Amy P Hsu

List of Publications by Year
in descending order

112 papers	10,344 citations	50276 46 h-index	33894 99 g-index
112 all docs	112 docs citations	112 times ranked	10463 citing authors

#	Article	IF	Citations
1	miR-181c regulates MCL1 and cell survival in GATA2 deficient cells. Journal of Leukocyte Biology, 2022, 111, 805-816.	3.3	3
2	Donor source and postâ€transplantation cyclophosphamide influence outcome in allogeneic stem cell transplantation for GATA2 deficiency. British Journal of Haematology, 2022, 196, 169-178.	2.5	18
3	ASXL1 and STAG2 are common mutations in GATA2 deficiency patients with bone marrow disease and myelodysplastic syndrome. Blood Advances, 2022, 6, 793-807.	5.2	24
4	Host genetics of innate immune system in infection. Current Opinion in Immunology, 2022, 74, 140-149.	5.5	8
5	Genetically programmed alternative splicing of NEMO mediates an autoinflammatory disease phenotype. Journal of Clinical Investigation, 2022, 132, .	8.2	15
6	A Novel RAC2 Variant Presenting as Severe Combined Immunodeficiency. Journal of Clinical Immunology, 2021, 41, 473-476.	3.8	9
7	Lost in Translation: Lack of CD4 Expression due to a Novel Genetic Defect. Journal of Infectious Diseases, 2021, 223, 645-654.	4.0	10
8	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
9	Pulmonary Manifestations of GATA2 Deficiency. Chest, 2021, 160, 1350-1359.	0.8	21
10	Hematopoietic Cell Transplantation Cures Adenosine Deaminase 2 Deficiency: Report on 30 Patients. Journal of Clinical Immunology, 2021, 41, 1633-1647.	3.8	43
11	Treatment of Relapsing HPV Diseases by Restored Function of Natural Killer Cells. New England Journal of Medicine, 2021, 385, 921-929.	27.0	22
12	Association of unbalanced translocation der(1;7) with germline GATA2 mutations. Blood, 2021, 138, 2441-2445.	1.4	12
13	Hematologically important mutations: X-linked chronic granulomatous disease (fourth update). Blood Cells, Molecules, and Diseases, 2021, 90, 102587.	1.4	22
14	Hematologically important mutations: The autosomal forms of chronic granulomatous disease (third) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	1.4	22
15	Prospective Study of a Novel, Radiation-Free, Reduced-Intensity Bone Marrow Transplantation Platform for Primary Immunodeficiency Diseases. Biology of Blood and Marrow Transplantation, 2020, 26, 94-106.	2.0	28
16	GATA2 deficiency and haematopoietic stem cell transplantation: challenges for the clinical practitioner. British Journal of Haematology, 2020, 188, 768-773.	2.5	27
17	Constructing and deconstructing GATA2-regulated cell fate programs to establish developmental trajectories. Journal of Experimental Medicine, 2020, 217, .	8.5	28
18	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

#	Article	IF	Citations
19	Brain Abscess as Severe Presentation of Specific Granule Deficiency. Frontiers in Pediatrics, 2020, 8, 117.	1.9	3
20	A Panoply of Rheumatological Manifestations in Patients with GATA2 Deficiency. Scientific Reports, 2020, 10, 8305.	3.3	8
21	Bone marrow failure syndromes. , 2020, , 411-441.		1
22	Sequencing of RNA in single cells reveals a distinct transcriptome signature of hematopoiesis in GATA2 deficiency. Blood Advances, 2020, 4, 2702-2716.	5.2	23
23	Impaired angiogenesis and extracellular matrix metabolism in autosomal-dominant hyper-IgE syndrome. Journal of Clinical Investigation, 2020, 130, 4167-4181.	8.2	13
24	STAT1 Gain-of-Function Mutations Cause High Total STAT1 Levels With Normal Dephosphorylation. Frontiers in Immunology, 2019, 10, 1433.	4.8	41
25	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
26	GATA-2â€“deficient mast cells limit IgE-mediated immediate hypersensitivity reactions in human subjects. Journal of Allergy and Clinical Immunology, 2019, 144, 613-617.e14.	2.9	21
27	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
28	Lymphocyte-driven regional immunopathology in pneumonitis caused by impaired central immune tolerance. Science Translational Medicine, 2019, 11, .	12.4	52
29	Pediatric CNS-isolated hemophagocytic lymphohistiocytosis. Neurology: Neuroimmunology and NeuroInflammation, 2019, 6, e560.	6.0	54
30	Heterozygous activating mutation in RAC2 causes infantile-onset combined immunodeficiency with susceptibility to viral infections. Clinical Immunology, 2019, 205, 1-5.	3.2	27
31	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
32	Dominant activating RAC2 mutation with lymphopenia, immunodeficiency, and cytoskeletal defects. Blood, 2019, 133, 1977-1988.	1.4	61
33	West Nile virus encephalitis in GATA2 deficiency. Allergy, Asthma and Clinical Immunology, 2019, 15, 5.	2.0	2
34	NCF1 (p47phox)â€“deficient chronic granulomatous disease: comprehensive genetic and flow cytometric analysis. Blood Advances, 2019, 3, 136-147.	5.2	20
35	MDS-associated mutations in germline GATA2 mutated patients with hematologic manifestations. Leukemia Research, 2019, 76, 70-75.	0.8	33
36	A review of innate and adaptive immunity to coccidioidomycosis. Medical Mycology, 2019, 57, S85-S92.	0.7	39

