

Amy P Hsu

List of Publications by Year in descending order

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112
papers

10,344
citations

50276

46
h-index

33894

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112
all docs

112
docs citations

112
times ranked

10463
citing authors

#	ARTICLE	IF	CITATIONS
1	<i>STAT3</i> Mutations in the Hyper-IgE Syndrome. <i>New England Journal of Medicine</i> , 2007, 357, 1608-1619.	27.0	1,098
2	Impaired TH17 cell differentiation in subjects with autosomal dominant hyper-IgE syndrome. <i>Nature</i> , 2008, 452, 773-776.	27.8	1,046
3	GATA2 deficiency: a protean disorder of hematopoiesis, lymphatics, and immunity. <i>Blood</i> , 2014, 123, 809-821.	1.4	599
4	Mutations in GATA2 are associated with the autosomal dominant and sporadic monocytopenia and mycobacterial infection (MonoMAC) syndrome. <i>Blood</i> , 2011, 118, 2653-2655.	1.4	572
5	Adult-Onset Immunodeficiency in Thailand and Taiwan. <i>New England Journal of Medicine</i> , 2012, 367, 725-734.	27.0	431
6	Autosomal dominant and sporadic monocytopenia with susceptibility to mycobacteria, fungi, papillomaviruses, and myelodysplasia. <i>Blood</i> , 2010, 115, 1519-1529.	1.4	299
7	Mutations in STAT3 and diagnostic guidelines for hyper-IgE syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2010, 125, 424-432.e8.	2.9	247
8	Loss-of-function germline GATA2 mutations in patients with MDS/AML or MonoMAC syndrome and primary lymphedema reveal a key role for GATA2 in the lymphatic vasculature. <i>Blood</i> , 2012, 119, 1283-1291.	1.4	244
9	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.	14.3	241
10	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.	2.9	222
11	Redefined clinical features and diagnostic criteria in autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy. <i>JCI Insight</i> , 2016, 1, .	5.0	219
12	GATA2 haploinsufficiency caused by mutations in a conserved intronic element leads to MonoMAC syndrome. <i>Blood</i> , 2013, 121, 3830-3837.	1.4	209
13	Mutations in GATA2 cause human NK cell deficiency with specific loss of the CD56bright subset. <i>Blood</i> , 2013, 121, 2669-2677.	1.4	208
14	Natural history of autoimmune lymphoproliferative syndrome associated with FAS gene mutations. <i>Blood</i> , 2014, 123, 1989-1999.	1.4	204
15	Autoimmune Lymphoproliferative Syndrome with Defective Fas: Genotype Influences Penetrance. <i>American Journal of Human Genetics</i> , 1999, 64, 1002-1014.	6.2	198
16	CARD9-Dependent Neutrophil Recruitment Protects against Fungal Invasion of the Central Nervous System. <i>PLoS Pathogens</i> , 2015, 11, e1005293.	4.7	184
17	Cis-element mutated in GATA2-dependent immunodeficiency governs hematopoiesis and vascular integrity. <i>Journal of Clinical Investigation</i> , 2012, 122, 3692-3704.	8.2	162
18	Extrapulmonary Aspergillus infection in patients with CARD9 deficiency. <i>JCI Insight</i> , 2016, 1, e89890.	5.0	141

