Reviewer's report

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

Version: 1 Date: 24 January 2014

Reviewer: Michael Gill

Reviewer's report:

This is a detailed case report with a great deal of clinical information. The characterisation of the de novo mutations found in this individual suggests that the mutation in SCN2A is likely to have an impact on the splicing and expression of the gene. The authors conclude that the mutation should be considered pathogenic in this case because it causes abnormal gene splicing, leading to significantly shortened protein produce (not tested) and/or an abnormal message that (may) be subject to nonsense-mediated mRNA decay. They also describe the gene as a “recently identified ASD gene”

The case is one of three previously reported as having different SCN2A mutations. The case for it as "an identified ASD gene" is circumstantial at best. The conclusions that the mutation is "pathogenic" is speculative.

Major compulsory revisions
The paper should be shortened to about 1/3 its size
The text should reflect the descriptive nature of the work and not describe it as pathogenic in a recently identified ASD gene.

Minor essential revisions
In the abstract the sentence beginning "to characterise a seven-year old...." is poorly written and should be revised to make better sense.

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.