Anne Puel

List of Publications by Year
in descending order

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219 papers	32,435 citations	5261 83 h-index	4427 172 g-index
235 all docs	235 docs citations	235 times ranked	29932 citing authors

#	Article	IF	Citations
1	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,983
2	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,749
3	Chronic Mucocutaneous Candidiasis in Humans with Inborn Errors of Interleukin-17 Immunity. Science, 2011, 332, 65-68.	6.0	1,482
4	Human CD14dim Monocytes Patrol and Sense Nucleic Acids and Viruses via TLR7 and TLR8 Receptors. Immunity, 2010, 33, 375-386.	6.6	1,060
5	TLR3 Deficiency in Patients with Herpes Simplex Encephalitis. Science, 2007, 317, 1522-1527.	6.0	970
6	Pyogenic Bacterial Infections in Humans with MyD88 Deficiency. Science, 2008, 321, 691-696.	6.0	844
7	Interleukin-36â€“Receptor Antagonist Deficiency and Generalized Pustular Psoriasis. New England Journal of Medicine, 2011, 365, 620-628.	13.9	836
8	Pyogenic Bacterial Infections in Humans with IRAK-4 Deficiency. Science, 2003, 299, 2076-2079.	6.0	820
9	Defective IL7R expression in T-B+NK+ severe combined immunodeficiency. Nature Genetics, 1998, 20, 394-397.	9.4	760
10	Gain-of-function human STAT1 mutations impair IL-17 immunity and underlie chronic mucocutaneous candidiasis. Journal of Experimental Medicine, 2011, 208, 1635-1648.	4.2	739
11	Herpes Simplex Virus Encephalitis in Human UNC-93B Deficiency. Science, 2006, 314, 308-312.	6.0	674
12	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, 291-297.	4.2	663
13	Heterozygous STAT1 gain-of-function mutations underlie an unexpectedly broad clinical phenotype. Blood, 2016, 127, 3154-3164.	0.6	465
14	Mycobacterial Disease and Impaired IFN-Î³ Immunity in Humans with Inherited ISG15 Deficiency. Science, 2012, 337, 1684-1688.	6.0	455
15	Systemic Human ILC Precursors Provide a Substrate for Tissue ILC Differentiation. Cell, 2017, 168, 1086-1100.e10.	13.5	420
16	Immunodeficiency, autoinflammation and amylopectinosis in humans with inherited HOIL-1 and LUBAC deficiency. Nature Immunology, 2012, 13, 1178-1186.	7.0	410
17	Mutations in STAT3 and IL12RB1 impair the development of human IL-17â€“producing T cells. Journal of Experimental Medicine, 2008, 205, 1543-1550.	4.2	406
18	Cloning of a receptor subunit required for signaling by thymic stromal lymphopoietin. Nature Immunology, 2000, 1, 59-64.	7.0	393

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19	Human Inborn Errors of Immunity: 2022 Update on the Classification from the International Union of Immunological Societies Expert Committee. Journal of Clinical Immunology, 2022, 42, 1473-1507.	2.0	389
20	Selective predisposition to bacterial infections in IRAK-4â€“deficient children: IRAK-4â€“dependent TLRs are otherwise redundant in protective immunity. Journal of Experimental Medicine, 2007, 204, 2407-2422.	4.2	374
21	Clinical Features and Outcome of Patients With IRAK-4 and MyD88 Deficiency. Medicine (United States), 2010, 89, 403-425.	0.4	366
22	Impairment of immunity to Candida and Mycobacterium in humans with bi-allelic RORC mutations. Science, 2015, 349, 606-613.	6.0	366
23	Deep Dermatophytosis and Inherited CARD9 Deficiency. New England Journal of Medicine, 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. Science Immunology, 2021, 6, .	5.6	357
25	Infectious Diseases in Patients with IRAK-4, MyD88, NEMO, or ÎºBÎ± Deficiency. Clinical Microbiology Reviews, 2011, 24, 490-497.	5.7	349
26	A hypermorphic ÎºBÎ± mutation is associated with autosomal dominant anhidrotic ectodermal dysplasia and T cell immunodeficiency. Journal of Clinical Investigation, 2003, 112, 1108-1115.	3.9	325
