

Investigation of a PAX6 gene mutation in a Malaysian family with congenital aniridia

Abstract

Mutations in the PAX6 gene that cause aniridia have been identified in various ethnicities but not in the Malaysian population. Therefore, the objective of this study was to investigate the PAX6 mutation in a Malaysian family with congenital aniridia. In this study, a complete ophthalmic examination was performed on a Dusun ethnic family with aniridia. Genomic DNA was extracted from the peripheral blood of the subjects and screened for the PAX6 gene mutation using polymerase chain reaction amplification high-resolution melting curve analysis (PCR-HRM) followed by confirmation via direct DNA sequencing. A heterozygous G deletion (c.857delG) in exon 7 causing a frame shift in PAX6 was identified in all affected family members. Genotype-phenotype correlation analysis revealed congenital cataract and all affected family members showed a similar spectrum of aniridia with no phenotypic variability but with differences in severity that were age-dependent. In summary, by using a PCR-HRM approach, this study is the first to report a PAX6 mutation in a Malaysian family. This mutation is the cause of the aniridia spectra observed in this family and of congenital cataract.