Jean-Laurent Casanova

List of Publications by Citations

747 papers	67,854 citations	133 h-index	236 g-index
819 ext. papers	81,715 ext. citations	11.5 avg, IF	7.57 L-index

#	Paper	IF	Citations
747	Gene therapy of human severe combined immunodeficiency (SCID)-X1 disease. *Science*, **2000**, 288, 669-72	33.3	2077
746	X-linked neonatal diabetes mellitus, enteropathy and endocrinopathy syndrome is the human equivalent of mouse scurfy. *Nature Genetics*, **2001**, 27, 18-20	36.3	1452
745	Chronic mucocutaneous candidiasis in humans with inborn errors of interleukin-17 immunity. *Science*, **2011**, 332, 65-8	33.3	1309
744	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. *Science*, **2020**, 370,	33.3	1090
743	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. *Science*, **2020**, 370,	33.3	994
742	Human CD14dim monocytes patrol and sense nucleic acids and viruses via TLR7 and TLR8 receptors. *Immunity*, **2010**, 33, 375-86	32.3	862
741	TLR3 deficiency in patients with herpes simplex encephalitis. *Science*, **2007**, 317, 1522-7	33.3	842
740	Genetic dissection of immunity to mycobacteria: the human model. *Annual Review of Immunology*, **2002**, 20, 581-620	34.7	771
739	Pyogenic bacterial infections in humans with IRAK-4 deficiency. *Science*, **2003**, 299, 2076-9	33.3	737
738	Interferon-gamma-receptor deficiency in an infant with fatal bacille Calmette-Guérin infection. *New England Journal of Medicine*, **1996**, 335, 1956-61	59.2	730
737	Severe mycobacterial and Salmonella infections in interleukin-12 receptor-deficient patients. *Science*, **1998**, 280, 1435-8	33.3	714
736	Impairment of mycobacterial immunity in human interleukin-12 receptor deficiency. *Science*, **1998**, 280, 1432-5	33.3	708
735	X-linked anhidrotic ectodermal dysplasia with immunodeficiency is caused by impaired NF-kappaB signaling. *Nature Genetics*, **2001**, 27, 277-85	36.3	681
734	Impaired response to interferon-alpha/beta and lethal viral disease in human STAT1 deficiency. *Nature Genetics*, **2003**, 33, 388-91	36.3	634
733	Interleukin-36-receptor antagonist deficiency and generalized pustular psoriasis. *New England Journal of Medicine*, **2011**, 365, 620-8	59.2	626
732	Human blood IgM "memory" B cells are circulating splenic marginal zone B cells harboring a prediversified immunoglobulin repertoire. *Blood*, **2004**, 104, 3647-54	2.2	612
731	Pyogenic bacterial infections in humans with MyD88 deficiency. *Science*, **2008**, 321, 691-6	33.3	608

730	Herpes simplex virus encephalitis in human UNC-93B deficiency. *Science*, **2006**, 314, 308-12	33.3	601
729	Gain-of-function human STAT1 mutations impair IL-17 immunity and underlie chronic mucocutaneous candidiasis. *Journal of Experimental Medicine*, **2011**, 208, 1635-48	16.6	599
728	Kawasaki-like multisystem inflammatory syndrome in children during the covid-19 pandemic in Paris, France: prospective observational study. *BMJ, The*, **2020**, 369, m2094	5.9	556
727	Autoantibodies against IL-17A, IL-17F, and IL-22 in patients with chronic mucocutaneous candidiasis and autoimmune polyendocrine syndrome type I. *Journal of Experimental Medicine*, **2010**, 207, 291-7	16.6	556
726	International Union of Immunological Societies: 2017 Primary Immunodeficiency Diseases Committee Report on Inborn Errors of Immunity. *Journal of Clinical Immunology*, **2018**, 38, 96-128	5.7	510
725	B cell-helper neutrophils stimulate the diversification and production of immunoglobulin in the marginal zone of the spleen. *Nature Immunology*, **2011**, 13, 170-80	19.1	501
