Author’s response to reviews

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

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Version: 4 Date: 04 May 2020

Author’s response to reviews:

Dear Editor and Reviewers,

Thank you for assessment and any valuable comments regarding presented manuscript. In revised version we took into account most comments from reviewers.

In response to the comments to NURL-D-19-00762R1:

Editor:

   We revised the text for language and grammar issues with a native.

Reviewer 1:

1) We added information about neurological examination, sensory and autonomic function. In all described patients this part of clinical picture was the same. (lines 133-134)
2) We clarified the confusion about „tonic epileptic seizures”, it should be „tonic non-epileptic seizures” from the beginning. (lines 189-191).
3) We revised the text for language and grammar issues with a native.

Reviewer 2:

1) We changed the genes names in Italic and reported gene and mutation according to HGVS recommendations. (all text)
2) We added informations from CLINVAR Database. (lines 153-154)
3) We added information about method of genetic test. (line 147)
4) We do not have access to the details of the genetic test. We only have the result of this tests.
5) Pedigree was changed according to the comments. (figure 1)
6) We added more details about mutations involved in PEPD. (lines 176-181)
7) Information that the mutations have been described in PEPD has been added. (lines 176-178)
8) We changed this lines due to the comments. (lines 185-186)
9) We changed this lines due to the comments. (lines 166-170)
10) We clarified this part of manuscript. (lines 181-185)
11) We changed nomenclature to clarify this point. (lines 187, 201-205)
12) We checked and added references where it was necessary.
In response to the comments to NURL-D-19-00762R2:

Editor:
We revised the text for language and grammar issues with a native.
Reviewer 1:
We revised the text for language and grammar issues with a native.
Reviewer 2:
1) In our opinion, the title should remain in this form.
2) We changed this lines due to the comments. (lines 58-60)
3) Rs number was added. (line 160)
4) Unfortunately we do not have access to the details of the genetic test. We only have the result of this tests, so we can not add picture of the chromatogram.
5) Individuals with genetic data is highlighted on the pedigree.
6) „Discussion and conclusions” part of the manuscript contains information about the gene SCN9A and mutations involved in PEPD.
7) Sentences were corrected where it was necessary.
8) We checked and added references where it was necessary.

In response to the comments to NURL-D-19-00762R3:
1) Figures names were corrected.
2) Revised manuscript is a final form for publication.

We hope that the improved version will meet your expectations.

Kind regards,
Daria Sałacińska