Luigi Daniele Notarangelo

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6383954/publications.pdf

Version: 2024-02-01

643 papers	51,561 citations	1530 106 h-index	2375 198 g-index
670 all docs	670 docs citations	670 times ranked	39393 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	Activation-Induced Cytidine Deaminase (AID) Deficiency Causes the Autosomal Recessive Form of the Hyper-IgM Syndrome (HIGM2). Cell, 2000, 102, 565-575.	13.5	1,489
4	Diagnostic Criteria for Primary Immunodeficiencies. Clinical Immunology, 1999, 93, 190-197.	1.4	964
5	Gene Therapy for Immunodeficiency Due to Adenosine Deaminase Deficiency. New England Journal of Medicine, 2009, 360, 447-458.	13.9	944
6	The X-linked lymphoproliferative-disease gene product SAP regulates signals induced through the co-receptor SLAM. Nature, 1998, 395, 462-469.	13.7	894
7	Mutations of Jak-3 gene in patients with autosomal severe combined immune deficiency (SCID). Nature, 1995, 377, 65-68.	13.7	864
8	Gene Therapy in Peripheral Blood Lymphocytes and Bone Marrow for ADA- Immunodeficient Patients. Science, 1995, 270, 470-475.	6.0	775
9	Immune dysregulation in human subjects with heterozygous germline mutations in CTLA4. Science, 2014, 345, 1623-1627.	6.0	745
10	Defective expression of T-cell CD40 ligand causes X-linked immunodeficiency with hyper-IgM. Nature, 1993, 361, 539-541.	13.7	703
11	Defects in TCIRG1 subunit of the vacuolar proton pump are responsible for a subset of human autosomal recessive osteopetrosis. Nature Genetics, 2000, 25, 343-346.	9.4	629
12	Clinical spectrum of X-linked hyper-IgM syndrome. Journal of Pediatrics, 1997, 131, 47-54.	0.9	604
13	Transplantation Outcomes for Severe Combined Immunodeficiency, 2000â€“2009. New England Journal of Medicine, 2014, 371, 434-446.	13.9	594
14	Newborn Screening for Severe Combined Immunodeficiency in 11 Screening Programs in the United States. JAMA - Journal of the American Medical Association, 2014, 312, 729.	3.8	586
15	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. Nucleic Acids Research, 2019, 47, D1018-D1027.	6.5	539
16	A three-dimensional model of human lung development and disease from pluripotent stem cells. Nature Cell Biology, 2017, 19, 542-549.	4.6	467
17	Primary immunodeficiency diseases: An update from the International Union of Immunological Societies Primary Immunodeficiency Diseases Classification Committee. Journal of Allergy and Clinical Immunology, 2007, 120, 776-794.	1.5	446
18	X-Linked Lymphoproliferative Disease. Journal of Experimental Medicine, 2000, 192, 337-346.	4.2	438

#	Article	IF	Citations
19	Human intracellular ISG15 prevents interferon-Î±/Î² over-amplification and auto-inflammation. Nature, 2015, 517, 89-93.	13.7	432
20	Partial V(D)J Recombination Activity Leads to Omenn Syndrome. Cell, 1998, 93, 885-896.	13.5	429
21	Primary immunodeficiencies: 2009 update. Journal of Allergy and Clinical Immunology, 2009, 124, 1161-1178.	1.5	416
22	Immunodeficiency, autoinflammation and amylopectinosis in humans with inherited HOIL-1 and LUBAC deficiency. Nature Immunology, 2012, 13, 1178-1186.	7.0	410
23	Life-threatening influenza and impaired interferon amplification in human IRF7 deficiency. Science, 2015, 348, 448-453.	6.0	389
24	Transplantation of hematopoietic stem cells and long-term survival for primary immunodeficiencies in Europe: Entering a new century, do we do better?. Journal of Allergy and Clinical Immunology, 2010, 126, 602-610.e11.	1.5	385
25	Impairment of immunity to Candida and Mycobacterium in humans with bi-allelic RORC mutations. Science, 2015, 349, 606-613.	6.0	366
26	Primary immunodeficiencies. Journal of Allergy and Clinical Immunology, 2010, 125, S182-S194.	1.5	358
27	A Modified Î³-Retrovirus Vector for X-Linked Severe Combined Immunodeficiency. New England Journal of Medicine, 2014, 371, 1407-1417.	13.9	358
28	Autoantibodies neutralizing type I IFNs are present in ~4% of uninfected individuals over 70 years old and account for ~20% of COVID-19 deaths. Science Immunology, 2021, 6, .	5.6	357
29	Mutations of CD40 gene cause an autosomal recessive form of immunodeficiency with hyper IgM. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 12614-12619.	3.3	347
30	V(D)J recombination defects in lymphocytes due to RAG mutations: severe immunodeficiency with a spectrum of clinical presentations. Blood, 2001, 97, 81-88.	0.6	324
31	Mutations of the Wiskott-Aldrich Syndrome Protein (WASP): hotspots, effect on transcription, and translation and phenotype/genotype correlation. Blood, 2004, 104, 4010-4019.	0.6	308
32	Establishing diagnostic criteria for severe combined immunodeficiency disease (SCID), leaky SCID, and Omenn syndrome: The Primary Immune Deficiency Treatment Consortium experience. Journal of Allergy and Clinical Immunology, 2014, 133, 1092-1098.	1.5	301
33	Clinical, Immunological, and Molecular Analysis in a Large Cohort of Patients with X-Linked Agammaglobulinemia: An Italian Multicenter Study. Clinical Immunology, 2002, 104, 221-230.	1.4	299
34	Long-term outcome and lineage-specific chimerism in 194 patients with Wiskott-Aldrich syndrome treated by hematopoietic cell transplantation in the period 1980-2009: an international collaborative study. Blood, 2011, 118, 1675-1684.	0.6	296
35	Review Guidelines Subscribe to Alerts Search Article Type Publication Date ..... Go Author Info Why Submit? Fees Article Types Author Guidelines Submission Checklist Contact Editorial Office Submit Manuscript Review ARTICLE Abstract Full Text PDF 0 Write a Comment Primary immunodeficiency diseases: an update on the classification from the International Union of Immunological Societies	2.2	294
36	Clinical features, long-term follow-up and outcome of a large cohort of patients with Chronic Granulomatous Disease: An Italian multicenter study. Clinical Immunology, 2008, 126, 155-164.	1.4	293

#	Article	IF	Citations
37	Inborn Errors of Human JAKs and STATs. Immunity, 2012, 36, 515-528.	6.6	290
38	Impaired intrinsic immunity to HSV-1 in human iPSC-derived TLR3-deficient CNS cells. Nature, 2012, 491, 769-773.	13.7	288
39	Gntb-A, a Novel Sh2d1a-Associated Surface Molecule Contributing to the Inability of Natural Killer Cells to Kill Epstein-Barr Virusâ€“Infected B Cells in X-Linked Lymphoproliferative Disease. Journal of Experimental Medicine, 2001, 194, 235-246.	4.2	287
40	Xâ€“linked thrombocytopenia and Wiskottâ€“Aldrich syndrome are allelic diseases with mutations in the WASP gene. Nature Genetics, 1995, 9, 414-417.	9.4	274
41	Human Osteoclast-Poor Osteopetrosis with Hypogammaglobulinemia due to TNFRSF11A (RANK) Mutations. American Journal of Human Genetics, 2008, 83, 64-76.	2.6	270
42	An immune-based biomarker signature is associated with mortality in COVID-19 patients. JCI Insight, 2021, 6, .	2.3	269
43	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
44	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
45	Human HOIP and LUBAC deficiency underlies autoinflammation, immunodeficiency, amylopectinosis, and lymphangiectasia. Journal of Experimental Medicine, 2015, 212, 939-951.	4.2	241
46	Lentiviral hematopoietic stem cell gene therapy for X-linked severe combined immunodeficiency. Science Translational Medicine, 2016, 8, 335ra57.	5.8	229
47	A missense mutation in TFRC, encoding transferrin receptor 1, causes combined immunodeficiency. Nature Genetics, 2016, 48, 74-78.	9.4	219
48	Guidelines for genetic studies in single patients: lessons from primary immunodeficiencies. Journal of Experimental Medicine, 2014, 211, 2137-2149.	4.2	218
49	Loss-of-function mutations in the C9ORF72 mouse ortholog cause fatal autoimmune disease. Science Translational Medicine, 2016, 8, 347ra93.	5.8	217
50	Bone Marrow Transplantation for Severe Combined Immune Deficiency. JAMA - Journal of the American Medical Association, 2006, 295, 508.	3.8	216
51	Long-term outcome following hematopoietic stem-cell transplantation in Wiskott-Aldrich syndrome: collaborative study of the European Society for Immunodeficiencies and European Group for Blood and Marrow Transplantation. Blood, 2008, 111, 439-445.	0.6	216
52	Human genetic and immunological determinants of critical COVID-19 pneumonia. Nature, 2022, 603, 587-598.	13.7	216
53	Omenn syndrome: Inflammation in leaky severe combined immunodeficiency. Journal of Allergy and Clinical Immunology, 2008, 122, 1082-1086.	1.5	213
54	Immune reconstitution and survival of 100 SCID patients postâ€“hematopoietic cell transplant: a PIDTC natural history study. Blood, 2017, 130, 2718-2727.	0.6	212

