Reviewer's report

Title: Characterization of a new full length TMPRSS3 isoform and identification of mutant alleles responsible for nonsyndromic recessive deafness in Newfoundland and Pakistan

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Reviewer: Edi L Sartorato

Reviewer's report:

Mutations in the TMPRSS3 gene are a rare cause of hearing loss in Caucasians patients but seems to be important in Pakistani families considering the heterogeneity of deafness in this population. The large number of individuals analyzed provides an epidemiological study and reveals a new isoform of the TMPRSS3 protein. The authors should analyze the possibility of to construct a minigene to study the intronic mutation IVS8+8insT in order to check changes in the splice sites. The results reported in the paper may contribute to understanding the etiology of deafness and well-characterize the genomic structure of the TMPRSS3 gene.

What next?: Accept after discretionary revisions

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Acceptable

Statistical review: No

Declaration of competing interests:

None