Reviewer's report

Title: Hair shaft structures in EDAR induced ectodermal dysplasia

Version: 1

Date: 9 July 2015

Reviewer: Andrew Jheon

Reviewer's report:

In this article, Stecksén-Blicks et. al. analyzed hair from 17 individuals with the same autosomal dominant c.1072C>T mutation in EDAR from 3 Swedish families compared to 20 unaffected, related individuals from the same families and 32 age and gender matched, non-related controls. The authors found that the minimum and maximum hair diameters were smaller in the affected group carrying the EDAR mutation compared to the control group, and the affected group had a higher incidence of hair shaft deformations compared to both the unaffected relatives and control group. In addition, 2 of the individuals with the EDAR mutation had an interesting twisted hair phenotype. The strength of this case report is that the authors describe an interesting phenotype in a genetically homogenous group affected by hypohidrotic ectodermal dysplasia (HED). And the authors clinically (re)confirm that EDAR is involved in hair follicle development and cycling. I would recommend that this manuscript be accept for publication with minor revisions.

Minor Essential Revisions

1. In the results section on page 6, paragraph 1, line 1, define “scale structure” of hair.

2. Also in the results section on page 6, paragraph 2, line 1, define severity of deformation. What is the difference between deep and superficial?

Discretionary comments

3. In the discussion section on page 8, paragraph 1, it would be interesting to add a comparison with Edar mouse models (e.g., Sleek). Does the Edar mouse have a hair phenotype similar to the decreased diameter or twist hair phenotypes in HED individuals?

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: Yes, but I do not feel adequately qualified to assess the statistics.

Declaration of competing interests:
I declare that I have no competing interests.