Luigi Daniele Notarangelo

List of Publications by Year in Descending Order

601 papers	38,678 citations	96 h-index	176 g-index
670 ext. papers	46,510 ext. citations	9.1 avg, IF	6.9 L-index

#	Paper	IF	Citations
601	Congenital and acquired defects of immunity: An ever-evolving story.. *Pediatric Allergy and Immunology*, **2022**, 33 Suppl 27, 61-64	4.2	0
600	Human genetic and immunological determinants of critical COVID-19 pneumonia.. *Nature*, **2022**,	50.4	23
599	SARS-CoV-2 infection in dialysis and kidney transplant patients: immunological and serological response.. *Journal of Nephrology*, **2022**, 1	4.8	0
598	Immunopathological signatures in multisystem inflammatory syndrome in children and pediatric COVID-19.. *Nature Medicine*, **2022**,	50.5	10
597	Case Report: Severe Rhabdomyolysis and Multiorgan Failure After ChAdOx1 nCoV-19 Vaccination.. *Frontiers in Immunology*, **2022**, 13, 845496	8.4	0
596	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	8.4	1
595	Granulocyte Transfusions in Patients with Chronic Granulomatous Disease Undergoing Hematopoietic Cell Transplantation or Gene Therapy.. *Journal of Clinical Immunology*, **2022**, 1	5.7	0
594	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	11.5	3
593	Human OTULIN haploinsufficiency impairs cell-intrinsic immunity to staphylococcal □toxin.. *Science* , **2022**, eabm6380	33.3	1
592	The Use of Induced Pluripotent Stem Cells to Study the Effects of Adenosine Deaminase Deficiency on Human Neutrophil Development. *Frontiers in Immunology*, **2021**, 12, 748519	8.4	1
591	Nfkb2 variants reveal a p100-degradation threshold that defines autoimmune susceptibility. *Journal of Experimental Medicine*, **2021**, 218,	16.6	5
590	Opinion and Special Articles: Cerebellar Ataxia and Liver Failure Complicating IPEX Syndrome. *Neurology*, **2021**, 96, e956-e959	6.5	0
589	RAG deficiencies: Recent advances in disease pathogenesis and novel therapeutic approaches. *European Journal of Immunology*, **2021**, 51, 1028-1038	6.1	3
588	Inhibition of HECT E3 ligases as potential therapy for COVID-19. *Cell Death and Disease*, **2021**, 12, 310	9.8	13
587	Gene Editing Rescues In vitro T Cell Development of RAG2-Deficient Induced Pluripotent Stem Cells in an Artificial Thymic Organoid System. *Journal of Clinical Immunology*, **2021**, 41, 852-862	5.7	5
586	Skewed TCR Alpha, but not Beta, Gene Rearrangements and Lymphoma Associated with a Pathogenic TRAC Variant. *Journal of Clinical Immunology*, **2021**, 41, 1395-1399	5.7	3
585	SARS-CoV-2-related MIS-C: A key to the viral and genetic causes of Kawasaki disease?. *Journal of Experimental Medicine*, **2021**, 218,	16.6	45

584	Preexisting autoantibodies to type I IFNs underlie critical COVID-19 pneumonia in patients with APS-1. *Journal of Experimental Medicine*, **2021**, 218,	16.6	79
583	SASH3 variants cause a novel form of X-linked combined immunodeficiency with immune dysregulation. *Blood*, **2021**, 138, 1019-1033	2.2	9
582	Time-resolved systems immunology reveals a late juncture linked to fatal COVID-19. *Cell*, **2021**, 184, 1836-1857.e22	56.2	64
581	HSCT corrects primary immunodeficiency and immune dysregulation in patients with POMP-related autoinflammatory disease. *Blood*, **2021**, 138, 1896-1901	2.2	6
580	Clinical Manifestations, Mutational Analysis, and Immunological Phenotype in Patients with RAG1/2 Mutations: First Cases Series from Mexico and Description of Two Novel Mutations. *Journal of Clinical Immunology*, **2021**, 41, 1291-1302	5.7	
579	Ten Years of Newborn Screening for Severe Combined Immunodeficiency (SCID) in Massachusetts. *Journal of Allergy and Clinical Immunology: in Practice*, **2021**, 9, 2060-2067.e2	5.4	6
578	Robust Antibody and T Cell Responses to SARS-CoV-2 in Patients with Antibody Deficiency. *Journal of Clinical Immunology*, **2021**, 41, 1146-1153	5.7	19
