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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Author’s response to reviews:

Dear Editor and Reviewers,

Thank you for valuable comments regarding presented manuscript. In revised version we took into account most comments from reviewers. We also revised the text for language and grammar.

In response to comments of the Reviewer 1 we added information about neurological examination, sensory and autonomic function. In all described patients this part of clinical picture was the same. We clarified the confusion about „tonic epileptic seizures”, it should be „tonic non-epileptic seizures” from the beginning.

In response to comments of Reviewer 2:
1) We changed the genes names in Italic and reported gene and mutation according to HGVS recommendations.
2) We added informations from CLINVAR Database.
3) We added information about method of genetic test.
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.
6) We added more details about mutations involved in PEPD.
7) Information that the mutations have been described in PEPD has been added.
8 i 9) We changed this lines due to the comments.
10) We clarified this part of manuscript.
11) We changed nomenclature to clarify this point.
12) We checked and added references where it was necessary.

We hope that the revised version meets the expectations of reviewers.

Kind regards,
Daria Sałacińska