Anne Puel

List of Publications by Year
in descending order

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Version: 2024-02-01

219 papers	32,435 citations	5268 83 h-index	4432 172 g-index
235 all docs	235 docs citations	235 times ranked	29932 citing authors

#	Article	IF	Citations
1	Vaccine breakthrough hypoxemic COVID-19 pneumonia in patients with auto-Abs neutralizing type I IFNs. Science Immunology, 2023, 8, .	11.9	35
2	A global effort to dissect the human genetic basis of resistance to SARS-CoV-2 infection. Nature Immunology, 2022, 23, 159-164.	14.5	41
3	CARD9 Expression Pattern, Gene Dosage, and Immunodeficiency Phenotype Revisited. Journal of Clinical Immunology, 2022, 42, 336-349.	3.8	6
4	Human genetic and immunological determinants of critical COVID-19 pneumonia. Nature, 2022, 603, 587-598.	27.8	216
5	Human autoantibodies underlying infectious diseases. Journal of Experimental Medicine, 2022, 219, .	8.5	55
6	Chronic mucocutaneous candidiasis with severe oral injury associated with aÂSTAT 1Âgain-of-function mutation. Advances in Oral and Maxillofacial Surgery, 2022, 6, 100272.	0.3	1
7	Inborn Errors of Immunity in Algerian Children and Adults: A Single-Center Experience Over a Period of 13 Years (2008â€“2021). Frontiers in Immunology, 2022, 13, 900091.	4.8	4
8	Invasive Rhinosinusitis Caused by Alternaria infectoria in a Patient with Autosomal Recessive CARD9 Deficiency and a Review of the Literature. Journal of Fungi (Basel, Switzerland), 2022, 8, 446.	3.5	2
9	Case Report: Invasive Cryptococcosis in French Guiana: Immune and Genetic Investigation in Six Non-HIV Patients. Frontiers in Immunology, 2022, 13, 881352.	4.8	1
10	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.	7.1	110
11	Chronic Granulomatous Disease-Like Presentation of a Child with Autosomal Recessive PKCÎ´ Deficiency. Journal of Clinical Immunology, 2022, 42, 1244-1253.	3.8	6
12	Human OTULIN haploinsufficiency impairs cell-intrinsic immunity to staphylococcal Î±-toxin. Science, 2022, 376, eabm6380.	12.6	25
13	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.	3.8	389
14	Functional analysis of two STAT1 gain-of-function mutations in two Iranian families with autosomal dominant chronic mucocutaneous candidiasis. Medical Mycology, 2021, 59, 180-188.	0.7	4
15	IRAK4 Deficiency Presenting with Anti-NMDAR Encephalitis and HHV6 Reactivation. Journal of Clinical Immunology, 2021, 41, 125-135.	3.8	10
16	A new case of deep dermatophytic disease with inherited CARD9 deficiency. International Journal of Dermatology, 2021, 60, e15-e16.	1.0	3
17	SARS-CoV-2 induces human plasmacytoid predendritic cell diversification via UNC93B and IRAK4. Journal of Experimental Medicine, 2021, 218, .	8.5	107
18	Inherited GATA2 Deficiency Is Dominant by Haploinsufficiency and Displays Incomplete Clinical Penetrance. Journal of Clinical Immunology, 2021, 41, 639-657.	3.8	30

