



Fig. S1. (A) A 4-mm punch skin biopsy from the index patient taken from her anterior chest shows a slight papillomatosis and acanthosis with a parakeratotic thickened stratum corneum. High power (*inset*) shows perinuclear vacuolization with intact cell membranes in the keratinocytes of the superficial epidermis. In superficial areas these changes are focally band like, but identical changes are seen in scattered single keratinocytes deeper in the stratum spinosum. The granular layer is missing. (B) A skin biopsy from the father taken from a linear hyperkeratotic streak at the edge of the right palm shows acanthosis and papillomatosis with columns of parakeratosis between the papillae. High power (*inset*) of areas with parakeratosis shows perinuclear vacuolar changes in the superficial keratinocytes focally in a confluent band, but there is no necrosis, and the cell membranes appear intact. Similar changes are seen in scattered keratinocytes deeper in the suprabasal epidermis. The thickness of the stratum granulosum is reduced. Light microscopy, hematoxylin-eosin staining. Original magnification $\times 25$, $\times 200$ (*insets*).