577	International retrospective study of allogeneic hematopoietic cell transplantation for activated PI3K-delta syndrome. *Journal of Allergy and Clinical Immunology*, **2021**,	11.5	9
576	Nodular regenerative hyperplasia in X-linked agammaglobulinemia: An underestimated and severe complication. *Journal of Allergy and Clinical Immunology*, **2021**,	11.5	2
575	CRISPR-targeted MAGT1 insertion restores XMEN patient hematopoietic stem cells and lymphocytes. *Blood*, **2021**,	2.2	1
574	Thymic Epithelial Cell Alterations and Defective Thymopoiesis Lead to Central and Peripheral Tolerance Perturbation in MHCII Deficiency. *Frontiers in Immunology*, **2021**, 12, 669943	8.4	2
573	Inherited PD-1 deficiency underlies tuberculosis and autoimmunity in a child. *Nature Medicine*, **2021**, 27, 1646-1654	50.5	17
572	Humans with inherited T´cell CD28 deficiency are susceptible to skin papillomaviruses but are otherwise healthy. *Cell*, **2021**, 184, 3812-3828.e30	56.2	18
571	POLD1 Deficiency Reveals a Role for POLD1 in DNA Repair and T and B Cell Development. *Journal of Clinical Immunology*, **2021**, 41, 270-273	5.7	6
570	Efficacy and safety of anti-CD45-saporin as conditioning agent for RAG deficiency. *Journal of Allergy and Clinical Immunology*, **2021**, 147, 309-320.e6	11.5	12
569	Infections in Infants with SCID: Isolation, Infection Screening, and Prophylaxis in PIDTC Centers. *Journal of Clinical Immunology*, **2021**, 41, 38-50	5.7	5
568	Novel Compound Heterozygous Mutations in ZAP70 Leading to a SCID Phenotype with Normal Downstream In vitro Signaling. *Journal of Clinical Immunology*, **2021**, 41, 470-472	5.7	1
567	Complete Absence of CD3 Protein Expression Is Responsible for Combined Immunodeficiency with Autoimmunity Rather than SCID. *Journal of Clinical Immunology*, **2021**, 41, 482-485	5.7	1

566	Reduction in the rate and improvement in the prognosis of COVID-19 in haematological patients over time. *Leukemia*, **2021**, 35, 632-634	10.7	2
565	Gut Microbiota-Host Interactions in Inborn Errors of Immunity. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	5
564	Infectious Complications Predict Premature CD8 T-cell Senescence in CD40 Ligand-Deficient Patients. *Journal of Clinical Immunology*, **2021**, 41, 795-806	5.7	1
563	TLR3 controls constitutive IFN-antiviral immunity in human fibroblasts and cortical neurons. *Journal of Clinical Investigation*, **2021**, 131,	15.9	19
562	Lost in Translation: Lack of CD4 Expression due to a Novel Genetic Defect. *Journal of Infectious Diseases*, **2021**, 223, 645-654	7	6
561	Aberrant type 1 immunity drives susceptibility to mucosal fungal infections. *Science*, **2021**, 371,	33.3	31
560	An immune-based biomarker signature is associated with mortality in COVID-19 patients. *JCI Insight* , **2021**, 6,	9.9	109
559	An appraisal of the Wilson & Jungner criteria in the context of genomic-based newborn screening for inborn errors of immunity. *Journal of Allergy and Clinical Immunology*, **2021**, 147, 428-438	11.5	3
558	Auto-antibodies to type I IFNs can underlie adverse reactions to yellow fever live attenuated vaccine. *Journal of Experimental Medicine*, **2021**, 218,	16.6	49
557	SARS-CoV-2 Spike Protein-Directed Monoclonal Antibodies May Ameliorate COVID-19 Complications in APECED Patients. *Frontiers in Immunology*, **2021**, 12, 720205	8.4	6
556	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,	15.9	7
555	IFNR1 deficiency presenting with visceral leishmaniasis and Mycobacterium Avium infections mimicking HLH. *Pediatric Allergy and Immunology*, **2021**,	4.2	1
554	Neutralizing type-I interferon autoantibodies are associated with delayed viral clearance and intensive care unit admission in patients with COVID-19. *Immunology and Cell Biology*, **2021**, 99, 917-921	5	17
553	Poor T-cell receptor repertoire diversity early posttransplant for severe combined immunodeficiency predicts failure of immune reconstitution. *Journal of Allergy and Clinical Immunology*, **2021**,	11.5	2