724	Human Inborn Errors of Immunity: 2019 Update on the Classification from the International Union of Immunological Societies Expert Committee. *Journal of Clinical Immunology*, **2020**, 40, 24-64	5.7	497
723	IL-12 and IL-23 cytokines: from discovery to targeted therapies for immune-mediated inflammatory diseases. *Nature Medicine*, **2015**, 21, 719-29	50.5	488
722	Primary Immunodeficiency Diseases: an Update on the Classification from the International Union of Immunological Societies Expert Committee for Primary Immunodeficiency 2015. *Journal of Clinical Immunology*, **2015**, 35, 696-726	5.7	478
721	Efficacy of gene therapy for X-linked severe combined immunodeficiency. *New England Journal of Medicine*, **2010**, 363, 355-64	59.2	471
720	IRF8 mutations and human dendritic-cell immunodeficiency. *New England Journal of Medicine*, **2011**, 365, 127-38	59.2	469
719	Impairment of mycobacterial but not viral immunity by a germline human STAT1 mutation. *Science*, **2001**, 293, 300-3	33.3	419
718	A human IFNGR1 small deletion hotspot associated with dominant susceptibility to mycobacterial infection. *Nature Genetics*, **1999**, 21, 370-8	36.3	402
717	Mendelian susceptibility to mycobacterial disease: genetic, immunological, and clinical features of inborn errors of IFN-γmmunity. *Seminars in Immunology*, **2014**, 26, 454-70	10.7	401
716	Inborn errors of IL-12/23- and IFN-gamma-mediated immunity: molecular, cellular, and clinical features. *Seminars in Immunology*, **2006**, 18, 347-61	10.7	366
715	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
714	Primary immunodeficiencies: 2009 update. *Journal of Allergy and Clinical Immunology*, **2009**, 124, 1161-78	11.5	361
713	Mutations in STAT3 and IL12RB1 impair the development of human IL-17-producing T cells. *Journal of Experimental Medicine*, **2008**, 205, 1543-50	16.6	361

712	Clinical features of dominant and recessive interferon gamma receptor 1 deficiencies. *Lancet, The*, **2004**, 364, 2113-21	40	359
711	Mycobacterial disease and impaired IFN-γ immunity in humans with inherited ISG15 deficiency. *Science*, **2012**, 337, 1684-8	33.3	348
710	The 2017 IUIS Phenotypic Classification for Primary Immunodeficiencies. *Journal of Clinical Immunology*, **2018**, 38, 129-143	5.7	345
709	De novo gain-of-function KCNT1 channel mutations cause malignant migrating partial seizures of infancy. *Nature Genetics*, **2012**, 44, 1255-9	36.3	344
708	Whole-genome sequencing is more powerful than whole-exome sequencing for detecting exome variants. *Proceedings of the National Academy of Sciences of the United States of America*, **2015**, 112, 5473-8	11.5	332
707	Selective predisposition to bacterial infections in IRAK-4-deficient children: IRAK-4-dependent TLRs are otherwise redundant in protective immunity. *Journal of Experimental Medicine*, **2007**, 204, 2407-22	16.6	329
706	Immunodeficiency, autoinflammation and amylopectinosis in humans with inherited HOIL-1 and LUBAC deficiency. *Nature Immunology*, **2012**, 13, 1178-86	19.1	320
705	Heterozygous STAT1 gain-of-function mutations underlie an unexpectedly broad clinical phenotype. *Blood*, **2016**, 127, 3154-64	2.2	314
704	Human intracellular ISG15 prevents interferon-α over-amplification and auto-inflammation. *Nature*, **2015**, 517, 89-93	50.4	311
703	Primary immunodeficiency diseases: an update on the classification from the international union of immunological societies expert committee for primary immunodeficiency. *Frontiers in Immunology*, **2014**, 5, 162	8.4	309
