A novel mutation of PAX6 identified in a Chinese twin family with congenital aniridia complicated with nystagmus

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ABSTRACT. Genetic variations within the paired box gene 6 (PAX6) gene are associated with congenital aniridia. To detect the genetic defects in a Chinese twin family with congenital aniridia and nystagmus, exons of PAX6 were amplified by polymerase chain reaction (PCR), sequenced and compared with a reference database. Six members from the family of three generations were included in the study. The twins’ father presented with congenital aniridia, nystagmus and cataract at birth, while the twins presented with congenital aniridia and nystagmus. A novel mutation c.888 insA in exon 10 of PAX6 was identified in all affected individuals. This study suggests that the novel mutation c.888 insA is likely responsible for the pathogenesis of the congenital aniridia and nystagmus in this family.
pedigree. To the best of our knowledge, this is the first report of this mutation in PAX6 gene in pedigree with aniridia. Furthermore, no PAX6 gene defect was reported in twins with congenital aniridia.

**Key words:** Congenital aniridia; Nystagmus; PAX6; Twin