Reviewer’s report

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

Version: 0 Date: 30 Mar 2019

Reviewer: Weimin Bi

Reviewer's report:

This manuscript reported a patient with a balanced translocation with one breakpoint located upstream of the MEF2C gene and further RNA studies showed the MEF2C gene expression was increased in a lymphoblastoid cell line. Some comments are listed below.

1. Translocations with breakpoints upstream of MEF2C have been reported in the literature. Some of these patients had reduced MEF2C expression. There is a complex neuronal enhancer network upstream MEF2C (PMID: 30445463). I suggest to discuss why expression level was increased in this patient while decreased in some reported patients. Comparing the breakpoint of this patient with the other reported patients may be helpful.

2. Please clarify which TAD identification tool was used in this study. Different TAD identification tool may produce quite different TAD. It is better to combine the Suppl. Fig.1 into Figure 1A so it is easier to compare the changed TAD with the original TAD in these regions.

3. For the RNA studies, it is unclear what the Y axis stands for. Are these controls used in this study gender- and age- matched with the patient?

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.
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.

Not relevant to this manuscript

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

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