Reviewer's report

Title: A Prevalent Mutation with Founder Effect in Spanish Recessive Dystrophic Epidermolysis Bullosa Families.

Version: 1 Date: 2 July 2010

Reviewer: Johann Bauer

Reviewer's report:

The authors report a population study in patients suffering from epidermolysis bullosa dystrophicans in Spain. Of 49 patients analysed, 27 carried at least one c.6527insC mutation. All of these c.6527insC mutations were associated with the H5 haplotype. This proves that these mutations were generated through a founder effect.

Minor comments:
Figure 1:
One would rather like to see the dermal part of the blister. Is there staining at the epidermal side of the blister?

Paucity and hypoplasia of hemidesmosomes: This is surprising. Usually in DEB there is absence of anchoring fibrils but no hypoplasia of hemidesmosomes.

Figure 2:
c. 6527insC in other countries is circled blue not purple (as described in the legend)

Literature:
There is a new classification paper of EB: Fine et al. JAAD, 2008

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

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

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