

Joshua D Milner

List of Publications by Year in descending order

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Version: 2024-02-01

106
papers

12,303
citations

34076

52
h-index

29127

104
g-index

129
all docs

129
docs citations

129
times ranked

19164
citing authors

#	ARTICLE	IF	CITATIONS
1	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. <i>Science</i> , 2020, 370, .	6.0	1,749
2	Impaired TH17 cell differentiation in subjects with autosomal dominant hyper-IgE syndrome. <i>Nature</i> , 2008, 452, 773-776.	13.7	1,046
3	Loss-of-function mutations in TNFAIP3 leading to A20 haploinsufficiency cause an early-onset autoinflammatory disease. <i>Nature Genetics</i> , 2016, 48, 67-73.	9.4	513
4	Multisystem Inflammatory Syndrome Related to COVID-19 in Previously Healthy Children and Adolescents in New York City. <i>JAMA - Journal of the American Medical Association</i> , 2020, 324, 294.	3.8	479
5	Heterozygous STAT1 gain-of-function mutations underlie an unexpectedly broad clinical phenotype. <i>Blood</i> , 2016, 127, 3154-3164.	0.6	465
6	Early-onset lymphoproliferation and autoimmunity caused by germline STAT3 gain-of-function mutations. <i>Blood</i> , 2015, 125, 591-599.	0.6	436
7	Distinct antibody responses to SARS-CoV-2 in children and adults across the COVID-19 clinical spectrum. <i>Nature Immunology</i> , 2021, 22, 25-31.	7.0	403
8	Cold Urticaria, Immunodeficiency, and Autoimmunity Related to <i>PLCG2</i> Deletions. <i>New England Journal of Medicine</i> , 2012, 366, 330-338.	13.9	391
9	A Hypermorphic Missense Mutation in <i>PLCG2</i> , Encoding Phospholipase C ^β 2, Causes a Dominantly Inherited Autoinflammatory Disease with Immunodeficiency. <i>American Journal of Human Genetics</i> , 2012, 91, 713-720.	2.6	327
10	Dominant gain-of-function STAT1 mutations in <i>FOXP3</i> wild-type immune dysregulationâ€“polyendocrinopathyâ€“enteropathyâ€“X-linkedâ€“like syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 131, 1611-1623.e3.	1.5	288
11	Elevated basal serum tryptase identifies a multisystem disorder associated with increased <i>TPSAB1</i> copy number. <i>Nature Genetics</i> , 2016, 48, 1564-1569.	9.4	279
12	A Critical Role for STAT3 Transcription Factor Signaling in the Development and Maintenance of Human T Cell Memory. <i>Immunity</i> , 2011, 35, 806-818.	6.6	241
13	Signal transducer and activator of transcription 1 (STAT1) gain-of-function mutations and disseminated coccidioidomycosis and histoplasmosis. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 131, 1624-1634.e17.	1.5	222
14	Biallelic hypomorphic mutations in a linear deubiquitinase define otulipenia, an early-onset autoinflammatory disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, 10127-10132.	3.3	206
15	Autosomal recessive phosphoglucomutase 3 (PGM3) mutations link glycosylation defects to atopy, immune deficiency, autoimmunity, and neurocognitive impairment. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 133, 1400-1409.e5.	1.5	193
16	Loss-of-function mutations in the IL-21 receptor gene cause a primary immunodeficiency syndrome. <i>Journal of Experimental Medicine</i> , 2013, 210, 433-443.	4.2	186
17	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. <i>Cell</i> , 2020, 181, 1194-1199.	13.5	185
18	Germline hypomorphic <i>CARD11</i> mutations in severe atopic disease. <i>Nature Genetics</i> , 2017, 49, 1192-1201.	9.4	174

