Helen C Su

List of Publications by Year
in descending order

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135 papers	17,588 citations	26610 56 h-index	15716 125 g-index
143 all docs	143 docs citations	143 times ranked	21067 citing authors

#	Article	IF	Citations
1	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,983
2	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,749
3	Regulation of an ATG7-beclin 1 Program of Autophagic Cell Death by Caspase-8. Science, 2004, 304, 1500-1502.	6.0	1,197
4	Immune dysregulation in human subjects with heterozygous germline mutations in CTLA4. Science, 2014, 345, 1623-1627.	6.0	745
5	Combined Immunodeficiency Associated withDOCK8Mutations. New England Journal of Medicine, 2009, 361, 2046-2055.	13.9	655
6	Patients with LRBA deficiency show CTLA4 loss and immune dysregulation responsive to abatacept therapy. Science, 2015, 349, 436-440.	6.0	580
7	Second messenger role for Mg2+ revealed by human T-cell immunodeficiency. Nature, 2011, 475, 471-476.	13.7	465
8	Early-onset lymphoproliferation and autoimmunity caused by germline STAT3 gain-of-function mutations. Blood, 2015, 125, 591-599.	0.6	436
9	Revised diagnostic criteria and classification for the autoimmune lymphoproliferative syndrome (ALPS): report from the 2009 NIH International Workshop. Blood, 2010, 116, e35-e40.	0.6	405
10	Requirement for Caspase-8 in NF-ÂB Activation by Antigen Receptor. Science, 2005, 307, 1465-1468.	6.0	404
11	Human Inborn Errors of Immunity: 2022 Update on the Classification from the International Union of Immunological Societies Expert Committee. Journal of Clinical Immunology, 2022, 42, 1473-1507.	2.0	389
12	Autoantibodies neutralizing type I IFNs are present in ~4% of uninfected individuals over 70 years old and account for ~20% of COVID-19 deaths. Science Immunology, 2021, 6, .	5.6	357
13	DOCK8 Deficiency: Clinical and Immunological Phenotype and Treatment Options - a Review of 136 Patients. Journal of Clinical Immunology, 2015, 35, 189-198.	2.0	284
14	Mg ²⁺ Regulates Cytotoxic Functions of NK and CD8 T Cells in Chronic EBV Infection Through NKG2D. Science, 2013, 341, 186-191.	6.0	269
15	An immune-based biomarker signature is associated with mortality in COVID-19 patients. JCI Insight, 2021, 6, .	2.3	269
16	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. Science Immunology, 2021, 6, .	5.6	267
17	Heterozygous splice mutation in PIK3R1 causes human immunodeficiency with lymphoproliferation due to dominant activation of PI3K. Journal of Experimental Medicine, 2014, 211, 2537-2547.	4.2	249
18	Human genetic and immunological determinants of critical COVID-19 pneumonia. Nature, 2022, 603, 587-598.	13.7	216

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19	Jakinibs for the treatment of immune dysregulation in patients with gain-of-function signal transducer and activator of transcription 1 (STAT1) or STAT3 mutations. Journal of Allergy and Clinical Immunology, 2018, 142, 1665-1669.	1.5	196
20	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
21	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. Cell, 2020, 181, 1194-1199.	13.5	185
22	Preexisting autoantibodies to type I IFNs underlie critical COVID-19 pneumonia in patients with APS-1. Journal of Experimental Medicine, 2021, 218, .	4.2	185
23	GENETIC DISORDERS OF PROGRAMMED CELL DEATH IN THE IMMUNE SYSTEM. Annual Review of Immunology, 2006, 24, 321-352.	9.5	178
