Reviewer’s report

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

Version: 1 Date: 26 Aug 2019

Reviewer: Reviewer 3

Reviewer's report:

"PEER REVIEWER ASSESSMENTS:

OBJECTIVE - Full research articles: is there a clear objective that addresses one or several testable research questions? (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 sufficient 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?

Yes - appropriate statistical analyses have been used in the study

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

Yes - the author's interpretation is reasonable

OVERALL MANUSCRIPT POTENTIAL - Has the author addressed your concerns sufficiently for you to now recommend the work as a technically sound contribution? If not, can further revisions be made to make the work technically sound?
Yes - current version is technically sound

PEER REVIEWER COMMENTS:

GENERAL COMMENTS: The manuscript titled "Pre-implantation genetic testing for a family with Usher syndrome through targeted sequencing and haplotype analysis" outlines the design and application of a PGT-M approach using MDA and capture sequencing for embryo selection. This paper presents an informative case study and example of a useful method for establishing custom preimplantation genetic testing for a monogenic disorder. The paper has been improved in response to the initial round of reviewer comments, and the research appears to be of sound quality. A few minor corrections are recommended.

ADDITIONAL REQUESTS/SUGGESTIONS:

The introduction should be further edited, particularly the text added during revision. Although it is useful to give some history and perspective on previous methods, this information should be integrated into the flow of the document rather than listing techniques. Sentence structure should be edited to ensure that meaning is clear and to avoid hanging or incomplete clauses.

Descriptions of evidence of pathology for identified mutations should be better defined to be accessible to readers unfamiliar with these classifications (page 9-10) and sources of this information should be cited.

With edits to the prenatal diagnosis section (page 12), the authors should still define which embryos were selected for transfer.

The paper uses many acronyms and abbreviations; these should be carefully re-checked before publication. PGD needs to be replaced with PGT in a couple spots. Some abbreviations need to be defined at first use (e.g., HMM in abstract) and/or added to the abbreviations list (CNV, SNP, correct PGT-M/PGT-A, check for others).

Correct "diagnosis" to "testing" in keywords.

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.

I am able to assess the statistics

Quality of written English

Please indicate the quality of language in the manuscript:

Needs some language corrections before being published

Declaration of competing interests

Please complete a declaration of competing interests, considering the following questions:

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5. Do you have any other financial competing interests?

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