

## **Curriculum vitae**

### **Prof. Dr. med. Cristina Has**

Fachärztin für Haut- und Geschlechtskrankheiten  
Qualifikation fachgebundene genetische Beratung

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#### **Berufliche Erfahrung / Schwerpunkte der Forschung**

- **>20 Jahre Erfahrung in der Dermatologie**
- **>15 Jahre Erfahrung in Genetik und molekulargenetischer Diagnostik**
- **>15 Jahre Erfahrung mit Genodermatosen**
- **>20 Erfahrung in akademischer Lehre**
- **Schwerpunkte der klinischen Tätigkeit und Forschung:**  
      **Genodermatosen, pädiatrische Dermatologie**
- **150 Publikationen (11.11.2017)**

<https://www.ncbi.nlm.nih.gov/pubmed/?term=has+c>

#### **Universitätsstudium, Promotion und Habilitation**

1985–1991	Medizinstudium, Universität Iuliu Hatieganu Cluj-Napoca, Rumänien
2001	Promotion (M.D.), Dermatologie, Universität Cluj-Napoca
2007	Habilitation, Experimentelle Dermatologie, Albert-Ludwigs-Universität Freiburg im Breisgau
2012	Außerplanmäßige Professur, Albert-Ludwigs-Universität Freiburg im Breisgau

#### **Weiterbildungen und Qualifikationen**

1991–1992	Ärztin im Praktikum, Universitätsklinik Cluj-Napoca
1992–1995	Assistenzärztin im Fach Dermatologie, Universitäts-Hautklinik Cluj-Napoca
1995	Fachärztin für Haut- und Geschlechtskrankheiten, Rumänien
10.2000–12.2000	Pädiatrische Dermatologie, Prof. F. Cambazard, CHU Saint-Etienne, Frankreich
2003	Oberärztin für Haut- und Geschlechtskrankheiten, Rumänien
2007	Approbation als Ärztin in der BRD
2009	Fachärztin für Haut- und Geschlechtskrankheiten, BRD
2014	Qualifikation fachgebundene genetische Beratung, BRD

#### **Wissenschaftlicher Werdegang und berufliche Tätigkeit**

1995–1998	Universitätsassistentin Genetik, Universität Cluj-Napoca
1998–2003	Universitätsassistentin Dermatologie, Fach- und Oberärztin für Dermatologie, Universitäts-Hautklinik Cluj-Napoca

1999–2000	Forschung zum Thema „Genetik des Conradi-Hünnerman-Happle-Syndroms“, Prof. H. Traupe, Universitäts-Hautklinik WWU Münster
01.2001–12.2001	Wissenschaftliche Mitarbeiterin AG Prof. L. Bruckner-Tuderman, Universitäts-Hautklinik WWU Münster
01.2002–03.2003	Postdoc, Prof. J. Fischer, Centre National de Génotypage Evry, Frankreich
seit 03.2003	Laborleiterin, Funktionsoberärztin, Leitung einer AG „Genetik der Hautfragilität“ Klinik für Dermatologie und Venerologie, Albert-Ludwigs-Universität Freiburg im Breisgau
01.2010–02.2010	Visiting scientist, Prof. B. Hinz, Universität Toronto, Kanada
Seit 2013	PI im Interdisziplinären Else Kröner-Forschungskolleg (EKFK) der Medizinischen Fakultät Freiburg, „Nierenfunktionsstörungen als Komplikation von Systemerkrankungen“
2014	Tertio loco - Head of the research unit for genodermatoses, EB-House Salzburg Austria
Seit 2014	PI im SFB 1140 KIDGEM

### **Auszeichnungen und andere Aktivitäten**

(in der Reihenfolge der Aktualität)

#### *Preise und Stipendien:*

2014	Ulmer Dermatologie - Preis
2014	„Quo vadis“-Vortrag der Arbeitsgemeinschaft Dermatologische Forschung
2007	Oscar-Gans - Preis
2004	„Research on Skin Dryness“- Preis
2001–2003	Stipendium Genopole, Frankreich
2000	Stipendium „Le Pont Neuf“, Paris

