

# Capucine Picard

## List of Publications by Year in descending order

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362  
papers

44,866  
citations

997

114  
h-index

2385

198  
g-index

384  
all docs

384  
docs citations

384  
times ranked

36196  
citing authors

#	ARTICLE	IF	CITATIONS
1	Chronic Mucocutaneous Candidiasis in Humans with Inborn Errors of Interleukin-17 Immunity. <i>Science</i> , 2011, 332, 65-68.	12.6	1,482
2	Human CD14 <sup>dim</sup> Monocytes Patrol and Sense Nucleic Acids and Viruses via TLR7 and TLR8 Receptors. <i>Immunity</i> , 2010, 33, 375-386.	14.3	1,060
3	TLR3 Deficiency in Patients with Herpes Simplex Encephalitis. <i>Science</i> , 2007, 317, 1522-1527.	12.6	970
4	Human Inborn Errors of Immunity: 2019 Update on the Classification from the International Union of Immunological Societies Expert Committee. <i>Journal of Clinical Immunology</i> , 2020, 40, 24-64.	3.8	881
5	Pyogenic Bacterial Infections in Humans with MyD88 Deficiency. <i>Science</i> , 2008, 321, 691-696.	12.6	844
6	Pyogenic Bacterial Infections in Humans with IRAK-4 Deficiency. <i>Science</i> , 2003, 299, 2076-2079.	12.6	820
7	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.	8.5	739
8	International Union of Immunological Societies: 2017 Primary Immunodeficiency Diseases Committee Report on Inborn Errors of Immunity. <i>Journal of Clinical Immunology</i> , 2018, 38, 96-128.	3.8	732
9	Herpes Simplex Virus Encephalitis in Human UNC-93B Deficiency. <i>Science</i> , 2006, 314, 308-312.	12.6	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.	8.5	663
11	Primary Immunodeficiency Diseases: an Update on the Classification from the International Union of Immunological Societies Expert Committee for Primary Immunodeficiency 2015. <i>Journal of Clinical Immunology</i> , 2015, 35, 696-726.	3.8	621
12	Efficacy of Gene Therapy for X-Linked Severe Combined Immunodeficiency. <i>New England Journal of Medicine</i> , 2010, 363, 355-364.	27.0	561
13	Phosphoinositide 3-Kinase $\hat{\gamma}$ Gene Mutation Predisposes to Respiratory Infection and Airway Damage. <i>Science</i> , 2013, 342, 866-871.	12.6	541
14	Human Inborn Errors of Immunity: 2019 Update of the IUIS Phenotypic Classification. <i>Journal of Clinical Immunology</i> , 2020, 40, 66-81.	3.8	525
15	The 2017 IUIS Phenotypic Classification for Primary Immunodeficiencies. <i>Journal of Clinical Immunology</i> , 2018, 38, 129-143.	3.8	488
16	Primary Immunodeficiency Diseases: An Update on the Classification from the International Union of Immunological Societies Expert Committee for Primary Immunodeficiency. <i>Frontiers in Immunology</i> , 2014, 5, 162.	4.8	466
17	Heterozygous <i>STAT1</i> gain-of-function mutations underlie an unexpectedly broad clinical phenotype. <i>Blood</i> , 2016, 127, 3154-3164.	1.4	465
18	<i>STIM1</i> Mutation Associated with a Syndrome of Immunodeficiency and Autoimmunity. <i>New England Journal of Medicine</i> , 2009, 360, 1971-1980.	27.0	459

