Jacinta Bustamante

List of Publications by Year in descending order

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	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	Pyogenic Bacterial Infections in Humans with IRAK-4 Deficiency. Science, 2003, 299, 2076-2079.	6.0	820
5	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
6	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
7	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
8	IRF8Mutations and Human Dendritic-Cell Immunodeficiency. New England Journal of Medicine, 2011, 365, 127-138.	13.9	564
9	Mycobacterial Disease and Impaired IFN-Î³ Immunity in Humans with Inherited ISG15 Deficiency. Science, 2012, 337, 1684-1688.	6.0	455
10	Human intracellular ISG15 prevents interferon-Î±/Î² over-amplification and auto-inflammation. Nature, 2015, 517, 89-93.	13.7	432
11	Inborn errors of IL-12/23- and IFN-Î³-mediated immunity: molecular, cellular, and clinical features. Seminars in Immunology, 2006, 18, 347-361.	2.7	422
12	Systemic Human ILC Precursors Provide a Substrate for Tissue ILC Differentiation. Cell, 2017, 168, 1086-1100.e10.	13.5	420
13	Revisiting Human IL-12RÎ²1 Deficiency. Medicine (United States), 2010, 89, 381-402.	0.4	367
14	Impairment of immunity to Candida and Mycobacterium in humans with bi-allelic RORC mutations. Science, 2015, 349, 606-613.	6.0	366
15	B cellâ€“intrinsic signaling through IL-21 receptor and STAT3 is required for establishing long-lived antibody responses in humans. Journal of Experimental Medicine, 2010, 207, 155-171.	4.2	346
16	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
17	Human TYK2 deficiency: Mycobacterial and viral infections without hyper-IgE syndrome. Journal of Experimental Medicine, 2015, 212, 1641-1662.	4.2	293
18	Low Penetrance, Broad Resistance, and Favorable Outcome of Interleukin 12 Receptor Î²1 Deficiency. Journal of Experimental Medicine, 2003, 197, 527-535.	4.2	286

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19	Inborn errors of interferon (IFN)â€mediated immunity in humans: insights into the respective roles of IFNâ€Î±/Î², IFNâ€Î³, and IFNâ€» in host defense. Immunological Reviews, 2008, 226, 29-40.	2.8	271
20	Functional STAT3 deficiency compromises the generation of human T follicular helper cells. Blood, 2012, 119, 3997-4008.	0.6	267
21	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
22	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
23	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
24	IL-12 drives functional plasticity of human group 2 innate lymphoid cells. Journal of Experimental Medicine, 2016, 213, 569-583.	4.2	246
25	Clinical, immunologic and genetic analysis of 29 patients with autosomal recessive hyper-IgM syndrome due to Activation-Induced Cytidine Deaminase deficiency. Clinical Immunology, 2004, 110, 22-29.	1.4	224
26	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
27	Gains of glycosylation comprise an unexpectedly large group of pathogenic mutations. Nature Genetics, 2005, 37, 692-700.	9.4	198
28	Primary immunodeficiencies underlying fungal infections. Current Opinion in Pediatrics, 2013, 25, 736-747.	1.0	190
29	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. Cell, 2020, 181, 1194-1199.	13.5	185
30	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
31	Inherited and acquired immunodeficiencies underlying tuberculosis in childhood. Immunological Reviews, 2015, 264, 103-120.	2.8	180
32	Hematologically important mutations: X-linked chronic granulomatous disease (third update). Blood Cells, Molecules, and Diseases, 2010, 45, 246-265.	0.6	179
33	Novel STAT1 Alleles in Otherwise Healthy Patients with Mycobacterial Disease. PLoS Genetics, 2006, 2, e131.	1.5	171
34	A partial form of recessive STAT1 deficiency in humans. Journal of Clinical Investigation, 2009, 119, 1502-1514.	3.9	167
35	Mendelian susceptibility to mycobacterial disease: 2014â€“2018 update. Immunology and Cell Biology, 2019, 97, 360-367.	1.0	163
36	Inherited IL-17RC deficiency in patients with chronic mucocutaneous candidiasis. Journal of Experimental Medicine, 2015, 212, 619-631.	4.2	162