552	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,	28	91
551	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. *Science Immunology*, **2021**, 6,	28	67
550	Treatment of Relapsing HPV Diseases by Restored Function of Natural Killer Cells. *New England Journal of Medicine*, **2021**, 385, 921-929	59.2	6
549	BTK inhibitors for severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2): A systematic review. *Clinical Immunology*, **2021**, 230, 108816	9	10

548	Response to Comments on "Aberrant type 1 immunity drives susceptibility to mucosal fungal infections". *Science*, **2021**, 373, eabi8835	33.3	1
547	Antibody responses to the SARS-CoV-2 vaccine in individuals with various inborn errors of immunity. *Journal of Allergy and Clinical Immunology*, **2021**, 148, 1192-1197	11.5	18
546	Temporal Dynamics of Anti-Type 1 Interferon Autoantibodies in COVID-19 Patients. *Clinical Infectious Diseases*, **2021**,	11.6	6
545	Evidence of SARS-CoV-2-Specific T-Cell-Mediated Myocarditis in a MIS-A Case.. *Frontiers in Immunology*, **2021**, 12, 779026	8.4	0
544	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. *Cell*, **2020**, 181, 1194-1199	56.2	113
543	Excellent outcomes following hematopoietic cell transplantation for Wiskott-Aldrich syndrome: a PIDTC report. *Blood*, **2020**, 135, 2094-2105	2.2	46
542	Artificial thymic organoids represent a reliable tool to study T-cell differentiation in patients with severe T-cell lymphopenia. *Blood Advances*, **2020**, 4, 2611-2616	7.8	27
541	The Clinical and Genetic Spectrum of 82 Patients With Deficiency Including a c.256_257delAA Founder Variant in Slavic Countries. *Frontiers in Immunology*, **2020**, 11, 900	8.4	6
540	Hematopoietic Cell Transplantation in Patients With Primary Immune Regulatory Disorders (PIRD): A Primary Immune Deficiency Treatment Consortium (PIDTC) Survey. *Frontiers in Immunology*, **2020**, 11, 239	8.4	25
539	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). *Biology of Blood and Marrow Transplantation*, **2020**, 26, S18-S19	4.7	2
538	Flow Cytometry Identifies Risk Factors and Dynamic Changes in Patients with COVID-19. *Journal of Clinical Immunology*, **2020**, 40, 970-973	5.7	21
537	Human inborn errors of immunity: An expanding universe. *Science Immunology*, **2020**, 5,	28	58
536	Severe combined immune deficiency **2020**, 153-205		2
535	PAX1 is essential for development and function of the human thymus. *Science Immunology*, **2020**, 5,	28	27
534	Allogeneic Hematopoietic Stem-Cell Transplantation in Patients with GATA 2 Deficiency: Influence of Donor Stem Cell Source and Post-Transplantation Cyclophosphamide. *Blood*, **2020**, 136, 37-38	2.2	
533	Impaired Sars-Cov-2 Specific Antibody Responses in Patients Treated with Anti-CD20 Antibodies. *Blood*, **2020**, 136, 47-48	2.2	
532	Longitudinal Serological Response to Sars-COV-2 in Patients Affected By Hematologic Diseases. *Blood*, **2020**, 136, 4-4	2.2	
531	A large-scale database of T-cell receptor beta (TCR) sequences and binding associations from natural and synthetic exposure to SARS-CoV-2 **2020**,		30

530	Magnitude and Dynamics of the T-Cell Response to SARS-CoV-2 Infection at Both Individual and Population Levels **2020**,		64
529	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	4.7	15
528	Novel Missense Mutation inSP110Associated with Combined Immunodeficiency and Advanced Liver Disease Without VOD. *Journal of Clinical Immunology*, **2020**, 40, 236-239	5.7	
527	IgG Fc glycosylation as an axis of humoral immunity in childhood. *Journal of Allergy and Clinical Immunology*, **2020**, 145, 710-713.e9	11.5	15
526	The immunologic features of patients with early-onset and polyautoimmunity. *Clinical Immunology*, **2020**, 211, 108326	9	4
525	Defining a new immune deficiency syndrome: MAN2B2-CDG. *Journal of Allergy and Clinical Immunology*, **2020**, 145, 1008-1011	11.5	9
