DEFICIENCY STATUS OF ALPHA-1-ANTITRYPSIN GENE IN CHRONIC OBSTRUCTIVE PULMONARY DISEASE (COPD)

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ABSTRACT

Chronic obstructive pulmonary disease (COPD) is the most prevalent clinical disorder. It is generally considered to be due to an imbalance between proteolytic enzymes and their inhibitors. A specific trypsin inhibitory protein was isolated along with the alpha-1-globulin of human serum. It was named as alpha-1-antitrypsin (α1-AT), as most of the serum trypsin inhibitory activity was found to be associated with the alpha-1 globulin fraction. Alpha-1-antitrypsin drew clinical interest when Laurell and Eriksson described the absence of plasma α1-AT in patients with degenerative lung disease leading to death in middle age. Genetic deficiencies resulting in the reduced levels of α1-AT in human plasma are particularly prevalent in individuals of north European descent. Deficiency of α1-AT is a recognized risk factor for COPD and is characterized by the progressive obstruction of airways, which is not fully reversible. The condition was hereditary and was likely to occur in individuals homozygous for mutated or deleted alpha-1-antitrypsin (***aat***) gene. Heterozygosity was found to exhibit half the normal proteolytic activity of α1-AT. Alpha-1-antityrpsin deficiency is widely under-diagnosed in many populations with majority of the individuals remaining undetected due to the delay in the onset and variability of respiratory symptoms. WHO stated that 2-3% of all alpha-1-antitrypsin deficient individuals were homozygous for PiZ and recommended screening for α1-AT deficiency in individuals with COPD, all adults and adolescents with asthma as well as neonates, children and adults with unexplained liver disease.

KEY WORDS: COPD, Genetic Predisposition, Alpha-1-Antitrypsin Deficiency