Reviewer’s report

Title: SCN8A mutations in Chinese patients with early onset epileptic encephalopathy and benign infantile seizures

Version: 0 Date: 01 Jul 2017

Reviewer: Aneek Das Bhowmik

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Manuscript number: MGTC-D-17-00139

Title: SCN8A mutations in Chinese patients with early onset epileptic encephalopathy and benign infantile seizures

Comment #1: The findings of the paper are well described by the authors in relevance to the different phenotype and mutation spectrum of SCN8A related neurodevelopmental disorders. Whole exome sequencing and targeted gene panel sequencing approaches were used to identify the mutations. Authors can include a table mentioning the different filtering steps used to shortlist the candidate gene variants from exome data. This will bring more clarity to the results described by the author.

Comment #2: Among the 178 sporadic cases studied, only in six patients authors found pathogenic variants in SCN8A gene. Naturally this will raise questions/curiosity in readers mind about the findings in the rest of the large number of patients where no SCN8A variants were observed. Authors can include few lines in brief by addressing this point. This will also strengthen the justifications made by the authors in support of their findings.

Comment #3: The 'Clinical and neurophysiological findings' part describing the phenotype details of the patients should come under 'Methods' section instead of 'Results'.

Comment #4: The manuscript needs some language correction. Some examples of poor sentences are, Page 7, Section- Results, First Paragraph, "Among these candidate genes, SCN8A deserves the most attention."; Page 8, Section- Results, First Paragraph, "Hence, we inferred the variants had low pathogenic possibility."; Page 12, Section- Discussion, First Paragraph, "De novo missense mutations of SCN8A were defined as EIEE13." etc.

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:

Needs some language corrections before being published

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