Author’s response to reviews

Title: Novel EYA4 variant in Slovak family with late onset autosomal dominant hearing loss: a case report

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Author’s response to reviews:

Isabelle Schrauwen
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Editor of BMC Medical Genetics

Dear Editor,

Herewith I would like to take the liberty of re-submitting our manuscript by Varga et al. entitled "Novel EYA4 variant in Slovak family with late onset autosomal dominant hearing loss" (MGTC-D-18-00511R1). The manuscript was revised as recommended by the Reviewers.

We like to thank You and the Reviewers for taking the time to evaluate our paper and the valuable and stimulating criticism as well.

Ad Reviewer 1

The manuscript was checked for typing errors. We apologize for a few mistakes in the previous version.
Ad Reviewer 2

1) Insisting on the fact that this is the first EYA4 mutation found in Slovakia also does not seem to be really significant since EYA4 mutations have been described in Caucasians so it is not surprising there may be a Slovak case and these sentences should be revised accordingly.

Authors response to point 1:

As suggested by the reviewer the sentence was omitted from the abstract. Our idea was to highlight that this is the first EYA4 variant not only in the Slovak population but also in the whole Slavic population, which is the largest ethnic and language population among European Caucasians.

2) The authors mention that a DFNB1 etiology was excluded in the family but fail to comment how in fact due to the clinical features presented by the affected individuals DFNB1 was not likely to be the problem.

Authors response to point 2:

The reviewer is correct that the DFNB1 etiology was unlikely in the presented family but the DFNB1 screening is a routine procedure performed by our laboratory. This is explained in page 4 lines 78-79.

3) The characterization of the variant, which by the way had in silico strong pathogenic predictions and segregated with a consistent HL phenotype in the family, includes RT-PCR from blood and urine despite being known that EYA4 is not expressed, at least in blood. This part of the analysis is not relevant.

Authors response to point 3:

Thank you for the reviewer's comment. The sentence was corrected as suggested by the reviewer (see page 10 lines 204-207).

4) The last sentence of the discussion is obvious and does not convey any sound conclusion for the work done.

Authors response to point 4:

The last sentence was removed from the conclusions.

5) The manuscript would benefit from language revision
Authors response to point 5:

The manuscript was checked for language errors.

Yours sincerely,

Daniela Gasperikova, D.Sc.

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