Reviewer's report

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

**Version:** 3 **Date:** 11 March 2013

**Reviewer:** Anne Slavotinek

**Reviewer's report:**

Minor essential revisions:

“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.

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.

Abstract:

“reveals a one base pair deletion”

“The deletion results in a frameshift change in the PAX6 gene and..”

Conclusion:

“….boy carrying PAX6 and KIF21A mutations,..”