Association of transforming growth factor-beta 1 gene polymorphism with genetic susceptibility to ossification of the posterior longitudinal ligament in Korean patients

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ABSTRACT. Ossification of the posterior longitudinal ligaments (OPLL) has been considered to be associated with abnormalities of bone metabolism, and transforming growth factor-β1 (TGF-β1) has been demonstrated to affect the bone remodeling process. We investigated two SNPs of the TGF-β1 promoter (-509C>T; rs1800469) and exon 1 (869T>C; rs1982073) in 298 Koreans (98 patients with OPLL and 200 control subjects). The promoter SNP -509C>T was determined by
PCR and RFLP, and the TaqMan probe assay was used to determine 869T>C polymorphism genotypes. The subjects were divided into OPLL continuous group (continuous type plus mixed type) and OPLL segmental group (segmental and localized type). We also separately analyzed this association according to gender difference. There was no significant difference in genotype distributions of -509C>T and 869T>C polymorphisms of the TGF-β1 gene between OPLL patients and controls. A combined analysis of TGF-β1 -509C>T and 869T>C polymorphisms showed no significant association with OPLL, and a subgroup analysis did not show any significant correlation between the SNP -509C>T or SNP 869T>C and OPLL subgroups. Stratification by gender demonstrated no significant effect. We conclude that promoter region (-509C>T) and exon 1 (869T>C) polymorphisms are not associated with OPLL in the Korean population.

**Key words:** Ossification; Posterior longitudinal ligament; Polymorphism; Transforming growth factor-β1