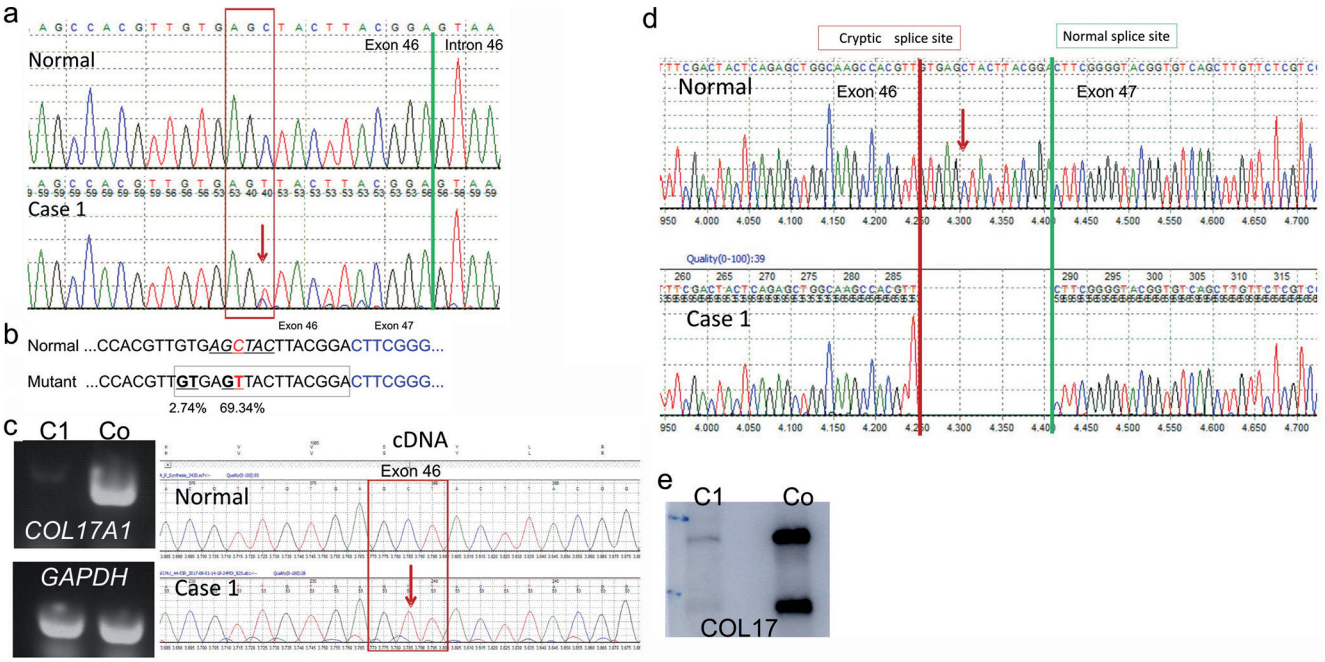

Fig. S1. *COL17A1* variant c.3198C>T and its consequences. (a) Sanger sequencing demonstrates the c.3198C>T variant (*arrow*) in the genomic DNA of case 1. (b) *In silico* predictions indicate: (*i*) increase of the activity of the exonic cryptic donor splice site GTTgtgagt (underlined, Mutation Taster: from 0.2185 to 0.2356; Human Splicing Finder http://www.umd.be/HSF3/ 2.74% increase); (*ii*) formation of a new donor splice site TGAgttact (underlined, Human Splicing Finder 69.34% increase), and (iii) alteration of an exonic splicing enhancer (ESE) site, *agctac* (italics, Human Splicing Finder). (c) RT-PCR (*left*) shows low levels of *COL17A1* mRNA in keratinocytes of case 1 (C1), compared with control keratinocytes (Co); *GAPDH* as a control. In the right panel the sequence shows decay of the allele carrying the mutation c.2407G>T. (d) The splicing anomaly leads to removal of 16 nucleotides c.3193_3208del, frame shift and formation of a premature termination codon, p.Val1065Leufs*35 in the NC5 domain of type XVII collagen. (e) Immunoblot showing type XVII collagen in lysates of keratinocytes of case 1 (C1) and control (Co) keratinocytes. The upper bands represent the full-length (180 kDa) and the lower bands represent the shed ectodomain (120 kDa) of type XVII collagen.