Joshua D Milner

List of Publications by Year
in descending order

106	12,303	34076 52	29127 104
papers	citations	h-index	g-index

129	129	129	19164
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Assessing the safety of bioactive ingredients in infant formula that affect the immune system: recommendations from an expert panel. American Journal of Clinical Nutrition, 2022, 115, 570-587.	2.2	3
2	Studying severe long COVID to understand post-infectious disorders beyond COVID-19. Nature Medicine, 2022, 28, 879-882.	15.2	72
3	Hereditary alpha-tryptasemia modifies clinical phenotypes among individuals with congenital hypermobility disorders. Human Genetics and Genomics Advances, 2022, 3, 100094.	1.0	1
4	Ex vivo effect of JAK inhibition on JAK-STAT1 pathway hyperactivation in patients with dominant-negative STAT3 mutations. Journal of Clinical Immunology, 2022, 42, 1193-1204.	2.0	8
5	Atopy as Immune Dysregulation: Offender Genes and Targets. Journal of Allergy and Clinical Immunology: in Practice, 2022, 10, 1737-1756.	2.0	15
6	Distinct antibody responses to SARS-CoV-2 in children and adults across the COVID-19 clinical spectrum. Nature Immunology, 2021, 22, 25-31.	7.0	403
7	Heritable risk for severe anaphylaxis associated with increased Î±-tryptaseâ€“encoding germline copy number at TPSAB1. Journal of Allergy and Clinical Immunology, 2021, 147, 622-632.	1.5	137
8	JAK inhibition in early-onset somatic, nonclonal STAT5B gain-of-function disease. Journal of Allergy and Clinical Immunology: in Practice, 2021, 9, 1008-1010.e2.	2.0	16
9	Recurrent lymphadenitis in a female XIAP/BIRC4 mutation carrier with normal lyonization. Journal of Allergy and Clinical Immunology: in Practice, 2021, 9, 1002-1005.e2.	2.0	1
10	Systematic evaluation of nine monogenic autoinflammatory diseases reveals common and disease-specific correlations with allergy-associated features. Annals of the Rheumatic Diseases, 2021, 80, 788-795.	0.5	12
11	Epicutaneous Staphylococcus aureus induces IL-36 to enhance IgE production and ensuing allergic disease. Journal of Clinical Investigation, 2021, 131, .	3.9	39
12	Inborn Error of Immunity or Atopic Dermatitis: When to be Concerned and How to Investigate. Journal of Allergy and Clinical Immunology: in Practice, 2021, 9, 1501-1507.	2.0	13
13	SARS-CoV-2â€“related MIS-C: A key to the viral and genetic causes of Kawasaki disease?. Journal of Experimental Medicine, 2021, 218, .	4.2	100
14	Potential mechanisms of anaphylaxis to COVID-19 mRNA vaccines. Journal of Allergy and Clinical Immunology, 2021, 147, 2075-2082.e2.	1.5	117
15	Clinical relevance of inherited genetic differences in human tryptases. Annals of Allergy, Asthma and Immunology, 2021, 127, 638-647.	0.5	30
16	Small intestinal immunopathology and GI-associated antibody formation in hereditary alpha-tryptasemia. Journal of Allergy and Clinical Immunology, 2021, 148, 813-821.e7.	1.5	17
17	Inherited SLP76 deficiency in humans causes severe combined immunodeficiency, neutrophil and platelet defects. Journal of Experimental Medicine, 2021, 218, .	4.2	20
18	Multisystem Inflammatory Syndrome in Children Associated With Coronavirus Disease 2019 in a Childrenâ€™s Hospital in New York City: Patient Characteristics and an Institutional Protocol for Evaluation, Management, and Follow-Up. Pediatric Critical Care Medicine, 2021, 22, e178-e191.	0.2	98

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19	STAT3 Gain-of-Function Mutations Underlie Deficiency in Human Nonclassical CD16+ Monocytes and CD141+ Dendritic Cells. Journal of Immunology, 2021, 207, 2423-2432.	0.4	11
