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 would like to thank you for your efforts in improving our manuscript entitled” Molecular and clinical analysis of Ellis-van Creveld syndrome in the United Arab Emirates” for publication in BMC Medical Genetics. We made all the requested changes to the manuscript as show bellow and we therefore hope that it will be accepted for publication in the journal. Here is a point-by-point response:

Abstract:
We made the grammatical corrections as suggested

Introduction:
"malformed or absence of some glands and ducts" has been removed as suggested.
We also made all the other suggested grammatical changes

Methods:
The paragraph starting by “The criteria for inclusion…” has been changed as suggested.

Results:
All the grammatical and style changes suggested by the editor have been accepted and carried out.
We now indicate in the modified manuscript (legend of figure 1) the members that have been tested by Tompson et al., 2007. We also indicate the parents who were tested by us and were found to be heterozygous (Figure 1 legend).

Discussion:
We made all the suggested grammatical changes.
We also modified the text to clarify the uncertainty about the diagnosis in family 4.

No RNA is available to demonstrate the effects of the novel mutation on RNA splicing and we indicate that in the text.

Radiographs for family 4 are not available and we indicate that in the text.

In addition, we adopted the nomenclature style recommended by the Human Genome Variation Society (http://www.hgvs.org) for the reported mutations and we also added the accession numbers of the genes used in this nomenclature. Furthermore, the references we checked against the BMC Medical Genetics format.

Consent forms will be sent by fax or e.mail very soon.

The approval of the ethics committee including the approval numbers are is now stated in the manuscript (methods section).

We hope that this response is satisfactory for the acceptance of this manuscript in BMC Medical Genetics and we are looking forward to hearing from you soon.

Best Regards, Bassam Ali