Luigi Daniele Notarangelo

List of Publications by Citations

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	Activation-induced cytidine deaminase (AID) deficiency causes the autosomal recessive form of the Hyper-IgM syndrome (HIGM2). *Cell*, **2000**, 102, 565-75	56.2	1283
600	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. *Science*, **2020**, 370,	33.3	1090
599	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. *Science*, **2020**, 370,	33.3	994
598	The X-linked lymphoproliferative-disease gene product SAP regulates signals induced through the co-receptor SLAM. *Nature*, **1998**, 395, 462-9	50.4	807
597	Diagnostic criteria for primary immunodeficiencies. Representing PAGID (Pan-American Group for Immunodeficiency) and ESID (European Society for Immunodeficiencies). *Clinical Immunology*, **1999**, 93, 190-7	9	793
596	Gene therapy for immunodeficiency due to adenosine deaminase deficiency. *New England Journal of Medicine*, **2009**, 360, 447-58	59.2	792
595	Mutations of Jak-3 gene in patients with autosomal severe combined immune deficiency (SCID). *Nature*, **1995**, 377, 65-8	50.4	742
594	Gene therapy in peripheral blood lymphocytes and bone marrow for ADA- immunodeficient patients. *Science*, **1995**, 270, 470-5	33.3	655
593	Defective expression of T-cell CD40 ligand causes X-linked immunodeficiency with hyper-IgM. *Nature*, **1993**, 361, 539-41	50.4	641
592	Immune dysregulation in human subjects with heterozygous germline mutations in CTLA4. *Science*, **2014**, 345, 1623-1627	33.3	563
591	Defects in TCIRG1 subunit of the vacuolar proton pump are responsible for a subset of human autosomal recessive osteopetrosis. *Nature Genetics*, **2000**, 25, 343-6	36.3	540
590	Clinical spectrum of X-linked hyper-IgM syndrome. *Journal of Pediatrics*, **1997**, 131, 47-54	3.6	528
589	Transplantation outcomes for severe combined immunodeficiency, 2000-2009. *New England Journal of Medicine*, **2014**, 371, 434-46	59.2	457
588	Newborn screening for severe combined immunodeficiency in 11 screening programs in the United States. *JAMA - Journal of the American Medical Association*, **2014**, 312, 729-38	27.4	426
587	X-linked lymphoproliferative disease. 2B4 molecules displaying inhibitory rather than activating function are responsible for the inability of natural killer cells to kill Epstein-Barr virus-infected cells. *Journal of Experimental Medicine*, **2000**, 192, 337-46	16.6	398
586	Partial V(D)J recombination activity leads to Omenn syndrome. *Cell*, **1998**, 93, 885-96	56.2	383
585	Primary immunodeficiency diseases: an update from the International Union of Immunological Societies Primary Immunodeficiency Diseases Classification Committee. *Journal of Allergy and Clinical Immunology*, **2007**, 120, 776-94	11.5	362

584	Primary immunodeficiencies: 2009 update. *Journal of Allergy and Clinical Immunology*, **2009**, 124, 1161-78	11.5	361
583	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. *Nucleic Acids Research*, **2019**, 47, D1018-D1027	20.1	333
582	Transplantation of hematopoietic stem cells and long-term survival for primary immunodeficiencies in Europe: entering a new century, do we do better?. *Journal of Allergy and Clinical Immunology*, **2010**, 126, 602-10.e1-11	11.5	328
581	Immunodeficiency, autoinflammation and amylopectinosis in humans with inherited HOIL-1 and LUBAC deficiency. *Nature Immunology*, **2012**, 13, 1178-86	19.1	320
580	Human intracellular ISG15 prevents interferon-α over-amplification and auto-inflammation. *Nature*, **2015**, 517, 89-93	50.4	311
579	A three-dimensional model of human lung development and disease from pluripotent stem cells. *Nature Cell Biology*, **2017**, 19, 542-549	23.4	297
578	Mutations of CD40 gene cause an autosomal recessive form of immunodeficiency with hyper IgM. *Proceedings of the National Academy of Sciences of the United States of America*, **2001**, 98, 12614-9	11.5	294