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19	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, .	7.1	33
20	Pediatric Demodicosis Associated with Gain-of-Function Variant in STAT1 Presenting as Rosacea-Type Rash. Journal of Clinical Immunology, 2021, 41, 698-700.	3.8	11
21	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.	3.8	165
22	Clinical and Molecular Findings in Mendelian Susceptibility to Mycobacterial Diseases: Experience From India. Frontiers in Immunology, 2021, 12, 631298.	4.8	36
23	Auto-antibodies to type I IFNs can underlie adverse reactions to yellow fever live attenuated vaccine. Journal of Experimental Medicine, 2021, 218, .	8.5	130
24	Inherited CARD9 Deficiency in a Child with Invasive Disease Due to Exophiala dermatitidis and Two Older but Asymptomatic Siblings. Journal of Clinical Immunology, 2021, 41, 975-986.	3.8	15
25	Human gut mycobiota tune immunity via CARD9-dependent induction of anti-fungal IgG antibodies. Cell, 2021, 184, 1017-1031.e14.	28.9	113
26	A Novel TRAF3IP2 Mutation Causing Chronic Mucocutaneous Candidiasis. Journal of Clinical Immunology, 2021, 41, 1376-1379.	3.8	11
27	SARS-CoV-2â€“related MIS-C: A key to the viral and genetic causes of Kawasaki disease?. Journal of Experimental Medicine, 2021, 218, .	8.5	100
28	Preexisting autoantibodies to type I IFNs underlie critical COVID-19 pneumonia in patients with APS-1. Journal of Experimental Medicine, 2021, 218, .	8.5	185
29	Detection of homozygous and hemizygous complete or partial exon deletions by whole-exome sequencing. NAR Genomics and Bioinformatics, 2021, 3, lqab037.	3.2	7
30	Human STAT3 variants underlie autosomal dominant hyper-IgE syndrome by negative dominance. Journal of Experimental Medicine, 2021, 218, .	8.5	30
31	High Th2 cytokine levels and upper airway inflammation in human inherited T-bet deficiency. Journal of Experimental Medicine, 2021, 218, .	8.5	25
32	Case Report: Interleukin-2 Receptor Common Gamma Chain Defect Presented as a Hyper-IgE Syndrome. Frontiers in Immunology, 2021, 12, 696350.	4.8	3
33	Impaired respiratory burst contributes to infections in PKCÎ´-deficient patients. Journal of Experimental Medicine, 2021, 218, .	8.5	23
34	Biallelic TRAF3IP2 variants causing chronic mucocutaneous candidiasis in a child harboring a STAT1 variant. Pediatric Allergy and Immunology, 2021, 32, 1804-1812.	2.6	7
35	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, .	11.9	357
36	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. Science Immunology, 2021, 6, .	11.9	267

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37	Inherited human c-Rel deficiency disrupts myeloid and lymphoid immunity to multiple infectious agents. Journal of Clinical Investigation, 2021, 131, .	8.2	21
38	Biochemically deleterious human NFKB1 variants underlie an autosomal dominant form of common variable immunodeficiency. Journal of Experimental Medicine, 2021, 218, .	8.5	32
39	Comment on â€œAberrant type 1 immunity drives susceptibility to mucosal fungal infectionsâ€. Science, 2021, 373, eabi5459.	12.6	8
40	Candidiasis in patients with APS-1: low IL-17, high IFN-Î³, or both?. Current Opinion in Immunology, 2021, 72, 318-323.	5.5	8
41	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.	6.2	21
42	Case Report: A New Gain-of-Function Mutation of STAT1 Identified in a Patient With Chronic Mucocutaneous Candidiasis and Rosacea-Like Demodicosis: An Emerging Association. Frontiers in Immunology, 2021, 12, 760019.	4.8	11
43	Delay in the Diagnosis of APECED: A Case Report and Review of Literature from Iran. Immunological Investigations, 2020, 49, 299-306.	2.0	3
44	Ruxolitinib Response in an Infant With Veryâ€earlyâ€onset Inflammatory Bowel Disease and Gainâ€ofâ€function STAT1 Mutation. Journal of Pediatric Gastroenterology and Nutrition, 2020, 71, e132-e133.	1.8	9
45	Disseminated Infectious Disease Caused by Histoplasma capsulatum in an Adult Patient as First Manifestation of Inherited IL-12RÎ²1 Deficiency. Journal of Clinical Immunology, 2020, 40, 1051-1054.	3.8	8
46	Dominant-negative mutations in human IL6ST underlie hyper-IgE syndrome. Journal of Experimental Medicine, 2020, 217, .	8.5	64
47	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science, 2020, 370, .	12.6	1,749
48	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. Science, 2020, 370, .	12.6	1,983
49	Human STAT1 Gain-of-Function Heterozygous Mutations: Chronic Mucocutaneous Candidiasis and Type I Interferonopathy. Journal of Clinical Immunology, 2020, 40, 1065-1081.	3.8	86
50	Human T-bet Governs Innate and Innate-like Adaptive IFN-Î³ Immunity against Mycobacteria. Cell, 2020, 183, 1826-1847.e31.	28.9	83
51	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. Cell, 2020, 181, 1194-1199.	28.9	185
52	Three Copies of Four Interferon Receptor Genes Underlie a Mild Type I Interferonopathy in Down Syndrome. Journal of Clinical Immunology, 2020, 40, 807-819.	3.8	44
53	Anti-GM-CSF Autoantibodies and Cryptococcus neoformans var. grubii CNS Vasculitis. Journal of Clinical Immunology, 2020, 40, 767-769.	3.8	11
54	The IL1RN Mutation Creating the Most-Upstream Premature Stop Codon Is Hypomorphic Because of a Reinitiation of Translation. Journal of Clinical Immunology, 2020, 40, 643-645.	3.8	1

