Reviewer's report

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

Version: 1 Date: 27 October 2006

Reviewer: Cord Drögemüller

Reviewer's report:

General

This paper describes an already known dominantly inherited EDAR mutation causing anhidrotic ectodermal dysplasia in two Swedish families. Therefore this paper represent a confirmation study providing supporting evidence for the relevance of these rare dominant mutation within EDAR towards the development of ectodermal structures. The authors also summarize all known human EDAR mutation from the literature. The short paper merits publication.

Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)

None

Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)

1. The authors cite OMIM entries for anhidrotic ectodermal dysplasia but use the old designation of hypohidrotic ectodermal dysplasia (HED), please change.
2. The authors should adapt the nomenclature for the description of sequence variations to international recommended standards of the human genome variation society (http://www.hgvs.org/mutnomen/).
3. The introduction doesn’t mention the two different ectodysplasin A1 and A2 proteins binding to two distinct receptors (EDAR and XEDAR), please clarify.
4. Are there already known EDAR mutations within other mammalian species causing anhidrotic ectodermal dysplasia, if so please report them.
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.
6. Figure 2: Please sign the girl within Figure 1 (e.g. using an arrow) and clarify which family is family A.
7. Table 1: The given wide range of optimal annealing temperatures should be explained.
8. Figure 3a: Please describe the difference between the two lines in more details or discard this part of the figure.
9. Figure 3b: Please indicate the influence of the SNP on the affected EDAR codon.

Discretionary Revisions (which the author can choose to ignore)

What next?: Accept after minor essential revisions

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

Quality of written English: Acceptable

Statistical review: No

Declaration of competing interests:

'I declare that I have no competing interests'