702	Optimal conditions for directly sequencing double-stranded PCR products with sequenase. *Nucleic Acids Research*, **1990**, 18, 4028	20.1	298
701	A three-dimensional model of human lung development and disease from pluripotent stem cells. *Nature Cell Biology*, **2017**, 19, 542-549	23.4	297
700	Clinical features and outcome of patients with IRAK-4 and MyD88 deficiency. *Medicine (United States)*, **2010**, 89, 403-425	1.8	297
699	Systemic Human ILC Precursors Provide a Substrate for Tissue ILC Differentiation. *Cell*, **2017**, 168, 1086-1100.e10	30.2	293
698	IMMUNODEFICIENCIES. Impairment of immunity to Candida and Mycobacterium in humans with bi-allelic RORC mutations. *Science*, **2015**, 349, 606-613	33.3	291
697	Infectious diseases in patients with IRAK-4, MyD88, NEMO, or IκBα deficiency. *Clinical Microbiology Reviews*, **2011**, 24, 490-7	34	286
696	Infectious disease. Life-threatening influenza and impaired interferon amplification in human IRF7 deficiency. *Science*, **2015**, 348, 448-53	33.3	285
695	IL-12 and IFN-gamma in host defense against mycobacteria and salmonella in mice and men. *Current Opinion in Immunology*, **1999**, 11, 346-51	7.8	278

694	B cell-intrinsic signaling through IL-21 receptor and STAT3 is required for establishing long-lived antibody responses in humans. *Journal of Experimental Medicine*, **2010**, 207, 155-71	16.6	277
693	Revisiting human IL-12Rβ1 deficiency: a survey of 141 patients from 30 countries. *Medicine (United States)*, **2010**, 89, 381-402	1.8	277
692	A hypermorphic IkappaBalpha mutation is associated with autosomal dominant anhidrotic ectodermal dysplasia and T cell immunodeficiency. *Journal of Clinical Investigation*, **2003**, 112, 1108-15	15.9	277
691	Evolutionary dynamics of human Toll-like receptors and their different contributions to host defense. *PLoS Genetics*, **2009**, 5, e1000562	6	272
690	Human Inborn Errors of Immunity: 2019 Update of the IUIS Phenotypical Classification. *Journal of Clinical Immunology*, **2020**, 40, 66-81	5.7	267
689	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
688	Human TRAF3 adaptor molecule deficiency leads to impaired Toll-like receptor 3 response and susceptibility to herpes simplex encephalitis. *Immunity*, **2010**, 33, 400-11	32.3	262
687	Low penetrance, broad resistance, and favorable outcome of interleukin 12 receptor beta1 deficiency: medical and immunological implications. *Journal of Experimental Medicine*, **2003**, 197, 527-35	16.6	256
686	Human TLRs and IL-1Rs in host defense: natural insights from evolutionary, epidemiological, and clinical genetics. *Annual Review of Immunology*, **2011**, 29, 447-91	34.7	255
685	Novel human immunodeficiencies reveal the essential role of type-I cytokines in immunity to intracellular bacteria. *Trends in Immunology*, **1998**, 19, 491-4		255
684	Clinical spectrum and features of activated phosphoinositide 3-kinase δ syndrome: A large patient cohort study. *Journal of Allergy and Clinical Immunology*, **2017**, 139, 597-606.e4	11.5	251
683	The transmembrane activator TACI triggers immunoglobulin class switching by activating B cells through the adaptor MyD88. *Nature Immunology*, **2010**, 11, 836-45	19.1	251
682	Autoimmunity in Wiskott-Aldrich syndrome: risk factors, clinical features, and outcome in a single-center cohort of 55 patients. *Pediatrics*, **2003**, 111, e622-7	7.4	248
681	Deep dermatophytosis and inherited CARD9 deficiency. *New England Journal of Medicine*, **2013**, 369, 1704-1714	59.2	245
680	Early and prolonged intravenous immunoglobulin replacement therapy in childhood agammaglobulinemia: a retrospective survey of 31 patients. *Journal of Pediatrics*, **1999**, 134, 589-96	3.6	245
