**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:** 13 Jul 2017

**Reviewer:** Conceicao Bettencourt

**Reviewer's report:**

The manuscript by Gao and colleagues has been substantially improved since the last submitted version, and they have addressed satisfactorily my previous comments. There are still minor issues though, including:

1. Further English revision is still needed, e.g. in the abstract where the authors say "The most variants were..." it should be "Most of the variants were..."; in the results where the authors say "Among the remained 16 rare SNVs..." it should be "Among the remaining 16 rare SNVs...", and where it is"...which was coincident with the recessive inherited model..." it should be "...which was coincident with the recessive inheritance model..."; in the discussions "The variants carrying in the patient..." it should be "The variants carried by the patient..."; the authors should mention "variants in CLN8" not "variants of CLN8" throughout the manuscript; and correct other minor things along the text.

2. There seem to be some incomplete sentences, e.g. in the first sentence of the background when the authors say "...cognitive decline, loss of motor and vision,...", what do they mean by loss of motor? In the discussion (end of second paragraph) when the authors say "...and his cognitive and vision was progressive deteriorated...", what do they mean by his cognitive was progressive deteriorated? Did they mean "...his cognitive function and vision have progressively deteriorated..."?

3. In the results, when the authors refer to "compound variants" they should say instead "compound heterozygous variants".
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.
No

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Not relevant to this manuscript

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