Reviewer's report

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

Version: 1 Date: 19 Jun 2018

Reviewer: Lydie Burglen

Reviewer's report:

The manuscript has been improved significantly according to the reviewers' comments.

The authors couldn't accede to our request of functional validation of the PIGW variants since they didn't have available samples from the deceased child. However, the data regarding software predictions of pathogenicity and frequency of the variants in databases are now reported and are sufficient together with the clinical correlation, to classify them as probably pathogenic.

The manuscript must be corrected by an English native and a lot of typographic errors are still there (spaces, references, legend of figures ("missen" instead of "missense"), "AKP" changed in "ALP" in some places but not in figures'legend and case checklist, etc.). Several sentences begin by "and" when it is not necessary. The term "variants" replace "mutations" which should be deleted (end of the paragraph "gene mutation analysis").

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:

Needs some language corrections before being published

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