Study of VSX1 Mutations in Patients with Keratoconus in Southwest Iran Using PCR-Single-Strand Conformation Polymorphism/Heteroduplex Analysis and Sequencing Method


Abstract:

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 are unknown. The disease incidence is approximately 1 in 2,000, and it is the most common cause of corneal transplantation in the USA. Many genes are involved in the disease, but evidence suggests a major role for VSX1 in the etiology of KC. This study aimed to determine the frequency of mutations in exons 2, 3 and 4 of the VSX1 gene in Chaharmahal va Bakhtiari province in the southwest of Iran.