24	Mapping of a major genetic modifier of embryonic lethality in TGFÎ²1 knockout mice. Nature Genetics, 1997, 15, 207-211.	9.4	168
25	Congenital B cell lymphocytosis explained by novel germline CARD11 mutations. Journal of Experimental Medicine, 2012, 209, 2247-2261.	4.2	167
26	Time-resolved systems immunology reveals a late juncture linked to fatal COVID-19. Cell, 2021, 184, 1836-1857.e22.	13.5	167
27	The Ever-Increasing Array of Novel Inborn Errors of Immunity: an Interim Update by the IUIS Committee. Journal of Clinical Immunology, 2021, 41, 666-679.	2.0	165
28	DOCK8 regulates lymphocyte shape integrity for skin antiviral immunity. Journal of Experimental Medicine, 2014, 211, 2549-2566.	4.2	150
29	XMEN disease: a new primary immunodeficiency affecting Mg2+ regulation of immunity against Epstein-Barr virus. Blood, 2014, 123, 2148-2152.	0.6	147
30	Life-threatening influenza pneumonitis in a child with inherited IRF9 deficiency. Journal of Experimental Medicine, 2018, 215, 2567-2585.	4.2	146
31	Immunopathological signatures in multisystem inflammatory syndrome in children and pediatric COVID-19. Nature Medicine, 2022, 28, 1050-1062.	15.2	144
32	CD55 Deficiency, Early-Onset Protein-Losing Enteropathy, and Thrombosis. New England Journal of Medicine, 2017, 377, 52-61.	13.9	138
33	Human inborn errors of immunity: An expanding universe. Science Immunology, 2020, 5, .	5.6	138
34	Essential Role for Caspase-8 in Toll-like Receptors and NFÎºB Signaling. Journal of Biological Chemistry, 2007, 282, 7416-7423.	1.6	137
35	Combined immunodeficiency and Epstein-Barr virusâ€“induced B cell malignancy in humans with inherited CD70 deficiency. Journal of Experimental Medicine, 2017, 214, 91-106.	4.2	134
36	Auto-antibodies to type I IFNs can underlie adverse reactions to yellow fever live attenuated vaccine. Journal of Experimental Medicine, 2021, 218, .	4.2	130

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37	Cutaneous Manifestations of <emph type="ital">DOCK8</emph> Deficiency Syndrome. Archives of Dermatology, 2012, 148, 79.	1.7	129
38	Restimulation-induced apoptosis of T cells is impaired in patients with X-linked lymphoproliferative disease caused by SAP deficiency. Journal of Clinical Investigation, 2009, 119, 2976-89.	3.9	126
39	CHAI and LATAIE: new genetic diseases of CTLA-4 checkpoint insufficiency. Blood, 2016, 128, 1037-1042.	0.6	124
40	Dedicator of cytokinesis 8 (DOCK8) deficiency. Current Opinion in Allergy and Clinical Immunology, 2010, 10, 515-520.	1.1	119
41	Recurrent rhinovirus infections in a child with inherited MDA5 deficiency. Journal of Experimental Medicine, 2017, 214, 1949-1972.	4.2	117
42	Life-Threatening COVID-19: Defective Interferons Unleash Excessive Inflammation. Med, 2020, 1, 14-20.	2.2	110
43	The risk of COVID-19 death is much greater and age dependent with type I IFN autoantibodies. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, e2200413119.	3.3	110
44	DOCK8 is essential for Tâ€cell survival and the maintenance of CD8⁺ Tâ€cell memory. European Journal of Immunology, 2011, 41, 3423-3435.	1.6	105
45	Expanded skin virome in DOCK8-deficient patients. Nature Medicine, 2018, 24, 1815-1821.	15.2	104
46	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
47	Additional Diverse Findings Expand the Clinical Presentation of DOCK8 Deficiency. Journal of Clinical Immunology, 2012, 32, 698-708.	2.0	84
