GMR Genetics and Molecular Research
Online Journal
ISSN:1676-5680

Association between *BMP-2* and *COL6A1* gene polymorphisms with susceptibility to ossification of the posterior longitudinal ligament of the cervical spine in Korean patients and family members

K.H. Kim, S.U. Kuh, J.Y. Park, S.J. Lee, H.S. Park, D.K. Chin, K.S. Kim and Y.E. Cho

Department of Neurosurgery, Spine and Spinal Cord Institute,
Gangnam Severance Hospital, Yonsei University College of Medicine,
Seoul, Korea

Corresponding author: Y.E. Cho
E-mail: yecho@yuhs.ac

Genet. Mol. Res. 13 (1): 2240-2247 (2014)
Received January 9, 2013
Accepted January 10, 2014
Published March 31, 2014
DOI http://dx.doi.org/10.4238/2014.March.31.4

ABSTRACT. *COL6A1* and *BMP-2* genes have been implicated in ossification of the posterior longitudinal ligament (OPLL) susceptibility in Japanese and Chinese Han populations. However, no study has yet investigated the DNA of unaffected family members of patients with OPLL. This study investigated differences in genetic polymorphisms of BMP-2 and COL6A1 between Korean patients with OPLL and their family members (with and without OPLL). A total of 321 subjects (110 patients with OPLL and 211 family members) were enrolled in the study. Associations between two single nucleotide polymorphisms (SNPs) of the *BMP-2* gene (Ser37Ala and Ser87Ser) and two SNPs of COL6A1 [promoter (-572) and intron 33 (+20)] with susceptibility to OPLL of the cervical spine were investigated between the two groups (OPLL+

and OPLL-). Of the 321 subjects, 162 had cervical OPLL (50.4%; 110 patients, 52 family members). There was a familial tendency of OPLL in 34 of the 110 families (30.9%). Allele and haplotype frequencies of the four SNPs in the *BMP-2* and *COL6A1* genes did not differ significantly between the OPLL+ and OPLL- groups, even when excluding participants over 50 years of age. This is the first report identifying SNPs of COL6A1 and BMP-2 in Korean patients and family members with OPLL. Although allele and haplotype frequencies were similar with those of a previous study in Japanese and Chinese patients, unaffected family members also showed similar rates of these SNPs in the present study. These results suggest that these SNPs may not directly influence the expression of OPLL.

Key words: Single nucleotide polymorphism; Genetic association; BMP-2; COL6A1; Ossification of the posterior longitudinal ligament

