



Fig. S4. Sequence chromatograms of *COL7A1* amplicons showing the novel glycine substitution mutations identified in patients with bullous dermolysis of the newborn (BDN): (a) p.Gly2431Val (dominant) in case 3, (b) p.Gly1830Arg (dominant) in case 4 and (c) p.Gly2216Glu (recessive) in case 1. The affected codon is underlined. Genotype-phenotype correlations in BDN are elusive. In our recessive cases, mutations c.4783-1G>A and c.497dupA combine with p.Pro1699Leu and p.Gly2216Glu, respectively. Both the c.4783-1G>A and c.497dupA are null mutations. The p.Pro1699Leu was previously reported in compound heterozygosity with a different splice site null mutation in an adult patient with the pretibial subtype of recessive dystrophic epidermolysis bullosa (DEB) and presenting keratinocyte cytoplasmic deposits of collagen VII in the skin (37). A similar mutation, p.Pro2259Leu, was recently described in another recessive BDN patient (see reference 28 in Table S1¹). It remains to be determined, however, why the p.Pro1699Leu mutation may result in different DEB subtypes. Interestingly, Murase et al. reported a family with dominant DEB due to the p.Gly2242Glu mutation (see reference 26 in Table S1¹). In this family the disease manifested as BDN in an infant and DEB pruriginosa in his mother. These examples underscore the impact of modifier genes or environmental factors in inter- and intra-familial clinical DEB variability. Glycine substitution mutations can have different degrees of severity and result in either dominant or recessive inheritance (38). In addition, the same glycine substitution may behave as a dominant mutation or be silent within the same nuclear family (see reference 24 in Table S1¹).