Reviewer's report

Title: Clinical utility of PKD2 mutation testing in a polycystic kidney disease cohort attending a specialist nephrology out-patient clinic.

Version: 2 Date: 13 March 2012

Reviewer: Carsten Bergmann

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Robinson and colleagues report mutation analysis of PKD2 in a clinically well-evaluated cohort of 142 patients with ADPKD and CDK stage 4 or less. Overall, this is a well-done study. Although ADPKD mutation analysis is incomplete and most aspects and results have been already discussed in the literature, studies like this are important and worth publishing. However, there are a few points that the authors are recommended to address and which may make the paper more robust.

- While there is no doubt on the usually milder clinical course of PKD2 compared to PKD1, the authors are asked to shortly comment on cases with early and severe disease manifestation.
- Page 7, 1st paragraph, balanced translocation: Please spend a few more words on the family history etc.
- Page 8, 1st paragraph: Please explain if also for these different (sub)cohorts age and other parameters were comparable?
- Page 10, 3rd paragraph, “not aware of a FH of ADPKD”: Please also discuss here other ways of inheritance and that PKD genetics becomes more complex.
- Overall, some statements should be more carefully (e.g., “patients with a FH of ESRF before age 50 can be excluded from PKD2 testing”).

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

I am an employer of Bioscientia as a member of Sonic Healthcare, but do not hold any stocks or shares.