

# Stephanie Boisson-Dupuis

## List of Publications by Year in descending order

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Version: 2024-02-01

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	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. <i>Science</i> , 2020, 370, .	6.0	1,983
2	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. <i>Science</i> , 2020, 370, .	6.0	1,749
3	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.	4.2	739
4	Mendelian susceptibility to mycobacterial disease: Genetic, immunological, and clinical features of inborn errors of IFN- $\gamma$ immunity. <i>Seminars in Immunology</i> , 2014, 26, 454-470.	2.7	582
5	<i>IRF8</i> Mutations and Human Dendritic-Cell Immunodeficiency. <i>New England Journal of Medicine</i> , 2011, 365, 127-138.	13.9	564
6	Mycobacterial Disease and Impaired IFN- $\gamma$ Immunity in Humans with Inherited ISG15 Deficiency. <i>Science</i> , 2012, 337, 1684-1688.	6.0	455
7	Human intracellular ISG15 prevents interferon- $\gamma$ over-amplification and auto-inflammation. <i>Nature</i> , 2015, 517, 89-93.	13.7	432
8	Inborn errors of IL-12/23- and IFN- $\gamma$ -mediated immunity: molecular, cellular, and clinical features. <i>Seminars in Immunology</i> , 2006, 18, 347-361.	2.7	422
9	Revisiting Human IL-12R $\beta$ 1 Deficiency. <i>Medicine (United States)</i> , 2010, 89, 381-402.	0.4	367
10	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.	6.0	366
11	B cell "intrinsic signaling through IL-21 receptor and STAT3 is required for establishing long-lived antibody responses in humans. <i>Journal of Experimental Medicine</i> , 2010, 207, 155-171.	4.2	346
12	Characterization of Greater Middle Eastern genetic variation for enhanced disease gene discovery. <i>Nature Genetics</i> , 2016, 48, 1071-1076.	9.4	314
13	Human TYK2 deficiency: Mycobacterial and viral infections without hyper-IgE syndrome. <i>Journal of Experimental Medicine</i> , 2015, 212, 1641-1662.	4.2	293
14	Inborn errors of interferon (IFN)-mediated immunity in humans: insights into the respective roles of IFN- $\alpha$ , IFN- $\beta$ , and IFN- $\gamma$ in host defense. <i>Immunological Reviews</i> , 2008, 226, 29-40.	2.8	271
15	Functional STAT3 deficiency compromises the generation of human T follicular helper cells. <i>Blood</i> , 2012, 119, 3997-4008.	0.6	267
16	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. <i>Science Immunology</i> , 2021, 6, .	5.6	267
17	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.	7.0	248
18	Inborn errors of human STAT1: allelic heterogeneity governs the diversity of immunological and infectious phenotypes. <i>Current Opinion in Immunology</i> , 2012, 24, 364-378.	2.4	245

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19	Human genetic and immunological determinants of critical COVID-19 pneumonia. <i>Nature</i> , 2022, 603, 587-598.	13.7	216
20	The human gene damage index as a gene-level approach to prioritizing exome variants. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, 13615-13620.	3.3	213
21	Induction of MxA Gene Expression by Influenza A Virus Requires Type I or Type III Interferon Signaling. <i>Journal of Virology</i> , 2007, 81, 7776-7785.	1.5	205
22	Gains of glycosylation comprise an unexpectedly large group of pathogenic mutations. <i>Nature Genetics</i> , 2005, 37, 692-700.	9.4	198
23	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. <i>Cell</i> , 2020, 181, 1194-1199.	13.5	185
24	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.	1.5	181
25	Inherited and acquired immunodeficiencies underlying tuberculosis in childhood. <i>Immunological Reviews</i> , 2015, 264, 103-120.	2.8	180
26	Partial Interferon- $\beta$ Receptor Signaling Chain Deficiency in a Patient with Bacille Calmette-Guérin and Mycobacterium abscessus Infection. <i>Journal of Infectious Diseases</i> , 2000, 181, 379-384.	1.9	171
27	Novel STAT1 Alleles in Otherwise Healthy Patients with Mycobacterial Disease. <i>PLoS Genetics</i> , 2006, 2, e131.	1.5	171
28	A partial form of recessive STAT1 deficiency in humans. <i>Journal of Clinical Investigation</i> , 2009, 119, 1502-1514.	3.9	167
29	Mendelian susceptibility to mycobacterial disease: 2014-2018 update. <i>Immunology and Cell Biology</i> , 2019, 97, 360-367.	1.0	163
30	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.	4.2	158
31	Human IFN- $\beta$ immunity to mycobacteria is governed by both IL-12 and IL-23. <i>Science Immunology</i> , 2018, 3, .	5.6	152
32	Inherited IL-12p40 Deficiency. <i>Medicine (United States)</i> , 2013, 92, 109-122.	0.4	151
33	Tuberculosis and impaired IL-23-dependent IFN- $\beta$ immunity in humans homozygous for a common TYK2 missense variant. <i>Science Immunology</i> , 2018, 3, .	5.6	148
34	IL-12 receptor $\beta$ 1 deficiency alters in vivo T follicular helper cell response in humans. <i>Blood</i> , 2013, 121, 3375-3385.	0.6	147
35	Life-threatening influenza pneumonitis in a child with inherited IRF9 deficiency. <i>Journal of Experimental Medicine</i> , 2018, 215, 2567-2585.	4.2	146
36	Genetic, immunological, and clinical features of patients with bacterial and fungal infections due to inherited IL-17RA deficiency. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, E8277-E8285.	3.3	137

