Reviewer's report

Title: Pathogenic substitution of IVS15+5G>A in SLC26A4 in patients of Okinawa Islands with enlarged vestibular aqueduct syndrome or Pendred syndrome

Version: 3 Date: 20 February 2013

Reviewer: Chia-Wen Hsu

Reviewer's report:

Major Compulsory Revisions:

1. Compared to several similar reports published in BMC Human Genetics, this study uses a relatively smaller sample size (n = 22) and lacks of potential controls (ie. healthy objects and cases with no PS or EVA). At least an equal number of control cases should be sampled. Considering the population of more than 1.4 millions of people in the Okinawa Islands, it is less convincing to draw conclusions from the 22 cases. Please give more detailed information regarding the geographic distribution of the test cases living in the Okinawa Islands, or include more patients with PS or EVA in the study.

2. Three patient samples per genotype should be used to produce meaningful standard deviation values in the Figure 5.

Minor Essential Revisions:

1. A control temporal bone CT should be included in the Figure 1. A scale bar in the figure or the measured value in the figure legend should be specified. It would be clearer to highlight the common crus and the external aperture with a circle or a point in white.

2. The molecular weight of the corresponding PCR product should be labeled in the Figure 4.

3. The numbering of the Figure 1 and the Figure 2 is mislabeled in the figures and the figure legends.

Discretionary Revisions:

None.

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' below.