Reviewer's report

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

Version: 1 Date: 13 November 2013

Reviewer: nicoletta landsberger

Reviewer's report:

Minor Essential Revisions
In this manuscript the authors have screened a large Chinese cohort of selected patients for CDKL5 mutations, revealing 10 de novo cases. In the manuscript the authors describe the clinical manifestations of the positive patients.

This is a case report paper that adds novel CDKL5 pathogenic mutations to clinician and scientists and strengthen the relevance of searching for CDKL5 mutations in patients affected by early seizures of both genders.

Although the manuscript does not add much to the field, I believe the manuscript deserves publication providing that the authors significantly implement the language and in general the text. The introduction describing symptoms so far associated to CDKL5 is quite poor; furthermore, they have to better explain the aim of the work that was to analyze for the first time a cohort of Chinese patients.

The gene/protein nomenclature is not respected (human gene goes in italics and capital letter, the human protein is capital but not in italics).

The all text contains a huge amount of misspelling and wrong sentences.

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

Quality of written English: Not suitable for publication unless extensively edited

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