ANALYSIS OF SNCA AND PARK2 MUTATIONS IN SPORADIC PARKINSON'S DISEASE

A. Polrolniczak\textsuperscript{1}, J. Dorszewska\textsuperscript{1}, J. Florczak\textsuperscript{2}, M. Owecki\textsuperscript{2}, A. Różycka\textsuperscript{3}, B. Rubis\textsuperscript{4}, P. Jagodzinski\textsuperscript{3}, W. Kozubski\textsuperscript{2}

\textsuperscript{1}Laboratory of Neurobiology, Department of Neurology, \textsuperscript{2}Chair and Department of Neurology, \textsuperscript{3}Department of Biochemistry and Molecular Biology, \textsuperscript{4}Department of Clinical Chemistry and Molecular Diagnostics, University of Medical Sciences, Poznan, Poland

Early diagnosis of Parkinson's disease (PD) is often problematic because clinically it can be difficult to distinguish idiopathic PD from the other extrapyramidal disorders.

The etiology of sporadic PD is not clear, but it is currently assumed that genetic susceptibilities, including SNCA and PARK2 mutations, may be involved.

The aim of the study was analysis of SNCA and PARK2 mutation in Polish patients with sporadic PD and in control group.

Peripheral blood was collected from 34 (15 male and 19 female) patients with sporadic PD clinical diagnosis (the average age 58 years) and from 25 (7 male and 18 female) healthy controls (the average age 60 years).

Restriction-enzyme digestion of polymerase-chain reaction (PCR) amplified genomic DNA fragment of SNCA exon 3 detected no G88C mutation. PCR-amplification of parkin exons 2 and 4 also detected no exon deletion. Moreover exons 4, 7 and 11 of PARK2 gene was screened using real-time PCR/HRM and exon sequencing.

Mutation in tested exons of PARK2 gene were identified in 21% patients with sporadic PD and in 4% control subjects. All detected mutations were heterozygous. One of the PD patients had two mutations in PARK2 gene (G1281A, G601A). It was also observed, that mutation in PARK2 gene are associated with slow progression of PD and positive L-dopa response.

Our results showed, that screening for PARK2 mutations (G1281A, C924T, G601A) may be a component of genetic testing for sporadic PD and may be prognostic factor both for efficacy of pharmacotherapy and PD progression.