Author's response to reviews

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

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Author's response to reviews: see over
Dear Editors,

We thank all reviewers for their thorough inspections and constructive comments on our manuscript “Inherited KIF21A and PAX6 gene mutations in a child with Congenital Fibrosis of Extraocular Muscles and Aniridia”. According to their suggestions, we revised the text as follows:

Reviewer #1

1. “The probability of developing both CFEOM1 and aniridia for their additional child would be 6.25%, should his parents produce an additional child” – this is incorrect – probability is 25% (1 in 2 x 1 in 2) as in the previous sentence for the proband.”

Thanks for your comments. We revised the incorrect statement.

2. “There are a number of minor grammatical problems – some changes are suggested below, but we would suggest that the authors recheck their English language usage again”.

Thanks. Professor J. Fielding Hejtmancik help us to revise this manuscript and change some errors in English usage.

Reviewer #2
1. “Conclusion statement regarding the importance of identifying PAX6 mutation. Individuals with PAX6 mutations are not at risk for Wilm's tumor and hence will not require routine screening for Wilm's tumor as needed in WAGR syndrome or where the molecular diagnosis for aniridia is not known.”

Thanks for your comments. We added these sentences of “However, since his aniridia is a consequence of an internal mutation within the PAX6 gene rather than a contiguous gene deletion including WT1, neither he nor his offspring will be at increased risk for the WAGR syndrome.” in the discussion for making the statements clear.