

Ulrike Häffmeier

List of Publications by Year in descending order

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76
papers

6,154
citations

109137

35
h-index

76769

74
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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. <i>Journal of Investigative Dermatology</i> , 2022, 142, 2149-2158.e10.	0.3	7
2	EIF3F-related neurodevelopmental disorder: refining the phenotypic and expanding the molecular spectrum. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 136.	1.2	5
3	<sc><i>ZMYND11</i></sc> variants are a novel cause of centrotemporal and generalised epilepsies with neurodevelopmental disorder. <i>Clinical Genetics</i> , 2021, 100, 412-429.	1.0	5
4	Genetic Analysis of MPO Variants in Four Psoriasis Subtypes in Patients from Germany. <i>Journal of Investigative Dermatology</i> , 2021, 141, 2079-2083.	0.3	3
5	Loss-of-Function Myeloperoxidase Mutations Are Associated with Increased Neutrophil Counts and Pustular Skin Disease. <i>American Journal of Human Genetics</i> , 2020, 107, 539-543.	2.6	44
6	Myeloperoxidase Modulates Inflammation in Generalized Pustular Psoriasis and Additional Rare Pustular Skin Diseases. <i>American Journal of Human Genetics</i> , 2020, 107, 527-538.	2.6	53
7	Genetic variants in FBLIM1 gene do not contribute to SAPHO syndrome and chronic recurrent multifocal osteomyelitis in typical patient groups. <i>BMC Medical Genetics</i> , 2020, 21, 102.	2.1	10
8	Rare Loss-of-Function Mutation in SERPINA3 in Generalized Pustular Psoriasis. <i>Journal of Investigative Dermatology</i> , 2020, 140, 1451-1455.e13.	0.3	48
9	Spatially clustering de novo variants in CYFIP2, encoding the cytoplasmic FMRP interacting protein 2, cause intellectual disability and seizures. <i>European Journal of Human Genetics</i> , 2019, 27, 747-759.	1.4	47
10	Analyses of association of psoriatic arthritis and psoriasis vulgaris with functional NCF1 variants. <i>Rheumatology</i> , 2019, 58, 915-917.	0.9	6
11	The mutational and phenotypic spectrum of TUBA1A-associated tubulinopathy. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 38.	1.2	48
12	Estimating the effect size of the 15Q11.2 BP1â€“BP2 deletion and its contribution to neurodevelopmental symptoms: recommendations for practice. <i>Journal of Medical Genetics</i> , 2019, 56, 701-710.	1.5	43
13	Successful treatment with interleukinâ€“17A antagonists of generalized pustular psoriasis in patients without <i>IL36RN</i> mutations. <i>Journal of Dermatology</i> , 2018, 45, 850-854.	0.6	31
14	Evidence for genetic overlap between adult onset Stillâ€“s disease and hereditary periodic fever syndromes. <i>Rheumatology International</i> , 2018, 38, 111-120.	1.5	20
15	The genetic basis for most patients with pustular skin disease remains elusive. <i>British Journal of Dermatology</i> , 2018, 178, 740-748.	1.4	82
16	Mannan-induced Nos2 in macrophages enhances IL-17â€“driven psoriatic arthritis by innate lymphocytes. <i>Science Advances</i> , 2018, 4, eaas9864.	4.7	33
17	Association Between Telomere Length and Risk of Cancer and Non-Neoplastic Diseases. <i>JAMA Oncology</i> , 2017, 3, 636.	3.4	376
18	Bainbridgeâ€“Ropers syndrome caused by loss-of-function variants in ASXL3: a recognizable condition. <i>European Journal of Human Genetics</i> , 2017, 25, 183-191.	1.4	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. <i>Human Molecular Genetics</i> , 2017, 26, 4301-4313.	1.4	41
20	Genome-wide association and targeted analysis of copy number variants with psoriatic arthritis in German patients. <i>BMC Medical Genetics</i> , 2017, 18, 92.	2.1	8
21	<i>KIF3A</i> and <i>IL-4</i> are disease-specific biomarkers for psoriatic arthritis susceptibility. <i>Oncotarget</i> , 2017, 8, 95401-95411.	0.8	12
22	SAT0011â€¦Replication of A Distinct Psoriatic Arthritis Risk Variant at IL23R. <i>Annals of the Rheumatic Diseases</i> , 2016, 75, 667.3-668.	0.5	0
23	Do the exome: A case of Williams-Beuren syndrome with severe epilepsy due to a truncating de novo variant in GABRA1. <i>European Journal of Medical Genetics</i> , 2016, 59, 549-553.	0.7	11
24	Association analysis of psoriasis vulgaris and psoriatic arthritis with loss-of-function mutations in IL36 RN in German patients. <i>British Journal of Dermatology</i> , 2016, 175, 639-641.	1.4	4
25	Replication of a distinct psoriatic arthritis risk variant at the IL23R locus. <i>Annals of the Rheumatic Diseases</i> , 2016, 75, 1417-1418.	0.5	9
26	OPO128â€¦PTPN22 is Associated with Susceptibility to Psoriatic Arthritis but not Psoriasis: Evidence for a Further PSA-Specific Risk Locus. <i>Annals of the Rheumatic Diseases</i> , 2015, 74, 116.3-117.	0.5	1
27	O53.â€¦PTPN22 is Associated with Susceptibility to Psoriatic Arthritis but not Psoriasis: Evidence for a Further PSA-Specific Risk Locus. <i>Rheumatology</i> , 2015, , .	0.9	1
28	Palmoplantar Pustular Psoriasis Is Associated with Missense Variants in CARD14 , but Not with Loss-of-Function Mutations in IL36RN in European Patients. <i>Journal of Investigative Dermatology</i> , 2015, 135, 2538-2541.	0.3	78
29	PTPN22 is associated with susceptibility to psoriatic arthritis but not psoriasis: evidence for a further PsA-specific risk locus. <i>Annals of the Rheumatic Diseases</i> , 2015, 74, 1882-1885.	0.5	64
30	Dense genotyping of immune-related susceptibility loci reveals new insights into the genetics of psoriatic arthritis. <i>Nature Communications</i> , 2015, 6, 6046.	5.8	149
31	Complex Role of TNF Variants in Psoriatic Arthritis and Treatment Response to Anti-TNF Therapy: Evidence and Concepts. <i>Journal of Investigative Dermatology</i> , 2014, 134, 2483-2485.	0.3	10
32	Successful therapy with anakinra in a patient with generalized pustular psoriasis carrying <i>IL36RN</i> mutations. <i>British Journal of Dermatology</i> , 2014, 170, 202-204.	1.4	95
33	Mutations in IL36RN in Patients with Generalized Pustular Psoriasis. <i>Journal of Investigative Dermatology</i> , 2013, 133, 2634-2637.	0.3	89
34	Variants in <i>RUNX3</i> Contribute to Susceptibility to Psoriatic Arthritis, Exhibiting Further Common Ground With Ankylosing Spondylitis. <i>Arthritis and Rheumatism</i> , 2013, 65, 1224-1231.	6.7	63
35	Association of Î²-Defensin Copy Number and Psoriasis in Three Cohorts of European Origin. <i>Journal of Investigative Dermatology</i> , 2012, 132, 2407-2413.	0.3	50
36	Conditional analysis identifies three novel major histocompatibility complex loci associated with psoriasis. <i>Human Molecular Genetics</i> , 2012, 21, 5185-5192.	1.4	58

