Title: Whole-exome sequencing identifies homozygous mutation in TTI2 in a child with primary microcephaly: a case report

Version: 0 Date: 15 Jan 2020

Reviewer: Taku Hatano

Reviewer's report:

This manuscript by Picher-Martel et al. reported the case with primary microcephaly due to novel TTI2 mutations. The patient showed microcephaly, short stature, severe speech delay, dysmorphic features, strabismus and dyskinesia. The authors confirmed cosegregation intra family. I feel that the paper is well written.

Minor

1. If possible, the authors show the results of predicting functional effect of the p.D317V mutation.

2. References of supplementary material should be separated from main manuscript.

3. The patient showed dyskinetic movement disorders. The authors should be more precisely stated. Choreic movements?, athetosis?, stereotypies?, Tremor?

4. Figure should be numbered in the order shown

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.

I am able to assess the statistics
Quality of written English
Please indicate the quality of language in the manuscript:

Acceptable

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