Reviewer’s report

Title: The low penetrance of retinoblastoma for the V654L mutation in RB1 gene

Version: 1 Date: 24 November 2010

Reviewer: Francisco Sanchez

Reviewer’s report:

In this article, the author describe a mutation in RB1 gene associated with a low penetrance retinoblastoma case. The following points need to be addressed:

1) The authors write in the abstract that the age of diagnosis varied from 16 and 46 years, but in results they write that the average age of the affected cases was 30 years old. They need to clarify this point. Retinoblastoma is diagnosed typically before the age of three years old. The are few cases published after five years old.

2) In methods, what are the primer sequences write in "polymerase chain reaction"?

3) In conclusion, the last sentence is not correct. The V654L mutation is itself sufficient to develop low penetrance retinoblastoma. It is a typical mutation associated to low penetrance.

4) In background, they write that low penetrance RB1 mutations are due "particularly from deletion of whole RB1 gene". This sentence is not correct.

5) Results chapter is confusing. For example, the number of heterozygous germ-line mutation members are not clear.

6) In discussion chapter, the authors do not comment anything about the cases described with V654L mutation.

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

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.