Reviewer's report

Title: High Resolution Melt analysis for mutation screening in PKD1 and PKD2.

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Reviewer: Sandro Rossetti

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High resolution melt analysis for mutation screening in PKD1 and PKD2, Bataille S, Berland Y, Fontes M, and Burtey S.

This manuscript describes the mutation analysis of a cohort of ADPKD patients by using high resolution melt analysis for most of the exons and direct sequencing for some remaining exons.

Overall, the work described here is well planned, methods are sound, and the mutations reported will be a worthwhile addition to the existing literature.

- Minor Essential Revisions:

1) I suggest using the word “mutation” for pathogenic changes only and “sequence variant” as a general word for any change in the DNA sequence. Right now, the authors mention 440 “mutations” in the abstract (which are more properly “sequence variants”, and similar elsewhere), and introduce a third definition, “sequence variation”, in the 3 tables.

2) It is unclear what exons are sequenced: PKD1 exons 1, 41 and 42 according to the discussion section, or PKD1 exons 1, 42 and 43 according to supplementary data 1 that describe primers and analysis conditions? This is important information for anyone wishing to adopt this approach. It is worth adding a couple of sentences explaining the technical issues that do not allow analysis of these exons by high resolution melt, this is again important information for anyone interested in evaluating such technology.

3) A paper has been recently published (Hoefele J et al. Nephrol Dial Transplant. 2010, describing a similar work) that the authors may want to include in the discussion.

Level of interest: An article of importance in its field

Quality of written English: Needs some language corrections before being published

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