48	Somatic reversion in dedicator of cytokinesis 8 immunodeficiency modulates disease phenotype. Journal of Allergy and Clinical Immunology, 2014, 133, 1667-1675.	1.5	82
49	Dedicator of cytokinesis 8â€“deficient patients have aÂbreakdown in peripheral B-cell tolerance and defectiveÂregulatory T cells. Journal of Allergy and Clinical Immunology, 2014, 134, 1365-1374.	1.5	79
50	DOCK8 deficiency. Annals of the New York Academy of Sciences, 2011, 1246, 26-33.	1.8	74
51	Germline CARD11 Mutation in a Patient with Severe Congenital B Cell Lymphocytosis. Journal of Clinical Immunology, 2015, 35, 32-46.	2.0	74
52	Defective glycosylation and multisystem abnormalities characterize the primary immunodeficiency XMEN disease. Journal of Clinical Investigation, 2019, 130, 507-522.	3.9	74
53	Dedicator of cytokinesis 8â€“deficient CD4 + TÂcells are biased to a T H 2 effector fate at the expense of T H 1 and T H 17Âcells. Journal of Allergy and Clinical Immunology, 2017, 139, 933-949.	1.5	69
54	Neutralizing typeâ€Ⅰinterferon autoantibodies are associated with delayed viral clearance and intensive care unit admission in patients with COVIDâ€19. Immunology and Cell Biology, 2021, 99, 917-921.	1.0	69

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55	DOCK8 is critical for the survival and function of NKT cells. Blood, 2013, 122, 2052-2061.	0.6	68
56	Hematopoietic Stem Cell Transplantation as Treatment for Patients with DOCK8 Deficiency. Journal of Allergy and Clinical Immunology: in Practice, 2019, 7, 848-855.	2.0	67
57	Extended clinical and immunological phenotype and transplant outcome in CD27 and CD70 deficiency. Blood, 2020, 136, 2638-2655.	0.6	64
58	Novel PIK3CD mutations affecting N-terminal residues of p110Î´ cause activated PI3KÎ´ syndrome (APDS) in humans. Journal of Allergy and Clinical Immunology, 2017, 140, 1152-1156.e10.	1.5	62
59	Genetic, Clinical, and Laboratory Markers for DOCK8 Immunodeficiency Syndrome. Disease Markers, 2010, 29, 131-139.	0.6	59
60	Recessive inborn errors of type I IFN immunity in children with COVID-19 pneumonia. Journal of Experimental Medicine, 2022, 219, .	4.2	59
61	Evaluation of IFITM3 rs12252 Association With Severe Pediatric Influenza Infection. Journal of Infectious Diseases, 2017, 216, 14-21.	1.9	58
62	A rapid flow cytometric screening test for Xâ€linked lymphoproliferative disease due to XIAP deficiency. Cytometry Part B - Clinical Cytometry, 2009, 76B, 334-344.	0.7	57
63	Dual Proteolytic Pathways Govern Glycolysis and Immune Competence. Cell, 2014, 159, 1578-1590.	13.5	54
64	Insights into immunity from clinical and basic science studies of <scp>DOCK</scp>8 immunodeficiency syndrome. Immunological Reviews, 2019, 287, 9-19.	2.8	52
65	Genetic, clinical, and laboratory markers for DOCK8 immunodeficiency syndrome. Disease Markers, 2010, 29, 131-9.	0.6	51
66	Aberrant tRNA processing causes an autoinflammatory syndrome responsive to TNF inhibitors. Annals of the Rheumatic Diseases, 2018, 77, 612-619.	0.5	49
67	Severe COVID-19 in the young and healthy: monogenic inborn errors of immunity?. Nature Reviews Immunology, 2020, 20, 455-456.	10.6	47
68	Matched Related and Unrelated Donor Hematopoietic Stem Cell Transplantation for DOCK8 Deficiency. Biology of Blood and Marrow Transplantation, 2015, 21, 1037-1045.	2.0	45
69	Mild B-cell lymphocytosis in patients with a CARD11 C49Y mutation. Journal of Allergy and Clinical Immunology, 2015, 136, 819-821.e1.	1.5	44
