



Figure S1. Keratin 1 (K1), K2 and K10 structural organization with the position of the pathogenic variants identified in this study. Novel mutations are underlined.

p.Ser598Lysfs*56	→	ATA AAG AGA TGC CCT CTG TTT CAT TAG
p.Tyr587Leufs*67		Ile Lys Arg Cys Pro Leu Phe His *
p.Gly585Trpfs*69		

Figure S2. *KRT1* frameshift mutations (listed on the left) that lead to stop codon readthrough resulting in an identical extension of 7 amino acids after the readthrough (right). The novel mutation described in this study is depicted in red; the natural stop codon is in bold.