List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Evolution and longâ€ŧerm outcomes of combined immunodeficiency due to CARMIL2 deficiency. Allergy: European Journal of Allergy and Clinical Immunology, 2022, 77, 1004-1019.	2.7	19
2	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
3	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
4	Hyper-IgE Syndrome due to an Elusive Novel Intronic Homozygous Variant in DOCK8. Journal of Clinical Immunology, 2022, 42, 119-129.	2.0	4
5	Human genetic and immunological determinants of critical COVID-19 pneumonia. Nature, 2022, 603, 587-598.	13.7	216
6	SARS-CoV-2 infection in dialysis and kidney transplant patients: immunological and serological response. Journal of Nephrology, 2022, , 1.	0.9	7
7	Immunopathological signatures in multisystem inflammatory syndrome in children and pediatric COVID-19. Nature Medicine, 2022, 28, 1050-1062.	15.2	144
8	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
9	Temporal Dynamics of Anti–Type 1 Interferon Autoantibodies in Patients With Coronavirus Disease 2019. Clinical Infectious Diseases, 2022, 75, e1192-e1194.	2.9	26
10	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
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	Respiratory viral infections in otherwise healthy humans with inherited IRF7 deficiency. Journal of Experimental Medicine, 2022, 219, .	4.2	21
13	Recessive inborn errors of type I IFN immunity in children with COVID-19 pneumonia. Journal of Experimental Medicine, 2022, 219, .	4.2	59
14	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
15	An immune-based biomarker signature is associated with mortality in COVID-19 patients. JCI Insight, 2021, 6, .	2.3	269
16	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
17	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
18	Inhibition of HECT E3 ligases as potential therapy for COVID-19. Cell Death and Disease, 2021, 12, 310.	2.7	33

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19	SARS-CoV-2–related MIS-C: A key to the viral and genetic causes of Kawasaki disease?. Journal of Experimental Medicine, 2021, 218, .	4.2	100
20	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
21	Time-resolved systems immunology reveals a late juncture linked to fatal COVID-19. Cell, 2021, 184, 1836-1857.e22.	13.5	167
22	GIMAP5 maintains liver endothelial cell homeostasis and prevents portal hypertension. Journal of Experimental Medicine, 2021, 218, .	4.2	22
23	IFIH1 loss-of-function variants contribute to very early-onset inflammatory bowel disease. Human Genetics, 2021, 140, 1299-1312.	1.8	17
24	Harnessing Type I IFN Immunity Against SARS-CoV-2 with Early Administration of IFN-Î ² . Journal of Clinical Immunology, 2021, 41, 1425-1442.	2.0	39
25	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
26	Neutralizing typeâ€l interferon 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
27	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
28	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
29	Biochemically deleterious human <i>NFKB1</i> variants underlie an autosomal dominant form of common variable immunodeficiency. Journal of Experimental Medicine, 2021, 218, .	4.2	32
30	PI3Kδ 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
31	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
32	Editorial overview: Human inborn errors of immunity to infection. Current Opinion in Immunology, 2021, 72, iii-v.	2.4	0
33	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
34	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
35	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
36	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

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37	Human Plasma-like Medium Improves T Lymphocyte Activation. IScience, 2020, 23, 100759.	1.9	44
38	Migration-induced cell shattering due to DOCK8 deficiency causes a type 2–biased helper T cell response. Nature Immunology, 2020, 21, 1528-1539.	7.0	21
39	Life-Threatening COVID-19: Defective Interferons Unleash Excessive Inflammation. Med, 2020, 1, 14-20.	2.2	110
40	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,749
41	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,983
42	The Growing Spectrum of Human Diseases Caused by InheritedCDC42 Mutations. Journal of Clinical Immunology, 2020, 40, 551-553.	2.0	14
43	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. Cell, 2020, 181, 1194-1199.	13.5	185
44	Severe COVID-19 in the young and healthy: monogenic inborn errors of immunity?. Nature Reviews Immunology, 2020, 20, 455-456.	10.6	47
45	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
46	Extended clinical and immunological phenotype and transplant outcome in CD27 and CD70 deficiency. Blood, 2020, 136, 2638-2655.	0.6	64
47	Human inborn errors of immunity: An expanding universe. Science Immunology, 2020, 5, .	5.6	138
48	Combined immune deficiencies (CIDs). , 2020, , 207-268.		2
49	Genetic determinants of host immunity against human rhinovirus infections. Human Genetics, 2020, 139, 949-959.	1.8	11
50	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
51	Haploinsufficiency of immune checkpoint receptor CTLA4 induces a distinct neuroinflammatory disorder. Journal of Clinical Investigation, 2020, 130, 5551-5561.	3.9	18
52	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
53	Evaluation of Mannose Binding Lectin Gene Variants in Pediatric Influenza Virus-Related Critical Illness. Frontiers in Immunology, 2019, 10, 1005.	2.2	6
54	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

