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: 02 Jan 2017

Reviewer: Dhavendra Kumar

Reviewer's report:

This is a clinical case report of a rare autosomal recessive skeletal dysplasia with immune deficiency.

It is a rare disorder and thus might be suitable for publication. However, following revisions are necessary:

1. The manuscript has considerable English grammar and spellinh errors- this needs detailed review and probably best re-written.

2. Clinical details are scatchy- there are no clinical photographs and X-ray pictures to demonstrate extent of skeletal dysplasia in the case.

3. There is no comparative analysis with other publishes cases

4. Discussion needs to provide clarification on molecular links with skeletal dysplasia and immune deficiency.
Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

No

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

No

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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None known

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