

Luigi Daniele Notarangelo

List of Publications by Year in descending order

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Version: 2024-02-01

643
papers

51,561
citations

1530

106
h-index

2375

198
g-index

670
all docs

670
docs citations

670
times ranked

39393
citing authors

#	ARTICLE	IF	CITATIONS
1	International retrospective study of allogeneic hematopoietic cell transplantation for activated PI3K-delta syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2022, 149, 410-421.e7.	1.5	34
2	Nodular regenerative hyperplasia in X-linked agammaglobulinemia: An underestimated and severe complication. <i>Journal of Allergy and Clinical Immunology</i> , 2022, 149, 400-409.e3.	1.5	8
3	IFN γ R1 deficiency presenting with visceral leishmaniasis and Mycobacterium Avium infections mimicking HLH. <i>Pediatric Allergy and Immunology</i> , 2022, 33, .	1.1	3
4	Poor T-cell receptor γ repertoire diversity early posttransplant for severe combined immunodeficiency predicts failure of immune reconstitution. <i>Journal of Allergy and Clinical Immunology</i> , 2022, 149, 1113-1119.	1.5	8
5	Donor source and posttransplantation cyclophosphamide influence outcome in allogeneic stem cell transplantation for GATA2 deficiency. <i>British Journal of Haematology</i> , 2022, 196, 169-178.	1.2	18
6	Congenital and acquired defects of immunity: An ever-evolving story. <i>Pediatric Allergy and Immunology</i> , 2022, 33, 61-64.	1.1	2
7	Human genetic and immunological determinants of critical COVID-19 pneumonia. <i>Nature</i> , 2022, 603, 587-598.	13.7	216
8	SARS-CoV-2 infection in dialysis and kidney transplant patients: immunological and serological response. <i>Journal of Nephrology</i> , 2022, , 1.	0.9	7
9	Chronic Granulomatous Disease With Inflammatory Bowel Disease: Clinical Presentation, Treatment, and Outcomes From the USIDNET Registry. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2022, 10, 1325-1333.e5.	2.0	11
10	Morbidity, Mortality, and Therapeutics in Combined Immunodeficiency: Data from the USIDNET Registry. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2022, , .	2.0	0
11	Immunopathological signatures in multisystem inflammatory syndrome in children and pediatric COVID-19. <i>Nature Medicine</i> , 2022, 28, 1050-1062.	15.2	144
12	Hematopoietic Cell Transplantation in 240 Patients with Chronic Granulomatous Disease: A PIDTC Report. <i>Transplantation and Cellular Therapy</i> , 2022, 28, S101-S103.	0.6	0
13	A Primary Immune Deficiency Treatment Consortium (PIDTC) Study of Chronic and Late Onset Medical Complications after Initial Hematopoietic Cell Transplantation (HCT) for Severe Combined Immunodeficiency Disease (SCID). <i>Transplantation and Cellular Therapy</i> , 2022, 28, S339-S340.	0.6	0
14	OPO19: A tertiary care clinical sequencing program for patients with suspected immune defects: Results from the first 1000 families. <i>Genetics in Medicine</i> , 2022, 24, S350.	1.1	0
15	Case Report: Severe Rhabdomyolysis and Multiorgan Failure After ChAdOx1 nCoV-19 Vaccination. <i>Frontiers in Immunology</i> , 2022, 13, 845496.	2.2	13
16	Sars-Cov-2 Infection and Vaccination in Recipients of Reduced-Intensity Conditioning, Posttransplantation Cyclophosphamide (PTCy)-Based Allogeneic Hematopoietic Cell Transplantation (HCT). <i>Transplantation and Cellular Therapy</i> , 2022, 28, S350-S351.	0.6	0
17	Autoantibodies Against Proteins Previously Associated With Autoimmunity in Adult and Pediatric Patients With COVID-19 and Children With MIS-C. <i>Frontiers in Immunology</i> , 2022, 13, 841126.	2.2	18
18	Outcomes after Hematopoietic Cell Transplant and Gene Therapy for Adenosine Deaminase (ADA) Severe Combined Immune Deficiency: A Combined Analysis from the Primary Immune Deficiency Treatment Consortium (PIDTC) 6901 and 6902 Studies. <i>Transplantation and Cellular Therapy</i> , 2022, 28, S100-S101.	0.6	0

