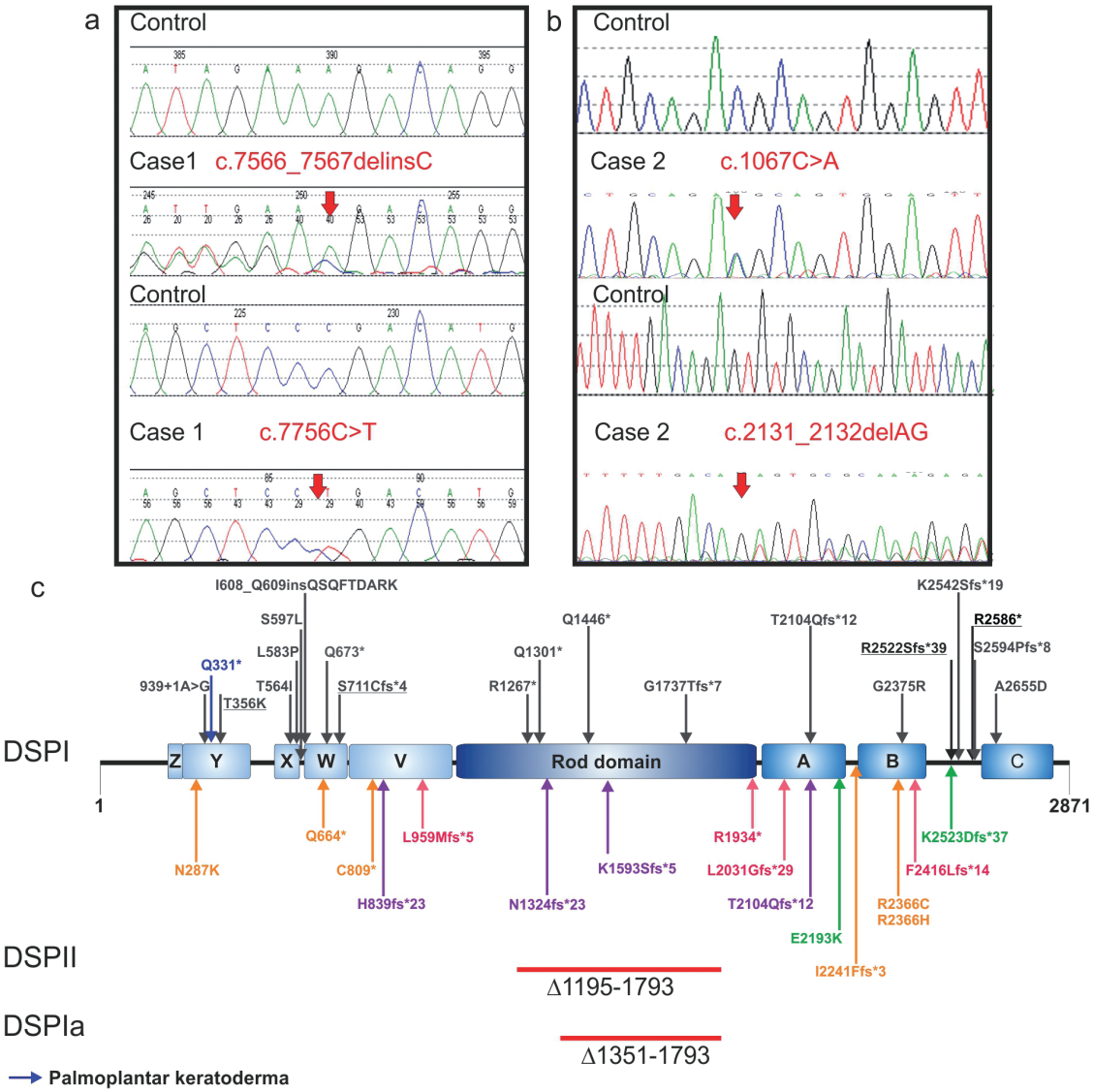

Fig. S1. Desmoplakin (*DSP)* mutations in this study. (a) Two *DSP* mutations, c.7566_7567delinsC (p.R2522Sfs*39) and c.7756C>T (p.R2586*) were disclosed in case 1. (b) Two heterozygous *DSP* mutations, c.1067C>A (p.T356K) in exon 9 and c.2131_2132delAG (p.S711Cfs*4) were disclosed in case 2 and 3. (c) Desmoplakin is composed of the N-terminal plakin domain with 5 α-helical bundles (Z, Y, X, W, V), the coiled-coiled rod domain, and the C-terminal plakin domain with the 3 plakin-repeat subdomains (A, B, C) (15, 16). The amino acids absent in the shorter isoforms, DSPII and DSPIa, are indicated below. *DSP* mutations are associated with a broad spectrum of clinical features, particularly involving the skin. The novel mutations identified in this study are underlined. Previously reported skin phenotypes include: palmoplantar keratoderma (blue), palmoplantar keratoderma, woolly hair (black), palmoplantar keratoderma, woolly hair, cardiac disease (grey), hair shaft abnormalities and cardiac disease (green), skin fragility, palmoplantar keratoderma, woolly hair, cardiac disease (pink), skin fragility, woolly hair, palmoplantar keratoderma (orange), and severe skin fragility (red).