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37	Naive and memory human B cells have distinct requirements for STAT3 activation to differentiate into antibody-secreting plasma cells. Journal of Experimental Medicine, 2013, 210, 2739-2753.	4.2	158
38	Human IFN-Î³ immunity to mycobacteria is governed by both IL-12 and IL-23. Science Immunology, 2018, 3, .	5.6	152
39	Inherited IL-12p40 Deficiency. Medicine (United States), 2013, 92, 109-122.	0.4	151
40	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
41	IL-12 receptor Î²1 deficiency alters in vivo T follicular helper cell response in humans. Blood, 2013, 121, 3375-3385.	0.6	147
42	Hematologically important mutations: The autosomal recessive forms of chronic granulomatous disease (second update). Blood Cells, Molecules, and Diseases, 2010, 44, 291-299.	0.6	143
43	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
44	Bacillus Calmette GuÃ©rin triggers the IL-12/IFN-Î³ axis by an IRAK-4- and NEMO-dependent, non-cognate interaction between monocytes, NK, and Tâ€„lymphocytes. European Journal of Immunology, 2004, 34, 3276-3284.	1.6	133
45	A recessive form of hyper-IgE syndrome by disruption of ZNF341-dependent STAT3 transcription and activity. Science Immunology, 2018, 3, .	5.6	132
46	Mendelian susceptibility to mycobacterial disease: recent discoveries. Human Genetics, 2020, 139, 993-1000.	1.8	132
47	Inherited disorders of human Toll-like receptor signaling: immunological implications. Immunological Reviews, 2005, 203, 10-20.	2.8	129
48	Natural history of GATA2 deficiency in a survey of 79 French and Belgian patients. Haematologica, 2018, 103, 1278-1287.	1.7	129
49	Primary immunodeficiencies associated with pneumococcal disease. Current Opinion in Allergy and Clinical Immunology, 2003, 3, 451-459.	1.1	128
50	Novel primary immunodeficiencies revealed by the investigation of paediatric infectious diseases. Current Opinion in Immunology, 2008, 20, 39-48.	2.4	127
51	IRAK4 and NEMO mutations in otherwise healthy children with recurrent invasive pneumococcal disease. Journal of Medical Genetics, 2006, 44, 16-23.	1.5	124
52	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
53	Inherited human OX40 deficiency underlying classic Kaposi sarcoma of childhood. Journal of Experimental Medicine, 2013, 210, 1743-1759.	4.2	119
54	BCG-osis and tuberculosis in a child with chronic granulomatous disease. Journal of Allergy and Clinical Immunology, 2007, 120, 32-38.	1.5	118

