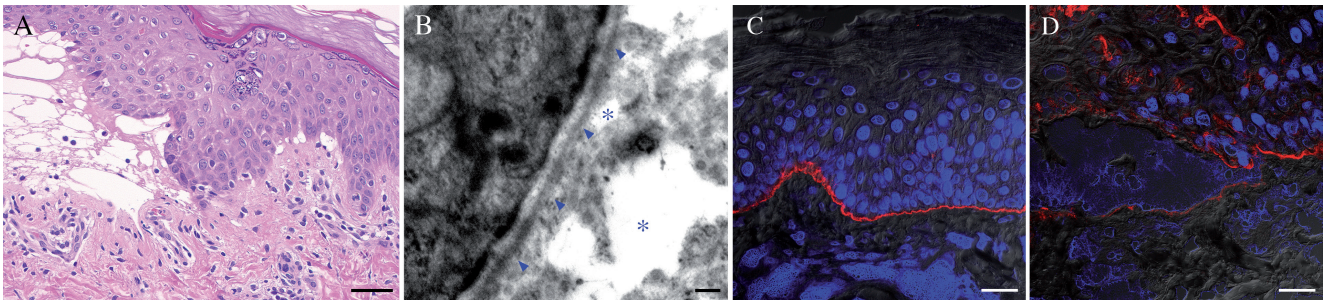

Fig. S1. Histology of the present case. (A) H&E stain. Subepidermal blisters and inflammatory infiltrates were noted in the upper dermis of a biopsy specimen from a dorsal lesion of her right foot; bar: 50 μm. (B) Ultrastructural analysis shows separation (asterisks) at the sublamina densa. Arrowheads: lamina densa; bar: 100 nm. (C, D) Localisation of type 7 collagen in intact skin (C) and in a blister (D). Sections were reacted with a monoclonal anti-type 7 collagen antibody (LH7.2) (1:2,000 dilution), a Cy3-labelled goat anti-mouse IgG (H+L) antibody (1:500 dilution) (Rockland Immunochemicals Inc., Gilbertsville, PA), and were then mounted using a ProLong® Gold antifade reagent with DAPI (Life Technologies Corp.). Confocal images were recorded using a laser scanning microscope LSM780 (Carl Zeiss Microscopy GmbH, Jena, Germany). Images are merged views for type 7 collagen (red), DAPI (blue) and differential interference contrast image (white). Type 7 collagen was positive at the basement membrane in intact skin (C), and was stained on both the epidermal and dermal sides in a blister (D); bar: 20 μm.

