Reviewer’s report

Title: Gaucher disease: Single gene molecular characterization of one-hundred Indian patients reveals novel variants and the most prevalent mutation.

Version: 0 Date: 06 Dec 2018

Reviewer: Mary Anne D. Chiong

Reviewer's report:

Please include all comments for the authors in this box rather than uploading your report as an attachment. Please only upload as attachments annotated versions of manuscripts, graphs, supporting materials or other aspects of your report which cannot be included in a text format. Please overwrite this text when adding your comments to the authors.

Very informative results and commending the comprehensiveness of the study.

1. Grammar, sentence construction and spelling should be improved/checked in the entire manuscript

2. why was the romanian study on mutations specified in the introduction? why not other asian population or other studies from indian ethnic groups?

3. in the methodology, when you say suspected, what were the inclusion criteria? just one manifestation like thrombocytopenia is already eligible for the study? please specifically enumerate your inclusion criteria- ie whether they will still be included even after a negative enzyme or other biochemical assays? or are they included even with normal assyas but with gaucher cells in the bone marrow? because you have patients whom you said had normal beta glucosidase activities

4. you should place the ages and male female ratio not in the methods but in the results.

5. in the results you said moderate splenomegaly, how would you define that? what is mild and severe splenomegaly then?

6. in the molecular analysis, sorry for my ignorance but is there a specific change for the complex C mutation? if yes, what is it?

7. in the discussion how were you able to classify that there 77 type 1, 12 type 2, etc. was it based on clinical manifestations or mutations or both?

8. in the discussion you said that there was a low prevalence of bone diseases in the patients investigated. how low? because this was one of your clinical inclusion criteria -WITH or
Without bone abnormality. so what were the predominant mutations you saw in those with bone abnormality if its not 84GG?

9. you mentioned about modifier genes in the discussion re Leu483Pro manifesting as type 1. have there been any studies before that elucidated these modifier genes for this mutation in relation to the diversity of the clinical manifestations? because it has always been thought as a severe mutations. if studies are existing maybe you should include that in your discussion

**Are the methods appropriate and well described?**
If not, please specify what is required in your comments to the authors.

Yes

**Does the work include the necessary controls?**
If not, please specify which controls are required in your comments to the authors.

No

**Are the conclusions drawn adequately supported by the data shown?**
If not, please explain in your comments to the authors.

Yes

**Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?**
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

I am able to assess the statistics

**Quality of written English**
Please indicate the quality of language in the manuscript:

Not suitable for publication unless extensively edited

**Declaration of competing interests**
Please complete a declaration of competing interests, considering the following questions:

1. Have you in the past five years received reimbursements, fees, funding, or salary from an organisation that may in any way gain or lose financially from the publication of this manuscript, either now or in the future?

2. Do you hold any stocks or shares in an organisation that may in any way gain or lose financially
from the publication of this manuscript, either now or in the future?

3. Do you hold or are you currently applying for any patents relating to the content of the manuscript?

4. Have you received reimbursements, fees, funding, or salary from an organization that holds or has applied for patents relating to the content of the manuscript?

5. Do you have any other financial competing interests?

6. Do you have any non-financial competing interests in relation to this paper?

If you can answer no to all of the above, write 'I declare that I have no competing interests' below. If your reply is yes to any, please give details below.

I declare that I have no competing interests

I agree to the open peer review policy of the journal. I understand that my name will be included on my report to the authors and, if the manuscript is accepted for publication, my named report including any attachments I upload will be posted on the website along with the authors' responses. I agree for my report to be made available under an Open Access Creative Commons CC-BY license (http://creativecommons.org/licenses/by/4.0/). I understand that any comments which I do not wish to be included in my named report can be included as confidential comments to the editors, which will not be published.

I agree to the open peer review policy of the journal