**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:** 13 Apr 2018

**Reviewer:** Shirley V. Hodgson

**Reviewer's report:**

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This is a case report of a boy with isolated Peutz-Jeghers syndrome and a novel germline mutation in the STK11 gene. The report focuses on the novel mutation and the gastrointestinal complications, which the authors indicate the need for more intensive screening for young children with this condition. The English is very poor throughout and requires extensive revision. Some examples include:

"got abdomen cramps", "but not in his parents, which is not recorded in databases.", "diagnosis of PJS was affirmed", "endoscopy was applied", "A survey of STK11 gene", "large defection and missense 17 mutation", etc.

The mutation is reasonably described, but 50 unrelated controls is an extremely small number to test to see whether it occurs in the general population. The structure predition seems good although the picture of the resultant protein is much smaller then the wild type protein. In the figures and the text there is no mention of the classical arborising histology seen in PJS polyps despite it being apparent in the histology slide shown.

The comment that extensive surveillance would be recommended for children from the age of 4-5y is not sufficiently discussed in the context of the disadvanateges of such screening in young children, and the fact that the patient described in this paper was found to have symptomatic polyps from the age of 7y. Also it seems worrying that one patient discussed from a quoted literature survey of 22 paediatric patients on surveillansve developed a post-Laparascopic-DBE pelvic abscess. A more critical review of the pros and cons of such screening in children would be appropriate.
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?
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Unable to assess

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