Reviewer's report

Title: Birt-Hogg-Dubé Syndrome in two Chinese families with Mutations in the FLCN Gene

Version: 2 Date: 03 Nov 2017

Reviewer: Xingnan Li

Reviewer's report:

In this study, Dr. Jiang et al. performed a family study of Birt-Hogg-Dubé Syndrome and indicated two mutations in FLCN gene associated with BHDS. Some of my concerns are:

Major:

(1) In Abstract, a Methods section is needed (separated from Results section).

(2) Authors mentioned that more than 200 mutations in FLCN have been identified (Table 1). Have all these mutations been checked in this study? What's the results (P values)? Especially, the three most common mutations being mentioned?

(3) Methods section is too brief and needs much more details. (3a) Which family members have been sequenced? (3b) How have they been sequenced? such as machine, alignment approach, and genotype call, etc. (3c) Statistical analysis method should be explained in detail, such as approach (family or case-control) and p-values, etc.

Minor:

(1) In Abstract (Line 25 and 28), please label exon numbers for two mutations.

(2) Figures 1-4 should have a general title.
Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

No

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Unable to assess

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

Yes

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

Not relevant to this manuscript

Quality of written English
Please indicate the quality of language in the manuscript:

Acceptable

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