20	A Novel Germline Heterozygous BCL11B Variant Causing Severe Atopic Disease and Immune Dysregulation. Frontiers in Immunology, 2021, 12, 788278.	2.2	9
21	Compound Heterozygous PGM3 Mutations in a Thai Patient with a Specific Antibody Deficiency Requiring Monthly IVIG Infusions. Journal of Clinical Immunology, 2020, 40, 227-231.	2.0	4
22	PLAID syndrome: Characteristic presentation and a novel therapeutic option. Pediatric Dermatology, 2020, 37, 147-149.	0.5	9
23	Asthma among hospitalized patients with COVID-19 and related outcomes. Journal of Allergy and Clinical Immunology, 2020, 146, 1027-1034.e4.	1.5	115
24	Dominant-negative mutations in human IL6ST underlie hyper-IgE syndrome. Journal of Experimental Medicine, 2020, 217, .	4.2	64
25	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,749
26	The Child with Elevated IgE and Infection Susceptibility. Current Allergy and Asthma Reports, 2020, 20, 65.	2.4	11
27	Multiplexed Functional Assessment of Genetic Variants in CARD11. American Journal of Human Genetics, 2020, 107, 1029-1043.	2.6	38
28	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. Cell, 2020, 181, 1194-1199.	13.5	185
29	Multisystem Inflammatory Syndrome Related to COVID-19 in Previously Healthy Children and Adolescents in New York City. JAMA - Journal of the American Medical Association, 2020, 324, 294.	3.8	479
30	Primary Atopic Disorders. Annual Review of Immunology, 2020, 38, 785-808.	9.5	40
31	Primary immune deficiencies associated with a Th2 diathesis. , 2020, , 393-410.		1
32	Impaired angiogenesis and extracellular matrix metabolism in autosomal-dominant hyper-IgE syndrome. Journal of Clinical Investigation, 2020, 130, 4167-4181.	3.9	13
33	Case Report: Secondary Hemophagocytic Lymphohistiocytosis With Disseminated Infection in Chronic Granulomatous Diseaseâ€"A Serious Cause of Mortality. Frontiers in Immunology, 2020, 11, 581475.	2.2	6
34	Human TH9 differentiation is dependent on signal transducer and activator of transcription (STAT) 3 to restrain STAT1-mediated inhibition. Journal of Allergy and Clinical Immunology, 2019, 143, 1108-1118.e4.	1.5	10
35	Geneâ€"environment interactions in primary atopic disorders. Current Opinion in Immunology, 2019, 60, 148-155.	2.4	11
36	Impact of naturally forming human Î±/Î²-tryptase heterotetramers in the pathogenesis of hereditary Î±-tryptasemia. Journal of Experimental Medicine, 2019, 216, 2348-2361.	4.2	85

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37	Editorial overview: Collusion between genes and environment in the pathogenesis of allergic disease. Current Opinion in Immunology, 2019, 60, iii-v.	2.4	1
38	Loss of the interleukin-6 receptor causes immunodeficiency, atopy, and abnormal inflammatory responses. Journal of Experimental Medicine, 2019, 216, 1986-1998.	4.2	153
39	The clinical and mechanistic intersection of primary atopic disorders and inborn errors of growth and metabolism. Immunological Reviews, 2019, 287, 135-144.	2.8	12
40	Loss-of-function mutations in caspase recruitment domain-containing protein 14 (CARD14) are associated with a severe variant of atopic dermatitis. Journal of Allergy and Clinical Immunology, 2019, 143, 173-181.e10.	1.5	60
41	Hypomorphic caspase activation and recruitment domain 11 (CARD11) mutations associated with diverse immunologic phenotypes with or without atopic disease. Journal of Allergy and Clinical Immunology, 2019, 143, 1482-1495.	1.5	116
