

# Laurent Abel

## List of Publications by Year in descending order

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Version: 2024-02-01

392  
papers

45,735  
citations

1231

110  
h-index

2558

195  
g-index

423  
all docs

423  
docs citations

423  
times ranked

39540  
citing authors

#	ARTICLE	IF	CITATIONS
1	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. <i>Science</i> , 2020, 370, .	6.0	1,983
2	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. <i>Science</i> , 2020, 370, .	6.0	1,749
3	Chronic Mucocutaneous Candidiasis in Humans with Inborn Errors of Interleukin-17 Immunity. <i>Science</i> , 2011, 332, 65-68.	6.0	1,482
4	TLR3 Deficiency in Patients with Herpes Simplex Encephalitis. <i>Science</i> , 2007, 317, 1522-1527.	6.0	970
5	GENETICDISSECTION OFIMMUNITY TOMYCOBACTERIA: The Human Model. <i>Annual Review of Immunology</i> , 2002, 20, 581-620.	9.5	900
6	Pyogenic Bacterial Infections in Humans with MyD88 Deficiency. <i>Science</i> , 2008, 321, 691-696.	6.0	844
7	Mutations of the RET proto-oncogene in Hirschsprung's disease. <i>Nature</i> , 1994, 367, 378-380.	13.7	750
8	Gain-of-function human <i>STAT1</i> mutations impair IL-17 immunity and underlie chronic mucocutaneous candidiasis. <i>Journal of Experimental Medicine</i> , 2011, 208, 1635-1648.	4.2	739
9	Herpes Simplex Virus Encephalitis in Human UNC-93B Deficiency. <i>Science</i> , 2006, 314, 308-312.	6.0	674
10	Autoantibodies against IL-17A, IL-17F, and IL-22 in patients with chronic mucocutaneous candidiasis and autoimmune polyendocrine syndrome type I. <i>Journal of Experimental Medicine</i> , 2010, 207, 291-297.	4.2	663
11	Mendelian susceptibility to mycobacterial disease: Genetic, immunological, and clinical features of inborn errors of IFN- $\gamma$ immunity. <i>Seminars in Immunology</i> , 2014, 26, 454-470.	2.7	582
12	<i>IRF8</i> Mutations and Human Dendritic-Cell Immunodeficiency. <i>New England Journal of Medicine</i> , 2011, 365, 127-138.	13.9	564
13	Whole-genome sequencing is more powerful than whole-exome sequencing for detecting exome variants. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, 5473-5478.	3.3	475
14	Mycobacterial Disease and Impaired IFN- $\gamma$ Immunity in Humans with Inherited ISG15 Deficiency. <i>Science</i> , 2012, 337, 1684-1688.	6.0	455
15	Human intracellular ISG15 prevents interferon- $\gamma$ over-amplification and auto-inflammation. <i>Nature</i> , 2015, 517, 89-93.	13.7	432
16	Susceptibility to leprosy is associated with PARK2 and PACRG. <i>Nature</i> , 2004, 427, 636-640.	13.7	426
17	Mutations in <i>STAT3</i> and <i>IL12RB1</i> impair the development of human IL-17-producing T cells. <i>Journal of Experimental Medicine</i> , 2008, 205, 1543-1550.	4.2	406
18	Life-threatening influenza and impaired interferon amplification in human IRF7 deficiency. <i>Science</i> , 2015, 348, 448-453.	6.0	389

