عنوان مقاله:

Studying VSX۱ Gene Mutations in Patients with Keratoconus of Chaharmahal and Bakhtiari Province, Iran

محل انتشار:

مجله دانشگاه علوم پزشکی کرمان, دوره 20, شماره 3 (سال: 1392)

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خلاصه مقاله:

Background & Aims: Keratoconus (KC) is an eye disorder in which the cornea is swollen, thinned and deformed. Despite extensive studies, the pathophysiological processes and genetic etiology of KC is unknown. The disease incidence is approximately ۱ in ۲۰۰۰ and is the most common cause of corneal transplantation in the US. Many genes are involved in the disease, but evidence suggests a major role for VSX۱ in the etiology of KC. This study aimed to determine the frequency of mutations in exons ۲, ۴ of the VSX۱ gene in Chaharmahal and Bakhtiari province, Iran. Methods: In this experimental study, mutations in two exons including exons ۲ and ۴ of VSX۱ were investigated in ۵۰ patients with KC. DNA was extracted using a standard phenol-chloroform method. PCRSSCP/HA was performed, followed by DNA sequencing to confirm the identified motility shift. Results: H۲۴۴R mutation was identified in exon ۴ of only one patient. Conclusion: Our investigation showed that the KC-related VSX۱ mutations are found in very small samples in the study subjects from Iran. Further investigations on other genes are needed to clarify their roles in KC pathogenesis.

کلمات کلیدی:
Keratoconus, Gene VSX۱, Mutation