#	Article	IF	Citations
37	IKBKG (NEMO) 5â€² Untranslated Splice Mutations Lead to Severe, Chronic Disseminated Mycobacterial Infections. Clinical Infectious Diseases, 2018, 67, 456-459.	5.8	6
38	Melanoma in patients with <scp>GATA</scp>2 deficiency. Pigment Cell and Melanoma Research, 2018, 31, 337-340.	3.3	13
39	Donor-derived MDS/AML in families with germline GATA2 mutation. Blood, 2018, 132, 1994-1998.	1.4	48
40	GATA2 deficiency and human hematopoietic development modeled using induced pluripotent stem cells. Blood Advances, 2018, 2, 3553-3565.	5.2	25
41	Aspergillosis, eosinophilic esophagitis, and allergic rhinitis in signal transducer and activator of transcription 3 haploinsufficiency. Journal of Allergy and Clinical Immunology, 2018, 142, 993-997.e3.	2.9	19
42	Dominant-negative IKZF1 mutations cause a T, B, and myeloid cell combined immunodeficiency. Journal of Clinical Investigation, 2018, 128, 3071-3087.	8.2	133
43	Pneumocystis jiroveci pneumonia and GATA2 deficiency: Expanding the spectrum of the disease. Journal of Infection, 2017, 74, 425-427.	3.3	9
44	Adaptive NK cells can persist in patients with GATA2 mutation depleted of stem and progenitor cells. Blood, 2017, 129, 1927-1939.	1.4	89
45	Multiple Opportunistic Infections in a Woman with GATA2 Mutation. International Journal of Infectious Diseases, 2017, 54, 89-91.	3.3	11
46	Hematopoietic stem cell transplantation rescues the hematological, immunological, and vascular phenotype in DADA2. Blood, 2017, 130, 2682-2688.	1.4	140
47	Gastrointestinal Manifestations of STAT3-Deficient Hyper-IgE Syndrome. Journal of Clinical Immunology, 2017, 37, 695-700.	3.8	52
48	Risks of Ruxolitinib in STAT1 Gain-of-Function-Associated Severe Fungal Disease. Open Forum Infectious Diseases, 2017, 4, ofx202.	0.9	56
49	Somatic mutations in children with GATA2-associated myelodysplastic syndrome who lack other features of GATA2 deficiency. Blood Advances, 2017, 1, 443-448.	5.2	23
50	Molecular Methods for Diagnosis of Genetic Diseases Involving the Immune System. , 2016, , 5-18.		0
51	Redefined clinical features and diagnostic criteria in autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy. JCI Insight, 2016, 1, .	5.0	219
52	Persistent nodal histoplasmosis in nuclear factor kappa B essential modulator deficiency: Report of a case and review of infection in primary immunodeficiencies. Journal of Allergy and Clinical Immunology, 2016, 138, 903-905.	2.9	14
53	Adenosine deaminase type 2 deficiency masquerading as GATA2 deficiency: Successful hematopoietic stem cell transplantation. Journal of Allergy and Clinical Immunology, 2016, 138, 628-630.e2.	2.9	41
54	Association of GATA2 Deficiency With Severe Primary Epstein-Barr Virus (EBV) Infection and EBV-associated Cancers. Clinical Infectious Diseases, 2016, 63, 41-47.	5.8	56

