Jacinta Bustamante

List of Publications by Year in Descending Order

231 papers	17,326 citations	65 h-index	129 g-index
245 ext. papers	21,483 ext. citations	9.9 avg, IF	5.87 L-index

#	Paper	IF	Citations
231	Pulmonary Alveolar Proteinosis and Multiple Infectious Diseases in a Child with Autosomal Recessive Complete IRF8 Deficiency.. *Journal of Clinical Immunology*, **2022**, 1	5.7	0
230	Diagnosis of APS-1 in Two Siblings Following Life-Threatening COVID-19 Pneumonia.. *Journal of Clinical Immunology*, **2022**, 1	5.7	0
229	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	6.2	0
228	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	5.6	0
227	SCID and Other Inborn Errors of Immunity with Low TRECs - the Brazilian Experience.. *Journal of Clinical Immunology*, **2022**, 1	5.7	0
226	Human OTULIN haploinsufficiency impairs cell-intrinsic immunity to staphylococcal □toxin.. *Science* , **2022**, eabm6380	33.3	1
225	Cutaneous Granulomatosis Revealing Whipple's Disease: Value of Polymerase Chain Reaction Assay for the Diagnosis. *Pathogens*, **2021**, 10,	4.5	0
224	Clinical and Genetic Spectrum of Inborn Errors of Immunity in a Tertiary Care Center in Southern India. *Indian Journal of Pediatrics*, **2021**, 1	3	1
223	GATA2 deficiency phenotype associated with tandem duplication GATA2 and over-expression of GATA2-AS1. *Blood Advances*, **2021**,	7.8	1
222	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	1.6	0
221	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,	11.5	16
220	Detection of homozygous and hemizygous complete or partial exon deletions by whole-exome sequencing. *NAR Genomics and Bioinformatics*, **2021**, 3, lqab037	3.7	2
219	Disseminated Tuberculosis in a Patient with Autosomal Recessive p47 Chronic Granulomatous Disease. *Journal of Clinical Immunology*, **2021**, 41, 1417-1419	5.7	0
218	Disseminated Mycobacterium simiae Infection in a Patient with Complete IL-12p40 Deficiency. *Iranian Journal of Allergy, Asthma and Immunology*, **2021**, 20, 376-381	1.1	
217	High Th2 cytokine levels and upper airway inflammation in human inherited T-bet deficiency. *Journal of Experimental Medicine*, **2021**, 218,	16.6	7
216	Inherited PD-1 deficiency underlies tuberculosis and autoimmunity in a child. *Nature Medicine*, **2021** , 27, 1646-1654	50.5	17
215	Genetic, Immunological, and Clinical Features of 32 Patients with Autosomal Recessive STAT1 Deficiency. *Journal of Immunology*, **2021**,	5.3	3

214	Improving the diagnostic efficiency of primary immunodeficiencies with targeted next-generation sequencing. *Journal of Allergy and Clinical Immunology*, **2021**, 147, 734-737	11.5	8
213	Multibatch Cytometry Data Integration for Optimal Immunophenotyping. *Journal of Immunology*, **2021**, 206, 206-213	5.3	13
212	Herpes simplex encephalitis in a patient with a distinctive form of inherited IFNAR1 deficiency. *Journal of Clinical Investigation*, **2021**, 131,	15.9	34
211	Inherited GATA2 Deficiency Is Dominant by Haploinsufficiency and Displays Incomplete Clinical Penetrance. *Journal of Clinical Immunology*, **2021**, 41, 639-657	5.7	9
210	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,	11.5	14
209	EDA-ID: a Severe Clinical Presentation Associated with a New IKBKG Mutation. *Journal of Clinical Immunology*, **2021**, 41, 1099-1102	5.7	1
208	Clinical and Molecular Findings in Mendelian Susceptibility to Mycobacterial Diseases: Experience From India. *Frontiers in Immunology*, **2021**, 12, 631298	8.4	8
207	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	6	3
206	Impaired respiratory burst contributes to infections in PKC□deficient patients. *Journal of Experimental Medicine*, **2021**, 218,	16.6	3
205	Leukocytoclastic vasculitis in patients with IL12B or IL12RB1 deficiency: case report and review of the literature. *Pediatric Rheumatology*, **2021**, 19, 121	3.5	2
204	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. *Science Immunology*, **2021**, 6,	28	67
203	Inherited human c-Rel deficiency disrupts myeloid and lymphoid immunity to multiple infectious agents. *Journal of Clinical Investigation*, **2021**, 131,	15.9	3
202	Biochemically deleterious human NFKB1 variants underlie an autosomal dominant form of common variable immunodeficiency. *Journal of Experimental Medicine*, **2021**, 218,	16.6	6
201	Hematologically important mutations: X-linked chronic granulomatous disease (fourth update). *Blood Cells, Molecules, and Diseases*, **2021**, 90, 102587	2.1	3
200	Mycobacterial diseases in patients with inborn errors of immunity. *Current Opinion in Immunology*, **2021**, 72, 262-271	7.8	4
199	Hematologically important mutations: The autosomal forms of chronic granulomatous disease (third update). *Blood Cells, Molecules, and Diseases*, **2021**, 92, 102596	2.1	3
198	Human T-bet Governs Innate and Innate-like Adaptive IFN-□mmunity against Mycobacteria. *Cell*, **2020**, 183, 1826-1847.e31	56.2	35
197	Human Lentiviral Gene Therapy Restores the Cellular Phenotype of Autosomal Recessive Complete IFN-R1 Deficiency. *Molecular Therapy - Methods and Clinical Development*, **2020**, 17, 785-795	6.4	5