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19	Inherited STING-activating mutation underlies a familial inflammatory syndrome with lupus-like manifestations. <i>Journal of Clinical Investigation</i> , 2014, 124, 5516-5520.	8.2	435
20	Clinical features of dominant and recessive interferon $\beta$ receptor 1 deficiencies. <i>Lancet</i> , The, 2004, 364, 2113-2121.	13.7	429
21	Inborn errors of IL-12/23- and IFN- $\beta$ -mediated immunity: molecular, cellular, and clinical features. <i>Seminars in Immunology</i> , 2006, 18, 347-361.	5.6	422
22	Immunodeficiency, autoinflammation and amylopectinosis in humans with inherited HOIL-1 and LUBAC deficiency. <i>Nature Immunology</i> , 2012, 13, 1178-1186.	14.5	410
23	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.	8.5	406
24	Life-threatening influenza and impaired interferon amplification in human IRF7 deficiency. <i>Science</i> , 2015, 348, 448-453.	12.6	389
25	Human Inborn Errors of Immunity: 2022 Update on the Classification from the International Union of Immunological Societies Expert Committee. <i>Journal of Clinical Immunology</i> , 2022, 42, 1473-1507.	3.8	389
26	Clinical spectrum and features of activated phosphoinositide 3-kinase $\gamma$ syndrome: A large patient cohort study. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 139, 597-606.e4.	2.9	377
27	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.	8.5	374
28	Revisiting Human IL-12R $\beta$ 1 Deficiency. <i>Medicine (United States)</i> , 2010, 89, 381-402.	1.0	367
29	Clinical Features and Outcome of Patients With IRAK-4 and MyD88 Deficiency. <i>Medicine (United States)</i> , 2010, 89, 403-425.	1.0	366
30	Impairment of immunity to <i>Candida</i> and <i>Mycobacterium</i> in humans with bi-allelic <i>RORC</i> mutations. <i>Science</i> , 2015, 349, 606-613.	12.6	366
31	Deep Dermatophytosis and Inherited CARD9 Deficiency. <i>New England Journal of Medicine</i> , 2013, 369, 1704-1714.	27.0	362
32	A Modified $\beta$ -Retrovirus Vector for X-Linked Severe Combined Immunodeficiency. <i>New England Journal of Medicine</i> , 2014, 371, 1407-1417.	27.0	358
33	Infectious Diseases in Patients with IRAK-4, MyD88, NEMO, or $\beta$ Deficiency. <i>Clinical Microbiology Reviews</i> , 2011, 24, 490-497.	13.6	349
34	Outcomes Following Gene Therapy in Patients With Severe Wiskott-Aldrich Syndrome. <i>JAMA - Journal of the American Medical Association</i> , 2015, 313, 1550.	7.4	327
35	Clinical similarities and differences of patients with X-linked lymphoproliferative syndrome type 1 (XLP-1/SAP deficiency) versus type 2 (XLP-2/XIAP deficiency). <i>Blood</i> , 2011, 117, 1522-1529.	1.4	320
36	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.	14.3	304

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37	Munc18-2 deficiency causes familial hemophagocytic lymphohistiocytosis type 5 and impairs cytotoxic granule exocytosis in patient NK cells. <i>Journal of Clinical Investigation</i> , 2009, 119, 3765-3773.	8.2	301
38	Long-term outcome and lineage-specific chimerism in 194 patients with Wiskott-Aldrich syndrome treated by hematopoietic cell transplantation in the period 1980-2009: an international collaborative study. <i>Blood</i> , 2011, 118, 1675-1684.	1.4	296
39	The transmembrane activator TACI triggers immunoglobulin class switching by activating B cells through the adaptor MyD88. <i>Nature Immunology</i> , 2010, 11, 836-845.	14.5	295
40	Human TYK2 deficiency: Mycobacterial and viral infections without hyper-IgE syndrome. <i>Journal of Experimental Medicine</i> , 2015, 212, 1641-1662.	8.5	293
41	ORAI1 deficiency and lack of store-operated Ca <sup>2+</sup> entry cause immunodeficiency, myopathy, and ectodermal dysplasia. <i>Journal of Allergy and Clinical Immunology</i> , 2009, 124, 1311-1318.e7.	2.9	289
42	Inborn errors of human IL-17 immunity underlie chronic mucocutaneous candidiasis. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2012, 12, 616-622.	2.3	288
43	DOCK8 Deficiency: Clinical and Immunological Phenotype and Treatment Options - a Review of 136 Patients. <i>Journal of Clinical Immunology</i> , 2015, 35, 189-198.	3.8	284
44	Autosomal Dominant STAT3 Deficiency and Hyper-IgE Syndrome. <i>Medicine (United States)</i> , 2012, 91, e1-e19.	1.0	274
45	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.	8.2	272
46	Inborn errors of interferon (IFN)-mediated immunity in humans: insights into the respective roles of IFN- $\alpha$ /IFN- $\beta$ , IFN- $\gamma$ , and IFN- $\lambda$ in host defense. <i>Immunological Reviews</i> , 2008, 226, 29-40.	6.0	271
47	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.	8.5	268
48	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.	6.2	265
49	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.	8.5	264
50	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.	8.5	262
51	An ACT1 Mutation Selectively Abolishes Interleukin-17 Responses in Humans with Chronic Mucocutaneous Candidiasis. <i>Immunity</i> , 2013, 39, 676-686.	14.3	262
52	Evidence of innate lymphoid cell redundancy in humans. <i>Nature Immunology</i> , 2016, 17, 1291-1299.	14.5	260
53	Herpes simplex encephalitis in children with autosomal recessive and dominant TRIF deficiency. <i>Journal of Clinical Investigation</i> , 2011, 121, 4889-4902.	8.2	254
54	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.	14.5	248