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55	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
56	Inherited GINS1 deficiency underlies growth retardation along with neutropenia and NK cell deficiency. Journal of Clinical Investigation, 2017, 127, 1991-2006.	3.9	115
57	IL-12RÎ²1 Deficiency in Two of Fifty Children with Severe Tuberculosis from Iran, Morocco, and Turkey. PLoS ONE, 2011, 6, e18524.	1.1	111
58	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
59	A novel form of complete IL-12/IL-23 receptor Â1 deficiency with cell surface-expressed nonfunctional receptors. Blood, 2004, 104, 2095-2101.	0.6	103
60	Partial recessive IFN-Î³R1 deficiency: genetic, immunological and clinical features of 14 patients from 11 kindreds. Human Molecular Genetics, 2011, 20, 1509-1523.	1.4	102
61	T-cell defects in patients with ARPC1B germline mutations account for combined immunodeficiency. Blood, 2018, 132, 2362-2374.	0.6	99
62	Inherited p40phox deficiency differs from classic chronic granulomatous disease. Journal of Clinical Investigation, 2018, 128, 3957-3975.	3.9	99
63	Clinical Features of Candidiasis in Patients With Inherited Interleukin 12 Receptor Î²1 Deficiency. Clinical Infectious Diseases, 2014, 58, 204-213.	2.9	98
64	Disruption of an antimycobacterial circuit between dendritic and helper T cells in human SPPL2a deficiency. Nature Immunology, 2018, 19, 973-985.	7.0	96
65	Hematopoietic stem cell transplantation in 29 patients hemizygous for hypomorphic IKBKG/NEMO mutations. Blood, 2017, 130, 1456-1467.	0.6	95
66	A novel form of human STAT1 deficiency impairing early but not late responses to interferons. Blood, 2010, 116, 5895-5906.	0.6	93
67	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
68	Inherited human IFN-Î³ deficiency underlies mycobacterial disease. Journal of Clinical Investigation, 2020, 130, 3158-3171.	3.9	89
69	Genetic lessons learned from Xâ€linked Mendelian susceptibility to mycobacterial diseases. Annals of the New York Academy of Sciences, 2011, 1246, 92-101.	1.8	85
70	Human T-bet Governs Innate and Innate-like Adaptive IFN-Î³ Immunity against Mycobacteria. Cell, 2020, 183, 1826-1847.e31.	13.5	83
71	IL-12RÎ²1 Deficiency: Mutation Update and Description of theIL12RB1Variation Database. Human Mutation, 2013, 34, 1329-1339.	1.1	81
72	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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73	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
74	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
75	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
76	JAK Inhibitor Therapy in a Child with Inherited USP18 Deficiency. New England Journal of Medicine, 2020, 382, 256-265.	13.9	69
77	NEMO Mutations in 2 Unrelated Boys With Severe Infections and Conical Teeth. Pediatrics, 2005, 115, e615-e619.	1.0	67
78	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
79	Primary immunodeficiencies of protective immunity to primary infections. Clinical Immunology, 2010, 135, 204-209.	1.4	65
80	Inherited PD-1 deficiency underlies tuberculosis and autoimmunity in a child. Nature Medicine, 2021, 27, 1646-1654.	15.2	65
81	Herpes simplex encephalitis in a patient with a distinctive form of inherited IFNAR1 deficiency. Journal of Clinical Investigation, 2021, 131, .	3.9	64
82	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
83	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
84	Inherited disorders of the IL-12-IFN-Î³ axis in patients with disseminated BCG infection. European Journal of Pediatrics, 2005, 164, 753-757.	1.3	59
85	Complementation of a pathogenic IFNGR2 misfolding mutation with modifiers of N-glycosylation. Journal of Experimental Medicine, 2008, 205, 1729-1737.	4.2	59
86	Haploinsufficiency at the human IFNGR2 locus contributes to mycobacterial disease. Human Molecular Genetics, 2013, 22, 769-781.	1.4	58
87	From idiopathic infectious diseases to novel primary immunodeficiencies. Journal of Allergy and Clinical Immunology, 2005, 116, 426-430.	1.5	57
88	Chronic Granulomatous Disease in Patients Reaching Adulthood: A Nationwide Study in France. Clinical Infectious Diseases, 2017, 64, 767-775.	2.9	57
89	Human autoantibodies underlying infectious diseases. Journal of Experimental Medicine, 2022, 219, .	4.2	55
90	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