196	Systemic Type I IFN Inflammation in Human ISG15 Deficiency Leads to Necrotizing Skin Lesions. *Cell Reports*, **2020**, 31, 107633	10.6	19
195	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. *Cell*, **2020**, 181, 1194-1199	56.2	113
194	Three Copies of Four Interferon Receptor Genes Underlie a Mild Type I Interferonopathy in Down Syndrome. *Journal of Clinical Immunology*, **2020**, 40, 807-819	5.7	16
193	Transient Decrease of Circulating and Tissular Dendritic Cells in Patients With Mycobacterial Disease and With Partial Dominant IFNR1 Deficiency. *Frontiers in Immunology*, **2020**, 11, 1161	8.4	3
192	Mendelian Susceptibility to Mycobacterial Disease (MSMD): Clinical and Genetic Features of 32 Iranian Patients. *Journal of Clinical Immunology*, **2020**, 40, 872-882	5.7	9
191	Autosomal recessive complete STAT1 deficiency caused by compound heterozygous intronic mutations. *International Immunology*, **2020**, 32, 663-671	4.9	9
190	Patient iPSC-Derived Macrophages to Study Inborn Errors of the IFN-Responsive Pathway. *Cells*, **2020**, 9,	7.9	8
189	Genetic, Immunological, and Clinical Features of the First Mexican Cohort of Patients with Chronic Granulomatous Disease. *Journal of Clinical Immunology*, **2020**, 40, 475-493	5.7	21
188	JAK Inhibitor Therapy in a Child with Inherited USP18 Deficiency. *New England Journal of Medicine*, **2020**, 382, 256-265	59.2	32
187	Inherited CARD9 Deficiency in a Patient with Both Exophiala spinifera and Aspergillus nomius Severe Infections. *Journal of Clinical Immunology*, **2020**, 40, 359-366	5.7	13
186	Fatal Cytomegalovirus Infection in an Adult with Inherited NOS2 Deficiency. *New England Journal of Medicine*, **2020**, 382, 437-445	59.2	21
185	Mendelian susceptibility to mycobacterial disease: recent discoveries. *Human Genetics*, **2020**, 139, 993-1000	6.9	63
184	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	2.3	4
183	Inherited human IFN-deficiency underlies mycobacterial disease. *Journal of Clinical Investigation*, **2020**, 130, 3158-3171	15.9	40
182	A New Patient with Inherited TYK2 Deficiency. *Journal of Clinical Immunology*, **2020**, 40, 232-235	5.7	11
181	Perianal Disease and Granulomas: Think Out of the Box *GE Portuguese Journal of Gastroenterology*, **2020**, 27, 119-123	1.1	
180	Mutual alteration of NOD2-associated Blau syndrome and IFNR1 deficiency. *Journal of Clinical Immunology*, **2020**, 40, 165-178	5.7	8
179	Interleukin 12-23 deficiency in the interferon gamma pathway in a 6-month-old toddler who has BCG vaccine complications. *Dermatologic Therapy*, **2020**, 33, e13999	2.2	1

