

## **EB-NET publication list**

### **Injury-Driven Stiffening of the Dermis Expedites Skin Carcinoma Progression**

Cancer Res. 2016 Feb 15;76(4):940-51. doi: 10.1158/0008-5472.CAN-15-1348.

Mittapalli VR, Madl J, Löffek S, Kiritsi D, Kern JS, Römer W, Nyström A, Bruckner-Tuderman L.

### **Losartan ameliorates dystrophic epidermolysis bullosa and uncovers new disease mechanisms**

EMBO Mol Med. 2015 Sep;7(9):1211-28. doi: 10.15252/emmm.201505061.

Nyström A, Thriene K, Mittapalli V, Kern JS, Kiritsi D, Dengel J, Bruckner-Tuderman L.

### **High Local Concentrations of Intradermal MSCs Restore Skin Integrity and Facilitate Wound Healing in Dystrophic Epidermolysis Bullosa.**

Mol Ther. 2015 Aug;23(8):1368-1379. doi: 10.1038/mt.2015.58.

Kühl T, Mezger M, Hausser I, Handgretinger R, Bruckner-Tuderman L, Nyström A.

### **Targeting epidermal lipids for treatment of Mendelian disorders of cornification**

Orphanet J Rare Dis. 2014 Mar 7;9:33

Kiritsi D, Schauer F, Wölflé U, Valari M, Bruckner-Tuderman L, Has C, Happé R.

### **Underrecognition of acral peeling skin syndrome: 59 new cases with 15 novel mutations.**

Br J Dermatol. 2014 Mar 13. doi: 10.1111/bjd.12964. [Epub ahead of print].

Szczecinska W, Nesteruk D, Wertheim-Tysarowska K, Greenblatt DT, Baty D, Browne F, Liu L, Ozoemena L, Terron-Kwiatkowski A, McGrath JA, Mellerio JE, Morton J, Woźniak K, Kowalewski C, Has C, Moss C.

### **RhoA activation by CNFy restores cell-cell adhesion in kindlin-2 deficient keratinocytes.**

J Pathol. 2014 Mar 10. doi: 10.1002/path.4350. [Epub ahead of print]

He Y, Sonnenwald T, Sprenger A, Hansen U, Dengel J, Bruckner-Tuderman L, Schmidt G, Has C.

### **Targeting epidermal lipids for treatment of Mendelian disorders of cornification.**

Orphanet J Rare Dis. 2014 Mar 7;9:33. doi: 10.1186/1750-1172-9-33.

Kiritsi D, Schauer F, Wölflé U, Valari M, Bruckner-Tuderman L, Has C, Happé R.

### **The missense mutation p.R1303Q in type XVII collagen underlies junctional epidermolysis bullosa resembling Kindler syndrome.**

J Invest Dermatol. 2014 Mar;134(3):845-9.

Has C, Kiritsi D, Mellerio JE, Franzke CW, Wedgeworth E, Tantcheva-Poor I, Kernland-Lang K, Itin P, Simpson MA, Dopping-Hepenstal PJ, Fujimoto W, McGrath JA, Bruckner-Tuderman L.

### **Novel and recurrent AAGAB mutations: clinical variability and molecular consequences.**

J Invest Dermatol. 2013 Oct;133(10):2483-6.

Kiritsi D, Chmel N, Arnold AW, Jakob T, Bruckner-Tuderman L, Has C.

### **Consistency of the proteome in primary human keratinocytes with respect to gender, age, and skin localization.**

Mol Cell Proteomics. 2013 Sep;12(9):2509-21

Sprenger A, Weber S, Zarai M, Engelke R, Nascimento JM, Gretzmeier C, Hilpert M, Boerries M, Has C, Busch H, Bruckner-Tuderman L, Dengel J.

### **Collagen VII plays a dual role in wound healing.**

J Clin Invest 123, 3489-3509, 2013 August 1

Nyström A, Velati D, Mittapalli VR, Fritsch A, Kern JS, Bruckner-Tuderman L.

### **Phenotypic spectrum associated with alpha6beta4 integrin mutations:**

Br J Dermatol. 2013 Jul;169(1):115-24.

Schumann H, Kiritsi D, Pigors M, Hausser I, Kohlhase J, Peters J, Ott H, Hyla-Klekot L, Gacka E, Sieron AL, Valari M, Leena Bruckner-Tuderman L, Has C.