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37	Bacillus Calmette Guérin triggers the IL-12/IFN- $\gamma$ axis by an IRAK-4- and NEMO-dependent, non-cognate interaction between monocytes, NK, and T $\gamma$ lymphocytes. <i>European Journal of Immunology</i> , 2004, 34, 3276-3284.	1.6	133
38	Novel primary immunodeficiencies revealed by the investigation of paediatric infectious diseases. <i>Current Opinion in Immunology</i> , 2008, 20, 39-48.	2.4	127
39	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.	0.6	121
40	Dual T cell $\beta$ and B cell $\beta$ intrinsic deficiency in humans with biallelic <i>RLTPR</i> mutations. <i>Journal of Experimental Medicine</i> , 2016, 213, 2413-2435.	4.2	117
41	IL-12R $\beta$ 2 Deficiency in Two of Fifty Children with Severe Tuberculosis from Iran, Morocco, and Turkey. <i>PLoS ONE</i> , 2011, 6, e18524.	1.1	111
42	ARHGAP10 is necessary for $\beta$ -catenin recruitment at adherens junctions and for Listeria invasion. <i>Nature Cell Biology</i> , 2005, 7, 954-960.	4.6	106
43	Mycobacterial disease in patients with chronic granulomatous disease: A retrospective analysis of 71 cases. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 241-248.e3.	1.5	106
44	A novel form of complete IL-12/IL-23 receptor $\beta$ 1 deficiency with cell surface-expressed nonfunctional receptors. <i>Blood</i> , 2004, 104, 2095-2101.	0.6	103
45	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.	1.4	102
46	Clinical Features of Candidiasis in Patients With Inherited Interleukin 12 Receptor $\beta$ 2 Deficiency. <i>Clinical Infectious Diseases</i> , 2014, 58, 204-213.	2.9	98
47	Disruption of an antimycobacterial circuit between dendritic and helper T cells in human SPPL2a deficiency. <i>Nature Immunology</i> , 2018, 19, 973-985.	7.0	96
48	A novel form of human STAT1 deficiency impairing early but not late responses to interferons. <i>Blood</i> , 2010, 116, 5895-5906.	0.6	93
49	A Patient with Tyrosine Kinase 2 Deficiency without Hyper-IgE Syndrome. <i>Journal of Pediatrics</i> , 2012, 160, 1055-1057.	0.9	92
50	ISG15: leading a double life as a secreted molecule. <i>Experimental and Molecular Medicine</i> , 2013, 45, e18-e18.	3.2	91
51	Inherited human IFN- $\gamma$ deficiency underlies mycobacterial disease. <i>Journal of Clinical Investigation</i> , 2020, 130, 3158-3171.	3.9	89
52	Homozygosity for <i>TYK2</i> P1104A underlies tuberculosis in about 1% of patients in a cohort of European ancestry. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019, 116, 10430-10434.	3.3	87
53	Human STAT1 Gain-of-Function Heterozygous Mutations: Chronic Mucocutaneous Candidiasis and Type I Interferonopathy. <i>Journal of Clinical Immunology</i> , 2020, 40, 1065-1081.	2.0	86
54	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.	1.8	85

