Reviewer's report

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

Version: 1 Date: 08 Sep 2017

Reviewer: Shagun Aggarwal

Reviewer's report:

The manuscript reports a Chinese family with NKH.

The following minor revisions need to be considered before it can be considered for publication:

Grammatical errors need to be corrected in multiple places preferably by a native English speaker.

Some information can be presented in a more concise manner eg. mutation pathogenicity prediction can be summarised in a table.

Overall the manuscript would benefit from editing.

Did the authors perform sequencing of all three genes followed by MLPA of GLDC? Logically, since GLDC mutations are predominant, it would make sense to do Sanger for GLDC, and as one mutation was identified followed by MLPA of GLDC to look for CNVs.

The authors need to clarify this in methods section, and suggest the best approach to molecular testing of NKH.
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.

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

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