Author’s response to reviews

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

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Author’s response to reviews:

Dear the editor of BMC Medical Genetics

Re: MGTC-D-18-00480R3

“Clinical course, mutations and its functional characteristics of infantile-onset Pompe disease in Thailand”
Thank you very much for the suggestion from the Editor. We finally come up with the modification of the data presented to increase the level of the patient/participant anonymity while keeping the scientific information unaffected/minimally affected. We delete the gender of the patients and provide age range at onset and at diagnosis instead of the exact age, as now shown in Table 2. We also made minor changes (related to the age data) in the result section accordingly. The detail of response is shown in the following page.

Again, we are greatly appreciated your thoughtful comments and giving us opportunity to edit the manuscript.

Sincerely yours,
Duangrurdee Wattanasirichaigoon, MD

Point-by-point Response

Editor Comments:

1. Identifying information

Currently, the age and gender information present in Table 2, in combination with other identifiers, may compromise patient/participant anonymity.

As you do not have consent to publish, please amend the file to address this issue, by removing this information or by providing ages as age-ranges.

Responses:

1) The gender of each patient is removed. The exact age at onset and at diagnosis are replaced by age ranges, as now shown in Table 2.

2) Since the gender of each patient is removed, we therefore provide aggregate data about the gender of the patient in this cohort in the beginning part of the result section as follow ‘Twelve patients (9 males and 3 females) with IOPD were included in the study (Table 2)’ (Results/Line 234-235/Page 10).

3) Result section

page 10, line 238-240: ‘Patient 2 was noted at age 15 days prompting referral to our hospital at 23 days due to a positive history of two previous siblings affected with cardiomyopathy of
unclear etiology.’ is replaced by ‘Patient 2 was noted to have mild tachypnea at two weeks, prompting referral to our hospital at 3-4 weeks of age due to a positive history of two previous siblings affected with cardiomyopathy of unclear etiology’ (Results/Line 238-240/Page 10).