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19	Hematopoietic stem cell transplantation rescues the hematological, immunological, and vascular phenotype in DADA2. <i>Blood</i> , 2017, 130, 2682-2688.	1.4	140
20	Pulmonary Nontuberculous Mycobacterial Infection. A Multisystem, Multigenic Disease. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2015, 192, 618-628.	5.6	136
21	Anti-cytokine autoantibodies are associated with opportunistic infection in patients with thymic neoplasia. <i>Blood</i> , 2010, 116, 4848-4858.	1.4	134
22	Dominant-negative IKZF1 mutations cause a T, B, and myeloid cell combined immunodeficiency. <i>Journal of Clinical Investigation</i> , 2018, 128, 3071-3087.	8.2	133
23	Successful allogeneic hematopoietic stem cell transplantation for GATA2 deficiency. <i>Blood</i> , 2011, 118, 3715-3720.	1.4	131
24	GATA2 deficiency-associated bone marrow disorder differs from idiopathic aplastic anemia. <i>Blood</i> , 2015, 125, 56-70.	1.4	131
25	Acquired ASXL1 mutations are common in patients with inherited GATA2 mutations and correlate with myeloid transformation. <i>Haematologica</i> , 2014, 99, 276-281.	3.5	119
26	GATA2 deficiency. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2015, 15, 104-109.	2.3	113
27	Gene therapy improves immune function in preadolescents with X-linked severe combined immunodeficiency. <i>Blood</i> , 2007, 110, 67-73.	1.4	97
28	Myelodysplasia in autosomal dominant and sporadic monocytopenia immunodeficiency syndrome: diagnostic features and clinical implications. <i>Haematologica</i> , 2011, 96, 1221-1225.	3.5	97
29	Invasive fungal disease in autosomal-dominant hyper-IgE syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2010, 125, 1389-1390.	2.9	91
30	Adaptive NK cells can persist in patients with GATA2 mutation depleted of stem and progenitor cells. <i>Blood</i> , 2017, 129, 1927-1939.	1.4	89
31	Interleukin-12 Receptor α 1 Deficiency Predisposing to Disseminated Coccidioidomycosis. <i>Clinical Infectious Diseases</i> , 2011, 52, e99-e102.	5.8	87
32	Reversion mutations in patients with leukocyte adhesion deficiency type-1 (LAD-1). <i>Blood</i> , 2008, 111, 209-218.	1.4	76
33	4 Primary immunodeficiency mutation databases. <i>Advances in Genetics</i> , 2001, 43, 103-188.	1.8	70
34	Highly Variable Clinical Phenotypes of Hypomorphic <i>RAG1</i> Mutations. <i>Pediatrics</i> , 2010, 126, e1248-e1252.	2.1	70
35	Genetic alterations in caspase-10 may be causative or protective in autoimmune lymphoproliferative syndrome. <i>Human Genetics</i> , 2006, 119, 284-294.	3.8	68
36	Coronary Artery Abnormalities in Hyper-IgE Syndrome. <i>Journal of Clinical Immunology</i> , 2011, 31, 338-345.	3.8	64

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37	A Novel STAT1 Mutation Associated with Disseminated Mycobacterial Disease. <i>Journal of Clinical Immunology</i> , 2012, 32, 681-689.	3.8	61
38	Dominant activating RAC2 mutation with lymphopenia, immunodeficiency, and cytoskeletal defects. <i>Blood</i> , 2019, 133, 1977-1988.	1.4	61
39	Progressive Multifocal Leukoencephalopathy in Primary Immune Deficiencies: Stat1 Gain of Function and Review of the Literature. <i>Clinical Infectious Diseases</i> , 2016, 62, 986-994.	5.8	59
40	Association of GATA2 Deficiency With Severe Primary Epstein-Barr Virus (EBV) Infection and EBV-associated Cancers. <i>Clinical Infectious Diseases</i> , 2016, 63, 41-47.	5.8	56
41	Risks of Ruxolitinib in STAT1 Gain-of-Function-Associated Severe Fungal Disease. <i>Open Forum Infectious Diseases</i> , 2017, 4, ofx202.	0.9	56
42	Pediatric CNS-isolated hemophagocytic lymphohistiocytosis. <i>Neurology: Neuroimmunology and Neuroinflammation</i> , 2019, 6, e560.	6.0	54
43	MonoMAC Syndrome in a Patient With a GATA2 Mutation: Case Report and Review of the Literature. <i>Clinical Infectious Diseases</i> , 2013, 57, 697-699.	5.8	53
44	Gastrointestinal Manifestations of STAT3-Deficient Hyper-IgE Syndrome. <i>Journal of Clinical Immunology</i> , 2017, 37, 695-700.	3.8	52
45	Lymphocyte-driven regional immunopathology in pneumonitis caused by impaired central immune tolerance. <i>Science Translational Medicine</i> , 2019, 11, .	12.4	52
46	Intermediate phenotypes in patients with autosomal dominant hyper-IgE syndrome caused by somatic mosaicism. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 131, 1586-1593.	2.9	50
47	Gain-of-function signal transducer and activator of transcription 1 (STAT1) mutation-related primary immunodeficiency is associated with disseminated mucormycosis. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 134, 236-239.	2.9	50
48	Clonal Diversification and Changes in Lipid Traits and Colony Morphology in Mycobacterium abscessus Clinical Isolates. <i>Journal of Clinical Microbiology</i> , 2015, 53, 3438-3447.	3.9	48
49	Donor-derived MDS/AML in families with germline GATA2 mutation. <i>Blood</i> , 2018, 132, 1994-1998.	1.4	48
50	Spectrum of myeloid neoplasms and immune deficiency associated with germline GATA2 mutations. <i>Cancer Medicine</i> , 2015, 4, 490-499.	2.8	43
51	Hematopoietic Cell Transplantation Cures Adenosine Deaminase 2 Deficiency: Report on 30 Patients. <i>Journal of Clinical Immunology</i> , 2021, 41, 1633-1647.	3.8	43
52	Autoimmune lymphoproliferative syndrome due to FAS mutations outside the signal-transducing death domain: molecular mechanisms and clinical penetrance. <i>Genetics in Medicine</i> , 2012, 14, 81-89.	2.4	41
53	Adenosine deaminase type 2 deficiency masquerading as GATA2 deficiency: Successful hematopoietic stem cell transplantation. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 628-630.e2.	2.9	41
54	STAT1 Gain-of-Function Mutations Cause High Total STAT1 Levels With Normal Dephosphorylation. <i>Frontiers in Immunology</i> , 2019, 10, 1433.	4.8	41

