



Fig. S2. Immunofluorescence antigen mapping and electron microscopy findings in cases 1 and 2. Immunofluorescence labelling for keratin 14 shows basal keratinocyte vacuolization and initial blistering in the epidermal basal layer (a) in case 1 at 7 years of age and (b) in case 2 at 2 weeks of age. Electron microscopy examination reveals reduced tonofilaments and initial vacuolization in basal keratinocytes in case 1 at 40 days of age (c, d), the reduction in tonofilaments persists at the age of 7 years in the same patient (e); cleavage within the epidermal basal layer in case 2 (f). Bars: (a, b) 200 μm , (c) 5 μm , (d) 1 μm , (e, f) 2 μm .