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19	Rubella Virus-associated Granulomas in Immunocompetent Adults—Possible Implications. <i>JAMA Dermatology</i> , 2022, .	2.0	5
20	Temporal Dynamics of Anti-Type 1 Interferon Autoantibodies in Patients With Coronavirus Disease 2019. <i>Clinical Infectious Diseases</i> , 2022, 75, e1192-e1194.	2.9	26
21	Granulocyte Transfusions in Patients with Chronic Granulomatous Disease Undergoing Hematopoietic Cell Transplantation or Gene Therapy. <i>Journal of Clinical Immunology</i> , 2022, 42, 1026-1035.	2.0	3
22	The risk of COVID-19 death is much greater and age dependent with type I IFN autoantibodies. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, e2200413119.	3.3	110
23	Human OTULIN haploinsufficiency impairs cell-intrinsic immunity to staphylococcal α -toxin. <i>Science</i> , 2022, 376, eabm6380.	6.0	25
24	Purine nucleoside phosphorylase deficiency induces p53-mediated intrinsic apoptosis in human induced pluripotent stem cell-derived neurons. <i>Scientific Reports</i> , 2022, 12, .	1.6	3
25	Outcomes following treatment for ADA-deficient severe combined immunodeficiency: a report from the PIDTC. <i>Blood</i> , 2022, 140, 685-705.	0.6	26
26	Respiratory viral infections in otherwise healthy humans with inherited IRF7 deficiency. <i>Journal of Experimental Medicine</i> , 2022, 219, .	4.2	21
27	Recessive inborn errors of type I IFN immunity in children with COVID-19 pneumonia. <i>Journal of Experimental Medicine</i> , 2022, 219, .	4.2	59
28	Lentivector cryptic splicing mediates increase in CD34+ clones expressing truncated HMGA2 in human X-linked severe combined immunodeficiency. <i>Nature Communications</i> , 2022, 13, .	5.8	19
29	Clonal hematopoiesis is not significantly associated with COVID-19 disease severity. <i>Blood</i> , 2022, 140, 1650-1655.	0.6	10
30	POLD1 Deficiency Reveals a Role for POLD1 in DNA Repair and T and B Cell Development. <i>Journal of Clinical Immunology</i> , 2021, 41, 270-273.	2.0	10
31	Efficacy and safety of anti-CD45 saporin as conditioning agent for RAG deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 147, 309-320.e6.	1.5	27
32	Infections in Infants with SCID: Isolation, Infection Screening, and Prophylaxis in PIDTC Centers. <i>Journal of Clinical Immunology</i> , 2021, 41, 38-50.	2.0	36
33	Novel Compound Heterozygous Mutations in ZAP70 Leading to a SCID Phenotype with Normal Downstream In vitro Signaling. <i>Journal of Clinical Immunology</i> , 2021, 41, 470-472.	2.0	2
34	Complete Absence of CD3 β Protein Expression Is Responsible for Combined Immunodeficiency with Autoimmunity Rather than SCID. <i>Journal of Clinical Immunology</i> , 2021, 41, 482-485.	2.0	1
35	Reduction in the rate and improvement in the prognosis of COVID-19 in haematological patients over time. <i>Leukemia</i> , 2021, 35, 632-634.	3.3	3
36	Gut Microbiota-Host Interactions in Inborn Errors of Immunity. <i>International Journal of Molecular Sciences</i> , 2021, 22, 1416.	1.8	18