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19	Selective predisposition to bacterial infections in IRAK-4-deficient children: IRAK-4-dependent TLRs are otherwise redundant in protective immunity. <i>Journal of Experimental Medicine</i> , 2007, 204, 2407-2422.	4.2	374
20	Revisiting Human IL-12R $\beta$ 1 Deficiency. <i>Medicine (United States)</i> , 2010, 89, 381-402.	0.4	367
21	Clinical Features and Outcome of Patients With IRAK-4 and MyD88 Deficiency. <i>Medicine (United States)</i> , 2010, 89, 403-425.	0.4	366
22	Impairment of immunity to <i>Candida</i> and <i>Mycobacterium</i> in humans with bi-allelic RORC mutations. <i>Science</i> , 2015, 349, 606-613.	6.0	366
23	Deep Dermatophytosis and Inherited CARD9 Deficiency. <i>New England Journal of Medicine</i> , 2013, 369, 1704-1714.	13.9	362
24	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
25	Evolutionary Dynamics of Human Toll-Like Receptors and Their Different Contributions to Host Defense. <i>PLoS Genetics</i> , 2009, 5, e1000562.	1.5	341
26	Genetic localization of a locus controlling the intensity of infection by <i>Schistosoma mansoni</i> on chromosome 5q31-q33. <i>Nature Genetics</i> , 1996, 14, 181-184.	9.4	326
27	Human TLRs and IL-1Rs in Host Defense: Natural Insights from Evolutionary, Epidemiological, and Clinical Genetics. <i>Annual Review of Immunology</i> , 2011, 29, 447-491.	9.5	316
28	Characterization of Greater Middle Eastern genetic variation for enhanced disease gene discovery. <i>Nature Genetics</i> , 2016, 48, 1071-1076.	9.4	314
29	Human TRAF3 Adaptor Molecule Deficiency Leads to Impaired Toll-like Receptor 3 Response and Susceptibility to Herpes Simplex Encephalitis. <i>Immunity</i> , 2010, 33, 400-411.	6.6	304
30	Human TYK2 deficiency: Mycobacterial and viral infections without hyper-IgE syndrome. <i>Journal of Experimental Medicine</i> , 2015, 212, 1641-1662.	4.2	293
31	Inborn errors of human IL-17 immunity underlie chronic mucocutaneous candidiasis. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2012, 12, 616-622.	1.1	288
32	Impaired intrinsic immunity to HSV-1 in human iPSC-derived TLR3-deficient CNS cells. <i>Nature</i> , 2012, 491, 769-773.	13.7	288
33	Low Penetrance, Broad Resistance, and Favorable Outcome of Interleukin 12 Receptor $\beta$ 1 Deficiency. <i>Journal of Experimental Medicine</i> , 2003, 197, 527-535.	4.2	286
34	Primary Immunodeficiencies: A Field in Its Infancy. <i>Science</i> , 2007, 317, 617-619.	6.0	280
35	Susceptibility to Leprosy Is Linked to the Human NRAMP1 Gene. <i>Journal of Infectious Diseases</i> , 1998, 177, 133-145.	1.9	273
36	Partial MCM4 deficiency in patients with growth retardation, adrenal insufficiency, and natural killer cell deficiency. <i>Journal of Clinical Investigation</i> , 2012, 122, 821-832.	3.9	272