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19	Atopic Dermatitis in Children. <i>Immunology and Allergy Clinics of North America</i> , 2015, 35, 161-183.	0.7	173
20	A biallelic mutation in <i>IL6ST</i> encoding the GP130 co-receptor causes immunodeficiency and craniosynostosis. <i>Journal of Experimental Medicine</i> , 2017, 214, 2547-2562.	4.2	158
21	Loss of the interleukin-6 receptor causes immunodeficiency, atopy, and abnormal inflammatory responses. <i>Journal of Experimental Medicine</i> , 2019, 216, 1986-1998.	4.2	153
22	Early Infant Multivitamin Supplementation Is Associated With Increased Risk for Food Allergy and Asthma. <i>Pediatrics</i> , 2004, 114, 27-32.	1.0	145
23	Hematopoietic prostaglandin D synthase defines a proeosinophilic pathogenic effector human TH2 cell subpopulation with enhanced function. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 137, 907-918.e9.	1.5	139
24	Mechanisms underlying helper T-cell plasticity: Implications for immune-mediated disease. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 131, 1276-1287.	1.5	138
25	Heritable risk for severe anaphylaxis associated with increased <i>±</i> -tryptase encoding germline copy number at <i>TPSAB1</i> . <i>Journal of Allergy and Clinical Immunology</i> , 2021, 147, 622-632.	1.5	137
26	The Ying and Yang of STAT3 in Human Disease. <i>Journal of Clinical Immunology</i> , 2015, 35, 615-623.	2.0	130
27	Potential mechanisms of anaphylaxis to COVID-19 mRNA vaccines. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 147, 2075-2082.e2.	1.5	117
28	Hypomorphic caspase activation and recruitment domain 11 (<i>CARD11</i>) mutations associated with diverse immunologic phenotypes with or without atopic disease. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 1482-1495.	1.5	116
29	Asthma among hospitalized patients with COVID-19 and related outcomes. <i>Journal of Allergy and Clinical Immunology</i> , 2020, 146, 1027-1034.e4.	1.5	115
30	Hypomorphic Rag mutations can cause destructive midline granulomatous disease. <i>Blood</i> , 2010, 116, 1263-1271.	0.6	110
31	Mendelian inheritance of elevated serum tryptase associated with atopy and connective tissue abnormalities. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 133, 1471-1474.	1.5	110
32	Diminished allergic disease in patients with STAT3 mutations reveals a role for STAT3 signaling in mast cell degranulation. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 132, 1388-1396.e3.	1.5	102
33	Sustained IL-4 exposure leads to a novel pathway for hemophagocytosis, inflammation, and tissue macrophage accumulation. <i>Blood</i> , 2010, 116, 2476-2483.	0.6	100
34	SARS-CoV-2 related MIS-C: A key to the viral and genetic causes of Kawasaki disease?. <i>Journal of Experimental Medicine</i> , 2021, 218, .	4.2	100
35	Multisystem Inflammatory Syndrome in Children Associated With Coronavirus Disease 2019 in a Children's Hospital in New York City: Patient Characteristics and an Institutional Protocol for Evaluation, Management, and Follow-Up. <i>Pediatric Critical Care Medicine</i> , 2021, 22, e178-e191.	0.2	98
36	Lymphopenic mice reconstituted with limited repertoire T cells develop severe, multiorgan, Th2-associated inflammatory disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2007, 104, 576-581.	3.3	94

