Reviewer’s report

Title: Clinical Characteristics and Mutation Spectrum of Neurofibromin 1 in 12 Chinese Families with Orbital/Periorbital Plexiform Neurofibromatosis Type 1

Version: 1 Date: 27 May 2019

Reviewer: Ashwin Dalal

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Title:

Clinical Characteristics and Mutation Spectrum of NF1 in 12 Chinese Families with 3 Orbital/Periorbital Plexiform Neurofibromatosis Type 1

Comments:

The authors have carried out a retrospective genetic analysis in 12 Neurofibromatosis families of Chinese origin with 26 family members, indicating earlier age at onset in successive generations.

The study revealed fourteen different NF1 mutations were identified. Eleven (91.7%) OPPN families presented NF1 mutations, and 7 identified mutations were novel. Nine (64.3%) out of the fourteen identified mutations were distributed in exons, three (21.4%) were found on splicing sites and one (7%) was found in an intron.

Comments:

1. Although there is nothing new about mutation analysis in NF1, it is interesting to see the clinical characterization of a subtype of NF1

2. There are many grammatical errors. Please check for the same.

3. Correct the grammar of this sentence on line number 19 in Discussion section"This is might due to the difference of age" - change to This might be due to the difference of age.

4. The conclusion regarding anticipation is far fetched and it is better if the authors do not draw such a conclusion without enough proof. Did the authors find any particular mutation which contributed more towards the anticipation or severity of the disease in successive generation?

4. Any genotype-phenotype correlation possible with available data.
5. Data needs to be provided about presence or absence of same/different mutation in parents along with zygosity.

6. Better resolution figures should be provided

**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.

Unable to assess

**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.

I am able to assess the statistics

**Quality of written English**
Please indicate the quality of language in the manuscript:

Needs some language corrections before being published

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