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-18-00480R2

“Clinical course, mutations and its functional characteristics of infantile-onset Pompe disease in Thailand”
Thank you very much for suggesting and giving us enough time for submission the variants identified to variant database. We have submitted those variants to ClinVar and received ClinVar SCV accession number per variant interpretation which are now present in Table 3. We also found that p.Ala261Thr, one of the pathogenic variants identified in the present study, which had not been reported in the literature was in fact listed in ClinVar database as a variant of unknown significance but without citable publication. Therefore, we cite ClinVar for this mutation and remove it from the list of novel variants identified in the present study. Minor changes related to this variant are made accordingly, as follows: Page 3 (line 66, lines 68-69), Page 7 (line 171, lines 172-173), Page 8 (line 190), Page 13 (line 303), Page 20 (lines 485-487) and Figure 2A.

Again, we are greatly appreciated the comments and suggestion from the reviewers and the editor. We look forward to hearing your final consideration.

Sincerely yours,

Duangrurdee Wattanasirichaigoon, MD

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