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-18-00480R4 “Clinical course, mutations and its functional characteristics of infantile-onset Pompe disease in Thailand”
We have edited the manuscript to improve the level of anonymity as much as possible. We removed individual ethnic data from Table 2 and provide an aggregate data instead. All the changes are provided in the following page.

Regarding the consent for publication of individual information, we were able to obtain the consent from some families especially those whose individual data are discussed. We have added the statement detailing that written informed consent to publish was obtained, into the Consent for publication section.

We are glad to inform the editors that the ClinVar ID provided in Table 3 is now accessible online.

Again, we totally appreciate your insightful advices to help us in resolving this difficult ethical issues. My colleagues and I have learnt much from the submission of this manuscript and would like to express our sincere gratefulness to you again. We are prepared for additional comments, if there is any.

Sincerely yours,

Duangrurdee Wattanasirichaigoon, MD

List of changes

1) IRB Protocol ID is added in the method section
   ‘…. (protocol ID 06-55-46).’ Page 7, line 151-152

2) Table 2: The column showing individual ethnic data is removed.

3) An aggregate data of ethnic background is provided in the beginning part of the result section.
   ‘Twelve patients including nine males and three females (10 Thai and 2 from other Asian ethnicities) with IOPD were included in the study (Table 2).’ Page 10, line 236-237

4) Discussion section.
Specific ethnicities other than Thai are deleted.

‘We described 12 patients with IOPD, including 10 Thai, one Burmese and one Vietnamese descendants. All patients had typical presentation of IOPD and fatal outcome early in life…’ is changed to ‘We described 12 patients with typical presentation of IOPD and fatal outcome early in life…’ Page 16, line 380-381

5) Other changes

Information are provided by more general term as follow.

‘The first sib, a 6-month old boy’ is changed to ‘The first sib, a 6-month old infant’ page 10, line 243

‘Patient 2 received ERT since the age of 1 month. She was able to sit…’ is changed to ‘Patient 2 received ERT since the age of 1 month. Patient 2 was able to sit…’ page 11, line 259

‘She said a meaningful word…’ is changed to ‘The patient said a meaningful word…’ page 11, line 261

‘she can speak in short sentences…’ is replaced by ‘the patient can speak in short sentences…’ page 11, line 263

‘Her weight and height…’ is replaced by ‘The patient’s weight and height…’ page 11, line 264

‘She never experienced…’ is changed to ‘Patient 2 never experienced…’ page 11, line 265

‘…when she was already on ventilator…’ is replaced by ‘… when the patient was already on ventilator…’ page 16, line 382

6) Consent for publication of individual data

We used BioMed Central format and translated it into Thai language so that the parents can read it. We were able to obtain the consent from some patients.

A statement ‘Written informed consent to publish was obtained from the parents or legal guardians of the participants whose individual data was detailed in the manuscript.’ is added into the Consent for publication section. Page 20, line 481-482

- Consents for publication were obtained from at least 3 families:

Patients 2 and 12, whose detail clinical data are provided much more as compared to the other patients;
Patient 4 who died 8 years ago.

- We definitely failed to locate the current address of 7 families, therefore the consent was not obtained.

Patients 3 and 5 who were from Myanmar and Vietnam, and dies years ago

Patients 6, 7, 8, 9, and 10 died years ago.

- We are still trying to locate 2 other families.

Patient 1 died 6 years ago.

Patient 11 died years ago.