#### *Mitgliedschaften und andere Aktivitäten:*

ESDR, EADV, ADF, AG Pädiatrische Dermatologie der DDG

seit 2017	Koordination der AG „Deep Genotyping“ des European Reference Network - Skin
seit 2016	Koordination des Projektes „Clinical Practice Guidelines EB Laboratory Diagnosis“ (Debra Austria)
seit 2014	Sprecherin der AG Dermatologische Genetik der „Arbeitsgemeinschaft Dermatologische Forschung“

#### *Ad hoc Reviewer:*

(Auswahl)

- Begutachtung Zeitschriften:  
Am J Hum Genet, Hum Mol Genet, J Invest Dermatol, PLoS ONE, Arch Derm Res, Exp Dermatol, Br J Dermatol, Clin Exp Dermatol, Acta Dermato Venerologica, Cancer Letters, Mol Biol Cell, JDDG, etc.
- Begutachtung Forschungsanträge und Abstracts:



Arbeitsgemeinschaft Dermatologische Forschung, European Society of Dermatologic Research, Deutsche Forschungsgemeinschaft, Telethon, Deutsche Forschungsgemeinschaft, DEBRA International, Health Research Board, Medical Research Council, Fondation Rene Touraine

### Publikationen

(Auswahl, in der Reihenfolge der Aktualität)

1. **Has C**, Schumann H, Leppert J, He Y, Hartmann B, Hausser I, Kohlhase J. Monoallelic large intragenic KRT5 deletions account for genetically unsolved cases of epidermolysis bullosa simplex. **J Invest Dermatol**. 2017 May 30. pii:S0022-202X(17)31562-2. doi: 10.1016/j.jid.2017.05.016. [Epub ahead of print] PubMed PMID: 28576738
2. Kroeger JK, Hofmann SC, Leppert J, **Has C**\*, Franzke CW#. Amino acid duplication in the coiled-coil structure of collagen XVII alters its maturation and trimerization causing mild junctional epidermolysis bullosa. **Hum Mol Genet** 1;26:479-488, 2017 \*corresponding author, # equal contribution
3. He Y, Leppert J, Steinke H, **Has C**. Homozygous Nonsense Mutation and Additional Deletion of an Amino Acid in BPAG1e Causing Mild Localized Epidermolysis Bullosa Simplex. **Acta Derm Venereol** 97:657-659 2017
4. Pazzagli C, He Y, Busch H, Esser P, Kiritsi D, Gache Y, Bruckner-Tuderman L, Boerries M, **Has C**. Absence of the integrin  $\alpha 3$  subunit induces an activated phenotype in human keratinocytes. **J Invest Dermatol** 137:1387-1391, 2017
5. Maier K, He Y, Wölfle U, Esser PR, Brummer T, Schempp C, Bruckner-Tuderman L, **Has C**. UV-B-induced cutaneous inflammation and prospects for antioxidant treatment in Kindler syndrome. **Hum Mol Genet** 25:5339-5352, 2016
6. He Y, Maier K, Leppert J, Hausser I, Schwieger-Briel A, Weibel L, Theiler M, Kiritsi D, Busch H, Boerries M, Hannula-Jouppi K, Heikkilä H, Tasanen K, Castiglia D, Zambruno G, **Has C**. Monoallelic Mutations in the Translation Initiation Codon of KLHL24 Cause Skin Fragility. **Am J Hum Genet** 99:1395-1404, 2016
7. **Has C**, Schwieger-Briel A, Schlipf N, Hausser I, Chmel N, Rösler B, Technau K, Jakob T, Zimmer A, Fischer J. Target-sequence Capture and High Throughput Sequencing Identify a De novo CARD14 Mutation in an Infant with Erythrodermic Pityriasis Rubra Pilaris. **Acta Derm Venereol** 96:989-990, 2016
8. He Y, Balasubramanian M, Humphreys N, Waruiru C, Brauner M, Kohlhase J, O'Reilly R, **Has C**. Intronic ITGA3 mutation impacts splicing regulation and causes interstitial lung disease, nephrotic syndrome and epidermolysis bullosa. **J Invest Dermatol** 136:1056-9, 2016
9. Maier K, He Y, Esser PR, Thriene K, Sarca D, Kohlhase J, Dengjel J, Martin L, **Has C**. Single Amino Acid Deletion in Kindlin-1 Results in Partial Protein Degradation Which Can Be Rescued by Chaperone Treatment. **J Invest Dermatol** 136:920-9, 2016
10. Yalcin EG, He Y, Orhan D, Pazzagli C, Emiralioglu N, **Has C**. Crucial role of posttranslational modifications of integrin  $\alpha 3$  in interstitial lung disease and nephrotic syndrome. **Hum Mol Genet** 24:3679-88, 2015
11. Chmel N, Danescu S, Gruler A, Kiritsi D, Bruckner-Tuderman L, Kreuter A, Kohlhase J, **Has C**. A Deep-Intronic FERMT1 Mutation Causes Kindler Syndrome: An Explanation for Genetically Unsolved Cases. **J Invest Dermatol**. 135:2876-9, 2015
12. Schwieger-Briel A, Weibel L, Chmel N, Leppert J, Kernland-Lang K, Grüniger G, **Has C**. A COL7A1 variant leading to in-frame skipping of exon 15 attenuates disease severity in recessive dystrophic epidermolysis bullosa. **Br J Dermatol** 173:1308-11, 2015

