Reviewer’s report

Title: An investigation of ribosomal protein L10 gene in autism spectrum disorders

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Reviewer: Astrid Vicente

Reviewer’s report:

Major compulsory revisions
The work reported in this article is a follow up to a previous report of mutations in the RPL10 gene in autistic subjects. In the present study no non-synonymous mutations were found, and the authors could not find any alterations in levels of mRNA transcripts from lymphoblasts associated with autism.

As the authors correctly state, there are important limitations in this study, namely the population sample size, which is too small and therefore has very limited power to detect rare mutations. The authors also report no significant differences in RPL10 mRNA levels, but these assays were carried out in lymphoblasts, and therefore relevance for a brain disorder is arguable. Finally, sequencing only included exons and flanking sequences, but not promoter and other regulatory regions.

While for complex disorders such as autism it is very important to report negative results, namely the non-replication of previous findings of mutations or associations, it is also fundamental that the follow up studies are appropriately designed. The present study is valuable in the context of autism, as it could provide further support for a new disease mechanisms, and eventually indication regarding mutation screening of autistic subjects. However, unless the sample size is increased to a reasonably powered sample, the study is not conclusive and does not advance the field.

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 not competing interests.