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55	Human T-bet Governs Innate and Innate-like Adaptive IFN- $\gamma$ Immunity against Mycobacteria. <i>Cell</i> , 2020, 183, 1826-1847.e31.	13.5	83
56	Septins Regulate Bacterial Entry into Host Cells. <i>PLoS ONE</i> , 2009, 4, e4196.	1.1	81
57	IL-12R $\beta$ 1 Deficiency: Mutation Update and Description of the IL12RB1 Variation Database. <i>Human Mutation</i> , 2013, 34, 1329-1339.	1.1	81
58	Simple diagnosis of STAT1 gain-of-function alleles in patients with chronic mucocutaneous candidiasis. <i>Journal of Leukocyte Biology</i> , 2013, 95, 667-676.	1.5	77
59	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.	4.2	77
60	Dominant-negative STAT1 SH2 domain mutations in unrelated patients with mendelian susceptibility to mycobacterial disease. <i>Human Mutation</i> , 2012, 33, 1377-1387.	1.1	71
61	A novel kindred with inherited STAT2 deficiency and severe viral illness. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 139, 1995-1997.e9.	1.5	71
62	STAT3 is a critical cell-intrinsic regulator of human unconventional T cell numbers and function. <i>Journal of Experimental Medicine</i> , 2015, 212, 855-864.	4.2	70
63	Primary immunodeficiencies of protective immunity to primary infections. <i>Clinical Immunology</i> , 2010, 135, 204-209.	1.4	65
64	Inherited PD-1 deficiency underlies tuberculosis and autoimmunity in a child. <i>Nature Medicine</i> , 2021, 27, 1646-1654.	15.2	65
65	Signal transducer and activator of transcription 3 (STAT3) mutations underlying autosomal dominant hyper-IgE syndrome impair human CD8+ T-cell memory formation and function. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 132, 400-411.e9.	1.5	63
66	THE CLINICAL SPECTRUM OF PATIENTS WITH DEFICIENCY OF SIGNAL TRANSDUCER AND ACTIVATOR OF TRANSCRIPTION-1. <i>Pediatric Infectious Disease Journal</i> , 2011, 30, 352-355.	1.1	61
67	Recessive inborn errors of type I IFN immunity in children with COVID-19 pneumonia. <i>Journal of Experimental Medicine</i> , 2022, 219, .	4.2	59
68	Haploinsufficiency at the human IFNGR2 locus contributes to mycobacterial disease. <i>Human Molecular Genetics</i> , 2013, 22, 769-781.	1.4	58
69	Human ancient DNA analyses reveal the high burden of tuberculosis in Europeans over the last 2,000 years. <i>American Journal of Human Genetics</i> , 2021, 108, 517-524.	2.6	58
70	Human primary immunodeficiencies of type I interferons. <i>Biochimie</i> , 2007, 89, 878-883.	1.3	57
71	A genome-wide association study of pulmonary tuberculosis in Morocco. <i>Human Genetics</i> , 2016, 135, 299-307.	1.8	57
72	Treatment of Disseminated Mycobacterial Infection with High-Dose IFN- $\gamma$ in a Patient with IL-12R $\beta$ 1 Deficiency. <i>Clinical and Developmental Immunology</i> , 2011, 2011, 1-5.	3.3	54

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73	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
74	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
75	Lethal Tuberculosis in a Previously Healthy Adult with IL-12 Receptor Deficiency. Journal of Clinical Immunology, 2011, 31, 537-539.	2.0	49
76	Heterozygosity for the Y701C STAT1 mutation in a multiplex kindred with multifocal osteomyelitis. Haematologica, 2013, 98, 1641-1649.	1.7	47
77	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
78	Age-Dependent Association between Pulmonary Tuberculosis and Common TOX Variants in the 8q12-13 Linkage Region. American Journal of Human Genetics, 2013, 92, 407-414.	2.6	46
79	A Variety of Alu-Mediated Copy Number Variations Can Underlie IL-12R $\beta$ 1 Deficiency. Journal of Clinical Immunology, 2018, 38, 617-627.	2.0	45
80	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
81	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
82	The monogenic basis of human tuberculosis. Human Genetics, 2020, 139, 1001-1009.	1.8	44
83	HGCS: an online tool for prioritizing disease-causing gene variants by biological distance. BMC Genomics, 2014, 15, 256.	1.2	43
84	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
85	IRF4 haploinsufficiency in a family with Whipple's disease. ELife, 2018, 7, .	2.8	43
86	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
87	Fatal Cytomegalovirus Infection in an Adult with Inherited NOS2 Deficiency. New England Journal of Medicine, 2020, 382, 437-445.	13.9	38
88	A novel form of cell type-specific partial IFN- $\gamma$ R1 deficiency caused by a germ line mutation of the IFNGR1 initiation codon. Human Molecular Genetics, 2010, 19, 434-444.	1.4	36
89	Partial IFN- $\gamma$ R2 deficiency is due to protein misfolding and can be rescued by inhibitors of glycosylation. Blood, 2013, 122, 2390-2401.	0.6	34
90	Multiple cutaneous squamous cell carcinomas in a patient with interferon $\gamma$ receptor 2 (IFN $\gamma$ R2) deficiency. Journal of Medical Genetics, 2010, 47, 631-634.	1.5	33

