Author's response to reviews

Title: Extending the spectrum of Ellis van Creveld syndrome: a large family with a mild mutation in the EVC gene

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Author's response to reviews: see over
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Melissa Norton, MD Editor-in-Chief
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Re: Ulucan et al, 3597171782052620 - Extending the spectrum of Ellis van Creveld syndrome: a large family with a mild mutation in the EVC gene

Dear Drs. Norton and Patel,

Thank you for the prompt and thoughtful review of our manuscript. We are grateful to the reviewers for their comments. We have revised the manuscript according to these comments, and have performed another check for grammatical errors and typos. A detailed response to the reviewer’s comments is provided below.

REVIEWER 1
The reviewer has provided a thoughtful and scholarly critique of the differences between Weyers and EvC. We have modified a number of features of the table as, on review of the published data, we agree with the reviewer’s comments. We rewrote our discussion to de-emphasize the distinction of EvC from Weyers based on features alone, and have emphasized inheritance, which is unambiguously distinct in these two disorders.
We corrected the location of Table 1.

REVIEWER 2
Tables 2 and 3 were merged into one table.
We are grateful to the reviewer for pointing out the paper by Temtamy et al, 2008 which was added to the discussion and new mutations are added in the new Table 3 (which was Table 4 in the original submission).

Thank you again for the excellent reviews.
Sincerely,

Leslie G. Biesecker, M.D.