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37	Infectious Complications Predict Premature CD8+ T-cell Senescence in CD40 Ligand-Deficient Patients. <i>Journal of Clinical Immunology</i> , 2021, 41, 795-806.	2.0	2
38	TLR3 controls constitutive IFN- γ antiviral immunity in human fibroblasts and cortical neurons. <i>Journal of Clinical Investigation</i> , 2021, 131, .	3.9	64
39	Lost in Translation: Lack of CD4 Expression due to a Novel Genetic Defect. <i>Journal of Infectious Diseases</i> , 2021, 223, 645-654.	1.9	10
40	Aberrant type 1 immunity drives susceptibility to mucosal fungal infections. <i>Science</i> , 2021, 371, .	6.0	84
41	An immune-based biomarker signature is associated with mortality in COVID-19 patients. <i>JCI Insight</i> , 2021, 6, .	2.3	269
42	An appraisal of the Wilson & Jungner criteria in the context of genomic-based newborn screening for inborn errors of immunity. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 147, 428-438.	1.5	19
43	Are we diagnosing too late? RAG deficiency in young adults with end organ damage. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 147, AB69.	1.5	0
44	Auto-antibodies to type I IFNs can underlie adverse reactions to yellow fever live attenuated vaccine. <i>Journal of Experimental Medicine</i> , 2021, 218, .	4.2	130
45	RAG deficiencies: Recent advances in disease pathogenesis and novel therapeutic approaches. <i>European Journal of Immunology</i> , 2021, 51, 1028-1038.	1.6	22
46	Inhibition of HECT E3 ligases as potential therapy for COVID-19. <i>Cell Death and Disease</i> , 2021, 12, 310.	2.7	33
47	Gene Editing Rescues In vitro T Cell Development of RAG2-Deficient Induced Pluripotent Stem Cells in an Artificial Thymic Organoid System. <i>Journal of Clinical Immunology</i> , 2021, 41, 852-862.	2.0	27
48	Skewed TCR Alpha, but not Beta, Gene Rearrangements and Lymphoma Associated with a Pathogenic TRAC Variant. <i>Journal of Clinical Immunology</i> , 2021, 41, 1395-1399.	2.0	4
49	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
50	Preexisting autoantibodies to type I IFNs underlie critical COVID-19 pneumonia in patients with APS-1. <i>Journal of Experimental Medicine</i> , 2021, 218, .	4.2	185
51	<i>SASH3</i> variants cause a novel form of X-linked combined immunodeficiency with immune dysregulation. <i>Blood</i> , 2021, 138, 1019-1033.	0.6	28
52	Time-resolved systems immunology reveals a late juncture linked to fatal COVID-19. <i>Cell</i> , 2021, 184, 1836-1857.e22.	13.5	167
53	HSCT corrects primary immunodeficiency and immune dysregulation in patients with POMP-related autoinflammatory disease. <i>Blood</i> , 2021, 138, 1896-1901.	0.6	14
54	Clinical Manifestations, Mutational Analysis, and Immunological Phenotype in Patients with RAG1/2 Mutations: First Cases Series from Mexico and Description of Two Novel Mutations. <i>Journal of Clinical Immunology</i> , 2021, 41, 1291-1302.	2.0	2