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37	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. <i>Immunological Reviews</i> , 2008, 226, 29-40.	2.8	271
38	Whole-exome sequencing-based discovery of STIM1 deficiency in a child with fatal classic Kaposi sarcoma. <i>Journal of Experimental Medicine</i> , 2010, 207, 2307-2312.	4.2	268
39	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
40	Inherited Interleukin-12 Deficiency: IL12B Genotype and Clinical Phenotype of 13 Patients from Six Kindreds. <i>American Journal of Human Genetics</i> , 2002, 70, 336-348.	2.6	265
41	X-linked susceptibility to mycobacteria is caused by mutations in NEMO impairing CD40-dependent IL-12 production. <i>Journal of Experimental Medicine</i> , 2006, 203, 1745-1759.	4.2	264
42	Herpes simplex virus encephalitis in a patient with complete TLR3 deficiency: TLR3 is otherwise redundant in protective immunity. <i>Journal of Experimental Medicine</i> , 2011, 208, 2083-2098.	4.2	262
43	An ACT1 Mutation Selectively Abolishes Interleukin-17 Responses in Humans with Chronic Mucocutaneous Candidiasis. <i>Immunity</i> , 2013, 39, 676-686.	6.6	262
44	Human herpesvirus 8 transmission from mother to child and between siblings in an endemic population. <i>Lancet, The</i> , 2000, 356, 1062-1065.	6.3	255
45	Herpes simplex encephalitis in children with autosomal recessive and dominant TRIF deficiency. <i>Journal of Clinical Investigation</i> , 2011, 121, 4889-4902.	3.9	254
46	The mutation significance cutoff: gene-level thresholds for variant predictions. <i>Nature Methods</i> , 2016, 13, 109-110.	9.0	249
47	The human model: a genetic dissection of immunity to infection in natural conditions. <i>Nature Reviews Immunology</i> , 2004, 4, 55-66.	10.6	248
48	Germline CYBB mutations that selectively affect macrophages in kindreds with X-linked predisposition to tuberculous mycobacterial disease. <i>Nature Immunology</i> , 2011, 12, 213-221.	7.0	248
49	Inborn errors of human STAT1: allelic heterogeneity governs the diversity of immunological and infectious phenotypes. <i>Current Opinion in Immunology</i> , 2012, 24, 364-378.	2.4	245
50	Primary Immunodeficiency Diseases Worldwide: More Common than Generally Thought. <i>Journal of Clinical Immunology</i> , 2013, 33, 1-7.	2.0	243
51	Genomic Signatures of Selective Pressures and Introgression from Archaic Hominins at Human Innate Immunity Genes. <i>American Journal of Human Genetics</i> , 2016, 98, 5-21.	2.6	243
52	Human HOIP and LUBAC deficiency underlies autoinflammation, immunodeficiency, amylopectinosis, and lymphangiectasia. <i>Journal of Experimental Medicine</i> , 2015, 212, 939-951.	4.2	241
53	Heterozygous <i>TBK1</i> mutations impair TLR3 immunity and underlie herpes simplex encephalitis of childhood. <i>Journal of Experimental Medicine</i> , 2012, 209, 1567-1582.	4.2	231
54	Guidelines for genetic studies in single patients: lessons from primary immunodeficiencies. <i>Journal of Experimental Medicine</i> , 2014, 211, 2137-2149.	4.2	218

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55	Human genetic and immunological determinants of critical COVID-19 pneumonia. <i>Nature</i> , 2022, 603, 587-598.	13.7	216
56	The human gene damage index as a gene-level approach to prioritizing exome variants. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, 13615-13620.	3.3	213
57	Tuberculosis in children and adults. <i>Journal of Experimental Medicine</i> , 2005, 202, 1617-1621.	4.2	209
58	Inherited CARD9 deficiency in otherwise healthy children and adults with <i>Candida</i> species-induced meningoencephalitis, colitis, or both. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 135, 1558-1568.e2.	1.5	208
59	Severe Hepatic Fibrosis in <i>Schistosoma mansoni</i> Infection Is Controlled by a Major Locus That Is Closely Linked to the Interferon- $\gamma$ Receptor Gene. <i>American Journal of Human Genetics</i> , 1999, 65, 709-721.	2.6	198
60	Gains of glycosylation comprise an unexpectedly large group of pathogenic mutations. <i>Nature Genetics</i> , 2005, 37, 692-700.	9.4	198
61	Human adenylate kinase 2 deficiency causes a profound hematopoietic defect associated with sensorineural deafness. <i>Nature Genetics</i> , 2009, 41, 106-111.	9.4	198
62	Functional Analysis via Standardized Whole-Blood Stimulation Systems Defines the Boundaries of a Healthy Immune Response to Complex Stimuli. <i>Immunity</i> , 2014, 40, 436-450.	6.6	192
63	A gene for Hirschsprung disease maps to the proximal long arm of chromosome 10. <i>Nature Genetics</i> , 1993, 4, 346-350.	9.4	190
64	Exome and genome sequencing for inborn errors of immunity. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 957-969.	1.5	187
65	Resistance to <i>Schistosoma mansoni</i> in Humans: Influence of the IgE/IgG4 Balance and IgG2 in Immunity to Reinfection after Chemotherapy. <i>Journal of Infectious Diseases</i> , 1993, 168, 1000-1008.	1.9	185
66	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
67	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
68	A Missense LRRK2 Variant Is a Risk Factor for Excessive Inflammatory Responses in Leprosy. <i>PLoS Neglected Tropical Diseases</i> , 2016, 10, e0004412.	1.3	181
69	Inherited and acquired immunodeficiencies underlying tuberculosis in childhood. <i>Immunological Reviews</i> , 2015, 264, 103-120.	2.8	180
70	A novel immunodeficiency associated with hypomorphic RAG1 mutations and CMV infection. <i>Journal of Clinical Investigation</i> , 2005, 115, 3291-3299.	3.9	177
71	Ribosomal Protein SA Haploinsufficiency in Humans with Isolated Congenital Asplenia. <i>Science</i> , 2013, 340, 976-978.	6.0	176
72	Whole-Exome-Sequencing-Based Discovery of Human FADD Deficiency. <i>American Journal of Human Genetics</i> , 2010, 87, 873-881.	2.6	171

