Reviewer's report

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

Version: 1 Date: 5 November 2006

Reviewer: WASIM AHMAD

Reviewer's report:

General

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

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

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

Lind et al has described two large Swedish families with hypohidrotic ectodermal dysplasia. A nonsense mutation Arg358X has been identified in both the families. The mutation was first described in 1999 by Monreal et al. in Nature Genetics. Clinical features of the patients are the same as reported earlier and so the mutation identified.

I do not agree that the mutation is in the hotspot region. The possibility that the American and Swedish families have the same ancestors can not be ruled out as described by the authors in the last para of the discussion.

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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 have no competing interests