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55	Paucity of genotype-phenotype correlations in STAT3 mutation positive Hyper IgE Syndrome (HIES). <i>Clinical Immunology</i> , 2011, 139, 75-84.	3.2	39
56	Lung Parenchyma Surgery in Autosomal Dominant Hyper-IgE Syndrome. <i>Journal of Clinical Immunology</i> , 2013, 33, 896-902.	3.8	39
57	A review of innate and adaptive immunity to coccidioidomycosis. <i>Medical Mycology</i> , 2019, 57, S85-S92.	0.7	39
58	GATA2 deficiency underlying severe <i>Aspergill</i> blastomycosis and fatal herpes simplex virus-associated hemophagocytic lymphohistiocytosis. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 137, 638-640.	2.9	36
59	MDS-associated mutations in germline GATA2 mutated patients with hematologic manifestations. <i>Leukemia Research</i> , 2019, 76, 70-75.	0.8	33
60	Plasma metalloproteinase levels are dysregulated in signal transducer and activator of transcription 3 mutated hyper-IgE syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2011, 128, 1124-1127.	2.9	32
61	IL2RG Reversion Event in a Common Lymphoid Progenitor Leads to Delayed Diagnosis and Milder Phenotype. <i>Journal of Clinical Immunology</i> , 2015, 35, 449-453.	3.8	32
62	Efficient Detection of Thirty-Seven New IL2RG Mutations in Human X-Linked Severe Combined Immunodeficiency. <i>Clinical Immunology</i> , 2000, 95, 33-38.	3.2	31
63	Mutations causing severe combined immunodeficiency: detection with a custom resequencing microarray. <i>Genetics in Medicine</i> , 2008, 10, 575-585.	2.4	31
64	Pulmonary nontuberculous mycobacterial infections in hyper-IgE syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2009, 124, 617-618.	2.9	31
65	Bone Density and Fractures in Autosomal Dominant Hyper IgE Syndrome. <i>Journal of Clinical Immunology</i> , 2014, 34, 260-264.	3.8	28
66	Prospective Study of a Novel, Radiation-Free, Reduced-Intensity Bone Marrow Transplantation Platform for Primary Immunodeficiency Diseases. <i>Biology of Blood and Marrow Transplantation</i> , 2020, 26, 94-106.	2.0	28
67	Constructing and deconstructing GATA2-regulated cell fate programs to establish developmental trajectories. <i>Journal of Experimental Medicine</i> , 2020, 217, .	8.5	28
68	Heterozygous activating mutation in RAC2 causes infantile-onset combined immunodeficiency with susceptibility to viral infections. <i>Clinical Immunology</i> , 2019, 205, 1-5.	3.2	27
69	GATA2 deficiency and haematopoietic stem cell transplantation: challenges for the clinical practitioner. <i>British Journal of Haematology</i> , 2020, 188, 768-773.	2.5	27
70	GATA2 deficiency and human hematopoietic development modeled using induced pluripotent stem cells. <i>Blood Advances</i> , 2018, 2, 3553-3565.	5.2	25
71	<i>ASXL1</i> and <i>STAG2</i> are common mutations in GATA2 deficiency patients with bone marrow disease and myelodysplastic syndrome. <i>Blood Advances</i> , 2022, 6, 793-807.	5.2	24
72	Distinct mutations at the same positions of STAT3 cause either loss or gain of function. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 1222-1224.e2.	2.9	23

