Reviewer's report

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

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Reviewer: Aneek Das Bhowmik

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Manuscript number: MGTC-D-17-00190R1

Title: A novel compound heterozygous mutation identified in GLDC gene in a Chinese family with nonketotic hyperglycinemia

Comment #1: Authors reported compound heterozygous variants in GLDC gene which includes a novel missense variant c.2680A>G and a previously reported exon 3 deletion, in a Chinese family affected with Non-ketotic hyperglycinemia (NKH). Importantly, the phenotype is consistent with mutations in GLDC gene and further delineates the mutation spectrum of this gene in MKH. The findings of the paper are well described however text (method and result sections) should be shortened and reorganized to make it more compact. The database and in silico analytical tools information and outcomes can be summarized in tabular forms and provided as a supplementary file. Figures 1, 2 and 3 should be clubbed up into one figure.

Comment #2: Page 4, 2nd line statement "However, few studies refer to the genetic profile for NKH patients in China have been reported." should be supported with appropriate references.

Comment #3: Page 5, Quantitative PCR (Q-PCR) section: The methods should be described elaborately for Q-PCR (SYBR green and other reagent details if any). What is the reference gene used for normalization in 2-ΔΔCT calculation?

Comment #4: If MRI pictures are available for the patients authors should provide that in the manuscript by clubbing up with figures 1, 2 and 3. If not available that should be clearly mentioned in the 3.1 Clinical data and auxiliary examination" subsection under "Results".
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

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Please indicate the quality of language in the manuscript:
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