



Fig. S1. Laboratory findings. (a) Indirect immunofluorescence of a skin biopsy shows broad reticular labelling for collagen IV extending from the dermal-epidermal junction (DEJ) to the upper dermis in patient skin, compared with (b) staining confined to the DEJ, vessels and adnexa in normal control skin. (c) Several re-duplications of the basement membrane surrounding a dermal blood vessel are visible on ultrastructural examination. (d) DNA sequence analysis of the region spanning mutation c.735_736delTG in exon 7 and c.1446_1450delCCCTG in exon 10 (arrows indicate the starting point of the frameshifts). Scale bars: (a, b) 20 μ m and (c) 2.5 μ m. The authors would like to thank Gabriele Bacile and Maurizio Inzillo for preparation of the figures.