70	Human Plasma-like Medium Improves T Lymphocyte Activation. IScience, 2020, 23, 100759.	1.9	44
71	Vaccine strain varicella-zoster virusâ€“induced central nervous system vasculopathy as the presenting feature of DOCK8 deficiency. Journal of Allergy and Clinical Immunology, 2014, 133, 1225-1227.	1.5	42
72	A global effort to dissect the human genetic basis of resistance to SARS-CoV-2 infection. Nature Immunology, 2022, 23, 159-164.	7.0	41

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73	Haploidentical Related Donor Hematopoietic Stem Cell Transplantation for Dedicator-of-Cytokinesis 8 Deficiency Using Post-Transplantation Cyclophosphamide. Biology of Blood and Marrow Transplantation, 2017, 23, 980-990.	2.0	39
74	Harnessing Type I IFN Immunity Against SARS-CoV-2 with Early Administration of IFN-Î². Journal of Clinical Immunology, 2021, 41, 1425-1442.	2.0	39
75	Early Cytokine Responses to Viral Infections and Their Roles in Shaping Endogenous Cellular Immunity. Advances in Experimental Medicine and Biology, 1998, 452, 143-149.	0.8	34
76	Function and Regulation of Natural Killer (NK) Cells during Viral Infections: Characterization of Responsesin Vivo. Methods, 1996, 9, 379-393.	1.9	33
77	Hyperimmunoglobulin E syndromes in pediatrics. Current Opinion in Pediatrics, 2011, 23, 653-658.	1.0	33
78	Inhibition of HECT E3 ligases as potential therapy for COVID-19. Cell Death and Disease, 2021, 12, 310.	2.7	33
79	Genetic Defects of Apoptosis and Primary Immunodeficiency. Immunology and Allergy Clinics of North America, 2008, 28, 329-351.	0.7	32
80	Biochemically deleterious human NFKB1 variants underlie an autosomal dominant form of common variable immunodeficiency. Journal of Experimental Medicine, 2021, 218, .	4.2	32
81	Recent Advances in DOCK8 Immunodeficiency Syndrome. Journal of Clinical Immunology, 2016, 36, 441-449.	2.0	31
82	RELA haploinsufficiency in CD4 lymphoproliferative disease with autoimmune cytopenias. Journal of Allergy and Clinical Immunology, 2018, 141, 1507-1510.e8.	1.5	31
83	Food allergies can persist after myeloablative hematopoietic stem cell transplantation in dedicator of cytokinesis 8â€“deficient patients. Journal of Allergy and Clinical Immunology, 2016, 137, 1895-1898.e5.	1.5	30
84	Genomics is rapidly advancing precision medicine for immunological disorders. Nature Immunology, 2015, 16, 1001-1004.	7.0	29
85	Evaluation of Recipients of Positive and Negative Secondary Findings Evaluations in a Hybrid CLIA-Research Sequencing Pilot. American Journal of Human Genetics, 2018, 103, 358-366.	2.6	29
86	Temporal Dynamics of Antiâ€“Type 1 Interferon Autoantibodies in Patients With Coronavirus Disease 2019. Clinical Infectious Diseases, 2022, 75, e1192-e1194.	2.9	26
87	A Unique Heterozygous CARD11 Mutation Combines Pathogenic Features of Both Gain- and Loss-of-Function Patients in a Four-Generation Family. Frontiers in Immunology, 2018, 9, 2944.	2.2	24
88	Multiplexed Proteomic Analysis for Diagnosis and Screening of Five Primary Immunodeficiency Disorders From Dried Blood Spots. Frontiers in Immunology, 2020, 11, 464.	2.2	24
89	Hematopoietic stem cell transplant effectively rescues lymphocyte differentiation and function in DOCK8-deficient patients. JCI Insight, 2019, 4, .	2.3	23
90	GIMAP5 maintains liver endothelial cell homeostasis and prevents portal hypertension. Journal of Experimental Medicine, 2021, 218, .	4.2	22

