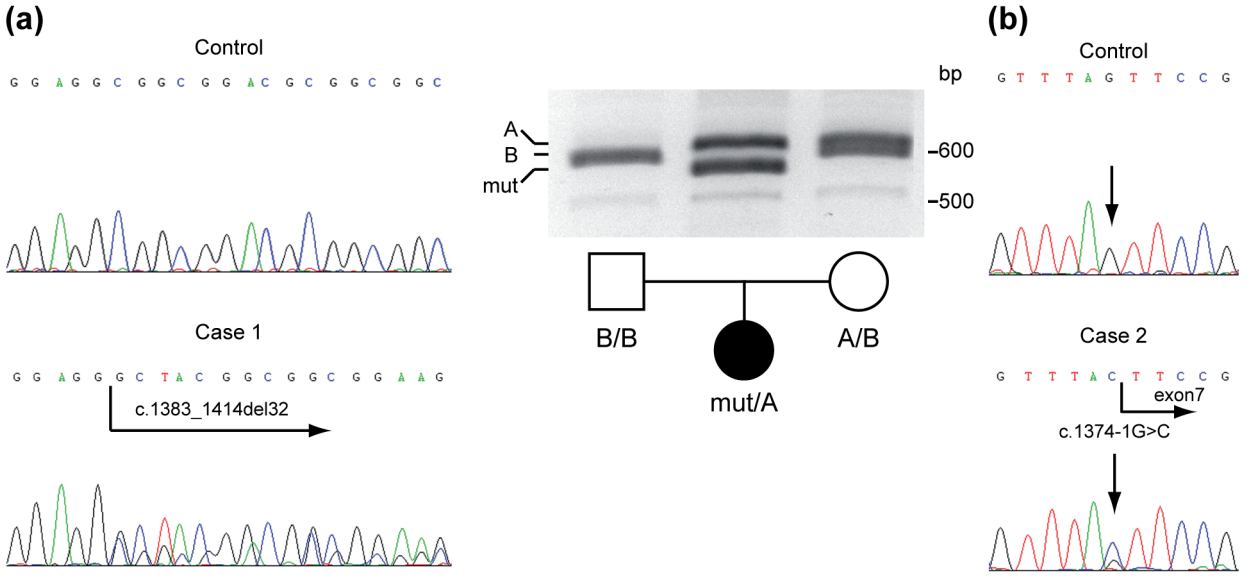

Fig. S2. Molecular analysis of the *KRT10* gene. (a) Case 1: sequence analysis of the genomic region spanning exon 7 and its intronic borders revealed the heterozygous c.1383_1414del32 frame-shift mutation (left panel). Length polymorphism analysis and gel electrophoresis of PCR amplified fragments encompassing exon 7 and flanking intronic borders. The family pedigree summarises allele segregation. Primers used to amplify the DNA are expected to generate a PCR product of 635 bp (allele A). This amplicon is shorter in the father, since he is homozygous for a known polymorphic in-frame variant c.1468_1479del12 (allele B). The mother is compound heterozygous for allele A and allele B. The proband inherits allele A from the mother, while the size of the other fragment (591 bp) demonstrates that the c.1383_1414del32 mutation arises *de novo* on the paternal allele in linkage with the c.1468_1479del12 polymorphic variant (allele mut) (right panel). (b) Case 2: sequence analysis of KRT10 intron 6/exon 7 border shows the heterozygous c.1374-1G>C mutation, which changes the obligate G of intron 6 acceptor site. A normal sequence is shown for comparison.