577	IMMUNODEFICIENCIES. Impairment of immunity to Candida and Mycobacterium in humans with bi-allelic RORC mutations. *Science*, **2015**, 349, 606-613	33.3	291
576	Primary immunodeficiencies. *Journal of Allergy and Clinical Immunology*, **2010**, 125, S182-94	11.5	289
575	Infectious disease. Life-threatening influenza and impaired interferon amplification in human IRF7 deficiency. *Science*, **2015**, 348, 448-53	33.3	285
574	A modified γ-retrovirus vector for X-linked severe combined immunodeficiency. *New England Journal of Medicine*, **2014**, 371, 1407-17	59.2	278
573	V(D)J recombination defects in lymphocytes due to RAG mutations: severe immunodeficiency with a spectrum of clinical presentations. *Blood*, **2001**, 97, 81-8	2.2	278
572	Primary immunodeficiency diseases: an update on the classification from the international union of immunological societies expert committee for primary immunodeficiency. *Frontiers in Immunology*, **2011**, 2, 54	8.4	266
571	NTB-A [correction of GNTB-A], a novel SH2D1A-associated surface molecule contributing to the inability of natural killer cells to kill Epstein-Barr virus-infected B cells in X-linked lymphoproliferative disease. *Journal of Experimental Medicine*, **2001**, 194, 235-46	16.6	261
570	X-linked thrombocytopenia and Wiskott-Aldrich syndrome are allelic diseases with mutations in the WASP gene. *Nature Genetics*, **1995**, 9, 414-7	36.3	251
569	Clinical, immunological, and molecular analysis in a large cohort of patients with X-linked agammaglobulinemia: an Italian multicenter study. *Clinical Immunology*, **2002**, 104, 221-30	9	248
568	Mutations of the Wiskott-Aldrich Syndrome Protein (WASP): hotspots, effect on transcription, and translation and phenotype/genotype correlation. *Blood*, **2004**, 104, 4010-9	2.2	244
567	Clinical features, long-term follow-up and outcome of a large cohort of patients with Chronic Granulomatous Disease: an Italian multicenter study. *Clinical Immunology*, **2008**, 126, 155-64	9	242

566	Impaired intrinsic immunity to HSV-1 in human iPSC-derived TLR3-deficient CNS cells. *Nature*, **2012**, 491, 769-73	50.4	240
565	Long-term outcome and lineage-specific chimerism in 194 patients with Wiskott-Aldrich syndrome treated by hematopoietic cell transplantation in the period 1980-2009: an international collaborative study. *Blood*, **2011**, 118, 1675-84	2.2	236
564	Human osteoclast-poor osteopetrosis with hypogammaglobulinemia due to TNFRSF11A (RANK) mutations. *American Journal of Human Genetics*, **2008**, 83, 64-76	11	231
563	Inborn errors of human JAKs and STATs. *Immunity*, **2012**, 36, 515-28	32.3	225
562	Establishing diagnostic criteria for severe combined immunodeficiency disease (SCID), leaky SCID, and Omenn syndrome: the Primary Immune Deficiency Treatment Consortium experience. *Journal of Allergy and Clinical Immunology*, **2014**, 133, 1092-8	11.5	222
561	X-linked susceptibility to mycobacteria is caused by mutations in NEMO impairing CD40-dependent IL-12 production. *Journal of Experimental Medicine*, **2006**, 203, 1745-59	16.6	222
560	Long-term outcome following hematopoietic stem-cell transplantation in Wiskott-Aldrich syndrome: collaborative study of the European Society for Immunodeficiencies and European Group for Blood and Marrow Transplantation. *Blood*, **2008**, 111, 439-45	2.2	195
559	Bone marrow transplantation for severe combined immune deficiency. *JAMA - Journal of the American Medical Association*, **2006**, 295, 508-18	27.4	191
558	Omenn syndrome: inflammation in leaky severe combined immunodeficiency. *Journal of Allergy and Clinical Immunology*, **2008**, 122, 1082-6	11.5	185
557	Chloride channel ClCN7 mutations are responsible for severe recessive, dominant, and intermediate osteopetrosis. *Journal of Bone and Mineral Research*, **2003**, 18, 1740-7	6.3	179
556	Immune reconstitution in ADA-SCID after PBL gene therapy and discontinuation of enzyme replacement. *Nature Medicine*, **2002**, 8, 423-5	50.5	173
