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: 2 March 2013

Reviewer: Pu Dai

Reviewer's report:

Minor revision needed:

P10. line 11: In the sentence of “a compound homozygous for IVS15+5G>A”, compound should be deleted.

P10. line 15-17: Neck examinations revealed thyroid goiters in 8 of 22 patients. Overall, 0% (0/11) and 73% (8/11) of the patients younger and older than 10 years of age, respectively, had a thyroid goiter. Their serum FT4 and TSH levels were within the normal ranges.

In this paragraph, the percentage of patients with SLC26A4 mutation having goiter is high. Considering incidence of pendred's syndrome is relative low in most Asian countries, the epidemiology of Goiter in general population of Okinawa should be reviewed and discussed.

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

Quality of written English: Acceptable

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

Declaration of competing interests:

'I declare that I have no competing interests'