Ulrike HÃ¼ffmeier

List of Publications by Year
in descending order

76 papers	6,154 citations	109321 35 h-index	76900 74 g-index
79 all docs	79 docs citations	79 times ranked	9489 citing authors

#	Article	IF	Citations
1	A genome-wide association study identifies new psoriasis susceptibility loci and an interaction between HLA-C and ERAP1. Nature Genetics, 2010, 42, 985-990.	21.4	918
2	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. Nature Genetics, 2012, 44, 1341-1348.	21.4	848
3	Psoriasis is associated with increased Î²-defensin genomic copy number. Nature Genetics, 2008, 40, 23-25.	21.4	587
4	Association Between Telomere Length and Risk of Cancer and Non-Neoplastic Diseases. JAMA Oncology, 2017, 3, 636.	7.1	376
5	Diagnostic yield of various genetic approaches in patients with unexplained developmental delay or mental retardation. American Journal of Medical Genetics, Part A, 2006, 140A, 2063-2074.	1.2	343
6	Common variants at TRAF3IP2 are associated with susceptibility to psoriatic arthritis and psoriasis. Nature Genetics, 2010, 42, 996-999.	21.4	334
7	Identification of ZNF313 / RNF114 as a novel psoriasis susceptibility gene. Human Molecular Genetics, 2008, 17, 1938-1945.	2.9	176
8	Dense genotyping of immune-related susceptibility loci reveals new insights into the genetics of psoriatic arthritis. Nature Communications, 2015, 6, 6046.	12.8	149
9	Systematic assessment of atypical deletions reveals genotype-phenotype correlation in 22q11.2. Journal of Medical Genetics, 2005, 42, 871-876.	3.2	118
10	Genetic Variants of the IL-23R Pathway: Association with Psoriatic Arthritis and Psoriasis Vulgaris, but No Specific Risk Factor for Arthritis. Journal of Investigative Dermatology, 2009, 129, 355-358.	0.7	97
11	Successful therapy with anakinra in a patient with generalized pustular psoriasis carryingIL36RNmutations. British Journal of Dermatology, 2014, 170, 202-204.	1.5	95
12	Meta-Analysis Confirms the LCE3C_LCE3B Deletion as a Risk Factor for Psoriasis in Several Ethnic Groups and Finds Interaction with HLA-Cw6. Journal of Investigative Dermatology, 2011, 131, 1105-1109.	0.7	89
13	Mutations in IL36RN in Patients with Generalized Pustular Psoriasis. Journal of Investigative Dermatology, 2013, 133, 2634-2637.	0.7	89
14	TNF polymorphisms in psoriasis: Association of psoriatic arthritis with the promoter polymorphismTNF*-857 independent of thePSORS1 risk allele. Arthritis and Rheumatism, 2007, 56, 2056-2064.	6.7	88
15	The genetic basis for most patients with pustular skin disease remains elusive. British Journal of Dermatology, 2018, 178, 740-748.	1.5	82
16	Palmoplantar Pustular Psoriasis Is Associated with Missense Variants in CARD14 , but Not with Loss-of-Function Mutations in IL36RN in European Patients. Journal of Investigative Dermatology, 2015, 135, 2538-2541.	0.7	78
17	Molecular karyotyping in patients with mental retardation using 100K single-nucleotide polymorphism arrays. Journal of Medical Genetics, 2007, 44, 629-636.	3.2	72
18	Cytokine balance in infants undergoing cardiac operation. Annals of Thoracic Surgery, 2002, 73, 601-608.	1.3	66