555	Missense Mutations in the Fas Gene Resulting in Autoimmune Lymphoproliferative Syndrome: A Molecular and Immunological Analysis. *Blood*, **1997**, 89, 902-909	2.2	172
554	Human HOIP and LUBAC deficiency underlies autoinflammation, immunodeficiency, amylopectinosis, and lymphangiectasia. *Journal of Experimental Medicine*, **2015**, 212, 939-51	16.6	171
553	DOCK8 functions as an adaptor that links TLR-MyD88 signaling to B cell activation. *Nature Immunology*, **2012**, 13, 612-20	19.1	170
552	How I treat ADA deficiency. *Blood*, **2009**, 114, 3524-32	2.2	168
551	The mutational spectrum of human malignant autosomal recessive osteopetrosis. *Human Molecular Genetics*, **2001**, 10, 1767-73	5.6	168
550	Lentiviral hematopoietic stem cell gene therapy for X-linked severe combined immunodeficiency. *Science Translational Medicine*, **2016**, 8, 335ra57	17.5	167
549	Guidelines for genetic studies in single patients: lessons from primary immunodeficiencies. *Journal of Experimental Medicine*, **2014**, 211, 2137-49	16.6	158

548	Loss-of-function mutations in the C9ORF72 mouse ortholog cause fatal autoimmune disease. *Science Translational Medicine*, **2016**, 8, 347ra93	17.5	157
547	Wiskott-Aldrich syndrome. *Current Opinion in Hematology*, **2008**, 15, 30-6	3.3	152
546	WASP regulates suppressor activity of human and murine CD4(+)CD25(+)FOXP3(+) natural regulatory T cells. *Journal of Experimental Medicine*, **2007**, 204, 369-80	16.6	149
545	Altered leukocyte response to CXCL12 in patients with warts hypogammaglobulinemia, infections, myelokathexis (WHIM) syndrome. *Blood*, **2004**, 104, 444-52	2.2	146
544	Primary immunodeficiency diseases: an update. *Journal of Allergy and Clinical Immunology*, **2004**, 114, 677-87	11.5	145
543	Interleukin-7 receptor alpha (IL-7Ralpha) deficiency: cellular and molecular bases. Analysis of clinical, immunological, and molecular features in 16 novel patients. *Immunological Reviews*, **2005**, 203, 110-26	11.3	142
542	A novel primary human immunodeficiency due to deficiency in the WASP-interacting protein WIP. *Journal of Experimental Medicine*, **2012**, 209, 29-34	16.6	138
541	Human RAG mutations: biochemistry and clinical implications. *Nature Reviews Immunology*, **2016**, 16, 234-46	36.5	131
540	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
539	AIRE deficiency in thymus of 2 patients with Omenn syndrome. *Journal of Clinical Investigation*, **2005**, 115, 728-732	15.9	128
538	A missense mutation in TFRC, encoding transferrin receptor 1, causes combined immunodeficiency. *Nature Genetics*, **2016**, 48, 74-8	36.3	127
537	Outcome of hematopoietic stem cell transplantation for adenosine deaminase-deficient severe combined immunodeficiency. *Blood*, **2012**, 120, 3615-24; quiz 3626	2.2	126
536	Inherited DOCK2 Deficiency in Patients with Early-Onset Invasive Infections. *New England Journal of Medicine*, **2015**, 372, 2409-22	59.2	125
535	Impaired natural and CD16-mediated NK cell cytotoxicity in patients with WAS and XLT: ability of IL-2 to correct NK cell functional defect. *Blood*, **2004**, 104, 436-43	2.2	120
534	Innate immunity defects in Hermansky-Pudlak type 2 syndrome. *Blood*, **2006**, 107, 4857-64	2.2	119
533	Early defects in human T-cell development severely affect distribution and maturation of thymic stromal cells: possible implications for the pathophysiology of Omenn syndrome. *Blood*, **2009**, 114, 105-8	2.2	117
532	Structural and Functional Basis for JAK3-Deficient Severe Combined Immunodeficiency. *Blood*, **1997**, 90, 3996-4003	2.2	117
531	Toll receptor-mediated regulation of NADPH oxidase in human dendritic cells. *Journal of Immunology*, **2004**, 173, 5749-56	5.3	117

530	Complex effects of naturally occurring mutations in the JAK3 pseudokinase domain: evidence for interactions between the kinase and pseudokinase domains. *Molecular and Cellular Biology*, **2000**, 20, 947-56	4.8	117
529	Signaling via IL-2 and IL-4 in JAK3-deficient severe combined immunodeficiency lymphocytes: JAK3-dependent and independent pathways. *Immunity*, **1996**, 5, 605-15	32.3	116
