



Fig. S2. Integrative genomics view of a single nucleotide variation (SNV) in chr. 3:g.48616827C>T (negative strand) in COL7A1 identified by next generation sequencing. The SNV corresponds to c.5282G>A in exon 60, which leads to a missense mutation of p.Gly1761Asp. No other novel SNV and indels were found in other genes examined, KRT5, KRT14, DST, EXPH5, DSP, PLEC, LAMA3, LAMB3, ITGA3, LAMC3, ITGB4, ITGB6, COL17A1, CD151 and FERMT1.