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73	Inherited DOCK2 Deficiency in Patients with Early-Onset Invasive Infections. <i>New England Journal of Medicine</i> , 2015, 372, 2409-2422.	13.9	169
74	A partial form of recessive STAT1 deficiency in humans. <i>Journal of Clinical Investigation</i> , 2009, 119, 1502-1514.	3.9	167
75	Chromosome 6q25 is linked to susceptibility to leprosy in a Vietnamese population. <i>Nature Genetics</i> , 2003, 33, 412-415.	9.4	164
76	Inherited IL-17RC deficiency in patients with chronic mucocutaneous candidiasis. <i>Journal of Experimental Medicine</i> , 2015, 212, 619-631.	4.2	162
77	Inborn errors of immunity to infection. <i>Journal of Experimental Medicine</i> , 2005, 202, 197-201.	4.2	161
78	Immunology in natura: clinical, epidemiological and evolutionary genetics of infectious diseases. <i>Nature Immunology</i> , 2007, 8, 1165-1171.	7.0	155
79	Life-threatening infectious diseases of childhood: single-gene inborn errors of immunity?. <i>Annals of the New York Academy of Sciences</i> , 2010, 1214, 18-33.	1.8	154
80	Human interferon-g-mediated immunity is a genetically controlled continuous trait that determines the outcome of mycobacterial invasion. <i>Immunological Reviews</i> , 2000, 178, 129-137.	2.8	153
81	Stepwise replication identifies a low-producing lymphotoxin- $\beta$ allele as a major risk factor for early-onset leprosy. <i>Nature Genetics</i> , 2007, 39, 517-522.	9.4	152
82	Human IFN- $\beta$ immunity to mycobacteria is governed by both IL-12 and IL-23. <i>Science Immunology</i> , 2018, 3, .	5.6	152
83	Inherited IL-12p40 Deficiency. <i>Medicine (United States)</i> , 2013, 92, 109-122.	0.4	151
84	Human genetics of infectious diseases: between proof of principle and paradigm. <i>Journal of Clinical Investigation</i> , 2009, 119, 2506-2514.	3.9	151
85	Tuberculosis and impaired IL-23-dependent IFN- $\beta$ immunity in humans homozygous for a common <i>TYK2</i> missense variant. <i>Science Immunology</i> , 2018, 3, .	5.6	148
86	Human Toll-like receptor-dependent induction of interferons in protective immunity to viruses. <i>Immunological Reviews</i> , 2007, 220, 225-236.	2.8	147
87	Life-threatening influenza pneumonitis in a child with inherited IRF9 deficiency. <i>Journal of Experimental Medicine</i> , 2018, 215, 2567-2585.	4.2	146
88	Human genetics of tuberculosis: a long and winding road. <i>Philosophical Transactions of the Royal Society B: Biological Sciences</i> , 2014, 369, 20130428.	1.8	144
89	Human genetics of infectious diseases: a unified theory. <i>EMBO Journal</i> , 2007, 26, 915-922.	3.5	143
90	Two loci control tuberculin skin test reactivity in an area hyperendemic for tuberculosis. <i>Journal of Experimental Medicine</i> , 2009, 206, 2583-2591.	4.2	142

