**Reviewer’s report**

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

**Version:** 2  **Date:** 28 Jan 2020

**Reviewer:** David A Weinstein

**Reviewer's report:**

The revised manuscript has improved. While the novelty of the manuscript remains in question, there are still some important points that are worth publishing due to the paucity of literature for GSD type VI.

**Important points:**

1. GSD VI is present in China, and the condition may be under-diagnosed. The fact that the patients come from different regions makes it even more likely that there is unrecognized GSD VI in China, and this discussion could be improved.

2. Patients with GSD VI respond very well to treatment with improved growth and laboratory improvement. This is an important point, and adds to the literature describing the impact of treatment. A table showing laboratory values at diagnosis and after treatment would strengthen this point.

3. The argument that the patients have a different phenotype is not convincing. I would recommend moving away from this discussion since it is not critical. The authors continue to ignore the abnormal hepatic transaminases in patient 1, and the elevated lactate was also not commented on. High lactate concentrations are not typically seen in GSD VI with fasting. The difference in growth at presentation could simply be due to the duration without treatment. With worse LFTs, comparable triglycerides, and similar response to treatment when compared with patient 2, it is difficult to state that the patients have a different phenotype. A genotype-phenotype correlation cannot be made based on one subject, and I would strongly recommend removing the discussion that patient 1 is mild. In this vain, the title should also be changed.

**Minor Comments:**

4. The authors state on page 6, line 52 that the AST normalized for patient 2. What about ALT?

5. The table should include CK concentrations if they were checked.

**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

**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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