Hans Christian Hennies

List of Publications by Year
in descending order

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90 papers	8,557 citations	66343 42 h-index	51608 86 g-index
91 all docs	91 docs citations	91 times ranked	10070 citing authors

#	Article	IF	Citations
1	Mutations in the gene encoding the serine protease inhibitor, Kazal type 1 are associated with chronic pancreatitis. Nature Genetics, 2000, 25, 213-216.	21.4	944
2	Revised nomenclature and classification of inherited ichthyoses: Results of the First Ichthyosis Consensus Conference in SorÃ¨ze 2009. Journal of the American Academy of Dermatology, 2010, 63, 607-641.	1.2	610
3	The centrosomal protein nephrocystin-6 is mutated in Joubert syndrome and activates transcription factor ATF4. Nature Genetics, 2006, 38, 674-681.	21.4	535
4	Positional cloning uncovers mutations in PLCE1 responsible for a nephrotic syndrome variant that may be reversible. Nature Genetics, 2006, 38, 1397-1405.	21.4	510
5	Linkage of familial hemophagocytic lymphohistiocytosis (FHL) type-4 to chromosome 6q24 and identification of mutations in syntaxin 11. Human Molecular Genetics, 2005, 14, 827-834.	2.9	502
6	Familial Hemophagocytic Lymphohistiocytosis Type 5 (FHL-5) Is Caused by Mutations in Munc18-2 and Impaired Binding to Syntaxin 11. American Journal of Human Genetics, 2009, 85, 482-492.	6.2	370
7	Increased Activity of Coagulation Factor XII (Hageman Factor) Causes Hereditary Angioedema Type III. American Journal of Human Genetics, 2006, 79, 1098-1104.	6.2	306
8	Mutation spectrum in children with primary hemophagocytic lymphohistiocytosis: molecular and functional analyses ofPRF1, UNC13D, STX11, andRAB27A. Human Mutation, 2006, 27, 62-68.	2.5	252
9	Loss of Corneodesmosin Leads to Severe Skin Barrier Defect, Pruritus, and Atopy: Unraveling the Peeling Skin Disease. American Journal of Human Genetics, 2010, 87, 274-281.	6.2	204
10	Wnt Signaling and Dupuytren's Disease. New England Journal of Medicine, 2011, 365, 307-317.	27.0	201
11	Genetic Variation in the Human Androgen Receptor Gene Is the Major Determinant of Common Early-Onset Androgenetic Alopecia. American Journal of Human Genetics, 2005, 77, 140-148.	6.2	198
12	A Gene Mutated in Nephronophthisis and Retinitis Pigmentosa Encodes a Novel Protein, Nephroretinin, Conserved in Evolution. American Journal of Human Genetics, 2002, 71, 1161-1167.	6.2	193
13	In vitro Modeling of Ryanodine Receptor 2 Dysfunction Using Human Induced Pluripotent Stem Cells. Cellular Physiology and Biochemistry, 2011, 28, 579-592.	1.6	179
14	A Truncating Mutation of CEP135 Causes Primary Microcephaly and Disturbed Centrosomal Function. American Journal of Human Genetics, 2012, 90, 871-878.	6.2	153
15	Mutations in AP3D1 associated with immunodeficiency and seizures define a new type of Hermansky-Pudlak syndrome. Blood, 2016, 127, 997-1006.	1.4	142
16	Gerodermia osteodysplastica is caused by mutations in SCYL1BP1, a Rab-6 interacting golgin. Nature Genetics, 2008, 40, 1410-1412.	21.4	138
17	Impaired Epidermal Ceramide Synthesis Causes Autosomal Recessive Congenital Ichthyosis and Reveals the Importance of Ceramide Acyl Chain Length. Journal of Investigative Dermatology, 2013, 133, 2202-2211.	0.7	138
18	Naegeli-Franceschetti-Jadassohn Syndrome and Dermatopathia Pigmentosa Reticularis: Two Allelic Ectodermal Dysplasias Caused by Dominant Mutations in KRT14. American Journal of Human Genetics, 2006, 79, 724-730.	6.2	114