13. Kiritsi D, Lorente AI, Happle R, Bernabeu Wittel J, **Has C**. Blaschko line acne on pre-existent hypomelanosis reflecting a mosaic FGFR2 mutation. **Br J Dermatol** 172(4):1125-7, 2015
14. **Has C**, Chmel N, Levati L, Neri I, Sonnenwald T, Pigors M, Godbole K, Dudhbhate A, Bruckner-Tuderman L, Zambruno G, Castiglia D. FERMT1 promoter mutations in patients with Kindler syndrome. **Clin Genet** 88(3):248-54, 2015
15. 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** 135(1):280-3, 2015
16. Pigors M, Schwieger-Briel A, Cosgarea R, Diaconeasa A, Bruckner-Tuderman L, Fleck T, **Has C**. Desmoplakin mutations with palmoplantar keratoderma, woolly hair and cardiomyopathy. **Acta Derm Venereol** 95(3):337-40, 2015
17. He Y, Sonnenwald T, Sprenger A, Hansen U, Dengjel J, Bruckner-Tuderman L, Schmidt G, **Has C**. RhoA activation by CNFY restores cell-cell adhesion in kindlin-2 deficient keratinocytes. **J Pathol** 233:269-80, 2014
18. Pigors M, Schwieger-Briel A, Leppert J, Kiritsi D, Kohlhase J, Bruckner-Tuderman L, **Has C**. Molecular heterogeneity of epidermolysis bullosa simplex: contribution of EXPH5 mutations. **J Invest Dermatol** 134:842-5, 2014
19. **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. The Missense Mutation p.R1303Q in Type XVII Collagen Underlies Junctional Epidermolysis Bullosa Resembling Kindler Syndrome. **J Invest Dermatol** 134:845-9, 2014
20. Kiritsi D, Nanda A, Kohlhase J, Bernhard C, Bruckner-Tuderman L, Happle R, **Has C**. Extensive Postzygotic Mosaicism for a Novel Keratin 10 Mutation in Epidermolytic Ichthyosis. **Acta Derm Venereol** 94(3):346-8, 2014
21. Kiritsi D, Chmel N, Arnold AW, Jakob T, Bruckner-Tuderman L, **Has C**. Novel and Recurrent AAGAB Mutations: Clinical Variability and Molecular Consequences. **J Invest Dermatol** 133: 2483-6, 2013
22. Kiritsi D, Pigors M, Tantcheva-Poor I, Wessel C, Arin MJ, Kohlhase J, Bruckner-Tuderman L, **Has C**. Epidermolysis bullosa simplex Ogná revisited. **J Invest Dermatol** 133: 270-3, 2013
23. Cifuentes L, Kiritsi D, Chen W, Pennino J, Ring J, Weidinger S, **Has C**. A case of junctional epidermolysis bullosa with prurigo-like lesions and reduction of collagen XVII and filaggrin. **Br J Dermatol** 169:195-8, 2013
24. Schumann H, Kiritsi D, Pigors M, Hausser I, Kohlhase J, Peters J, Ott H, Hyla-Klekot L, Gacka E, Sieron AL, Valari M, Bruckner-Tuderman L, **Has C**. Phenotypic spectrum of epidermolysis bullosa associated with  $\alpha 6\beta 4$  integrin mutations. **Br J Dermatol** 169:115-24, 2013
25. **Has C**, Sparta 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. Integrin  $\alpha 3$  mutations with kidney, lung and skin disease. **New Engl J Med** 366, 1508-1514, 2012
26. Kiritsi D, He Y, Pasmooij AMG, Onder M, Happle R, Jonkman MF, Bruckner-Tuderman L, and **Has C**. Revertant mosaicism in a human skin fragility disorder results from slipped mispairing and mitotic recombination. **J Clin Invest** 122: 1742-1746, 2012
27. Pigors M, Kiritsi D, Cobzaru C, Schwieger-Briel A, Suárez J, Faletra F, Aho H, Mäkelä L, Kern JS, Bruckner-Tuderman L, **Has C**. TGM5 Mutations Impact Epidermal Differentiation in Acral Peeling Skin Syndrome. **J Invest Dermatol** 132:2422-9, 2012
28. Arnold AW, Kern JS, Itin PH, Pigors M, Happle R, **Has C**. Acromelanosis albo-