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55	Human TLR-7-, -8-, and -9-Mediated Induction of IFN- $\beta$ and - $\gamma$ Is IRAK-4 Dependent and Redundant for Protective Immunity to Viruses. <i>Immunity</i> , 2005, 23, 465-478.	14.3	245
56	MST1 mutations in autosomal recessive primary immunodeficiency characterized by defective naive T-cell survival. <i>Blood</i> , 2012, 119, 3458-3468.	1.4	244
57	Human HOIP and LUBAC deficiency underlies autoinflammation, immunodeficiency, amylopectinosis, and lymphangiectasia. <i>Journal of Experimental Medicine</i> , 2015, 212, 939-951.	8.5	241
58	A human immunodeficiency caused by mutations in the PIK3R1 gene. <i>Journal of Clinical Investigation</i> , 2014, 124, 3923-3928.	8.2	239
59	Hematopoietic Stem Cell Transplantation in Hemophagocytic Lymphohistiocytosis: A Single-Center Report of 48 Patients. <i>Pediatrics</i> , 2006, 117, e743-e750.	2.1	230
60	Autoimmune and inflammatory manifestations occur frequently in patients with primary immunodeficiencies. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 140, 1388-1393.e8.	2.9	222
61	Long-term outcome after hematopoietic stem cell transplantation of a single-center cohort of 90 patients with severe combined immunodeficiency. <i>Blood</i> , 2009, 113, 4114-4124.	1.4	220
62	Immunotherapy of Familial Hemophagocytic Lymphohistiocytosis With Antithymocyte Globulins: A Single-Center Retrospective Report of 38 Patients. <i>Pediatrics</i> , 2007, 120, e622-e628.	2.1	218
63	Clinical and immunologic phenotype associated with activated phosphoinositide 3-kinase $\gamma$ syndrome 2: A cohort study. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 210-218.e9.	2.9	215
64	Tartrate-resistant acid phosphatase deficiency causes a bone dysplasia with autoimmunity and a type I interferon expression signature. <i>Nature Genetics</i> , 2011, 43, 127-131.	21.4	214
65	Inherited CARD9 deficiency in otherwise healthy children and adults with <i>Candida species</i> -induced meningoencephalitis, colitis, or both. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 135, 1558-1568.e2.	2.9	208
66	Intrinsic antiproliferative activity of the innate sensor STING in T lymphocytes. <i>Journal of Experimental Medicine</i> , 2017, 214, 1769-1785.	8.5	202
67	IRAK-4- and MyD88-Dependent Pathways Are Essential for the Removal of Developing Autoreactive B Cells in Humans. <i>Immunity</i> , 2008, 29, 746-757.	14.3	201
68	Long-Term Remissions of Severe Pemphigus After Rituximab Therapy Are Associated with Prolonged Failure of Desmoglein B Cell Response. <i>Science Translational Medicine</i> , 2013, 5, 175ra30.	12.4	200
69	The 2015 IUIS Phenotypic Classification for Primary Immunodeficiencies. <i>Journal of Clinical Immunology</i> , 2015, 35, 727-738.	3.8	199
70	Human adenylate kinase 2 deficiency causes a profound hematopoietic defect associated with sensorineural deafness. <i>Nature Genetics</i> , 2009, 41, 106-111.	21.4	198
71	Primary immunodeficiencies underlying fungal infections. <i>Current Opinion in Pediatrics</i> , 2013, 25, 736-747.	2.0	190
72	Interleukin (IL)-12 and IL-23 Are Key Cytokines for Immunity against <i>Salmonella</i> in Humans. <i>Journal of Infectious Diseases</i> , 2004, 190, 1755-1757.	4.0	189

