Keratoconus and VSX1 Polymorphism: A Preliminary Study in A Malaysian Population

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ABSTRACT

Keratoconus is a progressive disorder characterised by thinning of the cornea, and is associated with consanguinity, ultraviolet radiation, eye-rubbing and allergy. Visual system homeobox 1 (VSX1) gene is expressed in the human cornea and has been controversially implicated in keratoconus, wherein it was pathogenic in some populations while being a variation in gene frequency in the others. Molecular genetic analysis was conducted on 50 keratoconus patients and 100 unrelated controls. DNA extracted from EDTA blood samples were amplified via polymerase chain reaction (PCR) for the VSX1 polymorphisms at exons 2 and 4, and resulting PCR products were sequenced. The nucleotide sequences were aligned with the published VSX1 cDNA sequence (Genbank accession number NM_014588) using Sequencher 5.1 (Gene Codes Corporation). Polymorphisms in exon 2 of VSX1 were seen in two of the controls but not in keratoconus patients. The absence of mutation in keratoconus suggests that this is a polymorphism within the study’s population. Similar findings were reported in Italian, Slovenian and Saudi Arabian populations. Conversely, VSX1 has been reported to be pathogenic in other diverse populations such as in the Iranians, Indians, Canadians and Koreans. VSX1 gene with reference to exons 2 and 4 may not play a major role in the pathogenesis of keratoconus in the Malaysian population. Sample collection and genotyping is currently ongoing.