679	Impaired intrinsic immunity to HSV-1 in human iPSC-derived TLR3-deficient CNS cells. *Nature*, **2012**, 491, 769-73	50.4	240
678	Whole-exome sequencing-based discovery of STIM1 deficiency in a child with fatal classic Kaposi sarcoma. *Journal of Experimental Medicine*, **2010**, 207, 2307-12	16.6	236
677	Primary immunodeficiencies: a field in its infancy. *Science*, **2007**, 317, 617-9	33.3	236

676	Functional STAT3 deficiency compromises the generation of human T follicular helper cells. *Blood*, **2012**, 119, 3997-4008	2.2	230
675	Inherited interleukin-12 deficiency: IL12B genotype and clinical phenotype of 13 patients from six kindreds. *American Journal of Human Genetics*, **2002**, 70, 336-48	11	229
674	Human TLR-7-, -8-, and -9-mediated induction of IFN-alpha/beta and -lambda Is IRAK-4 dependent and redundant for protective immunity to viruses. *Immunity*, **2005**, 23, 465-78	32.3	228
673	Herpes simplex encephalitis in children with autosomal recessive and dominant TRIF deficiency. *Journal of Clinical Investigation*, **2011**, 121, 4889-902	15.9	227
672	Inborn errors of human JAKs and STATs. *Immunity*, **2012**, 36, 515-28	32.3	225
671	Herpes simplex virus encephalitis in a patient with complete TLR3 deficiency: TLR3 is otherwise redundant in protective immunity. *Journal of Experimental Medicine*, **2011**, 208, 2083-98	16.6	223
670	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
669	Treatment of the immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome (IPEX) by allogeneic bone marrow transplantation. *New England Journal of Medicine*, **2001**, 344, 1758-62	59.2	221
668	Inborn errors of interferon (IFN)-mediated immunity in humans: insights into the respective roles of IFN-alpha/beta, IFN-gamma, and IFN-lambda in host defense. *Immunological Reviews*, **2008**, 226, 29-40	11.3	220
667	The human model: a genetic dissection of immunity to infection in natural conditions. *Nature Reviews Immunology*, **2004**, 4, 55-66	36.5	212
666	Human TYK2 deficiency: Mycobacterial and viral infections without hyper-IgE syndrome. *Journal of Experimental Medicine*, **2015**, 212, 1641-62	16.6	209
665	Inborn errors of human IL-17 immunity underlie chronic mucocutaneous candidiasis. *Current Opinion in Allergy and Clinical Immunology*, **2012**, 12, 616-22	3.3	208
664	The NF-kappaB signalling pathway in human diseases: from incontinentia pigmenti to ectodermal dysplasias and immune-deficiency syndromes. *Human Molecular Genetics*, **2002**, 11, 2371-5	5.6	205
663	An ACT1 mutation selectively abolishes interleukin-17 responses in humans with chronic mucocutaneous candidiasis. *Immunity*, **2013**, 39, 676-86	32.3	204
662	Autosomal dominant STAT3 deficiency and hyper-IgE syndrome: molecular, cellular, and clinical features from a French national survey. *Medicine (United States)*, **2012**, 91, e1-e19	1.8	203
661	Germline CYBB mutations that selectively affect macrophages in kindreds with X-linked predisposition to tuberculous mycobacterial disease. *Nature Immunology*, **2011**, 12, 213-21	19.1	202
660	Partial MCM4 deficiency in patients with growth retardation, adrenal insufficiency, and natural killer cell deficiency. *Journal of Clinical Investigation*, **2012**, 122, 821-32	15.9	201
659	Hematopoietic stem cell transplantation in hemophagocytic lymphohistiocytosis: a single-center report of 48 patients. *Pediatrics*, **2006**, 117, e743-50	7.4	200