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73	Inherited disorders of NF- $\kappa$ B-mediated immunity in man. <i>Current Opinion in Immunology</i> , 2004, 16, 34-41.	5.5	188
74	Exome and genome sequencing for inborn errors of immunity. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 957-969.	2.9	187
75	Homozygous silencing of T-box transcription factor EOMES leads to microcephaly with polymicrogyria and corpus callosum agenesis. <i>Nature Genetics</i> , 2007, 39, 454-456.	21.4	181
76	Monogenic mutations differentially affect the quantity and quality of T follicular helper cells in patients with human primary immunodeficiencies. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 136, 993-1006.e1.	2.9	181
77	Ribosomal Protein SA Haploinsufficiency in Humans with Isolated Congenital Asplenia. <i>Science</i> , 2013, 340, 976-978.	12.6	176
78	DOCK8 deficiency impairs CD8 T cell survival and function in humans and mice. <i>Journal of Experimental Medicine</i> , 2011, 208, 2305-2320.	8.5	175
79	CTP synthase 1 deficiency in humans reveals its central role in lymphocyte proliferation. <i>Nature</i> , 2014, 510, 288-292.	27.8	174
80	Novel STAT1 Alleles in Otherwise Healthy Patients with Mycobacterial Disease. <i>PLoS Genetics</i> , 2006, 2, e131.	3.5	171
81	Experimental and natural infections in <i>Mycobacterium D88</i> and <i>IRAK4</i> deficient mice and humans. <i>European Journal of Immunology</i> , 2012, 42, 3126-3135.	2.9	169
82	Immunity to infection in <i>IL17A</i> deficient mice and humans. <i>European Journal of Immunology</i> , 2012, 42, 2246-2254.	2.9	167
83	The Ever-Increasing Array of Novel Inborn Errors of Immunity: an Interim Update by the IUIS Committee. <i>Journal of Clinical Immunology</i> , 2021, 41, 666-679.	3.8	165
84	Type I interferon-mediated autoinflammation due to DNase II deficiency. <i>Nature Communications</i> , 2017, 8, 2176.	12.8	164
85	Inherited IL-17RC deficiency in patients with chronic mucocutaneous candidiasis. <i>Journal of Experimental Medicine</i> , 2015, 212, 619-631.	8.5	162
86	Immunodeficiency due to mutations in ORAI1 and STIM1. <i>Clinical Immunology</i> , 2010, 135, 169-182.	3.2	159
87	Naive and memory human B cells have distinct requirements for STAT3 activation to differentiate into antibody-secreting plasma cells. <i>Journal of Experimental Medicine</i> , 2013, 210, 2739-2753.	8.5	158
88	Recurrent Staphylococcal Cellulitis and Subcutaneous Abscesses in a Child with Autoantibodies against IL-6. <i>Journal of Immunology</i> , 2008, 180, 647-654.	0.8	154
89	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.	6.0	153
90	A survey of 90 patients with autoimmune lymphoproliferative syndrome related to TNFRSF6 mutation. <i>Blood</i> , 2011, 118, 4798-4807.	1.4	153