178	Disseminated Infectious Disease Caused by Histoplasma capsulatum in an Adult Patient as First Manifestation of Inherited IL-12Rβ1 Deficiency. *Journal of Clinical Immunology*, **2020**, 40, 1051-1054	5.7	2
177	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	11.5	5
176	Homozygous STAT2 gain-of-function mutation by loss of USP18 activity in a patient with type I interferonopathy. *Journal of Experimental Medicine*, **2020**, 217,	16.6	34
175	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. *Science*, **2020**, 370,	33.3	994
174	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. *Science*, **2020**, 370,	33.3	1090
173	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	3.2	3
172	Nocardiosis Associated with Primary Immunodeficiencies (Nocar-DIP): an International Retrospective Study and Literature Review. *Journal of Clinical Immunology*, **2020**, 40, 1144-1155	5.7	6
171	Successful Hematopoietic Stem Cell Transplantation in a Patient with Complete IFN-γ Receptor 2 Deficiency: a Case Report and Literature Review. *Journal of Clinical Immunology*, **2020**, 40, 1191-1195	5.7	5
170	A Novel Recessive Mutation of Interferon-γ Receptor 1 in a Patient with Mycobacterium tuberculosis in Bone Marrow Aspirate. *Journal of Clinical Immunology*, **2019**, 39, 127-130	5.7	5
169	Molecular, Immunological, and Clinical Features of 16 Iranian Patients with Mendelian Susceptibility to Mycobacterial Disease. *Journal of Clinical Immunology*, **2019**, 39, 287-297	5.7	8
168	Life-Threatening Infections Due to Live-Attenuated Vaccines: Early Manifestations of Inborn Errors of Immunity. *Journal of Clinical Immunology*, **2019**, 39, 376-390	5.7	26
167	Recurrent Salmonella typhi Infection and Autoimmunity in a Young Boy with Complete IL-12 Receptor β1 Deficiency. *Journal of Clinical Immunology*, **2019**, 39, 358-362	5.7	6
166	Human DOCK2 Deficiency: Report of a Novel Mutation and Evidence for Neutrophil Dysfunction. *Journal of Clinical Immunology*, **2019**, 39, 298-308	5.7	17
165	Immunodeficiencies at the Interface of Innate and Adaptive Immunity **2019**, 509-522.e1		
164	LINE-1-Mediated AluYa5 Insertion Underlying Complete Autosomal Recessive IFN-γR1 Deficiency. *Journal of Clinical Immunology*, **2019**, 39, 739-742	5.7	4
163	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	5.7	0
162	Hematopoietic stem cell transplant effectively rescues lymphocyte differentiation and function in DOCK8-deficient patients. *JCI Insight*, **2019**, 5,	9.9	12
161	Rescue of recurrent deep intronic mutation underlying cell type-dependent quantitative NEMO deficiency. *Journal of Clinical Investigation*, **2019**, 129, 583-597	15.9	25

160	Skewed X-inactivation in a Female Carrier with X-linked Chronic Granulomatous Disease. *Iranian Journal of Allergy, Asthma and Immunology*, **2019**, 18, 447-451	1.1	1
159	IFN-γ and CD25 drive distinct pathologic features during hemophagocytic lymphohistiocytosis. *Journal of Allergy and Clinical Immunology*, **2019**, 143, 2215-2226.e7	11.5	33
158	Mendelian susceptibility to mycobacterial disease: 2014-2018 update. *Immunology and Cell Biology*, **2019**, 97, 360-367	5	99
157	Laboratory evaluation of the IFN-γ circuit for the molecular diagnosis of Mendelian susceptibility to mycobacterial disease. *Critical Reviews in Clinical Laboratory Sciences*, **2018**, 55, 184-204	9.4	27
156	Disseminated abscesses due to Mycoplasma faucium in a patient with activated PI3Kδ syndrome type 2. *Journal of Allergy and Clinical Immunology: in Practice*, **2018**, 6, 1796-1798.e2	5.4	4
155	Mendelian Susceptibility to Mycobacterial Disease Caused by a Novel Founder IL12B Mutation in Saudi Arabia. *Journal of Clinical Immunology*, **2018**, 38, 278-282	5.7	6
154	Severe BCG-osis Misdiagnosed as Multidrug-Resistant Tuberculosis in an IL-12Rβ1-Deficient Peruvian Girl. *Journal of Clinical Immunology*, **2018**, 38, 712-716	5.7	5
153	IRF4 haploinsufficiency in a family with Whipple's disease. *ELife*, **2018**, 7,	8.9	25
152	A Variety of Alu-Mediated Copy Number Variations Can Underlie IL-12Rβ1 Deficiency. *Journal of Clinical Immunology*, **2018**, 38, 617-627	5.7	26
151	Natural history of GATA2 deficiency in a survey of 79 French and Belgian patients. *Haematologica*, **2018**, 103, 1278-1287	6.6	74
150	Disruption of an antimycobacterial circuit between dendritic and helper T cells in human SPPL2a deficiency. *Nature Immunology*, **2018**, 19, 973-985	19.1	67
149	A purely quantitative form of partial recessive IFN-γR2 deficiency caused by mutations of the initiation or second codon. *Human Molecular Genetics*, **2018**, 27, 3919-3935	5.6	9
148	A recessive form of hyper-IgE syndrome by disruption of ZNF341-dependent STAT3 transcription and activity. *Science Immunology*, **2018**, 3,	28	82
147	Lethal Influenza in Two Related Adults with Inherited GATA2 Deficiency. *Journal of Clinical Immunology*, **2018**, 38, 513-526	5.7	24
146	Inherited p40phox deficiency differs from classic chronic granulomatous disease. *Journal of Clinical Investigation*, **2018**, 128, 3957-3975	15.9	65
145	Author response: IRF4 haploinsufficiency in a family with Whipple's disease **2018**,		3
144	Hematopoietic stem cell gene therapy for IFNγR1 deficiency protects mice from mycobacterial infections. *Blood*, **2018**, 131, 533-545	2.2	14
143	Human IFN-γ immunity to mycobacteria is governed by both IL-12 and IL-23. *Science Immunology*, **2018**, 3,	28	83

