

## **NIRK publication list**

### **Two novel mutations in the LOR gene in three families with loricrin keratoderma**

Br J Dermatol. 2014 Sep 18

Hotz A, Bourrat E, Hausser I, Haftek M, da Silva MV, Fischer J.

### **Palmoplantar keratoderma (PPK): acquired and genetic causes of a not so rare disease**

J Dtsch Dermatol Ges. 2014 Sep;12(9):781-8

Schiller S, Seebode C, Hennies HC, Giehl K, Emmert S.

### **A mouse organotypic tissue culture model for autosomal recessive congenital ichthyosis**

Br J Dermatol. 2014 Jul 31. doi: 10.1111/bjd.13308. [Epub ahead of print]

Rosenberger S, Dick A, Latzko S, Hausser I, Stark HJ, Rauh M, Schneider H, Krieg P

### **Loss of desmoglein 1 associated with palmoplantar keratoderma, dermatitis and multiple allergies**

Br J Dermatol. 2014 Jul 12. doi: 10.1111/bjd.13247. [Epub ahead of print]

Has C, Jakob T, He Y, Kiritsi D, Hausser I, Bruckner-Tuderman L.

### **Penetration of normal, damaged and diseased skin—an in vitro study on dendritic core-multishell nanotransporters**

J Control Release. 2014 Jul 10;185:45-50

Alnasif N, Zoschke C, Fleige E, Brodwolf R, Boreham A, Rühl E, Eckl KM, Merk HF, Hennies HC, Alexiev U, Haag R, Küchler S, Schäfer-Korting M.

### **White Sponge Nevus – A Rare Autosomal Dominant Keratinopathy**

Klin Padiatr. 2014 May 8. [Epub ahead of print] No abstract available

Benoit S, Schlipf N, Hausser I, Fischer J, Hamm H.

### **Increased cutaneous absorption reflects impaired barrier function of reconstructed skin models mimicking keratinisation disorders**

Exp Dermatol. 2014 Apr;23(4):286-8

Eckl KM, Weindl G, Ackermann K, Küchler S, Casper R, Radowski MR, Haag R, Hennies HC, Schäfer-Korting M.

### **Nonsyndromic types of ichthyoses – an update.**

J Dtsch Dermatol Ges. 2014 Feb;12(2):109-21. Traupe H, Fischer J, Oji V.

### **Annular atrophic plaques on the face in a father and a son: Christianson's disease, a real entity?**

Acta Derm Venereol. 2014 Jan;94(1):100-1

Peitsch WK, Orouji A, Starink TM, Hausser I, Figl R, Goerd S.

### **Oral liarozole in the treatment of patients with moderate/severe lamellar ichthyosis: results of a randomized, double-blind, multinational, placebo-controlled phase II/III trial**

Br J Dermatol. 2014 Jan;170(1):173-81

Vahlquist A, Blockhuys S, Steijlen P, van Rossem K, Didona B, Blanco D, Traupe H.

### **Topical Enzyme-Replacement Therapy Restores Transglutaminase 1 Activity and Corrects Architecture of Transglutaminase-1-Deficient Skin Grafts**

Am J Hum Genet. 2013 Sep 17

Aufenvenne K, Larcher F, Hausser I, Duarte B, Oji V, Nikolenko H, Del Rio M, Dathe M, Traupe H.

### **Functional and molecular genetic analyses of nine newly identified XPD-deficient patients reveal a novel mutation resulting in TTD as well as in XP/CS complex phenotypes.**

Exp Dermatol. 2013 Jul;22(7):486-9

Schäfer A, Gratchev A, Seebode C, Hofmann L, Schubert S, Laspe P, Apel A, Ohlenbusch A, Tzvetkov M, Weishaupt C, Oji V, Schön MP, Emmert S.

**Haplotype analysis in western European patients with mal de Meleda: founder effect for the W15R mutation in the SLURP1 gene.**

Br J Dermatol. 2013 Jun;168(6):1372-4.

Nellen RG, Steijlen PM, Hennies HC, Fischer J, Munro CS, Jonkman MF, van Steensel MA, van Geel M.

**Impaired Epidermal Ceramide Synthesis Causes Autosomal Recessive Congenital Ichthyosis and Reveals the Importance of Ceramide Acyl Chain Length**

J Invest Dermatol. 2013 Apr 2. doi: 10.1038/jid.2013.153. [Epub ahead of print] Eckl KM, Tidhar R, Thiele H, Oji V, Hausser I, Brodesser S, Preil ML, Onal-Akan A, Stock F, Müller D, Becker K, Casper R, Nürnberg G, Altmüller J, Nürnberg P, Traupe H, Futerman AH, Hennies HC

**Genotype-phenotype correlations emerging from the identification of missense mutations in MBTPS2**

Hum Mutat. 2013 Apr;34(4):587-94. doi: 10.1002/humu.22275. Epub 2013 Mar 8

Bornholdt D, Atkinson TP, Bouadjar B, Catteau B, Cox H, De Silva D, Fischer J, Gunasekera CN, Hadj-Rabia S, Happle R, Holder-Espinasse M, Kaminski E, König A, Mégarbané A, Mégarbané H, Neidel U, Oeffner F, Oji V, Theos A, Traupe H, Vahlquist A, van Bon BW, Virtanen M, Grzeschik KH.

