Reviewer's report

Title: Gorlin syndrome associated with small bowel carcinoma and neuroleiomyomatosis of the gastrointestinal tract: Is two-hit mutational inactivation of PTCH a rare pathomechanism in GI malignancies? Case report and review of literature

Version: 1 Date: 5 February 2010

Reviewer: Vesna Musani

Reviewer's report:

A few comments and suggestions:

1. Major Compulsory Revisions
Since the point of the article is to connect the two unusual symptoms with the Gorlin syndrome, and the authors suggest that the tumors are caused by second mutational hit of the PTCH gene, there should be more evidence of that connection. I would like to see the results of PTCH mutation screening in at least one of two tumors. If it is not possible (i.e. no more tumor sample), Discussion and Conclusion should be rewritten with less emphasis on the two-hit inactivation and it should be removed from the title.

2. Minor Essential Revisions
'stop codon' is misspelled several times as 'stopcodon' in the article.

3. Discretionary Revisions
It would be interesting to see some immunohistochemical PTCH staining.

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.