Reviewer’s report

Title: Intrafamilial phenotypic heterogeneity in a Taiwanese family with MAPT p.R5H mutation: a case report and literature review

Version: 0 Date: 04 Jul 2017

Reviewer: Alessio Di Fonzo

Reviewer’s report:

In this paper the authors describe the clinical features of a Taiwanese family displaying FTD, corticobasal degeneration (CBD), ALS and persistent depressive disorder (PDD) and highlighting the marked intrafamilial phenotypic variability and the association with a mutation in the MAPT gene p.R5H.

The paper is clear and the discussion is in agreement with the results.

Few remarks:

I suggest to screen the c.14G>A (p.R5H) variant a cohort of Taiwanese age-matched controls and/or to report data form SNP databases concerning the Chinese population.

Discussion:

I would add that MAPT mutations have also been found in pure motor neuron degeneration (Neurology. 2014 Jun 3;82(22):1990-8. doi: 10.1212/WNL.0000000000000476)

Conclusion:

Please revise the sentence as follows:

In summary, we present the first Taiwanese family presenting with PPA variant of FTD, CBD, ALS and PDD and carrying a MAPT p.R5H mutation. Our findings contribute to highlight the marked phenotypic variability within subjects with the same MAPT mutation and specifically extend the current knowledge of phenotypes associated to MAPT p.R5H mutation.

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

Yes

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Yes

**Are the conclusions drawn adequately supported by the data shown?**
If not, please explain in your comments to the authors.

Yes

**Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?**
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

Not relevant to this manuscript

**Quality of written English**
Please indicate the quality of language in the manuscript:

Needs some language corrections before being published

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