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91	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
92	A novel X-linked recessive form of Mendelian susceptibility to mycobaterial disease. Journal of Medical Genetics, 2006, 44, e65-e65.	1.5	52
93	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
94	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
95	Lethal Tuberculosis in a Previously Healthy Adult with IL-12 Receptor Deficiency. Journal of Clinical Immunology, 2011, 31, 537-539.	2.0	49
96	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
97	Systemic Type I IFN Inflammation in Human ISG15 Deficiency Leads to Necrotizing Skin Lesions. Cell Reports, 2020, 31, 107633.	2.9	47
98	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
99	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
100	Septicemia without Sepsis: Inherited Disorders of Nuclear Factor-kB-Mediated Inflammation. Clinical Infectious Diseases, 2005, 41, S436-S439.	2.9	45
101	A Variety of Alu-Mediated Copy Number Variations Can Underlie IL-12RÎ²1 Deficiency. Journal of Clinical Immunology, 2018, 38, 617-627.	2.0	45
102	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
103	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
104	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
105	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
106	IRF4 haploinsufficiency in a family with Whippleâ€™s disease. ELife, 2018, 7, .	2.8	43
107	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
108	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

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109	Interferon-Î³ Autoantibodies as Predisposing Factor for Nontuberculous Mycobacterial Infection. Emerging Infectious Diseases, 2016, 22, 1124-1126.	2.0	38
110	Fatal Cytomegalovirus Infection in an Adult with Inherited NOS2 Deficiency. New England Journal of Medicine, 2020, 382, 437-445.	13.9	38
111	Rescue of recurrent deep intronic mutation underlying cell typeâ€“dependent quantitative NEMO deficiency. Journal of Clinical Investigation, 2018, 129, 583-597.	3.9	38
112	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
113	Clinical and Molecular Findings in Mendelian Susceptibility to Mycobacterial Diseases: Experience From India. Frontiers in Immunology, 2021, 12, 631298.	2.2	36
114	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
115	ICON: The Early Diagnosis of Congenital Immunodeficiencies. Journal of Clinical Immunology, 2014, 34, 398-424.	2.0	34
116	Multiple cutaneous squamous cell carcinomas in a patient with interferon Â receptor 2 (IFNÂR2) deficiency. Journal of Medical Genetics, 2010, 47, 631-634.	1.5	33
117	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
118	Genetic, Immunological, and Clinical Features of 32 Patients with Autosomal Recessive STAT1 Deficiency. Journal of Immunology, 2021, 207, 133-152.	0.4	33
119	Biochemically deleterious human NFKB1 variants underlie an autosomal dominant form of common variable immunodeficiency. Journal of Experimental Medicine, 2021, 218, .	4.2	32
120	Clinical Disease Caused byKlebsiellain 2 Unrelated Patients With Interleukin 12 Receptor Î²1 Deficiency. Pediatrics, 2010, 126, e971-e976.	1.0	31
121	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
122	Human DOCK2 Deficiency: Report of a Novel Mutation and Evidence for Neutrophil Dysfunction. Journal of Clinical Immunology, 2019, 39, 298-308.	2.0	31
123	A Novel Homozygous p.R1105X Mutation of the AP4E1 Gene in Twins with Hereditary Spastic Paraplegia and Mycobacterial Disease. PLoS ONE, 2013, 8, e58286.	1.1	31
124	Inherited GATA2 Deficiency Is Dominant by Haploinsufficiency and Displays Incomplete Clinical Penetrance. Journal of Clinical Immunology, 2021, 41, 639-657.	2.0	30
125	Lethal Influenza in Two Related Adults with Inherited GATA2 Deficiency. Journal of Clinical Immunology, 2018, 38, 513-526.	2.0	29
126	Essential role of nuclear factor-ÎºB for NADPH oxidase activity in normal and anhidrotic ectodermal dysplasia leukocytes. Blood, 2008, 112, 1453-1460.	0.6	28