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19	Novel Gene Locus for Autosomal Dominant Left Ventricular Noncompaction Maps to Chromosome 11p15. Circulation, 2004, 109, 2720-2723.	1.6	112
20	Cohen Syndrome-associated Protein, COH1, Is a Novel, Giant Golgi Matrix Protein Required for Golgi Integrity. Journal of Biological Chemistry, 2011, 286, 37665-37675.	3.4	111
21	Diverse Regulation of Claudin-1 and Claudin-4 in Atopic Dermatitis. American Journal of Pathology, 2015, 185, 2777-2789.	3.8	105
22	Periodontal Ehlers-Danlos Syndrome Is Caused by Mutations in C1R and C1S , which Encode Subcomponents C1r and C1s of Complement. American Journal of Human Genetics, 2016, 99, 1005-1014.	6.2	100
23	Molecular Analysis of 250 Patients with Autosomal Recessive Congenital Ichthyosis: Evidence for Mutation Hotspots in ALOXE3 and Allelic Heterogeneity in ALOX12B. Journal of Investigative Dermatology, 2009, 129, 1421-1428.	0.7	96
24	Mutation spectrum and functional analysis of epidermis-type lipoxygenases in patients with autosomal recessive congenital ichthyosis. Human Mutation, 2005, 26, 351-361.	2.5	91
25	Mutations in CSTA, Encoding Cystatin A, Underlie Exfoliative Ichthyosis and Reveal a Role for This Protease Inhibitor in Cell-Cell Adhesion. American Journal of Human Genetics, 2011, 89, 564-571.	6.2	89
26	Ichthyosis, Follicular Atrophoderma, and Hypotrichosis Caused by Mutations in ST14 Is Associated with Impaired Profilaggrin Processing. Journal of Investigative Dermatology, 2009, 129, 862-869.	0.7	88
27	GJB2mutations in keratitis-ichthyosis-deafness syndrome including its fatal form. , 2005, 133A, 128-131.		85
28	Penetration of normal, damaged and diseased skin â€" An in vitro study on dendritic coreâ€"multishell nanotransporters. Journal of Controlled Release, 2014, 185, 45-50.	9.9	79
29	Genotype/Phenotype Correlation in Autosomal Recessive Lamellar Ichthyosis. American Journal of Human Genetics, 1998, 62, 1052-1061.	6.2	77
30	Bathing suit ichthyosis is caused by transglutaminase-1 deficiency: evidence for a temperature-sensitive phenotype. Human Molecular Genetics, 2006, 15, 3083-3097.	2.9	76
31	Thermosensitive dendritic polyglycerol-based nanogels for cutaneous delivery of biomacromolecules. Nanomedicine: Nanotechnology, Biology, and Medicine, 2015, 11, 1179-1187.	3.3	74
32	A novel in situ method for the detection of deficient transglutaminase activity in the skin. Archives of Dermatological Research, 1998, 290, 621-627.	1.9	73
33	Allelic Heterogeneity in the COH1 Gene Explains Clinical Variabilityin Cohen Syndrome. American Journal of Human Genetics, 2004, 75, 138-145.	6.2	72
34	A Second Locus for an Axonal Form of Autosomal Recessive Charcot-Marie-Tooth Disease Maps to Chromosome 19q13.3. American Journal of Human Genetics, 2001, 68, 269-274.	6.2	71
35	Cohen Syndrome-associated Protein COH1 Physically and Functionally Interacts with the Small GTPase RAB6 at the Golgi Complex and Directs Neurite Outgrowth. Journal of Biological Chemistry, 2015, 290, 3349-3358.	3.4	68
36	A Genome-wide Association Study of Dupuytren Disease Reveals 17 Additional Variants Implicated in Fibrosis. American Journal of Human Genetics, 2017, 101, 417-427.	6.2	67

