Author's response to reviews

Title: EDAR mutation in autosomal dominant hypohidrotic ectodermal dysplasia in two Swedish families

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Author's response to reviews: see over
Dear Editor,

We would like to thank for the encouraging immediate review of our article:

**EDAR mutation in autosomal dominant hypohidrotic ectodermal dysplasia in two Swedish families**

We appreciate the constructive criticism of the reviewers. Please see the attached point-by-point response to the reviewers concerns.

**Respone to Minor Essential Revisions from Reviewer 1 (Cord Drögemüller)**

<table>
<thead>
<tr>
<th>1. The authors cite OMIM entries for anhidrotic ectodermal dysplasia but use the old designation of hypohidrotic ectodermal dysplasia (HED) please change.</th>
<th>The OMIM entries all give the name hypohidrotic as an alternative to anhidrotic ectodermal dysplasia. Since the patients studied by us exhibit varying degree of hypohidrosis but not anhidrosis we considered it misleading to call their disorder anhidrotic ED. However, for the sake of uniformity we could change to anhidrotic ectodermal dysplasia - if desired.</th>
</tr>
</thead>
<tbody>
<tr>
<td>2. The authors should adapt the nomenclature for the description of sequence variations to international recommended standards of the human genome variation society (<a href="http://www.hgvs.org/mutnomen/">http://www.hgvs.org/mutnomen/</a>).</td>
<td>Done</td>
</tr>
<tr>
<td>3. The introduction doesn’t mention the two different ectodysplasin A1 and A2 proteins binding to two distinct receptors (EDAR and XEDAR), please clarify.</td>
<td>Done</td>
</tr>
</tbody>
</table>
4. Are there already known EDAR mutations within other mammalian species causing anhidrotic ectodermal dysplasia, if so please report them. | Done
---|---
5. The individuals of the two family should be numbered within Figure 1 (generation I, II, III, and individual 1, 2, 3, ...) and it should be stated in the text which four individuals (e.g. I.1) were chosen for the initial screening of the eleven coding EDAR exons. | Done
6. Figure 2: Please sign the girl within Figure 1 (e.g. using an arrow) and clarify which family is family A. | Done
7. Table 1: The given wide range of optimal annealing temperatures should be explained. | This is a common phenomenon when using gradient PCR to optimize the annealing temperatures. Some PCR reactions have a very narrow temperature optimum whereas other reactions are much more forgiving temperature wise.
8. Figure 3a: Please describe the difference between the two lines in more details or discard this part of the figure. | Figure 3a has been discarded.
9. Figure 3b: Please indicate the influence of the SNP on the affected EDAR codon. | Done

**Minor Essential Revisions from Reviewer 2 (Wasim Ahmad)**

1. I do not agree that the mutation is in the hotspot region. | Done. – We do no longer speculate about a hotspot region.

Yours sincerely,

Marcus Schmitt-Egenolf