Jacinta Bustamante

List of Publications by Year
in descending order

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228 papers	24,058 citations	10373 72 h-index	8384 147 g-index
245 all docs	245 docs citations	245 times ranked	25120 citing authors

#	Article	IF	Citations
1	Clinical and Genetic Spectrum of Inborn Errors of Immunity in a Tertiary Care Center in Southern India. Indian Journal of Pediatrics, 2022, 89, 233-242.	0.3	4
2	A partial form of inherited human USP18 deficiency underlies infection and inflammation. Journal of Experimental Medicine, 2022, 219, .	4.2	28
3	Human autoantibodies underlying infectious diseases. Journal of Experimental Medicine, 2022, 219, .	4.2	55
4	Pulmonary Alveolar Proteinosis and Multiple Infectious Diseases in a Child with Autosomal Recessive Complete IRF8 Deficiency. Journal of Clinical Immunology, 2022, 42, 975-985.	2.0	7
5	Diagnosis of APS-1 in Two Siblings Following Life-Threatening COVID-19 Pneumonia. Journal of Clinical Immunology, 2022, 42, 749-752.	2.0	10
6	Effective anti-mycobacterial treatment for BCG disease in patients with Mendelian Susceptibility to Mycobacterial Disease (MSMD): a case series. Annals of Clinical Microbiology and Antimicrobials, 2022, 21, 8.	1.7	7
7	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.	1.5	2
8	SCID and Other Inborn Errors of Immunity with Low TRECs â€" the Brazilian Experience. Journal of Clinical Immunology, 2022, 42, 1171-1192.	2.0	4
9	Chronic Granulomatous Disease-Like Presentation of a Child with Autosomal Recessive PKCÎ´ Deficiency. Journal of Clinical Immunology, 2022, 42, 1244-1253.	2.0	6
10	Human OTULIN haploinsufficiency impairs cell-intrinsic immunity to staphylococcal Î±-toxin. Science, 2022, 376, eabm6380.	6.0	25
11	Improving the diagnostic efficiency of primary immunodeficiencies with targeted next-generation sequencing. Journal of Allergy and Clinical Immunology, 2021, 147, 734-737.	1.5	17
12	Multibatch Cytometry Data Integration for Optimal Immunophenotyping. Journal of Immunology, 2021, 206, 206-213.	0.4	25
13	Herpes simplex encephalitis in a patient with a distinctive form of inherited IFNAR1 deficiency. Journal of Clinical Investigation, 2021, 131, .	3.9	64
14	Inherited GATA2 Deficiency Is Dominant by Haploinsufficiency and Displays Incomplete Clinical Penetrance. Journal of Clinical Immunology, 2021, 41, 639-657.	2.0	30
15	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
16	EDA-ID: a Severe Clinical Presentation Associated with a New IKBKG Mutation. Journal of Clinical Immunology, 2021, 41, 1099-1102.	2.0	2
17	Clinical and Molecular Findings in Mendelian Susceptibility to Mycobacterial Diseases: Experience From India. Frontiers in Immunology, 2021, 12, 631298.	2.2	36
18	Genome-wide association study of resistance to Mycobacterium tuberculosis infection identifies a locus at 10q26.2 in three distinct populations. PLoS Genetics, 2021, 17, e1009392.	1.5	17

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19	Clinical and immunological profile of children with Mendelian Susceptibility to Mycobacterial Diseases (MSMD) from an Indian tertiary care hospital. Indian Journal of Tuberculosis, 2021, 68, 292-297.	0.3	4
20	Inherited deficiency of stress granule ZNFX1 in patients with monocytosis and mycobacterial disease. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	47
21	Detection of homozygous and hemizygous complete or partial exon deletions by whole-exome sequencing. NAR Genomics and Bioinformatics, 2021, 3, lqab037.	1.5	7
22	Disseminated Tuberculosis in a Patient with Autosomal Recessive p47phox Chronic Granulomatous Disease. Journal of Clinical Immunology, 2021, 41, 1417-1419.	2.0	2