27	Human TRAF3 Adaptor Molecule Deficiency Leads to Impaired Toll-like Receptor 3 Response and Susceptibility to Herpes Simplex Encephalitis. Immunity, 2010, 33, 400-411.	6.6	304
28	The transmembrane activator TACI triggers immunoglobulin class switching by activating B cells through the adaptor MyD88. Nature Immunology, 2010, 11, 836-845.	7.0	295
29	Human TYK2 deficiency: Mycobacterial and viral infections without hyper-IgE syndrome. Journal of Experimental Medicine, 2015, 212, 1641-1662.	4.2	293
30	Inborn errors of human IL-17 immunity underlie chronic mucocutaneous candidiasis. Current Opinion in Allergy and Clinical Immunology, 2012, 12, 616-622.	1.1	288
31	Autosomal Dominant STAT3 Deficiency and Hyper-IgE Syndrome. Medicine (United States), 2012, 91, e1-e19.	0.4	274
32	Inborn errors of interferon (IFN)â€mediated immunity in humans: insights into the respective roles of IFNâ€Î±/Î², IFNâ€Î³, and IFNâ€Î» in host defense. Immunological Reviews, 2008, 226, 29-40.	2.8	271
33	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. Science Immunology, 2021, 6, .	5.6	267
34	X-linked susceptibility to mycobacteria is caused by mutations in NEMO impairing CD40-dependent IL-12 production. Journal of Experimental Medicine, 2006, 203, 1745-1759.	4.2	264
35	An ACT1 Mutation Selectively Abolishes Interleukin-17 Responses in Humans with Chronic Mucocutaneous Candidiasis. Immunity, 2013, 39, 676-686.	6.6	262
36	Herpes simplex encephalitis in children with autosomal recessive and dominant TRIF deficiency. Journal of Clinical Investigation, 2011, 121, 4889-4902.	3.9	254

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37	Germline CYBB mutations that selectively affect macrophages in kindreds with X-linked predisposition to tuberculous mycobacterial disease. Nature Immunology, 2011, 12, 213-221.	7.0	248
38	Human TLR-7-, -8-, and -9-Mediated Induction of IFN-Î±/Î² and -Î» Is IRAK-4 Dependent and Redundant for Protective Immunity to Viruses. Immunity, 2005, 23, 465-478.	6.6	245
39	Inborn errors of human STAT1: allelic heterogeneity governs the diversity of immunological and infectious phenotypes. Current Opinion in Immunology, 2012, 24, 364-378.	2.4	245
40	Human genetic and immunological determinants of critical COVID-19 pneumonia. Nature, 2022, 603, 587-598.	13.7	216
41	Tartrate-resistant acid phosphatase deficiency causes a bone dysplasia with autoimmunity and a type I interferon expression signature. Nature Genetics, 2011, 43, 127-131.	9.4	214
42	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, 13615-13620.	3.3	213
43	Inherited CARD9 deficiency in otherwise healthy children and adults with Candida speciesâ€“induced meningoencephalitis, colitis, or both. Journal of Allergy and Clinical Immunology, 2015, 135, 1558-1568.e2.	1.5	208
44	IRAK-4- and MyD88-Dependent Pathways Are Essential for the Removal of Developing Autoreactive B Cells in Humans. Immunity, 2008, 29, 746-757.	6.6	201
45	Primary immunodeficiencies underlying fungal infections. Current Opinion in Pediatrics, 2013, 25, 736-747.	1.0	190
46	Inherited disorders of NF-ÎºB-mediated immunity in man. Current Opinion in Immunology, 2004, 16, 34-41.	2.4	188
47	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. Cell, 2020, 181, 1194-1199.	13.5	185
48	Preexisting autoantibodies to type I IFNs underlie critical COVID-19 pneumonia in patients with APS-1. Journal of Experimental Medicine, 2021, 218, .	4.2	185
49	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.	1.5	181
50	Ribosomal Protein SA Haploinsufficiency in Humans with Isolated Congenital Asplenia. Science, 2013, 340, 976-978.	6.0	176
51	Whole-Exome-Sequencing-Based Discovery of Human FADD Deficiency. American Journal of Human Genetics, 2010, 87, 873-881.	2.6	171
