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:

Marseille, 26 August 2011

Dear editor,

Thank you to accept our manuscript called “High Resolution Melt analysis for mutation screening in PKD1 and PKD2”. We were very interested in the comments made by the reviewers. Please find in attached file a revised manuscript including asked modifications.

We include the emails of all the authors and we remove word count and keywords from the manuscript. We placed the table in the manuscript and not in additional files.

Point-by-point responses to reviewer’s comments are given below.

Again, we are very grateful for acceptance to publish our manuscript in BMC Nephrology because of the importance of the article in the field as stated by reviewers.

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-wrote 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 Qiagen 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