Reviewer’s report

Title: Molecular Testing for Adult Type Alport Syndrome

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Reviewer: Kandai Nozu

Reviewer’s report:

This report describes a molecular assay using a set of hybridization probes that identify the three mutations in the COL4A5 gene; C1564S, L1649R, and R1677Q. The system has a perfect sensitivity and specificity for the mutation screening, however, this system can detect only 3 mutations which may be common in US Alport syndrome patients because of the founder effects. Although this system may easily screen the patients with these mutations, this system has a severe limitation because there are no hot spots of mutations in the COL4A5 gene in previous reports. Moreover, about 15% of Alport syndrome patients are sporadic.

The authors described the L1649R mutation can account for 50% of adult type XLAS in the US. However, there are no reports to show this data correct. I can’t believe this mutation is so common in US population. The authors must show the data in this report.

As I described above, COL4A5 has no hot mutational spot and comparatively easy to cause mutations distributing all around this gene. Therefore, this mutational screening system is not adapted for COL4A5 screening.

Level of interest: An article of limited interest

Quality of written English: Acceptable

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