Serum levels and genotype distribution of α1-antitrypsin in the general population

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RATIONALE: alpha1-Antitrypsin (AAT) deficiency is one of the commonest rare respiratory disorders worldwide. Diagnosis, assessment of risk for developing chronic obstructive pulmonary disease (COPD), and management of replacement therapy require the availability of precise and updated ranges for protein serum levels. OBJECTIVE: This paper aims to provide ranges of serum AAT according to the main genotype classes in the general population. METHODS: The authors correlated mean AAT serum levels with the main SERPINA1 variants (M1Ala/M1Val (rs6647), M3 (rs1303), M2/M4 (rs709932), S (rs17580) and Z (rs28929474)) in 6057 individuals enrolled in the Swiss Cohort Study on Air Pollution and Lung Diseases in Adults (SAPALDIA) cohort. RESULTS: The following ranges (5th-95th percentile) of AAT were found in the serum (g/litre): 1.050-1.640 for PI*MM, 0.880-1.369 for PI*MS, 0.730-1.060 for PI*SS, 0.660-0.997 for PI*MZ and 0.490-0.660 for PI*SZ. There was very little overlap in AAT serum levels between genotype classes generally not believed to confer an enhanced health risk (MM and MS) and those associated with an intermediate AAT deficiency and a potentially mildly enhanced health risk (SS, MZ). CONCLUSION: This work resulted in three important findings: technically updated and narrower serum ranges for AAT according to PI genotype; a suggestion for a population-based 'protective threshold' of AAT serum level, used in decision-making for replacement therapy; and more precise ranges framing the intermediate AAT deficiency area, a potential target for future primary prevention
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