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55	Inherited disorders of TLR, IL-1R, and NFÎºB immunity. , 2020, , 869-883.		1
56	HumanÂinborn errors of immunity underlying superficial or invasive candidiasis. Human Genetics, 2020, 139, 1011-1022.	3.8	59
57	Inherited CARD9 Deficiency in a Patient with Both Exophiala spinifera and Aspergillus nomius Severe Infections. Journal of Clinical Immunology, 2020, 40, 359-366.	3.8	25
58	Large-scale genome mining allows identification of neutral polymorphisms and novel resistance mutations in genes involved in Candida albicans resistance to azoles and echinocandins. Journal of Antimicrobial Chemotherapy, 2020, 75, 835-848.	3.0	13
59	Human BCL10 Deficiency due to Homozygosity for a Rare Allele. Journal of Clinical Immunology, 2020, 40, 388-398.	3.8	17
60	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.	2.9	33
61	Efficacy of Dupilumab for Controlling Severe Atopic Dermatitis in a Patient with Hyper-IgE Syndrome. Journal of Clinical Immunology, 2020, 40, 418-420.	3.8	28
62	Aspergillus fumigatus Infection in Humans With STAT3-Deficiency Is Associated With Defective Interferon-Gamma and Th17 Responses. Frontiers in Immunology, 2020, 11, 38.	4.8	26
63	Immunodeficiencies at the Interface of Innate and Adaptive Immunity. , 2019, , 509-522.e1.		0
64	A deep intronic splice mutation of STAT3 underlies hyper IgE syndrome by negative dominance. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 16463-16472.	7.1	17
65	Deficiency of Interleukin-1 Receptor Antagonist: A Case with Late Onset Severe Inflammatory Arthritis, Nail Psoriasis with Onychomycosis and Well Responsive to Adalimumab Therapy. Case Reports in Immunology, 2019, 2019, 1-6.	0.4	8
66	Dominant negative CARD11 mutations: Beyond atopy. Journal of Allergy and Clinical Immunology, 2019, 143, 1345-1347.	2.9	8
67	Successful Allogenic Stem Cell Transplantation in Patients with Inherited CARD9 Deficiency. Journal of Clinical Immunology, 2019, 39, 462-469.	3.8	34
68	The nature of human IL-6. Journal of Experimental Medicine, 2019, 216, 1969-1971.	8.5	18
69	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.	5.5	44
70	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, .	11.9	45
71	A 7-Year-Old Child With Headaches and Prolonged Fever Associated With Oral and Nail Lesions. Open Forum Infectious Diseases, 2019, 6, ofz229.	0.9	6
72	Lessons learned from the study of human inborn errors of innate immunity. Journal of Allergy and Clinical Immunology, 2019, 143, 507-527.	2.9	46

