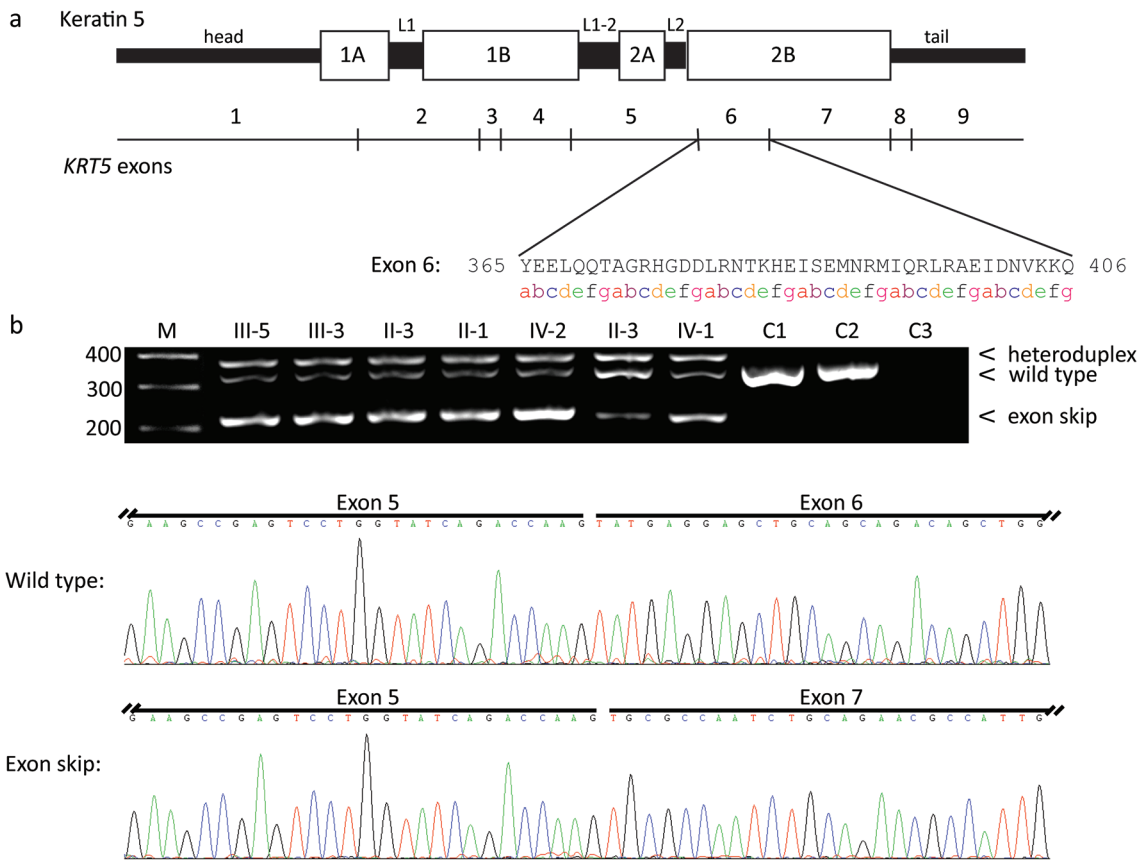

Fig. S1. Schematic representation of keratin 5 and mRNA studies. (a) Schematic representation of the keratin 5 polypeptide and, below, the 9 exons comprising the encoding gene *KRT5*. Exon 6 containing 42 amino acids (codons 365–406) comprises exactly 6 heptad repeats and is shown in coloured font. Using frozen skin biopsies, nested RT-PCR analysis (b) and Sanger sequencing of patient and healthy control mRNA. Lanes are marked with pedigree numbers of patients. C1 and C2: healthy control, C3: negative control. *KRT5* primers used: forward 5′-CGCAACCTGGACCTGGATAG- 3′ reverse: 5′-CCATGTCCTGCTTGGCCTTC- 3′ (wild-type: 349-bp length, exon skip 223-bp length).