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91	Juvenile myelomonocytic leukemia displays mutations in components of the RAS pathway and the PRC2 network. <i>Nature Genetics</i> , 2015, 47, 1334-1340.	21.4	152
92	Inherited IL-12p40 Deficiency. <i>Medicine (United States)</i> , 2013, 92, 109-122.	1.0	151
93	Human RTEL1 deficiency causes Hoyeraal-Hreidarsson syndrome with short telomeres and genome instability. <i>Human Molecular Genetics</i> , 2013, 22, 3239-3249.	2.9	150
94	SCID patients with ARTEMIS vs RAG deficiencies following HCT: increased risk of late toxicity in ARTEMIS-deficient SCID. <i>Blood</i> , 2014, 123, 281-289.	1.4	150
95	Mutations in CDCA7 and HELLS cause immunodeficiency-centromeric instability-facial anomalies syndrome. <i>Nature Communications</i> , 2015, 6, 7870.	12.8	148
96	Human Toll-like receptor-dependent induction of interferons in protective immunity to viruses. <i>Immunological Reviews</i> , 2007, 220, 225-236.	6.0	147
97	Somatic diversification in the absence of antigen-driven responses is the hallmark of the IgM+IgD+CD27+ B cell repertoire in infants. <i>Journal of Experimental Medicine</i> , 2008, 205, 1331-1342.	8.5	143
98	Inherited CARD9 Deficiency in 2 Unrelated Patients With Invasive Exophiala Infection. <i>Journal of Infectious Diseases</i> , 2015, 211, 1241-1250.	4.0	141
99	Disease Evolution and Response to Rapamycin in Activated Phosphoinositide 3-Kinase $\hat{\gamma}$ Syndrome: The European Society for Immunodeficiencies-Activated Phosphoinositide 3-Kinase $\hat{\gamma}$ Syndrome Registry. <i>Frontiers in Immunology</i> , 2018, 9, 543.	4.8	137
100	FAS-L, IL-10, and double-negative CD4 $\hat{\gamma}$ CD8 $\hat{\gamma}$ TCR $\hat{\gamma}$ / $\hat{\delta}$ + T cells are reliable markers of autoimmune lymphoproliferative syndrome (ALPS) associated with FAS loss of function. <i>Blood</i> , 2009, 113, 3027-3030.	1.4	134
101	Human RHOH deficiency causes T cell defects and susceptibility to EV-HPV infections. <i>Journal of Clinical Investigation</i> , 2012, 122, 3239-3247.	8.2	134
102	Bacillus Calmette Guérin triggers the IL-12/IFN- $\hat{\gamma}$ axis by an IRAK-4- and NEMO-dependent, non-cognate interaction between monocytes, NK, and T $\hat{\gamma}$ lymphocytes. <i>European Journal of Immunology</i> , 2004, 34, 3276-3284.	2.9	133
103	Dominant-negative IKZF1 mutations cause a T, B, and myeloid cell combined immunodeficiency. <i>Journal of Clinical Investigation</i> , 2018, 128, 3071-3087.	8.2	133
104	A recessive form of hyper-IgE syndrome by disruption of ZNF341-dependent STAT3 transcription and activity. <i>Science Immunology</i> , 2018, 3, .	11.9	132
105	Inherited disorders of human Toll-like receptor signaling: immunological implications. <i>Immunological Reviews</i> , 2005, 203, 10-20.	6.0	129
106	Primary immunodeficiencies associated with pneumococcal disease. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2003, 3, 451-459.	2.3	128
107	Morbidity and mortality from ataxia-telangiectasia are associated with ATM genotype. <i>Journal of Allergy and Clinical Immunology</i> , 2011, 128, 382-389.e1.	2.9	128
108	Novel primary immunodeficiencies revealed by the investigation of paediatric infectious diseases. <i>Current Opinion in Immunology</i> , 2008, 20, 39-48.	5.5	127

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109	Whole-exome sequencing identifies Coronin-1A deficiency in 3 siblings with immunodeficiency and EBV-associated B-cell lymphoproliferation. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 131, 1594-1603.e9.	2.9	127
110	Mutational, functional, and expression studies of the <i>TCF4</i> gene in Pitt-Hopkins syndrome. <i>Human Mutation</i> , 2009, 30, 669-676.	2.5	126
111	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.	5.5	126
112	Inherited MST1 Deficiency Underlies Susceptibility to EV-HPV Infections. <i>PLoS ONE</i> , 2012, 7, e44010.	2.5	125
113	Inborn errors in RNA polymerase III underlie severe varicella zoster virus infections. <i>Journal of Clinical Investigation</i> , 2017, 127, 3543-3556.	8.2	125
114	IRAK4 and NEMO mutations in otherwise healthy children with recurrent invasive pneumococcal disease. <i>Journal of Medical Genetics</i> , 2006, 44, 16-23.	3.2	124
115	Inherited CD70 deficiency in humans reveals a critical role for the CD70-CD27 pathway in immunity to Epstein-Barr virus infection. <i>Journal of Experimental Medicine</i> , 2017, 214, 73-89.	8.5	122
116	IL-21 signalling via STAT3 primes human naïve B cells to respond to IL-2 to enhance their differentiation into plasmablasts. <i>Blood</i> , 2013, 122, 3940-3950.	1.4	121
117	Inherited human OX40 deficiency underlying classic Kaposi sarcoma of childhood. <i>Journal of Experimental Medicine</i> , 2013, 210, 1743-1759.	8.5	119
118	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.	8.5	117
119	A Mendelian predisposition to B-cell lymphoma caused by IL-10R deficiency. <i>Blood</i> , 2013, 122, 3713-3722.	1.4	116
120	Inherited GINS1 deficiency underlies growth retardation along with neutropenia and NK cell deficiency. <i>Journal of Clinical Investigation</i> , 2017, 127, 1991-2006.	8.2	115
121	Onset of autoimmune lymphoproliferative syndrome (ALPS) in humans as a consequence of genetic defect accumulation. <i>Journal of Clinical Investigation</i> , 2011, 121, 106-112.	8.2	110
122	First use of thymus transplantation therapy for FOXP1 deficiency (nude/SCID): a report of 2 cases. <i>Blood</i> , 2011, 117, 688-696.	1.4	109
123	SARS-CoV-2 induces human plasmacytoid dendritic cell diversification via UNC93B and IRAK4. <i>Journal of Experimental Medicine</i> , 2021, 218, .	8.5	107
124	Mycobacterial disease in patients with chronic granulomatous disease: retrospective analysis of 71 cases. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 241-248.e3.	2.9	106
125	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.	2.9	102
126	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.	1.4	102



