**Appendix S1**

## SUPPLEMENTARY MATERIALS AND METHODS

*Whole-exome sequencing*

Exome capture was performed by in-solution hybridization using SureSelect Human All Exon V5 bait (Agilent Technologies, Santa Clara, CA, USA). Massively parallel sequencing was performed with the Illumina HiSeq2500 platform with 150-bp paired end-reads (Illumina, San Diego, CA, USA). The reads produced were aligned to the hg19 reference human genome using the Burrows–Wheeler Aligner software with default parameters and a –mem option. PCR duplicates were removed using MarkDuplicates in Picard tools (https://broadinstitute.github.io/picard/). Candidate variants were called using VarScan2 (http://massgenomics.org/varscan) and annotated using ANNOVAR (http://annovar.openbioinformatics.org/). Common variants defined by >1% minor allele frequency in ExAC (http://exac.broadinstitute.org/), 1000 genomes (http://www.1000genomes.org/), or ESP6500 (http://evs.gs.washington.edu/EVS/) were excluded from analysis.

*Immunohistochmistry*

Immunohistochemical analysis of skin samples from the participants was performed as described previously (S1), with slight modifications. Thin sections (3 μm) were cut from samples embedded in paraffin blocks. The sections were soaked for 20 min at room temperature in 0.3% $H_2O_2$/methanol to block endogenous peroxidase activity. After washing in PBS with 0.01% Triton X-100, the sections were incubated for 30 min in PBS with 4% BSA followed by incubation overnight with the primary antibodies, 2 anti-keratin 10 (K10) antibodies, ab218903, produced against full-length human K10 (Abcam, Cambridge, UK) and OAAB03764, produced against the amino acid sequence (150-179) from the N-terminal region of human K10 (Aviva Systems Biology Corporation, San Diego, CA, USA) and an anti-keratin 1 (K1) antibody, ab111471 (Abcam, Cambridge, UK), in phosphate-buffered saline (PBS) containing 1% BSA. After washing in PBS, the thin sections were stained with avidin-conjugated goat anti-mouse (for K10, ab218903) or rabbit (for K10 (OAAB03764) and K1 (ab111471)) immunoglobulin secondary antibodies for 1 h at room temperature and washed in PBS. The Vectastain Elite ABC-PO kit (Vector Laboratories, Burlingame, CA, USA) was used for staining.

## SUPPLEMENTARY REFERENCE

S1. Takeichi T, Sugiura K, Nomura T, Sakamoto T, Ogawa Y, Oiso N, et al. Pityriasis rubra pilaris type V as an autoinflammatory disease by CARD14 mutations. JAMA Dermatol 2017; 153: 66–70.