#	Article	IF	Citations
55	How I treat ADA deficiency. Blood, 2009, 114, 3524-3532.	0.6	206
56	Immune reconstitution in ADA-SCID after PBL gene therapy and discontinuation of enzyme replacement. Nature Medicine, 2002, 8, 423-425.	15.2	205
57	DOCK8 functions as an adaptor that links TLR-MyD88 signaling to B cell activation. Nature Immunology, 2012, 13, 612-620.	7.0	205
58	Chloride Channel ClCN7 Mutations Are Responsible for Severe Recessive, Dominant, and Intermediate Osteopetrosis. Journal of Bone and Mineral Research, 2003, 18, 1740-1747.	3.1	202
59	The mutational spectrum of human malignant autosomal recessive osteopetrosis. Human Molecular Genetics, 2001, 10, 1767-1773.	1.4	201
60	Human RAG mutations: biochemistry and clinical implications. Nature Reviews Immunology, 2016, 16, 234-246.	10.6	200
61	Wiskott-Aldrich syndrome. Current Opinion in Hematology, 2008, 15, 30-36.	1.2	186
62	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. Cell, 2020, 181, 1194-1199.	13.5	185
63	Preexisting autoantibodies to type I IFNs underlie critical COVID-19 pneumonia in patients with APS-1. Journal of Experimental Medicine, 2021, 218, .	4.2	185
64	Missense Mutations in the Fas Gene Resulting in Autoimmune Lymphoproliferative Syndrome: A Molecular and Immunological Analysis. Blood, 1997, 89, 902-909.	0.6	178
65	Primary immunodeficiency diseases: An update. Journal of Allergy and Clinical Immunology, 2004, 114, 677-687.	1.5	177
66	Severe combined immunodeficiencies and related disorders. Nature Reviews Disease Primers, 2015, 1, 15061.	18.1	173
67	Altered leukocyte response to CXCL12 in patients with warts hypogammaglobulinemia, infections, myelokathexis (WHIM) syndrome. Blood, 2004, 104, 444-452.	0.6	172
68	Inherited DOCK2 Deficiency in Patients with Early-Onset Invasive Infections. New England Journal of Medicine, 2015, 372, 2409-2422.	13.9	169
69	WASP regulates suppressor activity of human and murine CD4+CD25+FOXP3+ natural regulatory T cells. Journal of Experimental Medicine, 2007, 204, 369-380.	4.2	167
70	Time-resolved systems immunology reveals a late juncture linked to fatal COVID-19. Cell, 2021, 184, 1836-1857.e22.	13.5	167
71	Interleukin-7 receptor alpha (IL-7Ralpha) deficiency: cellular and molecular bases. Analysis of clinical, immunological, and molecular features in 16 novel patients. Immunological Reviews, 2005, 203, 110-126.	2.8	162
72	Broad-spectrum antibodies against self-antigens and cytokines in RAG deficiency. Journal of Clinical Investigation, 2015, 125, 4135-4148.	3.9	159

#	Article	IF	Citations
73	A novel primary human immunodeficiency due to deficiency in the WASP-interacting protein WIP. Journal of Experimental Medicine, 2012, 209, 29-34.	4.2	158
74	Outcome of hematopoietic stem cell transplantation for adenosine deaminaseâ€“deficient severe combined immunodeficiency. Blood, 2012, 120, 3615-3624.	0.6	151
75	Timely and spatially regulated maturation of B and T cell repertoire during human fetal development. Science Translational Medicine, 2015, 7, 276ra25.	5.8	148
76	Ruxolitinib reverses dysregulated T helper cell responses and controls autoimmunity caused by a novel signal transducer and activator of transcription 1 (STAT1) gain-of-function mutation. Journal of Allergy and Clinical Immunology, 2017, 139, 1629-1640.e2.	1.5	147
77	Life-threatening influenza pneumonitis in a child with inherited IRF9 deficiency. Journal of Experimental Medicine, 2018, 215, 2567-2585.	4.2	146
78	AIRE deficiency in thymus of 2 patients with Omenn syndrome. Journal of Clinical Investigation, 2005, 115, 728-732.	3.9	146
79	Hematopoietic Stem Cell Transplantation in Primary Immunodeficiency Diseases: Current Status and Future Perspectives. Frontiers in Pediatrics, 2019, 7, 295.	0.9	144
80	Immunopathological signatures in multisystem inflammatory syndrome in children and pediatric COVID-19. Nature Medicine, 2022, 28, 1050-1062.	15.2	144
81	Whole-exome sequencing identifies tetratricopeptide repeat domain 7A (TTC7A) mutations for combined immunodeficiency with intestinal atresias. Journal of Allergy and Clinical Immunology, 2013, 132, 656-664.e17.	1.5	140
82	Structural and Functional Basis for JAK3-Deficient Severe Combined Immunodeficiency. Blood, 1997, 90, 3996-4003.	0.6	138
83	Human inborn errors of immunity: An expanding universe. Science Immunology, 2020, 5, .	5.6	138
84	Innate immunity defects in Hermansky-Pudlak type 2 syndrome. Blood, 2006, 107, 4857-4864.	0.6	136
85	Early defects in human T-cell development severely affect distribution and maturation of thymic stromal cells: possible implications for the pathophysiology of Omenn syndrome. Blood, 2009, 114, 105-108.	0.6	135
86	Global study of primary immunodeficiency diseases (PI)â€”diagnosis, treatment, and economic impact: an updated report from the Jeffrey Modell Foundation. Immunologic Research, 2011, 51, 61-70.	1.3	135
87	Severe influenza pneumonitis in children with inherited TLR3 deficiency. Journal of Experimental Medicine, 2019, 216, 2038-2056.	4.2	134
88	IL-21 is the primary common Î³ chain-binding cytokine required for human B-cell differentiation in vivo. Blood, 2011, 118, 6824-6835.	0.6	132
89	A systematic analysis of recombination activity andÂgenotype-phenotype correlation in human recombination-activating gene 1 deficiency. Journal of Allergy and Clinical Immunology, 2014, 133, 1099-1108.e12.	1.5	132
90	Toll Receptor-Mediated Regulation of NADPH Oxidase in Human Dendritic Cells. Journal of Immunology, 2004, 173, 5749-5756.	0.4	131

