

Wichtige Publikationen nach 2013

Zimmer AD, Kim GJ, Hotz A, Bourrat E, Hausser I, Has C, Oji V, Stieler K, Vahlquist A, Kunde V, Weber B, Radner FP, Leclerc-Mercier S, Schlipf N, Demmer P, Küsel J, Fischer J. 16 novel mutations in PNPLA1 in patients with autosomal recessive congenital ichthyosis reveal the importance of an extended patatin domain in PNPLA1 that is essential for proper human skin barrier function. *Br J Dermatol.* 2017 Jan 17. doi: 10.1111/bjd.15308.

Kirchmeier P, Zimmer A, Bouadjar B, Rösler B, Fischer J. Whole-Exome-Sequencing Reveals Small Deletions in CASP14 in Patients with Autosomal Recessive Inherited Ichthyosis. *Acta Derm Venereol.* 2017 Jan 4;97(1):102-104.

Sánchez-Guijo A, Neunzig J, Gerber A, Oji V, Hartmann MF, Schuppe HC, Traupe H, Bernhardt R, Wudy SA. Role of steroid sulfatase in steroid homeostasis and characterization of the sulfated steroid pathway: Evidence from steroid sulfatase deficiency. *Mol Cell Endocrinol.* 2016 Dec 5;437:142-153.

Hotz A, Oji V, Bourrat E, Jonca N, Mazereeuw-Hautier J, Betz RC, Blume-Peytavi U, Stieler K, Morice-Picard F, Schönbuchner I, Markus S, Schlipf N, Fischer J. Expanding the Clinical and Genetic Spectrum of KRT1, KRT2 and KRT10 Mutations in Keratinopathic Ichthyosis. *Acta Derm Venereol.* 2016 May;96(4):473-8.

Pigors M, Sarig O, Heinz L, Plagnol V, Fischer J, Mohamad J, Malchin N, Rajpopat S, Kharfi M, Lestringant GG, Sprecher E, Kelsell DP, Blaydon DC. Loss-of-Function Mutations in SERPINB8 Linked to Exfoliative Ichthyosis with Impaired Mechanical Stability of Intercellular Adhesions. *Am J Hum Genet.* 2016 Aug 4;99(2):430-6.

Pigg MH, Bygum A, Gånemo A, Virtanen M, Brandrup F, Zimmer AD, Hotz A, Vahlquist A, Fischer J. Spectrum of Autosomal Recessive Congenital Ichthyosis in Scandinavia: Clinical Characteristics and Novel and Recurrent Mutations in 132 Patients. *Acta Derm Venereol.* 2016 Nov 2;96(7):932-937.

Gruber R, Rainer G, Weiss A, Udvardi A, Thiele H, Eckl KM, Schupart R, Nürnberg P, Zschocke J, Schmuth M, Volc-Platzer B, Hennies HC. Morphological alterations in two siblings with autosomal recessive congenital ichthyosis associated with CYP4F22 mutations. *Br J Dermatol.* 2017 Apr;176(4):1068-1073.

Schlipf NA, Traupe H, Gilaberte Y, Peitsch WK, Hausser I, Oji V, Schmieder A, Schneider SW, Demmer P, Rösler B, Fischer J. Association of Cole disease with novel heterozygous mutations in the somatomedin-B domains of the ENPP1 gene: necessary, but not always sufficient. *Br J Dermatol.* 2016 May;174(5):1152-6.

Hotz A, Oji V, Bourrat E, Jonca N, Mazereeuw-Hautier J, Betz RC, Blume-Peytavi U, Stieler K, Morice-Picard F, Schönbuchner I, Markus S, Schlipf N, Fischer J. Expanding the Clinical and Genetic Spectrum of KRT1, KRT2 and KRT10 Mutations in Keratinopathic Ichthyosis. *Acta Derm Venereol.* 2016 May;96(4):473-8.

Yang Z, Hartmann B, Xu Z, Ma L, Happle R, Schlipf N, Zhang LX, Xu ZG, Wang ZY, Fischer J. Large deletions in the NSDHL gene in two patients with CHILD syndrome. *Acta Derm Venereol.* 2015 Nov;95(8):1007-8.

Leclerc-Mercier S, Lemoine R, Bigorgne AE, Sepulveda F, Leveau C, Fischer A, Mahlaoui N, Hadj-Rabia S, de Saint Basile G. Ichthyosis as the dermatological phenotype associated with TTC7A mutations. *Br J Dermatol.* 2016 Nov;175(5):1061-1064.

Grond S, Eichmann TO, Dubrac S, Kolb D, Schmuth M, Fischer J, Crumrine D, Elias PM, Haemmerle G, Zechner R, Lass A, Radner FP. PNPLA1 Deficiency in Mice and Humans Leads to a Defect in the Synthesis of Omega-O-Acylceramides. *J Invest Dermatol*. 2017 Feb;137(2):394-402.

Sánchez-Guijo A, Oji V, Hartmann MF, Traupe H, Wudy SA. Simultaneous quantification of cholesterol sulfate, androgen sulfates, and progestogen sulfates in human serum by LC-MS/MS. *J Lipid Res*. 2015 Sep;56(9):1843-51.

Sánchez-Guijo A, Oji V, Hartmann MF, Schuppe HC, Traupe H, Wudy SA. High levels of oxysterol sulfates in serum of patients with steroid sulfatase deficiency. *J Lipid Res*. 2015 Feb;56(2):403-12.

Kiritsi D, Valari M, Fortugno P, Hausser I, Lykopoulou L, Zambruno G, Fischer J, Bruckner-Tuderman L, Jakob T, Has C. Whole-exome sequencing in patients with ichthyosis reveals modifiers associated with increased IgE levels and allergic sensitizations. *J Allergy Clin Immunol*. 2015 Jan;135(1):280-3.

Spoerri I, Brena M, De Mesmaeker J, Schlipf N, Fischer J, Tadini G, Itin PH, Burger B. The phenotypic and genotypic spectra of ichthyosis with confetti plus novel genetic variation in the 3' end of KRT10: from disease to a syndrome. *JAMA Dermatol*. 2015 Jan;151(1):64-9.

Gruber R, Sugarman JL, Crumrine D, Hupe M, Mauro TM, Mauldin EA, Thyssen JP, Brandner JM, Hennies HC, Schmuth M, Elias PM. Sebaceous gland, hair shaft, and epidermal barrier abnormalities in keratosis pilaris with and without filaggrin deficiency. *Am J Pathol*. 2015 Apr;185(4):1012-21.

Moosbrugger-Martinz V, Jalili A, Schossig AS, Jahn-Bassler K, Zschocke J, Schmuth M, Stingl G, Eckl KM, Hennies HC, Gruber R. Epidermal barrier abnormalities in exfoliative ichthyosis with a novel homozygous loss-of-function mutation in CSTA. *Br J Dermatol*. 2015 Jun;172(6):1628-32.

Traupe H, Fischer J, Oji V. Nonsyndromic types of ichthyoses - an update. *J Dtsch Dermatol Ges*. 2014 Feb;12(2):109-21.

Radner FP, Fischer J. The important role of epidermal triacylglycerol metabolism for maintenance of the skin permeability barrier function. *Biochim Biophys Acta*. 2014 Mar;1841(3):409-15.

Eckl KM, Weindl G, Ackermann K, Küchler S, Casper R, Radowski MR, Haag R, Hennies HC, Schäfer-Korting M. Increased cutaneous absorption reflects impaired barrier function of reconstructed skin models mimicking keratinisation disorders. *Exp Dermatol*. 2014 Apr;23(4):286-8.