**Appendix S1**

## MATERIALS AND METHODS

After obtaining informed consent, ethylenediaminetetraacetic acid (EDTA)-blood samples, and skin biopsies were taken from the index case. The study was approved by the ethics committee of the University of Freiburg, and conducted according to the principles of the Declaration of Helsinki.

Mutational analysis of the coding region and exon-intron boundaries of the *DST* gene was performed as described previously (9). Sanger sequences for the DNA were compared with reference sequences from the NCBI Entrez Nucleotide database (NM_001723.4; NG_029322.2). The primers used in this study are listed in **STable I**.

Normal skin specimens, obtained with the informed written consent of individuals who underwent surgery, were used for cell isolation and immunostaining. Keratinocytes derived from normal and patient's skin were immortalized with the HPV E6 and E7 genes (plasmids were gifts from Fernando Larcher and Stephanie Löffek) and cultured in keratinocyte growth medium (Invitrogen, Karlsruhe, Germany).

Immunofluorescence staining of the skin and cells was performed as described previously (10, 11). Confluent cell monolayers were lysed and immunoblotted as described previously (12). Primary and secondary antibodies are listed in **STable II**.

**STable I. Primers used in this study**

| Primer name | Sequence 5′-3′ | Primer name | Sequence 5′-3′ | Primer name | Sequence 5′-3′ |
|---|---|---|---|---|---|
| *Genomic DNA* | | BPAG1e-ex19R | ggaagatgaggagcatgactg | *cDNA* | |
| BPAG1e-ex1F | tgccacttttcaccgttaga | BPAG1e-ex20F | agtgcaaagggctaaggagc | GAPDH | gaaggtgaaggtcggagtca |
| BPAG1e-ex1R | aagtgctgcccatcaaaaag | BPAG1e-ex20R | aaacactgagtgaaatgacttgc | | ttgaggtcaatgaaggggtc |
| BPAG1e-ex2F | gcagcacttttcaggaatgg | BPAG1e-ex21F | gtaatgcattgtgccacgac | DST | ttgacacccttagagatagctaagaac |
| BPAG1e-ex2R | caaacgcacagtgactcctc | BPAG1e-ex21R | cccttcccctctttctctca | | tcatccctgactgaacatgc |
| BPAG1e-ex3F | tttcaggttgttttgaaacttg | BPAG1e-ex22F | gctgtctgaaggcctagttaatg | Col 17A | tctgaaggggatcatcaag |
| BPAG1e-ex3R | ttcaaatgcccatgaaacag | BPAG1e-ex22R | catgttttgctttcccaaac | | accctgaactccggataggt |
| BPAG1e-ex4F | ttcactctttgttacaagtgcc | BPAG1e-ex23aF | tggtgatagcgcttgttctg | ITGA6 | ggcggtgttatgtcctgagt |
| BPAG1e-ex4R | ggtccagttgtaccttcagacag | BPAG1e-ex23aR | tgcgaaaattcaggaggttc | | catgctcagtctctccacca |
| BPAG1e-ex5/6F | attgggttgatgagatgcag | BPAG1e-ex23bF | taccgcagggaacttgaaac | ITGB4 | gaggtaggtccaggacggg |
| BPAG1e-ex5/6R | tcttgattcctggatgtattctttag | BPAG1e-ex23bR | gcctttctattggcagctgtt | | ctgatcaaggctgccaggag |
| BPAG1e-ex7/8F | tctgtttcccttatctatatttggc | BPAG1e-ex23cF | gaaaatgcattgccagtgtg | FERMT1 | cattactgatatccctaaacttgc |
| BPAG1e-ex7/8R | caacttctcagcaataaaagaatctg | BPAG1e-ex23cR | catggcccttgtgattctg | | ttggatttgtgtctttttgc |
| BPAG1e-ex9F | cgatatcaaaaggtatgtgtgcc | BPAG1e-ex23dF | gaagaagccatgcaagaagc | FERMT2 | gaggggcctcttatcactcc |
| BPAG1e-ex9R | tcctgcaagttagccaaaatc | BPAG1e-ex23dR | tgtgctgcgtagctgatctt | | tctgcttggtttggtgacag |
| BPAG1e-ex10F | ggggtacacataggaccttgg | BPAG1e-ex23eF | gcactgaaaattcaggcaga | ITGA3 | ctggtgctgtgtacctgtgc |
| BPAG1e-ex10R | gccaataaaatacaaagttctgctc | BPAG1e-ex23eR | ttgctgcatttgttcacgat | | gctggtcttctgaccctgac |
| BPAG1e-ex11/12F | aaagtgctgaaggcatggag | BPAG1e-ex23fF | cagcagaggagtttcggaag | ITGB1 | taagatcaggggagccacag |
| BPAG1e-ex11/12R | ccttcaaccttcagagggc | BPAG1e-ex23fR | ctggcactccttcactgtca | | tcagaattggatttggctca |
| BPAG1e-ex13F | tgcagtgtgttttcagaggg | BPAG1e-ex23gF | aaagagcacagccaaagactg | | |
| BPAG1e-ex13R | caaactctacgacacatatgaaaagg | BPAG1e-ex23gR | caatgagccaatcacattcaa | | |
| BPAG1e-ex14F | ttggcatataccacacgacc | BPAG1e-ex24aF | ctgaaccctccttggtttga | | |
| BPAG1e-ex14R | cctaagattgagattcccattg | BPAG1e-ex24aR | attgaggtggctttcgtcag | | |
| BPAG1e-ex15F | tgcttgactagctgtgtccc | BPAG1e-ex24bF | aggcagttggtggaagctaa | | |
| BPAG1e-ex15R | tgtgcaaaactgaacattaagaag | BPAG1e-ex24bR | tgctggatggctcatgtaaa | | |
| BPAG1e-ex16F | cattggttcttaatgtgaaatgtg | BPAG1e-ex24cF | tgtgagaggcattcgtgttc | | |
| BPAG1e-ex16R | gcccagccaataatgtatttc | BPAG1e-ex24cR | actaaccggctcagcaaaga | | |
| BPAG1e-ex17F | tcagtgttccaaggtgaagaac | BPAG1e-ex24dF | caggaaggcctcatcacact | | |
| BPAG1e-ex17R | gagatcacagagacacaaatgg | BPAG1e-ex24dR | gttttgtggcaatgaggaca | | |
| BPAG1e-ex18F | gcaaattttattaggctgaacg | BPAG1e-ex24eF | aaatgggaatccgatgtttg | | |
| BPAG1e-ex18R | tcctccaagtaagaaagaaatatgg | BPAG1e-ex24eR | tttgaagggcattaaatcttatgTaa | | |
| BPAG1e-ex19F | catcttccaccctcctgatg | | | | |