**A novel X-chromosomal microdeletion encompassing congenital hemidysplasia with ichthyosiform erythroderma and limb defects.**

Pediatr Dermatol. 2013 Mar-Apr;30(2):250-2.

Raychaudhury T, George R, Mandal K, Srivastava VM, Thomas M, Bornholdt D, Grzeschik KH, Koehler A.

**First Symposium of Ichthyosis Experts**

Actas Dermosifiliogr. 2013 Jan 30. doi:pii: S0001-7310(12)00577-7. 10.1016/j.ad.2012.11.018. [Epub ahead of print]

Hernández-Martín A, Torrelo-Fernández A, de Lucas-Laguna R, Casco F, González-Sarmiento R, Vega A, Pedreira-Massa JL, de Unamuno-Pérez P, Larcher F, Arroyo I, Traupe H.

**Complete filaggrin deficiency in ichthyosis vulgaris is associated with only moderate changes in epidermal permeability barrier function profile**

J Eur Acad Dermatol Venereol. 2013 Jan 7. doi: 10.1111/jdv.12079. [Epub ahead of print]

Perusquía-Ortiz AM, Oji V, Sauerland MC, Tarinski T, Zaraeva I, Seller N, Metze D, Aufenvenne K, Hausser I, Traupe H.

**Ichthyosis keeps surprising us**

Actas Dermosifiliogr. 2013 May;104(4):267-9. doi: 10.1016/j.ad.2012.10.016. Epub 2013 Jan 3.

English, Spanish. No abstract available.

Traupe H.

**Induced pluripotent mesenchymal stromal cell clones retain donor-derived differences in DNA methylation profiles.**

Mol Ther. 2013 Jan;21(1):240-50

Shao K, Koch C, Gupta MK, Lin Q, Lenz M, Laufs S, Denecke B, Schmidt M, Linke M, Hennies HC, Hescheler J, Zenke M, Zechner U, Šarić T, Wagner W.

**Aloxe3 knockout mice reveal a function of epidermal lipoxygenase-3 as hepoxilin synthase and its pivotal role in barrier formation.**

J Invest Dermatol. 2013 Jan;133(1):172-80

Krieg P, Rosenberger S, de Juanes S, Latzko S, Hou J, Dick A, Kloz U, van der Hoeven F, Hausser I, Esposito I, Rauh M, Schneider H.

**The substrate degradome of meprin metalloproteases reveals an unexpected proteolytic link between meprin  $\beta$  and ADAM10.**

Cell Mol Life Sci. 2013 Jan;70(2):309-33.

Jefferson T, Auf dem Keller U, Bellac C, Metz VV, Broder C, Hedrich J, Ohler A, Maier W, Magdolen V, Sterchi E, Bond JS, Jayakumar A, Traupe H, Chalaris A, Rose-John S, Pietrzik CU, Postina R, Overall CM, Becker-Pauly C.

**Cystatin M/E knockdown by lentiviral delivery of shRNA impairs epidermal morphogenesis of human skin equivalents**

Exp Dermatol. 2012 Nov

Jansen PA, van den Bogaard EH, Kersten FF, Oostendorp C, van Vlijmen-Willems IM, Oji V, Traupe H, Hennies HC, Schalkwijk J, Zeeuwen PL

**The substrate degradome of meprin metalloproteases reveals an unexpected proteolytic link between meprin  $\beta$  and ADAM10**

Cell Mol Life Sci. 2013 Jan;70(2):309-33. doi: 10.1007/s00018-012-1106-2. Epub 2012 Sep 1.

Jefferson T, Auf dem Keller U, Bellac C, Metz VV, Broder C, Hedrich J, Ohler A, Maier W, Magdolen V, Sterchi E, Bond JS, Jayakumar A, Traupe H, Chalaris A, Rose-John S, Pietrzik CU, Postina R, Overall CM, Becker-Pauly C.

**Long-term faithful recapitulation of transglutaminase 1-deficient lamellar ichthyosis in a skin-humanized mouse model and insights from proteomic studies**

Aufvenne K, Rice RH, Hausser I, Oji V, Hennies HC, Del Rio M, Traupe H, Larcher F, J Invest Dermatol. 2012 Jul;132(7):1918-21.

**Beta-Actin is a Target for Transglutaminase Activity at Synaptic Endings in Chicken Telencephalic Cell Cultures**

Dolge L, Aufvenne K, Traupe H, Baumgartner W. J Mol Neurosci. 2012 Feb;46(2):410-9.

