Reviewer's report

Title: Inherited KIF21A and PAX6 gene Mutations in A Boy with Congenital Fibrosis of Extraocular Muscles and Aniridia

Version: 2 Date: 26 December 2012

Reviewer: Anne Slavotinek

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Ying et al.

This paper describes the detection of PAX6 and KIF2 mutations in a male with aniridia and congenital fibrosis of the extraocular muscles. The report is straightforward and the phenotypes and mutations are clearly described.

Major Compulsory Revisions:

“In addition, aniridia is usually associated with kidney nephroblastoma, genitourinary anomalies, or mental retardation and cerebellar ataxia[5,6].” – Aniridia can be associated with these findings but they are part of the WAGR spectrum and usually associated with a contiguous gene deletion syndrome or WT1 deletion – not related to PAX6 mutations. However, PAX6 mutations can be associated with brain malformations, for example, “PAX6 aniridia and interhemispheric brain anomalies.”, Abouzeid H, Youssef MA, ElShakankiri N, Hauser P, Munier FL, Schorderet DF.

Mol Vis. 2009 Oct 17;15:2074-83. The authors need to clarify this point.

Minor Essential Revisions:

“Here we report a boy carrying with dominant mutations in KIF21A and PAX6 gene – omit ‘with’ and italicize PAX6

7-years old boy – 7-year old boy

R954W - p.Arg954Trp

p.L249Yfs should be written p.Leu249TyrfsX22 according to what you have written in the text

bilateral optic nerve hapoplasia – bilateral optic nerve hypoplasia

Argnine (A) - Arginine

Gene names should be italicized

In Figure 1, the pedigree shading could be standardized – so that shading of the left hand side of the pedigree symbol represents CFEOM, regardless of gender.

Level of interest: An article whose findings are important to those with closely related research interests
Quality of written English: Needs some language corrections before being published

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:
I declare that I have no competing interests