Author’s response to reviews

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

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Author’s response to reviews: see over
To: Dr. Roser Torra  
Associate Editor  
BMC Nephrology  
Nephrology Department, Inherited Renal Disorders  
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Barcelona, Spain

Dear Dr. Torra,

We are submitting a revised version of the manuscript ID 4813445311078430 entitled "A novel SMARCAL1 mutation associated with a mild phenotype of Schimke Immuno-Osseous Dysplasia (SIOD)" along with a point-by-point answer to the referees’ comments. The manuscript was modified as suggested.  
We thank you for the useful suggestions and we hope to have adequately addressed the points raised during the review process.  
Looking forward to hearing from you.

Best regards,

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Answers to the major referees’ comments

Reviewer #2:

1. In the title: Schimke immuno-osseous displasia instead of osseus
We replaced “osseus” with “osseous” in the title (page 1 line 1), as requested.

2. In the abstract: (SIOD, OMIM #242900) instead of (SIOD, MIM 242900)
We revised the OMIM notation in the abstract (page 2 line 3) and in the background section (page 3 line 2), as requested.

3. 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.
The term “SMARCAL1” replaced the sentence “the gene encoding HepA-related protein (HARP) also known as SMARCAL1” in the abstract (page 2 line 5-6), as requested.

4. 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.
Stature, weight and OFC with related centiles were added in the case presentation (page 4 line 6-7), as requested.

5. 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 spondyloepiphyseal dysplasia
Phenotype description was completed (page 4, lines 9-10 and lines 13-16), as suggested.

6. 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.'.

The references were corrected following the author instructions (pages 10-12)

7. Finally, review the language.

English language was reviewed, as requested.