Reviewer's report

Title: Spectrum of PAH gene variants among a population of Han Chinese patients with phenylketonuria from Northern China

Version: 1 Date: 30 Jun 2017

Reviewer: Nenad Blau

Reviewer's report:
Gu et al. (PLoS One. 2014 Apr 4;9(4):e94100 did not correctly explain known genetic causes of PKU and BH4 deficiencies. They wrote " ..... six genes (PAH, PTS, GCH1, QDPR, PCBD1 and GFRP) involved in PKU and BH4 deficiency. .....", however there is so far no documented or published case of GFRP deficiency. Thus, I would suggest to modify the sentence on page 3, lines 54-56 to "Hyperphenylalaninemia (HPA) comprises a group of genetically heterogeneous disorders, including deficiencies in PAH, tetrahydrobiopterin (BH4) and DNAJC12 [2], which are associated with six genes (PAH, PTS, GCH1, QDPR, PCBD1, and DNAJC12). Deficiency of the GFRP was so far not reported and it is not clear if it would present with HPA as well."

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?
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