Reviewer’s report

Title: Case reports: three Novel variants in PCCA and PCCB Genes in Chinese Patients with propionic acidemia

Version: 1 Date: 17 Jan 2020

Reviewer: Dwi Nugrahamaningsih

Reviewer's report:

The manuscript present the new mutation on PCCA and PCCB gene related to propionic acidemia (PA). However, the manuscript should be improved since some information is not clear.

1. Background should explain what is the importance of genetics examination (PCCA and PCCB) for the case management or diagnosis.

2. the technique, material (reagents), software or sequence reference used should be explained more detail in methods section.

3. the discussion:

   a. how the author predict the effect of C.359-360 del AT and c.1398+1G>A mutation in PCCB gene (line 163-169)? please explain or cite the relevant article!

   b. c.1288C>T (p.R430X ) mutation in the PCCA gene, C.359-360 del AT and c.1398+1G>A mutation in PCCB gene also found in either mother ther of the patient. Is there any data regarding clinical or laboratory finding of the parents? if the mutation is related to PA, you might find something from their parent. please explain.

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

No

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

Unable to assess

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:

Needs some language corrections before being published

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