Reviewer's report

Title: Hair shaft structures in EDAR induced ectodermal dysplasia

Version: 1 Date: 17 July 2015

Reviewer: Naveed Wasif

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Major Compulsory Revisions

Though the mutation is recurrent but the data of three families is appreciable. the work presented on hair shaft has added novel information regarding nonsense mutation in EDAR gene. I have following suggestions

1- in materials and methods a male individual completely bald but WITH KNOWN MUTATION has been described. but he was excluded from the study. was he investigated by the expert dermatologist? Was he bald since childhood or later stages of life? Was his biopsy done? If No, then i would suggest that his biopsy should be performed so that hair structures like hair papilla, hair bulb and hair follicle, if present, are being studied. this would help to define phenotypic variability among affected members of one family or group of affected individuals of same race.

2- I suggest that clinical picture of affected member whose hair shaft deformations are shown should be presented along with mutation sequencing chromatogram.

Minor Essential Revisions

1- Legends of figures 2, 3, 4 a-d are incomplete or not properly described. i would suggest figures should be in a panel of a,b,c....

2- In discussion where author has given reference 9, it seems as if this is his work. these sentences should be rephrased.

Discretionary Revisions

None

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

Quality of written English: Acceptable

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