- punctata: a distinct inherited dermatosis with acral spotty dyspigmentation without systemic involvement. **Dermatology** 224:331-9, 2012
29. Heinemann A, He Y, Zimina E, Boerries M, Busch H, Chmel N, Kurz T, Bruckner-Tuderman L, and **Has C**. Induction of phenotype modifying cytokines by FERMT1 mutations. **Hum Mutat** 32: 397-406, 2011
  30. Has C, Castiglia D, del Rio M, Diez MG, Piccinni E, Kiritsi D, Kohlhase J, Itin P, Martin L, Fischer J, Zambruno G, Bruckner-Tuderman L. Kindler syndrome: extension of mutational spectrum and natural history. **Hum Mutat** 32:1204-1212, 2011
  31. He Y, Esser P, Schacht V, Bruckner-Tuderman L, **Has C**. Role of kindlin-2 in fibroblast functions: implications for wound healing. **J Invest Dermatol** 131:245-56, 2011
  32. Pigors M, Kiritsi D, Kruempelmann S, Wagner N, He Y, Podda M, Kohlhase J, Hausser I, Bruckner-Tuderman L, and **Has C**. Lack of plakoglobin leads to lethal congenital epidermolysis bullosa: a novel clinico-genetic entity. **Hum Mol Genet** 20:1811-1819, 2011
  33. He Y, Esser P, Heinemann A, 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
  34. Kiritsi D, Cosgarea I, Franzke CW, Schumann H, Oji V, Kohlhase J, Bruckner-Tuderman L, **Has C**. Acral peeling skin syndrome with TGM5 gene mutations may resemble epidermolysis bullosa simplex in young individuals. **J Invest Dermatol** 130:1741-6, 2010
  35. Arnold AW, Itin PH, Pigors M, Kohlhase J, Bruckner-Tuderman L, **Has C**. Poikiloderma with neutropenia: a novel C16orf57 mutation and clinical diagnostic criteria. **Br J Dermatol** 163:866-9, 2010
  36. Arin MJ, Grimberg G, Schumann H, De Almeida H Jr, Chang YR, Tadini G, Kohlhase J, Krieg T, Bruckner-Tuderman L, **Has C**. Identification of novel and known KRT5 and KRT14 mutations in 53 patients with epidermolysis bullosa simplex: correlation between genotype and phenotype. **Br J Dermatol** 162:1365-9, 2010
  37. **Has C**, Herz C, Zimina E, Qu HY, He Y, Zhang ZG, Wen TT, Gache Y, Aumailley M, Bruckner-Tuderman L. Kindlin-1 Is required for RhoGTPase-mediated lamellipodia formation in keratinocytes. **Am J Pathol** 175:1442-52, 2009
  38. Kern JS, Grüninger G, Imsak R, Müller ML, Schumann H, Kiritsi D, Emmert S, Borozdin W, Kohlhase J, Bruckner-Tuderman L, **Has C**. Forty-two novel COL7A1 mutations and the role of a frequent single nucleotide polymorphism in the MMP1 promoter in modulation of disease severity in a large European dystrophic epidermolysis bullosa cohort. **Br J Dermatol** 161:1089-97, 2009
  39. **Has C**, Yordanova I, Balabanova M, Kazandjieva J, Herz C, Kohlhase J, Bruckner-Tuderman L. A novel large FERMT1 (KIND1) gene deletion in Kindler syndrome. **J Dermatol Sci** 52:209-12, 2008
  40. **Has C**, Ludwig RJ, Herz C, Kern JS, Ussar S, Ochsendorf FR, Kaufmann R, Schumann H, Kohlhase J, Bruckner-Tuderman L. C-terminally truncated kindlin-1 leads to abnormal adhesion and migration of keratinocytes. **Br J Dermatol** 159:1192-6, 2008
  41. Kern JS, Herz C, Haan E, Moore D, Nottelmann S, von Lilien T, Greiner P, Schmitt-Graeff A, Opitz OG, Bruckner-Tuderman L, **Has C**. Chronic colitis due to an epithelial barrier defect: the role of kindlin-1 isoforms. **J Pathol** 213:462-70, 2007
  42. Mansur AT, Elcioglu NH, Aydingöz IE, Akkaya AD, Serdar ZA, Herz C, Bruckner-Tuderman L, **Has C**. Novel and recurrent KIND1 mutations in two patients with Kindler syndrome and severe mucosal involvement. **Acta Derm Venereol** 87:563-5, 2007
  43. **Has C**, Danescu S, Volz A, Nöh F, Technau K, Bruckner-Tuderman L. Incontinentia pigmenti in a newborn with a novel nonsense mutation in the NEMO gene. **Br J Dermatol** 156:392-3, 2007

