



Fig. S1. Results of genetic analysis. (a) Sanger sequencing confirms a heterozygous variant, c.1373+2T>C, in *KRT10* in case 2. It is not present in DNA from the unaffected father. (b) By using RNA extracted from hair samples of cases 2 and 3, PCR products from exon 6 to intron 6 of *KRT10* demonstrated a single band, whereas no PCR product was obtained from RNA of the normal control by using this primer pair. (c) Sequencing of cDNA from exon 6 to intron 6 of *KRT10* indicates aberrant splicing at the exon 6–intron 6 boundary leading to p.Ser458Argfs*6.