



Fig. S2. *FERMT1* mutation analysis reveals the novel mutation c.1564 delC in exon 12 of the *FERMT1* gene in the maternal allele (a). RT-PCR using mRNA from normal human keratinocytes (1), normal human fibroblasts (2) and fibroblasts from the patient (3). Some amount of *FERMT1* mRNA is detected in the normal human fibroblasts (arrow); in contrast, no PCR product is observed in the fibroblasts from the patient (b).