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Alpha-1 antitrypsin gene variants in patients without severe deficiency diagnosed with pulmonary emphysema on chest CT

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Abstract-

Introduction: Although pulmonary involvement due to alpha-1 antitrypsin (AAT) deficiency has been widely described, most studies focus on the genotypes causing severe deficiency (< 60 mg/dL).Objective: The aim of this study was to analyze the prevalence of the different AAT gene variants that do not cause severe deficiency in patients with pulmonary emphysema diagnosed by thoracic computed tomography (CT). Furthermore, we assessed the risk associated with a non-severe decrease in AAT values in the pathogenesis of emphysema.Methods: Case-control study design that included patients who had a CT scan available of the entire thorax. In total, 176 patients with emphysema (cases) and 100 control subjects without emphysema were analyzed.Results: The prevalence of variants was higher among cases (25.6%; 45/176) than controls (22%; 22/100), although the difference was not statistically significant (P=0.504) when analyzed globally. In the control group, all the variants detected were MS. Excluding this variant, statistically significant differences were observed in the remaining variants (MZ, SS and SZ). Only 18% of the controls (all MS) presented values below our limit of normality, and all had values very close to the reference value (90 mg/dL). In contrast, 76% of patients with the other variants presented pathological levels. In a logistic regression model, both smoking and a non-severe reduction in AAT (60 to 90 mg/dL) increased the probability of emphysema.Conclusion: Our study confirms an association between certain variants in the alpha-1 antitrypsin gene that do not cause severe deficiency and the presence of pulmonary emphysema. This association with variants that are associated with reductions in serum AAT values is statistically significant and independent of smoking habit.

Index Terms- emphysema; alpha-1 antitrypsin; deficiency; variants

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