Reviewer’s report

Title: Phenotype variability in a large Spanish family with Alport syndrome associated with novel mutations in COL4A3 gene

Version: 0 Date: 29 Nov 2016

Reviewer: Kandai Nozu

Reviewer's report:

The authors conducted genetic analysis for a large family with autosomal dominant Alport syndrome (ADAS) with a novel mutation in COL4A3 gene. They claim that the existence of non-pathogenic variants on another allele of the COL4A3 gene can affect the severities of ADAS phenotype in this family members. I strongly agree some additional gene variants including COL4- genes can affect the clinical presentation in ADAS and will be of interest. However, in this paper, this is shown in only one family members and investigated only for COL4A3 and COL4A4. The authors didn’t conduct any basic approach either. Therefore, this is just the case report and not the research article. I believe this paper should be published elsewhere, but not as a research article.

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Yes

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Unable to assess

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

No

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

Not relevant to this manuscript
Quality of written English
Please indicate the quality of language in the manuscript:

Needs some language corrections before being published

Declaration of competing interests
Please complete a declaration of competing interests, considering the following questions:

1. Have you in the past five years received reimbursements, fees, funding, or salary from an organisation that may in any way gain or lose financially from the publication of this manuscript, either now or in the future?

2. Do you hold any stocks or shares in an organisation that may in any way gain or lose financially from the publication of this manuscript, either now or in the future?

3. Do you hold or are you currently applying for any patents relating to the content of the manuscript?

4. Have you received reimbursements, fees, funding, or salary from an organization that holds or has applied for patents relating to the content of the manuscript?

5. Do you have any other financial competing interests?

6. Do you have any non-financial competing interests in relation to this paper?

If you can answer no to all of the above, write 'I declare that I have no competing interests' below. If your reply is yes to any, please give details below.

I declare that I have no competing interests.

I agree to the open peer review policy of the journal. I understand that my name will be included on my report to the authors and, if the manuscript is accepted for publication, my named report including any attachments I upload will be posted on the website along with the authors' responses. I agree for my report to be made available under an Open Access Creative Commons CC-BY license (http://creativecommons.org/licenses/by/4.0/). I understand that any comments which I do not wish to be included in my named report can be included as confidential comments to the editors, which will not be published.

I agree to the open peer review policy of the journal