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37	The CBM-opathies – A Rapidly Expanding Spectrum of Human Inborn Errors of Immunity Caused by Mutations in the CARD11-BCL10-MALT1 Complex. <i>Frontiers in Immunology</i> , 2018, 9, 2078.	2.2	92
38	The cup runneth over: lessons from the ever-expanding pool of primary immunodeficiency diseases. <i>Nature Reviews Immunology</i> , 2013, 13, 635-648.	10.6	91
39	Impact of naturally forming human $\hat{\iota}/\hat{\iota}^2$ -tryptase heterotetramers in the pathogenesis of hereditary $\hat{\iota}$ -tryptasemia. <i>Journal of Experimental Medicine</i> , 2019, 216, 2348-2361.	4.2	85
40	Gain-of-function STAT1 mutations are associated with PD-L1 overexpression and a defect in B-cell survival. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 131, 1691-1693.	1.5	82
41	PLAID: a Syndrome of Complex Patterns of Disease and Unique Phenotypes. <i>Journal of Clinical Immunology</i> , 2015, 35, 527-530.	2.0	76
42	Somatic STAT5b gain-of-function mutations in early onset nonclonal eosinophilia, urticaria, dermatitis, and diarrhea. <i>Blood</i> , 2017, 129, 650-653.	0.6	74
43	Primary atopic disorders. <i>Journal of Experimental Medicine</i> , 2018, 215, 1009-1022.	4.2	74
44	Studying severe long COVID to understand post-infectious disorders beyond COVID-19. <i>Nature Medicine</i> , 2022, 28, 879-882.	15.2	72
45	A mouse model of HIES reveals pro- and anti-inflammatory functions of STAT3. <i>Blood</i> , 2014, 123, 2978-2987.	0.6	71
46	Highly Variable Clinical Phenotypes of Hypomorphic <i>RAG1</i> Mutations. <i>Pediatrics</i> , 2010, 126, e1248-e1252.	1.0	70
47	ERBIN deficiency links STAT3 and TGF- $\hat{\iota}^2$ pathway defects with atopy in humans. <i>Journal of Experimental Medicine</i> , 2017, 214, 669-680.	4.2	70
48	Dominant-negative mutations in human <i>IL6ST</i> underlie hyper-IgE syndrome. <i>Journal of Experimental Medicine</i> , 2020, 217, .	4.2	64
49	Human IL-21 and IL-21R deficiencies. <i>Current Opinion in Pediatrics</i> , 2014, 26, 704-712.	1.0	63
50	Human syndromes of immunodeficiency and dysregulation are characterized by distinct defects in T-cell receptor repertoire development. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 133, 1109-1115.e14.	1.5	62
51	Loss-of-function mutations in caspase recruitment domain-containing protein 14 (CARD14) are associated with a severe variant of atopic dermatitis. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 173-181.e10.	1.5	60
52	First Identification of an Inherited TPSAB1 Quintuplication in a Patient with Clonal Mast Cell Disease. <i>Journal of Clinical Immunology</i> , 2018, 38, 457-459.	2.0	58
53	Glycans Instructing Immunity: The Emerging Role of Altered Glycosylation in Clinical Immunology. <i>Frontiers in Pediatrics</i> , 2015, 3, 54.	0.9	56
54	Genetics of allergy and allergic sensitization: common variants, rare mutations. <i>Current Opinion in Immunology</i> , 2015, 36, 115-126.	2.4	56