44. **Has C**, Wessagowit V, Pascucci M, Baer C, Didona B, Wilhelm C, Pedicelli C, Locatelli A, Kohlhase J, Ashton GH, Tadini G, Zambruno G, Bruckner-Tuderman L, McGrath JA, Castiglia D. Molecular basis of Kindler syndrome in Italy: novel and recurrent Alu/Alu recombination, splice site, nonsense, and frameshift mutations in the KIND1 gene. **J Invest Dermatol** 126:1776-83, 2006
45. **Has C**, Chang YR, Volz A, Hoeping D, Kohlhase J, Bruckner-Tuderman L. Novel keratin 14 mutations in patients with severe recessive epidermolysis bullosa simplex. **J Invest Dermatol** 126:1912-4, 2006
46. Kern JS, Kohlhase J, Bruckner-Tuderman L, **Has C**. Expanding the COL7A1 mutation database: novel and recurrent mutations and unusual genotype-phenotype constellations in 41 patients with dystrophic epidermolysis bullosa. **J Invest Dermatol** 126:1006-12, 2006
47. Herz C, Aumailley M, Schulte C, Schlötzer-Schrehardt U, Bruckner-Tuderman L, **Has C**. Kindlin-1 is a phosphoprotein involved in regulation of polarity, proliferation, and motility of epidermal keratinocytes. **J Biol Chem** 281: 36082-90, 2006
48. **Has C**, Bruckner-Tuderman L. A novel nonsense mutation in Kindler syndrome. **J Invest Dermatol** 122:84-6, 2004
49. **Has C**, Seedorf U, Kannenberg F, Bruckner-Tuderman L, Folkers E, Fölster-Holst R, Baric I, Traupe H. Gas chromatography-mass spectrometry and molecular genetic studies in families with the Conradi-Hünemann-Happle syndrome. **J Invest Dermatol** 118:851-8, 2002
50. **Has C**, Bruckner-Tuderman L, Müller D, Floeth M, Folkers E, Donnai D, Traupe H. The Conradi-Hünemann-Happle syndrome (CDPX2) and emopamil binding protein: novel mutations, and somatic and gonadal mosaicism. **Hum Mol Genet** 9:1951-5, 2000
51. **Has C**, He Y. Focal adhesions in the skin: lessons learned from skin fragility disorders. **Eur J Dermatol.** 27(S1):8-11, 2017
52. **Has C**. The "Kelch" Surprise: KLHL24, a New Player in the Pathogenesis of Skin Fragility. **J Invest Dermatol.** 2017 137(6):1211-1212, 2017
53. **Has C**, He Y. Renal-skin syndromes. **Cell Tissue Res.** 369(1):63-73, 2017
54. **Has C**. Mosaicism in the skin: lumping or splitting? **Br J Dermatol** 176(1):15-16, 2017
55. Schwieger-Briel A, **Has C**. Update Genodermatosen. **Hautnah Dermatologie** 33(1): 36-45, 2017
56. **Has C**. Molecular therapies for inherited epidermolysis bullosa. **G Ital Dermatol Venereol** 151(4):397-402, 2016
57. **Has C**, He Y. Research Techniques Made Simple: Immunofluorescence Antigen Mapping in Epidermolysis Bullosa. **J Invest Dermatol** 136(7):e65-71, 2016
58. Tantcheva-Poór I, Oji V, **Has C**. Ein mehrstufiger Algorithmus zur Diagnose seltener Genodermatosen. **J Dtsch Dermatol Ges** 14(10):969-987, 2016
59. **Has C**, Technau-Hafsi K. Palmoplantar keratodermas: clinical and genetic aspects. **J Dtsch Dermatol Ges** 14(2):123-40, 2016
60. **Has C**. Hemidesmosomes: how much plakins do they need? **Exp Dermatol** 25(4):263-4, 2016
61. **Has C**, He Y. [Practical aspects of molecular diagnostics in genodermatoses]. **Hautarzt** 67(1):53-8, 2016
62. Schwieger-Briel A., **Has C**. Blasenbildende Erkrankungen bei Kindern. **Pädiatrische Praxis** 86, 135–142, 2016
63. **Has C**, Nyström A. Epidermal Basement Membrane in Health and Disease. **Curr Top Membr** 76:117-70, 2015
64. **Has C**, Kiritsi D. Therapies for inherited skin fragility disorders. **Exp Dermatol** 24(5):325-31, 2015

