Reviewer’s report

Title: Clinical and whole-exome sequencing findings in two siblings from Hani ethnic minority with congenital glycosylation disorders

Version: 2 Date: 21 Jun 2019

Reviewer: Kamwing Jair

Reviewer's report:

Summary

Zhen Zhang and colleagues reported novel mutations identified in PMM2 and MYH9 from two siblings of Hani ethnic minority population, using whole-exome sequencing. These mutations linked to congenital glycosylation disorders.

Significant

These mutations are novel variation linked to refractory thrombocytopenia and PMM2 from this Hani ethnic minority living in Yunan, China. This finding broadens the spectrum of CDG in Chinese territory. Also, it helps to increase the awareness of this kind of disorder in China among healthcare providers so as to improve their health services by encouraging prenatal diagnosis in Yunnan Province, China.

Weakness

1. Even through number of reads from WES were mentioned, more detail information of how they perform the whole-exome sequencing followed by analytical strategy should be provided including quality and quantity measurement used such as the Q30 Score, filtering parameters, as well as software and its criteria used for bioinformatic analysis.

2. A functional assay, such as an enzymatic measurement of PMM2 gene, could be interesting to show the deficiency of enzymatic activity after mutation, provided that patient samples are available.
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.

Yes

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.

Not relevant to this manuscript

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

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