658	The genetic heterogeneity of mendelian susceptibility to mycobacterial diseases. *Journal of Allergy and Clinical Immunology*, **2008**, 122, 1043-51; quiz 1052-3	11.5	197
657	The European Society for Immunodeficiencies (ESID) Registry Working Definitions for the Clinical Diagnosis of Inborn Errors of Immunity. *Journal of Allergy and Clinical Immunology: in Practice*, **2019**, 7, 1763-1770	5.4	196
656	Heterozygous TBK1 mutations impair TLR3 immunity and underlie herpes simplex encephalitis of childhood. *Journal of Experimental Medicine*, **2012**, 209, 1567-82	16.6	196
655	IL-12 drives functional plasticity of human group 2 innate lymphoid cells. *Journal of Experimental Medicine*, **2016**, 213, 569-83	16.6	194
654	Inborn errors of human STAT1: allelic heterogeneity governs the diversity of immunological and infectious phenotypes. *Current Opinion in Immunology*, **2012**, 24, 364-78	7.8	194
653	Characterization of Greater Middle Eastern genetic variation for enhanced disease gene discovery. *Nature Genetics*, **2016**, 48, 1071-6	36.3	192
652	Long-term outcome after hematopoietic stem cell transplantation of a single-center cohort of 90 patients with severe combined immunodeficiency. *Blood*, **2009**, 113, 4114-24	2.2	184
651	Tuberculosis in children and adults: two distinct genetic diseases. *Journal of Experimental Medicine*, **2005**, 202, 1617-21	16.6	179
650	IRAK-4- and MyD88-dependent pathways are essential for the removal of developing autoreactive B cells in humans. *Immunity*, **2008**, 29, 746-57	32.3	178
649	Induction of MxA gene expression by influenza A virus requires type I or type III interferon signaling. *Journal of Virology*, **2007**, 81, 7776-85	6.6	175
648	Immunological conditions of children with BCG disseminated infection. *Lancet, The*, **1995**, 346, 581	40	174
647	Tartrate-resistant acid phosphatase deficiency causes a bone dysplasia with autoimmunity and a type I interferon expression signature. *Nature Genetics*, **2011**, 43, 127-31	36.3	173
646	The mutation significance cutoff: gene-level thresholds for variant predictions. *Nature Methods*, **2016**, 13, 109-10	21.6	171
645	Human HOIP and LUBAC deficiency underlies autoinflammation, immunodeficiency, amylopectinosis, and lymphangiectasia. *Journal of Experimental Medicine*, **2015**, 212, 939-51	16.6	171
644	Mutations in the TGF binding-protein-like domain 5 of FBN1 are responsible for acromicric and geleophysic dysplasias. *American Journal of Human Genetics*, **2011**, 89, 7-14	11	171
643	Interleukin (IL)-12 and IL-23 are key cytokines for immunity against Salmonella in humans. *Journal of Infectious Diseases*, **2004**, 190, 1755-7	7	169
642	Gains of glycosylation comprise an unexpectedly large group of pathogenic mutations. *Nature Genetics*, **2005**, 37, 692-700	36.3	168
641	Primary immunodeficiency diseases worldwide: more common than generally thought. *Journal of Clinical Immunology*, **2013**, 33, 1-7	5.7	166

640	Inherited disorders of NF-kappaB-mediated immunity in man. *Current Opinion in Immunology*, **2004**, 16, 34-41	7.8	166
639	Severe combined immunodeficiency and microcephaly in siblings with hypomorphic mutations in DNA ligase IV. *European Journal of Immunology*, **2006**, 36, 224-35	6.1	164
638	Single-cell PCR analysis of TCR repertoires selected by antigen in vivo: a high magnitude CD8 response is comprised of very few clones. *Immunity*, **1996**, 4, 47-55	32.3	164
637	Autoantibodies to interferon-gamma in a patient with selective susceptibility to mycobacterial infection and organ-specific autoimmunity. *Clinical Infectious Diseases*, **2004**, 38, e10-4	11.6	161
