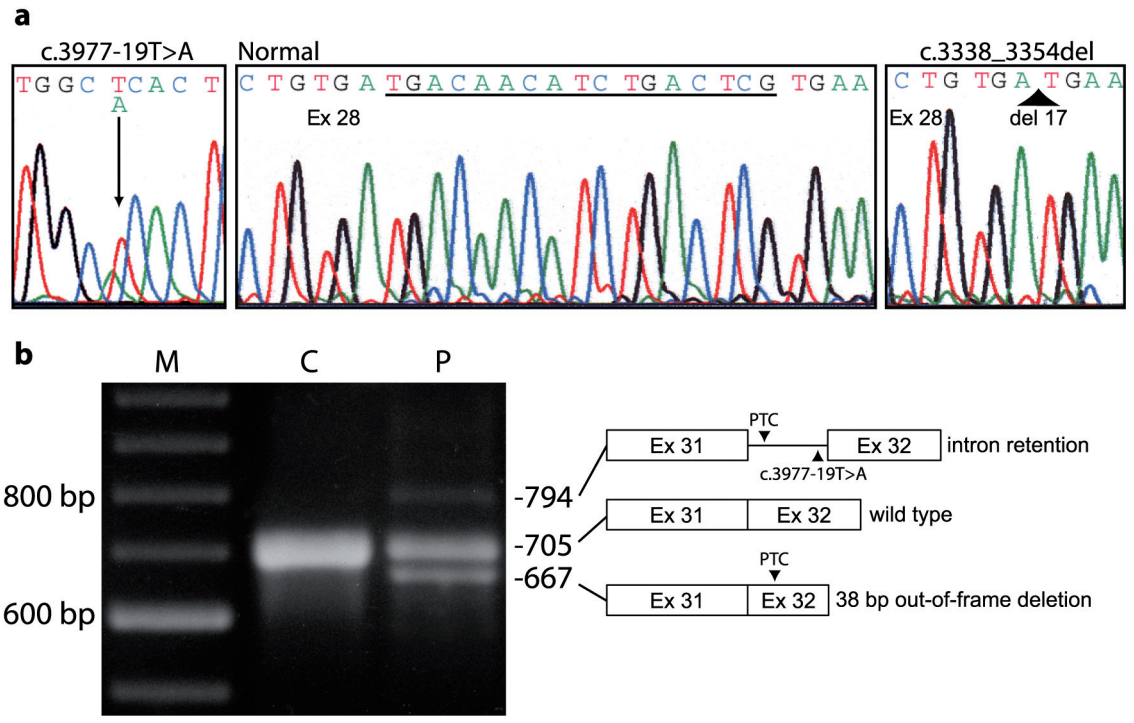

Fig. S2. Sequence chromatograms of compound heterozygous c.3977-19T>A (*arrow, left panel*) and c.3338_3354del (*middle and right panels*) mutations. (a) Sequence for c.3338_3354del is shown in the antisense orientation. (b) Reverse-transcriptase (RT)-PCR analysis in patient's keratinocytes. Agarose gel electrophoresis of the RT-PCR products (generated with primers 5'-gcgactatgagatgaaggtg and 5'-gtgagttgtagtcccgtgtg spanning nucleotides 3756 to 4460 of the ITGB4 mRNA) identifies a wild-type transcript (705-bp band) in both the patient (P) and a healthy control (C). In contrast, 2 abnormal bands of 794-bp and 667-bp are evident only in the patient. The top band corresponds to an mRNA transcript that retains intron 31 and bears a premature termination codon (PTC). The lower and more abundant band identifies transcripts with a 38-bp deletion, which results from the usage of a cryptic acceptor splice site in exon 32 (9).