



Fig. S1. Genetic data supporting mosaic mutations. Sequencing analysis of: (a) genomic DNA isolated from peripheral white blood cells of *KRT1* of the male patient; and (b) *KRT10* of the female patient. Genetic analysis was performed by (a) Sanger sequencing or (b) next-generation sequencing using a Personal Genome Machine (PGM, Ion Torrent, ThermoFisher Scientific) and an AmpliSeq Custom Panel. The chromatogram in (a) shows a sequence corresponding to the mutated allele in low proportion compared with the normal sequence of *KRT1*. In (b) 226 reads were aligned with the reference genome as provided by the Integrative Genomics Viewer (IGV v.2.1, Broad Institute) software; 84 reads were carrying the mutation in *KRT10*, indicated with a green square.