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55	Diminution of signal transducer and activator of transcription 3 signaling inhibits vascular permeability and anaphylaxis. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 187-199.	1.5	56
56	Distinct Cutaneous Manifestations and Cold-Induced Leukocyte Activation Associated With <i>PLCG2</i> Mutations. <i>JAMA Dermatology</i> , 2015, 151, 627.	2.0	55
57	PD-L1 up-regulation restrains Th17 cell differentiation in <i>STAT3</i> loss- and <i>STAT1</i> gain-of-function patients. <i>Journal of Experimental Medicine</i> , 2017, 214, 2523-2533.	4.2	55
58	FOXP3+ Tregs require WASP to restrain Th2-mediated food allergy. <i>Journal of Clinical Investigation</i> , 2016, 126, 4030-4044.	3.9	53
59	Eosinophilia Associated with Disorders of Immune Deficiency or Immune Dysregulation. <i>Immunology and Allergy Clinics of North America</i> , 2015, 35, 523-544.	0.7	48
60	Cutting Edge: Lack of High Affinity Competition for Peptide in Polyclonal CD4+ Responses Unmasks IL-4 Production. <i>Journal of Immunology</i> , 2010, 184, 6569-6573.	0.4	46
61	Repertoire-dependent immunopathology. <i>Journal of Autoimmunity</i> , 2007, 29, 257-261.	3.0	44
62	Primary Atopic Disorders. <i>Annual Review of Immunology</i> , 2020, 38, 785-808.	9.5	40
63	Epicutaneous <i>Staphylococcus aureus</i> induces IL-36 to enhance IgE production and ensuing allergic disease. <i>Journal of Clinical Investigation</i> , 2021, 131, .	3.9	39
64	IL-17 producing cells in host defense and atopy. <i>Current Opinion in Immunology</i> , 2011, 23, 784-788.	2.4	38
65	Multiplexed Functional Assessment of Genetic Variants in <i>CARD11</i> . <i>American Journal of Human Genetics</i> , 2020, 107, 1029-1043.	2.6	38
66	Autoimmunity in Severe Combined Immunodeficiency (SCID): Lessons from Patients and Experimental Models. <i>Journal of Clinical Immunology</i> , 2008, 28, 29-33.	2.0	37
67	The autoinhibitory C-terminal SH2 domain of phospholipase $\text{C}\beta 2$ stabilizes B cell receptor signalosome assembly. <i>Science Signaling</i> , 2014, 7, ra89.	1.6	32
68	Clinical relevance of inherited genetic differences in human tryptases. <i>Annals of Allergy, Asthma and Immunology</i> , 2021, 127, 638-647.	0.5	30
69	A common haplotype containing functional <i>CACNA1H</i> variants is frequently coinherited with increased <i>TPSAB1</i> copy number. <i>Genetics in Medicine</i> , 2018, 20, 503-512.	1.1	25
70	Elevated IgE and atopy in patients treated for early-onset ADA-SCID. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 132, 1444-1446.e5.	1.5	22
71	Inherited <i>SLP76</i> deficiency in humans causes severe combined immunodeficiency, neutrophil and platelet defects. <i>Journal of Experimental Medicine</i> , 2021, 218, .	4.2	20
72	Does Elevated Peak Bilirubin Protect from Retinopathy of Prematurity in Very Low Birthweight Infants. <i>Journal of Perinatology</i> , 2003, 23, 208-211.	0.9	19

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73	Pathogen-Sensing, Regulatory T Cells, and Responsiveness-Tuning Collectively Regulate Foreign- and Self-Antigen Mediated T-Cell Responses. <i>Cold Spring Harbor Symposia on Quantitative Biology</i> , 2013, 78, 265-276.	2.0	19
74	Small intestinal immunopathology and GI-associated antibody formation in hereditary alpha-tryptasemia. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 148, 813-821.e7.	1.5	17
75	The overlap between allergy and immunodeficiency. <i>Current Opinion in Pediatrics</i> , 2018, 30, 848-854.	1.0	16
76	JAK inhibition in early-onset somatic, nonclonal STAT5B gain-of-function disease. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2021, 9, 1008-1010.e2.	2.0	16
77	Altered T-cell receptor signaling in the pathogenesis of allergic disease. <i>Journal of Allergy and Clinical Immunology</i> , 2011, 127, 351-354.	1.5	15
78	Atopy as Immune Dysregulation: Offender Genes and Targets. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2022, 10, 1737-1756.	2.0	15
79	Functional characterization of phospholipase C- β 2 mutant protein causing both somatic ibrutinib resistance and a germline monogenic autoinflammatory disorder. <i>Oncotarget</i> , 2018, 9, 34357-34378.	0.8	14
80	Long-Acting Beta Agonists Enhance Allergic Airway Disease. <i>PLoS ONE</i> , 2015, 10, e0142212.	1.1	13
81	Atopic Dermatitis and Allergic Urticaria. <i>Immunology and Allergy Clinics of North America</i> , 2017, 37, 1-10.	0.7	13
82	Inborn Error of Immunity or Atopic Dermatitis: When to be Concerned and How to Investigate. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2021, 9, 1501-1507.	2.0	13
83	Impaired angiogenesis and extracellular matrix metabolism in autosomal-dominant hyper-IgE syndrome. <i>Journal of Clinical Investigation</i> , 2020, 130, 4167-4181.	3.9	13
84	The clinical and mechanistic intersection of primary atopic disorders and inborn errors of growth and metabolism. <i>Immunological Reviews</i> , 2019, 287, 135-144.	2.8	12
85	Systematic evaluation of nine monogenic autoinflammatory diseases reveals common and disease-specific correlations with allergy-associated features. <i>Annals of the Rheumatic Diseases</i> , 2021, 80, 788-795.	0.5	12
86	Gene-environment interactions in primary atopic disorders. <i>Current Opinion in Immunology</i> , 2019, 60, 148-155.	2.4	11
87	The Child with Elevated IgE and Infection Susceptibility. <i>Current Allergy and Asthma Reports</i> , 2020, 20, 65.	2.4	11
88	STAT3 Gain-of-Function Mutations Underlie Deficiency in Human Nonclassical CD16+ Monocytes and CD141+ Dendritic Cells. <i>Journal of Immunology</i> , 2021, 207, 2423-2432.	0.4	11
89	Detection of phosphoglucomutase-3 deficiency by lectin-based flow cytometry. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 140, 291-294.e4.	1.5	10
90	Human TH9 differentiation is dependent on signal transducer and activator of transcription (STAT) 3 to restrain STAT1-mediated inhibition. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 1108-1118.e4.	1.5	10

