Reviewer’s report

Title: Paroxysmal extreme pain disorder in family with c.3892G>T (p.Val1298Phe) in the SCN9A gene mutation - case report

Version: 2 Date: 29 Mar 2020

Reviewer: Bouchra Ouled Amar Bencheikh

Reviewer's report:

Comment #1
Titles: The sentence is not correct.

cmment#2:
line 59, 156: "missense" mutations to correct and the name of gene is not in italic
The sentence should be corrected: "Seven of them presented clinical manifestation of paroxysmal extreme pain disorder, of which in four were recorded missens mutation mutations in the SCN9A gene (NM_002977.3:c.3892G>T)."
The word "recorded" is not adapted, the authors should use "identified"

Comment #3:
The authors should mention the rs number of the mutation and describe the aminoacid change.

Comment #4:
The authors should add a picture of the chromatogram of the mutation obtained by sanger sequencing with the position of the mutation and the reference allele
In the paragraph: "The genetic test was performed in four family members. All four tests confirmed the heterozygous mutation NM_002977.3:c.3892G>T c.3892G&T; T (p.Val1298Phe) in the SCN9A gene (Fig.5). Genetic test was made with Sanger method"

Comment #5:
In figure 5: authors should use stars or other symbols to highlight individuals with genetic data. The number of patients should appear in the pedigree and mentioned in the manuscript when you describe any patient. for example (patient V.1 and patient VI.4 were investigated....) and the number of patient should appear in the pedigree

Comment #6:
authors should add more details about gene SCN9A and mutations involved in PERD and discuss the value of this case report even the mutation is known.

Comment #7:
line 192: "Symptoms of PEPD occur throughout the entire life of a patient...."
The authors should specify the reference for this sentence or correct the sentence if the "individuals" mean the patients of the family described.
line 181: "Later, a subject remains...". The authors should correct the sentence

Comment #8:
references are still missing for some sentences.
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.

Unable to assess

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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