Author's response to reviews

Title: Intragenic duplication in the PHKD1 gene in autosomal recessive polycystic kidney disease

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Response to reviewers
To reviewer #1
1. As the reviewer suggested, other exons also appeared to be duplicated. We did not use any criteria to select the exons 45/46 and rule out others, but we performed MLPA to validate the putative copy number changes.
2. Exons 45 and are located between MBC-2 and D6S1344. We have indicated the location of the exons in the figure and added a comment in the Figure legends.

To reviewer #2
We did not analyze the newly identified exons. We have added a comment of the new exons in the Discussion (page 15, line 235) and added a related reference (Boddu et al. 2014).