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55	Ten Years of Newborn Screening for Severe Combined Immunodeficiency (SCID) in Massachusetts. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2021, 9, 2060-2067.e2.	2.0	20
56	Robust Antibody and T Cell Responses to SARS-CoV-2 in Patients with Antibody Deficiency. <i>Journal of Clinical Immunology</i> , 2021, 41, 1146-1153.	2.0	45
57	CRISPR-targeted <i>MAGT1</i> insertion restores XMEN patient hematopoietic stem cells and lymphocytes. <i>Blood</i> , 2021, 138, 2768-2780.	0.6	20
58	Thymic Epithelial Cell Alterations and Defective Thymopoiesis Lead to Central and Peripheral Tolerance Perturbation in MHCII Deficiency. <i>Frontiers in Immunology</i> , 2021, 12, 669943.	2.2	8
59	Inherited PD-1 deficiency underlies tuberculosis and autoimmunity in a child. <i>Nature Medicine</i> , 2021, 27, 1646-1654.	15.2	65
60	Harnessing Type I IFN Immunity Against SARS-CoV-2 with Early Administration of IFN- β . <i>Journal of Clinical Immunology</i> , 2021, 41, 1425-1442.	2.0	39
61	Humans with inherited T β cell CD28 deficiency are susceptible to skin papillomaviruses but are otherwise healthy. <i>Cell</i> , 2021, 184, 3812-3828.e30.	13.5	53
62	SARS-CoV-2 Spike Protein-Directed Monoclonal Antibodies May Ameliorate COVID-19 Complications in APECED Patients. <i>Frontiers in Immunology</i> , 2021, 12, 720205.	2.2	16
63	Association of rare predicted loss-of-function variants of influenza-related type I IFN genes with critical COVID-19 pneumonia. <i>Journal of Clinical Investigation</i> , 2021, 131, .	3.9	12
64	Neutralizing type I interferon autoantibodies are associated with delayed viral clearance and intensive care unit admission in patients with COVID-19. <i>Immunology and Cell Biology</i> , 2021, 99, 917-921.	1.0	69
65	Autoantibodies neutralizing type I IFNs are present in ~4% of uninfected individuals over 70 years old and account for ~20% of COVID-19 deaths. <i>Science Immunology</i> , 2021, 6, .	5.6	357
66	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. <i>Science Immunology</i> , 2021, 6, .	5.6	267
67	Treatment of Relapsing HPV Diseases by Restored Function of Natural Killer Cells. <i>New England Journal of Medicine</i> , 2021, 385, 921-929.	13.9	22
68	BTK inhibitors for severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2): A systematic review. <i>Clinical Immunology</i> , 2021, 230, 108816.	1.4	17
69	Response to Comments on "Aberrant type 1 immunity drives susceptibility to mucosal fungal infections". <i>Science</i> , 2021, 373, eabi8835.	6.0	5
70	Antibody responses to the SARS-CoV-2 vaccine in individuals with various inborn errors of immunity. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 148, 1192-1197.	1.5	67
71	<i>Nfkb2</i> variants reveal a p100-degradation threshold that defines autoimmune susceptibility. <i>Journal of Experimental Medicine</i> , 2021, 218, .	4.2	16
72	The Use of Induced Pluripotent Stem Cells to Study the Effects of Adenosine Deaminase Deficiency on Human Neutrophil Development. <i>Frontiers in Immunology</i> , 2021, 12, 748519.	2.2	7

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73	In Memoriamâ€”Thomas Alexander Waldmann, M.D.. Journal of Clinical Immunology, 2021, , 1.	2.0	0
74	Evidence of SARS-CoV-2-Specific T-Cell-Mediated Myocarditis in a MIS-A Case. Frontiers in Immunology, 2021, 12, 779026.	2.2	15
75	Targeted Therapy with Biologicals and Small Molecules in Primary Immunodeficiencies. Medical Principles and Practice, 2020, 29, 101-112.	1.1	15
76	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
77	Novel Missense Mutation in SP110 Associated with Combined Immunodeficiency and Advanced Liver Disease Without VOD. Journal of Clinical Immunology, 2020, 40, 236-239.	2.0	0
78	IgG Fc glycosylation as an axis of humoral immunity in childhood. Journal of Allergy and Clinical Immunology, 2020, 145, 710-713.e9.	1.5	27
79	The immunologic features of patients with early-onset and polyautoimmunity. Clinical Immunology, 2020, 211, 108326.	1.4	8
80	Defining a new immune deficiency syndrome: MAN2B2-CDG. Journal of Allergy and Clinical Immunology, 2020, 145, 1008-1011.	1.5	19
81	Asymptomatic Infant With Atypical SCID and Novel Hypomorphic RAG Variant Identified by Newborn Screening: A Diagnostic and Treatment Dilemma. Frontiers in Immunology, 2020, 11, 1954.	2.2	9
82	Vasculitis as a Major Morbidity Factor in Patients With Partial RAG Deficiency. Frontiers in Immunology, 2020, 11, 574738.	2.2	10
83	Updates on new monogenic inborn errors of immunity. Pediatric Allergy and Immunology, 2020, 31, 57-59.	1.1	4
84	Life-Threatening COVID-19: Defective Interferons Unleash Excessive Inflammation. Med, 2020, 1, 14-20.	2.2	110
85	Opinion and Special Articles: Cerebellar ataxia and liver failure complicating IPEX syndrome. Neurology, 2020, 96, 10.1212/WNL.0000000000011195.	1.5	2
86	Phosphate Transporter Profiles in Murine and Human Thymi Identify Thymocytes at Distinct Stages of Differentiation. Frontiers in Immunology, 2020, 11, 1562.	2.2	3
87	Targeted pharmacologic immunomodulation for inborn errors of immunity. British Journal of Clinical Pharmacology, 2020, , .	1.1	2
88	Activated PI3KÎ± breaches multiple B cell tolerance checkpoints and causes autoantibody production. Journal of Experimental Medicine, 2020, 217, .	4.2	33
89	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,749
90	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,983