52	Experimental and natural infections in <scp>M</scp>y<scp>D</scp>88â€•and <scp>IRAK</scp>â€4â€deficient mice and humans. European Journal of Immunology, 2012, 42, 3126-3135.	1.6	169
53	Immunity to infection in <scp>IL</scp>â€17â€deficient mice and humans. European Journal of Immunology, 2012, 42, 2246-2254.	1.6	167
54	The Ever-Increasing Array of Novel Inborn Errors of Immunity: an Interim Update by the IUIS Committee. Journal of Clinical Immunology, 2021, 41, 666-679.	2.0	165

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55	Inherited IL-17RC deficiency in patients with chronic mucocutaneous candidiasis. Journal of Experimental Medicine, 2015, 212, 619-631.	4.2	162
56	Recurrent Staphylococcal Cellulitis and Subcutaneous Abscesses in a Child with Autoantibodies against IL-6. Journal of Immunology, 2008, 180, 647-654.	0.4	154
57	Human IFN-Î³ immunity to mycobacteria is governed by both IL-12 and IL-23. Science Immunology, 2018, 3, .	5.6	152
58	Chronic mucocutaneous candidiasis disease associated with inborn errors of ILâ€17 immunity. Clinical and Translational Immunology, 2016, 5, e114.	1.7	148
59	Tuberculosis and impaired IL-23â€“dependent IFN-Î³ immunity in humans homozygous for a common TYK2 missense variant. Science Immunology, 2018, 3, .	5.6	148
60	Human Tollâ€like receptorâ€dependent induction of interferons in protective immunity to viruses. Immunological Reviews, 2007, 220, 225-236.	2.8	147
61	Inherited CARD9 Deficiency in 2 Unrelated Patients With Invasive Exophiala Infection. Journal of Infectious Diseases, 2015, 211, 1241-1250.	1.9	141
62	Genetic, immunological, and clinical features of patients with bacterial and fungal infections due to inherited IL-17RA deficiency. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E8277-E8285.	3.3	137
63	A recessive form of hyper-IgE syndrome by disruption of ZNF341-dependent STAT3 transcription and activity. Science Immunology, 2018, 3, .	5.6	132
64	Inherited CARD9 Deficiency: Invasive Disease Caused by Ascomycete Fungi in Previously Healthy Children and Adults. Journal of Clinical Immunology, 2018, 38, 656-693.	2.0	130
65	Auto-antibodies to type I IFNs can underlie adverse reactions to yellow fever live attenuated vaccine. Journal of Experimental Medicine, 2021, 218, .	4.2	130
66	Inherited disorders of human Toll-like receptor signaling: immunological implications. Immunological Reviews, 2005, 203, 10-20.	2.8	129
67	Primary immunodeficiencies associated with pneumococcal disease. Current Opinion in Allergy and Clinical Immunology, 2003, 3, 451-459.	1.1	128
68	Novel primary immunodeficiencies revealed by the investigation of paediatric infectious diseases. Current Opinion in Immunology, 2008, 20, 39-48.	2.4	127
69	Inborn errors of mucocutaneous immunity to Candida albicans in humans: a role for IL-17 cytokines?. Current Opinion in Immunology, 2010, 22, 467-474.	2.4	126
70	IRAK4 and NEMO mutations in otherwise healthy children with recurrent invasive pneumococcal disease. Journal of Medical Genetics, 2006, 44, 16-23.	1.5	124
71	Nuclear factor ÎºB essential modulatorâ€deficient child with immunodeficiency yet without anhidrotic ectodermal dysplasia. Journal of Allergy and Clinical Immunology, 2004, 114, 1456-1462.	1.5	122
72	Dual T cellâ€ and B cellâ€intrinsic deficiency in humans with biallelic RLTPR mutations. Journal of Experimental Medicine, 2016, 213, 2413-2435.	4.2	117

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73	ZNF341 controls STAT3 expression and thereby immunocompetence. Science Immunology, 2018, 3, .	5.6	113
74	Human gut mycobiota tune immunity via CARD9-dependent induction of anti-fungal IgG antibodies. Cell, 2021, 184, 1017-1031.e14.	13.5	113
75	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.	3.3	110
