



**Fig. S1.** (a) Epidermal acanthosis with compact hyperkeratosis and hypergranulosis was noted. Dermal inflammatory cell infiltrates were mild. Bar=50  $\mu$ m. (b, c) Immunofluorescence for NIPAL4/ichthyin in: (b) healthy control skin, and (c) the patient's skin. A rabbit anti-ichthyin antibody (1:10 dilution) (Santa Cruz Biotechnology Inc., Santa Cruz, CA, USA) and a Cy<sup>3</sup>-labelled goat anti-rabbit IgG (1:500 dilution) (Kirkegaard & Perry Laboratories, Inc. Gaithersburg, MD, USA) were used. Sections were mounted using ProLong<sup>®</sup> Diamond anti-fade reagent with DAPI (Thermo Fisher Scientific Inc., Waltham, MA, USA). Fluorescence images were recorded using a confocal laser scanning microscope LSM780 (Carl Zeiss Microscopy GmbH, Jena, Germany). Images are merged views of NIPAL4/ichthyin (red), DAPI (blue) and DIC images (white). Bar: 20  $\mu$ m.