#	Article	IF	Citations
91	Impaired natural and CD16-mediated NK cell cytotoxicity in patients with WAS and XLT: ability of IL-2 to correct NK cell functional defect. Blood, 2004, 104, 436-443.	0.6	130
92	Interleukin-12 and Interleukin-23 Blockade in Leukocyte Adhesion Deficiency Type 1. New England Journal of Medicine, 2017, 376, 1141-1146.	13.9	130
93	Auto-antibodies to type I IFNs can underlie adverse reactions to yellow fever live attenuated vaccine. Journal of Experimental Medicine, 2021, 218, .	4.2	130
94	Recommendations for live viral and bacterial vaccines inÂimmunodeficient patients and their close contacts. Journal of Allergy and Clinical Immunology, 2014, 133, 961-966.	1.5	128
95	SCID genotype and 6-month posttransplant CD4 count predict survival and immune recovery. Blood, 2018, 132, 1737-1749.	0.6	128
96	C4b-Binding Protein (C4BP) Activates B Cells through the CD40 Receptor. Immunity, 2003, 18, 837-848.	6.6	126
97	Jak3, severe combined immunodeficiency, and a new class of immunosuppressive drugs. Immunological Reviews, 2005, 203, 127-142.	2.8	126
98	Complex Effects of Naturally Occurring Mutations in the JAK3 Pseudokinase Domain: Evidence for Interactions between the Kinase and Pseudokinase Domains. Molecular and Cellular Biology, 2000, 20, 947-956.	1.1	125
99	Primary immune deficiencies with aberrant IgE production. Journal of Allergy and Clinical Immunology, 2008, 122, 1054-1062.	1.5	124
100	Studies of the expression of the Wiskott-Aldrich syndrome protein.. Journal of Clinical Investigation, 1996, 97, 2627-2634.	3.9	124
101	Defective Expression of CD40 Ligand on T Cells Causes "X-Linked Immunodeficiency with Hyper-IgM (HIGM1)". Immunological Reviews, 1994, 138, 39-59.	2.8	122
102	Signaling via IL-2 and IL-4 in JAK3-Deficient Severe Combined Immunodeficiency Lymphocytes: JAK3-Dependent and Independent Pathways. Immunity, 1996, 5, 605-615.	6.6	120
103	Activation-induced cytidine deaminase (AID) is required for B-cell tolerance in humans. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 11554-11559.	3.3	118
104	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
105	Treatment of CD40 ligand deficiency by hematopoietic stem cell transplantation: a survey of the European experience, 1993-2002. Blood, 2003, 103, 1152-1157.	0.6	116
106	Improving cellular therapy for primary immune deficiency diseases: Recognition, diagnosis, and management. Journal of Allergy and Clinical Immunology, 2009, 124, 1152-1160.e12.	1.5	110
107	Hypomorphic Rag mutations can cause destructive midline granulomatous disease. Blood, 2010, 116, 1263-1271.	0.6	110
108	Life-Threatening COVID-19: Defective Interferons Unleash Excessive Inflammation. Med, 2020, 1, 14-20.	2.2	110

#	Article	IF	Citations
109	The risk of COVID-19 death is much greater and age dependent with type I IFN autoantibodies. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, e2200413119.	3.3	110
110	CD30 cell expression and abnormal soluble CD30 serum accumulation in Omenn's syndrome: Evidence for a T helper 2-mediated condition. European Journal of Immunology, 1996, 26, 329-334.	1.6	108
111	Expression of Inducible Nitric Oxide Synthase in Human Granulomas and Histiocytic Reactions. American Journal of Pathology, 1999, 154, 145-152.	1.9	108
112	Defects of class-switch recombination. Journal of Allergy and Clinical Immunology, 2006, 117, 855-864.	1.5	107
113	Long-term outcomes of 176 patients with X-linked hyper-IgM syndrome treated with or without hematopoietic cell transplantation. Journal of Allergy and Clinical Immunology, 2017, 139, 1282-1292.	1.5	107
114	Consensus approach for the management of severe combined immune deficiency caused by adenosine deaminase deficiency. Journal of Allergy and Clinical Immunology, 2019, 143, 852-863.	1.5	104
115	A novel 4-kb interleukin-13 receptor Î± mRNA expressed in human B, T, and endothelial cells encoding an alternate type-II interleukin-4/ interleukin-13 receptor. European Journal of Immunology, 1997, 27, 971-978.	1.6	103
116	Defective Th1 Cytokine Gene Transcription in CD4+ and CD8+ T Cells from Wiskott-Aldrich Syndrome Patients. Journal of Immunology, 2006, 177, 7451-7461.	0.4	103
117	A Phenotypic Approach for IUIS PID Classification and Diagnosis: Guidelines for Clinicians at the Bedside. Journal of Clinical Immunology, 2013, 33, 1078-1087.	2.0	103
118	Omenn syndrome in an infant with IL7RA gene mutation. Journal of Pediatrics, 2006, 148, 272-274.	0.9	102
119	Human Peripheral Lymphoid Tissues Contain Autoimmune Regulator-Expressing Dendritic Cells. American Journal of Pathology, 2010, 176, 1104-1112.	1.9	101
120	Mutations in severe combined immune deficiency (SCID) due to JAK3 deficiency. Human Mutation, 2001, 18, 255-263.	1.1	100
121	The Natural History of Children with Severe Combined Immunodeficiency: Baseline Features of the First Fifty Patients of the Primary Immune Deficiency Treatment Consortium Prospective Study 6901. Journal of Clinical Immunology, 2013, 33, 1156-1164.	2.0	100
122	SARS-CoV-2â€“related MIS-C: A key to the viral and genetic causes of Kawasaki disease?. Journal of Experimental Medicine, 2021, 218, .	4.2	100
123	Ineffective expression of CD40 ligand on cord blood T cells may contribute to poor immunoglobulin production in the newborn. European Journal of Immunology, 1994, 24, 1919-1924.	1.6	99
124	WASP confers selective advantage for specific hematopoietic cell populations and serves a unique role in marginal zone B-cell homeostasis and function. Blood, 2008, 112, 4139-4147.	0.6	99
125	B cellâ€“intrinsic deficiency of the Wiskott-Aldrich syndrome protein (WASp) causes severe abnormalities of the peripheral B-cell compartment in mice. Blood, 2012, 119, 2819-2828.	0.6	99
126	T-cell defects in patients with ARPC1B germline mutations account for combined immunodeficiency. Blood, 2018, 132, 2362-2374.	0.6	99

#	Article	IF	Citations
127	Mutational Analysis of Human BAFF Receptor TNFRSF13C (BAFF-R) in Patients with Common Variable Immunodeficiency. Journal of Clinical Immunology, 2005, 25, 496-502.	2.0	98
128	Of genes and phenotypes: the immunological and molecular spectrum of combined immune deficiency.Defects of the gc-JAK3 signaling pathway as a model. Immunological Reviews, 2000, 178, 39-48.	2.8	97
129	A hypomorphic R229Q Rag2 mouse mutant recapitulates human Omenn syndrome. Journal of Clinical Investigation, 2007, 117, 1260-1269.	3.9	97
130	Molecular Cloning of ILP-2 , a Novel Member of the Inhibitor of Apoptosis Protein Family. Molecular and Cellular Biology, 2001, 21, 4292-4301.	1.1	95
131	Long-term immune reconstitution and clinical outcome after stem cell transplantation for severe T-cell immunodeficiency. Journal of Allergy and Clinical Immunology, 2007, 120, 892-899.	1.5	95
132	Immature B cells preferentially switch to IgE with increased direct SÎ¼ to SÎµ recombination. Journal of Experimental Medicine, 2011, 208, 2733-2746.	4.2	95
133	Immunological and genetic bases of new primary immunodeficiencies. Nature Reviews Immunology, 2007, 7, 851-861.	10.6	94
134	Missense mutations of the WASP gene cause intermittent X-linked thrombocytopenia. Blood, 2002, 99, 2268-2269.	0.6	93
135	Clinical spectrum, pathophysiology and treatment of the Wiskottâ€“Aldrich syndrome. Current Opinion in Hematology, 2011, 18, 42-48.	1.2	93
136	Inborn Errors of RNA Lariat Metabolism in Humans with Brainstem Viral Infection. Cell, 2018, 172, 952-965.e18.	13.5	92
137	A hypomorphic recombination-activating gene 1 (RAG1) mutation resulting in a phenotype resembling common variable immunodeficiency. Journal of Allergy and Clinical Immunology, 2014, 134, 1375-1380.	1.5	91
138	Expansion of immunoglobulin-secreting cells and defects in B cell tolerance in Rag-dependent immunodeficiency. Journal of Experimental Medicine, 2010, 207, 1541-1554.	4.2	90
139	RAG Deficiency: Two Genes, Many Diseases. Journal of Clinical Immunology, 2018, 38, 646-655.	2.0	89
140	CD40Lbase: a database of CD40L gene mutations causing X-linked hyper-IgM syndrome. Trends in Immunology, 1996, 17, 511-516.	7.5	88
141	X-chromosome inactivation analysis in a female carrier of FOXP3 mutation. Clinical and Experimental Immunology, 2002, 130, 127-130.	1.1	88
142	Characterization of T and B cell repertoire diversity in patients with RAG deficiency. Science Immunology, 2016, 1, .	5.6	88
143	Immunology of Down syndrome: A review. American Journal of Medical Genetics Part A, 2005, 37, 204-212.	2.4	87
144	A combined immunodeficiency with severe infections, inflammation, and allergy caused by ARPC1B deficiency. Journal of Allergy and Clinical Immunology, 2019, 143, 2296-2299.	1.5	87

