Reviewer's report

Title: A novel SMARCAL1 mutation associated with a mild phenotype of Schimke Immuno-Osseous Dysplasia (SIOD)

Version: 2 Date: 26 January 2014

Reviewer: Encarna Guillen-Navarro

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The authors of this interesting manuscript “A novel SMARCAL1 mutation associated with a mild phenotype of Schimke Immuno-Osseous Dysplasia (SIOD)” report on a clinical case of this genetic disease which significantly expands the phenotypical spectrum of currently known affected patients with SIOD. This paper is also important for nephrologists to raise awareness of this rare disease.

In general, it is an interesting paper which may improve with some minor revisions.

Minor Essential Revisions

In the title: Schimke immuno-osseous displasia instead of osseus
In the abstract:
(SIOD, OMIM #242900) instead of (SIOD, MIM 242900)
-It is said that SIOD is caused by mutations in the gene encoding HepA-related protein (HARP) also known as SMARCAL1 which is correct, but if it is going to be used SMARCAL1 mainly in the body text, I would rather use it in the abstract
In the case presentation
Phenotype description should be completed. It is not mentioned patient’s stature, weight and OFC; at least, stature should be included with the corresponding centile. It is not specified if the patient presents short neck or trunk, hyperpigmented macules, corneal opacities or hypertension and this would be very important in order to evaluate this mild phenotype.

It is said that skeletal radiograph revealed dorsolumbar kyphoscoliosis with unbalanced iliac crests but the usual bone changes are not described. The most commonly observed radiologic abnormalities are ovoid and mildly flattened vertebral bodies, small deformed capital femoral epiphyses, and shallow dysplastic acetabular fossae. ¿were they found in this patient?. This findings are very relevant as a clue for the diagnosis of spondyloepiphysyeal dysplasia

The discussion is correct. May be the importance of recognizing this phenotype keeping in mind the characteristic growth retardation and bone anomalies might be emphasized.

Please, review references, and follow the instructions. Citations in the reference list should include all named authors, up to the first 30 before adding ’et al.’.
Finally, review the language.
In general, it is an interesting paper to publish which may improve with some minor revisions.

**Level of interest:** An article of importance in its field

**Quality of written English:** Needs some language corrections before being published

**Statistical review:** No, the manuscript does not need to be seen by a statistician.

**Declaration of competing interests:**
I declare that I have no competing interests