Reviewer’s report

Title: Rubinstein-Taybi 2 associated to novel EP300 mutations: deepening the clinical and genetic spectrum

Version: 3  Date: 20 Oct 2017

Reviewer: Walter E. Kaufmann

Reviewer's report:

The study describes an 8-subject Spanish cohort of individuals with Rubinstein-Taybi syndrome (RSTS) and mutations in the EP300 gene that, as the authors state is the less common genotype associated with RSTS. The affected subjects demonstrated an overall typical but milder RSTS phenotype and a variety of mutations, with frameshifts being the most common as reported in the literature. All major features of the cohort were in line with previous publications, with the exception of behavioral problems that were absent in the Spanish series.

The authors have been responsive to the initial review. In response to my comments, they have added a table comparing the reported series with the literature as well as information on parental genotyping. However, grammar issues still remain. For instance, in the new table, they use the awkward term "Autistiform" that should be replaced by "autistic."

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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