23	Disseminated Mycobacterium simiae Infection in a Patient with Complete IL-12p40 Deficiency. Iranian Journal of Allergy, Asthma and Immunology, 2021, 20, 376-381.	0.3	2
24	High Th2 cytokine levels and upper airway inflammation in human inherited T-bet deficiency. Journal of Experimental Medicine, 2021, 218, .	4.2	25
25	Inherited PD-1 deficiency underlies tuberculosis and autoimmunity in a child. Nature Medicine, 2021, 27, 1646-1654.	15.2	65
26	Genetic, Immunological, and Clinical Features of 32 Patients with Autosomal Recessive STAT1 Deficiency. Journal of Immunology, 2021, 207, 133-152.	0.4	33
27	Impaired respiratory burst contributes to infections in PKCÎ´-deficient patients. Journal of Experimental Medicine, 2021, 218, .	4.2	23
28	Leukocytoclastic vasculitis in patients with IL12B or IL12RB1 deficiency: case report and review of the literature. Pediatric Rheumatology, 2021, 19, 121.	0.9	8
29	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
30	Inherited human c-Rel deficiency disrupts myeloid and lymphoid immunity to multiple infectious agents. Journal of Clinical Investigation, 2021, 131, .	3.9	21
31	Biochemically deleterious human NFKB1 variants underlie an autosomal dominant form of common variable immunodeficiency. Journal of Experimental Medicine, 2021, 218, .	4.2	32
32	Hematologically important mutations: X-linked chronic granulomatous disease (fourth update). Blood Cells, Molecules, and Diseases, 2021, 90, 102587.	0.6	22
33	Mycobacterial diseases in patients with inborn errors of immunity. Current Opinion in Immunology, 2021, 72, 262-271.	2.4	23
34	Hematologically important mutations: The autosomal forms of chronic granulomatous disease (third) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	0.6	22
35	GATA2 deficiency phenotype associated with tandem duplication of GATA2 and overexpression of GATA2-AS1. Blood Advances, 2021, 5, 5631-5635.	2.5	5
36	Cutaneous Granulomatosis Revealing Whippleâ€™s Disease: Value of Tropheryma whipplei Polymerase Chain Reaction Assay for the Diagnosis. Pathogens, 2021, 10, 1438.	1.2	3

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37	A New Patient with Inherited TYK2 Deficiency. Journal of Clinical Immunology, 2020, 40, 232-235.	2.0	19
38	Perianal Disease and Granulomas: Think Out of the Boxâ€¦. GE Portuguese Journal of Gastroenterology, 2020, 27, 119-123.	0.3	0
39	Mutual alteration of NOD2-associated Blau syndrome and IFNÎ³R1 deficiency. Journal of Clinical Immunology, 2020, 40, 165-178.	2.0	11
40	Interleukin 12â€23 deficiency in the interferon gamma pathway in a 6â€monthâ€old toddler who has BCG vaccine complications. Dermatologic Therapy, 2020, 33, e13999.	0.8	2
41	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.	2.0	8
42	A genome-wide case-only test for the detection of digenic inheritance in human exomes. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 19367-19375.	3.3	15
43	Homozygous STAT2 gain-of-function mutation by loss of USP18 activity in a patient with type I interferonopathy. Journal of Experimental Medicine, 2020, 217, .	4.2	73
44	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,749
45	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,983
46	Inflammatory cutaneous lesions and pulmonary manifestations in a new patient with autosomal recessive ISG15 deficiency case report. Allergy, Asthma and Clinical Immunology, 2020, 16, 77.	0.9	7
47	Nocardiosis Associated with Primary Immunodeficiencies (Nocar-DIP): an International Retrospective Study and Literature Review. Journal of Clinical Immunology, 2020, 40, 1144-1155.	2.0	11
