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	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
2	EIF3F-related neurodevelopmental disorder: refining the phenotypic and expanding the molecular spectrum. Orphanet Journal of Rare Diseases, 2021, 16, 136.	2.7	5
3	<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
4	Genetic Analysis of MPO Variants in Four Psoriasis Subtypes in Patients from Germany. Journal of Investigative Dermatology, 2021, 141, 2079-2083.	0.7	3
5	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
6	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
7	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
8	Rare Loss-of-Function Mutation in SERPINA3 in Generalized Pustular Psoriasis. Journal of Investigative Dermatology, 2020, 140, 1451-1455.e13.	0.7	48
9	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
10	Analyses of association of psoriatic arthritis and psoriasis vulgaris with functional NCF1 variants. Rheumatology, 2019, 58, 915-917.	1.9	6
11	The mutational and phenotypic spectrum of TUBA1A-associated tubulinopathy. Orphanet Journal of Rare Diseases, 2019, 14, 38.	2.7	48
12	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
13	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
14	Evidence for genetic overlap between adult onset Stillâ€™s disease and hereditary periodic fever syndromes. Rheumatology International, 2018, 38, 111-120.	3.0	20
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	Mannan-induced Nos2 in macrophages enhances IL-17â€“driven psoriatic arthritis by innate lymphocytes. Science Advances, 2018, 4, eaas9864.	10.3	33
17	Association Between Telomere Length and Risk of Cancer and Non-Neoplastic Diseases. JAMA Oncology, 2017, 3, 636.	7.1	376
18	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

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19	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
20	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
21	KIF3A and IL-4 are disease-specific biomarkers for psoriatic arthritis susceptibility. Oncotarget, 2017, 8, 95401-95411.	1.8	12
22	SAT0011â€…Replication of A Distinct Psoriatic Arthritis Risk Variant at IL23R. Annals of the Rheumatic Diseases, 2016, 75, 667.3-668.	0.9	0
23	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
24	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
25	Replication of a distinct psoriatic arthritis risk variant at theIL23Rlocus. Annals of the Rheumatic Diseases, 2016, 75, 1417-1418.	0.9	9
26	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
27	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
28	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
29	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
30	Dense genotyping of immune-related susceptibility loci reveals new insights into the genetics of psoriatic arthritis. Nature Communications, 2015, 6, 6046.	12.8	149
31	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
32	Successful therapy with anakinra in a patient with generalized pustular psoriasis carryingIL36RNmutations. British Journal of Dermatology, 2014, 170, 202-204.	1.5	95
33	Mutations in IL36RN in Patients with Generalized Pustular Psoriasis. Journal of Investigative Dermatology, 2013, 133, 2634-2637.	0.7	89
34	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
35	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
36	Conditional analysis identifies three novel major histocompatibility complex loci associated with psoriasis. Human Molecular Genetics, 2012, 21, 5185-5192.	2.9	58

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37	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. Nature Genetics, 2012, 44, 1341-1348.	21.4	848
38	Identification of low frequency TRAF3IP2 coding variants in psoriatic arthritis patients and functional characterization. Arthritis Research and Therapy, 2012, 14, R84.	3.5	16
39	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
40	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
41	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
42	Common variants at TRAF3IP2 are associated with susceptibility to psoriatic arthritis and psoriasis. Nature Genetics, 2010, 42, 996-999.	21.4	334
43	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
44	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
45	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
46	Genotyping Microarray for CSNB-Associated Genes. , 2009, 50, 5919.		41
47	Neue Entwicklungen in der Psoriasisgenetik. Medizinische Genetik, 2009, 21, 498-504.	0.2	0
48	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
49	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
50	New Mutations of EXT1 and EXT2 Genes in German Patients with Multiple Osteochondromas. Annals of Human Genetics, 2009, 73, 283-291.	0.8	31
51	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
52	Psoriasis is associated with increased Î²-defensin genomic copy number. Nature Genetics, 2008, 40, 23-25.	21.4	587
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	Identification of ZNF313 / RNF114 as a novel psoriasis susceptibility gene. Human Molecular Genetics, 2008, 17, 1938-1945.	2.9	176

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55	Molecular karyotyping in patients with mental retardation using 100K single-nucleotide polymorphism arrays. Journal of Medical Genetics, 2007, 44, 629-636.	3.2	72
56	Severe skeletal dysplasia caused by undiagnosed hypothyroidism. European Journal of Medical Genetics, 2007, 50, 209-215.	1.3	30
57	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
58	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
59	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
60	Genetik der Psoriasis. Medizinische Genetik, 2007, 19, 350-355.	0.2	2
61	Male Restricted Genetic Association of Variant R620W in PTPN22 with Psoriatic Arthritis. Journal of Investigative Dermatology, 2006, 126, 936-938.	0.7	28
62	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
63	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
64	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
65	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
66	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
67	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
68	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
69	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
70	Novel autosomal recessive progressive hyperpigmentation syndrome. American Journal of Medical Genetics, Part A, 2005, 135A, 195-199.	1.2	2
71	Systematic assessment of atypical deletions reveals genotype-phenotype correlation in 22q11.2. Journal of Medical Genetics, 2005, 42, 871-876.	3.2	118
72	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

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73	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
74	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
75	Interleukin-10 promoter polymorphism IL10.G and familial early onset psoriasis. British Journal of Dermatology, 2003, 149, 381-385.	1.5	25
76	Cytokine balance in infants undergoing cardiac operation. Annals of Thoracic Surgery, 2002, 73, 601-608.	1.3	66