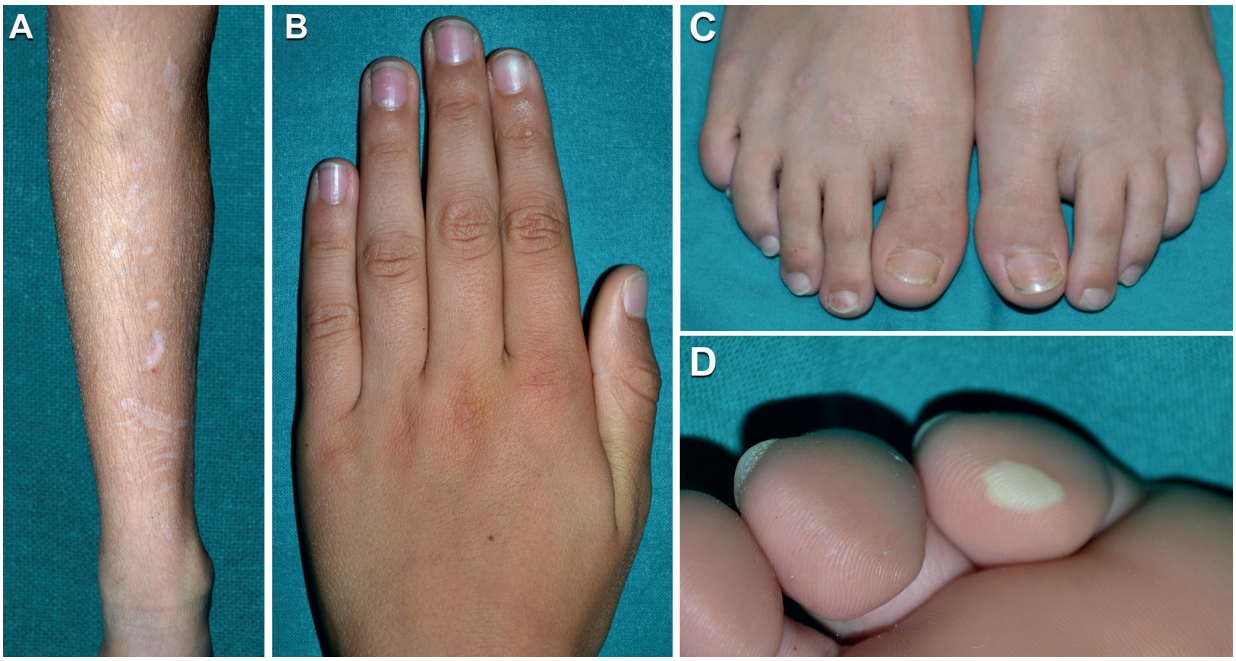

Fig. S1. Patient's clinical features at age 10 years. (A) Hypopigmented scars following trauma-induced blistering on the right leg. (B, C) Finger- and toe-nails are not dystrophic. (B, C) Note the normal appearance of the left hand and only minimal scars of the right foot. (D) A small blister on the ventral surface of the fifth left toe due to rubbing on footwear.