48	Successful Hematopoietic Stem Cell Transplantation in a Patient with Complete IFN-Î³ Receptor 2 Deficiency: a Case Report and Literature Review. Journal of Clinical Immunology, 2020, 40, 1191-1195.	2.0	7
49	Human T-bet Governs Innate and Innate-like Adaptive IFN-Î³ Immunity against Mycobacteria. Cell, 2020, 183, 1826-1847.e31.	13.5	83
50	Human Lentiviral Gene Therapy Restores the Cellular Phenotype of Autosomal Recessive Complete IFN-Î³R1 Deficiency. Molecular Therapy - Methods and Clinical Development, 2020, 17, 785-795.	1.8	10
51	Systemic Type I IFN Inflammation in Human ISG15 Deficiency Leads to Necrotizing Skin Lesions. Cell Reports, 2020, 31, 107633.	2.9	47
52	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. Cell, 2020, 181, 1194-1199.	13.5	185
53	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
54	Transient Decrease of Circulating and Tissular Dendritic Cells in Patients With Mycobacterial Disease and With Partial Dominant IFNÎ³R1 Deficiency. Frontiers in Immunology, 2020, 11, 1161.	2.2	5

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55	Mendelian Susceptibility to Mycobacterial Disease (MSMD): Clinical and Genetic Features of 32 Iranian Patients. Journal of Clinical Immunology, 2020, 40, 872-882.	2.0	22
56	Autosomal recessive complete STAT1 deficiency caused by compound heterozygous intronic mutations. International Immunology, 2020, 32, 663-671.	1.8	26
57	Patient iPSC-Derived Macrophages to Study Inborn Errors of the IFN-Î³ Responsive Pathway. Cells, 2020, 9, 483.	1.8	16
58	Genetic, Immunological, and Clinical Features of the First Mexican Cohort of Patients with Chronic Granulomatous Disease. Journal of Clinical Immunology, 2020, 40, 475-493.	2.0	45
59	JAK Inhibitor Therapy in a Child with Inherited USP18 Deficiency. New England Journal of Medicine, 2020, 382, 256-265.	13.9	69
60	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
61	Fatal Cytomegalovirus Infection in an Adult with Inherited NOS2 Deficiency. New England Journal of Medicine, 2020, 382, 437-445.	13.9	38
62	Mendelian susceptibility to mycobacterial disease: recent discoveries. Human Genetics, 2020, 139, 993-1000.	1.8	132
63	A novel variant in the neutrophil cytosolic factor 2 (NCF2) gene results in severe disseminated BCG infectious disease: A clinical report and literature review. Molecular Genetics & Genomic Medicine, 2020, 8, e1237.	0.6	5
64	Inherited human IFN-Î³ deficiency underlies mycobacterial disease. Journal of Clinical Investigation, 2020, 130, 3158-3171.	3.9	89
65	Immunodeficiencies at the Interface of Innate and Adaptive Immunity. , 2019, , 509-522.e1.		0
66	LINE-1-Mediated AluYa5 Insertion Underlying Complete Autosomal Recessive IFN-Î³R1 Deficiency. Journal of Clinical Immunology, 2019, 39, 739-742.	2.0	5
67	Disseminated Mycobacterial Disease in a Patient with 22q11.2 Deletion Syndrome: Case Report and Review of the Literature. Journal of Clinical Immunology, 2019, 39, 743-746.	2.0	1
68	A Novel Recessive Mutation of Interferon-Î³ Receptor 1 in a Patient with Mycobacterium tuberculosis in Bone Marrow Aspirate. Journal of Clinical Immunology, 2019, 39, 127-130.	2.0	8
69	Molecular, Immunological, and Clinical Features of 16 Iranian Patients with Mendelian Susceptibility to Mycobacterial Disease. Journal of Clinical Immunology, 2019, 39, 287-297.	2.0	13
70	Life-Threatening Infections Due to Live-Attenuated Vaccines: Early Manifestations of Inborn Errors of Immunity. Journal of Clinical Immunology, 2019, 39, 376-390.	2.0	50
