Reviewer's report

Title: Screening mutations of the Otoferlin gene (OTOF) in Chinese patients with auditory neuropathy, including a familial case of temperature-sensitive auditory neuropathy

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Reviewer: Thomas Friedman

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Review of Wang et al., 2009

The authors report a molecular genetic analysis of mutations of OTOF (DFNB9) in 73 Chinese with auditory neuropathy (AN) patients (92 ethnicity matched controls). One AN subject exhibited a temperature sensitive AN phenotype. The authors report three single mutant OTOF heterozygotes and one compound heterozygote (the temperature sensitive AN patient). Assuming this study is representative, mutations of OTOF associated with AN among the Chinese seems to be very rare compared to other populations that have been studied.

Major issues:

1. If recessive mutations of OTOF are responsible for AN, what are the possible reasons for finding only one mutant allele of OTOF in the three AN subjects?

2. Provide supporting clinical data in the manuscript to convince a reader that the subject described on page 8 has temperature sensitive AN.

Minor issues:

1. Page 4, OTOF wasn’t mapped to chromosome 2. AN was mapped to chromosome 2 and mutations were subsequently found in OTOF.

2. Page 5, How would the identification of more alleles of OTOF provide greater understanding of “the molecular pathway(s) for this complex neurological disorder”?

3. Figure legend 1b, what is meant by “intercepted ClustalW alignment”?

4. The quality of figure 1 could be improved.

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

Quality of written English: Needs some language corrections before being published
Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:
I declare that I have no competing interests' below