#	Article	IF	Citations
55	Complete Myeloperoxidase Deficiency: Beware the â€œFalse-Positiveâ€•Dihydrorhodamine Oxidation. Journal of Pediatrics, 2016, 176, 204-206.	1.8	21
56	Distinct mutations at the same positions of STAT3 cause either loss or gain of function. Journal of Allergy and Clinical Immunology, 2016, 138, 1222-1224.e2.	2.9	23
57	Progressive Multifocal Leukoencephalopathy in Primary Immune Deficiencies: Stat1 Gain of Function and Review of the Literature. Clinical Infectious Diseases, 2016, 62, 986-994.	5.8	59
58	GATA2 deficiency underlying severeÂblastomycosis and fatal herpes simplex virusâ€“associated hemophagocytic lymphohistiocytosis. Journal of Allergy and Clinical Immunology, 2016, 137, 638-640.	2.9	36
59	Extrapulmonary Aspergillus infection in patients with CARD9 deficiency. JCI Insight, 2016, 1, e89890.	5.0	141
60	Spectrum of myeloid neoplasms and immune deficiency associated with germline <scp>GATA</scp>2 mutations. Cancer Medicine, 2015, 4, 490-499.	2.8	43
61	GATA2 deficiency-associated bone marrow disorder differs from idiopathic aplastic anemia. Blood, 2015, 125, 56-70.	1.4	131
62	Pulmonary Nontuberculous Mycobacterial Infection. A Multisystem, Multigenic Disease. American Journal of Respiratory and Critical Care Medicine, 2015, 192, 618-628.	5.6	136
63	IL2RG Reversion Event in a Common Lymphoid Progenitor Leads to Delayed Diagnosis and Milder Phenotype. Journal of Clinical Immunology, 2015, 35, 449-453.	3.8	32
64	GATA2 deficiency. Current Opinion in Allergy and Clinical Immunology, 2015, 15, 104-109.	2.3	113
65	Clonal Diversification and Changes in Lipid Traits and Colony Morphology in Mycobacterium abscessus Clinical Isolates. Journal of Clinical Microbiology, 2015, 53, 3438-3447.	3.9	48
66	Pathologic Findings in NEMO Deficiency: A Surgical and Autopsy Survey. Pediatric and Developmental Pathology, 2015, 18, 387-400.	1.0	10
67	CARD9-Dependent Neutrophil Recruitment Protects against Fungal Invasion of the Central Nervous System. PLoS Pathogens, 2015, 11, e1005293.	4.7	184
68	Acquired ASXL1 mutations are common in patients with inherited GATA2 mutations and correlate with myeloid transformation. Haematologica, 2014, 99, 276-281.	3.5	119
69	Natural history of autoimmune lymphoproliferative syndrome associated with FAS gene mutations. Blood, 2014, 123, 1989-1999.	1.4	204
70	Gain-of-function signal transducer and activator of transcription 1 (STAT1) mutationâ€“related primary immunodeficiency is associated with disseminated mucormycosis. Journal of Allergy and Clinical Immunology, 2014, 134, 236-239.	2.9	50
71	Bone Density and Fractures in Autosomal Dominant Hyper IgE Syndrome. Journal of Clinical Immunology, 2014, 34, 260-264.	3.8	28
72	GATA2 deficiency: a protean disorder of hematopoiesis, lymphatics, and immunity. Blood, 2014, 123, 809-821.	1.4	599

#	Article	IF	Citations
73	Lung Parenchyma Surgery in Autosomal Dominant Hyper-IgE Syndrome. Journal of Clinical Immunology, 2013, 33, 896-902.	3.8	39
74	Intermediate phenotypes in patients with autosomal dominant hyper-IgE syndrome caused by somatic mosaicism. Journal of Allergy and Clinical Immunology, 2013, 131, 1586-1593.	2.9	50
75	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.	2.9	222
76	MonoMAC Syndrome in a Patient With a GATA2 Mutation: Case Report and Review of the Literature. Clinical Infectious Diseases, 2013, 57, 697-699.	5.8	53
77	GATA2 haploinsufficiency caused by mutations in a conserved intronic element leads to MonoMAC syndrome. Blood, 2013, 121, 3830-3837.	1.4	209
78	Mutations in GATA2 cause human NK cell deficiency with specific loss of the CD56bright subset. Blood, 2013, 121, 2669-2677.	1.4	208
79	Autoimmune lymphoproliferative syndrome due to FAS mutations outside the signal-transducing death domain: molecular mechanisms and clinical penetrance. Genetics in Medicine, 2012, 14, 81-89.	2.4	41
80	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. Blood, 2012, 119, 1283-1291.	1.4	244
81	Adult-Onset Immunodeficiency in Thailand and Taiwan. New England Journal of Medicine, 2012, 367, 725-734.	27.0	431
82	A Novel STAT1 Mutation Associated with Disseminated Mycobacterial Disease. Journal of Clinical Immunology, 2012, 32, 681-689.	3.8	61
83	Cis-element mutated in GATA2-dependent immunodeficiency governs hematopoiesis and vascular integrity. Journal of Clinical Investigation, 2012, 122, 3692-3704.	8.2	162
84	Regulatory Mutations in GATA2 Associated with Aplastic Anemia. Blood, 2012, 120, 3488-3488.	1.4	20
85	Plasma metalloproteinase levels are dysregulated in signal transducer and activator of transcription 3 mutated hyper-IgE syndrome. Journal of Allergy and Clinical Immunology, 2011, 128, 1124-1127.	2.9	32
86	Mutations in GATA2 are associated with the autosomal dominant and sporadic monocytopenia and mycobacterial infection (MonoMAC) syndrome. Blood, 2011, 118, 2653-2655.	1.4	572
87	Successful allogeneic hematopoietic stem cell transplantation for GATA2 deficiency. Blood, 2011, 118, 3715-3720.	1.4	131
88	A Critical Role for STAT3 Transcription Factor Signaling in the Development and Maintenance of Human T Cell Memory. Immunity, 2011, 35, 806-818.	14.3	241
89	Myelodysplasia in autosomal dominant and sporadic monocytopenia immunodeficiency syndrome: diagnostic features and clinical implications. Haematologica, 2011, 96, 1221-1225.	3.5	97
90	Paucity of genotypeâ€“phenotype correlations in STAT3 mutation positive Hyper IgE Syndrome (HIES). Clinical Immunology, 2011, 139, 75-84.	3.2	39