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91	PLAID syndrome: Characteristic presentation and a novel therapeutic option. <i>Pediatric Dermatology</i> , 2020, 37, 147-149.	0.5	9
92	A Novel Germline Heterozygous BCL11B Variant Causing Severe Atopic Disease and Immune Dysregulation. <i>Frontiers in Immunology</i> , 2021, 12, 788278.	2.2	9
93	Ex vivo effect of JAK inhibition on JAK-STAT1 pathway hyperactivation in patients with dominant-negative STAT3 mutations. <i>Journal of Clinical Immunology</i> , 2022, 42, 1193-1204.	2.0	8
94	TCR Signaling Abnormalities in Human Th2-Associated Atopic Disease. <i>Frontiers in Immunology</i> , 2018, 9, 719.	2.2	7
95	Case Report: Secondary Hemophagocytic Lymphohistiocytosis With Disseminated Infection in Chronic Granulomatous Disease—A Serious Cause of Mortality. <i>Frontiers in Immunology</i> , 2020, 11, 581475.	2.2	6
96	Transient environmental exposures on the developing immune system: implications for allergy and asthma. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2005, 5, 235-240.	1.1	5
97	Exogenous vitamin D might contribute to geographic variations in epinephrine prescriptions. <i>Journal of Allergy and Clinical Immunology</i> , 2008, 121, 265-266.	1.5	5
98	Peptide library-based evaluation of T-cell receptor breadth detects defects in global and regulatory activation in human immunologic diseases. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 8164-8169.	3.3	5
99	Low IgE Is Insufficiently Sensitive to Guide Genetic Testing of STAT3 Gain-of-Function Mutations. <i>Clinical Chemistry</i> , 2017, 63, 1539-1540.	1.5	4
100	Compound Heterozygous PGM3 Mutations in a Thai Patient with a Specific Antibody Deficiency Requiring Monthly IVIG Infusions. <i>Journal of Clinical Immunology</i> , 2020, 40, 227-231.	2.0	4
101	Assessing the safety of bioactive ingredients in infant formula that affect the immune system: recommendations from an expert panel. <i>American Journal of Clinical Nutrition</i> , 2022, 115, 570-587.	2.2	3
102	Learning from Job: A Rare Genetic Disease and Lessons of Biblical Proportions. <i>Rambam Maimonides Medical Journal</i> , 2018, 9, e0006.	0.4	2
103	Editorial overview: Collusion between genes and environment in the pathogenesis of allergic disease. <i>Current Opinion in Immunology</i> , 2019, 60, iii-v.	2.4	1
104	Primary immune deficiencies associated with a Th2 diathesis. , 2020, , 393-410.		1
105	Recurrent lymphadenitis in a female XIAP/BIRC4 mutation carrier with normal lyonization. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2021, 9, 1002-1005.e2.	2.0	1
106	Hereditary alpha-tryptasemia modifies clinical phenotypes among individuals with congenital hypermobility disorders. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100094.	1.0	1