Reviewer’s report

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

Version: 1 Date: 01 Dec 2019

Reviewer: Maria Nolano

Reviewer's report:

The authors described the clinical features of four out of seven members affected by PEPD from a family of 22 individuals. In the four individuals described, PEPD was linked to a known mutation in SCN9A (c.3892G>T, p.Val1298Phe).

PEPD is a rare disorder due to genetically determined Nav1.7 channel dysfunction. Due to its rarity it is important to gain information on the onset, on the disease course, on the most important clinical features, on the possible treatments as described in this paper. However there are some criticisms:
1) This is a condition characterized mainly by sensory and autonomic symptoms. The authors should make an effort to provide more information on neurological examination and on the assessment of sensory and autonomic function in these patients.
2) They described as normal the EEG as well as MRI and hematochemical analysis in these patients. However in the general clinical description of PEPD they mentioned "tonic epileptic seizures" among the other clinical features. This may be confounding since previous reports described episodes of tonic non-epileptic seizure in patients with PEPD. The authors should clarify this aspect.
3) The text should be revised for language and grammar.

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.

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:

Needs some language corrections before being published

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