Association between \textit{PARK16} gene polymorphisms and susceptibility of Parkinson’s disease in a Chinese population

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ABSTRACT. Recent genome-wide association studies identified 11 risk loci in different populations of familial and sporadic Parkinson’s disease (PD) patients. Few loci have been verified in different European and Asian populations. We also validated 2 new single-nucleotide polymorphisms, rs947211 and rs823144, in \textit{PARK16} to explore their association with susceptibility to PD in the Xinjiang Uygur and Han populations. This case-control study included 312 PD patients (130 Uygur and 182 Han) and 359 control subjects (179 Uygur and 180 Han). Polymerase chain reaction-restriction fragment length polymorphism analysis and DNA sequencing were used to detect the rs947211 and rs823144 polymorphism in the \textit{PARK16} gene between the Xinjiang Uygur and Han populations. Frequencies of the A allele and AA genotype (42.1 and 15.7\%, respectively) of rs947211 in PD patients were significantly lower than those in the control group (54.7 and 28.7\%, respectively, \(P < 0.01\)). A allele and AA genotype frequencies of
rs823144 were 56.8 and 31.8% in the PD patients group and were 54.1 and 29.3% in the control group; no significant difference was found (P > 0.05). In both the Han and Uygur groups, the rs947211 polymorphism was associated with PD. Haplotype analysis also indicated that the A-A and G-A haplotypes were associated with PD. We found that the rs947211 polymorphism may be a susceptibility marker for PD in the Chinese population; the A-A and G-A haplotypes may be a protective factor and a risk factor, respectively, for PD in the Chinese population.

**Key words:** Genetic polymorphism; Parkinson’s disease; PARK16