71	Recurrent Salmonella typhi Infection and Autoimmunity in a Young Boy with Complete IL-12 Receptor Î²1 Deficiency. Journal of Clinical Immunology, 2019, 39, 358-362.	2.0	9
72	Human DOCK2 Deficiency: Report of a Novel Mutation and Evidence for Neutrophil Dysfunction. Journal of Clinical Immunology, 2019, 39, 298-308.	2.0	31

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73	IFN-Î³ and CD25 drive distinct pathologic features during hemophagocytic lymphohistiocytosis. Journal of Allergy and Clinical Immunology, 2019, 143, 2215-2226.e7.	1.5	49
74	Mendelian susceptibility to mycobacterial disease: 2014â€“2018 update. Immunology and Cell Biology, 2019, 97, 360-367.	1.0	163
75	Hematopoietic stem cell transplant effectively rescues lymphocyte differentiation and function in DOCK8-deficient patients. JCI Insight, 2019, 4, .	2.3	23
76	Skewed X-inactivation in a Female Carrier with X-linked Chronic Granulomatous Disease. Iranian Journal of Allergy, Asthma and Immunology, 2019, 18, 447-451.	0.3	4
77	Laboratory evaluation of the IFN-Î³ circuit for the molecular diagnosis of Mendelian susceptibility to mycobacterial disease. Critical Reviews in Clinical Laboratory Sciences, 2018, 55, 184-204.	2.7	43
78	Disseminated abscesses due to Mycoplasma faucium in a patient with activated PI3KÎ´ syndrome type 2. Journal of Allergy and Clinical Immunology: in Practice, 2018, 6, 1796-1798.e2.	2.0	9
79	Mendelian Susceptibility to Mycobacterial Disease Caused by a Novel Founder IL12B Mutation in Saudi Arabia. Journal of Clinical Immunology, 2018, 38, 278-282.	2.0	9
80	Hematopoietic stem cell gene therapy for IFNÎ³R1 deficiency protects mice from mycobacterial infections. Blood, 2018, 131, 533-545.	0.6	19
81	Human IFN-Î³ immunity to mycobacteria is governed by both IL-12 and IL-23. Science Immunology, 2018, 3, .	5.6	152
82	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
83	T-cell defects in patients with ARPC1B germline mutations account for combined immunodeficiency. Blood, 2018, 132, 2362-2374.	0.6	99
84	Impaired IL-12- and IL-23-Mediated Immunity Due to IL-12RÎ²1 Deficiency in Iranian Patients with Mendelian Susceptibility to Mycobacterial Disease. Journal of Clinical Immunology, 2018, 38, 787-793.	2.0	13
85	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
86	Copy number variations and founder effect underlying complete IL-10RÎ² deficiency in Portuguese kindreds. PLoS ONE, 2018, 13, e0205826.	1.1	13
87	Autosomal Dominant IFN-Î³R1 Deficiency Presenting with both Atypical Mycobacteriosis and Tuberculosis in a BCG-Vaccinated South African Patient. Journal of Clinical Immunology, 2018, 38, 460-463.	2.0	8
88	Severe BCG-osis Misdiagnosed as Multidrug-Resistant Tuberculosis in an IL-12RÎ²1-Deficient Peruvian Girl. Journal of Clinical Immunology, 2018, 38, 712-716.	2.0	8
89	IRF4 haploinsufficiency in a family with Whippleâ€™s disease. ELife, 2018, 7, .	2.8	43
90	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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91	Natural history of GATA2 deficiency in a survey of 79 French and Belgian patients. Haematologica, 2018, 103, 1278-1287.	1.7	129
92	Disruption of an antimycobacterial circuit between dendritic and helper T cells in human SPPL2a deficiency. Nature Immunology, 2018, 19, 973-985.	7.0	96
93	A purely quantitative form of partial recessive IFN-Î³R2 deficiency caused by mutations of the initiation or second codon. Human Molecular Genetics, 2018, 27, 3919-3935.	1.4	14
94	A recessive form of hyper-IgE syndrome by disruption of ZNF341-dependent STAT3 transcription and activity. Science Immunology, 2018, 3, .	5.6	132
