Reviewer's report

Title: Whole-exome sequencing identifies MYO15A mutations as a cause of autosomal recessive nonsyndromic hearing loss in Korean families

Version: 1 Date: 29 March 2013

Reviewer: Tom Walsh

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Woo et al describe exome sequencing of 13 families with ARNSHL that had previously been excluded for GJB2 and SLC26A4 mutations. They report on two of the families in this manuscript. One family appears resolved by compound heterozygous mutations in MYO15A. One allele is a likely splice site alteration, although RT-PCR evidence is not presented. The second allele is a missense mutation that is also predicted to be damaging.

Major Compulsory Revisions

1) The second family is not resolved and should not be reported unless a convincing second MYO15A allele is identified. Exome based CNV calling could be attempted to determine if there is a MYO15A exonic deletion or duplication that could resolve the recessive hearing loss in this family.

Minor Essential Revisions

1) Ref#5 refers only to Iran. Add in refs for Pakistan and Turkey
2) Missing word in last sentence of page 5
3) Please state frequency of S3474G in the 1KG dataset
4) Please list the variants in GRHL2 and TECTA genes
5) Ref#14 should cite the original paper(s)

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