**Reviewer’s report**

**Title:** Identification of gene mutations in patients with primary periodic paralysis using targeted next-generation sequencing

**Version:** 1  **Date:** 07 Sep 2018

**Reviewer:** Saïd Bendahhou

**Reviewer’s report:**

The manuscript describes a panel of 60 patients with primary PP using the NGS technique for the identification of polymorphisms in 10 chosen ion channel genes and 245 muscular dystrophy- and myopathy-related genes. This strategy led to the identification of novel variants in the SCN4A, CACNA1S, and RYR1 genes. These variants were confirmed by the classical Sanger sequencing.

The major comment:

The authors did conduct any functional study on the novel identified mutations may turn out to be benign polymorphisms. The authors may have to be more cautious describing these mutations as disease-causing mutations unless they provide clear functional data correlating the genotype to the phenotype.

Minors:

Figure 3A: Mutation T704M is mislocated on the cartoon. It is usually shown on the segment S5 close to the intracellular face.

The cartoons (Figures 3 and 4) are of poor quality and could be improved.

Additional files: Table 1 is hard to read (Column titles). Please modify the layout accordingly.

**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:

Acceptable

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