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Lack of association between *COL1A1* and *COL9A2* single nucleotide polymorphisms and intervertebral disc degeneration

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Abstract

Background: Collagens are the main components of the extracellular matrix of intervertebral discs. The genetic mutations in collagen genes could potentially play a causal role in pathophysiology of intervertebral disc degeneration (IVDD). In this study, we investigate the association of COL1A1 and COL9A2 single nucleotide polymorphisms (SNPs) with IVDD. Material and Methods: ninety-six Iranian IVDD patients and 94 controls matched for age and sex were included. 5 cc of peripheral blood samples were obtained for DNA extraction using the Phenol–Chloroform method. The primers for SNPs COL1A1 rs909102 and COL9A2 were designed based on the TaqMan protocol and genotyped by real-time PCR with TaqMan. Results: The 'T' allele, 'CC' and 'TT' genotypes of COL1A1 rs909102 were more common among patients, however not significantly. Despite the similar allele distribution of COL9A2 rs137853213 in patients and controls, the homozygote genotypes were more frequent among patients, though this was not significant either. Conclusion: The allele and genotype distributions of COL1A1 rs909102 and COL9A2 rs137853213 SNPs were not significantly associated with IVDD in an Iranian population.

Keywords: Single nucleotide polymorphisms, intervertebral disc degeneration, collagen