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91	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
92	Excellent outcomes following hematopoietic cell transplantation for Wiskott-Aldrich syndrome: a PIDTC report. <i>Blood</i> , 2020, 135, 2094-2105.	0.6	87
93	Artificial thymic organoids represent a reliable tool to study T-cell differentiation in patients with severe T-cell lymphopenia. <i>Blood Advances</i> , 2020, 4, 2611-2616.	2.5	65
94	The Clinical and Genetic Spectrum of 82 Patients With RAG Deficiency Including a c.256_257delAA Founder Variant in Slavic Countries. <i>Frontiers in Immunology</i> , 2020, 11, 900.	2.2	16
95	Hematopoietic Cell Transplantation in Patients With Primary Immune Regulatory Disorders (PIRD): A Primary Immune Deficiency Treatment Consortium (PIDTC) Survey. <i>Frontiers in Immunology</i> , 2020, 11, 239.	2.2	57
96	Transplantation Outcomes for Children with Severe Combined Immune Deficiency (SCID) Have Improved over Time: A 36-Year Summary Report By the Primary Immune Deficiency Treatment Consortium (PIDTC). <i>Biology of Blood and Marrow Transplantation</i> , 2020, 26, S18-S19.	2.0	3
97	Flow Cytometry Identifies Risk Factors and Dynamic Changes in Patients with COVID-19. <i>Journal of Clinical Immunology</i> , 2020, 40, 970-973.	2.0	37
98	Human inborn errors of immunity: An expanding universe. <i>Science Immunology</i> , 2020, 5, .	5.6	138
99	Severe combined immune deficiency. , 2020, , 153-205.		7
100	PAX1 is essential for development and function of the human thymus. <i>Science Immunology</i> , 2020, 5, .	5.6	55
101	Low T-Cell Receptor \hat{I}^2 () Repertoire Diversity Early Post-Transplant for Severe Combined Immunodeficiency (SCID) Predicts Subsequent Failure of Immune Reconstitution. <i>Biology of Blood and Marrow Transplantation</i> , 2020, 26, S306.	2.0	1
102	International Retrospective Study of Allogeneic Hematopoietic Cell Transplantation (HCT) for Activated Phosphoinositide 3-Kinase Delta (PI3K) Syndrome. <i>Biology of Blood and Marrow Transplantation</i> , 2020, 26, S14-S15.	2.0	4
103	Cutaneous barrier leakage and gut inflammation drive skin disease in Omenn syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2020, 146, 1165-1179.e11.	1.5	13
104	Immune dysregulation in patients with RAG deficiency and other forms of combined immune deficiency. <i>Blood</i> , 2020, 135, 610-619.	0.6	37
105	2011 " MOUSE EMBRYONIC LYMPHOCYTES CONTRIBUTE TO AUTOIMMUNITY. <i>Experimental Hematology</i> , 2020, 88, S31.	0.2	0
106	Allogeneic Hematopoietic Stem-Cell Transplantation in Patients with GATA 2 Deficiency: Influence of Donor Stem Cell Source and Post-Transplantation Cyclophosphamide. <i>Blood</i> , 2020, 136, 37-38.	0.6	0
107	Impaired Sars-Cov-2 Specific Antibody Responses in Patients Treated with Anti-CD20 Antibodies. <i>Blood</i> , 2020, 136, 47-48.	0.6	0
108	Longitudinal Serological Response to Sars-COV-2 in Patients Affected By Hematologic Diseases. <i>Blood</i> , 2020, 136, 4-4.	0.6	0