95	Lethal Influenza in Two Related Adults with Inherited GATA2 Deficiency. Journal of Clinical Immunology, 2018, 38, 513-526.	2.0	29
96	Rescue of recurrent deep intronic mutation underlying cell typeâ€"dependent quantitative NEMO deficiency. Journal of Clinical Investigation, 2018, 129, 583-597.	3.9	38
97	Inherited p40phox deficiency differs from classic chronic granulomatous disease. Journal of Clinical Investigation, 2018, 128, 3957-3975.	3.9	99
98	Respiratory Complications Lead to the Diagnosis of Chronic Granulomatous Disease in Two Adult Patients. Journal of Clinical Immunology, 2017, 37, 113-116.	2.0	7
99	Systemic Human ILC Precursors Provide a Substrate for Tissue ILC Differentiation. Cell, 2017, 168, 1086-1100.e10.	13.5	420
100	Disseminated Bacillus Calmette-GuÃ©rin Osteomyelitis in Twin Sisters Related to STAT1 Gene Deficiency. Pediatric and Developmental Pathology, 2017, 20, 255-261.	0.5	11
101	Chronic Granulomatous Disease in Patients Reaching Adulthood: A Nationwide Study in France. Clinical Infectious Diseases, 2017, 64, 767-775.	2.9	57
102	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
103	An eQTL variant of ZXDC is associated with IFN-Î³ production following Mycobacterium tuberculosis antigen-specific stimulation. Scientific Reports, 2017, 7, 12800.	1.6	5
104	Inherited IL-12RÎ²1 Deficiency in a Child With BCG Adenitis and Oral Candidiasis: A Case Report. Pediatrics, 2017, 140, .	1.0	16
105	Paracoccidioidomycosis Associated With a Heterozygous STAT4 Mutation and Impaired IFN-Î³ Immunity. Journal of Infectious Diseases, 2017, 216, 1623-1634.	1.9	25
106	Severe Enteropathy and Hypogammaglobulinemia Complicating Refractory Mycobacterium tuberculosis Complex Disseminated Disease in a Child with IL-12RÎ²1 Deficiency. Journal of Clinical Immunology, 2017, 37, 732-738.	2.0	10
107	Hematopoietic stem cell transplantation in 29 patients hemizygous for hypomorphic IKBKG/NEMO mutations. Blood, 2017, 130, 1456-1467.	0.6	95
108	Visceral leishmaniasis in two patients with ILâ€12p40 and ILâ€12RÎ²1 deficiencies. Pediatric Blood and Cancer, 2017, 64, e26362.	0.8	25

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109	Microbial Disease Spectrum Linked to a Novel IL-12RÎ²1 N-Terminal Signal Peptide Stop-Gain Homozygous Mutation with Paradoxical Receptor Cell-Surface Expression. Frontiers in Microbiology, 2017, 8, 616.	1.5	18
110	Multifocal Recurrent Osteomyelitis and Hemophagocytic Lymphohistiocytosis in a Boy with Partial Dominant IFN-Î³R1 Deficiency: Case Report and Review of the Literature. Frontiers in Pediatrics, 2017, 5, 75.	0.9	24
111	Inherited GINS1 deficiency underlies growth retardation along with neutropenia and NK cell deficiency. Journal of Clinical Investigation, 2017, 127, 1991-2006.	3.9	115
112	IL12RÎ²1 defect presenting with massive intra-abdominal lymphadenopathy due to Mycobacterium intracellulare infection. Asian Pacific Journal of Allergy and Immunology, 2017, 35, 161-165.	0.2	3
113	Mendelian Susceptibility to Infections with Viruses, Mycobacteria, Bacteria, andÂ Candida. , 2016, , 407-415.		0
114	Interferon-Î³ Autoantibodies as Predisposing Factor for Nontuberculous Mycobacterial Infection. Emerging Infectious Diseases, 2016, 22, 1124-1126.	2.0	38
115	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
116	Disseminated BCG osteomyelitis related to STAT 1 gene deficiency mimicking a metastatic neuroblastoma. Pediatric and Developmental Pathology, 2016, , .	0.5	3
117	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
118	Role of Flow Cytometry in the Diagnosis of Chronic Granulomatous Disease: the Egyptian Experience. Journal of Clinical Immunology, 2016, 36, 610-618.	2.0	21