**PNPLA1 mutations cause autosomal recessive congenital ichthyosis in golden retriever dogs and humans.**

Grall A, Guaguère E, Planchais S, Grond S, Bourrat E, Hausser I, Hitte C, Le Gallo M, Derbois C, Kim GJ, Lagoutte L, Degorce-Rubiales F, Radner FP, Thomas A, Küry S, Bensignor E, Fontaine J, Pin D, Zimmermann R, Zechner R, Lathrop M, Galibert F, André C, Fischer J. Nat Genet. 2012 Jan 15;44(2):140-7

**Intronic mutations affecting splicing of MBTPS2 cause ichthyosis follicularis, alopecia and photophobia (IFAP) syndrome.**

Oeffner F, Martinez F, Schaffer J, Salhi A, Monfort S, Oltra S, Neidel U, Bornholdt D, van Bon B, König A, Happle R, Grzeschik KH. Exp Dermatol. 2011 May;20(5):447-9.

**rAAV2-mediated restoration of LEKTI in LEKTI-deficient cells from Netherton patients.**

Roedl D, Oji V, Buters JT, Behrendt H, Braun-Falco M. J Dermatol Sci. 2011 Mar;61(3):194-8.

**Genodermatoses.**

Happle R, Traupe H: Hautarzt. 2011 Feb;62(2):80-1.

**NSDHL-Related Disorders.**

du Souich C, Raymond FL, Grzeschik KH, König A, Boerkoel CF.

In: Pagon RA, Bird TD, Dolan CR, Stephens K, editors. GeneReviews [Internet]. Seattle (WA): University of Washington, Seattle; 2011 Feb 01

**Hereditäre Ichthyosen.**

Oji V. In: Braun-Falco's Dermatologie, Venerologie und Allergologie, 6. Aufl., Plewig, Wolff, Burgdorf, Landthaler, Springer Heidelberg (2011, imDruck)

**A family with dystrophic alopecia and keratosis pilaris.**

Scholz IM, Hausser I, Behnecke A, Helmbold P, J Dtsch Dermatol Ges 2011; 9:1055-6

**Palmoplantar hyperkeratoses and hypopigmentation. Cole disease.**

Schmieder A, Hausser I, Schneider SW, Goerdts S, Peitsch WK. Acta Derm Venereol. 2011; 91:737-8

**IGFBP7 as a novel therapeutic target in psoriasis.**

Nousbeck J, Ishida-Yamamoto A, Bidder M, Fuchs D, Eckl K, Hennies HC, Sagiv N, Gat A, Gini M, Filip I, Matz H, Goldberg I, Enk CD, Sarig O, Meilik B, Aberdam D, Gilhar A, Sprecher E J. Invest Dermatol 131:1767-1770 (2011)

**Hallmarks of atopic skin mimicked in vitro by means of a skin disease model based on FLG knock-down.**

Küchler S, Henkes D, Eckl KM, Ackermann K, Plendl J, Korting HC, Hennies HC, Schäfer-Korting M, Altern. Lab Anim 39:471-480 (2011)

**Ichthyosis prematurity syndrome caused by a novel fatty acid transport protein 4 gene mutation in a German patient.**

Inhoff O, Hausser I, Schneider SW, Khnykin D, Jahnsen F, Sartoris J, Goerdts S, Peitsch WK. Arch Dermatol 2011; 147:750-2

**Congenital ichthyosis in severe type II Gaucher disease with a homozygous null mutation.**

Haverkaemper S, Marquardt T, Hausser I, Timme K, Kuehn T, Hertzberg C, Rossi R. Neonatology 2011; 100:194-7

**A novel homozygous missense mutation in ARS (component B) encoding SLURP-1 causes Mal de Meleda with exceptional phenotype.**

Gruber R, Hennies HC, Romani N, Schmuth M, Arch Dermatol 147:748-750 (2011)

**CEDNIK syndrome results from loss-of-function mutations in SNAP29.**

Fuchs-Telem D, Stewart H, Rapaport D, Nousbeck J, Gat A, Gini M, Lugassy Y, Emmert S, Eckl KM, Hennies HC, Sarig O, Goldsher D, Meilik B, Horowitz M, Sprecher E Br J Dermatol 164:610-616 (2011)

**Full-thickness human skin models for congenital ichthyosis and related keratinisation disorders**

Eckl KM, Alef T, Torres S, Hennies HC, J Invest Dermatol 131:1938-1342 (2011)

**Mutations in CSTA, encoding Cystatin A, underlie exfoliative ichthyosis and reveal a role for this protease inhibitor in cell-cell adhesion.**

Blaydon DC, Nitoiu D, Eckl KM, Cabral RM, Bland P, Hausser I, van Heel DA, Rajpopat S, Fischer J, Oji V, Zvulunov, Traupe H, Hennies HC, Kelsell DP. Am J Hum Genet 2011; 89:564-71

**Expanding the keratin mutation database: novel and recurrent mutations and genotype-phenotype correlations in 28 patients with epidermolytic ichthyosis**

Arin MJ, Oji V, Emmert S, Hausser I, Traupe H, Krieg T, Grimberg G. Br J Dermatol 2011; 164:442-7