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127	Pediatric Evans syndrome is associated with a high frequency of potentially damaging variants in immune genes. <i>Blood</i> , 2019, 134, 9-21.	1.4	102
128	Characterization of Crohn disease in X-linked inhibitor of apoptosis-deficient male patients and female symptomatic carriers. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 134, 1131-1141.e9.	2.9	101
129	Occurrence of B-cell lymphomas in patients with activated phosphoinositide 3-kinase $\gamma$ syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 134, 233-236.e3.	2.9	101
130	HHV-8-associated Kaposi sarcoma in a child with IFN $\gamma$ R1 deficiency. <i>Journal of Pediatrics</i> , 2004, 144, 519-523.	1.8	99
131	Clinical Features of Candidiasis in Patients With Inherited Interleukin 12 Receptor $\gamma$ 21 Deficiency. <i>Clinical Infectious Diseases</i> , 2014, 58, 204-213.	5.8	98
132	Impaired Interferon Gamma-Mediated Immunity and Susceptibility to Mycobacterial Infection in Childhood. <i>Pediatric Research</i> , 2001, 50, 8-13.	2.3	97
133	Human iNKT and MAIT cells exhibit a PLZF-dependent proapoptotic propensity that is counterbalanced by XIAP. <i>Blood</i> , 2013, 121, 614-623.	1.4	97
134	Primary T-cell immunodeficiency with immunodysregulation caused by autosomal recessive LCK deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2012, 130, 1144-1152.e11.	2.9	96
135	Disruption of an antimycobacterial circuit between dendritic and helper T cells in human SPPL2a deficiency. <i>Nature Immunology</i> , 2018, 19, 973-985.	14.5	96
136	Hematopoietic stem cell transplantation in 29 patients hemizygous for hypomorphic IKBKG/NEMO mutations. <i>Blood</i> , 2017, 130, 1456-1467.	1.4	95
137	Interleukin receptor-associated kinase (IRAK-4) deficiency associated with bacterial infections and failure to sustain antibody responses. <i>Journal of Pediatrics</i> , 2004, 144, 524-526.	1.8	94
138	Hematopoietic stem cell transplantation for complete IFN $\gamma$ receptor 1 deficiency: A multi-institutional survey. <i>Journal of Pediatrics</i> , 2004, 145, 806-812.	1.8	92
139	Clinical, immunologic, and genetic spectrum of 696 patients with combined immunodeficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 141, 1450-1458.	2.9	90
140	Inherited disorders of IL-12- and IFN $\gamma$ -mediated immunity: a molecular genetics update. <i>Molecular Immunology</i> , 2002, 38, 903-909.	2.2	88
141	Hypomorphic mutation of <i>ZAP70</i> in human results in a late onset immunodeficiency and no autoimmunity. <i>European Journal of Immunology</i> , 2009, 39, 1966-1976.	2.9	88
142	Reduced Expression of FOXP3 and Regulatory T-Cell Function in Severe Forms of Early-onset Autoimmune Enteropathy. <i>Gastroenterology</i> , 2010, 139, 770-778.	1.3	88
143	IgM+IgD+CD27+ B cells are markedly reduced in IRAK-4-, MyD88-, and TIRAP- but not UNC-93B-deficient patients. <i>Blood</i> , 2012, 120, 4992-5001.	1.4	87
144	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.	3.8	85

