Reviewer's report

Title: Mutations in the PIGW gene associated with hyperphosphatasia and mental retardation syndrome: a case report

Version: 0 Date: 01 May 2018

Reviewer: Sylvie Jaillard

Reviewer's report:

The manuscript reports the identification of a compound hit in the PIGW gene and links this finding to the phenotype HPMRS. The manuscript is easily understandable but some points need to be clarified:

- The variants identified in the present study should be described more in depth. Some databases have been used for filtering, according to these databases, what is the description of the variants: frequency in the general population, pathogenicity...

- They should also been compared to the previous reported variants.

- There is also a lack of functional studies to assess the effects of the variants on the protein. They were performed in the study of Hogrebe et al: Transfection of the mutated allele into Pigw-defective CHO cells indicated impaired enzymatic activity of the mutated PIGW product. Is it possible to realize the same kind of analyses?

- The feature "mental retardation" is not suitable for a 70 day old baby: the author should use psychomotor retardation at this age or intellectual deficiency later.
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.

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.

I am able to assess the statistics

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

Needs some language corrections before being published

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