Analysis of SCN1A and SCN2A gene polymorphisms in epilepsy patients

K. Wize[1], U. Łagan-Jędrzejczyk[1], A. Gac[2], W. Kozubski[3], J. Dorszewska[1]
[1]*Poznan University of Medical Sciences, Laboratory of Neurobiology, Department of Neurology, Poland*
[2]*Poznan University of Medical Sciences, Students Scientific Neurobiological Association, Poland*
[3]*Poznan University of Medical Sciences, Chair and Department of Neurology, Poland*

Epilepsy is considered as a chronic neurological disorder, which requires long-lasting antiepileptic drugs (AEDs). The *SCN1A* and *SCN2A* genes encodes α subunits of neuronal voltage-gated sodium channel, which are targets for different AEDs. It is thought that various genetic variants of these genes are involved in the pathogenesis and treatment effectiveness of epilepsy. The aim of the study was to analyze the frequency of c.3184 A→G polymorphism of *SCN1A* gene and c.56 G→A polymorphism of *SCN2A* gene in Polish epilepsy patients and control group. There was taken into account kind of used treatment among epilepsy group. To this study we enrolled 46 epilepsy patients (20-66 aged) and 45 aged matches' controls (23-66 aged). Genetic study was conducted using HRM method. The study has showed that frequency of GG genotype of *SCN1A* 3184 A→G and AA *SCN2A* 56 A→G polymorphisms were higher in epilepsy patients than in control group. AG genotype of *SCN1A* 3184 A→G polymorphism was less frequent in epilepsy group as compared to controls (p=0,03) and patients with this genotype were mostly treated with polytheraphy consisting of newer and older AEDs. 2 of 3 epilepsy patients (67%) with AA genotype of *SCN2A* 56 A→G polymorphism were treated with newer AEDs in monotheraphy, despite of the duration of the disease more than 5 years. It seems that there is association between frequency of occurrence of *SCN1A* and *SCN2A* polymorphisms in epilepsy patients. Further study is needed to confirm the involvement of these genes in more personalized therapy