119	Leukocyte Adhesion Deficiency Type 1 (LAD1) with Expressed but Nonfunctional CD11/CD18. Journal of Clinical Immunology, 2016, 36, 627-630.	2.0	18
120	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
121	Use of corticosteroids as an alternative to surgical treatment for liver abscesses in chronic granulomatous disease. Pediatric Blood and Cancer, 2016, 63, 2254-2255.	0.8	5
122	Transduction of Herpesvirus saimiriâ€Transformed T Cells with Exogenous Genes of Interest. Current Protocols in Immunology, 2016, 115, 7.21C.1-7.21C.12.	3.6	17
123	Disseminated bacille Calmetteâ€“GuÃ©rin disease in an infant with a novel biallelic mutation in interferon gamma receptor-1 gene. Pediatric Infectious Disease, 2016, 8, 84-87.	0.1	1
124	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
125	Antiâ€“IFN-Î³ autoantibodies are strongly associated with HLA-DR*15:02/16:02 and HLA-DQ*05:01/05:02 across Southeast Asia. Journal of Allergy and Clinical Immunology, 2016, 137, 945-948.e8.	1.5	63
126	Severe Mycobacterial Diseases in a Patient with GOF IÎºBÎ± Mutation Without EDA. Journal of Clinical Immunology, 2016, 36, 12-15.	2.0	11

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127	Interferon-gamma-dependent Immunity in Bacillus Calmette-GuÃ©rin Vaccine Osteitis Survivors. Pediatric Infectious Disease Journal, 2016, 35, 690-694.	1.1	10
128	IL-12 drives functional plasticity of human group 2 innate lymphoid cells. Journal of Experimental Medicine, 2016, 213, 569-583.	4.2	246
129	Mycobacterial disease in patients with chronic granulomatous disease: AÂretrospective analysis of 71 cases. Journal of Allergy and Clinical Immunology, 2016, 138, 241-248.e3.	1.5	106
130	Major Loci on Chromosomes 8q and 3q Control Interferon Î³ Production Triggered by Bacillus Calmette-Guerin and 6-kDa Early Secretory Antigen Target, Respectively, in Various Populations. Journal of Infectious Diseases, 2016, 213, 1173-1179.	1.9	15
131	Infectious diseases, autoimmunity and midline defect in a patient with a novel bi-allelic mutation in IL12RB1 gene. Turkish Journal of Pediatrics, 2016, 58, 331-336.	0.3	15
132	Mendelian Susceptibility to Mycobacterial Disease due to IL-12RÎ²1 Deficiency in Three Iranian Children. Iranian Journal of Public Health, 2016, 45, 249-54.	0.3	6
133	Mendelian Susceptibility to Mycobacterial Disease due to IL-12RÎ²1 Deficiency in Three Iranian Children. Iranian Journal of Public Health, 2016, 45, 370-5.	0.3	6
134	Diagnostic and therapeutic challenges in a child with complete Interferonâ€Î³ Receptor 1 deficiency. Pediatric Blood and Cancer, 2015, 62, 2036-2039.	0.8	27
135	Inherited and acquired immunodeficiencies underlying tuberculosis in childhood. Immunological Reviews, 2015, 264, 103-120.	2.8	180
136	Phagocyte nicotinamide adenine dinucleotide phosphate oxidase activity in patients with inherited IFN-Î³R1 or IFN-Î³R2 deficiency. Journal of Allergy and Clinical Immunology, 2015, 135, 1393-1395.e1.	1.5	11
137	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
138	Impairment of immunity to Candida and Mycobacterium in humans with bi-allelic RORC mutations. Science, 2015, 349, 606-613.	6.0	366
139	Disseminated Mycobacterium avium Complex Infection in a Child with Partial Dominant Interferon Gamma Receptor 1 Deficiency in India. Journal of Clinical Immunology, 2015, 35, 459-462.	2.0	17
140	Tuberculin Skin Test Negativity Is Under Tight Genetic Control of Chromosomal Region 11p14-15 in Settings With Different Tuberculosis Endemicities. Journal of Infectious Diseases, 2015, 211, 317-321.	1.9	42