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73	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.	0.7	29
74	A CARD9 Founder Mutation Disrupts NF-ÎºB Signaling by Inhibiting BCL10 and MALT1 Recruitment and Signalosome Formation. Frontiers in Immunology, 2018, 9, 2366.	4.8	46
75	Somatic alterations compromised molecular diagnosis of DOCK8 hyper-IgE syndrome caused by a novel intronic splice site mutation. Scientific Reports, 2018, 8, 16719.	3.3	5
76	Human IFN-Î³ immunity to mycobacteria is governed by both IL-12 and IL-23. Science Immunology, 2018, 3, .	11.9	152
77	Tuberculosis and impaired IL-23â€“dependent IFN-Î³ immunity in humans homozygous for a common TYK2 missense variant. Science Immunology, 2018, 3, .	11.9	148
78	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.	3.8	40
79	Arid5a makes the IL-17A/Fâ€“responsive pathway less arid. Science Signaling, 2018, 11, .	3.6	5
80	Epithelial barrier dysfunction in desmoglein-1 deficiency. Journal of Allergy and Clinical Immunology, 2018, 142, 702-706.e7.	2.9	31
81	A novel AIRE gene mutation in a patient with autoimmune polyendocrinopathy candidiasis and ectodermal dystrophy revealed by alopecia areata. JAAD Case Reports, 2018, 4, 602-605.	0.8	2
82	A Variety of Alu-Mediated Copy Number Variations Can Underlie IL-12RÎ²1 Deficiency. Journal of Clinical Immunology, 2018, 38, 617-627.	3.8	45
83	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.	3.4	27
84	Inherited CARD9 Deficiency: Invasive Disease Caused by Ascomycete Fungi in Previously Healthy Children and Adults. Journal of Clinical Immunology, 2018, 38, 656-693.	3.8	130
85	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.	7.1	31
86	Human hyper-IgE syndrome: singular or plural?. Mammalian Genome, 2018, 29, 603-617.	2.2	55
87	A recessive form of hyper-IgE syndrome by disruption of ZNF341-dependent STAT3 transcription and activity. Science Immunology, 2018, 3, .	11.9	132
88	ZNF341 controls STAT3 expression and thereby immunocompetence. Science Immunology, 2018, 3, .	11.9	113
89	Rescue of recurrent deep intronic mutation underlying cell typeâ€“dependent quantitative NEMO deficiency. Journal of Clinical Investigation, 2018, 129, 583-597.	8.2	38
90	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.	7.1	49

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91	Systemic Human ILC Precursors Provide a Substrate for Tissue ILC Differentiation. Cell, 2017, 168, 1086-1100.e10.	28.9	420
92	Human Adaptive Immunity Rescues an Inborn Error of Innate Immunity. Cell, 2017, 168, 789-800.e10.	28.9	68
93	Primary Immunodeficiencies and Dermatophytosis. , 2017, , 121-133.		5
94	Self-reactive VH4-34â€“expressing IgG B cells recognize commensal bacteria. Journal of Experimental Medicine, 2017, 214, 1991-2003.	8.5	66
95	Human Ĩº BÎ± Gain of Function: a Severe and Syndromic Immunodeficiency. Journal of Clinical Immunology, 2017, 37, 397-412.	3.8	58
96	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.	2.9	43
97	A gain-of-function mutation of STAT1: A novel genetic factor contributing to chronic mucocutaneous candidiasis. Acta Microbiologica Et Immunologica Hungarica, 2017, 64, 191-201.	0.8	18
98	Inborn errors of immunity underlying fungal diseases in otherwise healthy individuals. Current Opinion in Microbiology, 2017, 40, 46-57.	5.1	101
99	Hematopoietic stem cell transplantation in 29 patients hemizygous for hypomorphic IKBKG/NEMO mutations. Blood, 2017, 130, 1456-1467.	1.4	95
100	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.	4.8	21
101	IRAK4 Deficiency in a Patient with Recurrent Pneumococcal Infections: Case Report and Review of the Literature. Frontiers in Pediatrics, 2017, 5, 83.	1.9	24
102	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.	0.4	29
103	Mendelian Susceptibility to Infections with Viruses, Mycobacteria, Bacteria, andÂ Candida. , 2016, , 407-415.		0
104	Severe Dermatophytosis and Acquired or Innate Immunodeficiency: A Review. Journal of Fungi (Basel,) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	3.5	84
105	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.	8.5	77
106	Novel <scp>STAT</scp>1 gainâ€ofâ€function mutation and suppurative infections. Pediatric Allergy and Immunology, 2016, 27, 220-223.	2.6	14
107	Chronic mucocutaneous candidiasis disease associated with inborn errors of ILâ€17 immunity. Clinical and Translational Immunology, 2016, 5, e114.	3.8	148
108	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.	7.1	137