76	SARS-CoV-2 induces human plasmacytoid predendritic cell diversification via UNC93B and IRAK4. Journal of Experimental Medicine, 2021, 218, .	4.2	107
77	New and recurrent gain-of-functionSTAT1mutations in patients with chronic mucocutaneous candidiasis from Eastern and Central Europe. Journal of Medical Genetics, 2013, 50, 567-578.	1.5	105
78	Inborn errors of immunity underlying fungal diseases in otherwise healthy individuals. Current Opinion in Microbiology, 2017, 40, 46-57.	2.3	101
79	SARS-CoV-2â€“related MIS-C: A key to the viral and genetic causes of Kawasaki disease?. Journal of Experimental Medicine, 2021, 218, .	4.2	100
80	Clinical Features of Candidiasis in Patients With Inherited Interleukin 12 Receptor Î²1 Deficiency. Clinical Infectious Diseases, 2014, 58, 204-213.	2.9	98
81	Chronic and Invasive Fungal Infections in a Family with CARD9 Deficiency. Journal of Clinical Immunology, 2016, 36, 204-209.	2.0	98
82	Hematopoietic stem cell transplantation in 29 patients hemizygous for hypomorphic IKBKG/NEMO mutations. Blood, 2017, 130, 1456-1467.	0.6	95
83	The NEMO Mutation Creating the Most-Upstream Premature Stop Codon Is Hypomorphic Because of a Reinitiation of Translation. American Journal of Human Genetics, 2006, 78, 691-701.	2.6	89
84	A Homozygous CARD9 Mutation in a Brazilian Patient with Deep Dermatophytosis. Journal of Clinical Immunology, 2015, 35, 486-490.	2.0	89
85	IgM+IgD+CD27+ B cells are markedly reduced in IRAK-4â€“, MyD88-, and TIRAP- but not UNC-93Bâ€“deficient patients. Blood, 2012, 120, 4992-5001.	0.6	87
86	Human STAT1 Gain-of-Function Heterozygous Mutations: Chronic Mucocutaneous Candidiasis and Type I Interferonopathy. Journal of Clinical Immunology, 2020, 40, 1065-1081.	2.0	86
87	TLR8-mediated NF-ÎºB and JNK Activation Are TAK1-independent and MEKK3-dependent. Journal of Biological Chemistry, 2006, 281, 21013-21021.	1.6	84
88	Severe Dermatophytosis and Acquired or Innate Immunodeficiency: A Review. Journal of Fungi (Basel,) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	1.5	84
89	Human T-bet Governs Innate and Innate-like Adaptive IFN-Î³ Immunity against Mycobacteria. Cell, 2020, 183, 1826-1847.e31.	13.5	83
90	Rhinoscleroma: A French National Retrospective Study of Epidemiological and Clinical Features. Clinical Infectious Diseases, 2008, 47, 1396-1402.	2.9	79

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91	Simple diagnosis ofSTAT1gain-of-function alleles in patients with chronic mucocutaneous candidiasis. Journal of Leukocyte Biology, 2013, 95, 667-676.	1.5	77
92	Unique and shared signaling pathways cooperate to regulate the differentiation of human CD4+ T cells into distinct effector subsets. Journal of Experimental Medicine, 2016, 213, 1589-1608.	4.2	77
93	Mutations in the gene for the IL-7 receptor result in Tâ€“B+NK+ severe combined immunodeficiency disease. Current Opinion in Immunology, 2000, 12, 468-473.	2.4	76
94	Invasive Pneumococcal Disease in Children Can Reveal a Primary Immunodeficiency. Clinical Infectious Diseases, 2014, 59, 244-251.	2.9	75
95	A narrow repertoire of transcriptional modules responsive to pyogenic bacteria is impaired in patients carrying loss-of-function mutations in MYD88 or IRAK4. Nature Immunology, 2014, 15, 1134-1142.	7.0	75
96	Isolated Congenital Asplenia: A French Nationwide Retrospective Survey of 20 Cases. Journal of Pediatrics, 2011, 158, 142-148.e1.	0.9	74
97	A Fast Procedure for the Detection of Defects in Toll-like Receptor Signaling. Pediatrics, 2006, 118, 2498-2503.	1.0	71
98	Posaconazole Treatment of Extensive Skin and Nail Dermatophytosis Due to Autosomal Recessive Deficiency of CARD9. JAMA Dermatology, 2015, 151, 192.	2.0	71
99	Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. Journal of Clinical Immunology, 2016, 36, 220-234.	2.0	71