524	Asymptomatic Infant With Atypical SCID and Novel Hypomorphic Variant Identified by Newborn Screening: A Diagnostic and Treatment Dilemma. *Frontiers in Immunology*, **2020**, 11, 1954	8.4	4
523	Vasculitis as a Major Morbidity Factor in Patients With Partial RAG Deficiency. *Frontiers in Immunology*, **2020**, 11, 574738	8.4	3
522	Updates on new monogenic inborn errors of immunity. *Pediatric Allergy and Immunology*, **2020**, 31 Suppl 26, 57-59	4.2	3
521	Phosphate Transporter Profiles in Murine and Human Thymi Identify Thymocytes at Distinct Stages of Differentiation. *Frontiers in Immunology*, **2020**, 11, 1562	8.4	2
520	Targeted pharmacologic immunomodulation for inborn errors of immunity. *British Journal of Clinical Pharmacology*, **2020**,	3.8	1
519	Activated PI3K breaches multiple B cell tolerance checkpoints and causes autoantibody production. *Journal of Experimental Medicine*, **2020**, 217,	16.6	18
518	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. *Science*, **2020**, 370,	33.3	994
517	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. *Science*, **2020**, 370,	33.3	1090
516	Targeted Therapy with Biologicals and Small Molecules in Primary Immunodeficiencies. *Medical Principles and Practice*, **2020**, 29, 101-112	2.1	8
515	International Retrospective Study of Allogeneic Hematopoietic Cell Transplantation (HCT) for Activated Phosphoinositide 3-Kinase Delta (PI3K) Syndrome. *Biology of Blood and Marrow Transplantation*, **2020**, 26, S14-S15	4.7	4
514	Cutaneous barrier leakage and gut inflammation drive skin disease in Omenn syndrome. *Journal of Allergy and Clinical Immunology*, **2020**, 146, 1165-1179.e11	11.5	8
513	Immune dysregulation in patients with RAG deficiency and other forms of combined immune deficiency. *Blood*, **2020**, 135, 610-619	2.2	13

512	Heterozygous FOXN1 Variants Cause Low TRECs and Severe T Cell Lymphopenia, Revealing a Crucial Role of FOXN1 in Supporting Early Thymopoiesis. *American Journal of Human Genetics*, **2019** , 105, 549-561	11	28
511	Inborn Errors of Immunity With Immune Dysregulation: From Bench to Bedside. *Frontiers in Pediatrics*, **2019**, 7, 353	3.4	41
510	Spatiotemporal Gradient of Cortical Neuron Death Contributes to Microcephaly in Knock-In Mouse Model of Ligase 4 Syndrome. *American Journal of Pathology*, **2019**, 189, 2440-2449	5.8	
509	Severe influenza pneumonitis in children with inherited TLR3 deficiency. *Journal of Experimental Medicine*, **2019**, 216, 2038-2056	16.6	83
508	Lymphocyte-driven regional immunopathology in pneumonitis caused by impaired central immune tolerance. *Science Translational Medicine*, **2019**, 11,	17.5	31
507	Outcomes and Treatment Strategies for Autoimmunity and Hyperinflammation in Patients with RAG Deficiency. *Journal of Allergy and Clinical Immunology: in Practice*, **2019**, 7, 1970-1985.e4	5.4	41
506	Cysteine and hydrophobic residues in CDR3 serve as distinct T-cell self-reactivity indices. *Journal of Allergy and Clinical Immunology*, **2019**, 144, 333-336	11.5	16
505	Human interleukin-2 receptor mutations associated with defects in immunity and peripheral tolerance. *Journal of Experimental Medicine*, **2019**, 216, 1311-1327	16.6	41
504	Two Unique Cases of X-linked SCID: A Diagnostic Challenge in the Era of Newborn Screening. *Frontiers in Pediatrics*, **2019**, 7, 55	3.4	9
503	Disseminated and Congenital Toxoplasmosis in a Mother and Child With Activated PI3-Kinase Syndrome Type 2 (APDS2): Case Report and a Literature Review of Toxoplasma Infections in Primary Immunodeficiencies. *Frontiers in Immunology*, **2019**, 10, 77	8.4	8
502	B cell-intrinsic requirement for STK4 in humoral immunity in mice and human subjects. *Journal of Allergy and Clinical Immunology*, **2019**, 143, 2302-2305	11.5	15
501	Human DOCK2 Deficiency: Report of a Novel Mutation and Evidence for Neutrophil Dysfunction. *Journal of Clinical Immunology*, **2019**, 39, 298-308	5.7	17
