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.

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 carriersonship?

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

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!

5) page 16, reference 22; Littink KW is mentioned twice.

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Acceptable

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.