142	Tuberculosis and impaired IL-23-dependent IFN-γ immunity in humans homozygous for a common missense variant. *Science Immunology*, **2018**, 3,	28	88
141	T-cell defects in patients with germline mutations account for combined immunodeficiency. *Blood*, **2018**, 132, 2362-2374	2.2	59
140	Impaired IL-12- and IL-23-Mediated Immunity Due to IL-12Rβ1 Deficiency in Iranian Patients with Mendelian Susceptibility to Mycobacterial Disease. *Journal of Clinical Immunology*, **2018**, 38, 787-793	5.7	10
139	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	5.7	20
138	Copy number variations and founder effect underlying complete IL-10Rβ deficiency in Portuguese kindreds. *PLoS ONE*, **2018**, 13, e0205826	3.7	6
137	Autosomal Dominant IFN-γR1 Deficiency Presenting with both Atypical Mycobacteriosis and Tuberculosis in a BCG-Vaccinated South African Patient. *Journal of Clinical Immunology*, **2018**, 38, 460-463	5.7	6
136	Respiratory Complications Lead to the Diagnosis of Chronic Granulomatous Disease in Two Adult Patients. *Journal of Clinical Immunology*, **2017**, 37, 113-116	5.7	4
135	Systemic Human ILC Precursors Provide a Substrate for Tissue ILC Differentiation. *Cell*, **2017**, 168, 1086-1100.e10	31.4	193
134	Disseminated Bacillus Calmette-Guérin Osteomyelitis in Twin Sisters Related to STAT1 Gene Deficiency. *Pediatric and Developmental Pathology*, **2017**, 20, 255-261	2.2	9
133	Chronic Granulomatous Disease in Patients Reaching Adulthood: A Nationwide Study in France. *Clinical Infectious Diseases*, **2017**, 64, 767-775	11.6	38
132	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	11.5	26
131	An eQTL variant of ZXDC is associated with IFN-γ production following Mycobacterium tuberculosis antigen-specific stimulation. *Scientific Reports*, **2017**, 7, 12800	4.9	3
130	Inherited IL-12Rβ1 Deficiency in a Child With BCG Adenitis and Oral Candidiasis: A Case Report. *Pediatrics*, **2017**, 140,	7.4	8
129	Paracoccidioidomycosis Associated With a Heterozygous STAT4 Mutation and Impaired IFN-γ Immunity. *Journal of Infectious Diseases*, **2017**, 216, 1623-1634	7	19
128	Severe Enteropathy and Hypogammaglobulinemia Complicating Refractory Mycobacterium tuberculosis Complex Disseminated Disease in a Child with IL-12Rβ1 Deficiency. *Journal of Clinical Immunology*, **2017**, 37, 732-738	5.7	9
127	Hematopoietic stem cell transplantation in 29 patients hemizygous for hypomorphic /NEMO mutations. *Blood*, **2017**, 130, 1456-1467	2.2	61
126	Visceral leishmaniasis in two patients with IL-12p40 and IL-12Rβ1 deficiencies. *Pediatric Blood and Cancer*, **2017**, 64, e26362	3	18
125	Microbial Disease Spectrum Linked to a Novel IL-12Rβ1 N-Terminal Signal Peptide Stop-Gain Homozygous Mutation with Paradoxical Receptor Cell-Surface Expression. *Frontiers in Microbiology*, **2017**, 8, 616	5.7	12

