Reviewer’s report

Title: Gorlin syndrome associated with small bowel carcinoma and mesenchymal proliferation of the gastrointestinal tract: Case report and review of literature

Version: 5 Date: 31 May 2010

Reviewer: Mariana Cajaiba

Reviewer’s report:

The paper reads really well, and after editing and modifications by the authors, I recommend publication. Congratulations to the authors for the thorough work.

Some minor suggestions that could improve the quality of the manuscript:

1. Stress the fact that this a new mutation that has never been described, and adds important information to the genetic spectrum of Gorlin syndrome.
2. Discuss possible pathogenic effects of the newly described mutation through correlation with the PTCH function/ expected functional defects in PTCH protein resulting from this mutation.
3. In the Case Presentation section, "Pathological and histopathological findings" is redundant; I suggest keeping only "Pathological findings".
4. In the future, the authors should consider further characterization of the mesenchymal lesion, maybe in the form of a "pathology-oriented", peer-reviewed short communication or letter to the editor. I suggest including broader histopathological documentation of the lesion, including gross pictures, high power photographs, pictures of the neuronal component, and additional ancillary stains that were not shown in this paper. With detailed documentation of this lesion, including a deeper discussion on the possible differential diagnoses, the authors could defend better their theory of a new lesion, and propose the term neuroleiomyomatosis based on more extensive scientific background. You should definitely explore your interesting pathology findings further.

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.