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91	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
92	Genetic, Immunological, and Clinical Features of 32 Patients with Autosomal Recessive STAT1 Deficiency. Journal of Immunology, 2021, 207, 133-152.	0.4	33
93	Biochemically deleterious human <i>NFKB1</i> variants underlie an autosomal dominant form of common variable immunodeficiency. Journal of Experimental Medicine, 2021, 218, .	4.2	32
94	Clinical Disease Caused by <i>Klebsiella</i> in 2 Unrelated Patients With Interleukin 12 Receptor $\beta 21$ Deficiency. Pediatrics, 2010, 126, e971-e976.	1.0	31
95	Association Study of Genes Controlling IL-12-dependent IFN- $\gamma$ Immunity: STAT4 Alleles Increase Risk of Pulmonary Tuberculosis in Morocco. Journal of Infectious Diseases, 2014, 210, 611-618.	1.9	31
96	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
97	Inherited GATA2 Deficiency Is Dominant by Haploinsufficiency and Displays Incomplete Clinical Penetrance. Journal of Clinical Immunology, 2021, 41, 639-657.	2.0	30
98	Human <i>STAT3</i> variants underlie autosomal dominant hyper-IgE syndrome by negative dominance. Journal of Experimental Medicine, 2021, 218, .	4.2	30
99	Paracoccidioidomycosis Associated With a Heterozygous STAT4 Mutation and Impaired IFN- $\gamma$ Immunity. Journal of Infectious Diseases, 2017, 216, 1623-1634.	1.9	25
100	Visceral leishmaniasis in two patients with IL-12p40 and IL-12R $\beta 21$ deficiencies. Pediatric Blood and Cancer, 2017, 64, e26362.	0.8	25
101	Multibatch Cytometry Data Integration for Optimal Immunophenotyping. Journal of Immunology, 2021, 206, 206-213.	0.4	25
102	Mycobacterial diseases in patients with inborn errors of immunity. Current Opinion in Immunology, 2021, 72, 262-271.	2.4	23
103	Paternal uniparental isodisomy of chromosome 6 causing a complex syndrome including complete IFN- $\gamma$ receptor 1 deficiency. American Journal of Medical Genetics, Part A, 2010, 152A, 622-629.	0.7	22
104	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
105	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
106	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
107	Granulomatous skin lesions, severe scrotal and lower limb edema due to mycobacterial infections in a child with complete IFN- $\gamma$ receptor-1 deficiency. Immunotherapy, 2012, 4, 1121-1127.	1.0	20
108	Mycobacterium simiae Infection in Two Unrelated Patients with Different Forms of Inherited IFN- $\gamma$ R2 Deficiency. Journal of Clinical Immunology, 2014, 34, 904-909.	2.0	20