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91	Impaired Control of Epsteinâ€“Barr Virus Infection in B-Cell Expansion with NF-Î²B and T-Cell Anergy Disease. Frontiers in Immunology, 2018, 9, 198.	2.2	21
92	F-BAR domain only protein 1 (FCHO1) deficiency is a novel cause of combined immune deficiency in human subjects. Journal of Allergy and Clinical Immunology, 2019, 143, 2317-2321.e12.	1.5	21
93	Migration-induced cell shattering due to DOCK8 deficiency causes a type 2â€“biased helper T cell response. Nature Immunology, 2020, 21, 1528-1539.	7.0	21
94	Respiratory viral infections in otherwise healthy humans with inherited IRF7 deficiency. Journal of Experimental Medicine, 2022, 219, .	4.2	21
95	Unrelated Hematopoietic Cell Transplantation in a Patient with Combined Immunodeficiency with Granulomatous Disease and Autoimmunity Secondary to RAG Deficiency. Journal of Clinical Immunology, 2016, 36, 725-732.	2.0	19
96	Evolution and longâ€term outcomes of combined immunodeficiency due to CARMIL2 deficiency. Allergy: European Journal of Allergy and Clinical Immunology, 2022, 77, 1004-1019.	2.7	19
97	Haploinsufficiency of immune checkpoint receptor CTLA4 induces a distinct neuroinflammatory disorder. Journal of Clinical Investigation, 2020, 130, 5551-5561.	3.9	18
98	Autoantibodies Against Proteins Previously Associated With Autoimmunity in Adult and Pediatric Patients With COVID-19 and Children With MIS-C. Frontiers in Immunology, 2022, 13, 841126.	2.2	18
99	IFIH1 loss-of-function variants contribute to very early-onset inflammatory bowel disease. Human Genetics, 2021, 140, 1299-1312.	1.8	17
100	Haploidentical related donor hematopoietic stem cell transplantation withÂpost-transplantation cyclophosphamide for DOCK8 deficiency. Journal of Allergy and Clinical Immunology: in Practice, 2016, 4, 1239-1242.e1.	2.0	16
101	New immunodeficiency syndromes that help us understand the IFN-mediated antiviral immune response. Current Opinion in Pediatrics, 2019, 31, 815-820.	1.0	16
102	DOCK 8 Deficiency, EBV+ Lymphomatoid Granulomatosis, and Intrafamilial Variation in Presentation. Frontiers in Pediatrics, 2017, 5, 38.	0.9	15
103	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.	1.5	14
104	The Growing Spectrum of Human Diseases Caused by InheritedCDC42 Mutations. Journal of Clinical Immunology, 2020, 40, 551-553.	2.0	14
105	A Double-Blind, Placebo-Controlled, Crossover Study of Magnesium Supplementation in Patients with XMEN Disease. Journal of Clinical Immunology, 2022, 42, 108-118.	2.0	14
106	PI3KÎ´ coordinates transcriptional, chromatin, and metabolic changes to promote effector CD8+ TÂcells at the expense of central memory. Cell Reports, 2021, 37, 109804.	2.9	13
107	Association of rare predicted loss-of-function variants of influenza-related type I IFN genes with critical COVID-19 pneumonia. Journal of Clinical Investigation, 2021, 131, .	3.9	12
108	Inborn errors of TLR3- or MDA5-dependent type I IFN immunity in children with enterovirus rhombencephalitis. Journal of Experimental Medicine, 2021, 218, .	4.2	12

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109	Genetic determinants of host immunity against human rhinovirus infections. Human Genetics, 2020, 139, 949-959.	1.8	11
110	Compound Heterozygous DOCK8 Mutations in a Patient with B Lymphoblastic Leukemia and EBV-Associated Diffuse Large B Cell Lymphoma. Journal of Clinical Immunology, 2019, 39, 592-595.	2.0	10
