Reviewer’s report

Title: Exon deletions and intragenic insertions as a frequent cause for ataxia oculomotor-apraxia 2

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Reviewer: Filippo M. Santorelli

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The manuscript is not too original and lack any type of genotype-phenotype correlations.

The following major points are mandatory and need to be addressed.

1. The authors should list primers and probes used in qPCR studies to permit replication (even as a supplemental table).

2. The clinical phenotypes outlined in table 1 can be improved. It should be clarified if the neurological features as well as the AFP levels presented in table 1 refer to the latest examination and if the age is that of onset or at last observation.

3. It seems important to clarify if cerebellar atrophy at brain MRI is global or limited to the upper vermis.

4. The title appears to be misleading. If one combines data from this work and recent literature, “large” mutations account for about 8% of the total. Is this the case for a “frequent cause”? Certainly not, though a 10% is a number to be considered in diagnostic laboratories. I propose to modify the title as “large mutations in SETX are not rare in AOA2”.

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

Quality of written English: Needs some language corrections before being published

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