Reviewer's report

Title: Sequence variations in the human PAX6 gene with Aniridia in South Indian population

Version: 1 Date: 12 January 2004

Reviewer: Noriyuki Azuma

Reviewer's report:

General
It is well known that haploinsufficiency of the PAX6 gene causes the aniridia phenotype. Most mutations detected in aniridia result in premature translational termination on one of the alleles. The authors identified novel PAX6 mutations in South Indian population. However, the data only confirmed the haploinsufficiency theory and does not provide any novel finding on genetic pathogenesis of aniridia.

Minor Essential Revisions

In the Result session, the gene accession number for use of nucleotide numbering was not provided.

In the last paragraph on page 7: Did the proband 16-1 show only lens dislocation or also systemic anomalies matched with Marfan syndrome?

What next?: Unable to decide on acceptance or rejection until the authors have responded to the major compulsory revisions

Level of interest: An article of limited interest

Quality of written English: Needs some language corrections before being published

Statistical review: No