Supplementary material to article by K. B. Gostyńska et al. "In-frame Exon Skipping in KRT5 due to Novel Intronic Deletion Causes Epidermolysis Bullosa Simplex, Generalized Severe"

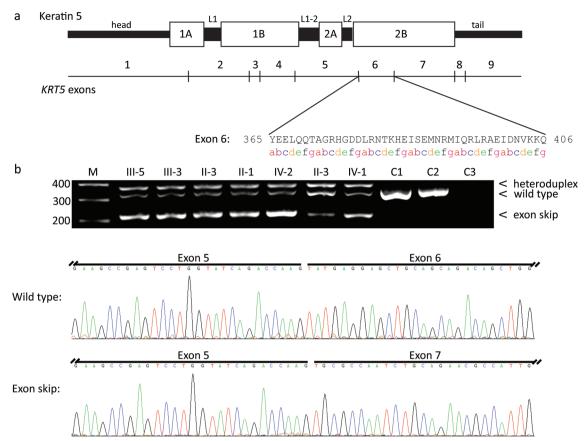


Fig. S1. Schematic representation of keratin 5 and mRNA studies. (a) Schematic representation of the keratin 5 polypeptide and, below, the 9 exons comprising the encoding gene KRT5. Exon 6 containing 42 amino acids (codons 365-406) comprises exactly 6 heptad repeats and is shown in coloured font. Using frozen skin biopsies, nested RT-PCR analysis (b) and Sanger sequencing of patient and healthy control mRNA. Lanes are marked with pedigree numbers of patients. C1 and C2: healthy control, C3: negative control. KRT5 primers used: forward 5'-CGCAACCTGGACCTGGATAG- 3' reverse: 5'-CCATGTCCTGCTTGGCCTTC- 3' (wild-type: 349bp length, exon skip 223-bp length).

Acta Derm Venereol 97, 2017