Reviewer’s report

Title: Massive parallel sequencing as a new diagnostic approach for phenylketonuria and tetrahydrobiopterin-deficiency in Thailand

Version: 1 Date: 20 Jul 2017

Reviewer: Dhanya Lakshmi

Reviewer’s report:

Your attempt to use NGS for diagnosis of hyperphenylalaninemia is appreciable.

There are some disparities with the written text and the legend to the figure given.

In the text it is mentioned that patient II had novel mutation at a site which is conserved across species. (Page 9, line 12-14). But in the table it is given that patient 2 has known mutations.

The depth for patient III is very less. This makes the pathogenicity of the novel variants doubtful.

May be this is the reason for the higher incidence of BH4 deficiency in your cohort.

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.

Unable to assess

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

No

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:
Not suitable for publication unless extensively edited

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