Reviewer's report

Title: Low penetrance of retinoblastoma for V654L mutation of the RB1 gene

Version: 2 Date: 29 April 2011

Reviewer: Annemarie Hedwig H van der Hout

Reviewer's report:

Major Compulsory Revisions:
In the methods section, the authors mention only one set of primers for amplification of the coding region of the RB1 gene. I guess these are only the primers for amplification of exon 19 in which the mutation is located? This point should be clarified.

page 5. What is meant by non-intermediary germline mutations?

page 11. What is meant by "none of them exhibited variable severity"?

Minor essential revisions:
In fig. 1 individual III3 is not marked * although according to the text he tested positive for the mutation.

the correct nomenclature for the mutation is p.V654L

Discretionary Revisions:
The authors stress that in this particular family the penetrance of the mutation V654L is much lower than in other families carrying this same mutation. They still only discuss why V654L is a mutation with reduced penetrance, compared to other mutations, but not why the penetrance in this family is so much lower than in other families carrying this same mutation.

Level of interest: An article of limited interest

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.

Declaration of competing interests:
'I declare that I have no competing interests'