124	Multifocal Recurrent Osteomyelitis and Hemophagocytic Lymphohistiocytosis in a Boy with Partial Dominant IFN-γR1 Deficiency: Case Report and Review of the Literature. *Frontiers in Pediatrics*, **2017**, 5, 75	3.4	22
123	Inherited GINS1 deficiency underlies growth retardation along with neutropenia and NK cell deficiency. *Journal of Clinical Investigation*, **2017**, 127, 1991-2006	15.9	73
122	IL12Rβ1 defect presenting with massive intra-abdominal lymphadenopathy due to Mycobacterium intracellulare infection. *Asian Pacific Journal of Allergy and Immunology*, **2017**, 35, 161-165	5.4	2
121	Dual T cell- and B cell-intrinsic deficiency in humans with biallelic RLTPR mutations. *Journal of Experimental Medicine*, **2016**, 213, 2413-2435	16.6	75
120	Use of corticosteroids as an alternative to surgical treatment for liver abscesses in chronic granulomatous disease. *Pediatric Blood and Cancer*, **2016**, 63, 2254-2255	3	3
119	Transduction of Herpesvirus saimiri-Transformed T Cells with Exogenous Genes of Interest. *Current Protocols in Immunology*, **2016**, 115, 7.21C.1-7.21C.12	4	15
118	Disseminated bacille Calmette-Guérin disease in an infant with a novel biallelic mutation in interferon gamma receptor-1 gene. *Pediatric Infectious Disease*, **2016**, 8, 84-87		1
117	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, 6713-8	11.5	37
116	Anti-IFN-γ 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, 945-8.e8	11.5	41
115	Severe Mycobacterial Diseases in a Patient with GOF IKBA Mutation Without EDA. *Journal of Clinical Immunology*, **2016**, 36, 12-5	5.7	11
114	Interferon-gamma-dependent Immunity in Bacillus Calmette-Guérin Vaccine Osteitis Survivors. *Pediatric Infectious Disease Journal*, **2016**, 35, 690-4	3.4	9
113	IL-12 drives functional plasticity of human group 2 innate lymphoid cells. *Journal of Experimental Medicine*, **2016**, 213, 569-83	16.6	194
112	Mycobacterial disease in patients with chronic granulomatous disease: A´retrospective analysis of 71 cases. *Journal of Allergy and Clinical Immunology*, **2016**, 138, 241-248.e3	11.5	76
111	Major Loci on Chromosomes 8q and 3q Control Interferon-γ Production Triggered by Bacillus Calmette-Guerin and 6-kDa Early Secretory Antigen Target, Respectively, in Various Populations. *Journal of Infectious Diseases*, **2016**, 213, 1173-9	7	14
110	Mendelian Susceptibility to Mycobacterial Disease due to IL-12Rβ1 Deficiency in Three Iranian Children. *Iranian Journal of Public Health*, **2016**, 45, 249-54	0.7	6
109	Mendelian Susceptibility to Mycobacterial Disease due to IL-12Rβ1 Deficiency in Three Iranian Children. *Iranian Journal of Public Health*, **2016**, 45, 370-5	0.7	5
108	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	0.7	11
107	Mendelian Susceptibility to Infections with Viruses, Mycobacteria, Bacteria, and Candida **2016**, 407-415		

106	Interferon-□Autoantibodies as Predisposing Factor for Nontuberculous Mycobacterial Infection. *Emerging Infectious Diseases*, **2016**, 22, 1124-1126	10.2	30
105	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, 1589-608	16.6	51
104	Disseminated BCG osteomyelitis related to STAT 1 gene deficiency mimicking a metastatic neuroblastoma. *Pediatric and Developmental Pathology*, **2016**,	2.2	3
103	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	11.5	94
102	Role of Flow Cytometry in the Diagnosis of Chronic Granulomatous Disease: the Egyptian Experience. *Journal of Clinical Immunology*, **2016**, 36, 610-8	5.7	15
101	Leukocyte Adhesion Deficiency Type 1 (LAD1) with Expressed but Nonfunctional CD11/CD18. *Journal of Clinical Immunology*, **2016**, 36, 627-30	5.7	7
100	Inherited and acquired immunodeficiencies underlying tuberculosis in childhood. *Immunological Reviews*, **2015**, 264, 103-20	11.3	133
99	Phagocyte nicotinamide adenine dinucleotide phosphate oxidase activity in patients with inherited IFN-R1 or IFN-R2 deficiency. *Journal of Allergy and Clinical Immunology*, **2015**, 135, 1393-5.e1	11.5	9
98	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	11.5	126
97	IMMUNODEFICIENCIES. Impairment of immunity to Candida and Mycobacterium in humans with bi-allelic RORC mutations. *Science*, **2015**, 349, 606-613	33.3	291
96	Disseminated Mycobacterium avium complex infection in a child with partial dominant interferon gamma receptor 1 deficiency in India. *Journal of Clinical Immunology*, **2015**, 35, 459-62	5.7	15
95	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, 317-21	7	34
94	Inherited IL-17RC deficiency in patients with chronic mucocutaneous candidiasis. *Journal of Experimental Medicine*, **2015**, 212, 619-31	16.6	130
93	STAT3 is a critical cell-intrinsic regulator of human unconventional T cell numbers and function. *Journal of Experimental Medicine*, **2015**, 212, 855-64	16.6	54
92	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, 13615-20	11.5	152
91	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, 2101-7	3	46
90	Human TYK2 deficiency: Mycobacterial and viral infections without hyper-IgE syndrome. *Journal of Experimental Medicine*, **2015**, 212, 1641-62	16.6	209
89	Human intracellular ISG15 prevents interferon-□over-amplification and auto-inflammation. *Nature*, **2015**, 517, 89-93	50.4	311