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109	Heterozygous STAT1 gain-of-function mutations underlie an unexpectedly broad clinical phenotype. Blood, 2016, 127, 3154-3164.	1.4	465
110	Dual T cellâ€“ and B cellâ€“intrinsic deficiency in humans with biallelic RLTPR mutations. Journal of Experimental Medicine, 2016, 213, 2413-2435.	8.5	117
111	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.	7.1	53
112	Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. Journal of Clinical Immunology, 2016, 36, 220-234.	3.8	71
113	Chronic and Invasive Fungal Infections in a Family with CARD9 Deficiency. Journal of Clinical Immunology, 2016, 36, 204-209.	3.8	98
114	Posaconazole Treatment of Extensive Skin and Nail Dermatophytosis Due to Autosomal Recessive Deficiency of CARD9. JAMA Dermatology, 2015, 151, 192.	4.1	71
115	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.	2.9	208
116	Orf Infection in a Patient with Stat1 Gain-of-Function. Journal of Clinical Immunology, 2015, 35, 80-83.	3.8	25
117	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.	2.9	181
118	Impairment of immunity to Candida and Mycobacterium in humans with bi-allelic RORC mutations. Science, 2015, 349, 606-613.	12.6	366
119	A Homozygous CARD9 Mutation in a Brazilian Patient with Deep Dermatophytosis. Journal of Clinical Immunology, 2015, 35, 486-490.	3.8	89
120	Inherited IL-17RC deficiency in patients with chronic mucocutaneous candidiasis. Journal of Experimental Medicine, 2015, 212, 619-631.	8.5	162
121	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.	7.1	213
122	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.	2.9	65
123	Human TYK2 deficiency: Mycobacterial and viral infections without hyper-IgE syndrome. Journal of Experimental Medicine, 2015, 212, 1641-1662.	8.5	293
124	Inherited CARD9 Deficiency in 2 Unrelated Patients With Invasive Exophiala Infection. Journal of Infectious Diseases, 2015, 211, 1241-1250.	4.0	141
125	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.	0.7	21
126	Addressing diagnostic challenges in primary immunodeficiencies: Laboratory evaluation of Toll-like receptor- and NF-Î°B-mediated immune responses. Critical Reviews in Clinical Laboratory Sciences, 2014, 51, 112-123.	6.1	20