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109	Recombination activity of human recombination-activating gene 2 (RAG2) mutations and correlation with clinical phenotype. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 726-735.	1.5	39
110	Gain-of-function CEBPE mutation causes noncanonical autoinflammatory inflammasomopathy. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 144, 1364-1376.	1.5	37
111	Chronic Granulomatous Disease-Associated IBD Resolves and Does Not Adversely Impact Survival Following Allogeneic HCT. <i>Journal of Clinical Immunology</i> , 2019, 39, 653-667.	2.0	41
112	Hematopoietic Stem Cell Transplantation in Primary Immunodeficiency Diseases: Current Status and Future Perspectives. <i>Frontiers in Pediatrics</i> , 2019, 7, 295.	0.9	144
113	From Natural Killer Cell Receptor Discovery to Characterization of Natural Killer Cell Defects in Primary Immunodeficiencies. <i>Frontiers in Immunology</i> , 2019, 10, 1757.	2.2	2
114	Generation of human induced pluripotent stem cell lines from patients with selective IgA deficiency. <i>Stem Cell Research</i> , 2019, 41, 101613.	0.3	0
115	Lack of specific T- and B-cell clonal expansions in multiple sclerosis patients with progressive multifocal leukoencephalopathy. <i>Scientific Reports</i> , 2019, 9, 16605.	1.6	4
116	Heterozygous FOXP1 Variants Cause Low TRECs and Severe T Cell Lymphopenia, Revealing a Crucial Role of FOXP1 in Supporting Early Thymopoiesis. <i>American Journal of Human Genetics</i> , 2019, 105, 549-561.	2.6	52
117	Inborn Errors of Immunity With Immune Dysregulation: From Bench to Bedside. <i>Frontiers in Pediatrics</i> , 2019, 7, 353.	0.9	85
118	Spatiotemporal Gradient of Cortical Neuron Death Contributes to Microcephaly in Knock-In Mouse Model of Ligase 4 Syndrome. <i>American Journal of Pathology</i> , 2019, 189, 2440-2449.	1.9	2
119	Severe influenza pneumonitis in children with inherited TLR3 deficiency. <i>Journal of Experimental Medicine</i> , 2019, 216, 2038-2056.	4.2	134
120	Lymphocyte-driven regional immunopathology in pneumonitis caused by impaired central immune tolerance. <i>Science Translational Medicine</i> , 2019, 11, .	5.8	52
121	Outcomes and Treatment Strategies for Autoimmunity and Hyperinflammation in Patients with RAG Deficiency. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2019, 7, 1970-1985.e4.	2.0	64
122	Cysteine and hydrophobic residues in CDR3 serve as distinct T-cell self-reactivity indices. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 144, 333-336.	1.5	31
123	Human interleukin-2 receptor $\hat{1}^2$ mutations associated with defects in immunity and peripheral tolerance. <i>Journal of Experimental Medicine</i> , 2019, 216, 1311-1327.	4.2	62
124	Two Unique Cases of X-linked SCID: A Diagnostic Challenge in the Era of Newborn Screening. <i>Frontiers in Pediatrics</i> , 2019, 7, 55.	0.9	10
125	Disseminated and Congenital Toxoplasmosis in a Mother and Child With Activated PI3-Kinase $\hat{1}$ Syndrome Type 2 (APDS2): Case Report and a Literature Review of Toxoplasma Infections in Primary Immunodeficiencies. <i>Frontiers in Immunology</i> , 2019, 10, 77.	2.2	16
126	B cell intrinsic requirement for STK4 in humoral immunity in mice and human subjects. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 2302-2305.	1.5	21

