Reviewer's report

Title: Targeted exome sequencing reveals novel USH2A mutations in Chinese patients with simplex Usher syndrome

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Reviewer: Kari Branham

Reviewer's report:

General Review: The authors provide an example of the application of targeted exome sequencing to a disease (Usher Syndrome) with extensive genetic heterogeneity. The only novel information presented in the paper is one of the mutations—there have been multiple other papers using exome sequencing to identify mutations in patients with Usher Syndrome.

Minor Essential Revisions:

1. Background:
   Line 50: the hearing is patients with Usher Syn II is generally described as congenital hearing loss that is mild-moderate in low tones and moderate to severe at higher frequencies.
   Line 52: regarding Usher III—should remove the word “also” not born deaf.
   Line 60: there are 12 Usher Syndrome genes: CDH23, CEP250, CIB2, CLRN1, DFNB31, GPR98, HARS, MYO7A, PCDH15, USH1C, USH1G, USH2A

2. Results
   Candidate mutations: Line 143: more specific information should be provided about a list of candidate gene mutations. Were there any other possible Usher Syndrome genes with mutations?
   Analysis of USH2A mutations: Line 163: more specific information about the prediction software used and what the results were should be provided. As written it is very vague.

Discretionary Revisions

1. Results-clinical findings:
   It would be much more useful to other researchers if additional information was provided about clinical features, such as age of onset of disease. Moreover, virtually no information about the phenotype of the proband in F2 is given.

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

Quality of written English: Acceptable
**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.