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19	PTPN22 is associated with susceptibility to psoriatic arthritis but not psoriasis: evidence for a further PsA-specific risk locus. Annals of the Rheumatic Diseases, 2015, 74, 1882-1885.	0.9	64
20	Variants in RUNX3 Contribute to Susceptibility to Psoriatic Arthritis, Exhibiting Further Common Ground With Ankylosing Spondylitis. Arthritis and Rheumatism, 2013, 65, 1224-1231.	6.7	63
21	Replication of LCE3Câ€“LCE3B CNV as a Risk Factor for Psoriasis and Analysis of Interaction with Other Genetic Risk Factors. Journal of Investigative Dermatology, 2010, 130, 979-984.	0.7	61
22	Conditional analysis identifies three novel major histocompatibility complex loci associated with psoriasis. Human Molecular Genetics, 2012, 21, 5185-5192.	2.9	58
23	Myeloperoxidase Modulates Inflammation in Generalized Pustular Psoriasis and Additional Rare Pustular Skin Diseases. American Journal of Human Genetics, 2020, 107, 527-538.	6.2	53
24	Association of Î²-Defensin Copy Number and Psoriasis in Three Cohorts of European Origin. Journal of Investigative Dermatology, 2012, 132, 2407-2413.	0.7	50
25	The mutational and phenotypic spectrum of TUBA1A-associated tubulinopathy. Orphanet Journal of Rare Diseases, 2019, 14, 38.	2.7	48
26	Rare Loss-of-Function Mutation in SERPINA3 in Generalized Pustular Psoriasis. Journal of Investigative Dermatology, 2020, 140, 1451-1455.e13.	0.7	48
27	Spatially clustering de novo variants in CYFIP2, encoding the cytoplasmic FMRP interacting protein 2, cause intellectual disability and seizures. European Journal of Human Genetics, 2019, 27, 747-759.	2.8	47
28	A variable combination of features of Noonan syndrome and neurofibromatosis type I are caused by mutations in the NF1 gene. American Journal of Medical Genetics, Part A, 2006, 140A, 2749-2756.	1.2	46
29	Loss-of-Function Myeloperoxidase Mutations Are Associated with Increased Neutrophil Counts and Pustular Skin Disease. American Journal of Human Genetics, 2020, 107, 539-543.	6.2	44
30	Estimating the effect size of the 15Q11.2 BP1â€“BP2 deletion and its contribution to neurodevelopmental symptoms: recommendations for practice. Journal of Medical Genetics, 2019, 56, 701-710.	3.2	43
31	Genotyping Microarray for CSNB-Associated Genes. , 2009, 50, 5919.		41
32	Exome-wide association study reveals novel psoriasis susceptibility locus at TNFSF15 and rare protective alleles in genes contributing to type I IFN signalling. Human Molecular Genetics, 2017, 26, 4301-4313.	2.9	41
33	Ichthyosis vulgaris: novelFLGmutations in the German population and high presence of CD1a+ cells in the epidermis of the atopic subgroup. British Journal of Dermatology, 2009, 160, 771-781.	1.5	40
34	Loss-of-Function Variants of the Filaggrin Gene Are Not Major Susceptibility Factors for Psoriasis Vulgaris or Psoriatic Arthritis in German Patients. Journal of Investigative Dermatology, 2007, 127, 1367-1370.	0.7	39
35	Tumor necrosis factor receptor-associated periodic syndrome characterized by a mutation affecting the cleavage site of the receptor: Implications for pathogenesis. Arthritis and Rheumatism, 2003, 48, 2386-2388.	6.7	38
36	Systematic Linkage Disequilibrium Analysis of SLC12A8 at PSORS5 Confirms a Role in Susceptibility to Psoriasis Vulgaris. Journal of Investigative Dermatology, 2005, 125, 906-912.	0.7	38

