Reviewer's report

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

Version: 1 Date: 13 July 2010

Reviewer: Cristina Has

Reviewer's report:

The manuscript is well written and describes a COL7A1 founder mutation in the Spanish population. The question posed by the authors is well defined, the methods are appropriate.

Minor points:

1. The number of COL7A1 mutations differs in Abstract and Background. Actually, more than 500 mutations are known (https://portal.biobase-international.com/hgmd/pro/allmut.php.)

2. Page 4, Background, 5th sentence: is not clear to me

3. Page 5, SNPs selection and haplotyping assay: The novel polymorphism NM_000094.3 c.11639C>T in intron 19 is designated differently in Table 1 and 2. Please check this.

4. Page 6, Results: polymorphisms are described in respect to the sequence NM_000094.3, whereas on page 5 in respect to NG_007065.

5. Page 8, Discussion second paragraph: how do the authors know that the allele CCGCTCAAA_6527insC is found in other populations? Did the cited authors describe this haplotype?

6. Legend to figure 1:
   (A) a “C” should be included after 7 and before peaks. The mutation is actually a duplication.
   (B) Immunofluorescent staining with LH7.2 shows in the picture strongly reduced staining but not complete lack. Please mention if this is an unspecific staining at the blister margin.
   (C) Why are the hemidesmosomes hypoplastic?

7. Figure 3 is not explained properly in the legend.

8. Figure 4 is not absolutely necessary. Why was rs9878950 not considered?

9. Tables lack legends.

Level of interest: An article of importance in its field

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