100	Molecular mechanisms of mucocutaneous immunity against Candida and Staphylococcus species. Journal of Allergy and Clinical Immunology, 2012, 130, 1019-1027.	1.5	68
101	Human Adaptive Immunity Rescues an Inborn Error of Innate Immunity. Cell, 2017, 168, 789-800.e10.	13.5	68
102	Shigella sonnei Meningitis Due to Interleukin-1 Receptor–Associated Kinase–4 Deficiency: First Association with a Primary Immune Deficiency. Clinical Infectious Diseases, 2005, 40, 1227-1231.	2.9	66
103	NEMO is a key component of NF-ÎºBâ€“ and IRF-3â€“dependent TLR3-mediated immunity to herpes simplex virus. Journal of Allergy and Clinical Immunology, 2011, 128, 610-617.e4.	1.5	66
104	Herpes in STAT1 gain-of-function mutation. Lancet, The, 2012, 379, 2500.	6.3	66
105	Self-reactive VH4-34â€“expressing IgG B cells recognize commensal bacteria. Journal of Experimental Medicine, 2017, 214, 1991-2003.	4.2	66
106	Primary immunodeficiencies of protective immunity to primary infections. Clinical Immunology, 2010, 135, 204-209.	1.4	65
107	Genetic errors of the human caspase recruitment domainâ€“B-cell lymphoma 10â€“mucosa-associated lymphoid tissue lymphoma-translocation gene 1 (CBM) complex: Molecular, immunologic, and clinical heterogeneity. Journal of Allergy and Clinical Immunology, 2015, 136, 1139-1149.	1.5	65
108	Dominant-negative mutations in human IL6ST underlie hyper-IgE syndrome. Journal of Experimental Medicine, 2020, 217, .	4.2	64

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109	A Novel Gain-of-Function IKBA Mutation Underlies Ectodermal Dysplasia with Immunodeficiency and Polyendocrinopathy. Journal of Clinical Immunology, 2013, 33, 1088-1099.	2.0	60
110	HumanÂinborn errors of immunity underlying superficial or invasive candidiasis. Human Genetics, 2020, 139, 1011-1022.	1.8	59
111	Human Î¹BÎ± Gain of Function: a Severe and Syndromic Immunodeficiency. Journal of Clinical Immunology, 2017, 37, 397-412.	2.0	58
112	Human primary immunodeficiencies of type I interferons. Biochimie, 2007, 89, 878-883.	1.3	57
113	Human hyper-IgE syndrome: singular or plural?. Mammalian Genome, 2018, 29, 603-617.	1.0	55
114	Human autoantibodies underlying infectious diseases. Journal of Experimental Medicine, 2022, 219, .	4.2	55
115	A 1-Year-Old Girl with a Gain-of-Function STAT1 Mutation Treated with Hematopoietic Stem Cell Transplantation. Journal of Clinical Immunology, 2013, 33, 1273-1275.	2.0	54
116	Whole-exome sequencing to analyze population structure, parental inbreeding, and familial linkage. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 6713-6718.	3.3	53
117	Successful Allogeneic Hemopoietic Stem Cell Transplantation in a Child Who Had Anhidrotic Ectodermal Dysplasia With Immunodeficiency. Pediatrics, 2006, 118, e205-e211.	1.0	52
118	New mechanism of X-linked anhidrotic ectodermal dysplasia with immunodeficiency: impairment of ubiquitin binding despite normal folding of NEMO protein. Blood, 2011, 118, 926-935.	0.6	52
119	A Mild Form of SLC29A3 Disorder: A Frameshift Deletion Leads to the Paradoxical Translation of an Otherwise Noncoding mRNA Splice Variant. PLoS ONE, 2012, 7, e29708.	1.1	50
120	Interleukin 1/Toll-like Receptor-induced Autophosphorylation Activates Interleukin 1 Receptor-associated Kinase 4 and Controls Cytokine Induction in a Cell Type-specific Manner. Journal of Biological Chemistry, 2014, 289, 10865-10875.	1.6	50
121	Inherited human IRAK-1 deficiency selectively impairs TLR signaling in fibroblasts. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E514-E523.	3.3	49
122	Autosomal recessive Interleukin-1 receptor-associated kinase 4 deficiency in fourth-degree relatives. Journal of Pediatrics, 2006, 148, 549-551.	0.9	48