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127	A partial form of inherited human USP18 deficiency underlies infection and inflammation. Journal of Experimental Medicine, 2022, 219, .	4.2	28
128	Heritable defects of the human TLR signalling pathways. Journal of Endotoxin Research, 2005, 11, 220-224.	2.5	27
129	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
130	Autosomal recessive complete STAT1 deficiency caused by compound heterozygous intronic mutations. International Immunology, 2020, 32, 663-671.	1.8	26
131	From Infectious Diseases to Primary Immunodeficiencies. Immunology and Allergy Clinics of North America, 2008, 28, 235-258.	0.7	25
132	Paracoccidioidomycosis Associated With a Heterozygous STAT4 Mutation and Impaired IFN-Î³ Immunity. Journal of Infectious Diseases, 2017, 216, 1623-1634.	1.9	25
133	Visceral leishmaniasis in two patients with ILâ€12p40 and ILâ€12RÎ²1 deficiencies. Pediatric Blood and Cancer, 2017, 64, e26362.	0.8	25
134	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
135	Multibatch Cytometry Data Integration for Optimal Immunophenotyping. Journal of Immunology, 2021, 206, 206-213.	0.4	25
136	High Th2 cytokine levels and upper airway inflammation in human inherited T-bet deficiency. Journal of Experimental Medicine, 2021, 218, .	4.2	25
137	Human OTULIN haploinsufficiency impairs cell-intrinsic immunity to staphylococcal Î±-toxin. Science, 2022, 376, eabm6380.	6.0	25
138	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
139	Impaired respiratory burst contributes to infections in PKCÎ´-deficient patients. Journal of Experimental Medicine, 2021, 218, .	4.2	23
140	Mycobacterial diseases in patients with inborn errors of immunity. Current Opinion in Immunology, 2021, 72, 262-271.	2.4	23
141	Hematopoietic stem cell transplant effectively rescues lymphocyte differentiation and function in DOCK8-deficient patients. JCI Insight, 2019, 4, .	2.3	23
142	Paternal uniparental isodisomy of chromosome 6 causing a complex syndrome including complete IFNâ€Î³ receptor 1 deficiency. American Journal of Medical Genetics, Part A, 2010, 152A, 622-629.	0.7	22
143	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
144	Hematologically important mutations: X-linked chronic granulomatous disease (fourth update). Blood Cells, Molecules, and Diseases, 2021, 90, 102587.	0.6	22

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145	Hematologically important mutations: The autosomal forms of chronic granulomatous disease (third) Tj ETQq1 1 0.784314 rgBT /Ove	0.6	22
146	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
147	Inherited human c-Rel deficiency disrupts myeloid and lymphoid immunity to multiple infectious agents. Journal of Clinical Investigation, 2021, 131, .	3.9	21
148	Granulomatous skin lesions, severe scrotal and lower limb edema due to mycobacterial infections in a child with complete IFN-Î³ receptor-1 deficiency. Immunotherapy, 2012, 4, 1121-1127.	1.0	20
149	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
150	Alu-repeat-induced deletions within theNCF2gene causing p67-phox-deficient chronic granulomatous disease (CGD). Human Mutation, 2010, 31, 151-158.	1.1	19
151	Hematopoietic stem cell gene therapy for IFNÎ³R1 deficiency protects mice from mycobacterial infections. Blood, 2018, 131, 533-545.	0.6	19
152	A New Patient with Inherited TYK2 Deficiency. Journal of Clinical Immunology, 2020, 40, 232-235.	2.0	19
153	Leukocyte Adhesion Deficiency Type 1 (LAD1) with Expressed but Nonfunctional CD11/CD18. Journal of Clinical Immunology, 2016, 36, 627-630.	2.0	18
154	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
155	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
156	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
157	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
158	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
159	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
160	Inherited IL-12RÎ²1 Deficiency in a Child With BCG Adenitis and Oral Candidiasis: A Case Report. Pediatrics, 2017, 140, .	1.0	16
161	Patient iPSC-Derived Macrophages to Study Inborn Errors of the IFN-Î³ Responsive Pathway. Cells, 2020, 9, 483.	1.8	16
162	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

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163	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
164	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
165	Accounting for genetic heterogeneity in homozygosity mapping: application to Mendelian susceptibility to mycobacterial disease. Journal of Medical Genetics, 2011, 48, 567-571.	1.5	14
166	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
167	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
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