Reviewer’s report

Title: Disruption of chromatin organisation causes MEF2C gene overexpression in intellectual disability: a case report.

Version: 0 Date: 28 Feb 2019

Reviewer: Anna Hackett

Reviewer's report:

This paper is interesting, but needs some major revisions.

These include
1. The English is substandard. It is necessary that the authors ask a native English speaker to edit the paper, for sentence construction, present and past tense and readability.
2. FISH or other studies should be done to confirm that MEF2C and LINC01266 genes are in the same TAD.
3. It is not clear from the paper what the hypothesis is for the increase in MEF2C expression - Is this due to a loss of regulatory elements or loss of the wild type TAD boundaries? Perhaps a diagram with a proposed model for pathogenicity could be included.

Minor comments:
1. In the abstract, spell out what TAD stands for
2. Line 74, it is not clear who the "half-sister from the same father" is. Needs to be clarified or include a pedigree.
3. Line 81 - "language deficiency word association at five years" - needs clarification. Was this a conclusion from a particular IQ / developmental assessment?
4. Line 95 - please expalin what the significance of a micro-homology of 3 nucleotides (TGC) is and how it is relevant.
5. Line 97-98. needs clarification that this is the predicted TAD. If this has been demonstrated, it needs to be more clearly spelt out.
6. Line 108-110. References needed for this sentence 
Line 118. reference needed in relation to patients with CNTN6.

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.

Yes

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

No

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:

Not suitable for publication unless extensively edited

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