#	ARTICLE	IF	CITATIONS
145	PRKDC mutations associated with immunodeficiency, granuloma, and autoimmune regulator-dependent autoimmunity. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 135, 1578-1588.e5.	2.9	84
146	Polymerase $\beta$ 1 mutation in a human syndrome with facial dysmorphism, immunodeficiency, livedo, and short stature (FILS syndrome). <i>Journal of Experimental Medicine</i> , 2012, 209, 2323-2330.	8.5	83
147	Characteristics and outcome of early-onset, severe forms of Wiskott-Aldrich syndrome. <i>Blood</i> , 2013, 121, 1510-1516.	1.4	82
148	Prevention of Infections During Primary Immunodeficiency. <i>Clinical Infectious Diseases</i> , 2014, 59, 1462-1470.	5.8	81
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151	Unique and shared signaling pathways cooperate to regulate the differentiation of human CD4+ T cells into distinct effector subsets. <i>Journal of Experimental Medicine</i> , 2016, 213, 1589-1608.	8.5	77
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155	Early-onset autoimmunity associated with SOCS1 haploinsufficiency. <i>Nature Communications</i> , 2020, 11, 5341.	12.8	74
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158	A prospective study on the natural history of patients with profound combined immunodeficiency: An interim analysis. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 139, 1302-1310.e4.	2.9	71
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161	Human Adaptive Immunity Rescues an Inborn Error of Innate Immunity. <i>Cell</i> , 2017, 168, 789-800.e10.	28.9	68
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164	NEMO Mutations in 2 Unrelated Boys With Severe Infections and Conical Teeth. <i>Pediatrics</i> , 2005, 115, e615-e619.	2.1	67
165	Hematopoietic Stem Cell Transplantation as Treatment for Patients with DOCK8 Deficiency. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2019, 7, 848-855.	3.8	67
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179	Thromboxane synthase mutations in an increased bone density disorder (Ghosal syndrome). <i>Nature Genetics</i> , 2008, 40, 284-286.	21.4	61
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183	Complementation of a pathogenic <i>IFNGR2</i> misfolding mutation with modifiers of N-glycosylation. <i>Journal of Experimental Medicine</i> , 2008, 205, 1729-1737.	8.5	59
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257	Diagnosis of autoimmune lymphoproliferative syndrome caused by FAS deficiency in adults. <i>Haematologica</i> , 2013, 98, 389-392.	3.5	25
258	Germline genes hypomethylation and expression define a molecular signature in peripheral blood of ICF patients: implications for diagnosis and etiology. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 56.	2.7	25
259	Characteristics of HIV-infected children recently diagnosed in Paris, France. <i>European Journal of Pediatrics</i> , 2006, 165, 684-687.	2.7	24
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262	Occurrence of Aortic Aneurysms in 5 Cases of Wiskott-Aldrich Syndrome. <i>Pediatrics</i> , 2011, 127, e498-e504.	2.1	23
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265	Mechanisms of genotype-phenotype correlation in autosomal dominant anhidrotic ectodermal dysplasia with immune deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 141, 1060-1073.e3.	2.9	22
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273	Progressive Multifocal Leukoencephalopathy in Primary Immunodeficiencies. <i>Journal of Clinical Immunology</i> , 2019, 39, 55-64.	3.8	20
274	Genetic diagnosis of primary immunodeficiencies: A survey of the French national registry. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 1646-1649.e10.	2.9	20
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276	Pneumococcal Meningitis Vaccine Breakthroughs and Failures After Routine 7-Valent and 13-Valent Pneumococcal Conjugate Vaccination in Children in France. <i>Pediatric Infectious Disease Journal</i> , 2015, 34, e260-e263.	2.0	19
277	Analysis of the interleukin-12/interferon- $\gamma$ pathway in children with non-tuberculous mycobacterial cervical lymphadenitis. <i>European Journal of Pediatrics</i> , 2007, 166, 835-841.	2.7	18
278	DOCK8 Drives Src-Dependent NK Cell Effector Function. <i>Journal of Immunology</i> , 2017, 199, 2118-2127.	0.8	18
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281	Somatic reversion of pathogenic DOCK8 variants alters lymphocyte differentiation and function to effectively cure DOCK8 deficiency. <i>Journal of Clinical Investigation</i> , 2021, 131, .	8.2	18
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286	Improving the diagnostic efficiency of primary immunodeficiencies with targeted next-generation sequencing. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 147, 734-737.	2.9	17
287	<i>Burkholderia pseudomallei</i> infection in chronic granulomatous disease. <i>European Journal of Pediatrics</i> , 2006, 165, 175-177.	2.7	15
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