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127	Clinical Features of Candidiasis in Patients With Inherited Interleukin 12 Receptor Î²1 Deficiency. Clinical Infectious Diseases, 2014, 58, 204-213.	5.8	98
128	Invasive Pneumococcal Disease in Children Can Reveal a Primary Immunodeficiency. Clinical Infectious Diseases, 2014, 59, 244-251.	5.8	75
129	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.	14.5	75
130	The Differential Regulation of Human ACT1 Isoforms by Hsp90 in IL-17 Signaling. Journal of Immunology, 2014, 193, 1590-1599.	0.8	22
131	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.	3.4	50
132	A Novel Gain-of-Function IKBA Mutation Underlies Ectodermal Dysplasia with Immunodeficiency and Polyendocrinopathy. Journal of Clinical Immunology, 2013, 33, 1088-1099.	3.8	60
133	The Role of Human IL-17 Immunity in Fungal Disease. Current Fungal Infection Reports, 2013, 7, 132-137.	2.6	2
134	An ACT1 Mutation Selectively Abolishes Interleukin-17 Responses in Humans with Chronic Mucocutaneous Candidiasis. Immunity, 2013, 39, 676-686.	14.3	262
135	Deep Dermatophytosis and Inherited CARD9 Deficiency. New England Journal of Medicine, 2013, 369, 1704-1714.	27.0	362
136	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.	3.8	54
137	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.	3.2	105
138	Primary immunodeficiencies underlying fungal infections. Current Opinion in Pediatrics, 2013, 25, 736-747.	2.0	190
139	Ribosomal Protein SA Haploinsufficiency in Humans with Isolated Congenital Asplenia. Science, 2013, 340, 976-978.	12.6	176
140	Autoantibodies against cytokines: back to human genetics. Blood, 2013, 121, 1246-1247.	1.4	14
141	Simple diagnosis ofSTAT1gain-of-function alleles in patients with chronic mucocutaneous candidiasis. Journal of Leukocyte Biology, 2013, 95, 667-676.	3.3	77
142	Inherited disorders of IFN-Î³-, IFN-Î±/Î²/Î»-, and NF-ÎºB-mediated immunity. , 2013, , 454-464.		1
143	Inborn errors of human IL-17 immunity underlie chronic mucocutaneous candidiasis. Current Opinion in Allergy and Clinical Immunology, 2012, 12, 616-622.	2.3	288
144	Autosomal Dominant STAT3 Deficiency and Hyper-IgE Syndrome. Medicine (United States), 2012, 91, e1-e19.	1.0	274

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145	Herpes in STAT1 gain-of-function mutation. Lancet, The, 2012, 379, 2500.	13.7	66
146	Immunodeficiency, autoinflammation and amylopectinosis in humans with inherited HOIL-1 and LUBAC deficiency. Nature Immunology, 2012, 13, 1178-1186.	14.5	410
147	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.	1.4	87
148	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.	2.9	169
149	Mycobacterial Disease and Impaired IFN-Î³ Immunity in Humans with Inherited ISG15 Deficiency. Science, 2012, 337, 1684-1688.	12.6	455
150	Immunity to infection in <scp>IL</scp>â€17â€deficient mice and humans. European Journal of Immunology, 2012, 42, 2246-2254.	2.9	167
151	Molecular mechanisms of mucocutaneous immunity against Candida and Staphylococcus species. Journal of Allergy and Clinical Immunology, 2012, 130, 1019-1027.	2.9	68
152	Inborn errors of human STAT1: allelic heterogeneity governs the diversity of immunological and infectious phenotypes. Current Opinion in Immunology, 2012, 24, 364-378.	5.5	245
153	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.	2.5	50
154	Chronic Mucocutaneous Candidiasis in Humans with Inborn Errors of Interleukin-17 Immunity. Science, 2011, 332, 65-68.	12.6	1,482
155	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.	2.9	66
156	PS2-010. Human TIRAP deficiency in eight individuals from a large consanguineous family: A cause for predisposition to staphylococcal diseases?. Cytokine, 2011, 56, 66.	3.2	0
157	CS16-7. A novel autosomal recessive and autosomal dominant deficiency in the TLR3 pathway underlying susceptibility to Herpes Simplex Encephalitis. Cytokine, 2011, 56, 106.	3.2	0
158	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.	1.4	52
159	Tartrate-resistant acid phosphatase deficiency causes a bone dysplasia with autoimmunity and a type I interferon expression signature. Nature Genetics, 2011, 43, 127-131.	21.4	214
160	Germline CYBB mutations that selectively affect macrophages in kindreds with X-linked predisposition to tuberculous mycobacterial disease. Nature Immunology, 2011, 12, 213-221.	14.5	248
161	Isolated Congenital Asplenia: A French Nationwide Retrospective Survey of 20 Cases. Journal of Pediatrics, 2011, 158, 142-148.e1.	1.8	74
162	Interleukin-36â€“Receptor Antagonist Deficiency and Generalized Pustular Psoriasis. New England Journal of Medicine, 2011, 365, 620-628.	27.0	836

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163	Infectious Diseases in Patients with IRAK-4, MyD88, NEMO, or IÎºBÎ± Deficiency. Clinical Microbiology Reviews, 2011, 24, 490-497.	13.6	349
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