DO SERPINA1 GENE VARIANTS AFFECT THE CLINICAL COURSE OF GRANULOMATOSIS WITH POLYANGIITIS

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Alpha -antitripsin A AT deficiency is one of the most common genetic disorders in Caucasians There is a link between granulomatosis with polyangiitis GPA and most frequent variants of SERPINA gene encoding severe A AT deficiency activity However the potential effect of Pi Z Pi S and other SERPINA variants on clinical course of vasculitis are not well understood The aim of the study was to assess the possible effect of allelic variants of SERPINA gene on the activity and clinical course of GPA The study group consisted of patients with GPA Patients were stratified according to the disease severity patients during active phase n patients during remission on treatment n and patients during remission n A AT and hsCRP blood concentration was assessed by nephelometry A AT serum protein phenotyped by isoelectrofocusing and SERPINA genotyped by real-time PCR or direct sequencing Normal Pi MM genotype was detected in patients Pi MZ genotype in patients and Pi IM Pi MS Pi SZ in patient respectively Patients with abnormal Pi Z S or I genotype constitute in group I in group II and in group III In of patients with the GPA and in with lung lesions Pi MZ Pi MS and Pi SZ phenotypes were found The nodules pulmonary fibrosis have been the most frequent radiologic changes in patients with abnormal phenotype of A AT In patients with abnormal genotype Pi MZ or Pi MI acute onset of the GPA multi-organ involvement of upper and lower respiratory tract eyes