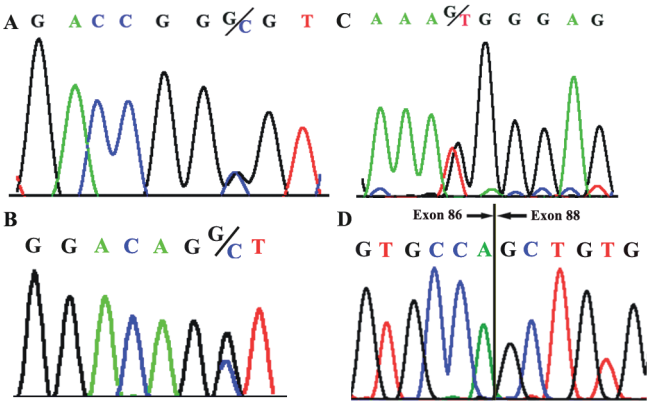

Fig. S2. Sequencing results of the three DEB-pr probands. (A) Proband 1, heterozygous transversion mutation of c.5317G>C in exon 61, resulting in the amino acid change of G1773R. (B) Proband 2, heterozygous transversion mutation of c.6900+1G>C in intron 87, causing abnormal splicing of COL7A1 mRNA. (C) Proband 3, heterozygous transversion mutation of c.8101G>T in exon 109, resulting in the amino acid change of G2701W. (D) Proband 2, sequencing result of the lower band from RT-PCR (Fig. S1) showing abnormal splicing of COL7A1 mRNA, the 3' end of exon 86 connecting to the 5' end of exon 88.