Author’s response to reviews

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

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Version: 4 Date: 05 Dec 2017

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Dear editor,

My responses are as follows according to the questions.

(1) It’s true that we chose whole exome sequencing on the two subjects as the first step to detect the suspected mutations of the two families in our study. The two mutations c.1570_1580insA (exon 14) and c.649C>T (exon 7) weren’t chosen artificially, but depended on the sequencing results. These two mutations are the only mutations we detected in these two subjects.

(2) It’s correct that the second step is Sanger sequencing of corresponding exons (only two mutations on exon 7 and exon 14) specifically on the basis of the first step. Genotypes of
the two mutations for control subjects (II10, III10, III11, III13, IV2) have been clarified at the end of the results section (outlined in red).

(3) In regard to the statistical analysis, it’s permissible that all cases has mutation and all control do not inherit that mutation used as the criterion of significance.

Thank you very much for your careful revision.

Best wishes,
Hong Jiang