Reviewer’s report

Title: Identification of two novel null variants of CLN8 by targeted next-generation sequencing: first report of a Chinese patient with neuronal ceroid lipofuscinosis due to CLN8 variants

Version: 1 Date: 19 Jul 2017

Reviewer: Manoj Menezes

Reviewer's report:
I have only 2 comments on the revised manuscript

1. There are still grammatical errors that need correction.
2. As stated by the authors, there are no specific NCL genetic variant databases for Chinese patients, and we all accept that update of disease-specific databases is suboptimal. Other Chinese patients with NCL mutations may not have been reported. Hence I would still prefer statements like variants in CLN8 have never been 'identified' (page 3), first NCL patient due to CLN8 (page 3), first Chinese NCL patient (page 7) and second Asian NCL patient, to be all qualified with the with 'reported' to have mutations in CLN8'.

I think that the evidence from a small group of previously reported patients with CLN8 and individual in this report that null variants cause earlier onset and progressive disease, and this being the first reported Chinese patient, both add value to this manuscript.

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.
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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