Reviewer’s report

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

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Reviewer: Kenji Kurosawa

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RE: Yamada et al., A wide spectrum of clinical and brain MRI findings in patients with SLC19A3

This is an interesting clinical report describing a pedigree with four patients who presented with epileptic spasms in early infancy, severe psychomotor retardation, and characteristic brain MRI findings. The authors performed molecular analysis on the pedigree and determined the causative homozygous mutation in SLC19A3 gene, which is responsible for two clinical phenotypes, biotin-responsive basal ganglia disease and Wernicke’s-like encephalopathy. The authors demonstrated the broad phenotypic spectrum caused by mutations of SLC19A3, and suggested the potential benefit of biotin and/or thiamin treatments. This is a well-written paper containing interesting results.

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.

Level of interest: An article of outstanding merit and interest in its field

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.