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37	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. <i>Nature Genetics</i> , 2012, 44, 1341-1348.	9.4	848
38	Identification of low frequency TRAF3IP2 coding variants in psoriatic arthritis patients and functional characterization. <i>Arthritis Research and Therapy</i> , 2012, 14, R84.	1.6	16
39	Deletion of LCE3C and LCE3B is a susceptibility factor for psoriatic arthritis: A study in Spanish and Italian populations and meta-analysis. <i>Arthritis and Rheumatism</i> , 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. <i>Arthritis and Rheumatism</i> , 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. <i>Journal of Investigative Dermatology</i> , 2011, 131, 1105-1109.	0.3	89
42	Common variants at TRAF3IP2 are associated with susceptibility to psoriatic arthritis and psoriasis. <i>Nature Genetics</i> , 2010, 42, 996-999.	9.4	334
43	A genome-wide association study identifies new psoriasis susceptibility loci and an interaction between HLA-C and ERAP1. <i>Nature Genetics</i> , 2010, 42, 985-990.	9.4	918
44	Replication of LCE3C/LCE3B CNV as a Risk Factor for Psoriasis and Analysis of Interaction with Other Genetic Risk Factors. <i>Journal of Investigative Dermatology</i> , 2010, 130, 979-984.	0.3	61
45	Deletion of LCE3C and LCE3B genes at PSORS4 does not contribute to susceptibility to psoriatic arthritis in German patients. <i>Annals of the Rheumatic Diseases</i> , 2010, 69, 876-878.	0.5	34
46	Genotyping Microarray for CSNB-Associated Genes. , 2009, 50, 5919.		41
47	Neue Entwicklungen in der Psoriasisgenetik. <i>Medizinische Genetik</i> , 2009, 21, 498-504.	0.1	0
48	Ichthyosis vulgaris: novel FLG mutations in the German population and high presence of CD1a+ cells in the epidermis of the atopic subgroup. <i>British Journal of Dermatology</i> , 2009, 160, 771-781.	1.4	40
49	Genetic Variants of the IL-23R Pathway: Association with Psoriatic Arthritis and Psoriasis Vulgaris, but No Specific Risk Factor for Arthritis. <i>Journal of Investigative Dermatology</i> , 2009, 129, 355-358.	0.3	97
50	New Mutations of EXT1 and EXT2 Genes in German Patients with Multiple Osteochondromas. <i>Annals of Human Genetics</i> , 2009, 73, 283-291.	0.3	31
51	Characterisation of psoriasis susceptibility locus 6 (PSORS6) in patients with early onset psoriasis and evidence for interaction with PSORS1. <i>Journal of Medical Genetics</i> , 2009, 46, 736-744.	1.5	34
52	Psoriasis is associated with increased β -defensin genomic copy number. <i>Nature Genetics</i> , 2008, 40, 23-25.	9.4	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. <i>Annals of the Rheumatic Diseases</i> , 2008, 67, 131-133.	0.5	21
54	Identification of ZNF313 / RNF114 as a novel psoriasis susceptibility gene. <i>Human Molecular Genetics</i> , 2008, 17, 1938-1945.	1.4	176