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127	Human DOCK2 Deficiency: Report of a Novel Mutation and Evidence for Neutrophil Dysfunction. <i>Journal of Clinical Immunology</i> , 2019, 39, 298-308.	2.0	31
128	Activating mutations in PIK3CD disrupt the differentiation and function of human and murine CD4+ T cells. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 144, 236-253.	1.5	44
129	F-BAR domain only protein 1 (FCHO1) deficiency is a novel cause of combined immune deficiency in human subjects. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 2317-2321.e12.	1.5	21
130	A combined immunodeficiency with severe infections, inflammation, and allergy caused by ARPC1B deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 2296-2299.	1.5	87
131	Thymic Epithelium Abnormalities in DiGeorge and Down Syndrome Patients Contribute to Dysregulation in T Cell Development. <i>Frontiers in Immunology</i> , 2019, 10, 447.	2.2	64
132	Second Case of HOIP Deficiency Expands Clinical Features and Defines Inflammatory Transcriptome Regulated by LUBAC. <i>Frontiers in Immunology</i> , 2019, 10, 479.	2.2	54
133	Increased proportions of $\gamma\delta$ T lymphocytes in atypical SCID associate with disease manifestations. <i>Clinical Immunology</i> , 2019, 201, 30-34.	1.4	6
134	Successful Allogeneic Hematopoietic Cell Transplantation (HCT) with Low Toxicity and Gvhd in a Heterogeneous Cohort of Primary Immunodeficiency (PID) Patients. <i>Biology of Blood and Marrow Transplantation</i> , 2019, 25, S42-S43.	2.0	0
135	Invasive and Allergic Complications Due to <i>Aspergillus fumigatus</i> in Allogeneic Hematopoietic Cell Transplantation (HCT) Primary Immunodeficiency (PID) Patients. <i>Biology of Blood and Marrow Transplantation</i> , 2019, 25, S356.	2.0	2
136	Human SNORA31 variations impair cortical neuron-intrinsic immunity to HSV-1 and underlie herpes simplex encephalitis. <i>Nature Medicine</i> , 2019, 25, 1873-1884.	15.2	76
137	Hematopoietic stem cell transplantation for activated phosphoinositide 3-kinase γ syndrome: Who, when, and how?. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 91-93.	1.5	9
138	<i>RAG</i> gene defects at the verge of immunodeficiency and immune dysregulation. <i>Immunological Reviews</i> , 2019, 287, 73-90.	2.8	44
139	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. <i>Nucleic Acids Research</i> , 2019, 47, D1018-D1027.	6.5	539
140	Consensus approach for the management of severe combined immune deficiency caused by adenosine deaminase deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 852-863.	1.5	104
141	Immune Reconstitution Therapy for Immunodeficiency. , 2019, , 1115-1128.e1.		0
142	Wiskott-Aldrich syndrome protein (WASP) is a tumor suppressor in T cell lymphoma. <i>Nature Medicine</i> , 2019, 25, 130-140.	15.2	57
143	The genetic landscape of severe combined immunodeficiency in the United States and Canada in the current era (2010-2018). <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 405-407.	1.5	64
144	Enhanced Transduction Lentivector Gene Therapy for Treatment of Older Patients with X-Linked Severe Combined Immunodeficiency. <i>Blood</i> , 2019, 134, 608-608.	0.6	7

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145	Primary immunodeficiencies: novel genes and unusual presentations. Hematology American Society of Hematology Education Program, 2019, 2019, 443-448.	0.9	18
146	Inborn Errors of RNA Lariat Metabolism in Humans with Brainstem Viral Infection. Cell, 2018, 172, 952-965.e18.	13.5	92
147	Autonomous role of Wiskott-Aldrich syndrome platelet deficiency in inducing autoimmunity and inflammation. Journal of Allergy and Clinical Immunology, 2018, 142, 1272-1284.	1.5	28
148	Prevalence and clinical challenges among adults with primary immunodeficiency and recombination-activating gene deficiency. Journal of Allergy and Clinical Immunology, 2018, 141, 2303-2306.	1.5	40
149	Patients with CD3G mutations reveal a role for human CD3 \hat{I} 3 in Treg diversity and suppressive function. Blood, 2018, 131, 2335-2344.	0.6	51
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