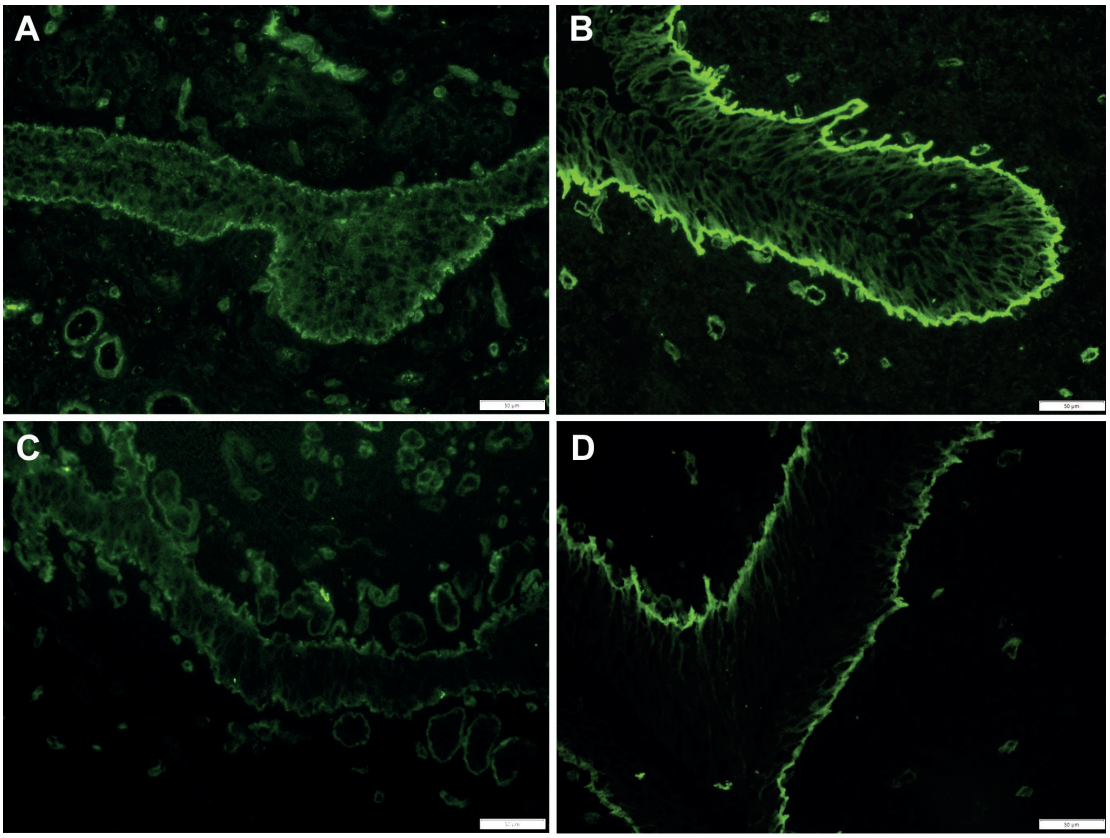

Fig. S2. Immunofluorescence antigen mapping on ureter sample. The expression of β4 (A, B monoclonal antibody 3E1) and α6 (C, D, monoclonal antibody G0H3) integrin subunits along the urothelial-chorion junction is markedly reduced in the patient (A, C) compared with normal control ureter (B, D) (indirect immunofluorescence, bars= 50 μm).