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73	Somatic mutations in children with GATA2-associated myelodysplastic syndrome who lack other features of GATA2 deficiency. <i>Blood Advances</i> , 2017, 1, 443-448.	5.2	23
74	Sequencing of RNA in single cells reveals a distinct transcriptome signature of hematopoiesis in GATA2 deficiency. <i>Blood Advances</i> , 2020, 4, 2702-2716.	5.2	23
75	Treatment of Relapsing HPV Diseases by Restored Function of Natural Killer Cells. <i>New England Journal of Medicine</i> , 2021, 385, 921-929.	27.0	22
76	Hematologically important mutations: X-linked chronic granulomatous disease (fourth update). <i>Blood Cells, Molecules, and Diseases</i> , 2021, 90, 102587.	1.4	22
77	Hematologically important mutations: The autosomal forms of chronic granulomatous disease (third) Tj ETQq1 1 0,784314 rgBT /Ovelde	1.4	22
78	Complete Myeloperoxidase Deficiency: Beware the "False-Positive" Dihydrorhodamine Oxidation. <i>Journal of Pediatrics</i> , 2016, 176, 204-206.	1.8	21
79	GATA-2-deficient mast cells limit IgE-mediated immediate hypersensitivity reactions in human subjects. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 144, 613-617.e14.	2.9	21
80	Pulmonary Manifestations of GATA2 Deficiency. <i>Chest</i> , 2021, 160, 1350-1359.	0.8	21
81	NCF1 (p47phox)-deficient chronic granulomatous disease: comprehensive genetic and flow cytometric analysis. <i>Blood Advances</i> , 2019, 3, 136-147.	5.2	20
82	Regulatory Mutations in GATA2 Associated with Aplastic Anemia. <i>Blood</i> , 2012, 120, 3488-3488.	1.4	20
83	Aspergillosis, eosinophilic esophagitis, and allergic rhinitis in signal transducer and activator of transcription 3 haploinsufficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 142, 993-997.e3.	2.9	19
84	Donor source and post-transplantation cyclophosphamide influence outcome in allogeneic stem cell transplantation for GATA2 deficiency. <i>British Journal of Haematology</i> , 2022, 196, 169-178.	2.5	18
85	Coronary Abnormalities in Hyper-IgE Recurrent Infection Syndrome: Depiction at Coronary MDCT Angiography. <i>American Journal of Roentgenology</i> , 2009, 193, W478-W481.	2.2	16
86	Mutation analysis in primary immunodeficiency diseases: case studies. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2009, 9, 517-524.	2.3	15
87	Genetically programmed alternative splicing of NEMO mediates an autoinflammatory disease phenotype. <i>Journal of Clinical Investigation</i> , 2022, 132, .	8.2	15
88	Retroviral transduction of IL2RG into CD34+ cells from X-linked severe combined immunodeficiency patients permits human T- and B-cell development in sheep chimeras. <i>Blood</i> , 2002, 100, 72-79.	1.4	14
89	Persistent nodal histoplasmosis in nuclear factor kappa B essential modulator deficiency: Report of a case and review of infection in primary immunodeficiencies. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 903-905.	2.9	14
90	A novel IL2RG mutation associated with maternal T lymphocyte engraftment in a patient with severe combined immunodeficiency. <i>Journal of Human Genetics</i> , 2006, 51, 495-497.	2.3	13