65. **Has C**, Giehl K. [Rare diseases are common]. **Hautarzt** 65(6):488-9, 2014
66. **Has C**, Kiritsi D. [The many facets of inherited skin fragility]. **Hautarzt** 65(6):490-8, 2014
67. **Has C**, Bruckner-Tuderman L. The genetics of skin fragility. **Annu Rev Genomics Hum Genet** 15:245-68, 2014
68. **Has C**. Processing of the laminin-332  $\alpha$  chain: from bedside to bench. **Br J Dermatol** 170(5):1008-9, 2014
69. Bruckner-Tuderman L, **Has C**. Disorders of the cutaneous basement membrane zone-The paradigm of epidermolysis bullosa. **Matrix Biol** 33C:29-34, 2014
70. **Has C**, Kiritsi D. Molecular therapies for epidermolysis bullosa. **G Ital Dermatol Venereol** 148:65-72, 2013
71. **Has C**, Sitaru C. Molecular dermatology comes of age. **Methods Mol Biol** 961:1-16, 2013
72. Bruckner-Tuderman L, **Has C**. Molecular heterogeneity of blistering disorders: the paradigm of epidermolysis bullosa. **J Invest Dermatol** 15;132(E1):E2-5, 2012
73. **Has C**. Mosaicism in the skin: the importance of mild or minimal skin lesions. **Arch Dermatol** 147:1094-6, 2011
74. **Has C**. Molecular genetic assays for inherited epidermolysis bullosa. **Clin Dermatol** 29:420-6, 2011
75. **Has C**, Bruckner-Tuderman L. [Epidermolysis bullosa: Diagnosis and therapy]. **Hautarzt** 62:82-90, 2011
76. **Has C**, Kern JS. Collagen XVII. **Dermatol Clin** 28(1):61-6, 2010
77. **Has C**. [Kindler syndrome. A new bullous dermatosis]. **Hautarzt** 60(8):622-6, 2009
78. **Has C**, Bruckner-Tuderman L. Molecular and diagnostic aspects of genetic skin fragility. **J Dermatol Sci** 44(3):129-44, 2006
79. **Has C**, Kern JS, Bruckner-Tuderman L. [Hereditary blistering disorders]. **Hautarzt** 55(10):920, 922-30, 2004
80. Traupe H, **Has C**. The Conradi-Hünemann-Happle syndrome is caused by mutations in the gene that encodes a 8-7 sterol isomerase and is biochemically related to the CHILD syndrome. **Eur J Dermatol** 10(6):425-8, 2000