**Keratins control intercellular adhesion involving PKC- $\alpha$ -mediated desmoplakin phosphorylation**

J Cell Biol 201, 681-92, 2013

Kröger C, Loschke F, Schwarz N, Windoffer R, Leube RE, Magin TM

**Rat model for DDEB: glycine substitution reduces C7 stability and shows gene-dosage effect**

PloS One 8, e64243, 2013

Nyström A, Buttgereit J, Bader M, Shmidt T, Özcelik C, Hausser I, Bruckner-Tuderman L, Kern JS.

**Global remodeling of cellular microenvironment due to loss of collagen VII**

Mol Syst Biol, Online Publication 16.04.2013, doi: 10.1038/msb.2013.17

Küttner V, Mack C, Rigbolt KTG, Kern JS, Schilling O, Busch H, Bruckner-Tuderman L, Dengjel J.

**Molecular heterogeneity of epidermolysis bullosa simplex: contribution of EXPH5 mutations.**

J Invest Dermatol. 2014 Mar;134(3):842-5.

Pigors M, Schwieger-Briel A, Leppert J, Kiritsi D, Kohlhase J, Bruckner-Tuderman L, Has C.

**Passive transfer of collagen XVII-specific antibodies induces sustained blistering disease in adult mice.**

Orphanet J Rare Dis. 2013 Jan 29;8:17.

Chiriac MT, Licarete E, Sas AG, Rados AM, Lupan I, Chiriac AM, Speth H, Pop-Vancia V, Domsa I, Sesarman A, Popescu O, Sitaru C.

**Keratins mediate localization of hemidesmosomes and represses cell motility**

J Invest Dermatol 133, 181-90, 2013

Seltmann K, Roth W, Kröger C, Loschke F, Magin TM.

**Epidermolysis bullosa simplex Ogna revisited**

J Invest Dermatol 133, 270-3, 2013 (\*equal contribution)

Kiritsi D\*, Pigors M\*, Tantcheva-Poor I, Wessel C, Arin MJ, Kohlhase J, Bruckner-Tuderman L, Has C.

**Novel and recurrent AAGAB mutations in patients with punctate palmoplantar keratoderma.**

J Invest Dermatol, 2013; 133: 2483-2486.

Kiritsi D, Chmel N, Arnold AW, Jakob T, Bruckner-Tuderman L, Has C.

**Molecular fingerprinting of the podocyte reveals novel gene and protein regulatory networks.**

Kidney Int 83 (6), 1052-1064, 2013.

Boerries M, Grahammer F, Eiselein S, Buck M, Meyer C, Goedel M, Bechtel W, Zschiedrich S, Pfeifer D, Laloe D, Arrondel C, Goncalves S, Krüger M, Harvey SJ, Busch H, Dengjel J, Huber TB.

**Involvement of mitochondrial dynamics in the segregation of mitochondrial matrix proteins during stationary phase mitophagy.**

INat Commun 4, 2789, 2013.

Abeliovich H, Zarei M, Rigbolt KTG, Youle RJ, Dengjel J.

**Keratins significantly contribute to cell stiffness and impact on invasive behavior.**

Proc Natl Acad Sc 110, 18507-18512, 2013.

Seltmann K, Fritsch A, Käs J, Magin TM.

**The ubiquitin-proteasome system regulates desmoplakin transport to desmosomes in an actin-dependent manner**

Exp Dermatol 21, 791-3, 2012.

Löffek S, Bruckner-Tuderman L, Magin TM.

**A novel keratin 5 mutation causes severe EBS accompanied by keratoderma and squamous cell carcinoma**

Br J Dermatol 167, 929-36, 2012

Schumann H, Roth W, Has C, Volz A, Erfurt-Berge C, Magin TM, Bruckner-Tuderman L.

**TGM5 mutations impact epidermal differentiation in acral peeling skin syndrome**

J Invest Dermatol 132, 2422-2429, 2012

Pigors M, Kiritsi D, Schwieger-Briel A, Suarez J, Faletra F, Aho H, Makela L, Kern JS, Bruckner-Tuderman L, Has C.

**Type 1 segmental Galli-Galli disease resulting from a previously unreported keratin 5 mutation**

J Invest Dermatol 132, 2100-3, 2012 (\*equal contribution)

Arnold AW\*, Kiritsi D\*, Happle R, Kohlhase J, Hausser I, Bruckner-Tuderman L, Has C, Itin PH.

