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

Authors:
Mohammad Al-Qattan (moqattan@hotmail.com)
Abdulaziz Jarman (ajarman@kfshrc.edu.sa)
Atif Rafique (drafiq_2@yahoo.com)
Zuhair Al-Hassnan (zhassnan@kfshrc.edu.sa)
Heba Al-Qattan (heba.qattan96@gmail.com)

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

Mohammad Manna Al-Qattan; Abdulaziz Jarman; Atif Rafique; Zuhair N Al-Hassnan; Heba M Al-Qattan

BMC Medical Genetics

Dear Editor

I have revised the manuscript as follows:

Editor Comments:

Thank you for revising the manuscript according reviewers’ suggestions. However, there are several concerns should be addressed further before considering for publication, as follow:
1. please provide the ethical approval number for the study

Response:

The study was approved by the research committee at Riyadh Care (#22-2018)

This is now added to the Declarations section

2. kindly attach the English editing service approval

Response: Done. We have this free service at our University in Riyadh, Saudi Arabia since all teaching staff are Arabic nationals. This is done through Our Prince Naif Research Center. I have attached the required documents for your review under Supplementary Materials (The first file is my application to the center requesting the language review and the second is the Editor letter).

I have included all the recommended grammar/language corrections in the revised manuscript.

3. please acknowledge anyone who have already helped your study, such as patients family that agreed to participate, doctor/nurse that taken care of the patients or technician that performed the experiment, but not sufficient contribution to be considered as a co-author, into the Acknowledgement section

Response: Done. See Acknowledgement section

3. reviewer #3 (Jennifer Below) asked authors to provide the Sanger sequencing results of both variants. please add them as figure, including the sequencing of parents as well.

4. authors stated that all differences from the reference sequences (sequence variants) were assigned to one of the five interpretation categories as per the ACMG guidelines.

please categorize both variants according to five interpretation of ACMG guidelines, and explain briefly why authors suggest so.

Response: I am the corresponding author for the paper and I am a Hand Surgeon; and I have special interest in the genetics of congenital hand defect. Dr Zuhair N. Al-Hassnan is my co-author and he is the geneticist in the team. He is currently away on holidays and will not come back till after the NEW YEAR. You have requested the Sanger sequencing results/categorization; and you stated that the revision should be submitted by Dec 12th. I am not able to
provide this until he comes back. The aim of the paper is the delineation of several unique features of the syndrome. The same mutation was previously described by Bentivegna et al. and that was published in your respected Journal (this is mentioned in our paper in line 122 and the reference is given as ref#7)


If the Editor thinks this is absolutely necessary, please extent the time limit till end of January so that we will have time to prepare the detailed Sanger sequencing results after Dr Al-Hassnan comes back.

Thank you

The corresponding author (on behalf of all co-authors)