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55	New immunodeficiency syndromes that help us understand the IFN-mediated antiviral immune response. Current Opinion in Pediatrics, 2019, 31, 815-820.	1.0	16
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	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
58	Hematopoietic stem cell transplant effectively rescues lymphocyte differentiation and function in DOCK8-deficient patients. JCI Insight, 2019, 4, .	2.3	23
59	Defective glycosylation and multisystem abnormalities characterize the primary immunodeficiency XMEN disease. Journal of Clinical Investigation, 2019, 130, 507-522.	3.9	74
60	Aberrant tRNA processing causes an autoinflammatory syndrome responsive to TNF inhibitors. Annals of the Rheumatic Diseases, 2018, 77, 612-619.	0.5	49
61	RELA haploinsufficiency in CD4 lymphoproliferative disease with autoimmune cytopenias. Journal of Allergy and Clinical Immunology, 2018, 141, 1507-1510.e8.	1.5	31
62	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
63	Expanded skin virome in DOCK8-deficient patients. Nature Medicine, 2018, 24, 1815-1821.	15.2	104
64	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
65	Impaired Control of Epstein–Barr Virus Infection in B-Cell Expansion with NF-κB and T-Cell Anergy Disease. Frontiers in Immunology, 2018, 9, 198.	2.2	21
66	Life-threatening influenza pneumonitis in a child with inherited IRF9 deficiency. Journal of Experimental Medicine, 2018, 215, 2567-2585.	4.2	146
67	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
68	Novel PIK3CD mutations affecting N-terminal residues of p110δ cause activated PI3Kδ syndrome (APDS) in humans. Journal of Allergy and Clinical Immunology, 2017, 140, 1152-1156.e10.	1.5	62
69	Evaluation of IFITM3 rs12252 Association With Severe Pediatric Influenza Infection. Journal of Infectious Diseases, 2017, 216, 14-21.	1.9	58
70	Recurrent rhinovirus infections in a child with inherited MDA5 deficiency. Journal of Experimental Medicine, 2017, 214, 1949-1972.	4.2	117
71	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
72	Combined immunodeficiency and Epstein-Barr virus–induced B cell malignancy in humans with inherited CD70 deficiency. Journal of Experimental Medicine, 2017, 214, 91-106.	4.2	134

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73	CD55 Deficiency, Early-Onset Protein-Losing Enteropathy, and Thrombosis. New England Journal of Medicine, 2017, 377, 52-61.	13.9	138
74	Dedicator of cytokinesis 8–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
75	DOCK 8 Deficiency, EBV+ Lymphomatoid Granulomatosis, and Intrafamilial Variation in Presentation. Frontiers in Pediatrics, 2017, 5, 38.	0.9	15
76	Studying human immunodeficiencies in humans: advances in fundamental concepts and therapeutic interventions. F1000Research, 2017, 6, 318.	0.8	1
77	Food allergies can persist after myeloablative hematopoietic stem cell transplantation in dedicator of cytokinesis 8–deficient patients. Journal of Allergy and Clinical Immunology, 2016, 137, 1895-1898.e5.	1.5	30
78	Recent Advances in DOCK8 Immunodeficiency Syndrome. Journal of Clinical Immunology, 2016, 36, 441-449.	2.0	31
79	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
80	CHAI and LATAIE: new genetic diseases of CTLA-4 checkpoint insufficiency. Blood, 2016, 128, 1037-1042.	0.6	124
81	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
82	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
83	Early-onset lymphoproliferation and autoimmunity caused by germline STAT3 gain-of-function mutations. Blood, 2015, 125, 591-599.	0.6	436
84	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
85	Germline CARD11 Mutation in a Patient with Severe Congenital B Cell Lymphocytosis. Journal of Clinical Immunology, 2015, 35, 32-46.	2.0	74
86	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
87	Clinical utility gene card for: X-linked immunodeficiency with magnesium defect, Epstein–Barr virus infection, and neoplasia (XMEN). European Journal of Human Genetics, 2015, 23, 889-889.	1.4	5
88	Patients with LRBA deficiency show CTLA4 loss and immune dysregulation responsive to abatacept therapy. Science, 2015, 349, 436-440.	6.0	580
89	Genomics is rapidly advancing precision medicine for immunological disorders. Nature Immunology, 2015, 16, 1001-1004.	7.0	29
90	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