88	Diagnostic and therapeutic challenges in a child with complete interferon-γ receptor 1 deficiency. *Pediatric Blood and Cancer*, **2015**, 62, 2036-9	3	23
87	Chronic granulomatous disease in Morocco: genetic, immunological, and clinical features of 12 patients from 10 kindreds. *Journal of Clinical Immunology*, **2014**, 34, 452-8	5.7	12
86	ICON: the early diagnosis of congenital immunodeficiencies. *Journal of Clinical Immunology*, **2014**, 34, 398-424	5.7	26
85	Clinical features of Candidiasis in patients with inherited interleukin 12 receptor β1 deficiency. *Clinical Infectious Diseases*, **2014**, 58, 204-13	11.6	81
84	Association study of genes controlling IL-12-dependent IFN-γ immunity: STAT4 alleles increase risk of pulmonary tuberculosis in Morocco. *Journal of Infectious Diseases*, **2014**, 210, 611-8	7	24
83	Mendelian susceptibility to mycobacterial disease: genetic, immunological, and clinical features of inborn errors of IFN-γ immunity. *Seminars in Immunology*, **2014**, 26, 454-70	10.7	401
82	Mycobacterium simiae infection in two unrelated patients with different forms of inherited IFN-γR2 deficiency. *Journal of Clinical Immunology*, **2014**, 34, 904-9	5.7	16
81	Recurrent Salmonellosis in a Child with Complete IL-12Rβ1 Deficiency **2014**, 3,		7
80	IL-12Rβ1 deficiency: mutation update and description of the IL12RB1 variation database. *Human Mutation*, **2013**, 34, 1329-39	4.7	56
79	Partial IFN-γR2 deficiency is due to protein misfolding and can be rescued by inhibitors of glycosylation. *Blood*, **2013**, 122, 2390-401	2.2	27
78	Haploinsufficiency at the human IFNGR2 locus contributes to mycobacterial disease. *Human Molecular Genetics*, **2013**, 22, 769-81	5.6	53
77	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, 400-11.e9	11.5	48
76	IL-12Rβ1 deficiency and disseminated Mycobacterium tilburgii disease. *Journal of Clinical Immunology*, **2013**, 33, 1285-8	5.7	10
75	A 1-year-old girl with a gain-of-function STAT1 mutation treated with hematopoietic stem cell transplantation. *Journal of Clinical Immunology*, **2013**, 33, 1273-5	5.7	41
74	IL-21 signalling via STAT3 primes human naive B cells to respond to IL-2 to enhance their differentiation into plasmablasts. *Blood*, **2013**, 122, 3940-50	2.2	84
73	Age-dependent association between pulmonary tuberculosis and common TOX variants in the 8q12-13 linkage region. *American Journal of Human Genetics*, **2013**, 92, 407-14	11	42
72	Primary immunodeficiencies underlying fungal infections. *Current Opinion in Pediatrics*, **2013**, 25, 736-47	3.2	149
71	Inherited human OX40 deficiency underlying classic Kaposi sarcoma of childhood. *Journal of Experimental Medicine*, **2013**, 210, 1743-59	16.6	99

70	Variant of X-Linked Chronic Granulomatous Disease Revealed by a Severe Burkholderia cepacia Invasive Infection in an Infant. *Case Reports in Immunology*, **2013**, 2013, 323614	1.9	0
69	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, 2739-53	16.6	121
68	IL-12 receptor 1 deficiency alters in vivo T follicular helper cell response in humans. *Blood*, **2013**, 121, 3375-85	2.2	121
67	Inherited IL-12p40 deficiency: genetic, immunologic, and clinical features of 49 patients from 30 kindreds. *Medicine (United States)*, **2013**, 92, 109-122	1.8	121
66	A novel homozygous p.R1105X mutation of the AP4E1 gene in twins with hereditary spastic paraplegia and mycobacterial disease. *PLoS ONE*, **2013**, 8, e58286	3.7	27
65	Inherited disorders of IFN- IFN- and NF-B-mediated immunity **2013**, 454-464		1
64	Functional STAT3 deficiency compromises the generation of human T follicular helper cells. *Blood*, **2012**, 119, 3997-4008	2.2	230
63	Mycobacterial disease and impaired IFN- immunity in humans with inherited ISG15 deficiency. *Science*, **2012**, 337, 1684-8	33.3	348
62	Clinical immunology Disseminated Mycobacterium tuberculosis complex infection in a girl with partial dominant IFN- receptor 1 deficiency. *Central-European Journal of Immunology*, **2012**, 4, 378-381	1.6	3
61	Mendelian susceptibility to mycobacterial disease in egyptian children. *Mediterranean Journal of Hematology and Infectious Diseases*, **2012**, 4, e2012033	3.2	9
60	Granulomatous skin lesions, severe scrotal and lower limb edema due to mycobacterial infections in a child with complete IFN- receptor-1 deficiency. *Immunotherapy*, **2012**, 4, 1121-7	3.8	17
59	Hodgkin lymphoma in 2 children with chronic granulomatous disease. *Journal of Allergy and Clinical Immunology*, **2011**, 127, 543-544.e1-3	11.5	9
58	IRF8 mutations and human dendritic-cell immunodeficiency. *New England Journal of Medicine*, **2011**, 365, 127-38	59.2	469
57	New mechanism of X-linked anhidrotic ectodermal dysplasia with immunodeficiency: impairment of ubiquitin binding despite normal folding of NEMO protein. *Blood*, **2011**, 118, 926-35	2.2	45
56	Germline CYBB mutations that selectively affect macrophages in kindreds with X-linked predisposition to tuberculous mycobacterial disease. *Nature Immunology*, **2011**, 12, 213-21	19.1	202
55	Genetic lessons learned from X-linked Mendelian susceptibility to mycobacterial diseases. *Annals of the New York Academy of Sciences*, **2011**, 1246, 92-101	6.5	67
54	Lethal tuberculosis in a previously healthy adult with IL-12 receptor deficiency. *Journal of Clinical Immunology*, **2011**, 31, 537-9	5.7	44
53	Partial recessive IFN-R1 deficiency: genetic, immunological and clinical features of 14 patients from 11 kindreds. *Human Molecular Genetics*, **2011**, 20, 1509-23	5.6	75

