Reviewer’s report

Title: Novel PYGL mutations in Chinese children leading to glycogen storage disease type VI: two case reports

Version: 2 Date: 16 Jan 2020

Reviewer: Andreas Janecke

Reviewer's report:

The detailed clinical findings in the identified two patients with GSD VI are the most interesting and worth reporting.

I do not accept calling that one patient has a "milder" form disease, based on previous comments. This comparision is not necessary. Reporting the different patient disease courses per se is fine and CONTRIBUTE TO THE DELINEATION OF THIS RARE DISORDER which is helpful for the clinicians.

Still, there is the unnecessary mentioning of earlier testing (panel testing) and comparision.

WES is an well-established method for SNV and CNV detection; there is too much emphasis in the manuscript on this old method, included the mentioning of WES in the conclusion of the abstract (this is not new enough).

In my opinion, all the patient details reported in the comments to the reviewers should be carefully included into the manuscript, and more general information should be dismissed, as the discussion is much too long.

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:

Acceptable

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