528	IL-21 is the primary common chain-binding cytokine required for human B-cell differentiation in vivo. *Blood*, **2011**, 118, 6824-35	2.2	115
527	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. *Cell*, **2020**, 181, 1194-1199	56.2	113
526	Timely and spatially regulated maturation of B and T cell repertoire during human fetal development. *Science Translational Medicine*, **2015**, 7, 276ra25	17.5	112
525	C4b-binding protein (C4BP) activates B cells through the CD40 receptor. *Immunity*, **2003**, 18, 837-48	32.3	112
524	Whole-exome sequencing identifies tetratricopeptide repeat domain 7A (TTC7A) mutations for combined immunodeficiency with intestinal atresias. *Journal of Allergy and Clinical Immunology*, **2013**, 132, 656-664.e17	11.5	109
523	Studies of the expression of the Wiskott-Aldrich syndrome protein. *Journal of Clinical Investigation*, **1996**, 97, 2627-34	15.9	109
522	An immune-based biomarker signature is associated with mortality in COVID-19 patients. *JCI Insight* , **2021**, 6,	9.9	109
521	Severe combined immunodeficiencies and related disorders. *Nature Reviews Disease Primers*, **2015**, 1, 15061	51.1	108
520	Global study of primary immunodeficiency diseases (PI)--diagnosis, treatment, and economic impact: an updated report from the Jeffrey Modell Foundation. *Immunologic Research*, **2011**, 51, 61-70	4.3	108
519	Primary immune deficiencies with aberrant IgE production. *Journal of Allergy and Clinical Immunology*, **2008**, 122, 1054-62; quiz 1063-4	11.5	107
518	Jak3, severe combined immunodeficiency, and a new class of immunosuppressive drugs. *Immunological Reviews*, **2005**, 203, 127-42	11.3	107
517	Defective expression of CD40 ligand on T cells causes "X-linked immunodeficiency with hyper-IgM (HIGM1)". *Immunological Reviews*, **1994**, 138, 39-59	11.3	107
516	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
515	Activation-induced cytidine deaminase (AID) is required for B-cell tolerance in humans. *Proceedings of the National Academy of Sciences of the United States of America*, **2011**, 108, 11554-9	11.5	102
514	A novel 4-kb interleukin-13 receptor alpha mRNA expressed in human B, T, and endothelial cells encoding an alternate type-II interleukin-4/interleukin-13 receptor. *European Journal of Immunology*, **1997**, 27, 971-8	6.1	101
513	Treatment of CD40 ligand deficiency by hematopoietic stem cell transplantation: a survey of the European experience, 1993-2002. *Blood*, **2004**, 103, 1152-7	2.2	101

512	CD30 cell expression and abnormal soluble CD30 serum accumulation in Omenn syndrome: evidence for a T helper 2-mediated condition. *European Journal of Immunology*, **1996**, 26, 329-34	6.1	101
511	A systematic analysis of recombination activity and genotype-phenotype correlation in human recombination-activating gene 1 deficiency. *Journal of Allergy and Clinical Immunology*, **2014**, 133, 1099-108	11.5	100
510	Recommendations for live viral and bacterial vaccines in immunodeficient patients and their close contacts. *Journal of Allergy and Clinical Immunology*, **2014**, 133, 961-6	11.5	100
509	Expression of inducible nitric oxide synthase in human granulomas and histiocytic reactions. *American Journal of Pathology*, **1999**, 154, 145-52	5.8	99
508	Life-threatening influenza pneumonitis in a child with inherited IRF9 deficiency. *Journal of Experimental Medicine*, **2018**, 215, 2567-2585	16.6	98
507	Improving cellular therapy for primary immune deficiency diseases: recognition, diagnosis, and management. *Journal of Allergy and Clinical Immunology*, **2009**, 124, 1152-60.e12	11.5	96
506	Hypomorphic Rag mutations can cause destructive midline granulomatous disease. *Blood*, **2010**, 116, 1263-71	2.2	96
505	Omenn syndrome in an infant with IL7RA gene mutation. *Journal of Pediatrics*, **2006**, 148, 272-4	3.6	94
504	Broad-spectrum antibodies against self-antigens and cytokines in RAG deficiency. *Journal of Clinical Investigation*, **2015**, 125, 4135-48	15.9	94