#	Article	IF	Citations
145	Excellent outcomes following hematopoietic cell transplantation for Wiskott-Aldrich syndrome: a PIDTC report. Blood, 2020, 135, 2094-2105.	0.6	87
146	Structural basis for chromosome X-linked agammaglobulinemia: a tyrosine kinase disease.. Proceedings of the National Academy of Sciences of the United States of America, 1994, 91, 12803-12807.	3.3	85
147	X-linked immunodeficiency with hyper-IgM (XHIM). Clinical and Experimental Immunology, 2000, 120, 399-405.	1.1	85
148	Inborn Errors of Immunity With Immune Dysregulation: From Bench to Bedside. Frontiers in Pediatrics, 2019, 7, 353.	0.9	85
149	Management options for adenosine deaminase deficiency; proceedings of the EBMT satellite workshop (Hamburg, March 2006). Clinical Immunology, 2007, 123, 139-147.	1.4	84
150	Severe impairment of IFN-Î³ and IFN-Î± responses in cells of a patient with a novel STAT1 splicing mutation. Blood, 2011, 118, 1806-1817.	0.6	84
151	PRKDC mutations associated with immunodeficiency, granuloma, and autoimmune regulatorâ€“dependent autoimmunity. Journal of Allergy and Clinical Immunology, 2015, 135, 1578-1588.e5.	1.5	84
152	Aberrant type 1 immunity drives susceptibility to mucosal fungal infections. Science, 2021, 371, .	6.0	84
153	A singleâ€center experience in 20 patients with infantile malignant osteopetrosis. American Journal of Hematology, 2009, 84, 473-479.	2.0	83
154	Defect of regulatory T cells in patients with Omenn syndrome. Journal of Allergy and Clinical Immunology, 2010, 125, 209-216.	1.5	83
155	Small RNAs derived from lncRNA RNase MRP have gene-silencing activity relevant to human cartilageâ€“hair hypoplasia. Human Molecular Genetics, 2014, 23, 368-382.	1.4	83
156	Comparison of outcomes of hematopoietic stem cell transplantation without chemotherapy conditioning by using matched sibling and unrelated donors for treatment ofÂsevere combined immunodeficiency. Journal of Allergy and Clinical Immunology, 2014, 134, 935-943.e15.	1.5	82
157	PTX3 genetic variations affect the risk of Pseudomonas aeruginosa airway colonization in cystic fibrosis patients. Genes and Immunity, 2010, 11, 665-670.	2.2	81
158	Global overview of primary immunodeficiencies: a report from Jeffrey Modell Centers worldwide focused on diagnosis, treatment, and discovery. Immunologic Research, 2014, 60, 132-144.	1.3	81
159	A single amino acid change, A91V, leads to conformational changes that can impair processing to the active form of perforin. Blood, 2005, 106, 932-937.	0.6	80
160	G-CSF treatment of severe congenital neutropenia reverses neutropenia but does not correct the underlying functional deficiency of the neutrophil in defending against microorganisms. Blood, 2007, 109, 4716-4723.	0.6	80
161	Primary immunodeficiencies: AÂrapidly evolving story. Journal of Allergy and Clinical Immunology, 2013, 131, 314-323.	1.5	80
162	Intrathymic Restriction and Peripheral Expansion of the T-Cell Repertoire in Omenn Syndrome. Blood, 1999, 94, 3468-3478.	0.6	79

#	Article	IF	Citations
163	WASPbase: a database of WAS- and XLT-causing mutations. Trends in Immunology, 1996, 17, 496-502.	7.5	78
164	Wiskott-Aldrich syndrome protein (WASP) and N-WASP are critical for peripheral B-cell development and function. Blood, 2012, 119, 3966-3974.	0.6	78
165	Disseminated cryptosporidium infection in an infant with hyper-IgM syndrome caused by CD40 deficiency. Journal of Pediatrics, 2003, 142, 194-196.	0.9	77
166	Hyperactivated PI3KÎ´ promotes self and commensal reactivity at the expense of optimal humoral immunity. Nature Immunology, 2018, 19, 986-1000.	7.0	77
167	Hyper IgM syndromes. Current Opinion in Rheumatology, 2003, 15, 422-429.	2.0	76
168	EXTL3 mutations cause skeletal dysplasia, immune deficiency, and developmental delay. Journal of Experimental Medicine, 2017, 214, 623-637.	4.2	76
169	Human SNORA31 variations impair cortical neuron-intrinsic immunity to HSV-1 and underlie herpes simplex encephalitis. Nature Medicine, 2019, 25, 1873-1884.	15.2	76
170	Activity of Classical and Alternative Pathways of Complement in Preterm and Small for Gestational Age Infants. Pediatric Research, 1984, 18, 281-285.	1.1	75
171	Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy Syndrome: Time to Review Diagnostic Criteria?. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 3146-3148.	1.8	75
172	Stem cell transplantation for the Wiskottâ€“Aldrich syndrome: a single-center experience confirms efficacy of matched unrelated donor transplantation. Bone Marrow Transplantation, 2006, 38, 671-679.	1.3	74
173	Omenn syndrome: a disorder of Rag1 and Rag2 genes. Journal of Clinical Immunology, 1999, 19, 87-97.	2.0	73
174	In vitro correction of JAK3-deficient severe combined immunodeficiency by retroviral-mediated gene transduction.. Journal of Experimental Medicine, 1996, 183, 2687-2692.	4.2	71
175	Novel Munc13-4 mutations in children and young adult patients with haemophagocytic lymphohistiocytosis. Journal of Medical Genetics, 2006, 43, 953-960.	1.5	71
176	A prospective study on the natural history of patients with profound combined immunodeficiency: An interim analysis. Journal of Allergy and Clinical Immunology, 2017, 139, 1302-1310.e4.	1.5	71
177	4 Primary immunodeficiency mutation databases. Advances in Genetics, 2001, 43, 103-188.	0.8	70
178	Progressive Neurodegeneration in Patients with Primary Immunodeficiency Disease on IVIG Treatment. Clinical Immunology, 2002, 102, 19-24.	1.4	70
179	Allogeneic hematopoietic cell transplantation for primary immune deficiency diseases: Current status and critical needs. Journal of Allergy and Clinical Immunology, 2008, 122, 1087-1096.	1.5	70
180	Identification of Patients with RAG Mutations Previously Diagnosed with Common Variable Immunodeficiency Disorders. Journal of Clinical Immunology, 2015, 35, 119-124.	2.0	70

