ALPHA-1-ANTITRYPSIN DEFICIENCY IN PATIENTS WITH GRANULOMATOSIS WITH POLYANGITIS

M. Hadzik-Blaszczyk[1] , T.M. Zielonka[1], R.Krupa[1], A. Rozy[2], K. A. Wardyn[1], J. Chorostowska-Wynimko[2], K. Zycinska[1]

[1]Department of Family Medicine , Internal and Metabolic Diseases, Medical University of Warsaw Czerniakowski Hospital, Stepinska Street 19/25 Warsaw Poland , Tel ./Fax + 48 22 318 63 25, e-mail : pb.mb@wp.pl

[2]Laboratory of Molecular Diagnostics and Immunology, National Institute of Tuberculosis and Lung Diseases, Warsaw

Alpha-1-antitrypsin deficiency (A1AT) is one of the most common genetic disorders. It has been suggested that A1AT deficiency might be occurred in ANCA+ vasculitis. The aim of the study was to estimate the frequency of A1AT deficiency and also the blood concentration of A1AT and hsCRP related to activity of disease. The survey was conducted on a group of 64 patients (41 women, 23 men) suffered from granulomatosis with polyangitis (GPA), treated at Czerniakowski Hospital between 2004 and 2014. Diagnosis was established according to ACR criteria. A1AT and hsCRP blood concentration assessment by nephelometry, phenotyping A1AT by isoelectrofocusing and real-time PCR genotyping were performed. Normal gene code MM were detected in 59 patients, mutated MZ allele in 2, and IM, MS, SZ in following 1 patients. According to the severity of disorder 3 groups were created: during induction or relapse (n=12), during remission (n=40), and during remission without treatment (n=12). Blood A1AT concentration in the following groups was: 172±50, 156±31 and 145±30 mg/dL, and concentration of hsCRP respectively 1.56±3.9, 0.64±0.77 and 0.86±1.27 mg/dL. Correlation between serum level of hsCRP and A1AT was observed (p=0.52). A1AT deficiency is observed in the group of patients with GPA and leads to lung lesion.