503	WASP confers selective advantage for specific hematopoietic cell populations and serves a unique role in marginal zone B-cell homeostasis and function. *Blood*, **2008**, 112, 4139-47	2.2	92
502	Defective Th1 cytokine gene transcription in CD4+ and CD8+ T cells from Wiskott-Aldrich syndrome patients. *Journal of Immunology*, **2006**, 177, 7451-61	5.3	91
501	Molecular cloning of ILP-2, a novel member of the inhibitor of apoptosis protein family. *Molecular and Cellular Biology*, **2001**, 21, 4292-301	4.8	91
500	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
499	Defects of class-switch recombination. *Journal of Allergy and Clinical Immunology*, **2006**, 117, 855-64	11.5	90
498	Ineffective expression of CD40 ligand on cord blood T cells may contribute to poor immunoglobulin production in the newborn. *European Journal of Immunology*, **1994**, 24, 1919-24	6.1	89
497	Long-term outcomes of 176 patients with X-linked hyper-IgM syndrome treated with or without hematopoietic cell transplantation. *Journal of Allergy and Clinical Immunology*, **2017**, 139, 1282-1292	11.5	88
496	SCID genotype and 6-month posttransplant CD4 count predict survival and immune recovery. *Blood* , **2018**, 132, 1737-1749	2.2	88
495	B cell-intrinsic deficiency of the Wiskott-Aldrich syndrome protein (WASp) causes severe abnormalities of the peripheral B-cell compartment in mice. *Blood*, **2012**, 119, 2819-28	2.2	87

494	A hypomorphic R229Q Rag2 mouse mutant recapitulates human Omenn syndrome. *Journal of Clinical Investigation*, **2007**, 117, 1260-9	15.9	87
493	Long-term immune reconstitution and clinical outcome after stem cell transplantation for severe T-cell immunodeficiency. *Journal of Allergy and Clinical Immunology*, **2007**, 120, 892-9	11.5	86
492	Interleukin-12 and Interleukin-23 Blockade in Leukocyte Adhesion Deficiency Type 1. *New England Journal of Medicine*, **2017**, 376, 1141-1146	59.2	84
491	Of genes and phenotypes: the immunological and molecular spectrum of combined immune deficiency. Defects of the gamma(c)-JAK3 signaling pathway as a model. *Immunological Reviews*, **2000**, 178, 39-48	11.3	84
490	CD40Lbase: a database of CD40L gene mutations causing X-linked hyper-IgM syndrome. *Trends in Immunology*, **1996**, 17, 511-516		84
489	Severe influenza pneumonitis in children with inherited TLR3 deficiency. *Journal of Experimental Medicine*, **2019**, 216, 2038-2056	16.6	83
488	Mutations in severe combined immune deficiency (SCID) due to JAK3 deficiency. *Human Mutation*, **2001**, 18, 255-63	4.7	83
487	Missense mutations of the WASP gene cause intermittent X-linked thrombocytopenia. *Blood*, **2002**, 99, 2268-9	2.2	83
486	Immature B cells preferentially switch to IgE with increased direct Sμ to Sε recombination. *Journal of Experimental Medicine*, **2011**, 208, 2733-46	16.6	81
485	Immunological and genetic bases of new primary immunodeficiencies. *Nature Reviews Immunology*, **2007**, 7, 851-61	36.5	81
484	A phenotypic approach for IUIS PID classification and diagnosis: guidelines for clinicians at the bedside. *Journal of Clinical Immunology*, **2013**, 33, 1078-87	5.7	79
483	Mutational analysis of human BAFF receptor TNFRSF13C (BAFF-R) in patients with common variable immunodeficiency. *Journal of Clinical Immunology*, **2005**, 25, 496-502	5.7	79
482	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
481	The natural history of children with severe combined immunodeficiency: baseline features of the first fifty patients of the primary immune deficiency treatment consortium prospective study 6901. *Journal of Clinical Immunology*, **2013**, 33, 1156-64	5.7	78
480	A single amino acid change, A91V, leads to conformational changes that can impair processing to the active form of perforin. *Blood*, **2005**, 106, 932-7	2.2	78
479	Expansion of immunoglobulin-secreting cells and defects in B cell tolerance in Rag-dependent immunodeficiency. *Journal of Experimental Medicine*, **2010**, 207, 1541-54	16.6	77