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37	Localisation of a gene for Papillon-LefÃ¨vre syndrome to chromosome 11q14-q21 by homozygosity mapping. Human Genetics, 1997, 101, 376-382.	3.8	66
38	Mal de Meleda (MDM) caused by mutations in the gene for SLURP-1 in patients from Germany, Turkey, Palestine, and the United Arab Emirates. Human Genetics, 2003, 112, 50-56.	3.8	63
39	Genome-wide Scan and Fine-Mapping Linkage Study of Androgenetic Alopecia Reveals a Locus on Chromosome 3q26. American Journal of Human Genetics, 2008, 82, 737-743.	6.2	62
40	Induced Pluripotent Mesenchymal Stromal Cell Clones Retain Donor-derived Differences in DNA Methylation Profiles. Molecular Therapy, 2013, 21, 240-250.	8.2	54
41	Genetic Aspects of Familial MÃ©niÃ¨re's Disease. Otology and Neurotology, 2011, 32, 695-700.	1.3	52
42	Genome-wide pooling approach identifies SPATA5 as a new susceptibility locus for alopecia areata. European Journal of Human Genetics, 2012, 20, 326-332.	2.8	48
43	MYO15A (DFNB3) mutations in Turkish hearing loss families and functional modeling of a novel motor domain mutation. American Journal of Medical Genetics, Part A, 2007, 143A, 2382-2389.	1.2	45
44	Expanded mutational spectrum in Cohen syndrome, tissue expression, and transcript variants ofCOH1. Human Mutation, 2009, 30, E404-E420.	2.5	44
45	Transglutaminase-1 and Bathing Suit Ichthyosis: Molecular Analysis of Gene/Environment Interactions. Journal of Investigative Dermatology, 2009, 129, 2068-2071.	0.7	41
46	Vestibular dysfunction of patients with mutations of Connexin 26. NeuroReport, 2005, 16, 1179-1181.	1.2	40
47	Epidermolytic palmoplantar keratoderma of VÃ¶rner: re-evaluation of VÃ¶rner's original family and identification of a novel keratin 9 mutation. Archives of Dermatological Research, 2002, 294, 268-272.	1.9	39
48	Localization of the Gene Causing Keratolytic Winter Erythema to Chromosome 8p22-p23, and Evidence for a Founder Effect in South African Afrikaans-Speakers. American Journal of Human Genetics, 1997, 61, 370-378.	6.2	38
49	Identification, by Homozygosity Mapping, of a Novel Locus for Autosomal Recessive Congenital Ichthyosis on Chromosome 17p, and Evidence for Further Genetic Heterogeneity. American Journal of Human Genetics, 2001, 69, 216-222.	6.2	37
50	Role of epidermis-type lipoxygenases for skin barrier function and adipocyte differentiation. Prostaglandins and Other Lipid Mediators, 2007, 82, 128-134.	1.9	37
51	Effective Immunological Guidance of Genetic Analyses Including Exome Sequencing in Patients Evaluated for Hemophagocytic Lymphohistiocytosis. Journal of Clinical Immunology, 2017, 37, 770-780.	3.8	37
52	Mutations in SNRPE, which Encodes a Core Protein of the Spliceosome, Cause Autosomal-Dominant Hypotrichosis Simplex. American Journal of Human Genetics, 2013, 92, 81-87.	6.2	36
53	Palmoplantar keratoderma (PPK): acquired and genetic causes of a not so rare disease. JDDG - Journal of the German Society of Dermatology, 2014, 12, 781-788.	0.8	35
54	A Novel Locus for Dilated Cardiomyopathy, Diffuse Myocardial Fibrosis, and Sudden Death on Chromosome 10q25-26. Journal of the American College of Cardiology, 2006, 48, 106-111.	2.8	34

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55	Medullary cystic kidney disease type 1: mutational analysis in 37 genes based on haplotype sharing. Human Genetics, 2006, 119, 649-658.	3.8	34
56	Full-Thickness Human Skin Models for Congenital Ichthyosis and Related Keratinization Disorders. Journal of Investigative Dermatology, 2011, 131, 1938-1942.	0.7	34
57	SPINK1 mutations in chronic pancreatitis. Gastroenterology, 2001, 120, 1060-1061.	1.3	29
58	Genetic and immunohistochemical detection of mutations inactivating the keratinocyte transglutaminase in patients with lamellar ichthyosis. Human Genetics, 1998, 102, 314-318.	3.8	28
59	Homozygosity mapping as a screening tool for the molecular diagnosis of hereditary skin diseases in consanguineous populations. Journal of the American Academy of Dermatology, 2006, 55, 393-401.	1.2	27
60	Rapid detection of homozygous mutations in congenital recessive ichthyosis. Archives of Dermatological Research, 2008, 300, 81-85.	1.9	27
61	Long-Term Faithful Recapitulation of Transglutaminase 1â€“Deficient Lamellar Ichthyosis in a Skin-Humanized Mouse Model, and Insights from Proteomic Studies. Journal of Investigative Dermatology, 2012, 132, 1918-1921.	0.7	27
62	Mapping a new suggestive gene locus for autosomal dominant nephrolithiasis to chromosome 9q33.2â€“q34.2 by total genome search for linkage. Nephrology Dialysis Transplantation, 2005, 20, 909-914.	0.7	26
63	Meta-Analysis of Genome-Wide Association Studies and Network Analysis-Based Integration with Gene Expression Data Identify New Suggestive Loci and Unravel a Wnt-Centric Network Associated with Dupuytrenâ€™s Disease. PLoS ONE, 2016, 11, e0158101.	2.5	26
64	Transglutaminase 1 Replacement Therapy Successfully Mitigates the Autosomal Recessive Congenital Ichthyosis Phenotype in Full-Thickness Skin Disease Equivalents. Journal of Investigative Dermatology, 2019, 139, 1191-1195.	0.7	24
65	Telomeric refinement of the MCKD1 locuson chromosome 1q21**See Editorial by Bichet and Fujiwara, p. 864.. Kidney International, 2004, 66, 580-585.	5.2	20
66	Assignment of the Gene for a New Hereditary Nail Disorder, Isolated Congenital Nail Dysplasia, to Chromosome 17p13. Journal of Investigative Dermatology, 2000, 115, 664-667.	0.7	17
67	Focal palmoplantar and gingival keratosis: A distinct palmoplantar ectodermal dysplasia with epidermolytic alterations but lack of mutations in known keratins. Journal of the American Academy of Dermatology, 2005, 52, 403-409.	1.2	15
68	Physical and transcriptional map of the critical region for keratolytic winter erythema (KWE) on chromosome 8p22-p23 between D8S550 and D8S1759. European Journal of Human Genetics, 2002, 10, 17-25.	2.8	14
69	IGFBP7 as a Potential Therapeutic Target in Psoriasis. Journal of Investigative Dermatology, 2011, 131, 1767-1770.	0.7	14
70	Increased cutaneous absorption reflects impaired barrier function of reconstructed skin models mimicking keratinisation disorders. Experimental Dermatology, 2014, 23, 286-288.	2.9	14
71	S1 guidelines for the diagnosis and treatment of ichthyoses â€“ update. JDDG - Journal of the German Society of Dermatology, 2017, 15, 1053-1065.	0.8	14
72	Genetic and Clinical Heterogeneity in Transgressive Palmoplantar Keratoderma. Journal of Investigative Dermatology, 2001, 116, 825-827.	0.7	13