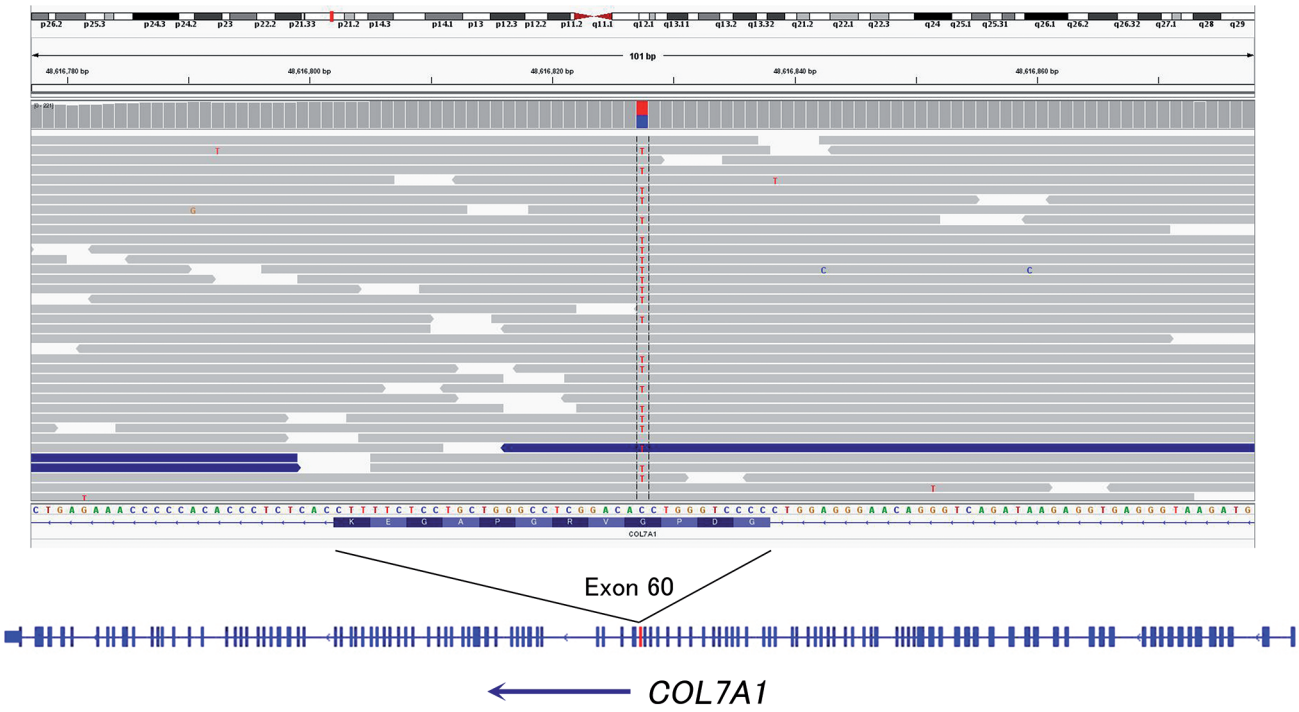

Fig. S2. Integrative genomics vie of a single nucleotide variation (SNV) in chr. 3:g.48616827C>T (negative strand) in *COL7A1* identified by next generation sequencing. The SNV corresponds to c.5282G>A in exon 60, which leads to a missense mutation of p.Gly1761Asp. No other novel SNV and indels were found in other genes examined, *KRT5, KRT14, DST, EXPH5, DSP, PLEC, LAMA3, LAMB3, ITGA3, LAMC3, ITGB4, ITGB6, COL17A1, CD151* and *FERMT1*.

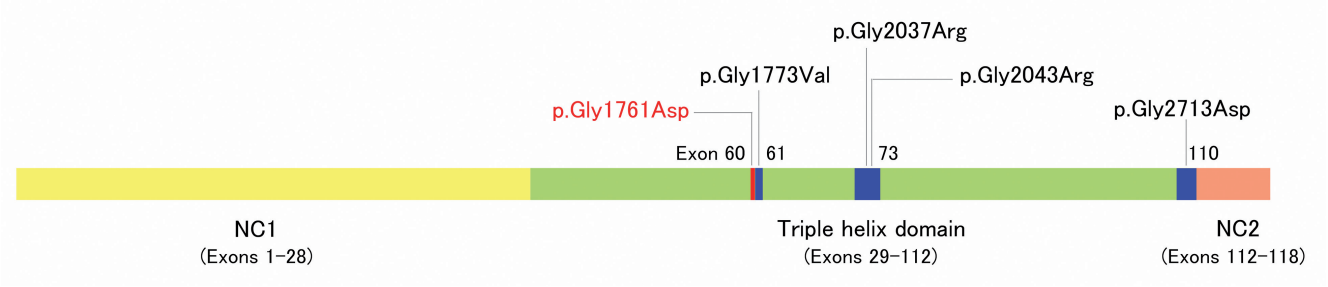

Fig S3. Mutations of *COL7A1* in localised DDBE including 4 previously reported mutations and a novel mutation in the present case. Those mutations occur at a Gly residue of Gly-X-Y triplet repeats in the triple helix domain of type 7 collagen, which is encoded by exons 60 (the present case), 61, 73 and 110 of *COL7A1*.