478	Hematopoietic Stem Cell Transplantation in Primary Immunodeficiency Diseases: Current Status and Future Perspectives. *Frontiers in Pediatrics*, **2019**, 7, 295	3.4	76
477	Dual T cell- and B cell-intrinsic deficiency in humans with biallelic RLTPR mutations. *Journal of Experimental Medicine*, **2016**, 213, 2413-2435	16.6	75

476	Management options for adenosine deaminase deficiency; proceedings of the EBMT satellite workshop (Hamburg, March 2006). *Clinical Immunology*, **2007**, 123, 139-47	9	75
475	PTX3 genetic variations affect the risk of Pseudomonas aeruginosa airway colonization in cystic fibrosis patients. *Genes and Immunity*, **2010**, 11, 665-70	4.4	74
474	Small RNAs derived from lncRNA RNase MRP have gene-silencing activity relevant to human cartilage-hair hypoplasia. *Human Molecular Genetics*, **2014**, 23, 368-82	5.6	73
473	X-linked immunodeficiency with hyper-IgM (XHIM). *Clinical and Experimental Immunology*, **2000**, 120, 399-405	6.2	73
472	WASPbase: a database of WAS- and XLT-causing mutations. *Trends in Immunology*, **1996**, 17, 496-502		73
471	A hypomorphic recombination-activating gene 1 (RAG1) mutation resulting in a phenotype resembling common variable immunodeficiency. *Journal of Allergy and Clinical Immunology*, **2014**, 134, 1375-1380	11.5	72
470	Clinical spectrum, pathophysiology and treatment of the Wiskott-Aldrich syndrome. *Current Opinion in Hematology*, **2011**, 18, 42-8	3.3	71
469	Intrathymic Restriction and Peripheral Expansion of the T-Cell Repertoire in Omenn Syndrome. *Blood*, **1999**, 94, 3468-3478	2.2	71
468	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
467	Defect of regulatory T cells in patients with Omenn syndrome. *Journal of Allergy and Clinical Immunology*, **2010**, 125, 209-16	11.5	70
466	Human peripheral lymphoid tissues contain autoimmune regulator-expressing dendritic cells. *American Journal of Pathology*, **2010**, 176, 1104-12	5.8	70
465	X-chromosome inactivation analysis in a female carrier of FOXP3 mutation. *Clinical and Experimental Immunology*, **2002**, 130, 127-30	6.2	70
464	Comparison of outcomes of hematopoietic stem cell transplantation without chemotherapy conditioning by using matched sibling and unrelated donors for treatment of severe combined immunodeficiency. *Journal of Allergy and Clinical Immunology*, **2014**, 134, 935-943.e15	11.5	69
463	G-CSF treatment of severe congenital neutropenia reverses neutropenia but does not correct the underlying functional deficiency of the neutrophil in defending against microorganisms. *Blood*, **2007**, 109, 4716-23	2.2	69
462	Primary immunodeficiencies: a rapidly evolving story. *Journal of Allergy and Clinical Immunology*, **2013**, 131, 314-23	11.5	68
461	Structural basis for chromosome X-linked agammaglobulinemia: a tyrosine kinase disease. *Proceedings of the National Academy of Sciences of the United States of America*, **1994**, 91, 12803-7	11.5	68
460	Activity of classical and alternative pathways of complement in preterm and small for gestational age infants. *Pediatric Research*, **1984**, 18, 281-5	3.2	68
459	A single-center experience in 20 patients with infantile malignant osteopetrosis. *American Journal of Hematology*, **2009**, 84, 473-9	7.1	67

458	Stem cell transplantation for the Wiskott-Aldrich syndrome: a single-center experience confirms efficacy of matched unrelated donor transplantation. *Bone Marrow Transplantation*, **2006**, 38, 671-9	4.4	67
457	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. *Science Immunology*, **2021**, 6,	28	67
456	Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy syndrome: time to review diagnostic criteria?. *Journal of Clinical Endocrinology and Metabolism*, **2003**, 88, 3146-8	5.6	66
455	Wiskott-Aldrich syndrome protein (WASP) and N-WASP are critical for peripheral B-cell development and function. *Blood*, **2012**, 119, 3966-74	2.2	65
454	Inborn Errors of RNA Lariat Metabolism in Humans with Brainstem Viral Infection. *Cell*, **2018**, 172, 952-965.e18	56.2	64
