Reviewer’s report

Title: De novo frameshift mutation in ASXL3 in a patient with global developmental delay, microcephaly, and craniofacial anomalies

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Reviewer: Zubair Ahmed

Reviewer’s report:

In the manuscript by Dinwiddle et al., authors report a new allele of ASXL3 associated with number of clinical feature in a six year old female. The manuscript is well written and data presented support the conclusions.

Minor Essential Revisions

(1) specify that this is trio analysis and not candidate gene filters.
(2) state that no ASXL1 mutations were detected.
(3) report if any additional pathogenic de novo heterozygous mutations were detected in this six year old female sample and how they were eliminated as causative.
(4) describe more completely whether expression pattern and function studies support ASXL3 as the causative gene.

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

I declare that I have no competing interests