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37	Association between protein tyrosine phosphatase 22 variant R620W in conjunction with the HLAâ€“DRB1 shared epitope and humoral autoimmunity to an immunodominant epitope of cartilage-specific type II collagen in early rheumatoid arthritis. Arthritis and Rheumatism, 2006, 54, 82-89.	6.7	36
38	Bainbridgeâ€“Ropers syndrome caused by loss-of-function variants in ASXL3: a recognizable condition. European Journal of Human Genetics, 2017, 25, 183-191.	2.8	35
39	Characterisation of psoriasis susceptibility locus 6 (PSORS6) in patients with early onset psoriasis and evidence for interaction with PSORS1. Journal of Medical Genetics, 2009, 46, 736-744.	3.2	34
40	Deletion of LCE3C and LCE3B genes at PSORS4 does not contribute to susceptibility to psoriatic arthritis in German patients. Annals of the Rheumatic Diseases, 2010, 69, 876-878.	0.9	34
41	Mannan-induced Nos2 in macrophages enhances IL-17â€“driven psoriatic arthritis by innate lymphocytes. Science Advances, 2018, 4, eaas9864.	10.3	33
42	Evidence for susceptibility determinant(s) to psoriasis vulgaris in or near PTPN22 in German patients. Journal of Medical Genetics, 2006, 43, 517-522.	3.2	31
43	New Mutations of EXT1 and EXT2 Genes in German Patients with Multiple Osteochondromas. Annals of Human Genetics, 2009, 73, 283-291.	0.8	31
44	Deletion of LCE3C and LCE3B is a susceptibility factor for psoriatic arthritis: A study in Spanish and Italian populations and meta-analysis. Arthritis and Rheumatism, 2011, 63, 1860-1865.	6.7	31
45	Successful treatment with interleukinâ€17A antagonists of generalized pustular psoriasis in patients without IL36RN mutations. Journal of Dermatology, 2018, 45, 850-854.	1.2	31
46	Severe skeletal dysplasia caused by undiagnosed hypothyroidism. European Journal of Medical Genetics, 2007, 50, 209-215.	1.3	30
47	Lack of genetic association of the three more common polymorphisms of CARD15 with psoriatic arthritis and psoriasis in a German cohort. Annals of the Rheumatic Diseases, 2005, 64, 951-954.	0.9	29
48	Male Restricted Genetic Association of Variant R620W in PTPN22 with Psoriatic Arthritis. Journal of Investigative Dermatology, 2006, 126, 936-938.	0.7	28
49	Association scan of the novel psoriasis susceptibility region on chromosome 19: evidence for both susceptible and protective loci. Experimental Dermatology, 2003, 12, 490-496.	2.9	26
50	Interleukin-10 promoter polymorphism IL10.G and familial early onset psoriasis. British Journal of Dermatology, 2003, 149, 381-385.	1.5	25
51	Tumor necrosis factor promoter polymorphism TNF*-857 is a risk allele for psoriatic arthritis independent of the PSORS1 locus. Arthritis and Rheumatism, 2011, 63, 3801-3806.	6.7	25
52	Lack of Evidence for Genetic Association to RUNX1 Binding Site at PSORS2 in Different German Psoriasis Cohorts. Journal of Investigative Dermatology, 2005, 124, 107-110.	0.7	22
53	Loss-of-function mutations in the filaggrin gene: no contribution to disease susceptibility, but to autoantibody formation against citrullinated peptides in early rheumatoid arthritis. Annals of the Rheumatic Diseases, 2008, 67, 131-133.	0.9	21
54	Evidence for genetic overlap between adult onset Stillâ€™s disease and hereditary periodic fever syndromes. Rheumatology International, 2018, 38, 111-120.	3.0	20