52	Chronic mucocutaneous candidiasis in humans with inborn errors of interleukin-17 immunity. *Science*, **2011**, 332, 65-8	33.3	1309
51	Accounting for genetic heterogeneity in homozygosity mapping: application to Mendelian susceptibility to mycobacterial disease. *Journal of Medical Genetics*, **2011**, 48, 567-71	5.8	14
50	Gain-of-function human STAT1 mutations impair IL-17 immunity and underlie chronic mucocutaneous candidiasis. *Journal of Experimental Medicine*, **2011**, 208, 1635-48	16.6	599
49	IL-12Rβ1 deficiency in two of fifty children with severe tuberculosis from Iran, Morocco, and Turkey. *PLoS ONE*, **2011**, 6, e18524	3.7	91
48	A novel form of cell type-specific partial IFN-gammaR1 deficiency caused by a germ line mutation of the IFNGR1 initiation codon. *Human Molecular Genetics*, **2010**, 19, 434-44	5.6	34
47	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, 291-7	16.6	556
46	B cell-intrinsic signaling through IL-21 receptor and STAT3 is required for establishing long-lived antibody responses in humans. *Journal of Experimental Medicine*, **2010**, 207, 155-71	16.6	277
45	Clinical disease caused by Klebsiella in 2 unrelated patients with interleukin 12 receptor beta1 deficiency. *Pediatrics*, **2010**, 126, e971-6	7.4	25
44	Multiple cutaneous squamous cell carcinomas in a patient with interferon gamma receptor 2 (IFN gamma R2) deficiency. *Journal of Medical Genetics*, **2010**, 47, 631-4	5.8	27
43	Hematologically important mutations: the autosomal recessive forms of chronic granulomatous disease (second update). *Blood Cells, Molecules, and Diseases*, **2010**, 44, 291-9	2.1	127
42	Hematologically important mutations: X-linked chronic granulomatous disease (third update). *Blood Cells, Molecules, and Diseases*, **2010**, 45, 246-65	2.1	160
41	Revisiting human IL-12Rβ1 deficiency: a survey of 141 patients from 30 countries. *Medicine (United States)*, **2010**, 89, 381-402	1.8	277
40	A novel form of human STAT1 deficiency impairing early but not late responses to interferons. *Blood*, **2010**, 116, 5895-906	2.2	77
39	Primary immunodeficiencies of protective immunity to primary infections. *Clinical Immunology*, **2010**, 135, 204-9	9	57
38	Alu-repeat-induced deletions within the NCF2 gene causing p67-phox-deficient chronic granulomatous disease (CGD). *Human Mutation*, **2010**, 31, 151-8	4.7	16
37	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, 622-9	2.5	18
36	Human TRAF3 adaptor molecule deficiency leads to impaired Toll-like receptor 3 response and susceptibility to herpes simplex encephalitis. *Immunity*, **2010**, 33, 400-11	32.3	262
35	Comment on "Impaired priming and activation of the neutrophil NADPH oxidase in patients with IRAK4 or NEMO deficiency". *Journal of Immunology*, **2009**, 183, 3559	5.3	