#	Article	IF	Citations
91	Coronary Artery Abnormalities in Hyper-IgE Syndrome. Journal of Clinical Immunology, 2011, 31, 338-345.	3.8	64
92	Interleukin-12 Receptor Â1 Deficiency Predisposing to Disseminated Coccidioidomycosis. Clinical Infectious Diseases, 2011, 52, e99-e102.	5.8	87
93	Autosomal dominant and sporadic monocytopenia with susceptibility to mycobacteria, fungi, papillomaviruses, and myelodysplasia. Blood, 2010, 115, 1519-1529.	1.4	299
94	Anti-cytokine autoantibodies are associated with opportunistic infection in patients with thymic neoplasia. Blood, 2010, 116, 4848-4858.	1.4	134
95	Highly Variable Clinical Phenotypes of Hypomorphic RAG1 Mutations. Pediatrics, 2010, 126, e1248-e1252.	2.1	70
96	Mutations in STAT3 and diagnostic guidelines for hyper-IgE syndrome. Journal of Allergy and Clinical Immunology, 2010, 125, 424-432.e8.	2.9	247
97	Invasive fungal disease in autosomal-dominant hyper-IgE syndrome. Journal of Allergy and Clinical Immunology, 2010, 125, 1389-1390.	2.9	91
98	Coronary Abnormalities in Hyper-IgE Recurrent Infection Syndrome: Depiction at Coronary MDCT Angiography. American Journal of Roentgenology, 2009, 193, W478-W481.	2.2	16
99	Pulmonary nontuberculous mycobacterial infections in hyper-IgE syndrome. Journal of Allergy and Clinical Immunology, 2009, 124, 617-618.	2.9	31
100	Mutation analysis in primary immunodeficiency diseases: case studies. Current Opinion in Allergy and Clinical Immunology, 2009, 9, 517-524.	2.3	15
101	Impaired TH17 cell differentiation in subjects with autosomal dominant hyper-IgE syndrome. Nature, 2008, 452, 773-776.	27.8	1,046
102	Mutations causing severe combined immunodeficiency: detection with a custom resequencing microarray. Genetics in Medicine, 2008, 10, 575-585.	2.4	31
103	Reversion mutations in patients with leukocyte adhesion deficiency type-1 (LAD-1). Blood, 2008, 111, 209-218.	1.4	76
104	Gene therapy improves immune function in preadolescents with X-linked severe combined immunodeficiency. Blood, 2007, 110, 67-73.	1.4	97
105	STAT3Mutations in the Hyper-IgE Syndrome. New England Journal of Medicine, 2007, 357, 1608-1619.	27.0	1,098
106	Single nucleotide polymorphisms in the apoptosis receptor gene TNFRSF6. Molecular and Cellular Probes, 2006, 20, 21-26.	2.1	12
107	A novel IL2RG mutation associated with maternal T lymphocyte engraftment in a patient with severe combined immunodeficiency. Journal of Human Genetics, 2006, 51, 495-497.	2.3	13
108	Genetic alterations in caspase-10 may be causative or protective in autoimmune lymphoproliferative syndrome. Human Genetics, 2006, 119, 284-294.	3.8	68

#	Article	IF	Citations
109	Retroviral transduction of IL2RG into CD34+ cells from X-linked severe combined immunodeficiency patients permits human T- and B-cell development in sheep chimeras. Blood, 2002, 100, 72-79.	1.4	14
110	4 Primary immunodeficiency mutation databases. Advances in Genetics, 2001, 43, 103-188.	1.8	70
111	Efficient Detection of Thirty-Seven New IL2RG Mutations in Human X-Linked Severe Combined Immunodeficiency. Clinical Immunology, 2000, 95, 33-38.	3.2	31
112	Autoimmune Lymphoproliferative Syndrome with Defective Fas: Genotype Influences Penetrance. American Journal of Human Genetics, 1999, 64, 1002-1014.	6.2	198