Reviewer’s report

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

Version: 3 Date: 29 April 2015

Reviewer: Hyojin Chae

Reviewer’s report:

Discretionary Revisions

1. The supplementary figure shows other exons which could also regarded as candidate exons to be ruled out for copy number variations (deletions/duplications). Please describe the criteria by which exon 45 and 46 were selected as candidates.

2. The locations of the haplotype markers could be given in more detailed manner in regard to the genomic locations of intron44~46. Is it possible to modify figure 2A accordingly? If not please address in the manuscript.

Level of interest: An article of outstanding merit and interest 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.