42	Primary atopic disorders. Journal of Experimental Medicine, 2018, 215, 1009-1022.	4.2	74
43	A common haplotype containing functional CACNA1H variants is frequently coinherited with increased TPSAB1 copy number. Genetics in Medicine, 2018, 20, 503-512.	1.1	25
44	The overlap between allergy and immunodeficiency. Current Opinion in Pediatrics, 2018, 30, 848-854.	1.0	16
45	The CBM-opathiesâ€"A Rapidly Expanding Spectrum of Human Inborn Errors of Immunity Caused by Mutations in the CARD11-BCL10-MALT1 Complex. Frontiers in Immunology, 2018, 9, 2078.	2.2	92
46	TCR Signaling Abnormalities in Human Th2-Associated Atopic Disease. Frontiers in Immunology, 2018, 9, 719.	2.2	7
47	First Identification of an Inherited TPSAB1 Quintuplication in a Patient with Clonal Mast Cell Disease. Journal of Clinical Immunology, 2018, 38, 457-459.	2.0	58
48	Functional characterization of phospholipase C-Î³2 mutant protein causing both somatic ibrutinib resistance and a germline monogenic autoinflammatory disorder. Oncotarget, 2018, 9, 34357-34378.	0.8	14
49	Learning from Job: A Rare Genetic Disease and Lessons of Biblical Proportions. Rambam Maimonides Medical Journal, 2018, 9, e0006.	0.4	2
50	Detection of phosphoglucomutase-3 deficiency by lectin-based flow cytometry. Journal of Allergy and Clinical Immunology, 2017, 140, 291-294.e4.	1.5	10
51	ERBIN deficiency links STAT3 and TGF-Î² pathway defects with atopy in humans. Journal of Experimental Medicine, 2017, 214, 669-680.	4.2	70
52	Germline hypomorphic CARD11 mutations in severe atopic disease. Nature Genetics, 2017, 49, 1192-1201.	9.4	174
53	Somatic STAT5b gain-of-function mutations in early onset nonclonal eosinophilia, urticaria, dermatitis, and diarrhea. Blood, 2017, 129, 650-653.	0.6	74
54	PD-L1 up-regulation restrains Th17 cell differentiation in STAT3 loss- and STAT1 gain-of-function patients. Journal of Experimental Medicine, 2017, 214, 2523-2533.	4.2	55

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55	A biallelic mutation in IL6ST encoding the GP130 co-receptor causes immunodeficiency and craniosynostosis. Journal of Experimental Medicine, 2017, 214, 2547-2562.	4.2	158
56	Low IgE Is Insufficiently Sensitive to Guide Genetic Testing of STAT3 Gain-of-Function Mutations. Clinical Chemistry, 2017, 63, 1539-1540.	1.5	4
57	Atopic Dermatitis and Allergic Urticaria. Immunology and Allergy Clinics of North America, 2017, 37, 1-10.	0.7	13
58	Heterozygous STAT1 gain-of-function mutations underlie an unexpectedly broad clinical phenotype. Blood, 2016, 127, 3154-3164.	0.6	465
59	Biallelic hypomorphic mutations in a linear deubiquitinase define otulipenia, an early-onset autoinflammatory disease. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 10127-10132.	3.3	206
60	Elevated basal serum tryptase identifies a multisystem disorder associated with increased TPSAB1 copy number. Nature Genetics, 2016, 48, 1564-1569.	9.4	279
61	Loss-of-function mutations in TNFAIP3 leading to A20 haploinsufficiency cause an early-onset autoinflammatory disease. Nature Genetics, 2016, 48, 67-73.	9.4	513
62	Hematopoietic prostaglandin D synthase defines a proeosinophilic pathogenic effector human TH2 cell subpopulation with enhanced function. Journal of Allergy and Clinical Immunology, 2016, 137, 907-918.e9.	1.5	139
63	Diminution of signal transducer and activator of transcription 3 signaling inhibits vascular permeability and anaphylaxis. Journal of Allergy and Clinical Immunology, 2016, 138, 187-199.	1.5	56