**Epidermal ADAM17 maintains the skin barrier by regulating EGFR ligand-dependent terminal keratinocyte differentiation**

J Exp Med 209, 1105-19, 2012

Franzke CW, Cobzaru C, Triantafyllopoulou A, Loeffek S, Horiuchi K, Threadgill DW, Kurz T, Bruckner-Tuderman L, Blobel CP.

**The epidermal basement membrane is a composite of separate laminin- and collagen IV-containing networks connected by aggregated perlecan, but not by nidogens**

J Biol Chem 287, 18700-9, 2012

Behrens D, Villone D, Koch M, Brunner G, Sorokin L, Robenek H, Bruckner-Tuderman L, Bruckner P, Hansen U.

**Revertant mosaicism in a human skin fragility disorder results from slippedmispairing and mitotic recombination**

J Clin Invest 122, 1742-1746, 2012

Kiritsi D, He Y, Pasmoij AMG, Onder M, Happle R, Jonkman M, Bruckner-Tuderman L, Has C.

**Integrin α3 mutations with kidney, lung and skin disease**

New Engl J Med 366, 1508-1514, 2012 (\*, §, equal contribution)

Has C\*, Spartà G\*, Kiritsi D\*, Weibel L, Moeller A, Vega-Warner V, Waters A, He Y, Anikster Y, Esser P, Straub BK, Hausser I, Bockenhauer D, Dekel B, Hildebrandt F, Bruckner-Tuderman L§, Laube GF§

**Partial loss of epithelial phenotype in kindlin-1-deficient keratinocytes**

Am J Pathol. 180, 1581-92, 2012

Qu H, Wen T, Pesch M, Aumailley M.

**Kindler Syndrome: Extension of mutational spectrum and natural history**

Hum Mutat 32, 1204-12, 2011

Has C, Castiglia D, del Rio M, Garcia Diez M, Piccinni E, Kiritsi D, Kohlhase J, Itin P, Martin L, Fischer J, Zambruno G, Bruckner-Tuderman L.

**Efficiency of translation termination in humans is highly dependent upon nucleotides in the neighbourhood of a (premature) termination codon**

J Med Genet 48, 640-4, 2011

Pacho F, Zambruno G, Calabresi V, Kiritsi D, Schneider H.

**Dynamic interactions of epidermal collagen XVII with the extracellular matrix: laminin 332 as a major binding partner**

Am J Pathol 79, 829-37, 2011

Nishie W, Kiritsi D, Nyström A, Hofmann SC, Bruckner-Tuderman L.

**Laminin-332 coordinates mechanotransduction and growth cone bifurcation in sensory neurons**

Nat Neurosci 14, 993-1000, 2011

Chiang LY, Poole K, Oliveira BE, Duarte N, Sierra YA, Bruckner-Tuderman L, Koch M, Hu J, Lewin GR.

**Molecular mechanisms of phenotypic variability in junctional epidermolysis bullosa**

J Med Genet 48, 450-7, 2011

Kiritsi D, Kern JS, Schumann H, Kohlhase J, Has C, Bruckner-Tuderman L.

**Lack of Plakoglobin Leads to Lethal Congenital Epidermolysis Bullosa**

Hum Mol Genet 20, 1811-9, 2011.

Pigors M, Kiritsi D, Krümpelmann S, Wagner N, He Y, Podda M, Kohlhase J, Hausser I, Bruckner-Tuderman L, Has C.

**Induction of phenotype modifying cytokines by FERMT1 mutations**

Hum Mutat 32, 397-406, 2011

Heinemann A, He Y, Zimina E, Boerries M, Busch H, Chmel N, Kurz T, Bruckner-Tuderman L, Has C.

**Kindlin-1 and -2 have overlapping functions in epithelial cells implications for phenotype modification**

Am J Pathol 178, 975-82, 2011

He Y, Esser P, Heinemann A, Bruckner-Tuderman L, Has C.

**A secreted variant of cartilage oligomeric matrix protein (COMP) carrying a chondrodysplasia-causing mutation (p.H587R) disrupts collagen fibrillogenesis**

Arthritis Rheum 63,159-167, 2011

Hansen U, Platz N, Becker A, Bruckner P, Paulsson M, Zaucke F.