Reviewer's report

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

Version: 1 Date: 7 July 2009

Reviewer: Florens G.A. Versteegh

Reviewer's report:

General
-the paper should be shortened.
-figures could be ommitted

Major Compulsory Revisions

Introduction:
-first sentence: Ellis and van Creveld described in their paper already previous reports. To mention McIntosh separately seems unnecessary.
-"300 cases reported worldwide being most prevalent in the Amish population of USA (Baujat and Le Merrer, 2007) with cases reported in Mexico, Brazil and Ecuador": saying worldwide and then mentioning 4 countries in the Americas seems contradictory.

Materials and Method:
-"Genomic DNA was extracted from whole blood.
: since the authors state that opportunities in UAE are limited and 4 out of 6 patients died they should describe how, where and when the blood was collected/sampled and stored and where the tests were performed.

Results:
-from all patients should be mentioned when they were born and where.

-since in family 4 no mutations were found, the question remains whether these patients suffered from EvC syndrome or from another condition. May be this family should be ommited. Or a very good reason should be described to keep this family included.

References:
-I could not find all references mentioned in the paper. The authors should check whether everything is correct

Minor Essential Revisions

Introduction:
“Affected individuals with mutations in EVC and EVC2 have the typical spectrum of features and are phenotypically indistinguishable (Ruiz-Perez et al. 2003); although EvC patient present typical phenotype in most cases. Recent findings by Uluncan et al…”: unclear to me what the authors try to say. Sentence should be refrased.

Discussion:
- “Therefore our findings might have impact on diagnosis and prevention of this condition in neighboring countries.”: a little overestimated may be, but true for the whole world probably, not only the region.

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Needs some language corrections before being published

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