Reviewer’s report

Title: A novel compound heterozygous mutation of the SMARCAL1 gene leading to mild Schimke immune-osseous dysplasia: A case report

Version: 1 Date: 14 Dec 2016

Reviewer: Peter Stenzel

Reviewer's report:

The authors report a novel and important mutation in the SMARCAL1 gene associated with a previously reported mutation. The compound heterozygous patient is well described. The molecular findings are clearly documented. The discussion contains a good analysis.

The English is not acceptable.

References 11 and 14 are redundant.

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:

Not suitable for publication unless extensively edited

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