**Author’s response to reviews**

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

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**Version:** 1  **Date:** 10 Oct 2018

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MGTC-D-18-00379

A SAUDI BOY WITH RUBINSTEIN-TAYBI SYNDROME AND DISTINCT FEATURES: MIDLINE NOTCH OF THE UPPER LIP, BIFID TIP OF THE TONGUE, MIDLINE GROOVE OF THE LOWER LIP, PLUMP FINGERS WITH BROAD / FLAT FINGERTIPS, AND BRACHYDACTYLY. A CASE REPORT

Dear Editor

We have revised the paper as follows:

1) revise the title because the title doesn’t imply any gene has been analyzed by the authors

2) title is too long, make it concise but informative for the readers

3) title is not necessarily written in big letter for whole title
Response: The new title is now:
Rubinstein-Taybi syndrome in a Saudi boy with distinct features and variants in both the CREBBP and EP300 genes: A case report

4) revise the abstract and add the gene analyzed in the study

5) the authors should determine which variant might be the cause of the disease

Response: The abstract was modified adding the genes analyzed under “case presentation” and determining the disease-causing variant under “conclusion”:

Case presentation: we report on a Saudi boy with RSTS Type 1 and the following distinct features: a midline notch of the upper lip, a bifid tip of the tongue, a midline groove of the lower lip, plump fingers with broad / flat fingertips, and brachydactyly. The child was found to be heterozygous in the CREBBP gene for a sequence variant designated c.4963del, which is predicted to result in premature protein termination p.Leu1655Cysfs*89. The child and his father were also found to be heterozygous in the EP300 gene for a sequence variant designated c.586A>G, which is predicted in the amino-acid substitution p.Ile196Val.

Conclusion: Our report expands the clinical spectrum of RSTS to include several distinct facial and limb features. The variant in the CREBBP gene is known to be causative of RSTS Type 1. The variant in the EP300 gene is benign since the father carried the same variant and had no abnormalities. However, functional studies are required to investigate if this benign EP300 variant has an effect on the phenotype in the presence of disease-causing CREBBP gene mutations.

Thank you

The authors