64	FOXP3+ Tregs require WASP to restrain Th2-mediated food allergy. Journal of Clinical Investigation, 2016, 126, 4030-4044.	3.9	53
65	Early-onset lymphoproliferation and autoimmunity caused by germline STAT3 gain-of-function mutations. Blood, 2015, 125, 591-599.	0.6	436
66	Glycans Instructing Immunity: The Emerging Role of Altered Glycosylation in Clinical Immunology. Frontiers in Pediatrics, 2015, 3, 54.	0.9	56
67	Long-Acting Beta Agonists Enhance Allergic Airway Disease. PLoS ONE, 2015, 10, e0142212.	1.1	13
68	Distinct Cutaneous Manifestations and Cold-Induced Leukocyte Activation Associated WithPLCG2Mutations. JAMA Dermatology, 2015, 151, 627.	2.0	55
69	PLAID: a Syndrome of Complex Patterns of Disease and Unique Phenotypes. Journal of Clinical Immunology, 2015, 35, 527-530.	2.0	76
70	Eosinophilia Associated with Disorders of Immune Deficiency or Immune Dysregulation. Immunology and Allergy Clinics of North America, 2015, 35, 523-544.	0.7	48
71	Genetics of allergy and allergic sensitization: common variants, rare mutations. Current Opinion in Immunology, 2015, 36, 115-126.	2.4	56
72	The Ying and Yang of STAT3 in Human Disease. Journal of Clinical Immunology, 2015, 35, 615-623.	2.0	130

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73	Atopic Dermatitis in Children. Immunology and Allergy Clinics of North America, 2015, 35, 161-183.	0.7	173
74	A mouse model of HIES reveals pro- and anti-inflammatory functions of STAT3. Blood, 2014, 123, 2978-2987.	0.6	71
75	The autoinhibitory C-terminal SH2 domain of phospholipase Câ€“Î³2 stabilizes B cell receptor signalosome assembly. Science Signaling, 2014, 7, ra89.	1.6	32
76	Human IL-21 and IL-21R deficiencies. Current Opinion in Pediatrics, 2014, 26, 704-712.	1.0	63
77	Autosomal recessive phosphoglucomutase 3 (PGM3) mutations link glycosylation defects to atopy, immune deficiency, autoimmunity, and neurocognitive impairment. Journal of Allergy and Clinical Immunology, 2014, 133, 1400-1409.e5.	1.5	193
78	Human syndromes of immunodeficiency and dysregulation are characterized by distinct defects in T-cell receptor repertoire development. Journal of Allergy and Clinical Immunology, 2014, 133, 1109-1115.e14.	1.5	62
79	Mendelian inheritance of elevated serum tryptase associated with atopy and connective tissue abnormalities. Journal of Allergy and Clinical Immunology, 2014, 133, 1471-1474.	1.5	110
80	The cup runneth over: lessons from the ever-expanding pool of primary immunodeficiency diseases. Nature Reviews Immunology, 2013, 13, 635-648.	10.6	91
81	Elevated IgE and atopy in patients treated for early-onset ADA-SCID. Journal of Allergy and Clinical Immunology, 2013, 132, 1444-1446.e5.	1.5	22
82	Diminished allergic disease in patients with STAT3 mutations reveals a role for STAT3 signaling in mast cellÂdegranulation. Journal of Allergy and Clinical Immunology, 2013, 132, 1388-1396.e3.	1.5	102
83	Dominant gain-of-function STAT1 mutations in FOXP3 wild-type immune dysregulationâ€“polyendocrinopathyâ€“enteropathyâ€“X-linkedâ€“like syndrome. Journal of Allergy and Clinical Immunology, 2013, 131, 1611-1623.e3.	1.5	288
84	Gain-of-function STAT1 mutations are associated with PD-L1 overexpression and a defect in B-cell survival. Journal of Allergy and Clinical Immunology, 2013, 131, 1691-1693.	1.5	82
85	Signal transducer and activator of transcription 1 (STAT1) gain-of-function mutations and disseminated coccidioidomycosis and histoplasmosis. Journal of Allergy and Clinical Immunology, 2013, 131, 1624-1634.e17.	1.5	222