453	Magnitude and Dynamics of the T-Cell Response to SARS-CoV-2 Infection at Both Individual and Population Levels **2020**,		64
452	Time-resolved systems immunology reveals a late juncture linked to fatal COVID-19. *Cell*, **2021**, 184, 1836-1857.e22	56.2	64
451	Severe impairment of IFN-α and IFN-β responses in cells of a patient with a novel STAT1 splicing mutation. *Blood*, **2011**, 118, 1806-17	2.2	63
450	Characterization of T and B cell repertoire diversity in patients with RAG deficiency. *Science Immunology*, **2016**, 1,	28	62
449	Global overview of primary immunodeficiencies: a report from Jeffrey Modell Centers worldwide focused on diagnosis, treatment, and discovery. *Immunologic Research*, **2014**, 60, 132-44	4.3	61
448	Allogeneic hematopoietic cell transplantation for primary immune deficiency diseases: current status and critical needs. *Journal of Allergy and Clinical Immunology*, **2008**, 122, 1087-96	11.5	61
447	Novel Munc13-4 mutations in children and young adult patients with haemophagocytic lymphohistiocytosis. *Journal of Medical Genetics*, **2006**, 43, 953-60	5.8	61
446	Six novel mutations in the PRF1 gene in children with haemophagocytic lymphohistiocytosis. *Journal of Medical Genetics*, **2001**, 38, 643-6	5.8	61
445	In vitro correction of JAK3-deficient severe combined immunodeficiency by retroviral-mediated gene transduction. *Journal of Experimental Medicine*, **1996**, 183, 2687-92	16.6	61
444	Organization of the human CD40L gene: implications for molecular defects in X chromosome-linked hyper-IgM syndrome and prenatal diagnosis. *Proceedings of the National Academy of Sciences of the United States of America*, **1994**, 91, 2110-4	11.5	61
443	Disseminated cryptosporidium infection in an infant with hyper-IgM syndrome caused by CD40 deficiency. *Journal of Pediatrics*, **2003**, 142, 194-6	3.6	60
442	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
441	Identification of patients with RAG mutations previously diagnosed with common variable immunodeficiency disorders. *Journal of Clinical Immunology*, **2015**, 35, 119-24	5.7	59

440	A new case of IPEX receiving bone marrow transplantation. *Bone Marrow Transplantation*, **2005**, 35, 1033-4	1.4	59
439	T-cell defects in patients with germline mutations account for combined immunodeficiency. *Blood*, **2018**, 132, 2362-2374	2.2	59
438	Human inborn errors of immunity: An expanding universe. *Science Immunology*, **2020**, 5,	28	58
437	Unexpected and variable phenotypes in a family with JAK3 deficiency. *Genes and Immunity*, **2001**, 2, 422-32	1.4	58
436	Primary immunodeficiency mutation databases. *Advances in Genetics*, **2001**, 43, 103-88	3.3	58
435	Close linkage of probe p212 (DXS178) to X-linked agammaglobulinemia. *Human Genetics*, **1989**, 84, 19-21	6.3	58
434	Omenn syndrome: a disorder of Rag1 and Rag2 genes. *Journal of Clinical Immunology*, **1999**, 19, 87-97	5.7	57
433	AIRE deficiency in thymus of 2 patients with Omenn syndrome. *Journal of Clinical Investigation*, **2005**, 115, 728-32	15.9	56
432	Altered BCR and TLR signals promote enhanced positive selection of autoreactive transitional B cells in Wiskott-Aldrich syndrome. *Journal of Experimental Medicine*, **2015**, 212, 1663-77	16.6	55
431	Reduced thymic output, increased spontaneous apoptosis and oligoclonal B cells in polyethylene glycol-adenosine deaminase-treated patients. *European Journal of Immunology*, **2005**, 35, 3376-86	6.1	55
430	mutations cause skeletal dysplasia, immune deficiency, and developmental delay. *Journal of Experimental Medicine*, **2017**, 214, 623-637	16.6	54
429	Hyperactivated PI3K promotes self and commensal reactivity at the expense of optimal humoral immunity. *Nature Immunology*, **2018**, 19, 986-1000	19.1	54
428	Reversible severe combined immunodeficiency phenotype secondary to a mutation of the proton-coupled folate transporter. *Clinical Immunology*, **2009**, 133, 287-94	9	54
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