Reviewer’s report

Title: NAA10 dysfunction with normal NatA-complex activity in a girl with non-syndromic ID and a de novo NAA10 p.(V111G) variant - a case report

Version: 1 Date: 08 Feb 2018

Reviewer: John Moffett

Reviewer’s report:

The manuscript "A case report of NAA10 dysfunction with normal NatA-complex activity in a girl with non-syndromic ID and a de novo NAA10 p.(V111G) variant" by McTiernan et al. describes a case study of a patient with a single missense mutation in the NAA10 gene and studies into the effects of this mutation on the protein's function. The study is well done and no serious issues were found.

In figures 2-4, error bars are included in the graphs, but no statistical calculations are given. While the differences between NAA-WT and NAA10-V111G are clear, the authors should provide the statistical analysis of these differences.

Further, the author's results show that only the monomeric form has compromised function, and therefore it may be useful to discuss the issues concerning the monomer more thoroughly. For example, the authors state that several studies have indicated that the monomeric form of NAA10 may function as a lysine acetyltransferase (KNAT), and as such they should also discuss the negative findings of Magin et al. 2016 (1). These investigators failed to find any KNAT activity in recombinant NAA10 enzyme in vitro. This discrepancy should be discussed considering the importance of the issue of NAA10 possibly functioning as a KNAT in the monomeric form in vivo.


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 recommend additional statistical review

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

Acceptable

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