A rare $PAX6$ mutation in a Chinese family with congenital aniridia


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ABSTRACT. Aniridia is an autosomal dominant disorder characterized by the complete or partial loss of the iris and is almost associated with mutations in the paired box gene 6 ($PAX6$). We examined three generations of a Chinese family with congenital aniridia and observed genetic defects. Exons of $PAX6$ from 12 family members were amplified by polymerase chain reaction, sequenced, and compared with reference sequences in NCBI reference sequence database (http://www.ncbi.nlm.nih.gov/nuccore/NG_008679.1?from=5001&to=38170&report=genbank). A rare mutation c.2T>A (M1K) in exon 4 of $PAX6$ was identified in all affected family members but not in unaffected family members. Our results suggest that the c.2T>A (M1K) mutation may be responsible for the pathogenesis of congenital aniridia in this family. To our knowledge, this is the first report of the M1K mutation in $PAX6$ in a Chinese family with this disease and the second report worldwide.

Key words: Congenital aniridia; $PAX6$; Gene mutation; Eye development