

Association analysis of a polymorphism of interleukin 1 beta (IL-1 β) gene in temporal lobe epilepsy with hippocampal sclerosis in a Korean population

K Heo, *MK Chu, MA Kim, MA Shin, GW Kim, BI Lee

Department of Neurology, Yonsei University College of Medicine, Seoul, Korea; *Department of Neurology, Hallym University College of Medicine, Chunchon, Korea

Background and Objective: Temporal lobe epilepsy (TLE) with hippocampal sclerosis (HS) is the most widely recognized symptomatic focal epilepsy and frequently manifests resistance to drug therapy. The homozygotes for allele 2 at position-511 of IL-1 β gene promoter region (IL-1 β -511*2/2) has been reported as a genetic risk factor in the development of TLE with HS in a Japanese population.¹ However, this has not been confirmed in European and Chinese populations.² We investigated the association between IL-1 β -511 polymorphism and TLE with HS in a Korean population.

Methods: Forty-seven TLE patients with evidence of HS of Korean ancestry (31 patients with pathologically proven HS and 16 with HS on MRI) and 49 ethnically matched control subjects were selected for this study. A 304-base pair fragment in position of IL-1 β -511 was amplified by polymerase chain reaction (PCR). The PCR products were digested with restriction enzyme *Ava*I, and run on a 12% polyacrylamide gel. A single base pair polymorphism at position 511 in the promoter region of the IL-1 β gene was analyzed by the PCR-restricted fragment length polymorphism (RFLP) method.

Results: The mean age at seizure onset in the TLE with HS patients was 16.6 years (SD 8.3), which was similar to that of the Chinese patients (16.46 years) reported by Jin *et al.*² and older than that of the Japanese patients (9.4 years) reported by Kanemoto *et al.*¹ Allele 2 frequency in the controls was 48.0%, which was similar to that of Japanese (47.9%) and Chinese patients (50.0%). The analysis of genotype and allele distribution showed no significant difference between the TLE with HS patients and the controls. The heterozygous type (1/2) was the most common genotype in the TLE with HS patients (55.3%) and the controls (55.1%); 1/1, 14.9% and 2/2, 29.8% in the TLE with HS patients, and 1/1, 24.5% and 2/2, 20.4% in the controls. Allele 2 frequency in the TLE with HS patients was 57.4%. The distribution of genotypes and allele frequency were not significantly different among the TLE with HS patients with febrile convulsion (n=20), those without febrile convulsion, and the controls, and among the TLE with HS with the age at seizure onset <15 years (n=22), those with the age at seizure onset =15 years, and the controls, although the frequency of allele 2 seemed higher in TLE with HS patients with febrile convulsion than the controls (65.0% versus 48.0%, p=0.069).

Conclusion: We did not find a strong association between IL-1 β -511 polymorphism and the development of HS in this Korean population.

References

1. Kanemoto K, Kawasaki J, Yuasa S, *et al.* Increased frequency of interleukin-1beta-511T allele in patients with temporal lobe epilepsy, hippocampal sclerosis, and prolonged febrile convulsion. *Epilepsia* 2003; 44: 796-9
2. Jin L, Jia Y, Zhang B *et al.* Association analysis of a polymorphism of interleukin 1 beta (IL-1 beta) gene with temporal lobe epilepsy in a Chinese population. *Epilepsia* 2003; 44: 1306-9