Reviewer's report

Title: De novo SCN2A splice site mutation in a male participant with Autism Spectrum Disorder

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Reviewer: Sean Ennis

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The article by Tavassoli et al ‘De novo SCN2A splice site mutation in a male participant with Autism Spectrum Disorder’ is an interesting case report. It describes an individual with ASD with a high de novo mutation rate including a de novo SCN2A splice site mutation producing a stop codon which is the likely cause of the individual's ASD. Although SCN2A mutations have been previously reported it is often associated with epilepsy, interestingly this individual does not have any history of seizures. Apart from late onset this is an important finding. This case may also help to reduce phenotypic heterogeneity in individuals with ASD and aid further studies. I therefore recommend that this paper be published with discretionary revisions.

1. The article would benefit from more detail on which chromosomal microarray analysis (CMA) was used aCGH or SNP and at which density.
2. The authors obviously have looked at the rest of the exome variants, a statement about other inherited mutations present or not would be of benefit here.
3. A possible ‘Typo’ The de novo SCN2A splice site mutation produced a stop codon 10 amino acid downstream, possible resulting in a truncated protein and/or a nonsense-mediated mRNA decay. Should this not read possibly?

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