Author’s response to reviews

Title: Clinical course, mutations and its functional characteristics of infantile-onset Pompe disease in Thailand

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Dear the editor of BMC Medical Genetics

Re: MGTC-D-18-00480R1

Thank you very much for giving us opportunity for polishing and fixing our mistake for the final manuscript entitled “Clinical course, mutations and its functional characteristics of infantile-
onset Pompe disease in Thailand”. In the following pages are point-by-point responses to each of the comments raised by the editor/reviewers.

In addition, we update the clinical information of patient 2 and 12 since they it is now 8 months since the beginning of initial submission of this manuscript. This information appears on We are appreciated your consideration and look forward to receiving your final decision soon.

Sincerely yours,
Duangrurdee Wattanasirichaigoon, MD
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Point-by-point Response

Editor Comments:

1. Overlap

We note that the current submission contains some textual overlap with other previously published works, in particular:


Since the overlap is contained within the Methods section, and rephrasing the methods would not be conducive to future reproducibility of your study, we ask that, for these sections of textual overlap, you provide a summarizing statement explaining this methodology has been published previously and provide the attribution to the sources mentioned above. For instance, at the start of the "Functional analysis by expression in COS-7 cells and Western blot analysis" paragraph, you could write: “We used the methodology previously described by ________”

Alternatively, please re-phrase these sections to minimize overlap.

Response. We total appreciated for the editorial team for checking this out and we are so sorry for this mistake. We have shortened the methods and referred to the detailed methods/techniques used as published earlier (Ref. Annals of human genetics, 82(3), pp.150-157).
Most of the changes are in the subsections: Leukocyte preparation and enzyme assay (page 7, line 153), Functional analysis by expression in COS-7 cells and construction of vectors for GAA mutants and expression in COS-7 cells (page 8, line 180), and Western blot analysis (page 8, line 183).

2. Consent to participate

In your “Ethical Approval and Consent to Participate” section of your Declarations, please confirm whether informed consent, written or verbal, was obtained from all participants and clearly state it in this section. If verbal, please state the reason and whether the ethics committee approved this procedure. If the need for consent was waived by an IRB or is deemed unnecessary according to national regulations, please clearly state this, including the name of the IRB or a reference to the relevant legislation.

In the case of this study which involved minors please confirm whether consent written or verbal was obtained from the parents/guardians of the participants of this study.

Response. We clearly stated that the written inform consent were obtained for each participants.

‘Written informed consents were obtained from all participants and the parents of the minor participants, following the Ramathibodi Hospital Institutional Review Board (protocol ID 06-55-46) (page 20, lines 475-477).’

3. Author contributions

We note the initials PS are given twice for the same contribution (i.e. in the same sentence). Please ensure this is corrected.

We also note that authors AK and SP are missing from this section. The individual contributions of all authors to the manuscript should be specified in the Authors’ Contributions section. Guidance and criteria for authorship can be found here:

http://www.biomedcentral.com/submissions/editorial-policies#authorship

Response. Our apology for this simple mistake.

The second ‘PS’ was a typo error. It is replaced by ‘AK’

‘AK’ had role in performing experiments.
‘SP’ is already appeared in the text and was involved in clinical data collection and interpretation.

The roles in performing experiments are now clearly elaborated for each coauthor (page 21, lines 493-499).

4. Declarations heading

Please provide the heading "Declarations" above the corresponding section.

Response. The heading "Declarations" is added above the corresponding section (page 20, line 473).

5. Funding

In the Funding section, please also describe the role of the funding body in the design of the study and collection, analysis, and interpretation of data and in writing the manuscript.

Response. We have added the statement about the role of funding body.

‘The funding body had no influence in the design of the study and collection, analysis, and interpretation of data and in writing the manuscript (page 20, lines 487-491 )’.

6. Genetic data

As part of our editorial policies we strongly encourage all authors to deposit their sequencing data before publication. Please deposit the sequencing data generated in this study in a suitable public repository such as the NCBI SRA database. Once you have deposited your data, ensure you provide the information on deposition and how to access these data in the ‘Availability of Data and Materials’ section in Declarations, including the permanent link or the unique identifier associated to it.

Response. Regarding newly identified variants (5 pathogenic and 2 benign) we are in the process of registering into ClinVar. However, as the first time registration for a new institute it takes a week at least before we can get approval and access for submitting variants. This may prohibit us from completing the resubmission of the manuscript by the deadline which is June 20th. Therefore, may I ask that we submit the revised manuscript first and add ClinVar submission ID later as earlier as possible. Table 3 is our target site to add the ID. However, if the editor/reviewer may have other advice, we would appreciate your suggestion.
7. Clean manuscript

At this stage, please upload your manuscript as a single, final, clean version that does not contain any tracked changes, comments, highlights, strikethroughs or text in different colours.

All relevant tables/figures/additional files should also be clean versions. Figures (and additional files) should remain uploaded as separate files.

Please remove any files that should not be published along with your manuscript e.g. cover letters, reviewer responses, guidelines etc., ready for publication.

Response. We make sure that the manuscript main body and tables are now clean.

Figures and supplemental data are uploaded in separate files.

Cover letter and response to reviewer are not uploaded. Only the content is provided in the specific section during the submission process.

8. Additional information

In addition, we update the clinical information of patients 2 and 12 since they are now 8 months older than when describe in the initial manuscript. This information appears on (page 11, line 258 and line 267) and Table 2, column ERT/Clinical outcome. These data do not affect the interpretation of clinical data and discussion.