Reviewer's report

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

Version: 1 Date: 24 October 2008

Reviewer: Dai Zhang

Reviewer's report:

BMC Medical Genetics considers the following article types: Database, Debate, Research, Software, Study protocol and Technical advance articles. The journal does not generally consider narrative review articles.

When assessing the work, please consider the following points:

1. Is the question posed by the authors well defined? Yes
2. Are the methods appropriate and well described? Need more description in detail.
3. Are the data sound? Yes
4. Does the manuscript adhere to the relevant standards for reporting and data deposition? Yes
5. Are the discussion and conclusions well balanced and adequately supported by the data? Need more description in detail.
6. Are limitations of the work clearly stated? Need more details.
7. Do the authors clearly acknowledge any work upon which they are building, both published and unpublished? Yes
8. Do the title and abstract accurately convey what has been found? Yes
9. Is the writing acceptable? Yes

- Discretionary Revisions
  None
- Minor Essential Revisions Yes
- Major Compulsory Revisions (which the author must respond to before a decision on publication can be reached)

The comments are as follow:

Previous literature has suggested that RPL10 might play a role in the pathogenesis of autism. Gong et al. explored the correlation between RPL10 and ASD using the re-sequenced and quantified PCR methods, however, they found that the RPL10 has no major effect on the susceptibility to ASD. There are following questions should be set forth in detail.

1. In the part of Methods, the authors described as “Of 141 ASD patients, 109
subjects showed mental retardation”. How did the authors identify the mental retardation? Did the authors mean that 109 patients met the diagnostic criteria of MR or these patients showed much lower IQ levels? What was the intellectual level for the 48 patients and 27 controls who received quantitative PCR examination? How could the authors exclude the mixture effect of mental retardation on the correlation between ASD and RPL10?

2. The authors sequenced all RPL10 exons and flanking junctions in 141 ASD patients, however, they did not identify any missense mutation in their patients. On the other hand, Klauck et al. had identified two non-synonymous mutations, L206M and H213Q, in the C-terminal domain of RPL10 in four boys with ASD from 345 patients. As both the two study used the European populations, what the other reasons might cause the polymorphic difference across the two samples? They authors should explore what potential factors caused this difference in the Discussion.

3. The authors mentioned that based on their sample size they had 56% of chance to detect at least one mutation in their sample. How did they get the 56%, what are the parameters for this estimation?

4. What was significant level the authors set for the statistic analysis?

Quality of written English: Acceptable