### **Buchkapiteln**

(Auswahl, in der Reihenfolge der Aktualität)

1. **Has C**. Hereditäre Epidermolysen in Braun-Falco's Dermatologie, Venerologie und Allergologie Springer 2017, doi:10.1007/978-3-662-49546-9\_48-2
2. **Has C**, Nyström A, Epidermal Basement Membrane in Health and Disease. Basement membranes Ed. Jeffrey Miner, Curr Top Membr. 76:117-70, Elsevier 2015
3. **Has C**, Kapiteln in Blistering disorders, Ed. Dedee Murrell, Springer 2015  
 Acantholytic Forms of Epidermolysis Bullosa  
 Kindlin-1 and Its Role in Kindler Syndrome  
 Collagen XVII and Its Role in Junctional Epidermolysis Bullosa  
 Junctional Epidermolysis Bullosa with Renal and Respiratory Involvement: Integrin  $\alpha 3$  Mutations
4. **Has C**, Genodermatosen, in 1000 Fragen Dermatologie, Springer 2014
5. **Has C**, Bruckner-Tuderman L, Uitto J, Chapter 146 – Epidermolysis Bullosa, in Emery & Rimoin's Essential Medical Genetics Eds. Rimoin, Pyeritz & Korf, Elsevier Ltd. 2013

6. **Eds. Has C**, Sitaru C, Humana Press, Molecular Dermatology, Humana Press 2013
7. Bruckner-Tuderman L, **Has C**. Inherited bullous diseases in Therapy of skin diseases, Eds. Krieg T, Miyachi Y, Bickers DR, Springer 2010
8. Bruckner-Tuderman L, **Has C**. Epidermolysis bullosa in Pädiatrische Dermatologie Eds. Traupe H, Hamm H, Springer 2006

### Vorträge

(Auswahl, in der Reihenfolge der Aktualität)

1. **Schwieger-Briel A**, He Y, Has C. Monoallelic mutations in the translation initiation codon of KLHL24 cause skin fragility. World Congress of Pediatric Dermatology, Chicago 6.07.2017
2. **Has C**. Epidermolysis bullosa: from genes to therapies. British Association of Dermatologists, Liverpool 5.07.2017 (invited lecture)
3. **Has C**. Genodermatoses: case reports, 11<sup>th</sup> Genodermatoses Meeting, Freiburg, 29.06.2017
4. **Has C**. The kelch surprise: monoallelic mutations in the translation initiation codon of KLHL24 cause skin fragility. Summer School: Translational research for rare diseases. München 21.-22.06.2017
5. **Has C**. Palmoplantarkeratosen. Ärztliche Fortbildung, Freiburg 9.05.2017
6. **Has C**. Erbkrankheiten. 49. DDG-Tagung, Berlin 27.04.2017
7. **Has C**. The Kelch surprise: KLHL24 mutations cause skin fragility. Arbeitsgemeinschaft Dermatologische Forschung Göttingen, AG Genetik, 9.03.2017
8. **Has C**. Monoallelic mutations in KLHL24 cause skin fragility. Seminar GRK 1727 Klinik für Dermatologie Lübeck, 19.01.2017
9. **Has C**. Klinische und genetische Klassifikation der Epidermolysis bullosa. Beratertreffen Humangenetik, Freiburg, 13.01.2017
10. **Has C**. Syndromale Ichthyosen, Fortbildung, Klinik für Dermatologie und Venerologie Freiburg, 30.11.2016
11. **Has C**. Focal adhesions in the skin: lessons from skin Fragility disorders, Skin Physiology International Meeting, Vichy, 17.11.2016 (invited lecture)
12. **Has C**. Genodermatoses: case reports, 10<sup>th</sup> Genodermatoses Meeting, Zürich, 10.11.2016
13. **Has C**. Hereditary Ichthyoses and Palmoplantar keratodermas, Cluj Romania, 02.11.2016
14. **Has C**. Hereditary Ichthyoses, Iasi Romania, 31.10.2016
15. **Has C**. Vorstellung der Aktivitäten im Kompetenzzentrum, Mitgliederversammlung Kompetenzzentrum Seltene Erkrankungen Baden-Württemberg, Ulm, 24.10.2016
16. Kristin Maier, Yinghong He, Ute Wölfle, Philipp R. Esser, Tilman Brummer, Christoph Schempp, Leena Bruckner-Tuderman, **C Has**. Kindlin-1 protects keratinocytes against UV-B: prospects for antioxidant treatment in Kindler syndrome 46<sup>th</sup> Annual ESDR Meeting poster walk Presentation, München Germany 7-10.09.2016
17. He Y, ..., **Has C**. Alterations of the composition of the extracellular matrix of integrin  $\alpha 3$  deficient keratinocytes reflect basement membrane abnormalities in ILNEB 46th Annual ESDR Meeting, Poster walk presentation, München Germany, 7-10.09.2016