34	A partial form of recessive STAT1 deficiency in humans. *Journal of Clinical Investigation*, **2009**, 119, 1502-14	15.9	140
33	Complementation of a pathogenic IFNGR2 misfolding mutation with modifiers of N-glycosylation. *Journal of Biotechnology*, **2008**, 136, S176	3.7	
32	From infectious diseases to primary immunodeficiencies. *Immunology and Allergy Clinics of North America*, **2008**, 28, 235-58, vii	3.3	23
31	Complementation of a pathogenic IFNGR2 misfolding mutation with modifiers of N-glycosylation. *Journal of Experimental Medicine*, **2008**, 205, 1729-37	16.6	52
30	Rhinoscleroma: a French national retrospective study of epidemiological and clinical features. *Clinical Infectious Diseases*, **2008**, 47, 1396-402	11.6	65
29	Essential role of nuclear factor-kappaB for NADPH oxidase activity in normal and anhidrotic ectodermal dysplasia leukocytes. *Blood*, **2008**, 112, 1453-60	2.2	22
28	Novel primary immunodeficiencies revealed by the investigation of paediatric infectious diseases. *Current Opinion in Immunology*, **2008**, 20, 39-48	7.8	113
27	Inborn errors of interferon (IFN)-mediated immunity in humans: insights into the respective roles of IFN-alpha/beta, IFN-gamma, and IFN-lambda in host defense. *Immunological Reviews*, **2008**, 226, 29-40	11.3	220
26	Complementation of a pathogenicIFNGR2misfolding mutation with modifiers of N-glycosylation. *Journal of Cell Biology*, **2008**, 182, i6-i6	7.3	
25	BCG-osis and tuberculosis in a child with chronic granulomatous disease. *Journal of Allergy and Clinical Immunology*, **2007**, 120, 32-8	11.5	105
24	IRAK4 and NEMO mutations in otherwise healthy children with recurrent invasive pneumococcal disease. *Journal of Medical Genetics*, **2007**, 44, 16-23	5.8	111
23	A novel X-linked recessive form of Mendelian susceptibility to mycobaterial disease. *Journal of Medical Genetics*, **2007**, 44, e65	5.8	44
22	Novel STAT1 alleles in otherwise healthy patients with mycobacterial disease. *PLoS Genetics*, **2006**, 2, e131	6	138
21	X-linked susceptibility to mycobacteria is caused by mutations in NEMO impairing CD40-dependent IL-12 production. *Journal of Experimental Medicine*, **2006**, 203, 1745-59	16.6	222
20	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	11	80
19	Inborn errors of IL-12/23- and IFN-gamma-mediated immunity: molecular, cellular, and clinical features. *Seminars in Immunology*, **2006**, 18, 347-61	10.7	366
18	From idiopathic infectious diseases to novel primary immunodeficiencies. *Journal of Allergy and Clinical Immunology*, **2005**, 116, 426-30	11.5	44
17	Gains of glycosylation comprise an unexpectedly large group of pathogenic mutations. *Nature Genetics*, **2005**, 37, 692-700	36.3	168

16	Inherited disorders of human Toll-like receptor signaling: immunological implications. *Immunological Reviews*, **2005**, 203, 10-20	11.3	115
15	Inherited disorders of the IL-12-IFN-gamma axis in patients with disseminated BCG infection. *European Journal of Pediatrics*, **2005**, 164, 753-7	4.1	53
14	Heritable defects of the human TLR signalling pathways. *Journal of Endotoxin Research*, **2005**, 11, 220-4		25
13	NEMO mutations in 2 unrelated boys with severe infections and conical teeth. *Pediatrics*, **2005**, 115, e615-9	7.4	57
12	Septicemia without sepsis: inherited disorders of nuclear factor-kappa B-mediated inflammation. *Clinical Infectious Diseases*, **2005**, 41 Suppl 7, S436-9	11.6	39
11	Bacillus Calmette Guerin triggers the IL-12/IFN-gamma axis by an IRAK-4- and NEMO-dependent, non-cognate interaction between monocytes, NK, and T lymphocytes. *European Journal of Immunology*, **2004**, 34, 3276-84	6.1	119
10	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, 22-9	9	195
9	A novel form of complete IL-12/IL-23 receptor beta1 deficiency with cell surface-expressed nonfunctional receptors. *Blood*, **2004**, 104, 2095-101	2.2	90
8	Low penetrance, broad resistance, and favorable outcome of interleukin 12 receptor beta1 deficiency: medical and immunological implications. *Journal of Experimental Medicine*, **2003**, 197, 527-35	16.6	256
7	Primary immunodeficiencies associated with pneumococcal disease. *Current Opinion in Allergy and Clinical Immunology*, **2003**, 3, 451-9	3.3	104
6	Pyogenic bacterial infections in humans with IRAK-4 deficiency. *Science*, **2003**, 299, 2076-9	33.3	737
5	Homozygous STAT2 gain-of-function mutation by loss of USP18 activity in a patient with type I interferonopathy		
4	Negative selection on human genes causing severe inborn errors depends on disease outcome and both the mode and mechanism of inheritance		1
3	Genome-wide association study of resistance to Mycobacterium tuberculosis infection identifies a locus at 10q26.2 in three distinct populations		1
2	Multi-batch cytometry data integration for optimal immunophenotyping		2
1	Human T-bet governs innate and innate-like adaptive IFN-γ immunity against mycobacteria		2