Reviewer’s report

Title: A novel compound heterozygous variant identified in GLDC gene in a Chinese family with non-ketotic hyperglycinemia

Version: 2 Date: 07 Nov 2017

Reviewer: Elaine B Spector-Christensen

Reviewer's report:

This manuscript reports the results of sequencing and deletion/duplication testing in a family with three siblings affected with NKH. The methods used were appropriate. It is interesting that deletions occur in so many different ethnic populations. What are the approximate chromosomal locations of the beginning and end of the deletion in exon 3? I do not recall if this information is available from the MLPA kit that was utilized. Was a DNA sample with a known deletion used for a control for the MLPA? The ACMG/AMP 2015 Standards and Guidelines for the interpretation of sequence variants strongly discourages the use of the word mutation. It should be substituted by sequence variant or pathogenic variant etc. I will leave this up to the editors. Please use the 3 letter amino acid code when describing the sequence variant. Some editing of the manuscript is still needed.

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.

Yes

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

Yes

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.

I am able to assess the statistics
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 organisation 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