Reviewer's report

Title: Molecular Testing for Adult Type Alport Syndrome

Version: 1 Date: 8 June 2009

Reviewer: jie ding

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Major Compulsory Revisions

1. The major concerns on this method are the possibility of detecting other mutations on exons 49, 50 and 51, which definitely exist in other patients of Alport syndrome not from these 3 families. It is no problem to detect mutations R1677Q, L1649R and C1564S according to the established experimental conditions by identifying the expected melting peak(s). Thus the significance of the assay described in this manuscript will be diminished. And, the assay can not “help elucidate otherwise unexplained hematuria or chronic kidney disease in men, and unexplained hematuria in woman with a family history of chronic kidney disease.”

2. In addition, even for the majority of adult type AS in the US, this assay dose not display more advantages compared with the method of conventional PCR and direct sequencing.

Minor Essential Revisions

1. There is a mis-label on melting peaks of R1677Q in Figure 1. Labels for the male and female were reversed.

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.