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109	A New Patient with Inherited TYK2 Deficiency. <i>Journal of Clinical Immunology</i> , 2020, 40, 232-235.	2.0	19
110	Microbial Disease Spectrum Linked to a Novel IL-12R $\beta$ 1 N-Terminal Signal Peptide Stop-Gain Homozygous Mutation with Paradoxical Receptor Cell-Surface Expression. <i>Frontiers in Microbiology</i> , 2017, 8, 616.	1.5	18
111	Transduction of <i>Herpesvirus saimiri</i> -Transformed T Cells with Exogenous Genes of Interest. <i>Current Protocols in Immunology</i> , 2016, 115, 7.21C.1-7.21C.12.	3.6	17
112	A deep intronic splice mutation of <i>STAT3</i> underlies hyper IgE syndrome by negative dominance. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019, 116, 16463-16472.	3.3	17
113	Prevalence and risk factors for latent tuberculosis infection among healthcare workers in Morocco. <i>PLoS ONE</i> , 2019, 14, e0221081.	1.1	17
114	Genome-wide association study of resistance to <i>Mycobacterium tuberculosis</i> infection identifies a locus at 10q26.2 in three distinct populations. <i>PLoS Genetics</i> , 2021, 17, e1009392.	1.5	17
115	SUCCESSFUL HEMATOPOIETIC STEM CELL TRANSPLANTATION FROM AN UNRELATED DONOR IN A CHILD WITH INTERFERON GAMMA RECEPTOR DEFICIENCY. <i>Pediatric Infectious Disease Journal</i> , 2009, 28, 658-660.	1.1	16
116	Inherited IL-12R $\beta$ 1 Deficiency in a Child With BCG Adenitis and Oral Candidiasis: A Case Report. <i>Pediatrics</i> , 2017, 140, .	1.0	16
117	Patient iPSC-Derived Macrophages to Study Inborn Errors of the IFN- $\beta$ Responsive Pathway. <i>Cells</i> , 2020, 9, 483.	1.8	16
118	Major Loci on Chromosomes 8q and 3q Control Interferon $\beta$ Production Triggered by <i>Bacillus Calmette-Guerin</i> and 6-kDa Early Secretory Antigen Target, Respectively, in Various Populations. <i>Journal of Infectious Diseases</i> , 2016, 213, 1173-1179.	1.9	15
119	Accounting for genetic heterogeneity in homozygosity mapping: application to Mendelian susceptibility to mycobacterial disease. <i>Journal of Medical Genetics</i> , 2011, 48, 567-571.	1.5	14
120	A purely quantitative form of partial recessive IFN- $\beta$ 2 deficiency caused by mutations of the initiation or second codon. <i>Human Molecular Genetics</i> , 2018, 27, 3919-3935.	1.4	14
121	Pineal Germinoma in a Child with Interferon- $\beta$ Receptor 1 Deficiency. Case Report and Literature Review. <i>Journal of Clinical Immunology</i> , 2014, 34, 922-927.	2.0	13
122	Impaired IL-12- and IL-23-Mediated Immunity Due to IL-12R $\beta$ 1 Deficiency in Iranian Patients with Mendelian Susceptibility to Mycobacterial Disease. <i>Journal of Clinical Immunology</i> , 2018, 38, 787-793.	2.0	13
123	STAT1-dependent IgG cell-surface expression in a human B cell line derived from a STAT1-deficient patient. <i>Journal of Leukocyte Biology</i> , 2010, 87, 1145-1152.	1.5	11
124	MENDELIAN SUSCEPTIBILITY TO MYCOBACTERIAL DISEASE IN EGYPTIAN CHILDREN. <i>Mediterranean Journal of Hematology and Infectious Diseases</i> , 2012, 4, e2012033.	0.5	11
125	Severe Mycobacterial Diseases in a Patient with GOF $\beta$ 1 Mutation Without EDA. <i>Journal of Clinical Immunology</i> , 2016, 36, 12-15.	2.0	11
126	Pott's disease in Moroccan children: clinical features and investigation of the interleukin-12/interferon- $\beta$ pathway. <i>International Journal of Tuberculosis and Lung Disease</i> , 2015, 19, 1455-1462.	0.6	10



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127	Utility of the QuantiFERON <sup>®</sup> -TB Gold In-Tube assay for the diagnosis of tuberculosis in Moroccan children. <i>International Journal of Tuberculosis and Lung Disease</i> , 2016, 20, 1639-1646.	0.6	9
128	Recurrent Salmonellosis in a Child with Complete IL-12 $\beta$ 1 Deficiency. <i>Journal of Immunodeficiency &amp; Disorders</i> , 2014, 03, .	0.4	9
129	Autosomal Dominant IFN- $\gamma$ 1 Deficiency Presenting with both Atypical Mycobacteriosis and Tuberculosis in a BCG-Vaccinated South African Patient. <i>Journal of Clinical Immunology</i> , 2018, 38, 460-463.	2.0	8
130	An eQTL variant of ZXDC is associated with IFN- $\gamma$ production following Mycobacterium tuberculosis antigen-specific stimulation. <i>Scientific Reports</i> , 2017, 7, 12800.	1.6	5
131	AD Hyper-IgE Syndrome Due to a Novel Loss-of-Function Mutation in STAT3: a Diagnostic Pursuit Won by Clinical Acuity. <i>Journal of Clinical Immunology</i> , 2017, 37, 12-17.	2.0	5
132	Multifocal Tuberculous Osteomyelitis: Possible Inherited Interferon Gamma Axis Defect. <i>Indian Journal of Pediatrics</i> , 2013, 80, 505-508.	0.3	4
133	Corrigendum to "Inborn errors of IL-12/23- and IFN- $\gamma$ -mediated immunity: Molecular, cellular, and clinical features" [Semin. Immunol. 18 (2006) 347-361]. <i>Seminars in Immunology</i> , 2007, 19, 136-137.	2.7	2
134	Human Genetics of Tuberculosis of the Nervous System. , 2017, , 11-22.		1
135	Complementation of a pathogenic IFNGR2 misfolding mutation with modifiers of N-glycosylation. <i>Journal of Biotechnology</i> , 2008, 136, S176.	1.9	0