Stephanie Boisson-Dupuis

List of Publications by Year
in descending order

135 papers	17,633 citations	18436 62 h-index	15218 126 g-index
145 all docs	145 docs citations	145 times ranked	21803 citing authors

#	Article	IF	Citations
1	Human genetic and immunological determinants of critical COVID-19 pneumonia. Nature, 2022, 603, 587-598.	13.7	216
2	Recessive inborn errors of type I IFN immunity in children with COVID-19 pneumonia. Journal of Experimental Medicine, 2022, 219, .	4.2	59
3	Multibatch Cytometry Data Integration for Optimal Immunophenotyping. Journal of Immunology, 2021, 206, 206-213.	0.4	25
4	Inherited GATA2 Deficiency Is Dominant by Haploinsufficiency and Displays Incomplete Clinical Penetrance. Journal of Clinical Immunology, 2021, 41, 639-657.	2.0	30
5	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
6	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
7	Human ancient DNA analyses reveal the high burden of tuberculosis in Europeans over the last 2,000 years. American Journal of Human Genetics, 2021, 108, 517-524.	2.6	58
8	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
9	Human STAT3 variants underlie autosomal dominant hyper-IgE syndrome by negative dominance. Journal of Experimental Medicine, 2021, 218, .	4.2	30
10	Inherited PD-1 deficiency underlies tuberculosis and autoimmunity in a child. Nature Medicine, 2021, 27, 1646-1654.	15.2	65
11	Genetic, Immunological, and Clinical Features of 32 Patients with Autosomal Recessive STAT1 Deficiency. Journal of Immunology, 2021, 207, 133-152.	0.4	33
12	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
13	Biochemically deleterious human NFKB1 variants underlie an autosomal dominant form of common variable immunodeficiency. Journal of Experimental Medicine, 2021, 218, .	4.2	32
14	Mycobacterial diseases in patients with inborn errors of immunity. Current Opinion in Immunology, 2021, 72, 262-271.	2.4	23
15	Identification of discriminative gene-level and protein-level features associated with pathogenic gain-of-function and loss-of-function variants. American Journal of Human Genetics, 2021, 108, 2301-2318.	2.6	21
16	A New Patient with Inherited TYK2 Deficiency. Journal of Clinical Immunology, 2020, 40, 232-235.	2.0	19
17	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,749
18	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,983

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19	Human STAT1 Gain-of-Function Heterozygous Mutations: Chronic Mucocutaneous Candidiasis and Type I Interferonopathy. Journal of Clinical Immunology, 2020, 40, 1065-1081.	2.0	86
20	Human T-bet Governs Innate and Innate-like Adaptive IFN-Î³ Immunity against Mycobacteria. Cell, 2020, 183, 1826-1847.e31.	13.5	83
21	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. Cell, 2020, 181, 1194-1199.	13.5	185
22	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
23	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
24	Patient iPSC-Derived Macrophages to Study Inborn Errors of the IFN-Î³ Responsive Pathway. Cells, 2020, 9, 483.	1.8	16
25	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
26	The monogenic basis of human tuberculosis. Human Genetics, 2020, 139, 1001-1009.	1.8	44
27	Fatal Cytomegalovirus Infection in an Adult with Inherited NOS2 Deficiency. New England Journal of Medicine, 2020, 382, 437-445.	13.9	38
28	Inherited human IFN-Î³ deficiency underlies mycobacterial disease. Journal of Clinical Investigation, 2020, 130, 3158-3171.	3.9	89
29	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.	3.3	17
30	Prevalence and risk factors for latent tuberculosis infection among healthcare workers in Morocco. PLoS ONE, 2019, 14, e0221081.	1.1	17
31	Homozygosity for TYK2 P1104A underlies tuberculosis in about 1% of patients in a cohort of European ancestry. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 10430-10434.	3.3	87
32	Mendelian susceptibility to mycobacterial disease: 2014â€“2018 update. Immunology and Cell Biology, 2019, 97, 360-367.	1.0	163
33	Human IFN-Î³ immunity to mycobacteria is governed by both IL-12 and IL-23. Science Immunology, 2018, 3, .	5.6	152
34	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
35	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
36	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

