Author's response to reviews

Title: A wide spectrum of clinical and brain MRI findings in patients with SLC19A3 mutations

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Author's response to reviews:

Professor Tadao Arinami Editor, BMC Medical Genetics,

Dear Professor Tadao Arinami

MS: 1849205714464792


Thank you very much for your letter of November 29, 2010 with the reviewers’ comments and your kind consideration of our manuscript (MS: 1849205714464792). We have revised the manuscript as far as possible in line with the reviewers’ kind suggestions. The changes we made are as follows: 1. Responses to comments of Reviewer 1

Item: None

2. Responses to comments of Reviewer 2

Item: For the clue of better understanding of readers, a concise table comparing the three categories involving the BBGD phenotype, Wernicke’s-like encephalopathy, and severe infantile spasm phenotype may be required.

According to the suggestion of Reviewer 2, we made new table1 including the
summary of clinical features and brain MRI findings of the present four patients, BBGD and Wernicke’s-like encephalopathy caused by SLC19A3 mutations. Since the clinical features of reported cases of BBGD are very broad. For example, nystagmus was seen only one patient. Thus, we described typical features of BBGD.

3: Minor corrections.

We found several minor errors. The corrections are as follows.

1) We changed all “Fig” to “Figure”.

2) We corrected the word “Fig. 1” to “Figure 2” at “Patient V-2” (P5L4 and L5).

3) We corrected “Fig 3B” to “Figure 3A and B” at “result”. (P9L18).

4) We added the word “table 1” at “DISCUSSION” (P11 L2 and L14 ).

5) We corrected (1028G>C, E320Q) to (c.958G>C, [p.E320Q]) at “Abstract”, “Results”, “Conclusion” and “Figure Legends” (P3L13, P9L16, P12L20, P16L22, P17L1 and L3).

6) We changed the word “MRI” to “brain MRI” at “Patient V-6” (P6L23) and “result” (P910L).

7) We corrected the word “The axial views” to “The sagittal and axial views” at “Figure Legends” (P16L12).

8) We corrected “1028” to “958” at “Figure Legends” (P17L2).

Thank you for your consideration.

Yours sincerely,

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