleaved into 228, 83 and 49bp products, showing that the mobility shift was due to some other sequence change and not from PiM1 Val213 to PiM1 Ala213.

DNA Sequencing was carried out for the SSCP variant (sample no.176) and two controls (sample no.85 and 152). In the sense strand of the SSCP variant at the 7489\(^{th}\) base sequence ‘Guanine’ was substituted by ‘Adenine’. The codon change was from AAG to AAA - Lys201Lys (both coding for lysine). Hence a silent mutation was observed. Such silent mutations or polymorphism are predicted to be evolutionary intermediates between \textit{aat} PiM1 Ala213 and PiM1 Val213alleles.

Absence of severe \textit{aat} deficient alleles in south Indian population indicates that \(\alpha1\)-AT deficiency due to PiS and PiZ occur mainly among European populations of Caucasian descent.
The PiM1 allele present in the present study was found to be solely PiM1 Val213 allele. Absence of PiM1 Ala213 allele among the Asian populations accounts for the extreme rarity of the PiZ gene in those populations as the PiZ allele has developed on the PiM1 Ala213 base allele. This further account for the absence of Z variants in the COPD samples screened in the present study.

The present work indicates that COPD in the patients screened was not due to \textit{aat} gene deficiency variants. It may have other genetic, environmental, occupational and lifestyle components.