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37	IRF4 haploinsufficiency in a family with Whippleâ€™s disease. ELife, 2018, 7, .	2.8	43
38	A Variety of Alu-Mediated Copy Number Variations Can Underlie IL-12RÎ²1 Deficiency. Journal of Clinical Immunology, 2018, 38, 617-627.	2.0	45
39	Disruption of an antimycobacterial circuit between dendritic and helper T cells in human SPPL2a deficiency. Nature Immunology, 2018, 19, 973-985.	7.0	96
40	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
41	Life-threatening influenza pneumonitis in a child with inherited IRF9 deficiency. Journal of Experimental Medicine, 2018, 215, 2567-2585.	4.2	146
42	A novel kindred with inherited STAT2 deficiency and severe viral illness. Journal of Allergy and Clinical Immunology, 2017, 139, 1995-1997.e9.	1.5	71
43	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
44	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
45	Inherited IL-12RÎ²1 Deficiency in a Child With BCG Adenitis and Oral Candidiasis: A Case Report. Pediatrics, 2017, 140, .	1.0	16
46	Paracoccidioidomycosis Associated With a Heterozygous STAT4 Mutation and Impaired IFN-Î³ Immunity. Journal of Infectious Diseases, 2017, 216, 1623-1634.	1.9	25
47	AD Hyper-IgE Syndrome Due to a Novel Loss-of-Function Mutation in STAT3: a Diagnostic Pursuit Won by Clinical Acuity. Journal of Clinical Immunology, 2017, 37, 12-17.	2.0	5
48	Visceral leishmaniasis in two patients with ILâ€12p40 and ILâ€12RÎ²1 deficiencies. Pediatric Blood and Cancer, 2017, 64, e26362.	0.8	25
49	Disseminated Tuberculosis and Chronic Mucocutaneous Candidiasis in a Patient with a Gain-of-Function Mutation in Signal Transduction and Activator of Transcription 1. Frontiers in Immunology, 2017, 8, 1651.	2.2	21
50	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
51	Human Genetics of Tuberculosis of the Nervous System. , 2017, , 11-22.		1
52	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
53	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
54	Utility of the QuantiFERON^{Â®}-TB Gold In-Tube assay for the diagnosis of tuberculosis in Moroccan children. International Journal of Tuberculosis and Lung Disease, 2016, 20, 1639-1646.	0.6	9

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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	Characterization of Greater Middle Eastern genetic variation for enhanced disease gene discovery. Nature Genetics, 2016, 48, 1071-1076.	9.4	314
57	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
58	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
59	Severe Mycobacterial Diseases in a Patient with GOF ÎºBÎ± Mutation Without EDA. Journal of Clinical Immunology, 2016, 36, 12-15.	2.0	11
60	A genome-wide association study of pulmonary tuberculosis in Morocco. Human Genetics, 2016, 135, 299-307.	1.8	57
61	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
62	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
63	Inherited and acquired immunodeficiencies underlying tuberculosis in childhood. Immunological Reviews, 2015, 264, 103-120.	2.8	180
64	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
65	Impairment of immunity to Candida and Mycobacterium in humans with bi-allelic RORC mutations. Science, 2015, 349, 606-613.	6.0	366
66	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
67	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
68	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
69	Human TYK2 deficiency: Mycobacterial and viral infections without hyper-IgE syndrome. Journal of Experimental Medicine, 2015, 212, 1641-1662.	4.2	293
70	Pott's disease in Moroccan children: clinical features and investigation of the interleukin-12/interferon-Î³ pathway. International Journal of Tuberculosis and Lung Disease, 2015, 19, 1455-1462.	0.6	10
71	Human intracellular ISG15 prevents interferon-Î±/Î² over-amplification and auto-inflammation. Nature, 2015, 517, 89-93.	13.7	432
72	Clinical Features of Candidiasis in Patients With Inherited Interleukin 12 Receptor Î²1 Deficiency. Clinical Infectious Diseases, 2014, 58, 204-213.	2.9	98