#	Article	IF	Citations
181	Six novel mutations in the PRF1 gene in children with haemophagocytic lymphohistiocytosis. Journal of Medical Genetics, 2001, 38, 643-646.	1.5	69
182	Activation-Induced Cytidine Deaminase Expression in Human B Cell Precursors Is Essential for Central B Cell Tolerance. Immunity, 2015, 43, 884-895.	6.6	69
183	Neutralizing typeâ€I interferon autoantibodies are associated with delayed viral clearance and intensive care unit admission in patients with COVIDâ€19. Immunology and Cell Biology, 2021, 99, 917-921.	1.0	69
184	AIRE deficiency in thymus of 2 patients with Omenn syndrome. Journal of Clinical Investigation, 2005, 115, 728-732.	3.9	69
185	Organization of the human CD40L gene: implications for molecular defects in X chromosome-linked hyper-IgM syndrome and prenatal diagnosis.. Proceedings of the National Academy of Sciences of the United States of America, 1994, 91, 2110-2114.	3.3	68
186	Altered BCR and TLR signals promote enhanced positive selection of autoreactive transitional B cells in Wiskott-Aldrich syndrome. Journal of Experimental Medicine, 2015, 212, 1663-1677.	4.2	67
187	Antibody responses to the SARS-CoV-2 vaccine in individuals with various inborn errors of immunity. Journal of Allergy and Clinical Immunology, 2021, 148, 1192-1197.	1.5	67
188	A new case of IPEX receiving bone marrow transplantation. Bone Marrow Transplantation, 2005, 35, 1033-1034.	1.3	66
189	Primary Immune Deficiency Treatment Consortium (PIDTC) report. Journal of Allergy and Clinical Immunology, 2014, 133, 335-347.e11.	1.5	65
190	Artificial thymic organoids represent a reliable tool to study T-cell differentiation in patients with severe T-cell lymphopenia. Blood Advances, 2020, 4, 2611-2616.	2.5	65
191	Inherited PD-1 deficiency underlies tuberculosis and autoimmunity in a child. Nature Medicine, 2021, 27, 1646-1654.	15.2	65
192	Immunodeficiencies with Autoimmune Consequences. Advances in Immunology, 2006, 89, 321-370.	1.1	64
193	Outcomes and Treatment Strategies for Autoimmunity and Hyperinflammation in Patients with RAG Deficiency. Journal of Allergy and Clinical Immunology: in Practice, 2019, 7, 1970-1985.e4.	2.0	64
194	Thymic Epithelium Abnormalities in DiGeorge and Down Syndrome Patients Contribute to Dysregulation in T Cell Development. Frontiers in Immunology, 2019, 10, 447.	2.2	64
195	The genetic landscape of severe combined immunodeficiency in the United States and Canada in the current era (2010-2018). Journal of Allergy and Clinical Immunology, 2019, 143, 405-407.	1.5	64
196	TLR3 controls constitutive IFN-Î² antiviral immunity in human fibroblasts and cortical neurons. Journal of Clinical Investigation, 2021, 131, .	3.9	64
197	Close linkage of probe p212 (DXS178) to X-linked agammaglobulinemia. Human Genetics, 1989, 84, 19-21.	1.8	63
198	Unexpected and variable phenotypes in a family with JAK3 deficiency. Genes and Immunity, 2001, 2, 422-432.	2.2	63

#	Article	IF	Citations
199	Functional analysis of naturally occurring DCLRE1C mutations and correlation with the clinical phenotype of ARTEMIS deficiency. Journal of Allergy and Clinical Immunology, 2015, 136, 140-150.e7.	1.5	63
200	Human interleukin-2 receptor Î² mutations associated with defects in immunity and peripheral tolerance. Journal of Experimental Medicine, 2019, 216, 1311-1327.	4.2	62
201	Reversible severe combined immunodeficiency phenotype secondary to a mutation of the proton-coupled folate transporter. Clinical Immunology, 2009, 133, 287-294.	1.4	61
202	Guidelines for implementation of populationâ€based newborn screening for severe combined immunodeficiency. Journal of Inherited Metabolic Disease, 2010, 33, 273-281.	1.7	60
203	Clinical, immunologic and genetic profiles of DOCK8-deficient patients in Kuwait. Clinical Immunology, 2012, 143, 266-272.	1.4	60
204	A common single nucleotide polymorphism impairs B-cell activating factor receptor's multimerization, contributing to common variable immunodeficiency. Journal of Allergy and Clinical Immunology, 2014, 133, 1222-1225.e10.	1.5	60
205	Wiskottâ€”Aldrich Syndrome: a model for defective actin reorganization, cell trafficking and synapse formation. Current Opinion in Immunology, 2003, 15, 585-591.	2.4	59
206	Reduced thymic output, increased spontaneous apoptosis and oligoclonal B cells in polyethylene glycol-adenosine deaminase-treated patients. European Journal of Immunology, 2005, 35, 3376-3386.	1.6	59
207	Expanding the spectrum of recombination-activating gene 1 deficiency: AÂfamily with early-onset autoimmunity. Journal of Allergy and Clinical Immunology, 2013, 132, 969-971.e2.	1.5	59
208	SNX10 mutations define a subgroup of human autosomal recessive osteopetrosis with variable clinical severity. Journal of Bone and Mineral Research, 2013, 28, 1041-1049.	3.1	59
209	Recessive inborn errors of type I IFN immunity in children with COVID-19 pneumonia. Journal of Experimental Medicine, 2022, 219, .	4.2	59
210	RAG and RAG defects. Current Opinion in Immunology, 1999, 11, 435-442.	2.4	58
211	Mutations in Conserved Regions of the Predicted RAG2 Kelch Repeats Block Initiation of V(D)J Recombination and Result in Primary Immunodeficiencies. Molecular and Cellular Biology, 2000, 20, 5653-5664.	1.1	58
212	Variability of clinical and laboratory features among patients with ribonuclease mitochondrial RNA processing endoribonuclease gene mutations. Journal of Allergy and Clinical Immunology, 2008, 122, 1178-1184.	1.5	58
213	Human iPSC-derived trigeminal neurons lack constitutive TLR3-dependent immunity that protects cortical neurons from HSV-1 infection. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E8775-E8782.	3.3	58
214	Wiskottâ€“Aldrich syndrome protein (WASP) is a tumor suppressor in T cell lymphoma. Nature Medicine, 2019, 25, 130-140.	15.2	57
215	Hematopoietic Cell Transplantation in Patients With Primary Immune Regulatory Disorders (PIRD): A Primary Immune Deficiency Treatment Consortium (PIDTC) Survey. Frontiers in Immunology, 2020, 11, 239.	2.2	57
216	X-linked primary immunodeficiencies as a bridge to better understandingX-chromosome related autoimmunity. Journal of Autoimmunity, 2009, 33, 17-24.	3.0	56

#	Article	IF	Citations
217	Diagnosis of immunodeficiency caused by a purine nucleoside phosphorylase defect by using tandem mass spectrometry on dried blood spots. Journal of Allergy and Clinical Immunology, 2014, 134, 155-159.e3.	1.5	56
218	Deficiency of base excision repair enzyme NEIL3 drives increased predisposition to autoimmunity. Journal of Clinical Investigation, 2016, 126, 4219-4236.	3.9	56
219	PAX1 is essential for development and function of the human thymus. Science Immunology, 2020, 5, .	5.6	55
220	Transfer of the ADA Gene into Bone Marrow Cells and Peripheral Blood Lymphocytes for the Treatment of Patients Affected by ADA-Deficient SCID. Human Gene Therapy, 1993, 4, 513-520.	1.4	54
221	Functional T Cell Immunodeficiencies (with T Cells Present). Annual Review of Immunology, 2013, 31, 195-225.	9.5	54
222	Autoimmunity due to RAG deficiency and estimated disease incidence in RAG1/2 mutations. Journal of Allergy and Clinical Immunology, 2014, 133, 880-882.e10.	1.5	54
223	Targeted strategies directed at the molecular defect: Toward precision medicine for select primary immunodeficiency disorders. Journal of Allergy and Clinical Immunology, 2017, 139, 715-723.	1.5	54
224	Architecture of the human PI4KIIIÎ± lipid kinase complex. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 13720-13725.	3.3	54
225	Second Case of HOIP Deficiency Expands Clinical Features and Defines Inflammatory Transcriptome Regulated by LUBAC. Frontiers in Immunology, 2019, 10, 479.	2.2	54
226	Primary immunodeficiencies (PIDs) presenting with cytopenias. Hematology American Society of Hematology Education Program, 2009, 2009, 139-143.	0.9	53
227	Humans with inherited TÂcell CD28 deficiency are susceptible to skin papillomaviruses but are otherwise healthy. Cell, 2021, 184, 3812-3828.e30.	13.5	53
228	Clinical, Immunological and Molecular Characteristics of 37 Iranian Patients with X-Linked Agammaglobulinemia. International Archives of Allergy and Immunology, 2006, 141, 408-414.	0.9	52
229	Combined decrease of defined B and T cell subsets in a group of common variable immunodeficiency patients. Clinical Immunology, 2006, 121, 203-214.	1.4	52
230	Clinical and genetic diagnosis of warts, hypogammaglobulinemia, infections, and myelokathexis syndrome in 10 patients. Journal of Allergy and Clinical Immunology, 2009, 123, 1170-1173.e3.	1.5	52
231	Heterozygous FOXN1 Variants Cause Low TRECs and Severe T Cell Lymphopenia, Revealing a Crucial Role of FOXN1 in Supporting Early Thymopoiesis. American Journal of Human Genetics, 2019, 105, 549-561.	2.6	52
232	Lymphocyte-driven regional immunopathology in pneumonitis caused by impaired central immune tolerance. Science Translational Medicine, 2019, 11, .	5.8	52
233	Defective actin polymerization in EBV-transformed B-cell lines from patients with the Wiskott-Aldrich syndrome. , 1998, 185, 99-107.		51
234	Applications of flow cytometry for the study of primary immune deficiencies. Current Opinion in Allergy and Clinical Immunology, 2008, 8, 499-509.	1.1	51