111	Another Fork in the Road. Immunity, 2004, 21, 133-134.	6.6	9
112	Combined Immunodeficiency Associated with DOCK8 Mutations and Related Immunodeficiencies. Disease Markers, 2010, 29, 121-122.	0.6	7
113	SARS-CoV-2 infection in dialysis and kidney transplant patients: immunological and serological response. Journal of Nephrology, 2022, , 1.	0.9	7
114	Case Report: Fatal Complications of BK Virus-Hemorrhagic Cystitis and Severe Cytokine Release Syndrome Following BK Virus-Specific T-Cells. Frontiers in Immunology, 2021, 12, 801281.	2.2	7
115	Evaluation of Mannose Binding Lectin Gene Variants in Pediatric Influenza Virus-Related Critical Illness. Frontiers in Immunology, 2019, 10, 1005.	2.2	6
116	Cryptosporidium infection in dedicator of cytokinesis 8 (DOCK 8) deficiency. Journal of Allergy and Clinical Immunology: in Practice, 2020, 8, 3663-3666.e1.	2.0	6
117	Clinical utility gene card for: X-linked immunodeficiency with magnesium defect, Epsteinâ€“Barr virus infection, and neoplasia (XMEN). European Journal of Human Genetics, 2015, 23, 889-889.	1.4	5
118	Hyper-IgE Syndrome due to an Elusive Novel Intronic Homozygous Variant in DOCK8. Journal of Clinical Immunology, 2022, 42, 119-129.	2.0	4
119	Exome sequencing study in a clinical research setting finds general acceptance of study returning secondary genomic findings with little decisional conflict. Journal of Genetic Counseling, 2021, 30, 766-773.	0.9	4
120	Combined Immune Deficiencies. , 2014, , 143-169.		3
121	The Influence of Immune Immaturity on Outcome After Virus Infections. Journal of Allergy and Clinical Immunology: in Practice, 2021, 9, 641-650.	2.0	3
122	Lessons from autoimmune lymphoproliferative syndrome. Drug Discovery Today Disease Mechanisms, 2005, 2, 495-502.	0.8	2
123	Combined immune deficiencies (CIDs). , 2020, , 207-268.		2
124	450. Type I Interferon Autoantibodies Are Detected in Those with Critical COVID-19, Including a Young Female Patient. Open Forum Infectious Diseases, 2021, 8, S325-S326.	0.4	2
125	The technological transformation of patient-driven human immunology research. Immunologic Research, 2009, 43, 167-171.	1.3	1
126	Studying human immunodeficiencies in humans: advances in fundamental concepts and therapeutic interventions. F1000Research, 2017, 6, 318.	0.8	1

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127	A Novel Gain-Of-Function Mutation In The CARD Domain Of CARD11 (C49Y) Results In Benta Disease. Blood, 2013, 122, 3485-3485.	0.6	1
128	Programmed cell death in lymphocytes. , 2008, , 225-234.		0
129	Chediak-Steinbrinck-Higashi Syndrome. , 2009, , 314-314.		0
130	Designs for Massively Parallel Sequencing Approaches to Identify Causal Mutations in Human Immune Disorders. Methods in Molecular Biology, 2013, 979, 175-187.	0.4	0
131	Monogenic Autoimmune Lymphoproliferative Syndromes. , 2014, , 695-709.		0
132	Programmed cell death in lymphocytes and associated disorders. , 2013, , 172-180.		0
133	DOCK8 regulates lymphocyte shape integrity for skin antiviral immunity. Journal of Cell Biology, 2014, 207, 2075OIA223.	2.3	0
134	Editorial overview: Human inborn errors of immunity to infection. Current Opinion in Immunology, 2021, 72, iii-v.	2.4	0
135	126. Magnitude and Dynamics of the T-Cell Response to SARS-CoV-2 Infection and Vaccination. Open Forum Infectious Diseases, 2021, 8, S77-S77.	0.4	0