Author's response to reviews

Title: Molecular and clinical analysis of Ellis-van Creveld syndrome in the United Arab Emirates

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Author's response to reviews:

Dear Editor,

We thank you and the reviewers for the constructive comments on our manuscript entitled “Molecular and clinical analysis of Ellis-van Creveld syndrome in the United Arab Emirates” (Authors: Bassam R. Ali, Nadia A. Akawi, Faris Chadid, Mohmood Bakir, Moghis Ur Rehman, Aiman Rahmani and Lihadh Al-Gazali). We believe that we fully addressed all the comments as detailed below and we therefore hope that the manuscript is now acceptable for publication in BMC Medical Genetics.

Point by point responses

Response to the Editor comments

Point 1. We now include sub-headings in table 1 separating the skeletal from facial features for more clarity.

Point 2. We now include a new column in table 3 (previously table 4) to indicate the effects of the mutations (including the frame shift) on the protein.

Point 3. We now added to table 3 (now table 2) the dbSNP numbers, minor allele frequency and average heterozygosity score of the previously reported SNPs.

Point 4. We don’t have RNA from members of this family and therefore the effect on RNA splicing was not tested.

Point 5. Yes, we sequenced members of the same families reported by Tompson et al 2007

Point 6. We accept the comment by the reviewer regarding family 4 and now indicate that they have EvC-like phenotype.
Point 7. The promoter regions within EVC and EVC2 were predicted using the free online Proscan (version 1.7) software (http://www-bimas.cit.nih.gov/molbio/proscan/). A statement to this effect is now included in the methods section.

Point 8. “Malformed or absence of some glands and ducts” has been removed

Other comments addressed

Reviewer 1 (Z. Bian) comments

Families 2 and 3 in this manuscript are the same as reported in Tompson et al. 2007. We clarify that in the text.

We now moved table 1 (primers used) to the supplementary material section as suggested by the reviewer.

Gene names have been italicized in the legend of Figure 3 and throughout the text

Figure 1. We have standardized the symbols for the pedigrees as suggested

References formatted according to the journal style

Reviewer 2 (F.G.A. Verteegh) comments

Page 8. The name Juene has been changed to Jeune as suggested

Description of the 4th family was kept in the manuscript but we included clear statements that the diagnosis of EvC in this family is likely but not 100% confirmed and therefore we refer to it as “EvC-like”.

References: Van Hagen et al have been removed from the reference list.

Looking forward to hearing from you positively

Best Regards,

Bassam Ali