#	Article	IF	Citations
235	Patients with CD3G mutations reveal a role for human CD3Î³ in Treg diversity and suppressive function. Blood, 2018, 131, 2335-2344.	0.6	51
236	Functional defects of dendritic cells in patients with CD40 deficiency. Blood, 2003, 102, 4099-4106.	0.6	50
237	Cartilage-hair hypoplasia: molecular basis and heterogeneity of the immunological phenotype. Current Opinion in Allergy and Clinical Immunology, 2008, 8, 534-539.	1.1	50
238	Genetic and demographic features of X-linked agammaglobulinemia in Eastern and Central Europe: A cohort study. Molecular Immunology, 2009, 46, 2140-2146.	1.0	50
239	Impaired NK-cell migration in WAS/XLT patients: role of Cdc42/WASp pathway in the control of chemokine-induced Î²2 integrin high-affinity state. Blood, 2010, 115, 2818-2826.	0.6	50
240	Clinical and immunologic outcome of patients with cartilage hair hypoplasia after hematopoietic stem cell transplantation. Blood, 2010, 116, 27-35.	0.6	50
241	Hematopoietic Cell Transplantation for Wiskott-Aldrich Syndrome: Advances in Biology and Future Directions for Treatment. Immunology and Allergy Clinics of North America, 2010, 30, 179-194.	0.7	50
242	Lymphoid abnormalities in CD40 ligand transgenic mice suggest the need for tight regulation in gene therapy approaches to hyper immunoglobulin M (IgM) syndrome. Cancer Gene Therapy, 2000, 7, 1299-1306.	2.2	49
243	Clinical heterogeneity and diagnostic delay of autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy syndrome. Clinical Immunology, 2011, 139, 6-11.	1.4	49
244	Wiskott-Aldrich syndrome proteinâ€“mediated actin dynamics control type-I interferon production in plasmacytoid dendritic cells. Journal of Experimental Medicine, 2013, 210, 355-374.	4.2	49
245	Reticular dysgenesisâ€“associated AK2 protects hematopoietic stem and progenitor cell development from oxidative stress. Journal of Experimental Medicine, 2015, 212, 1185-1202.	4.2	49
246	A novel mutation in the POLE2 geneÂcausing combined immunodeficiency. Journal of Allergy and Clinical Immunology, 2016, 137, 635-638.e1.	1.5	49
247	Current Knowledge and Priorities for Future Research in Late Effects after Hematopoietic Stem Cell Transplantation (HCT) for Severe Combined Immunodeficiency Patients: A Consensus Statement from the Second Pediatric Blood and Marrow Transplant Consortium International Conference on Late Effects after Pediatric HCT. Biology of Blood and Marrow Transplantation, 2017, 23, 379-387.	2.0	49
248	Identification of an infant with severe combined immunodeficiency by newborn screening. Journal of Allergy and Clinical Immunology, 2010, 126, 1073-1074.	1.5	48
249	Hematopoietic stem cell transplantation outcomes for 11 patients with dedicator of cytokinesis 8 deficiency. Journal of Allergy and Clinical Immunology, 2016, 138, 852-859.e3.	1.5	48
250	Linker for Activation of T Cells (LAT), a Novel Immunohistochemical Marker for T Cells, NK Cells, Mast Cells, and Megakaryocytes. American Journal of Pathology, 1999, 154, 1037-1046.	1.9	46
251	An international study examining therapeutic options used in treatment of Wiskottâ€“Aldrich syndrome. Clinical Immunology, 2003, 109, 272-277.	1.4	46
252	Genetic Causes of Bronchiectasis: Primary Immune Deficiencies and the Lung. Respiration, 2007, 74, 264-275.	1.2	46

#	Article	IF	Citations
253	Decreased somatic hypermutation induces an impaired peripheral B cell tolerance checkpoint. Journal of Clinical Investigation, 2016, 126, 4289-4302.	3.9	46
254	Advances in the understanding of cytokine signal transduction: the role of Jaks and STATs in immunoregulation and the pathogenesis of immunodeficiency. Journal of Clinical Immunology, 1997, 17, 431-447.	2.0	45
255	Comparison of American and European practices in the management of patients with primary immunodeficiencies. Clinical and Experimental Immunology, 2012, 169, 57-69.	1.1	45
256	RAG1 Deficiency May Present Clinically as Selective IgA Deficiency. Journal of Clinical Immunology, 2015, 35, 280-288.	2.0	45
257	Modeling altered T-cell development with induced pluripotent stem cells from patients with RAG1-dependent immune deficiencies. Blood, 2016, 128, 783-793.	0.6	45
258	Robust Antibody and T Cell Responses to SARS-CoV-2 in Patients with Antibody Deficiency. Journal of Clinical Immunology, 2021, 41, 1146-1153.	2.0	45
259	The Spleen in the Wiskott-Aldrich Syndrome. American Journal of Surgical Pathology, 1999, 23, 182-191.	2.1	45
260	Glucose metabolism and diet-based prevention of liver dysfunction in MPV17 mutant patients. Journal of Hepatology, 2009, 50, 215-221.	1.8	44
261	Recommendations for Screening and Management of Late Effects in Patients with Severe Combined Immunodeficiency after Allogenic Hematopoietic Cell Transplantation: A Consensus Statement from the Second Pediatric Blood and Marrow Transplant Consortium International Conference on Late Effects after Pediatric HCT. Biology of Blood and Marrow Transplantation, 2017, 23, 1229-1240.	2.0	44
262	Activating mutations in PIK3CD disrupt the differentiation and function of human and murine CD4+ T cells. Journal of Allergy and Clinical Immunology, 2019, 144, 236-253.	1.5	44
263	<scp>RAG</scp> gene defects at the verge of immunodeficiency and immune dysregulation. Immunological Reviews, 2019, 287, 73-90.	2.8	44
264	A novel homozygous mutation in recombination activating gene 2 in 2 relatives with different clinical phenotypes: Omenn syndrome and hyper-IgM syndrome. Journal of Allergy and Clinical Immunology, 2012, 130, 1414-1416.	1.5	43
265	Severe eczema and Hyper-IgE in Loeysâ€“Dietz-syndrome â€" Contribution to new findings of immune dysregulation in connective tissue disorders. Clinical Immunology, 2014, 150, 43-50.	1.4	43
266	Development of Autologous T Lymphocytes in Two Males with X-Linked Severe Combined Immune Deficiency: Molecular and Cellular Characterization. Clinical Immunology, 2000, 95, 39-50.	1.4	42
267	Hematopoietic stem cell transplantation in Omenn syndrome: a single-center experience. Bone Marrow Transplantation, 2005, 36, 107-114.	1.3	42
268	Nextâ€Generation Sequencing Reveals Restriction and Clonotypic Expansion of Treg Cells in Juvenile Idiopathic Arthritis. Arthritis and Rheumatology, 2016, 68, 1758-1768.	2.9	42
269	Primary immunodeficiencies worldwide: an updated overview from the Jeffrey Modell Centers Global Network. Immunologic Research, 2016, 64, 736-753.	1.3	42
270	Prospects for ultraviolet A1 phototherapy as a treatment for chronic cutaneous graft-versus-host disease. Haematologica, 2003, 88, 1169-75.	1.7	42