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55	Molecular karyotyping in patients with mental retardation using 100K single-nucleotide polymorphism arrays. <i>Journal of Medical Genetics</i> , 2007, 44, 629-636.	1.5	72
56	Severe skeletal dysplasia caused by undiagnosed hypothyroidism. <i>European Journal of Medical Genetics</i> , 2007, 50, 209-215.	0.7	30
57	Tumor necrosis factor polymorphisms in psoriatic arthritis: association with the promoter polymorphism TNF-857 independent of the PSORS1 risk allele. <i>Arthritis Research and Therapy</i> , 2007, 9, P18.	1.6	2
58	TNF polymorphisms in psoriasis: Association of psoriatic arthritis with the promoter polymorphism TNF*-857 independent of the PSORS1 risk allele. <i>Arthritis and Rheumatism</i> , 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. <i>Journal of Investigative Dermatology</i> , 2007, 127, 1367-1370.	0.3	39
60	Genetik der Psoriasis. <i>Medizinische Genetik</i> , 2007, 19, 350-355.	0.1	2
61	Male Restricted Genetic Association of Variant R620W in PTPN22 with Psoriatic Arthritis. <i>Journal of Investigative Dermatology</i> , 2006, 126, 936-938.	0.3	28
62	Association between protein tyrosine phosphatase 22 variant R620W in conjunction with the HLA*DRB1 shared epitope and humoral autoimmunity to an immunodominant epitope of cartilage-specific type II collagen in early rheumatoid arthritis. <i>Arthritis and Rheumatism</i> , 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. <i>Arthritis and Rheumatism</i> , 2006, 54, 4023-4024.	6.7	4
64	Diagnostic yield of various genetic approaches in patients with unexplained developmental delay or mental retardation. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 2063-2074.	0.7	343
65	A variable combination of features of Noonan syndrome and neurofibromatosis type I are caused by mutations in the NF1 gene. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 2749-2756.	0.7	46
66	Evidence for susceptibility determinant(s) to psoriasis vulgaris in or near PTPN22 in German patients. <i>Journal of Medical Genetics</i> , 2006, 43, 517-522.	1.5	31
67	Lack of Evidence for Genetic Association to RUNX1 Binding Site at PSORS2 in Different German Psoriasis Cohorts. <i>Journal of Investigative Dermatology</i> , 2005, 124, 107-110.	0.3	22
68	Fine Mapping of the Psoriasis Susceptibility Gene PSORS1: A Reassessment of Risk Associated with a Putative Risk Haplotype Lacking HLA-Cw6. <i>Journal of Investigative Dermatology</i> , 2005, 124, 921-930.	0.3	18
69	Systematic Linkage Disequilibrium Analysis of SLC12A8 at PSORS5 Confirms a Role in Susceptibility to Psoriasis Vulgaris. <i>Journal of Investigative Dermatology</i> , 2005, 125, 906-912.	0.3	38
70	Novel autosomal recessive progressive hyperpigmentation syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2005, 135A, 195-199.	0.7	2
71	Systematic assessment of atypical deletions reveals genotype-phenotype correlation in 22q11.2. <i>Journal of Medical Genetics</i> , 2005, 42, 871-876.	1.5	118
72	Lack of genetic association of the three more common polymorphisms of CARD15 with psoriatic arthritis and psoriasis in a German cohort. <i>Annals of the Rheumatic Diseases</i> , 2005, 64, 951-954.	0.5	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. <i>Arthritis and Rheumatism</i> , 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. <i>Experimental Dermatology</i> , 2003, 12, 490-496.	1.4	26
75	Interleukin-10 promoter polymorphism IL10.G and familial early onset psoriasis. <i>British Journal of Dermatology</i> , 2003, 149, 381-385.	1.4	25
76	Cytokine balance in infants undergoing cardiac operation. <i>Annals of Thoracic Surgery</i> , 2002, 73, 601-608.	0.7	66