EARLY-ONSET PARKINSON’S DISEASE IN CZECH SLAVIC PATIENTS AND PARK2 MUTATIONS.

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INTRODUCTION
Mutations in the parkin gen (PARK2) have been associated with autosomal recessive early-onset Parkinson’s disease with various frequencies in different populations.

OBJECTIVES
The study was designed to determine the frequency of parkin allelic variants in Czech early-onset Parkinson’s disease patients and age-matched healthy controls.

MATERIAL AND METHODS
A total of 70 (47 males, 23 females) early-onset Parkinson’s disease patients (age at onset ≤ 40 years) and 75 (52 males, 23 females) controls were screened for the sequence variants and exon rearrangements in the parkin gene. All 12 exons of the PARK2 gene were amplified from the patient’s genomic DNA by the PCR.

DISCUSSION AND RESULTS
Previously described non-pathogenic polymorphisms p.S167N and p.D394N were seen in similar percentage in patients (7.1%, 10.0%) and controls (9.3%, 8.0%). Polymorphism p.V380L was almost twice as frequent in controls (25.3%) as in patients (14.3%). A novel sequence variant p.V380I (c.1138G>A) was identified in one control (1.3%). One patient (1.4%) and three controls (4.0%) were carriers of two different polymorphisms (p.S167N + p.D394N, p.S167N + p.V380L, p.V380I + p.D394N). Parkin mutations were identified in five patients (7.1%): the p.R334C point mutation was present in one patient, four patients had exon deletions. The detected mutations were observed in the heterozygous state except one homozygous deletion of the exon 4. No mutations were obtained in control subjects. A novel sequence variant p.V380I (c.1138G>A) was identified in one control.

CONCLUSIONS
Our study contributes to the growing body of evidence on the low frequency of the parkin mutations in the early-onset Parkinson’s disease suggesting the potential role of other genes in the pathogenesis of the disease.

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Key Words: Genetics; Parkinson’s disease