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91	Genome-Wide Association Study Identifies Variants Associated With Progression of Liver Fibrosis From HCV Infection. <i>Gastroenterology</i> , 2012, 143, 1244-1252.e12.	0.6	142
92	TLR3 immunity to infection in mice and humans. <i>Current Opinion in Immunology</i> , 2013, 25, 19-33.	2.4	141
93	Inherited CARD9 Deficiency in 2 Unrelated Patients With Invasive <i>Exophiala</i> Infection. <i>Journal of Infectious Diseases</i> , 2015, 211, 1241-1250.	1.9	141
94	IL28B alleles associated with poor hepatitis C virus (HCV) clearance protect against inflammation and fibrosis in patients infected with non-1 HCV genotypes. <i>Hepatology</i> , 2012, 55, 384-394.	3.6	138
95	Genetic, immunological, and clinical features of patients with bacterial and fungal infections due to inherited IL-17RA deficiency. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, E8277-E8285.	3.3	137
96	Malaria in Humans: <i>Plasmodium falciparum</i> Blood Infection Levels Are Linked to Chromosome 5q31-q33. <i>American Journal of Human Genetics</i> , 1998, 63, 498-505.	2.6	136
97	Human Mannose-binding Lectin in Immunity. <i>Journal of Experimental Medicine</i> , 2004, 199, 1295-1299.	4.2	135
98	Severe influenza pneumonitis in children with inherited TLR3 deficiency. <i>Journal of Experimental Medicine</i> , 2019, 216, 2038-2056.	4.2	134
99	Human RHOH deficiency causes T cell defects and susceptibility to EV-HPV infections. <i>Journal of Clinical Investigation</i> , 2012, 122, 3239-3247.	3.9	134
100	A recessive form of hyper-IgE syndrome by disruption of ZNF341-dependent STAT3 transcription and activity. <i>Science Immunology</i> , 2018, 3, .	5.6	132
101	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
102	The Genetic Theory of Infectious Diseases: A Brief History and Selected Illustrations. <i>Annual Review of Genomics and Human Genetics</i> , 2013, 14, 215-243.	2.5	129
103	TLR3 deficiency in herpes simplex encephalitis. <i>Neurology</i> , 2014, 83, 1888-1897.	1.5	128
104	Novel primary immunodeficiencies revealed by the investigation of paediatric infectious diseases. <i>Current Opinion in Immunology</i> , 2008, 20, 39-48.	2.4	127
105	Inherited IFNAR1 deficiency in otherwise healthy patients with adverse reaction to measles and yellow fever live vaccines. <i>Journal of Experimental Medicine</i> , 2019, 216, 2057-2070.	4.2	127
106	Inborn errors of mucocutaneous immunity to <i>Candida albicans</i> in humans: a role for IL-17 cytokines?. <i>Current Opinion in Immunology</i> , 2010, 22, 467-474.	2.4	126
107	Inherited MST1 Deficiency Underlies Susceptibility to EV-HPV Infections. <i>PLoS ONE</i> , 2012, 7, e44010.	1.1	125
108	Inborn errors in RNA polymerase III underlie severe varicella zoster virus infections. <i>Journal of Clinical Investigation</i> , 2017, 127, 3543-3556.	3.9	125

