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: 23 Jan 2018
Reviewer: Andrzej Plawski

Reviewer's report:

The authors made good work.

They diagnosed correctly the PJS patients and confirmed their clinical diagnosis by detection of frameshift mutation c.243delG in STK11. Due to the c.243delG in novel mutation they wide analyze an impact of the mutation on the protein product and its activity. The main point of the report is the age of the baseline colonoscopy and upper GI endoscopy should be performed. There are recommendation, to perform it at the age of 8. But in the presented case the symptoms occurred earlier. Screening strategy must avoid most of patients from severe conditions and in the case of PJS it may be worthy to discuss the base age. The PJS is not frequent condition and another cases, deepen knowledge and may led to improve the patients care.

The PJS is a syndrome increasing cancer risk in various organs. As we know in young boy there is the increased risk of testicular tumors and it may occur at age of 5, but the authors did not mention about any other examination of the patient. It should be added

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?
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Yes
Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
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Not relevant to this manuscript

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