Reviewer's report

Title: Rubinstein-Taybi syndrome in a Saudi boy with distinct features and variants in both the CREBBP and EP300 genes: A case report

Version: 1 Date: 06 Nov 2018

Reviewer: Katrina Fontana

Reviewer's report:

Dear Editor,

I have revised this case report and it presents new and important informations about Rubinstein-Taybi syndrome. However, some issues must be evaluated before considering for publication, as follow:

1) I suggest including in the "Background" informations about the main CREBBP and EP300 genes variants that are already known to be associated with the Rubinstein-Taybi syndrome. The authors should make a brief genotype/phenotype correlation.

2) I suggest including in the "Mutational Analysis" a short description of the molecular methods used for the study of CREBBP and EP300 gene variants.

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

No

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:

Acceptable

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