18. **Has C.** Epidermolysis bullosa: Klassifikation und Diagnostik, Fortbildung, Klinik für Dermatologie und Venerologie Freiburg, 3.08.2016
19. **Has C.** Vom Leitsymptom zur Diagnose der fragilen Haut, 25. Fortbildungswoche, München 27.07.2016 (invited lecture)
20. **Has C.** Nicht-syndromale Ichthyosen, Fortbildung, Klinik für Dermatologie und Venerologie Freiburg, 13.07.2016
21. **Has C.** Neues zu der Klassifikation und Genetik der hereditären Palmoplantarkeratosen, Universitäts-Hautklinik Homburg, 06.07.2016 (invited lecture)
22. **Has C.** Atypical Epidermolysis bullosa phenotypes of the Freiburg Clinic. Dept. Dermatology, University Groningen, Groningen, 6.06.2016
23. **Has C.** Genodermatoses: case reports, 9<sup>th</sup> Genodermatoses Meeting, Basel, 02.06.2016
24. **Has C.** Skin blistering disorders, European Society for Pediatric Dermatology, Paris, 25.05.2016
25. **Has C.** Molecular mechanisms in ILNEB, SFB 1140 Retreat 16.03.2016
26. **Has C.** Red scaly baby, Arbeitsgemeinschaft Dermatologische Forschung Wien, AG Genetik, 10.03.2016
27. **Has C.** Single amino acid deletion in kindlin-1 results in partial protein degradation which can be rescued by chaperone treatment, Arbeitsgemeinschaft Dermatologische Forschung Wien, AG Genetik, 10.03.2016
28. **Has C.** Horizonte in der Diagnostik und Behandlung der fragilen Haut, 25. Jahrestagung der AG Pädiatrische Dermatologie in der DDG und 11. Kölner Symposium „Haut und Kind“, Köln, 19.02.2016
29. **Has C.** Genodermatosen, Fortbildung Hautklinik Freiburg, 13.01.2016
30. **Has C.** Molecular mechanisms underlying interstitial lung disease, nephrotic syndrome, epidermolysis bullosa, 45<sup>th</sup> Annual ESDR Meeting, Rotterdam, 09.–12.09.2015
31. **Has C.** The genetics of skin fragility, EADV/ESDR Summer School, Marburg, 20.–21.07.2015 (invited lecture)
32. **Has C.** New forms of skin fragility, 23. World Congress of Dermatology, Vancouver, 09.06.2015 (invited lecture)
33. **Has C.** New EB classification, 23. World Congress of Dermatology, Vancouver, 09.06.2015 (invited lecture)
34. **Has C.** The Odyssey of genetically unsolved EB cases, 2015 Research Symposium, Debra International, Atlanta, 04.05.2015
35. **Has C.** Molecular diagnosis of bullous diseases, European Academy of Dermatology and Venereology, Amsterdam, 09.10.2014
36. **Has C.** Old and new treatment for genodermatoses, European Academy of Dermatology and Venereology, Amsterdam, 09.10.2014
37. **Has C.** FERMT1 promoter mutations in patients with Kindler syndrome, 44<sup>th</sup> Annual ESDR Meeting, Kopenhagen, 09.–11.09.2014
38. **Has C.** New cases of Epidermolysis bullosa, European Society for Pediatric Dermatology, Kiel, 12.06.2014
39. **Has C.** Focal adhesions and skin fragility disorders, Gordon Research Conference, New London NH, 2013