123	A CARD9 Founder Mutation Disrupts NF-Î¹B Signaling by Inhibiting BCL10 and MALT1 Recruitment and Signalosome Formation. Frontiers in Immunology, 2018, 9, 2366.	2.2	46
124	Lessons learned from the study of human inborn errors of innate immunity. Journal of Allergy and Clinical Immunology, 2019, 143, 507-527.	1.5	46
125	Septicemia without Sepsis: Inherited Disorders of Nuclear Factor-kB-Mediated Inflammation. Clinical Infectious Diseases, 2005, 41, S436-S439.	2.9	45
126	A Variety of Alu-Mediated Copy Number Variations Can Underlie IL-12RÎ²1 Deficiency. Journal of Clinical Immunology, 2018, 38, 617-627.	2.0	45

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127	Chronic mucocutaneous candidiasis and connective tissue disorder in humans with impaired JNK1-dependent responses to IL-17A/F and TGF-Î². Science Immunology, 2019, 4, .	5.6	45
128	Human inborn errors of immunity to infection affecting cells other than leukocytes: from the immune system to the whole organism. Current Opinion in Immunology, 2019, 59, 88-100.	2.4	44
129	Three Copies of Four Interferon Receptor Genes Underlie a Mild Type I Interferonopathy in Down Syndrome. Journal of Clinical Immunology, 2020, 40, 807-819.	2.0	44
130	Alanine-scanning mutagenesis of human signal transducer and activator of transcription 1 to estimate loss- or gain-of-function variants. Journal of Allergy and Clinical Immunology, 2017, 140, 232-241.	1.5	43
131	IRAK-4 Mutation (Q293X): Rapid Detection and Characterization of Defective Post-Transcriptional TLR/IL-1R Responses in Human Myeloid and Non-Myeloid Cells. Journal of Immunology, 2006, 177, 8202-8211.	0.4	42
132	A global effort to dissect the human genetic basis of resistance to SARS-CoV-2 infection. Nature Immunology, 2022, 23, 159-164.	7.0	41
133	Inherited human IRAK-4 deficiency: an update. Immunologic Research, 2007, 38, 347-352.	1.3	40
134	Early-Onset Invasive Infection Due to Corynespora cassiicola Associated with Compound Heterozygous CARD9 Mutations in a Colombian Patient. Journal of Clinical Immunology, 2018, 38, 794-803.	2.0	40
135	Rescue of recurrent deep intronic mutation underlying cell typeâ€“dependent quantitative NEMO deficiency. Journal of Clinical Investigation, 2018, 129, 583-597.	3.9	38
136	A novel form of cell type-specific partial IFN-Î³R1 deficiency caused by a germ line mutation of the IFNGR1 initiation codon. Human Molecular Genetics, 2010, 19, 434-444.	1.4	36
137	Clinical and Molecular Findings in Mendelian Susceptibility to Mycobacterial Diseases: Experience From India. Frontiers in Immunology, 2021, 12, 631298.	2.2	36
138	Vaccine breakthrough hypoxemic COVID-19 pneumonia in patients with auto-Abs neutralizing type I IFNs. Science Immunology, 2023, 8, .	5.6	35
139	Successful Allogenic Stem Cell Transplantation in Patients with Inherited CARD9 Deficiency. Journal of Clinical Immunology, 2019, 39, 462-469.	2.0	34
140	A toxic palmitoylation of Cdc42 enhances NF-ÎºB signaling and drives a severe autoinflammatory syndrome. Journal of Allergy and Clinical Immunology, 2020, 146, 1201-1204.e8.	1.5	33
141	Negative selection on human genes underlying inborn errors depends on disease outcome and both the mode and mechanism of inheritance. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	33
142	Biochemically deleterious human NFKB1 variants underlie an autosomal dominant form of common variable immunodeficiency. Journal of Experimental Medicine, 2021, 218, .	4.2	32
143	Familial NK Cell Deficiency Associated with Impaired IL-2- and IL-15-Dependent Survival of Lymphocytes. Journal of Immunology, 2006, 177, 8835-8843.	0.4	31
144	Epithelial barrier dysfunction in desmoglein-1 deficiency. Journal of Allergy and Clinical Immunology, 2018, 142, 702-706.e7.	1.5	31

