Reviewer’s report

Title: The identification and characterization of the p.G91 deletion in CRYBA1 in a Chinese family with congenital cataracts

Version: 1 Date: 26 May 2019

Reviewer: Zhang Yikui

Reviewer's report:

This manuscript confirmed that small in-frame one amino acid deletion in the beta-crystalline gene CRYBA1 was the genetic cause of congenital cataract in a Chinese family by sequencing; the authors also performed several in vitro experiments to try to explain why this indel mutation in gene CRYBA1 would cause congenital cataract.

Here following are my comments on this manuscript:

1. Whether the esotropia and nystagmus of these patients with congenital cataract were caused directly by this indel mutation or were just as a consequence of congenital cataract? Is it possible that this indel mutation may also affect ocular muscular movement genetically?

2. Line 49 of Abstract: "more aggregation" should be corrected as "more aggregate", because "aggregate" is an adjective while "aggregation" is not.

3. Line 60 of Abstract: the authors didn't provide sufficient evidence proving that this mutation was able to "destabilize the protein". According to the results of this manuscript, it will be more appropriate that this mutation results in abnormal expression and distribution of CRYBA1 protein.

4. Line 29 of Results: the authors should state clearly that whether both eyes of these patients developed cataract or not. If both eyes were opaque due to congenital cataract, the expression of "showed no other abnormalities in THE OTHER eye structures" is quitly confusing and misleading; if only one eye developed cataract while the other eye is fine, how to explain that the other eye could spare the pathogenetic indel mutation of the beta-crystalline gene CRYBA1?
Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Yes

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Yes

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

No

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

I am able to assess the statistics

Quality of written English
Please indicate the quality of language in the manuscript:

Acceptable

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