636	The 2015 IUIS Phenotypic Classification for Primary Immunodeficiencies. *Journal of Clinical Immunology*, **2015**, 35, 727-38	5.7	160
635	Loss of B Cells in Patients with Heterozygous Mutations in IKAROS. *New England Journal of Medicine*, **2016**, 374, 1032-1043	59.2	159
634	Human complete Stat-1 deficiency is associated with defective type I and II IFN responses in vitro but immunity to some low virulence viruses in vivo. *Journal of Immunology*, **2006**, 176, 5078-83	5.3	159
633	Guidelines for genetic studies in single patients: lessons from primary immunodeficiencies. *Journal of Experimental Medicine*, **2014**, 211, 2137-49	16.6	158
632	Partial T and B lymphocyte immunodeficiency and predisposition to lymphoma in patients with hypomorphic mutations in Artemis. *Journal of Clinical Investigation*, **2003**, 111, 381-7	15.9	158
631	Genetic Diagnosis Using Whole Exome Sequencing in Common Variable Immunodeficiency. *Frontiers in Immunology*, **2016**, 7, 220	8.4	158
630	The human gene damage index as a gene-level approach to prioritizing exome variants. *Proceedings of the National Academy of Sciences of the United States of America*, **2015**, 112, 13615-20	11.5	152
629	Parsing the Interferon Transcriptional Network and Its Disease Associations. *Cell*, **2016**, 164, 564-78	56.2	151
628	Genomic Signatures of Selective Pressures and Introgression from Archaic Hominins at Human Innate Immunity Genes. *American Journal of Human Genetics*, **2016**, 98, 5-21	11	149
627	Primary immunodeficiencies underlying fungal infections. *Current Opinion in Pediatrics*, **2013**, 25, 736-47	3.2	149
626	A novel immunodeficiency associated with hypomorphic RAG1 mutations and CMV infection. *Journal of Clinical Investigation*, **2005**, 115, 3291-9	15.9	149
625	Whole-exome-sequencing-based discovery of human FADD deficiency. *American Journal of Human Genetics*, **2010**, 87, 873-81	11	148
624	Severe infectious diseases of childhood as monogenic inborn errors of immunity. *Proceedings of the National Academy of Sciences of the United States of America*, **2015**, 112, E7128-37	11.5	147
623	Ribosomal protein SA haploinsufficiency in humans with isolated congenital asplenia. *Science*, **2013**, 340, 976-8	33.3	145

622	Primary immunodeficiency diseases: an update. *Journal of Allergy and Clinical Immunology*, **2004**, 114, 677-87	11.5	145
621	Inherited CARD9 deficiency in otherwise healthy children and adults with Candida species-induced meningoencephalitis, colitis, or both. *Journal of Allergy and Clinical Immunology*, **2015**, 135, 1558-68.e2	11.5	143
620	DOCK8 deficiency impairs CD8 T cell survival and function in humans and mice. *Journal of Experimental Medicine*, **2011**, 208, 2305-20	16.6	140
619	A partial form of recessive STAT1 deficiency in humans. *Journal of Clinical Investigation*, **2009**, 119, 1502-14	15.9	140
618	The role of interleukin-12 in human infectious diseases: only a faint signature. *European Journal of Immunology*, **2003**, 33, 1461-4	6.1	139
617	Novel STAT1 alleles in otherwise healthy patients with mycobacterial disease. *PLoS Genetics*, **2006**, 2, e131	6	138
616	Inherited and acquired immunodeficiencies underlying tuberculosis in childhood. *Immunological Reviews*, **2015**, 264, 103-20	11.3	133
615	Inborn errors of immunity to infection: the rule rather than the exception. *Journal of Experimental Medicine*, **2005**, 202, 197-201	16.6	133
614	Human interferon-gamma-mediated immunity is a genetically controlled continuous trait that determines the outcome of mycobacterial invasion. *Immunological Reviews*, **2000**, 178, 129-37	11.3	133
