Reviewer's report

Title: High prevalence of germline STK11 mutations in Hungarian Peutz-Jeghers Syndrome patients

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Reviewer: Waltraut Friedl

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The manuscript 'High prevalence of germline STK11 mutations in Hungarian Peutz-Jeghers Syndrome patients' by Janos Papp et al. describes the detection of STK11 mutations in a cohort of 13 Hungarian PJS families. Of note, the mutation detection rate is 100%, when direct sequencing and MLPA deletion screening were applied. Moreover, each family has a different mutation, and most of the identified mutations are novel.

The laboratory work is well performed, the results are clearly presented and competently discussed. In addition to the detection of mutations on genomic DNA the work includes semiquantitative analysis of mutated mRNA expression.

Of interest is the observation that the mutation in the splice acceptor site of exon 2 results in the deletion of both exons 2 and 3 as shown by mRNA analysis; moreover, the large deletion of exons 3-7 (detected by MLPA analysis and exactly defined by identification of the breaking point on genomic DNA) results in the additional deletion of exon 2 on mRNA level. As the authors point out, one reason for this might be the presence of the minor U12-dependent spliceosom (AT-AC) in intron 2.

Minor comment:
In table 1 the relationship in family HP03 (uncle&cousin) seems to be not correctly defined. Should it be uncle and nephew? Or two cousins?

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