Reviewer's report

Title: Whole exome sequencing reveals novel COL4A3 and COL4A4 mutations and resolves diagnosis in Chinese families with kidney disease

Version: 2 Date: 7 August 2014

Reviewer: Judy Savige

Reviewer's report:

Major changes
1. Most Pro to Leu changes are NOT pathogenic so there needs to be more vigorous discussion of this point.
2. There should be some more discussion of the ability of this technique to pick up compound heterozygotes eg COL4A5/NPHS2.
3. It would also be good to have some discussion of comparison of WES with a 'panel' capture assay.

Minor changes
1. The EM is not very supportive of the diagnosis of Alport syndrome.
2. What was the significance of the CUB variant?

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:

None