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91	Heterozygous splice mutation in <i>PIK3R1</i> causes human immunodeficiency with lymphoproliferation due to dominant activation of PI3K. Journal of Experimental Medicine, 2014, 211, 2537-2547.	4.2	249
92	Combined Immune Deficiencies. , 2014, , 143-169.		3
93	Dual Proteolytic Pathways Govern Glycolysis and Immune Competence. Cell, 2014, 159, 1578-1590.	13.5	54
94	DOCK8 regulates lymphocyte shape integrity for skin antiviral immunity. Journal of Experimental Medicine, 2014, 211, 2549-2566.	4.2	150
95	Dedicator of cytokinesis 8–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
96	XMEN disease: a new primary immunodeficiency affecting Mg2+ regulation of immunity against Epstein-Barr virus. Blood, 2014, 123, 2148-2152.	0.6	147
97	Vaccine strain varicella-zoster virus–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
98	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
99	Monogenic Autoimmune Lymphoproliferative Syndromes. , 2014, , 695-709.		0
100	Immune dysregulation in human subjects with heterozygous germline mutations in <i>CTLA4</i> . Science, 2014, 345, 1623-1627.	6.0	745
101	Somatic reversion in dedicator of cytokinesis 8 immunodeficiency modulates disease phenotype. Journal of Allergy and Clinical Immunology, 2014, 133, 1667-1675.	1.5	82
102	DOCK8 regulates lymphocyte shape integrity for skin antiviral immunity. Journal of Cell Biology, 2014, 207, 2075OIA223.	2.3	0
103	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
104	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
105	DOCK8 is critical for the survival and function of NKT cells. Blood, 2013, 122, 2052-2061.	0.6	68
106	Programmed cell death in lymphocytes and associated disorders. , 2013, , 172-180.		0
107	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
108	Congenital B cell lymphocytosis explained by novel germline <i>CARD11</i> mutations. Journal of Experimental Medicine, 2012, 209, 2247-2261.	4.2	167

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109	Cutaneous Manifestations of <emph type="ital">DOCK8</emph> Deficiency Syndrome. Archives of Dermatology, 2012, 148, 79.	1.7	129
110	Additional Diverse Findings Expand the Clinical Presentation of DOCK8 Deficiency. Journal of Clinical Immunology, 2012, 32, 698-708.	2.0	84
111	Second messenger role for Mg2+ revealed by human T-cell immunodeficiency. Nature, 2011, 475, 471-476.	13.7	465
112	DOCK8 deficiency. Annals of the New York Academy of Sciences, 2011, 1246, 26-33.	1.8	74
113	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
114	Hyperimmunoglobulin E syndromes in pediatrics. Current Opinion in Pediatrics, 2011, 23, 653-658.	1.0	33
115	Dedicator of cytokinesis 8 (DOCK8) deficiency. Current Opinion in Allergy and Clinical Immunology, 2010, 10, 515-520.	1.1	119
116	Combined Immunodeficiency Associated with DOCK8 Mutations and Related Immunodeficiencies. Disease Markers, 2010, 29, 121-122.	0.6	7
117	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
118	Genetic, Clinical, and Laboratory Markers for DOCK8 Immunodeficiency Syndrome. Disease Markers, 2010, 29, 131-139.	0.6	59
119	Genetic, clinical, and laboratory markers for DOCK8 immunodeficiency syndrome. Disease Markers, 2010, 29, 131-9.	0.6	51
120	Combined Immunodeficiency Associated with <i>DOCK8</i> Mutations. New England Journal of Medicine, 2009, 361, 2046-2055.	13.9	655
121	A rapid flow cytometric screening test for Xâ€ŀinked lymphoproliferative disease due to XIAP deficiency. Cytometry Part B - Clinical Cytometry, 2009, 76B, 334-344.	0.7	57
122	The technological transformation of patient-driven human immunology research. Immunologic Research, 2009, 43, 167-171.	1.3	1
123	Chediak-Steinbrinck-Higashi Syndrome. , 2009, , 314-314.		0
124	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
125	Genetic Defects of Apoptosis and Primary Immunodeficiency. Immunology and Allergy Clinics of North America, 2008, 28, 329-351.	0.7	32
126	Programmed cell death in lymphocytes. , 2008, , 225-234.		0

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127	Essential Role for Caspase-8 in Toll-like Receptors and NFκB Signaling. Journal of Biological Chemistry, 2007, 282, 7416-7423.	1.6	137
128	GENETIC DISORDERS OF PROGRAMMED CELL DEATH IN THE IMMUNE SYSTEM. Annual Review of Immunology, 2006, 24, 321-352.	9.5	178
129	Requirement for Caspase-8 in NF-ÂB Activation by Antigen Receptor. Science, 2005, 307, 1465-1468.	6.0	404
130	Lessons from autoimmune lymphoproliferative syndrome. Drug Discovery Today Disease Mechanisms, 2005, 2, 495-502.	0.8	2
131	Another Fork in the Road. Immunity, 2004, 21, 133-134.	6.6	9
132	Regulation of an ATG7-beclin 1 Program of Autophagic Cell Death by Caspase-8. Science, 2004, 304, 1500-1502.	6.0	1,197
133	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
134	Mapping of a major genetic modifier of embryonic lethality in TGFβ1 knockout mice. Nature Genetics, 1997, 15, 207-211.	9.4	168
135	Function and Regulation of Natural Killer (NK) Cells during Viral Infections: Characterization of Responsesin Vivo. Methods, 1996, 9, 379-393.	1.9	33