Author's response to reviews

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

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Author's response to reviews: see over
Dear editor,

Thank you to accept our manuscript called “High Resolution Melt analysis for mutation screening in *PKD1* and *PKD2*”. Please find in attached file a revised manuscript including asked modifications.

**Major revisions** (we require the author to make these changes)

References: The references have only been cited up to 32, please cite the rest in the text or remove them from the references

We made no changes, all the references are cited in the text or in the tables.

Additional files:

We correct the additional files as asked.

**Minor**

Tables: please remove the visible vertical lines from your tables

We done this

Figures: It is important for the final layout of the manuscript that the figures are cropped as closely as possible to minimise white space around the image

We cropped the figures

We hope now our manuscript is ready to be published in BMC nephrology.

Sincerely yours.

Professor Stéphane Burtey
Responses to reviewer’s comments

Please check mutation nomenclature through all text and tables. Splice site mutations in the text and mainly in tables should be corrected according to nomenclature recommended by the HGVS. Other mutation nomenclature should also be corrected (i.e. mutation p.ins3781_3782Glu should be named p.Asp3781_Val3782insGlu, or p.2894_2896delANS should be named p.Ala2894_Ser2896del).

We checked and corrected all the mutation nomenclature through text and tables.

Abstract:
Results “we identified 440 sequence variants in the 37 patients; 138 were different sequence variants”. Re-write the sentence.

We re-writed the sentence.

Methods:
- DNA samples: control sample number and description should be included here.
  We included the control group here.
- Reverse transcription analysis:
  commercial name is Qiagen instead of Quiagen Use the same style for primer sequences.
  We corrected these two points.
- Sequencing: state kit and sequencer used.
  We added theses data in the manuscript.

Results:
First and second paragraph: the results are not clearly stated and figures are difficult to follow. In the abstract it is stated that 440 variants were detected, but this figure cannot be obtained from the ones shown in the results section.

We re-writed the second paragraph to ease the lecture. We identified 440 variants, 410 in PKD1 and 30 in PKD2.

4th paragraph: “In addition, this mutation was previously described associated with lack of expression of PC-1 in primary cilia ()”. No reference is given.
We added the reference.

Last sentence of results is difficult to understand, please re-write.
We re-writed it

Discussion:
3rd paragraph: “54 new sequence variants in PKD1: 20 were classified as mutation”. It should be stated that 52 were the variants detected and 18 of them were mutations.
We corrected it