86	Loss-of-function mutations in the IL-21 receptor gene cause a primary immunodeficiency syndrome. Journal of Experimental Medicine, 2013, 210, 433-443.	4.2	186
87	Mechanisms underlying helper T-cell plasticity: Implications for immune-mediated disease. Journal of Allergy and Clinical Immunology, 2013, 131, 1276-1287.	1.5	138
88	Pathogen-Sensing, Regulatory T Cells, and Responsiveness-Tuning Collectively Regulate Foreign- and Self-Antigen Mediated T-Cell Responses. Cold Spring Harbor Symposia on Quantitative Biology, 2013, 78, 265-276.	2.0	19
89	Peptide library-based evaluation of T-cell receptor breadth detects defects in global and regulatory activation in human immunologic diseases. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 8164-8169.	3.3	5
90	Cold Urticaria, Immunodeficiency, and Autoimmunity Related toPLCG2Deletions. New England Journal of Medicine, 2012, 366, 330-338.	13.9	391

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91	A Hypermorphic Missense Mutation in PLCG2 , Encoding Phospholipase Cî³2, Causes a Dominantly Inherited Autoinflammatory Disease with Immunodeficiency. American Journal of Human Genetics, 2012, 91, 713-720.	2.6	327
92	Altered T-cell receptor signaling in the pathogenesis of allergic disease. Journal of Allergy and Clinical Immunology, 2011, 127, 351-354.	1.5	15
93	IL-17 producing cells in host defense and atopy. Current Opinion in Immunology, 2011, 23, 784-788.	2.4	38
94	A Critical Role for STAT3 Transcription Factor Signaling in the Development and Maintenance of Human T Cell Memory. Immunity, 2011, 35, 806-818.	6.6	241
95	Hypomorphic Rag mutations can cause destructive midline granulomatous disease. Blood, 2010, 116, 1263-1271.	0.6	110
96	Sustained IL-4 exposure leads to a novel pathway for hemophagocytosis, inflammation, and tissue macrophage accumulation. Blood, 2010, 116, 2476-2483.	0.6	100
97	Cutting Edge: Lack of High Affinity Competition for Peptide in Polyclonal CD4+ Responses Unmasks IL-4 Production. Journal of Immunology, 2010, 184, 6569-6573.	0.4	46
98	Highly Variable Clinical Phenotypes of Hypomorphic RAG1 Mutations. Pediatrics, 2010, 126, e1248-e1252.	1.0	70
99	Autoimmunity in Severe Combined Immunodeficiency (SCID): Lessons from Patients and Experimental Models. Journal of Clinical Immunology, 2008, 28, 29-33.	2.0	37
100	Impaired TH17 cell differentiation in subjects with autosomal dominant hyper-IgE syndrome. Nature, 2008, 452, 773-776.	13.7	1,046
101	Exogenous vitamin D might contribute to geographic variations in epinephrine prescriptions. Journal of Allergy and Clinical Immunology, 2008, 121, 265-266.	1.5	5
102	Lymphopenic mice reconstituted with limited repertoire T cells develop severe, multiorgan, Th2-associated inflammatory disease. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 576-581.	3.3	94
103	Repertoire-dependent immunopathology. Journal of Autoimmunity, 2007, 29, 257-261.	3.0	44
104	Transient environmental exposures on the developing immune system: implications for allergy and asthma. Current Opinion in Allergy and Clinical Immunology, 2005, 5, 235-240.	1.1	5
105	Early Infant Multivitamin Supplementation Is Associated With Increased Risk for Food Allergy and Asthma. Pediatrics, 2004, 114, 27-32.	1.0	145
106	Does Elevated Peak Bilirubin Protect from Retinopathy of Prematurity in Very Low Birthweight Infants. Journal of Perinatology, 2003, 23, 208-211.	0.9	19