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145	Incomplete penetrance for isolated congenital asplenia in humans with mutations in translated and untranslated RPSA exons. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E8007-E8016.	3.3	31
146	Inherited GATA2 Deficiency Is Dominant by Haploinsufficiency and Displays Incomplete Clinical Penetrance. Journal of Clinical Immunology, 2021, 41, 639-657.	2.0	30
147	Human STAT3 variants underlie autosomal dominant hyper-IgE syndrome by negative dominance. Journal of Experimental Medicine, 2021, 218, .	4.2	30
148	Gain-of-Function Mutations inSTAT1: A Recently Defined Cause for Chronic Mucocutaneous Candidiasis Disease Mimicking Combined Immunodeficiencies. Case Reports in Immunology, 2017, 2017, 1-6.	0.2	29
149	STAT1 Gain-of-Function and Dominant Negative STAT3 Mutations Impair IL-17 and IL-22 Immunity Associated with CMC. Journal of Investigative Dermatology, 2018, 138, 711-714.	0.3	29
150	Efficacy of Dupilumab for Controlling Severe Atopic Dermatitis in a Patient with Hyper-IgE Syndrome. Journal of Clinical Immunology, 2020, 40, 418-420.	2.0	28
151	Heritable defects of the human TLR signalling pathways. Journal of Endotoxin Research, 2005, 11, 220-224.	2.5	27
152	Very Lateâ€Onset Group BStreptococcusMeningitis, Sepsis, and Systemic Shigellosis due to Interleukinâ€1 Receptorâ€“Associated Kinaseâ€4 Deficiency. Clinical Infectious Diseases, 2009, 49, 1393-1396.	2.9	27
153	Mechanism of dysfunction of human variants of the IRAK4 kinase and a role for its kinase activity in interleukin-1 receptor signaling. Journal of Biological Chemistry, 2018, 293, 15208-15220.	1.6	27
154	Aspergillus fumigatus Infection in Humans With STAT3-Deficiency Is Associated With Defective Interferon-Gamma and Th17 Responses. Frontiers in Immunology, 2020, 11, 38.	2.2	26
155	Orf Infection in a Patient with Stat1 Gain-of-Function. Journal of Clinical Immunology, 2015, 35, 80-83.	2.0	25
156	Inherited CARD9 Deficiency in a Patient with Both Exophiala spinifera and Aspergillus nomius Severe Infections. Journal of Clinical Immunology, 2020, 40, 359-366.	2.0	25
157	High Th2 cytokine levels and upper airway inflammation in human inherited T-bet deficiency. Journal of Experimental Medicine, 2021, 218, .	4.2	25
158	Human OTULIN haploinsufficiency impairs cell-intrinsic immunity to staphylococcal Î±-toxin. Science, 2022, 376, eabm6380.	6.0	25
159	IRAK4 Deficiency in a Patient with Recurrent Pneumococcal Infections: Case Report and Review of the Literature. Frontiers in Pediatrics, 2017, 5, 83.	0.9	24
160	Impaired respiratory burst contributes to infections in PKCÎ´-deficient patients. Journal of Experimental Medicine, 2021, 218, .	4.2	23
161	The Differential Regulation of Human ACT1 Isoforms by Hsp90 in IL-17 Signaling. Journal of Immunology, 2014, 193, 1590-1599.	0.4	22
162	IL-17 T Cellsâ€™ Defective Differentiation In Vitro Despite Normal Range Ex Vivo in Chronic Mucocutaneous Candidiasis Due to STAT1 Mutation. Journal of Investigative Dermatology, 2014, 134, 1155-1157.	0.3	21

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163	Disseminated Tuberculosis and Chronic Mucocutaneous Candidiasis in a Patient with a Gain-of-Function Mutation in Signal Transduction and Activator of Transcription 1. Frontiers in Immunology, 2017, 8, 1651.	2.2	21
164	Inherited human c-Rel deficiency disrupts myeloid and lymphoid immunity to multiple infectious agents. Journal of Clinical Investigation, 2021, 131, .	3.9	21
165	Identification of discriminative gene-level and protein-level features associated with pathogenic gain-of-function and loss-of-function variants. American Journal of Human Genetics, 2021, 108, 2301-2318.	2.6	21
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