613	Inherited IL-17RC deficiency in patients with chronic mucocutaneous candidiasis. *Journal of Experimental Medicine*, **2015**, 212, 619-31	16.6	130
612	Immunity to infection in IL-17-deficient mice and humans. *European Journal of Immunology*, **2012**, 42, 2246-54	6.1	130
611	Life-threatening infectious diseases of childhood: single-gene inborn errors of immunity?. *Annals of the New York Academy of Sciences*, **2010**, 1214, 18-33	6.5	129
610	Immunology in natura: clinical, epidemiological and evolutionary genetics of infectious diseases. *Nature Immunology*, **2007**, 8, 1165-71	19.1	129
609	Human genetics of infectious diseases: between proof of principle and paradigm. *Journal of Clinical Investigation*, **2009**, 119, 2506-14	15.9	128
608	In a novel form of IFN-gamma receptor 1 deficiency, cell surface receptors fail to bind IFN-gamma. *Journal of Clinical Investigation*, **2000**, 105, 1429-36	15.9	127
607	Monogenic mutations differentially affect the quantity and quality of T follicular helper cells in patients with human primary immunodeficiencies. *Journal of Allergy and Clinical Immunology*, **2015**, 136, 993-1006.e1	11.5	126
606	Two loci control tuberculin skin test reactivity in an area hyperendemic for tuberculosis. *Journal of Experimental Medicine*, **2009**, 206, 2583-91	16.6	126
605	Inherited DOCK2 Deficiency in Patients with Early-Onset Invasive Infections. *New England Journal of Medicine*, **2015**, 372, 2409-22	59.2	125

604	Severe cutaneous papillomavirus disease after haemopoietic stem-cell transplantation in patients with severe combined immune deficiency caused by common gammac cytokine receptor subunit or JAK-3 deficiency. *Lancet, The*, **2004**, 363, 2051-4	40	125
603	Recurrent staphylococcal cellulitis and subcutaneous abscesses in a child with autoantibodies against IL-6. *Journal of Immunology*, **2008**, 180, 647-54	5.3	122
602	Human Toll-like receptor-dependent induction of interferons in protective immunity to viruses. *Immunological Reviews*, **2007**, 220, 225-36	11.3	122
601	Experimental and natural infections in MyD88- and IRAK-4-deficient mice and humans. *European Journal of Immunology*, **2012**, 42, 3126-35	6.1	121
600	Naive and memory human B cells have distinct requirements for STAT3 activation to differentiate into antibody-secreting plasma cells. *Journal of Experimental Medicine*, **2013**, 210, 2739-53	16.6	121
599	IL-12 receptor 1 deficiency alters in vivo T follicular helper cell response in humans. *Blood*, **2013**, 121, 3375-85	2.2	121
598	Inherited IL-12p40 deficiency: genetic, immunologic, and clinical features of 49 patients from 30 kindreds. *Medicine (United States)*, **2013**, 92, 109-122	1.8	121
597	Invasive pulmonary infection due to Scedosporium apiospermum in two children with chronic granulomatous disease. *Clinical Infectious Diseases*, **1998**, 27, 1437-41	11.6	120
596	Bacillus Calmette Guerin triggers the IL-12/IFN-gamma axis by an IRAK-4- and NEMO-dependent, non-cognate interaction between monocytes, NK, and T lymphocytes. *European Journal of Immunology*, **2004**, 34, 3276-84	6.1	119
595	Evolutionary genetic dissection of human interferons. *Journal of Experimental Medicine*, **2011**, 208, 2747-59	16.6	118
594	Inherited disorders of human Toll-like receptor signaling: immunological implications. *Immunological Reviews*, **2005**, 203, 10-20	11.3	115
593	Human genetics of tuberculosis: a long and winding road. *Philosophical Transactions of the Royal Society B: Biological Sciences*, **2014**, 369, 20130428	5.8	114