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109	Clinical and epidemiological assessment of steroid-resistant nephrotic syndrome associated with the NPHS2 R229Q variant. <i>Kidney International</i> , 2009, 75, 727-735.	2.6	119
110	Inherited human OX40 deficiency underlying classic Kaposi sarcoma of childhood. <i>Journal of Experimental Medicine</i> , 2013, 210, 1743-1759.	4.2	119
111	Genetics of human susceptibility to active and latent tuberculosis: present knowledge and future perspectives. <i>Lancet Infectious Diseases</i> , The, 2018, 18, e64-e75.	4.6	119
112	Dual T cell and B cell intrinsic deficiency in humans with biallelic <i>RLTPR</i> mutations. <i>Journal of Experimental Medicine</i> , 2016, 213, 2413-2435.	4.2	117
113	Recurrent rhinovirus infections in a child with inherited MDA5 deficiency. <i>Journal of Experimental Medicine</i> , 2017, 214, 1949-1972.	4.2	117
114	Revisiting Crohn's disease as a primary immunodeficiency of macrophages. <i>Journal of Experimental Medicine</i> , 2009, 206, 1839-1843.	4.2	116
115	Host genetics of severe influenza: from mouse Mx1 to human IRF7. <i>Current Opinion in Immunology</i> , 2016, 38, 109-120.	2.4	115
116	Inherited GINS1 deficiency underlies growth retardation along with neutropenia and NK cell deficiency. <i>Journal of Clinical Investigation</i> , 2017, 127, 1991-2006.	3.9	115
117	Evolutionary insights into the high worldwide prevalence of MBL2 deficiency alleles. <i>Human Molecular Genetics</i> , 2006, 15, 2650-2658.	1.4	114
118	A Novel Primary Immunodeficiency with Specific Natural-Killer Cell Deficiency Maps to the Centromeric Region of Chromosome 8. <i>American Journal of Human Genetics</i> , 2006, 78, 721-727.	2.6	113
119	Human genetic variants and age are the strongest predictors of humoral immune responses to common pathogens and vaccines. <i>Genome Medicine</i> , 2018, 10, 59.	3.6	113
120	Estimating the age of rare disease mutations: the example of Triple-A syndrome. <i>Journal of Medical Genetics</i> , 2004, 41, 445-449.	1.5	112
121	IL-12R $\beta$ 1 Deficiency in Two of Fifty Children with Severe Tuberculosis from Iran, Morocco, and Turkey. <i>PLoS ONE</i> , 2011, 6, e18524.	1.1	111
122	Life-Threatening COVID-19: Defective Interferons Unleash Excessive Inflammation. <i>Med</i> , 2020, 1, 14-20.	2.2	110
123	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
124	Alleles of the NRAMP1 gene are risk factors for pediatric tuberculosis disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2005, 102, 12183-12188.	3.3	108
125	The gene for incontinentia pigmenti is assigned to Xq28. <i>Genomics</i> , 1989, 4, 427-429.	1.3	107
126	Mycobacterial disease in patients with chronic granulomatous disease: A retrospective analysis of 71 cases. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 241-248.e3.	1.5	106