141	Inherited IL-17RC deficiency in patients with chronic mucocutaneous candidiasis. Journal of Experimental Medicine, 2015, 212, 619-631.	4.2	162
142	STAT3 is a critical cell-intrinsic regulator of human unconventional T cell numbers and function. Journal of Experimental Medicine, 2015, 212, 855-864.	4.2	70
143	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
144	Clinical and Genotypic Spectrum of Chronic Granulomatous Disease in 71 Latin American Patients: First Report from the LASID Registry. Pediatric Blood and Cancer, 2015, 62, 2101-2107.	0.8	67

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145	Human TYK2 deficiency: Mycobacterial and viral infections without hyper-IgE syndrome. Journal of Experimental Medicine, 2015, 212, 1641-1662.	4.2	293
146	Human intracellular ISG15 prevents interferon-Î±/Î² over-amplification and auto-inflammation. Nature, 2015, 517, 89-93.	13.7	432
147	Chronic Granulomatous Disease in Morocco: Genetic, Immunological, and Clinical Features of 12 Patients from 10 Kindreds. Journal of Clinical Immunology, 2014, 34, 452-8.	2.0	17
148	ICON: The Early Diagnosis of Congenital Immunodeficiencies. Journal of Clinical Immunology, 2014, 34, 398-424.	2.0	34
149	Clinical Features of Candidiasis in Patients With Inherited Interleukin 12 Receptor Î²1 Deficiency. Clinical Infectious Diseases, 2014, 58, 204-213.	2.9	98
150	Association Study of Genes Controlling IL-12-dependent IFN-Î³ Immunity: STAT4 Alleles Increase Risk of Pulmonary Tuberculosis in Morocco. Journal of Infectious Diseases, 2014, 210, 611-618.	1.9	31
151	Mendelian susceptibility to mycobacterial disease: Genetic, immunological, and clinical features of inborn errors of IFN-Î³ immunity. Seminars in Immunology, 2014, 26, 454-470.	2.7	582
152	Mycobacterium simiae Infection in Two Unrelated Patients with Different Forms of Inherited IFN-Î³R2 Deficiency. Journal of Clinical Immunology, 2014, 34, 904-909.	2.0	20
153	Recurrent Salmonellosis in a Child with Complete IL-12RÎ²1 Deficiency. Journal of Immunodeficiency & Disorders, 2014, 03, .	0.4	9
154	IL-12RÎ²1 Deficiency: Mutation Update and Description of theIL12RB1Variation Database. Human Mutation, 2013, 34, 1329-1339.	1.1	81
155	Partial IFN-Î³R2 deficiency is due to protein misfolding and can be rescued by inhibitors of glycosylation. Blood, 2013, 122, 2390-2401.	0.6	34
156	Haploinsufficiency at the human IFNGR2 locus contributes to mycobacterial disease. Human Molecular Genetics, 2013, 22, 769-781.	1.4	58
157	Signal transducer and activator of transcription 3 (STAT3) mutations underlying autosomal dominant hyper-IgE syndrome impair human CD8+ T-cell memory formation and function. Journal of Allergy and Clinical Immunology, 2013, 132, 400-411.e9.	1.5	63
158	IL-12RÎ²1 Deficiency and Disseminated Mycobacterium tilburgii Disease. Journal of Clinical Immunology, 2013, 33, 1285-1288.	2.0	11
159	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
160	IL-21 signalling via STAT3 primes human naÃ¯ve B cells to respond to IL-2 to enhance their differentiation into plasmablasts. Blood, 2013, 122, 3940-3950.	0.6	121
161	Age-Dependent Association between Pulmonary Tuberculosis and Common TOX Variants in the 8q12â€“13 Linkage Region. American Journal of Human Genetics, 2013, 92, 407-414.	2.6	46
162	Primary immunodeficiencies underlying fungal infections. Current Opinion in Pediatrics, 2013, 25, 736-747.	1.0	190

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163	Inherited human OX40 deficiency underlying classic Kaposi sarcoma of childhood. Journal of Experimental Medicine, 2013, 210, 1743-1759.	4.2	119
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