



Fig. S2. Allogeneic haematopoietic cell transplantation outcome in Patient 2: (1A–F) Clinical photographs of patient 2 at baseline (Day 0) showing cutis aplasia (1A); skin blisters and nail dystrophy (1B–E) and extensive mucosal erosions (1F). (2A–E): Clinical photographs of patient 2 (day +180) showing completely healed skin lesions (2A–D) and partially healed mucosal erosions (2E). (3A–E) Immunofluorescence antigen mapping (IFM) showing normal staining for type VII collagen (green) (clone: monoclonal antibody LH7:2, Sigma-Aldrich, Poole, UK) in control tissue (3A) and complete absence in unaffected skin of patient 2 at Day 0 (3B), Day +180 (3C) and post-mortem (Day +283, 3E). IFM from healed skin at Day +180 also did not show any Col7 expression (3D). (4A–E) Transmission electron micrographs (TEM) showing lack of anchoring fibrils at the dermo–epidermal junction in unaffected skin of patient 2 at Day 0 (4B), Day +180 (4C) and post-mortem (Day +283, 4E) compared with control tissue (4A). TEM from healed skin at Day +180 also did not show any anchoring fibril formation (4D).