Reviewer’s report

Title: Early screening the small bowel is key to protect Peutz-Jeghers Syndrome patients from surgery: a novel mutation c.243delG in STK11 gene

Version: 0 Date: 20 Apr 2018

Reviewer: Randall W Burt

Reviewer's report:

This case report describes a child with Peutz-Jeghers syndrome. The patient developed symptoms related to colonic and small bowel polyps, which were found and treated. The child also had a de novo and unique STK11 mutation that was carefully found and verified.

Several items would be very helpful to this report.

The authors should give:

1. The fraction of expected de novo mutations in this disease (often stated to be about 25%).

2. A clear statement of the generally agreed upon screening and surveillance recommendations for the large and small bowel in Peutz-Jegher's syndrome---a table would be good for this. Several papers are quoted, but a summary recommendation would be very helpful.

Not as relevant to this paper, but nonetheless very important to the management of Peutz-Jeghers syndrome would be a table summarizing the cancer risks and screening guidelines for GI and non-GI malignancies. This is optional but may be very helpful. At least a reference to this information should be given.

Are the methods appropriate and well described?  
If not, please specify what is required in your comments to the authors.

Yes

Does the work include the necessary controls?  
If not, please specify which controls are required in your comments to the authors.

Yes

Are the conclusions drawn adequately supported by the data shown?  
If not, please explain in your comments to the authors.

Yes
Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

Not relevant to this manuscript

Quality of written English
Please indicate the quality of language in the manuscript:

Acceptable

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