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73	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
74	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
75	Pineal Germinoma in a Child with Interferon-Î³ Receptor 1 Deficiency. Case Report and Literature Review. Journal of Clinical Immunology, 2014, 34, 922-927.	2.0	13
76	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
77	HGCS: an online tool for prioritizing disease-causing gene variants by biological distance. BMC Genomics, 2014, 15, 256.	1.2	43
78	Recurrent Salmonellosis in a Child with Complete IL-12RÎ²1 Deficiency. Journal of Immunodeficiency & Disorders, 2014, 03, .	0.4	9
79	IL-12RÎ²1 Deficiency: Mutation Update and Description of theIL12RB1Variation Database. Human Mutation, 2013, 34, 1329-1339.	1.1	81
80	Multifocal Tuberculous Osteomyelitis: Possible Inherited Interferon Gamma Axis Defect. Indian Journal of Pediatrics, 2013, 80, 505-508.	0.3	4
81	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
82	Haploinsufficiency at the human IFNGR2 locus contributes to mycobacterial disease. Human Molecular Genetics, 2013, 22, 769-781.	1.4	58
83	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
84	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
85	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
86	ISG15: leading a double life as a secreted molecule. Experimental and Molecular Medicine, 2013, 45, e18-e18.	3.2	91
87	Heterozygosity for the Y701C STAT1 mutation in a multiplex kindred with multifocal osteomyelitis. Haematologica, 2013, 98, 1641-1649.	1.7	47
88	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
89	IL-12 receptor Î²1 deficiency alters in vivo T follicular helper cell response in humans. Blood, 2013, 121, 3375-3385.	0.6	147
90	Inherited IL-12p40 Deficiency. Medicine (United States), 2013, 92, 109-122.	0.4	151

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91	Simple diagnosis ofSTAT1gain-of-function alleles in patients with chronic mucocutaneous candidiasis. Journal of Leukocyte Biology, 2013, 95, 667-676.	1.5	77
92	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
93	MENDELIAN SUSCEPTIBILITY TO MYCOBACTERIAL DISEASE IN EGYPTIAN CHILDREN. Mediterranean Journal of Hematology and Infectious Diseases, 2012, 4, e2012033.	0.5	11
94	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
95	Functional STAT3 deficiency compromises the generation of human T follicular helper cells. Blood, 2012, 119, 3997-4008.	0.6	267
96	A Patient with Tyrosine Kinase 2 Deficiency without Hyper-IgE Syndrome. Journal of Pediatrics, 2012, 160, 1055-1057.	0.9	92
97	Mycobacterial Disease and Impaired IFN-Î³ Immunity in Humans with Inherited ISG15 Deficiency. Science, 2012, 337, 1684-1688.	6.0	455
98	Inborn errors of human STAT1: allelic heterogeneity governs the diversity of immunological and infectious phenotypes. Current Opinion in Immunology, 2012, 24, 364-378.	2.4	245
99	Dominant-negative STAT1 SH2 domain mutations in unrelated patients with mendelian susceptibility to mycobacterial disease. Human Mutation, 2012, 33, 1377-1387.	1.1	71
100	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
101	IRF8Mutations and Human Dendritic-Cell Immunodeficiency. New England Journal of Medicine, 2011, 365, 127-138.	13.9	564
102	THE CLINICAL SPECTRUM OF PATIENTS WITH DEFICIENCY OF SIGNAL TRANSDUCER AND ACTIVATOR OF TRANSCRIPTION-1. Pediatric Infectious Disease Journal, 2011, 30, 352-355.	1.1	61
103	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
104	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
105	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
106	Lethal Tuberculosis in a Previously Healthy Adult with IL-12 Receptor Deficiency. Journal of Clinical Immunology, 2011, 31, 537-539.	2.0	49
107	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
108	Treatment of Disseminated Mycobacterial Infection with High-Dose IFN-Î³in a Patient with IL-12RÎ²1 Deficiency. Clinical and Developmental Immunology, 2011, 2011, 1-5.	3.3	54