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73	Identification and Localization of a New Human Myotubularin-Related Protein Gene, MTMR8, on 8p22â€“p23. Genomics, 2001, 75, 6-8.	2.9	12
74	A Novel Homozygous Missense Mutation in SLURP1 Causing Mal de Meleda With an Atypical Phenotype. Archives of Dermatology, 2011, 147, 748.	1.4	12
75	Bathing suit ichthyosis. European Journal of Dermatology, 2010, 20, 447-450.	0.6	11
76	Morphological alterations in two siblings with autosomal recessive congenital ichthyosis associated with CYP4F22 mutations. British Journal of Dermatology, 2017, 176, 1068-1073.	1.5	10
77	Cystatin <scp>M/E</scp> knockdown by lentiviral delivery of sh<scp>RNA</scp> impairs epidermal morphogenesis of human skin equivalents. Experimental Dermatology, 2012, 21, 889-891.	2.9	9
78	Unknown mutations and genotype/phenotype correlations of autosomal recessive congenital ichthyosis in patients from Saudi Arabia and Pakistan. Molecular Genetics & Genomic Medicine, 2019, 7, e539.	1.2	9
79	A Gene Locus for Steroid-Resistant Nephrotic Syndrome with Deafness Maps to Chromosome 14q24.2. Journal of the American Society of Nephrology: JASN, 2003, 14, 1519-1522.	6.1	8
80	All is balanced: interâ€Î±â€trypsin inhibitors as unseen extracellular matrix proteins in epidermal morphology and differentiation. Experimental Dermatology, 2015, 24, 661-662.	2.9	8
81	Skin Disease Models In Vitro and Inflammatory Mechanisms: Predictability for Drug Development. Handbook of Experimental Pharmacology, 2021, 265, 187-218.	1.8	8
82	Mutation analysis of theM6bgene in patients with Pelizaeus-Merzbacher-like syndrome. , 2004, 128A, 156-158.		7
83	S1â€Leitlinie zur Diagnostik und Therapie der Ichthyosen â€“ Aktualisierung. JDDG - Journal of the German Society of Dermatology, 2017, 15, 1053-1065.	0.8	7
84	Netherton syndrome previously misdiagnosed as hyper ige syndrome caused by a probable mutation in spink5 c. Turkish Journal of Pediatrics, 2019, 61, 604.	0.6	7
85	Genome-Wide Association Scan of Dupuytren's Disease. Journal of Hand Surgery, 2011, 36, 755-756.	1.6	4
86	hiPSC-Derived Epidermal Keratinocytes from Ichthyosis Patients Show Altered Expression of Cornification Markers. International Journal of Molecular Sciences, 2021, 22, 1785.	4.1	4
87	Hypoplastic thyroid, growth hormone deficiency, corneal opacities, cataract and hyperkeratotic skin disease: a possible new ichthyosis syndrome associated with endocrinopathies. Archives of Dermatological Research, 2005, 296, 585-587.	1.9	3
88	Bathing Suit Variant of Autosomal Recessive Congenital Ichthyosis (ARCI) in Two Indian Patients. Case Reports in Dermatological Medicine, 2018, 2018, 1-4.	0.3	2
89	Nanogel-Mediated Protein Replacement Therapy for Autosomal Recessive Congenital Ichthyosis (ARCI). , 0, , .		2
90	A novel eicosanoid pathway is essential for the epidermal barrier function: lessons from congenital ichthyosis and 12R-lipoxygenase deficient mice. Chemistry and Physics of Lipids, 2009, 160, S16.	3.2	0