Reviewer’s report

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

Version: 2 Date: 28 January 2013

Reviewer: Suma Shankar

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Major Compulsory Revisions:

1. In Conclusion: Include tahn both CFEOM type I and aniridia are AD disorders. The proband's children have 50% risk of inheriting each of these disorders. His children can inherit both CFEOM and aniridia from him.

2. Risk for parents to have additonal children with teh 2 disorders should also be mentioned in discussion.

3. In isolated aniridia with mutations in PAX6 -- THERE IS NO INCREASED RISK OF WILMS tumor. WILMs tumor risk is increased only in individuals with deletion of PAX6 along with involvement of neighbouring gene "WT1". Explain about importance of differentiating the 2 so that now that a PAX6 mutation has been identified, screening for abdominal tumors by routine U?S not required any longer and this is important for families and reduces anxiety about tumors.

4. Describe other clinical features and recommnedationsfor follow up of these patients.

5. Please correct on page 3, 2nd para last line -- "WAGR" caused by contiguous gene deletion syndrome.

7. In Materials and methods:

8. Change "accepted" to "underwent" strabismus surgery.

9. Change line 7 & 8 to proband was found to have limited extraocular movment soon after birth.

10. Page 5 paragraph 3 - please provide additional details as to why the authors feel the transcript will be subject to NMDA. Describe when a truncated protein may be formed versus when it will be haploinsufficiency because of NMDA.

11. indications and prognosis in CFEOM1 when surgical options are sought.

Level of interest: An article of limited interest
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