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91	Melanoma in patients with <sc>GATA</sc>2 deficiency. Pigment Cell and Melanoma Research, 2018, 31, 337-340.	3.3	13
92	Outcomes of Related and Unrelated Donor Searches Among Patients with Primary Immunodeficiency Diseases Referred for Allogeneic Hematopoietic Cell Transplantation. Biology of Blood and Marrow Transplantation, 2019, 25, 1666-1673.	2.0	13
93	Impaired angiogenesis and extracellular matrix metabolism in autosomal-dominant hyper-IgE syndrome. Journal of Clinical Investigation, 2020, 130, 4167-4181.	8.2	13
94	Single nucleotide polymorphisms in the apoptosis receptor gene TNFRSF6. Molecular and Cellular Probes, 2006, 20, 21-26.	2.1	12
95	Association of unbalanced translocation der(1;7) with germline GATA2 mutations. Blood, 2021, 138, 2441-2445.	1.4	12
96	Multiple Opportunistic Infections in a Woman with GATA2 Mutation. International Journal of Infectious Diseases, 2017, 54, 89-91.	3.3	11
97	Rapid progression to AML in a patient with germline GATA2 mutation and acquired NRAS Q61K mutation. Leukemia Research Reports, 2019, 12, 100176.	0.4	11
98	Pathologic Findings in NEMO Deficiency: A Surgical and Autopsy Survey. Pediatric and Developmental Pathology, 2015, 18, 387-400.	1.0	10
99	Lost in Translation: Lack of CD4 Expression due to a Novel Genetic Defect. Journal of Infectious Diseases, 2021, 223, 645-654.	4.0	10
100	Pneumocystis jiroveci pneumonia and GATA2 deficiency: Expanding the spectrum of the disease. Journal of Infection, 2017, 74, 425-427.	3.3	9
101	A Novel RAC2 Variant Presenting as Severe Combined Immunodeficiency. Journal of Clinical Immunology, 2021, 41, 473-476.	3.8	9
102	Hematopoietic Cell Transplantation and Outcomes Related to Human Papillomavirus Disease in GATA2 Deficiency. Transplantation and Cellular Therapy, 2021, 27, 435.e1-435.e11.	1.2	9
103	A Panoply of Rheumatological Manifestations in Patients with GATA2 Deficiency. Scientific Reports, 2020, 10, 8305.	3.3	8
104	Host genetics of innate immune system in infection. Current Opinion in Immunology, 2022, 74, 140-149.	5.5	8
105	IKBKG (NEMO) 5' Untranslated Splice Mutations Lead to Severe, Chronic Disseminated Mycobacterial Infections. Clinical Infectious Diseases, 2018, 67, 456-459.	5.8	6
106	Generation of human induced pluripotent stem cell lines (NIHTVBi011-A, NIHTVBi012-A, NIHTVBi013-A) from autosomal dominant Hyper IgE syndrome (AD-HIES) patients carrying STAT3 mutation. Stem Cell Research, 2019, 41, 101586.	0.7	5
107	Patients With Natural Killer (NK) Cell Chronic Active Epstein-Barr Virus Have Immature NK Cells and Hyperactivation of PI3K/Akt/mTOR and STAT1 Pathways. Journal of Infectious Diseases, 2020, 222, 1170-1179.	4.0	5
108	Brain Abscess as Severe Presentation of Specific Granule Deficiency. Frontiers in Pediatrics, 2020, 8, 117.	1.9	3

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109	miR-181c regulates MCL1 and cell survival in GATA2 deficient cells. <i>Journal of Leukocyte Biology</i> , 2022, 111, 805-816.	3.3	3
110	West Nile virus encephalitis in GATA2 deficiency. <i>Allergy, Asthma and Clinical Immunology</i> , 2019, 15, 5.	2.0	2
111	Bone marrow failure syndromes. , 2020, , 411-441.		1
112	Molecular Methods for Diagnosis of Genetic Diseases Involving the Immune System. , 2016, , 5-18.		0