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: 20 March 2012

Reviewer: Curie Ahn

Reviewer's report:

• Major Compulsory Revisions

1) Although it is a good idea to screen PKD2 mutation in the suspected ADPKD patients, the predictive value of FH of ESRF seems more economical way to clinically predict patients’ outcome. You should clarify this point in your discussion.

2) What is the novel finding of your study? We already know patients with PKD2 mutation have slow disease progression and low number of cysts. You should clarify the strength of your study in your discussion section.

3) The aim of this study was to identify factors that can be used to offer targeted gene testing and to provide patients with improved prognostic information. You concluded that PKD2 mutation testing can be useful to all but those whose relatives have developed ESRF before the sixth decade. However, even without PKD2 mutation analysis, we already know from the previous study that patients with FH of ESRF <50 more likely to have PKD1 mutation and FH of ESRF >70 more likely to have PKD2 mutation. What is the additional role of PKD2 mutation analysis?

4) Your results demonstrated that there was no difference in progression rate from CKD 3 between PKD2 and non-PKD2 groups. As you may know, the initial GFR or renal function is the main contributing factor of later renal function progression. In addition, the amount of albuminuria also affects renal function deterioration. Therefore, I recommend to analyze the result in a subgroup divided by the initial GFR. Also, when you perform the statistical analyses, you should adjust the model for albuminuria along with gender, age, hypertension.

• Minor Essential Revision

1) In the method section, it is better to describe the median or mean follow-up duration of your cohort.

2) It is better to mention the limitation of your study (small number of patients, retrospective study, etc.) in the discussion section.

3) Figure legends should describe the contents of the figures. Please add the description of each figure in your figure legends.

4) There are some typos in the title of tables. Please recheck they have the same format ‘Table O. Title.’ For example, Supplementary Table 1 has description rather than the title. Supplementary Table 2 does not have the title.
• Discretionary Revisions
None.

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

**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.