500	Activating mutations in PIK3CD disrupt the differentiation and function of human and murine CD4 T cells. *Journal of Allergy and Clinical Immunology*, **2019**, 144, 236-253	11.5	31
499	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	11.5	17
498	A combined immunodeficiency with severe infections, inflammation, and allergy caused by ARPC1B deficiency. *Journal of Allergy and Clinical Immunology*, **2019**, 143, 2296-2299	11.5	59
497	Thymic Epithelium Abnormalities in DiGeorge and Down Syndrome Patients Contribute to Dysregulation in T Cell Development. *Frontiers in Immunology*, **2019**, 10, 447	8.4	33
496	Second Case of HOIP Deficiency Expands Clinical Features and Defines Inflammatory Transcriptome Regulated by LUBAC. *Frontiers in Immunology*, **2019**, 10, 479	8.4	31
495	Increased proportions of T lymphocytes in atypical SCID associate with disease manifestations. *Clinical Immunology*, **2019**, 201, 30-34	9	3

494	Recombination activity of human recombination-activating gene 2 (RAG2) mutations and correlation with clinical phenotype. *Journal of Allergy and Clinical Immunology*, **2019**, 143, 726-735	11.5	24
493	Gain-of-function CEBPE mutation causes noncanonical autoinflammatory inflammasomopathy. *Journal of Allergy and Clinical Immunology*, **2019**, 144, 1364-1376	11.5	20
492	Chronic Granulomatous Disease-Associated IBD Resolves and Does Not Adversely Impact Survival Following Allogeneic HCT. *Journal of Clinical Immunology*, **2019**, 39, 653-667	5.7	21
491	Hematopoietic Stem Cell Transplantation in Primary Immunodeficiency Diseases: Current Status and Future Perspectives. *Frontiers in Pediatrics*, **2019**, 7, 295	3.4	76
490	From Natural Killer Cell Receptor Discovery to Characterization of Natural Killer Cell Defects in Primary Immunodeficiencies. *Frontiers in Immunology*, **2019**, 10, 1757	8.4	1
489	Generation of human induced pluripotent stem cell lines from patients with selective IgA deficiency. *Stem Cell Research*, **2019**, 41, 101613	1.6	
488	Lack of specific T- and B-cell clonal expansions in multiple sclerosis patients with progressive multifocal leukoencephalopathy. *Scientific Reports*, **2019**, 9, 16605	4.9	2
487	Enhanced Transduction Lentivector Gene Therapy for Treatment of Older Patients with X-Linked Severe Combined Immunodeficiency. *Blood*, **2019**, 134, 608-608	2.2	3
486	Primary immunodeficiencies: novel genes and unusual presentations. *Hematology American Society of Hematology Education Program*, **2019**, 2019, 443-448	3.1	10
485	Invasive and Allergic Complications Due to Aspergillus fumigatus in Allogeneic Hematopoietic Cell Transplantation (HCT) Primary Immunodeficiency (PID) Patients. *Biology of Blood and Marrow Transplantation*, **2019**, 25, S356	4.7	2
484	Human SNORA31 variations impair cortical neuron-intrinsic immunity to HSV-1 and underlie herpes simplex encephalitis. *Nature Medicine*, **2019**, 25, 1873-1884	50.5	49
483	RAG gene defects at the verge of immunodeficiency and immune dysregulation. *Immunological Reviews*, **2019**, 287, 73-90	11.3	28
482	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. *Nucleic Acids Research*, **2019**, 47, D1018-D1027	20.1	333
481	Consensus approach for the management of severe combined immune deficiency caused by adenosine deaminase deficiency. *Journal of Allergy and Clinical Immunology*, **2019**, 143, 852-863	11.5	71
480	Immune Reconstitution Therapy for Immunodeficiency **2019**, 1115-1128.e1		
479	Wiskott-Aldrich syndrome protein (WASP) is a tumor suppressor in T cell lymphoma. *Nature Medicine*, **2019**, 25, 130-140	50.5	36
478	The genetic landscape of severe combined immunodeficiency in the United States and Canada in the current era (2010-2018). *Journal of Allergy and Clinical Immunology*, **2019**, 143, 405-407	11.5	35
