



Fig. S1. (a, b) Agarose gel analysis and (c, d) direct sequencing of mRNA amplified by nested PCRs in the proband and her mother. The mutation c.488delG in exon 5 of *LAMA3* has been amplified using a forward primer located in exon 1 (bp 175) and a reverse primer located in exon 8 (bp 815). The mutation c.4484C>T in exon 33 of *LAMA3* has been amplified using a forward primer located in exon 31 (bp 4177) and a reverse primer located in exon 35 (bp 4810). Gel analysis of the exons surrounding (a) mutation c.488delG, and (b) mutation c.4484C>T, showed no alternatively spliced products in the proband (lane 2) and her mother (lane 3), compared with control (lane 1). (c) Direct sequencing of the amplified mRNA showed the presence of mRNA carrying the mutation c.488delG in the proband (P) and her mother (M). (d) In the proband (P) both mRNA with the mutation c.4484C>T as well as mRNA with the mutation c.488delG are produced. Her mothers sequence showed no mutation (M).