Reviewer’s report

Title: A novel compound mutation in GLRA1 cause hyperekplexia in a Chinese boy- a case report and review of the literature

Version: 0 Date: 01 Aug 2017

Reviewer: Barbara Steinborn

Reviewer's report:

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Hereditary startle disease is caused by genetic defects in inhibitory glycine receptor and transporter genes. Missense, nonsense, frameshift, splice site mutations, and large deletions in the human glycine receptor α1 subunit gene (GLRA1) are the major known causes of this disorder. However, mutations are also found in the genes encoding the glycine receptor β subunit (GLRB) and the presynaptic Na+/Cl−-dependent glycine transporter GlyT2 (SLC6A5). The reasons of hyperekplexia may be connected with other mutation in other genes. We report for the first time a clear association of mutation in CTNNB1 with an atypical syndromic hyperekplexia expanding the phenotype of CTNNB1-related syndrome. Consequently CTNNB1 should be added to the growing list of genes to be considered as a cause of startle disease or syndromic hyperekplexia.[1]

I proposed to mention in this article about other mutation in hyperekplexia.


Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Yes

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I am able to assess the statistics

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