Reviewer’s report

Title: Whole exome sequencing reveals a stop-gain mutation of PKD2 in an autosomal dominant polycystic kidney disease family complicated with aortic dissection

Version: 0 Date: 18 Jul 2017

Reviewer: Andrew Paterson

Reviewer's report:
This is a report of a stop gain pkd2 mutation in a family with 3 PKD-affected individuals. the proband has aortic dissection, but it is not clear whether the pkd2 carriers in the family have been phenotyped for aortic dissection.

Whether pkd2 has anything to do with aortic dissection in this family is unclear. Currently, as stated, only 1 out of 3 individuals in this family with the pkd2 mutation have aortic dissection.

What about other genes for aortic dissection? it appears as though no other variants were identified from exome sequencing of the proband.

What was father's creatinine?
How was rare and pathogenic defined. Did they functionally assess all variants?
What about aortic dissection in other individuals in the family, including the probands mother?
Has she had Thoracic CT?

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.
Yes

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.
Yes
Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.
No

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.
Not relevant to this manuscript

Quality of written English
Please indicate the quality of language in the manuscript:
Acceptable

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