Reviewer's report

Title: Clinical features and gene mutational spectrum of CDKL5-related diseases in a cohort of Chinese patients

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Reviewer: Renzo Guerrini

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Zhao et al. describe the clinical features and mutational spectrum of CDKL5-related diseases in a cohort of 102 Chinese patients with early-onset epileptic encephalopathies and ‘atypical Rett syndrome’. The authors identified a CDKL5 mutation in ten patients, nine girls and one boy. The methods are appropriate and well described and the data are convincing.

Major Compulsory Revisions

The paper is too long and should be shortened. In particular, the results section “clinical manifestation” should be made available as online supplemental material. A brief clinical synopsis of the patients should then be added to the same section.

Authors use the terms early-seizure of RTT or Hanefeld variant of RTT. However, recent publications have clarified that CDKL5-related disorder should be considered as separate from RTT, rather than another variant (Guerrini and Parrini, 2012 Epilepsia. 2012 Dec;53(12):2067-78; Fehr et al. (Eur J Hum Genet. 2013;21(3):266-73). Please revise the paper taking into the account this view.

Minor Essential Revisions

Please add a sentence in the methods section indicating which CDKL5 gene accession numbers have been used to describe the mutations.

“Non-syndrome” should be changed in “non-syndromic”

The text should be revised for typos and grammatical errors.

Table 1 should be revised for formatting and typos. A legend should be added and the text modified accordingly (i.e. months : m; years : y) to improve the reading. Please clearly state the meaning of “_” or “-“ or alternatively use symbols.

Patient 7 had two XCI values. Is this correct?

Level of interest: An article whose findings are important to those with closely related research interests
Quality of written English: Needs some language corrections before being published

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