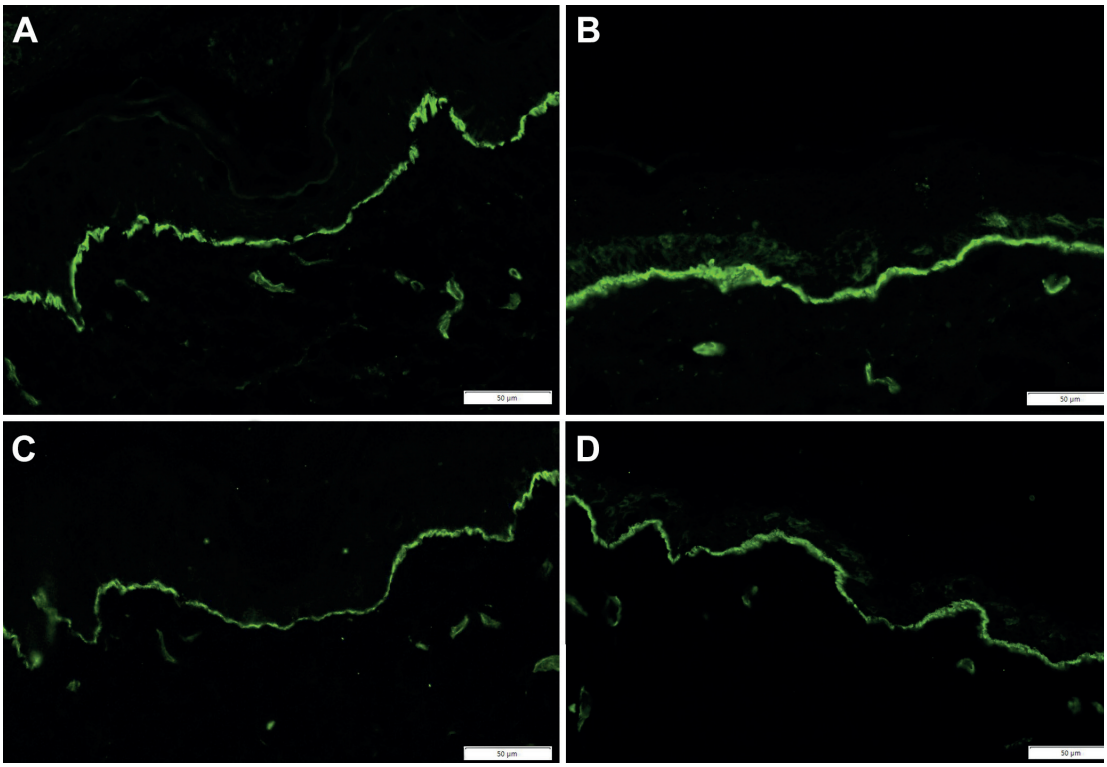

Fig. S3. Immunofluorescence antigen mapping on skin biopsy. The expression of β4 (A, B, monoclonal antibody 3E1) and α6 (C, D, monoclonal antibody G0H3) integrin subunits along the cutaneous basement membrane zone is slightly reduced in the patient's skin (A, C) compared with normal human control skin (B, D). Expression of other epidermal major adhesion protein components (keratins 5 and 14, plectin, collagen XVII, laminin-332 chains and collagen VII) was comparable with control skin (not shown) (indirect immunofluorescence, bars=50 μm).

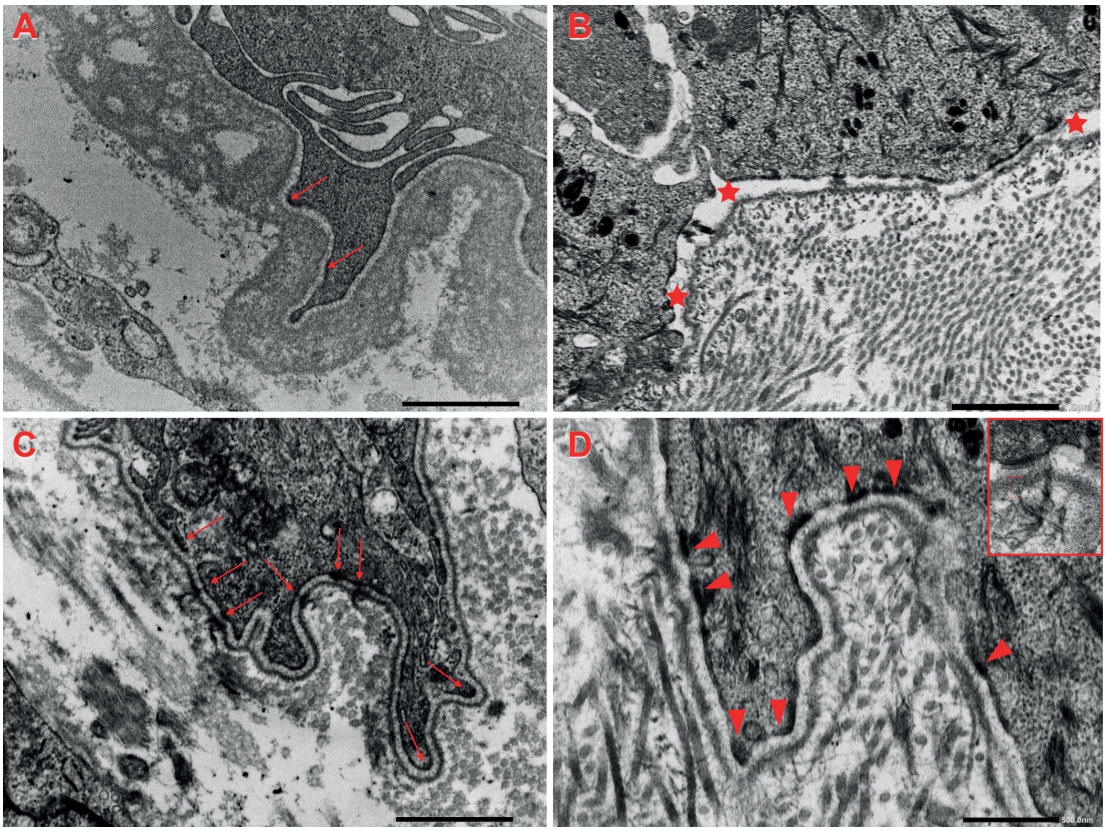

Fig. S4. Ultrastructural findings in ureteral and skin samples. Reduced number of hypoplastic hemidesmosomes (*arrows*) along the plasma membrane of urothelial cells, together with markedly thickened urothelial basement membrane in (A) the patient compared with (C) a normal control ureter. (B) Initial cleavage within the lamina lucida of the cutaneous membrane zone (*asterisks*) in patient's skin diagnostic for junctional epidermolysis bullosa. Basal keratinocyte hemidesmosomes appear well represented and only mildly hypoplastic, mostly lacking sub-basal dense plates (*arrowheads*) (D); the inset in (D) shows a typical hemidesmosome with sub-basal dense plate in normal human skin. Bars=1.0 μm in (A–C), and 0.5 μm in (D).