#	Article	IF	Citations
271	Isolation of Cosmid and cDNA Clones in the Region Surrounding the BTK Gene at Xq21.3-q22. Genomics, 1994, 21, 517-524.	1.3	41
272	Distinct Interactions of the X-Linked Lymphoproliferative Syndrome Gene Product SAP with Cytoplasmic Domains of Members of the CD2 Receptor Family. Clinical Immunology, 2001, 100, 15-23.	1.4	41
273	Structure and function of the Wiskott-Aldrich syndrome protein. Current Opinion in Hematology, 2005, 12, 284-291.	1.2	41
274	Cord Blood Banking for Potential Future Transplantation. Pediatrics, 2017, 140, e20172695.	1.0	41
275	Natural Killer Cells from Patients with Recombinase-Activating Gene and Non-Homologous End Joining Gene Defects Comprise a Higher Frequency of CD56bright NKG2A+++ Cells, and Yet Display Increased Degranulation and Higher Perforin Content. Frontiers in Immunology, 2017, 8, 798.	2.2	41
276	Chronic Granulomatous Disease-Associated IBD Resolves and Does Not Adversely Impact Survival Following Allogeneic HCT. Journal of Clinical Immunology, 2019, 39, 653-667.	2.0	41
277	Next Generation Sequencing Reveals Skewing of the T and B Cell Receptor Repertoires in Patients with WiskottÃ¢â‚¬â€œAldrich Syndrome. Frontiers in Immunology, 2014, 5, 340.	2.2	40
278	Stem cell transplantation for tetratricopeptide repeat domain 7A deficiency: long-term follow-up. Blood, 2016, 128, 1306-1308.	0.6	40
279	Prevalence and clinical challenges among adults with primary immunodeficiency and recombination-activating gene deficiency. Journal of Allergy and Clinical Immunology, 2018, 141, 2303-2306.	1.5	40
280	WASP-mediated regulation of anti-inflammatory macrophages is IL-10 dependent and is critical for intestinal homeostasis. Nature Communications, 2018, 9, 1779.	5.8	40
281	Homozygous DNA ligase IV R278H mutation in mice leads to leaky SCID and represents a model for human LIG4 syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 3024-3029.	3.3	39
282	Combined Immunodeficiencies with Nonfunctional T Lymphocytes. Advances in Immunology, 2014, 121, 121-190.	1.1	39
283	Recombination activity of human recombination-activating gene 2 (RAG2) mutations and correlation with clinical phenotype. Journal of Allergy and Clinical Immunology, 2019, 143, 726-735.	1.5	39
284	Harnessing Type I IFN Immunity Against SARS-CoV-2 with Early Administration of IFN-Î². Journal of Clinical Immunology, 2021, 41, 1425-1442.	2.0	39
285	Recent Advances in Primary Immunodeficiencies: Identification of Novel Genetic Defects and Unanticipated Phenotypes. Pediatric Research, 2009, 65, 3R-12R.	1.1	38
286	Development of Autologous, Oligoclonal, Poorly Functioning T Lymphocytes in a Patient With Autosomal Recessive Severe Combined Immunodeficiency Caused by Defects of the Jak3 Tyrosine Kinase. Blood, 1998, 91, 949-955.	0.6	37
287	Impaired thymic output and restricted T-cell repertoire in two infants with immunodeficiency and early-onset generalized dermatitis. Immunology Letters, 2003, 86, 93-97.	1.1	37
288	Induced pluripotent stem cells: AÂnovel frontier in the study of human primary immunodeficiencies. Journal of Allergy and Clinical Immunology, 2011, 127, 1400-1407.e4.	1.5	37

#	Article	IF	Citations
289	Adult-onset manifestation of idiopathic T-cell lymphopenia due to a heterozygous RAG1 mutation. Journal of Allergy and Clinical Immunology, 2013, 131, 1421-1423.	1.5	37
290	B-cell differentiation and IL-21 response in IL2RG/JAK3 SCID patients after hematopoietic stem cell transplantation. Blood, 2018, 131, 2967-2977.	0.6	37
291	Gain-of-function CEBPE mutation causes noncanonical autoinflammatory inflammasomopathy. Journal of Allergy and Clinical Immunology, 2019, 144, 1364-1376.	1.5	37
292	Comprehensive Genetic Results for Primary Immunodeficiency Disorders in a Highly Consanguineous Population. Frontiers in Immunology, 2018, 9, 3146.	2.2	37
293	Flow Cytometry Identifies Risk Factors and Dynamic Changes in Patients with COVID-19. Journal of Clinical Immunology, 2020, 40, 970-973.	2.0	37
294	Immune dysregulation in patients with RAG deficiency and other forms of combined immune deficiency. Blood, 2020, 135, 610-619.	0.6	37
295	Founder effect for a 26-bp deletion in the RFXANK gene in North African major histocompatibility complex class II-deficient patients belonging to complementation group B. Immunogenetics, 2000, 51, 261-267.	1.2	36
296	Reduced thymic output, cell cycle abnormalities, and increased apoptosis of T lymphocytes in patients with cartilage-hair hypoplasia. Journal of Allergy and Clinical Immunology, 2011, 128, 139-146.	1.5	36
297	Infections in Infants with SCID: Isolation, Infection Screening, and Prophylaxis in PIDTC Centers. Journal of Clinical Immunology, 2021, 41, 38-50.	2.0	36
298	Characterization of a CD38-like 78-kilodalton soluble protein released from B cell lines derived from patients with X-linked agammaglobulinemia.. Journal of Clinical Investigation, 1998, 101, 2821-2830.	3.9	36
299	Gene therapy for primary immunodeficiencies: Looking ahead, toward gene correction. Journal of Allergy and Clinical Immunology, 2011, 127, 1344-1350.	1.5	35
300	Editorial: NK Cell Subsets in Health and Disease: New Developments. Frontiers in Immunology, 2017, 8, 1363.	2.2	35
301	WASPbase: a database of WAS- and XLT-causing mutations. Trends in Immunology, 1996, 17, 496-502.	7.5	35
302	Insulin-dependent diabetes mellitus and severe atopic dermatitis in a child with adenosine deaminase deficiency. European Journal of Pediatrics, 1992, 151, 811-814.	1.3	34
303	Mutations of the X-linked lymphoproliferative disease gene SH2D1A mimicking common variable immunodeficiency. European Journal of Pediatrics, 2002, 161, 656-659.	1.3	34
304	First report of successful stem cell transplantation in a child with CD40 deficiency. Bone Marrow Transplantation, 2007, 40, 279-281.	1.3	34
305	Mutations affecting mRNA splicing are the most common molecular defect in patients with familial hemophagocytic lymphohistiocytosis type 3. Haematologica, 2008, 93, 1086-1090.	1.7	34
306	Immune deficiency caused by impaired expression of nuclear factor-ÎºB essential modifier (NEMO) because of a mutation in the 5â€² untranslated region of the NEMO gene. Journal of Allergy and Clinical Immunology, 2010, 126, 127-132.e7.	1.5	34