477	Comprehensive Genetic Results for Primary Immunodeficiency Disorders in a Highly Consanguineous Population. *Frontiers in Immunology*, **2018**, 9, 3146	8.4	27

476	Inborn Errors of RNA Lariat Metabolism in Humans with Brainstem Viral Infection. *Cell*, **2018**, 172, 952-965.e18	36.2	64
475	Autonomous role of Wiskott-Aldrich syndrome platelet deficiency in inducing autoimmunity and inflammation. *Journal of Allergy and Clinical Immunology*, **2018**, 142, 1272-1284	11.5	17
474	Prevalence and clinical challenges among adults with primary immunodeficiency and recombination-activating gene deficiency. *Journal of Allergy and Clinical Immunology*, **2018**, 141, 2303-2306	11.5	25
473	Patients with mutations reveal a role for human CD3 in T diversity and suppressive function. *Blood* , **2018**, 131, 2335-2344	2.2	28
472	Neutrophils drive type I interferon production and autoantibodies in patients with Wiskott-Aldrich syndrome. *Journal of Allergy and Clinical Immunology*, **2018**, 142, 1605-1617.e4	11.5	13
471	Efficacy of lentivirus-mediated gene therapy in an Omenn syndrome recombination-activating gene 2 mouse model is not hindered by inflammation and immune dysregulation. *Journal of Allergy and Clinical Immunology*, **2018**, 142, 928-941.e8	11.5	16
470	WASP-mediated regulation of anti-inflammatory macrophages is IL-10 dependent and is critical for intestinal homeostasis. *Nature Communications*, **2018**, 9, 1779	17.4	22
469	CD40 ligand deficiency causes functional defects of peripheral neutrophils that are improved by exogenous IFN- *Journal of Allergy and Clinical Immunology*, **2018**, 142, 1571-1588.e9	11.5	19
468	DNA recombination defects in Kuwait: Clinical, immunologic and genetic profile. *Clinical Immunology*, **2018**, 187, 68-75	9	7
467	Future of Care for Patients With Chronic Granulomatous Disease: Gene Therapy and Targeted Molecular Medicine. *Journal of the Pediatric Infectious Diseases Society*, **2018**, 7, S40-S44	4.8	19
466	HPV: CIB1 is for EVER and EVER. *Journal of Experimental Medicine*, **2018**, 215, 2229-2231	16.6	2
465	Clinical and molecular features of X-linked hyper IgM syndrome - An experience from North India. *Clinical Immunology*, **2018**, 195, 59-66	9	11
464	Disrupted N-linked glycosylation as a disease mechanism in deficiency of ADA2. *Journal of Allergy and Clinical Immunology*, **2018**, 142, 1363-1365.e8	11.5	20
463	RAG Deficiency: Two Genes, Many Diseases. *Journal of Clinical Immunology*, **2018**, 38, 646-655	5.7	43
462	Congenital Disorders of Lymphocyte Function **2018**, 710-723.e3		1
461	Hypomorphic mutations alter the preimmune repertoire at early stages of lymphoid development. *Blood*, **2018**, 132, 281-292	2.2	16
460	Hyperactivated PI3K promotes self and commensal reactivity at the expense of optimal humoral immunity. *Nature Immunology*, **2018**, 19, 986-1000	19.1	54
459	Life-threatening influenza pneumonitis in a child with inherited IRF9 deficiency. *Journal of Experimental Medicine*, **2018**, 215, 2567-2585	16.6	98

458	Human iPSC-derived trigeminal neurons lack constitutive TLR3-dependent immunity that protects cortical neurons from HSV-1 infection. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, E8775-E8782	11.5	46
457	Biallelic mutations in DNA ligase 1 underlie a spectrum of immune deficiencies. *Journal of Clinical Investigation*, **2018**, 128, 5489-5504	15.9	19
456	RAG deficiency with ALPS features successfully treated with TCR$\alpha\beta$/CD19 cell depleted haploidentical stem cell transplant. *Clinical Immunology*, **2018**, 187, 102-103	9	9
455	T-cell defects in patients with germline mutations account for combined immunodeficiency. *Blood*, **2018**, 132, 2362-2374	2.2	59
454	T cell dynamics and response of the microbiota after gene therapy to treat X-linked severe combined immunodeficiency. *Genome Medicine*, **2018**, 10, 70	14.4	19
