



Fig. S1. Histopathological and immunofluorescence analysis of a skin biopsy in Case 2. Acanthotic and hyperkeratotic epidermis with a reduced granular layer and focal parakeratosis (a). Note the presence of cytoplasmic vacuolisation in several suprabasal keratinocytes (a). Immunofluorescence labelling for keratin 10 reveals a markedly reduced staining of suprabasal epidermis (b) as compared to normal control skin (c). Note the dotted nuclear labelling for keratin 10 of numerous suprabasal keratinocytes (b). (a) HE staining, original magnification X200(b, c), nuclear DAPI counterstaining, original magnification $\times 200$.