Reviewer's report

Title: Combined examination of sequence and copy number variations in human deafness genes better explained the cause of genetic deafness

Version: 3  Date: 14 May 2014

Reviewer: David J Lim

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GENERAL COMMENTS:
This study represents the first attempt to examine the deafness genes associated with a group of patients who have diagnosis of deafness by the combination of data obtained by sequence variations and copy number variations using next generation sequence. The major strengths of this study is the utilization of the state of art technologies available, multiple validations of the data, and excellent discussion concerning advantage and disadvantage/limitations of various techniques currently being used. This study revealed possible molecular mechanisms involved in the genetic deafness. The newly generated data are highly informative in understanding human deafness and deafness gene functions and dysfunctions.

SPECIFIC COMMENTS:
Minor Essential Revision:
1. In page 3, line 83, “otitis media were ruled out”. Perhaps, “excluded” may be better choice of word to describe patients with otitis media were not included in the study.

Minor Discretionary Revisions:
2. In page 6, line 144, a symbol indicating greater than or less than is missing in front of 0.005. This is most likely due to the web-based manuscript retrieval.
3. In page 7, line 171, “Ct” could be plainly explained or defined for unsophisticated readers in genetics or statistics.

Level of interest: An article of outstanding merit and interest in its field

Quality of written English: Acceptable

Statistical review: Yes, but I do not feel adequately qualified to assess the statistics.

Declaration of competing interests:
I have no competing interest