453	Adult-Onset Myopathy in a Patient with Hypomorphic RAG2 Mutations and Combined Immune Deficiency. *Journal of Clinical Immunology*, **2018**, 38, 642-645	5.7	2
452	SCID genotype and 6-month posttransplant CD4 count predict survival and immune recovery. *Blood* , **2018**, 132, 1737-1749	2.2	88
451	B-cell differentiation and IL-21 response in SCID patients after hematopoietic stem cell transplantation. *Blood*, **2018**, 131, 2967-2977	2.2	22
450	Current Knowledge and Priorities for Future Research in Late Effects after Hematopoietic Stem Cell Transplantation (HCT) for Severe Combined Immunodeficiency Patients: A Consensus Statement from the Second Pediatric Blood and Marrow Transplant Consortium International Conference on Late Effects after Pediatric HCT. *Biology of Blood and Marrow Transplantation*, **2017**, 23, 1979-1987	4.7	34
449	Ruxolitinib reverses dysregulated T helper cell responses and controls autoimmunity caused by a novel signal transducer and activator of transcription 1 (STAT1) gain-of-function mutation. *Journal of Allergy and Clinical Immunology*, **2017**, 139, 1629-1640.e2	11.5	104
448	mutations cause skeletal dysplasia, immune deficiency, and developmental delay. *Journal of Experimental Medicine*, **2017**, 214, 623-637	16.6	54
447	Analysis of clinical and immunologic phenotype in a large cohort of children and adults with cartilage-hair hypoplasia. *Journal of Allergy and Clinical Immunology*, **2017**, 140, 612-614.e5	11.5	21
446	Targeted strategies directed at the molecular defect: Toward precision medicine for select primary immunodeficiency disorders. *Journal of Allergy and Clinical Immunology*, **2017**, 139, 715-723	11.5	33
445	First Case of X-Linked Moesin Deficiency Identified After Newborn Screening for SCID. *Journal of Clinical Immunology*, **2017**, 37, 336-338	5.7	18
444	A three-dimensional model of human lung development and disease from pluripotent stem cells. *Nature Cell Biology*, **2017**, 19, 542-549	23.4	297
443	Recommendations for Screening and Management of Late Effects in Patients with Severe Combined Immunodeficiency after Allogenic Hematopoietic Cell Transplantation: A Consensus Statement from the Second Pediatric Blood and Marrow Transplant Consortium International Conference on Late Effects after Pediatric HCT. *Biology of Blood and Marrow Transplantation*, **2017**, 23, 1988-2003	4.7	27
442	Expanding the spectrum of skeletal dysplasia with immunodeficiency: a commentary on identification of biallelic EXTL3 mutations in a novel type of spondylo-epi-metaphyseal dysplasia. *Journal of Human Genetics*, **2017**, 62, 737-738	4.3	6
441	Genotype, Phenotype and T Cell Counts at One Year Predict Survival and Long Term Immune Reconstitution after Transplantation in Severe Combined Immune Deficiency (SCID)—The Primary Immune Deficiency Treatment Consortium (PIDTC). *Biology of Blood and Marrow Transplantation*, **2017**, 23, S133-S134	4.7	4

440	Reticular dysgenesis: international survey on clinical presentation, transplantation, and outcome. *Blood*, **2017**, 129, 2928-2938	2.2	26
439	Interleukin-12 and Interleukin-23 Blockade in Leukocyte Adhesion Deficiency Type 1. *New England Journal of Medicine*, **2017**, 376, 1141-1146	59.2	84
438	Cord Blood Banking for Potential Future Transplantation. *Pediatrics*, **2017**, 140,	7.4	26
437	Immune reconstitution and survival of 100 SCID patients post-hematopoietic cell transplant: a PIDTC natural history study. *Blood*, **2017**, 130, 2718-2727	2.2	129
436	Detection of Sp110 by Flow Cytometry and Application to Screening Patients for Veno-occlusive Disease with Immunodeficiency. *Journal of Clinical Immunology*, **2017**, 37, 707-714	5.7	8
435	Abnormalities of T-cell receptor repertoire in CD4 regulatory and conventional T cells in patients with RAG mutations: Implications for autoimmunity. *Journal of Allergy and Clinical Immunology*, **2017**, 140, 1739-1743.e7	11.5	22
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