Author's response to reviews

Title: Identification of a novel homozygous nonsense mutation in EYS in a Chinese family with autosomal recessive retinitis pigmentosa

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Version: 5 Date: 14 July 2010

Author's response to reviews: see over
Dear Prof. Wasim Ahmad,

Thank you for reviewing our manuscript and recommending us to submit a revised version. We appreciate the excellent comments made by three reviewers. We have revised the manuscript based on the editorial and reviewer suggestions. The details are listed below. The changes made are in blue in the text.

**Reviewer's report**

**Title:** Identification of a novel homozygous nonsense mutation in EYS in a Chinese family with autosomal recessive retinitis pigmentosa  
**Version:** 3  
**Date:** 14 June 2010  
**Reviewer:** Rob WJ Collin

**Reviewer's report:**

The authors have addressed the majority of points raised by all the referees in an adequate manner. However, there are still some small issues that have to be addressed.

1) page 5, line 14. The authors put ‘encoding the protein SPAM’ between parentheses. Yet, from the next sentence onwards, they use EYS as the name of the protein throughout the remainder of the manuscript. Please correct.

The mistake has been corrected.  

2) page 8, line 12. The authors state that the unaffected brother II4 inherits one affected allele. Figure 1 however suggests that II4 has neither of the two disease alleles. Did the authors sequence all individuals including non-affected relatives, to determine carriership?

The figure is right, and we did sequence all family members. II4 inherited neither of the mutations from his parents. In the revision, the sentence has been changed to “whereas their unaffected brother II4 inherited two normal chromosomes, and the proband’s daughter III1 only one affected allele”.

3) page 9, line 16, and page 10, line 15; pT3156X should state pY3156X. Please correct.

The mistake has been corrected.

4) page 15, note added to proof. The authors state that besides three recently published papers, they additionally have identified a mutation in the Chinese population. What do the authors mean? If they refer to the p.E1836X mutation described in their manuscript, they shouldn't mention it in the Note added to proof. If they mean that they have identified a second EYS mutation in the Chinese population, I would recommend that they add that family to this manuscript. Please clarify!

The sentence “Additionally, we have identified a mutation in the Chinese population” has been deleted in the revision.
5) page 16, reference 22; Littink KW is mentioned twice. 
The error has been corrected in the revision.

Thank you very much for improving our manuscript. If you have any questions, please feel free to contact me.

Sincerely yours,

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