Reviewer's report

Title: Sequence variations in the human PAX6 gene with aniridia in south Indian population

Version: 2 Date: 24 March 2004

Reviewer: Patrick CALVAS

Reviewer's report:

General

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Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)

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Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)

Abstract: p2 l7 were instead of was
p2 l17 stop codons (physically present) instead of premature termination (putative functional effect)

background:
p3 l25 of the pax6 protein causing (instead of due to) haploinsufficiency

discussion:
p10 l11 the mutations if translated, would result in...

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Discretionary Revisions (which the author can choose to ignore)

discretionary revisions
p2 l17 here we add 3 novel gene mutations in South Indian population to the...
p2 l18 mutations in the pax6 gene correlate...

Once again, the association of proven aniridia (PAX6 mutation) with Marfan's syndrome (now convincing) should not be interpreted in the same way as aniridia in Marfan's children. According to my point of view the presence of aniridia in a Marfan's syndrome (without PAX6 mutation) is a very interesting and rare fact that might be interpreted as an intriguing effect of lens development disruption (see Collinson et al. PNAS 2001; 98:9688-9693.

What next?: Accept after minor essential revisions

Level of interest: An article of limited interest

Quality of written English: Acceptable
Statistical review: No

Declaration of competing interests: None