Author’s response to reviews

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

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**Author’s response to reviews:**

Dear Editors and Reviewers:

We appreciate the detailed and valuable comments from you and reviewers. Those comments are all valuable and very helpful for revising and improving our paper. The manuscript has been carefully revised and the major revisions were indicated using track changes in revised manuscript. The point-by-point response to the comments and suggestions were listed as below. We sincerely hope this manuscript will be finally acceptable to be published on BMC Medical Genomics.

Responds to the reviewer’s comments:

Technical Comments:

Comment 1: Please change the header 'Objective' to 'Background' in the Abstract, in line with our submission guidelines.

(https://bmcmedgenomics.biomedcentral.com/submission-guidelines/preparing-your-manuscript/research-article).

Response: Thank you for your useful comments. We have made correction and re-writen this part according to the submission guidelines (Abstract section, line 41-44, page 3).

Comment 2: Please change the headers 'Introduction' to 'Background' and 'Materials and Methods' to 'Methods'.

Response: Thank you for your useful comments. We have made correction according to the reviewer’s suggestion (Page 4, Line 60; Page 6, Line 119).

Comment 3: In the 'Ethics approval and consent to participate' statement, please provide the full name of the Institutional Review Board that approved the study, as well as the approval number (if applicable). We also ask you to add that the consent to participate obtained from the subjects was written, as already stated in the Methods.

Response: Thank you for your useful comments. As the reviewer's good advice, the full name of the Institutional Review Board, the approval number and the written consent was added in the revised manuscript ('Ethics approval and consent to participate statement' section, line 337-339, page 14).
Comment 4: In the 'Funding' statement, please declare the role of the funding body in the design of the study and collection, analysis, and interpretation of data and in writing the manuscript.

Response: Thank you for your useful comments. The role of the funding body in the design of the study and collection, analysis, and interpretation of data and in writing the manuscript was added in the revised manuscript ('Funding statement' section, line 352-353, page 15).

Comment 5: Please include a statement in the Authors' contributions section to the effect that all authors have read and approved the manuscript, and ensure that this is the case.

Response: Thank you for your useful comments. The statement that all authors have read and approved the manuscript was added in the Authors' contributions section. (Line 359, Page 15).

Reviewer 2

Comment 1: The authors implemented suggestions. I think that the authors did a good job. Actually I do agree with the reviewer #1: "Methods section is very long and contains a lot of technical details, authors can skip part of it, which will make the paper easier to read." I think that removal of technical details may be a problem. I would add those things to a supplement instead of removal. However, I do not insist on this. I think that the paper is acceptable.

Response: Thank you for your useful comments. As the reviewer's good advice, the technical details in methods section were moved to supplemental materials (Additional file 2).

Comment 2: I would add removed technical details to a supplement but I do not insist. I think that the paper is acceptable as is.

Response: Thank you for your useful comments. As the reviewer's good advice, the technical details in methods section were moved to supplemental materials (Additional file 2).

Reviewer 3

Comment 1: 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.
Response: Thank you for your useful comments. The list techniques have been integrated into the flow of the document (Background section, line 66-82, page 4). We also modified the sentence structure to make it clear to reviewer.

Comment 2: 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.

Response: Thank you for your useful comments. Descriptions of evidence of pathogenicity for identified mutations have been added in supplemental materials according to the reviewer’s suggestion (Additional file 4: Table S3). The cited reference was added in the revised manuscript (Page 8, Line 200, reference 17).

Comment 3: With edits to the prenatal diagnosis section (page 12), the authors should still define which embryos were selected for transfer.

Response: Thank you for your useful comments. The embryo selected for transfer was added in the revised manuscript (Results section, line 248-249, page 10).

Comment 4: 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).

Response: Thank you for your useful comments. The acronyms and abbreviations were carefully re-checked throughout the paper. The abbreviations were defined at first use and added to the abbreviations list.

Comment 5: Correct "diagnosis" to "testing" in keywords.

Response: Thank you for your useful comments. The "diagnosis" in keywords has been corrected as "testing" in the revised manuscript according to the reviewer’s suggestion (Keywords section, line 59, page 3).

We tried our best to improve the manuscript and made some changes in the manuscript. These changes will not influence the content and framework of the paper. We appreciate for editors and reviewers’ warm work earnestly, and hope that the correction will meet with approval. Once again, thank you very much for your comments and suggestions.

Sincerely Yours