Author’s response to reviews

Title: HDR Syndrome with a Novel Mutation in GATA3 Mimicking a Congenital X-linked Stapes Gusher: a case report

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Author’s response to reviews:

Response to Review Comments

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Thank you for the positive feedback and potentially accepting our paper. We wish to express our appreciation for your in-depth comments, suggestions, and corrections, which have greatly improved the manuscript. I hope my revision has improved the paper to a level of their satisfaction. Number wise answers to their specific comments/suggestions/queries are as follows.

Response to Editor Comments

Editor Comments:

1. Please amend the headings in the main text, they should be: 1) Background, 2) Case presentation, 3) Discussion & Conclusions.

Response: As suggested, we revised the headings.

2. Please remove the funding information from the Acknowledgements and include it in the Funding section instead. If you have no further acknowledgements please put “Not Applicable” in the Acknowledgements section.
Response: As suggested, we revised as below.

Acknowledgements

Not Applicable

Funding

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3. We would also like to ask for you to provide more justification for the contributions of JK and SHH, as currently they do not automatically qualify for authorship. Contribution to one aspect of the manuscript, alone, does not usually justify authorship.

An 'author' is generally considered to be someone who has made substantive intellectual contributions to a published study. According to the ICMJE guidelines, to qualify as an author one should have:

a) made substantial contributions to conception and design, or acquisition of data, or analysis and interpretation of data; AND

b) been involved in drafting the manuscript or revising it critically for important intellectual content; AND

c) given final approval of the version to be published. Each author should have participated sufficiently in the work to take public responsibility for appropriate portions of the content; AND

d) agreed to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

Anyone listed as an author must be included in this section. If you choose to change your author list you will need to fill out a change in authorship form and send it by email to the Editorial office to be approved by the Editor. The form can be found here: https://www.biomedcentral.com/getpublished/editorial-policies#authorship.

Anyone who contributed towards the article who does not meet the criteria for authorship can be acknowledged in the ‘Acknowledgements’ section.

Response: Thank you for your precious comments. As suggested, we revised as below.

SHH assisted in developing figures and performed evaluation of hearing loss referring the patient to our clinic. SHH also involved in revising the manuscript. JK was the study coordinator and
involved in revising the manuscript. He was responsible for the collection of clinical data and enrolled the patient and his family.

4. Please clarify in the Consent for publication what the patient's parents consented to being published, e.g. medical data, images, etc.

Response: As suggested, we revised as below.

Consent for publication

The parent of the patient consented to the publication of the case and any accompanying images with written consent.

5. Thank you for including the CARE checklist, please remove it as it is no longer required, and please include a statement in the Case presentation section stating that the CARE guidelines were followed.

Response: As suggested, we removed the CARE checklist and included a statement at the end of case presentation section stating that “The CARE guidelines were followed in this study”. You can see the statement at case presentation section, line 97, page 5.

6. Please remove the response letter, as it is no longer needed at this stage.

Response: As suggested, we removed the response letter.

7. Please include the heading "Additional File List" above the legend for SUPPL.FIG.1.

Response: As suggested, we included the heading "Additional File List" above the legend for SUPPL.FIG.1.

8. At this stage, please upload your manuscript as a single, final, clean version that does not contain any tracked changes, comments, highlights, strikethroughs or text in different colours. All relevant tables/figures/additional files should also be clean versions. Figures (and additional files) should remain uploaded as separate files.

Response: As suggested, we reuploaded revised manuscript as a final, single, clean version.

Response to Reviewer #1 Comments
"140 The molecular analysis of our patient revealed a heterozygous deletion of AT nucleotides in exon 6 (c.1201_1202delAT) of the GATA3 gene causing a frameshift at the 401st codon with a premature stop codon at the 106th codon of the reading frame (p.Met401Valfs*106)."

This cannot be described as a premature stop codon as the normal stop codon is at 444. Here there is a C-terminal extension until 106 codons after the frameshift at codon 401. Thus, the total protein is 506 amino acids long which includes 106 missense amino acids at the C-terminus. Your nomenclature for the mutation is correct, your interpretation for what this means is not.

Please revise text accordingly.

Response: We appreciate the reviewer for taking the time to offer us comments and insights. Your precious comment improved the quality of our paper. As suggested, we revised the text as below. You can see revised version at discussion section, line 139-141, page 7.

The molecular analysis of our patient revealed a heterozygous deletion of AT nucleotides in exon 6 (c.1201_1202delAT) of the GATA3 gene, which is predicted to cause a frameshift at the 401st codon for the methionine and the new reading frame ending in a stop at postion 106 (p.Met401Valfs*106).

We thank your kind comments.

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CLOSING COMMENTS TO THE EDITOR:

Again, we appreciate the opportunity to revise our work for consideration for publication in BMC medical genetics. We hope our revision meet your approval.