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127	Polyclonal expansion of TCR V $\beta$ 21.3 CD4 and CD8 T cells is a hallmark of multisystem inflammatory syndrome in children. <i>Science Immunology</i> , 2021, 6, .	5.6	105
128	An autosomal dominant major gene confers predisposition to pulmonary tuberculosis in adults. <i>Journal of Experimental Medicine</i> , 2006, 203, 1679-1684.	4.2	104
129	Susceptibility to Periportal (Symmers) Fibrosis in Human <i>Schistosoma mansoni</i> Infections: Evidence That Intensity and Duration of Infection, Gender, and Inherited Factors Are Critical in Disease Progression. <i>Journal of Infectious Diseases</i> , 1999, 180, 1298-1306.	1.9	103
130	Partial recessive IFN- $\gamma$ R1 deficiency: genetic, immunological and clinical features of 14 patients from 11 kindreds. <i>Human Molecular Genetics</i> , 2011, 20, 1509-1523.	1.4	102
131	Major histocompatibility complex class II expression deficiency caused by a RFXANK founder mutation: a survey of 35 patients. <i>Blood</i> , 2011, 118, 5108-5118.	0.6	102
132	Genetic susceptibility to herpes simplex virus 1 encephalitis in mice and humans. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2007, 7, 495-505.	1.1	101
133	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
134	Neutralizing Autoantibodies to Type I IFNs in >10% of Patients with Severe COVID-19 Pneumonia Hospitalized in Madrid, Spain. <i>Journal of Clinical Immunology</i> , 2021, 41, 914-922.	2.0	100
135	Inherited p40phox deficiency differs from classic chronic granulomatous disease. <i>Journal of Clinical Investigation</i> , 2018, 128, 3957-3975.	3.9	99
136	Clinical Features of Candidiasis in Patients With Inherited Interleukin 12 Receptor $\beta$ 1 Deficiency. <i>Clinical Infectious Diseases</i> , 2014, 58, 204-213.	2.9	98
137	Disruption of an antimycobacterial circuit between dendritic and helper T cells in human SPPL2a deficiency. <i>Nature Immunology</i> , 2018, 19, 973-985.	7.0	96
138	Leprosy as a genetic disease. <i>Mammalian Genome</i> , 2011, 22, 19-31.	1.0	94
139	Clinical epidemiology of laboratory-confirmed Buruli ulcer in Benin: a cohort study. <i>The Lancet Global Health</i> , 2014, 2, e422-e430.	2.9	94
140	Autosomal Recessive Cardiomyopathy Presenting as Acute Myocarditis. <i>Journal of the American College of Cardiology</i> , 2017, 69, 1653-1665.	1.2	94
141	A novel form of human STAT1 deficiency impairing early but not late responses to interferons. <i>Blood</i> , 2010, 116, 5895-5906.	0.6	93
142	A Patient with Tyrosine Kinase 2 Deficiency without Hyper-IgE Syndrome. <i>Journal of Pediatrics</i> , 2012, 160, 1055-1057.	0.9	92
143	Inborn Errors of RNA Lariat Metabolism in Humans with Brainstem Viral Infection. <i>Cell</i> , 2018, 172, 952-965.e18.	13.5	92
144	The human CIB1-EVER1-EVER2 complex governs keratinocyte-intrinsic immunity to $\beta$ -papillomaviruses. <i>Journal of Experimental Medicine</i> , 2018, 215, 2289-2310.	4.2	92

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145	Homozygous <i>NLRP1</i> gain-of-function mutation in siblings with a syndromic form of recurrent respiratory papillomatosis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019, 116, 19055-19063.	3.3	92
146	Inherited human IFN- $\gamma$ deficiency underlies mycobacterial disease. <i>Journal of Clinical Investigation</i> , 2020, 130, 3158-3171.	3.9	89
147	The interplay between environmental and host factors during an outbreak of visceral leishmaniasis in eastern Sudan. <i>Microbes and Infection</i> , 2002, 4, 1449-1457.	1.0	88
148	Homozygosity for <i>TYK2</i> P1104A underlies tuberculosis in about 1% of patients in a cohort of European ancestry. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019, 116, 10430-10434.	3.3	87
149	Merkel cell polyomavirus infection occurs during early childhood and is transmitted between siblings. <i>Journal of Clinical Virology</i> , 2013, 58, 288-291.	1.6	86
150	The impact of host genetics on susceptibility to human infectious diseases. <i>Current Opinion in Immunology</i> , 1997, 9, 509-516.	2.4	85
151	Age-Dependent Mendelian Predisposition to Herpes Simplex Virus Type 1 Encephalitis in Childhood. <i>Journal of Pediatrics</i> , 2010, 157, 623-629.e1.	0.9	85
152	Genetic lessons learned from X-linked Mendelian susceptibility to mycobacterial diseases. <i>Annals of the New York Academy of Sciences</i> , 2011, 1246, 92-101.	1.8	85
153	Standardized Whole-Blood Transcriptional Profiling Enables the Deconvolution of Complex Induced Immune Responses. <i>Cell Reports</i> , 2016, 16, 2777-2791.	2.9	84
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