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109	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
110	IL-12RÎ²1 Deficiency in Two of Fifty Children with Severe Tuberculosis from Iran, Morocco, and Turkey. PLoS ONE, 2011, 6, e18524.	1.1	111
111	Revisiting Human IL-12RÎ²1 Deficiency. Medicine (United States), 2010, 89, 381-402.	0.4	367
112	A novel form of human STAT1 deficiency impairing early but not late responses to interferons. Blood, 2010, 116, 5895-5906.	0.6	93
113	Primary immunodeficiencies of protective immunity to primary infections. Clinical Immunology, 2010, 135, 204-209.	1.4	65
114	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
115	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
116	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
117	Clinical Disease Caused byKlebsiellain 2 Unrelated Patients With Interleukin 12 Receptor Î²1 Deficiency. Pediatrics, 2010, 126, e971-e976.	1.0	31
118	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
119	STAT1-dependent IgG cell-surface expression in a human B cell line derived from a STAT1-deficient patient. Journal of Leukocyte Biology, 2010, 87, 1145-1152.	1.5	11
120	Septins Regulate Bacterial Entry into Host Cells. PLoS ONE, 2009, 4, e4196.	1.1	81
121	SUCCESSFUL HEMATOPOIETIC STEM CELL TRANSPLANTATION FROM AN UNRELATED DONOR IN A CHILD WITH INTERFERON GAMMA RECEPTOR DEFICIENCY. Pediatric Infectious Disease Journal, 2009, 28, 658-660.	1.1	16
122	A partial form of recessive STAT1 deficiency in humans. Journal of Clinical Investigation, 2009, 119, 1502-1514.	3.9	167
123	Novel primary immunodeficiencies revealed by the investigation of paediatric infectious diseases. Current Opinion in Immunology, 2008, 20, 39-48.	2.4	127
124	Inborn errors of interferon (IFN)â€mediated immunity in humans: insights into the respective roles of IFNâ€Î±/Î², IFNâ€Î³, and IFNâ€Î» in host defense. Immunological Reviews, 2008, 226, 29-40.	2.8	271
125	Complementation of a pathogenic IFNGR2 misfolding mutation with modifiers of N-glycosylation. Journal of Biotechnology, 2008, 136, S176.	1.9	0
126	Induction of MxA Gene Expression by Influenza A Virus Requires Type I or Type III Interferon Signaling. Journal of Virology, 2007, 81, 7776-7785.	1.5	205

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127	Corrigendum to â€œInborn errors of IL-12/23- and IFN-Î³-mediated immunity: Molecular, cellular, and clinical featuresâ€•[Semin. Immunol. 18 (2006) 347â€“361]. Seminars in Immunology, 2007, 19, 136-137.	2.7	2
128	Human primary immunodeficiencies of type I interferons. Biochimie, 2007, 89, 878-883.	1.3	57
129	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
130	Novel STAT1 Alleles in Otherwise Healthy Patients with Mycobacterial Disease. PLoS Genetics, 2006, 2, e131.	1.5	171
131	ARHGAP10 is necessary for Î±-catenin recruitment at adherens junctions and for Listeria invasion. Nature Cell Biology, 2005, 7, 954-960.	4.6	106
132	Gains of glycosylation comprise an unexpectedly large group of pathogenic mutations. Nature Genetics, 2005, 37, 692-700.	9.4	198
133	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
134	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
135	Partial Interferonâ€Î³ Receptor Signaling Chain Deficiency in a Patient with Bacille Calmetteâ€GuÃ©rin andMycobacterium abscessusInfection. Journal of Infectious Diseases, 2000, 181, 379-384.	1.9	171