Reviewer's report

Title: Preimplantation genetic testing for a family with Usher syndrome through targeted sequencing and haplotype analysis

Version: 0 Date: 12 Mar 2019

Reviewer: Reviewer 2

Reviewer's report:

PEER REVIEWER ASSESSMENTS:

OBJECTIVE - Full research articles: is there a clear objective that addresses a testable research question(s) (brief or other article types: is there a clear objective)?

Yes - there is a clear objective

DESIGN - Is the current approach (including controls and analysis protocols) appropriate for the objective?

Yes - the approach is appropriate

EXECUTION - Are the experiments and analyses performed with technical rigor to allow confidence in the results?

Yes - experiments and analyses were performed appropriately

Statistics - Is the use of statistics in the manuscript appropriate?

N/A - there are no statistics in this study

INTERPRETATION - Is the current interpretation/discussion of the results reasonable and not overstated?

Yes - the author's interpretation is reasonable

OVERALL MANUSCRIPT POTENTIAL - Is the current version of this work technically sound? If not, can revisions be made to make the work technically sound?

Probably - with minor revisions
PEER REVIEWER COMMENTS:

GENERAL COMMENTS: I think that this a useful contribution to the field. I see two issues. 1) The authors did not describe a biopsy procedure, this is all I see "...11 embryos were subsequently biopsied at the blastocyst stage". I think that more details will help. 2) The clinical importance of this study is not clear. The c.10740+7G>A SNP in USH2A may or may not be pathogenic. However the methodology looks promising thus the study looks attractive.

REQUESTED REVISIONS:

The authors need to describe the biopsy procedure in details, this is important for reproducibility of this study.

ADDITIONAL REQUESTS/SUGGESTIONS:

"...mutation taster..." is not good, the name of the program is "MutationTaster", the authors should add a reference.

Note: This reviewer report can be downloaded - see attached pdf file.

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.

Yes

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.

Not relevant to this manuscript

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

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