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: 08 Nov 2018

Reviewer: Jennifer Below

Reviewer's report:

The authors present a case report with notable clinical characteristics of a child with RSTS and describe a likely causal mutation in CREBBP as well as a possibly benign mutation in EP300. The author suggest that some of the phenotypes observed may be due to interacting effects of the two observed variants. The paper is clear and concise and is generally well written (there is a minor typo on page 5, where the authors say 'evaluation' when I think the might have meant 'evolution').

I have few comments, which I believe would strengthen this case report.

1) it would be nice to see a table comparing features of RSTS from other published reported cases to this new case

2) the methods used to genotype the EP300 and CREBBP variants is completely absent- results and quality control measures from sanger sequencing should be shown (or taqman genotyping, or whatever methods the authors used). It would also be important for the reader to know exactly what portion of the genome was interrogated- for example comparisons are made to other cranio-facial abnormalities, but it is not explicitly clear to the readers whether or the the authors checked genes known to cause those cranio-facial abnormalities for mutations in this proband

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.

Unable to assess

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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