Reviewer's report

Title: A Novel De Novo Mutation in the Serine-Threonine Kinase STK11 Gene in a Korean patient with Peutz-Jeghers Syndrome

Version: 1 Date: 5 November 2007

Reviewer: Giriraj Chandak

Reviewer's report:

General

The paper reports a mutation in STK11 gene in a patient with Peutz Jeghers Syndrome. Several mutations in this gene have been reported to be associated with PJS and the present study and this report adds another one to the existing list. The mutation is novel but it leads to the effect that has been predicted by an earlier report of mutation (Thakur et al, 2006), which unfortunately is not quoted in the text.

The manuscript does not present anything novel apart from adding to the existing list of mutations in STK11 gene and hence may be submitted as a letter or a short report and should pool the results and discussion section in one.

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Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)

The language of the paper is poor and several grammatical inconsistencies are present. I suggest that the authors consult somebody well-versed with English literature and get the manuscript corrected for all the language and grammar related issues. To illustrate the point, following are the examples:

In abstract, Patient with PJS are at an increase risk of developing multi-organ cancer, most frequently gastrointestinal tracts-'increase' should be 'increased'

In discussion, “These included histopathologically hamartoma together with classical mucocutaneous hyperpigmentation and small-bowel polyposis”, ‘histopathologically should be ‘histopathologically proven hamartoma’.

In background section, the statement “The relative cancer risk is 15.2 and no significant difference in overall cancer risk between genders [3]” is not clear and may be reframed.

Was the polyp tissue also analysed for the mutation?

‘Case presentation’ should be substituted with methodology and ‘identification of
mutation should be included in the results and combined with discussion section.

The number of references are too many for one topic for example ‘Human STK11 consists of nine coding exons with a 433 amino acid coding sequence and on non-coding exon 10 spanning 23 kb [9, 10, 19, 20, 25].’ They may be appropriately reduced.

The effect of this mutation with reference to generation of a stop codon at codon 286 has already been well described in paper by Thakur et al, BMC Med Genet;7:73,2006 and hence this should be referred to in the text.

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Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)

LKB should be expanded too and included in the ‘list of abbreviations.’

The paper should be modified as per the abovementioned suggestions.

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Discretionary Revisions (which the author can choose to ignore)

What next?: Unable to decide on acceptance or rejection until the authors have responded to the major compulsory revisions

Level of interest: An article of limited interest

Quality of written English: Not suitable for publication unless extensively edited

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'