Reviewer's report

Title: Clinical and GAA gene mutation analysis in mainland Chinese patients with late-onset Pompe disease: identifying c.2238G>C as the most common mutation

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Reviewer: Fernando Scaglia

Reviewer's report:

The authors present the clinical and genetic characteristics of 27 mainland Chinese late-onset Pompe patients from 24 families. Their findings indicate that c.2238G>C (p.W746C) is the most common mutation in mainland Chinese late-onset Pompe patients, as observed in Taiwanese patients. The novel mutations identified in this study expand the genetic spectrum of late-onset Pompe disease, and the prevalence of respiratory dysfunction highlights the importance of monitoring pulmonary function in late-onset Pompe patients.

Major Compulsory Revisions:

Results Section: the authors state that two patients were diagnosed with either abnormal liver function or elevated CK. The authors need to clarify whether by elevated liver function they imply elevated liver transaminases that also may originate in skeletal muscle. Abnormal liver function should not be used in the manuscript.

Discussion: The authors state that muscle biopsy should be considered to be a very useful tool as the diagnosis of Pompe disease can be challenging. However, this could be a point of contention. The authors need to acknowledge that up to 30% of individuals with late onset Pompe disease may not show specific changes.

Minor Essential Revisions: when the authors present data on ethnicity they need to state that Portuguese are Caucasians. They can talk about data presented on individuals of Portuguese descent and the rest can be named: subjects belonging to other Caucasian groups.

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:
I declare that I have no competing interests.