Total RNA was isolated from keratinocytes using RNAeasy® FFPE kit (QIAGEN, Hilden, Germany), transcribed into cDNA (Fermentas, St Leon-Rot, Germany), and subjected to quantitative real time PCR (qPCR) using iQTM SYBR® Green Supermix, Biorad CFX96 and BioRad CFX Manager Software (version 1.5). Primers are listed in STable II.

**STable II. Antibodies used in this study**

| Primary antibodies/Antigen | Clone/Cat. no. | Company | Application and dilution |
|---|---|---|---|
| BPAG1 | 279 | Cosmo BIO | IF 1:50 |
| Phallidin, TRITC-conjugated | FAK100 | Millipore (Darmstadt, Germany) | IF 1:1000 |
| Collagen XVII | NC16A, polyclonal rabbit | Schäcke et.al. (S1) | IF 1:1000<br>IB 1:1000 |
| Collagen XVII | NC16A-3 | Abcam (Cambridge, UK) | IF 1:1000 |
| Collagen XVII | Endo 2 | Franzike et al. (S2) | IB 1:1000 |
| Plectin | 31 | BD Biosciences | IF 1:250 |
| GAPDH | 6C5 | Merck (Darmstadt, Germany) | IB 1:2000 |
| Integrin α6 | GOH3 | Progen (Heidelberg, Germany) | IF 1:50 |
| Integrin β4 | 3E1 | Millipore (Darmstadt, Germany) | IF 1:100 |
| Integrin α3 | P1B5 | Chemicon | IF 1:100 |
| Integrin β1 | 4B7R | Abcam (Cambridge, UK) | IF 1:50 |
| Keratin 5/6 | D5/16 B4 | Dako (Hamburg, Germany) | IB, IF 1.500 |
| Keratin 5 | Polyclonal, ab53121 | Abcam (Cambridge, UK) | IF 1:1000 |
| Keratin 14 | LL002 | Abcam (Cambridge, UK) | IB 1:500<br>IF 1:100 |
| Keratin 15 | EPR1614Y | Abcam (Cambridge, UK) | IB 1:400<br>IF 1:100 |
| Fibronectin | Polyclonal | Abcam (Cambridge, UK) | IB 1:2000<br>IF 1:1000 |
| Laminin | Polyclonal | Abcam (Cambridge, UK) | IF 1:1000 |
| **Secondary antibodies** | | **Company** | **Dilution** |
| Alexa Fluor® 488 goat anti-mouse IgG | | Invitrogen, Darmstadt, Germany | 1:1000 |
| Alexa Fluor® 488 goat anti-rabbit IgG | | Invitrogen, Darmstadt, Germany | 1:1000 |
| Alexa Fluor® 594 goat anti-rabbit IgG | | Invitrogen, Darmstadt, Germany | 1:1000 |
| Alexa Fluor® 568 goat anti-mouse IgG | | Invitrogen, Darmstadt, Germany | 1:1000 |
| Horseradish peroxidase labeled goat anti-mouse | | Merck, Darmstadt, Germany | 1: 5000 |
| Horseradish peroxidase labeled goat anti-rabbit | | KPL, Gaitherburg, MA, USA | 1:10000 |

IB: immunoblotting; IF: immunofluorescence staining; Cat. no.: catalogue number.

SUPPLEMENTARY REFERENCES

S1. Schäcke H, Schumann H, Hammami-Hauasli N, Raghunath M, Bruckner-Tuderman L. Two forms of collagen XVII in keratinocytes. A full-length transmembrane protein and a soluble ectodomain. J Biol Chem 1998; 273: 25937–25943.

S2. Franzke C-W, Tasanen K, Schäcke H, Zhou Z, Tryggvason K, Mauch C, et al. Transmembrane collagen XVII, an epithelial adhesion protein, is shed from the cell surface by ADAMs. EMBO J 2002; 21: 5026–5035.