Reviewer's report

Title: Mutations in the STK11 gene in Czech Peutz-Jeghers families

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Reviewer: D Nageshwar Reddy

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Authors of the present study investigated 5 families with individuals affected with Peutz-Jeghers syndrome (PJS) for mutations in the promoter, coding region and splice-site boundaries of STK11 gene, by sequencing and MLPA assay. They report one frameshift and two large deletions in 6 individuals out of eight individuals screened. The study is well designed and lucidly presented.

Major Compulsory Revisions:
None

Minor Essential Revisions:
1. In a statement in paragraph 7 in the results and discussion section, ‘The mean age of Konishi et al. reviewed 103 …’ the mean values probably should have been 31.2 and 39.7 instead of 31,2 and 39,7.

Discretionary Revisions:
1. Representative chromatogram of MLPA analysis in the figures can be more descriptive and self explanatory (X- and Y-axes may be defined).

2. An interesting observation was a sporadic case (C-1) with hyperpigmentation and one adenomatous polyp harbouring a germline deletion of entire STK11 gene. Do they have any hypothesis to explain the subdued manifestation of the condition in the patient?

Level of interest: An article of importance in its field

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.