592	TLR3 immunity to infection in mice and humans. *Current Opinion in Immunology*, **2013**, 25, 19-33	7.8	114
591	Human genetic basis of interindividual variability in the course of infection. *Proceedings of the National Academy of Sciences of the United States of America*, **2015**, 112, E7118-27	11.5	114
590	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. *Cell*, **2020**, 181, 1194-1199	56.2	113
589	Novel primary immunodeficiencies revealed by the investigation of paediatric infectious diseases. *Current Opinion in Immunology*, **2008**, 20, 39-48	7.8	113
588	Human Mannose-binding Lectin in Immunity: Friend, Foe, or Both?. *Journal of Experimental Medicine* , **2004**, 199, 1295-9	16.6	113
587	Inborn errors of mucocutaneous immunity to Candida albicans in humans: a role for IL-17 cytokines?. *Current Opinion in Immunology*, **2010**, 22, 467-74	7.8	112

586	Fatal varicella associated with selective natural killer cell deficiency. *Journal of Pediatrics*, **2005**, 146, 423-5	3.6	112
585	Combined immunodeficiency and Epstein-Barr virus-induced B cell malignancy in humans with inherited CD70 deficiency. *Journal of Experimental Medicine*, **2017**, 214, 91-106	16.6	111
584	Genome-wide association study identifies variants associated with progression of liver fibrosis from HCV infection. *Gastroenterology*, **2012**, 143, 1244-1252.e12	13.3	111
583	IRAK4 and NEMO mutations in otherwise healthy children with recurrent invasive pneumococcal disease. *Journal of Medical Genetics*, **2007**, 44, 16-23	5.8	111
582	Exome and genome sequencing for inborn errors of immunity. *Journal of Allergy and Clinical Immunology*, **2016**, 138, 957-969	11.5	111
581	FAS-L, IL-10, and double-negative CD4- CD8- TCR alpha/beta+ T cells are reliable markers of autoimmune lymphoproliferative syndrome (ALPS) associated with FAS loss of function. *Blood*, **2009**, 113, 3027-30	2.2	110
580	Human RHOH deficiency causes T cell defects and susceptibility to EV-HPV infections. *Journal of Clinical Investigation*, **2012**, 122, 3239-47	15.9	109
579	Mutations at a single codon in Mad homology 2 domain of SMAD4 cause Myhre syndrome. *Nature Genetics*, **2011**, 44, 85-8	36.3	107
578	Revisiting Crohn's disease as a primary immunodeficiency of macrophages. *Journal of Experimental Medicine*, **2009**, 206, 1839-43	16.6	107
577	Human genetics of infectious diseases: a unified theory. *EMBO Journal*, **2007**, 26, 915-22	13	107
576	Nuclear factor kappaB essential modulator-deficient child with immunodeficiency yet without anhidrotic ectodermal dysplasia. *Journal of Allergy and Clinical Immunology*, **2004**, 114, 1456-62	11.5	107
575	Functional consequences of perforin gene mutations in 22 patients with familial haemophagocytic lymphohistiocytosis. *British Journal of Haematology*, **2002**, 117, 965-72	4.5	107
574	TLR3 deficiency in herpes simplex encephalitis: high allelic heterogeneity and recurrence risk. *Neurology*, **2014**, 83, 1888-97	6.5	105
573	Exome sequencing identifies PDE4D mutations as another cause of acrodysostosis. *American Journal of Human Genetics*, **2012**, 90, 740-5	11	105
572	BCG-osis and tuberculosis in a child with chronic granulomatous disease. *Journal of Allergy and Clinical Immunology*, **2007**, 120, 32-8	11.5	105
571	Osteopetrosis, lymphedema, anhidrotic ectodermal dysplasia, and immunodeficiency in a boy and incontinentia pigmenti in his mother. *Pediatrics*, **2002**, 109, e97	7.4	105
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