Reviewer's report

Title: A novel deletion mutation in KMT2A identified in a child with ID/DD and blood eosinophilia

Version: 0 Date: 14 Dec 2018

Reviewer: Yongguo Yu

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Zhang et al. reported the clinical phenotypes of a 3.5-year-old boy, including intellectual disability, developmental delay and blood eosinophilia, and then found that he carried a de novo heterozygous frameshift mutation (c.74delG;p:25Rfs) in the KMT2A gene by trio-based whole exome sequencing. These findings will not only contribute to expanding the phenotypical spectrum in patients with KMT2A mutations, but also shed new insight into the role of KMT2A in eosinophil metabolism. However, some problems need to be revised:

1. supplying ACMG/AMP assessment results;
2. English language needs to be modified, such as the last paragraph in Case presentation, "To analysis the relation between phenotypes of .....";
3. According to authors' description and the current evidence, persistently elevated eosinophil may result from mysterious variants in blood disease-associated gene.

Thus authors should supply candidate variants associated with blood disease in trio-wes as supplementary, and provide reasons for exclusion.

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.

No

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:

Needs some language corrections before being published

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