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Novel visual system homeobox 1 gene mutations in Turkish patients with keratoconus

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ABSTRACT. The aim of this study was to screen the visual system homeobox 1 (*VSX1*) gene in Turkish patients with keratoconus (KC). The patient group consisted of 44 patients who had undergone corneal transplant surgery before the age of 30, for advanced and rapidly progressive KC. The control group comprised 250 healthy individuals. We detected two missense mutations, D144N and D295Y, in exon 2 and exon 5 of the *VSX1* gene, respectively, using next-generation sequencing analysis. The pathologic effects of the D144N and D295Y missense mutations on protein function were determined with bioinformatic

analysis tools, SIFT, PolyPhen, and MutationTaster. Aspartic acid at the 144th position was more preserved among species than aspartic acid at the 295th position of the VSX1 protein. In the control group, five different genetic variations were detected, two of which (rs8123716 and rs12480307) were synonymous with variations in the patient group. Our results suggested that the D144N and D295Y mutations might have a role in the pathogenesis of KC disease.

Key words: Keratoconus; Mutation; Next-generation sequencing; *VSX1*