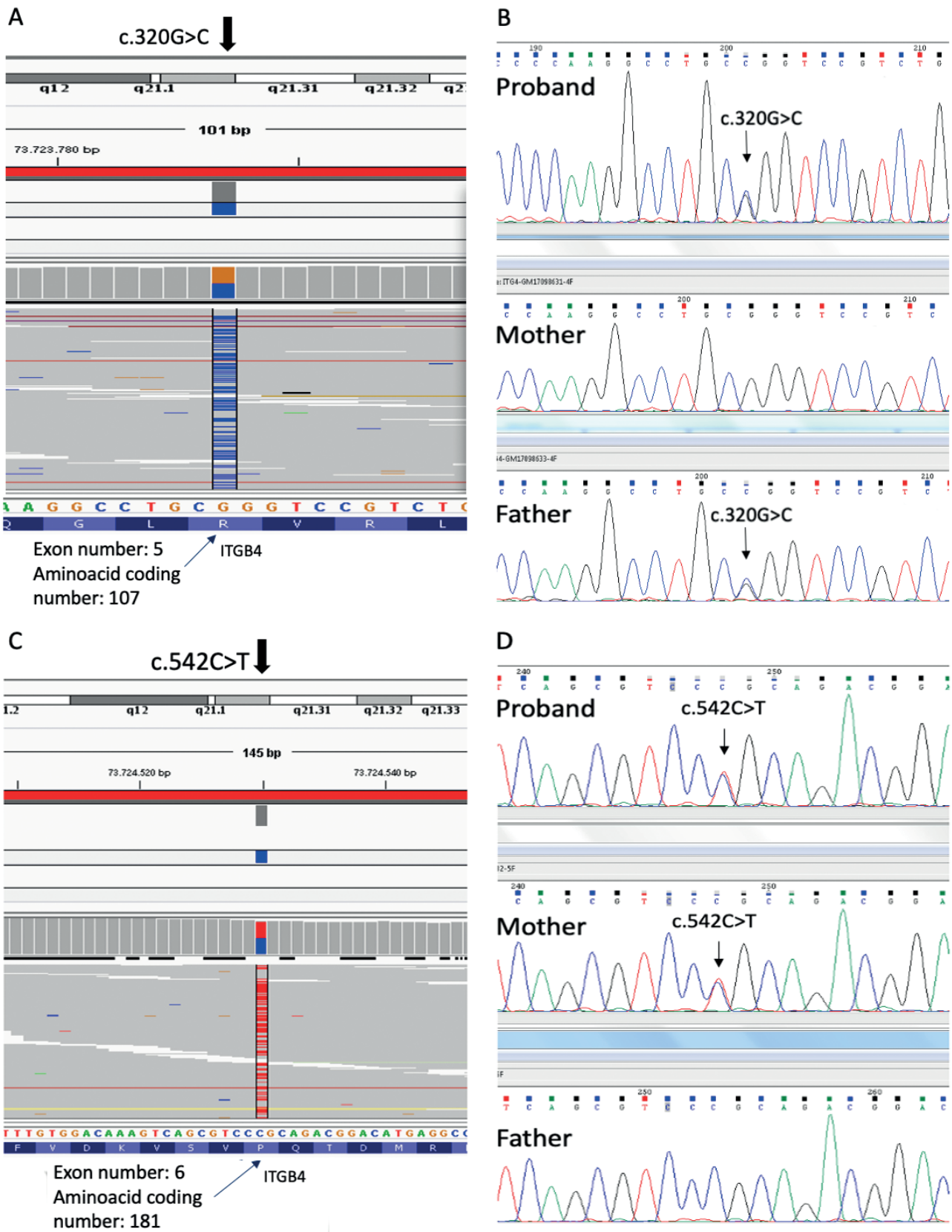

Fig. S5. Molecular genetic testing. Next generation sequencing (NGS) analysis shows the *ITGB4* (NM_000213.3) variant c.320G>C, p.Arg107Pro at the heterozygous state in (A) the patient, and Sanger sequencing confirms its presence at the heterozygous state in (B) the patient and his healthy father. (C) NGS identifies the heterozygous variant c.542C>T, p.Pro181Leu in the *ITGB4* gene in the patient, and (D) Sanger sequencing shows its presence at the heterozygous state in both the patient and his healthy mother.