Reviewer's report

Title: Genetic Heterogeneity of Ellis-van Creveld in the United Arab Emirates

Version: 1 Date: 25 June 2009

Reviewer: Hakan Ulucan

Reviewer's report:

Minor essential revisions:
1. The title should be "Genetic Heterogeneity of Ellis-van Creveld syndrome in the United Arab Emirates"
2. The term "polydactyly" could be added to the keywords
3. After the use of abbreviations do not repeat the long expression, just use EvC in the last line of the abstract.
4. Introduction 10th line of the 1st paragraph: Uluncan to Ulucan
5. Introduction 5th line from the end: delete part "although EvC patient present typical phenotype in most cases"
6. Introduction 4th line from the end: Uluncan to Ulucan
7. Materials/Met: 1st line: neonatologists to neonatologist
8. Results: percentiles should be added to cases in families 2, 3, 4.
9. Results: Ethnic diversity in UAE is mentioned in the discussion but not all ethnicities are given in the patients phenotypes. It is important for this paper.
10. Results Family 3: I spostaxial polydactyly is bilateral?
11. Results Family 4: 3rd line: should be "her birth weight"
12. Results family 4 line 10: She is 2-year-old
13. Results family 4 line 11: The uncle..... should be paragraph.
14. Results family 4: 3rd line from end: Is it "there were short extra frenula" or "Her upper frenulum and lower frenulum were short". Which one is correct?
15. Results 3rd line from the end: murmer to "murmur"
16. Patients genotypes 6th line EVC2 should be italic type.
17. Patient genotypes 3rd and 2nd lines: family number 2 and family number 3 to family 2 and family 3.
18. Discussion 6th line from end: EVC and EVC2 should be italic.
19. Discussion: You say the patients are typical in the introduction but in the sentence "In this paper, we report the clinical and molecular heterogeneity of this condition in UAE population (table 3)." The cases also seems typical to me. I this case you should say" In this paper, we report a novel mutation and also confirm two previously reported mutations in the EvC, indicating molecular heterogeneity in the disorder."
20. You should get Tables 2 and 3 together and show the mutations highlighted and novel mutation and previously reported mutations marked in the box and referred in the legend. You have to make necessary changes in the text as well after the table merging.

**Level of interest:** An article of importance in its field

**Quality of written English:** Acceptable

**Statistical review:** No, the manuscript does not need to be seen by a statistician.

**Declaration of competing interests:**

I declare that I have no competing interests