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55	Fine Mapping of the Psoriasis Susceptibility Gene PSORS1: A Reassessment of Risk Associated with a Putative Risk Haplotype Lacking HLA-Cw6. Journal of Investigative Dermatology, 2005, 124, 921-930.	0.7	18
56	Identification of low frequency TRAF3IP2 coding variants in psoriatic arthritis patients and functional characterization. Arthritis Research and Therapy, 2012, 14, R84.	3.5	16
57	KIF3A and IL-4 are disease-specific biomarkers for psoriatic arthritis susceptibility. Oncotarget, 2017, 8, 95401-95411.	1.8	12
58	Do the exome: A case of Williams-Beuren syndrome with severe epilepsy due to a truncating de novo variant in GABRA1. European Journal of Medical Genetics, 2016, 59, 549-553.	1.3	11
59	Complex Role of TNF Variants in Psoriatic Arthritis and Treatment Response to Anti-TNF Therapy: Evidence and Concepts. Journal of Investigative Dermatology, 2014, 134, 2483-2485.	0.7	10
60	Genetic variants in FBLIM1 gene do not contribute to SAPHO syndrome and chronic recurrent multifocal osteomyelitis in typical patient groups. BMC Medical Genetics, 2020, 21, 102.	2.1	10
61	Replication of a distinct psoriatic arthritis risk variant at theIL23Rlocus. Annals of the Rheumatic Diseases, 2016, 75, 1417-1418.	0.9	9
62	Genome-wide association and targeted analysis of copy number variants with psoriatic arthritis in German patients. BMC Medical Genetics, 2017, 18, 92.	2.1	8
63	Transcriptomes of MPO-Deficient Patients with Generalized Pustular Psoriasis Reveals Expansion of CD4+ Cytotoxic T Cells and an Involvement of the Complement System. Journal of Investigative Dermatology, 2022, 142, 2149-2158.e10.	0.7	7
64	Analyses of association of psoriatic arthritis and psoriasis vulgaris with functional NCF1 variants. Rheumatology, 2019, 58, 915-917.	1.9	6
65	EIF3F-related neurodevelopmental disorder: refining the phenotypic and expanding the molecular spectrum. Orphanet Journal of Rare Diseases, 2021, 16, 136.	2.7	5
66	<scp>ZMYND11</scp> variants are a novel cause of centrotemporal and generalised epilepsies with neurodevelopmental disorder. Clinical Genetics, 2021, 100, 412-429.	2.0	5
67	Lack of genetic association of the interleukin-4 receptor single-nucleotide polymorphisms I50V and Q551R with erosive disease in psoriatic arthritis. Arthritis and Rheumatism, 2006, 54, 4023-4024.	6.7	4
68	Association analysis of psoriasis vulgaris and psoriatic arthritis with lossâ€ofâ€function mutations in IL 36 RN in German patients. British Journal of Dermatology, 2016, 175, 639-641.	1.5	4
69	Genetic Analysis of MPO Variants in Four Psoriasis Subtypes in Patients from Germany. Journal of Investigative Dermatology, 2021, 141, 2079-2083.	0.7	3
70	Novel autosomal recessive progressive hyperpigmentation syndrome. American Journal of Medical Genetics, Part A, 2005, 135A, 195-199.	1.2	2
71	Tumor necrosis factor polymorphisms in psoriatic arthritis: association with the promoter polymorphism TNF-857 independent of the PSORS1 risk allele. Arthritis Research and Therapy, 2007, 9, P18.	3.5	2
72	Genetik der Psoriasis. Medizinische Genetik, 2007, 19, 350-355.	0.2	2

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73	OP0128â€…PTPN22 is Associated with Susceptibility to Psoriatic Arthritis but not Psoriasis: Evidence for a Further PSA-Specific Risk Locus. Annals of the Rheumatic Diseases, 2015, 74, 116.3-117.	0.9	1
74	O53.â€ƒPTPN22 is Associated with Susceptibility to Psoriatic Arthritis but not Psoriasis: Evidence for a Further PSA-Specific Risk Locus. Rheumatology, 2015, , .	1.9	1
75	Neue Entwicklungen in der Psoriasisgenetik. Medizinische Genetik, 2009, 21, 498-504.	0.2	0
76	SAT0011â€…Replication of A Distinct Psoriatic Arthritis Risk Variant at IL23R. Annals of the Rheumatic Diseases, 2016, 75, 667.3-668.	0.9	0