



Fig. S1. (a) Next-generation sequencing identified the deletion of whole exon 6 of ectodysplasin-A (*EDA*). (b) PCR products for the female control (*lane 2*), father (*lane 3*), mother (*lane 4*) and proband (*lane 5*). The 232-bp band represents *GAPDH* and 362-bp band represents the exon 6 of *EDA*. (c) qPCR shows the copy number of *EDA* is 2 for female controls (PC), 1 for the proband's father and mother, and 0 for the proband. (d) Protein structure of *EDA*. The tumour necrosis factor (TNF) domain of mutant type *EDA* was deleted and the new reading frame stopped early with only 14 amino acid residues (IFQVECSMTGLASL).