#	Article	IF	Citations
307	Abnormalities of Thymic Stroma may Contribute to Immune Dysregulation in Murine Models of Leaky Severe Combined Immunodeficiency. Frontiers in Immunology, 2011, 2, .	2.2	34
308	Differential role of nonhomologous end joining factors in the generation, DNA damage response, and myeloid differentiation of human induced pluripotent stem cells. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 8889-8894.	3.3	34
309	International retrospective study of allogeneic hematopoietic cell transplantation for activated PI3K-delta syndrome. Journal of Allergy and Clinical Immunology, 2022, 149, 410-421.e7.	1.5	34
310	WASP and the phenotypic range associated with deficiency. Current Opinion in Allergy and Clinical Immunology, 2005, 5, 485-490.	1.1	33
311	Fatal autoimmunity in mice reconstituted with human hematopoietic stem cells encoding defective FOXP3. Blood, 2015, 125, 3886-3895.	0.6	33
312	Primary Immune Deficiency Treatment Consortium (PIDTC) update. Journal of Allergy and Clinical Immunology, 2016, 138, 375-385.	1.5	33
313	The Second Pediatric Blood and Marrow Transplant Consortium International Consensus Conference on Late Effects after Pediatric Hematopoietic Cell Transplantation: Defining the Unique Late Effects of Children Undergoing Hematopoietic Cell Transplantation for Immune Deficiencies, Inherited Marrow Failure Disorders, and Hemoglobinopathies. Biology of Blood and Marrow Transplantation, 2017, 23, 24-29.	2.0	33
314	Activated PI3KÎ´ breaches multiple B cell tolerance checkpoints and causes autoantibody production. Journal of Experimental Medicine, 2020, 217, .	4.2	33
315	Inhibition of HECT E3 ligases as potential therapy for COVID-19. Cell Death and Disease, 2021, 12, 310.	2.7	33
316	Pulmonary and Sinusal Changes in 45 Patients With Primary Immunodeficiencies. Journal of Computer Assisted Tomography, 2007, 31, 620-628.	0.5	32
317	First reported case of Omenn syndrome in a patient with reticular dysgenesis. Journal of Allergy and Clinical Immunology, 2013, 131, 1227-1230.e3.	1.5	32
318	Biallelic mutations in DNA ligase 1 underlie a spectrum of immune deficiencies. Journal of Clinical Investigation, 2018, 128, 5489-5504.	3.9	32
319	USIDNET: A Strategy to Build a Community of Clinical Immunologists. Journal of Clinical Immunology, 2014, 34, 428-435.	2.0	31
320	Disease specificity of anti-tryptophan hydroxylase-1 and anti-AIE-75 autoantibodies in APECED and IPEX syndrome. Clinical Immunology, 2015, 156, 36-42.	1.4	31
321	Reticular dysgenesis: international survey on clinical presentation, transplantation, and outcome. Blood, 2017, 129, 2928-2938.	0.6	31
322	Cysteine and hydrophobic residues in CDR3 serve as distinct T-cell self-reactivity indices. Journal of Allergy and Clinical Immunology, 2019, 144, 333-336.	1.5	31
323	Human DOCK2 Deficiency: Report of a Novel Mutation and Evidence for Neutrophil Dysfunction. Journal of Clinical Immunology, 2019, 39, 298-308.	2.0	31
324	Damaging-agent sensitivity of Artemis-deficient cell lines. European Journal of Immunology, 2005, 35, 1250-1256.	1.6	30

#	Article	IF	Citations
325	X-linked hypogammaglobulinemia and isolated growth hormone deficiency: an update. Immunologic Research, 2008, 40, 262-270.	1.3	30
326	Management of Primary Antibody Deficiency with Replacement Therapy: Summary of Guidelines. Immunology and Allergy Clinics of North America, 2008, 28, 875-876.	0.7	30
327	Parental Consanguinity and the Risk of Primary Immunodeficiency Disorders: Report from the Kuwait National Primary Immunodeficiency Disorders Registry. International Archives of Allergy and Immunology, 2011, 154, 76-80.	0.9	30
328	Impaired receptor editing and heterozygous RAG2 mutation in a patient with systemic lupus erythematosus and erosive arthritis. Journal of Allergy and Clinical Immunology, 2015, 135, 272-273.	1.5	30
329	DNA-PKcs Is Involved in Ig Class Switch Recombination in Human B Cells. Journal of Immunology, 2015, 195, 5608-5615.	0.4	30
330	Ligase-4 Deficiency Causes Distinctive Immune Abnormalities in Asymptomatic Individuals. Journal of Clinical Immunology, 2016, 36, 341-353.	2.0	30
331	Leukocyte adhesion deficiency in a child with severe oral involvement. Oral Surgery Oral Medicine Oral Pathology Oral Radiology and Endodontics, 1999, 87, 691-694.	1.6	29
332	Structural Basis for SH2D1A Mutations in X-Linked Lymphoproliferative Disease. Biochemical and Biophysical Research Communications, 2000, 269, 124-130.	1.0	29
333	Functional characterization of natural killer cells in type I leukocyte adhesion deficiency. Blood, 2007, 109, 4873-4881.	0.6	29
334	Different molecular behavior of CD40 mutants causing hyper-IgM syndrome. Blood, 2010, 116, 5867-5874.	0.6	29
335	Multiple intestinal atresia with combined immune deficiency. Current Opinion in Pediatrics, 2014, 26, 690-696.	1.0	29
336	Analysis of clinical and immunologic phenotype in a large cohort of children and adults with cartilage-hair hypoplasia. Journal of Allergy and Clinical Immunology, 2017, 140, 612-614.e5.	1.5	29
337	Hypomorphic Rag1 mutations alter the preimmune repertoire at early stages of lymphoid development. Blood, 2018, 132, 281-292.	0.6	29
338	A PCR-based non-radioactive X-chromosome inactivation assay for genetic counseling in X-linked primary immunodeficiencies. Life Sciences, 1997, 61, 1405-1411.	2.0	28
339	Mutation analysis by a nonâ€radioactive singleâ€strand conformation polymorphism assay in nine families with Xâ€linked severe combined immunodeficiency (SCIDX1). British Journal of Haematology, 1998, 101, 582-587.	1.2	28
340	Combined Immunodeficiencies Due to Defects in Signal Transduction: Defects of the Î³c-JAK3 Signaling Pathway as a Model. Immunobiology, 2000, 202, 106-119.	0.8	28
341	Eleven novel JAK3 mutations in patients with severe combined immunodeficiency?including the first patients with mutations in the kinase domain. Human Mutation, 2001, 18, 355-356.	1.1	28
342	Application and implementation of the GINA asthma guidelines by specialist and primary care physicians: a longitudinal follow-up study on 264 children. Primary Care Respiratory Journal: Journal of the General Practice Airways Group, 2007, 16, 357-362.	2.5	28

#	Article	IF	Citations
343	Lack of iNKT cells in patients with combined immune deficiency due to hypomorphic RAG mutations. Blood, 2008, 111, 271-274.	0.6	28
344	Leukocyte trafficking in primary immunodeficiencies. Journal of Leukocyte Biology, 2009, 85, 335-343.	1.5	28
345	Immunodeficiency and immune dysregulation associated with proximal defects of T cell receptor signaling. Current Opinion in Immunology, 2014, 31, 97-101.	2.4	28
346	First Case of X-Linked Moesin Deficiency Identified After Newborn Screening for SCID. Journal of Clinical Immunology, 2017, 37, 336-338.	2.0	28
347	Abnormalities of T-cell receptor repertoire in CD4+ regulatory and conventional T cells in patients with RAG mutations: Implications for autoimmunity. Journal of Allergy and Clinical Immunology, 2017, 140, 1739-1743.e7.	1.5	28
348	Autonomous role of Wiskott-Aldrich syndrome platelet deficiency in inducing autoimmunity and inflammation. Journal of Allergy and Clinical Immunology, 2018, 142, 1272-1284.	1.5	28
349	Efficacy of lentivirus-mediated gene therapy in an Omenn syndrome recombination-activating gene 2 mouse model is not hindered by inflammation and immune dysregulation. Journal of Allergy and Clinical Immunology, 2018, 142, 928-941.e8.	1.5	28
350	T cell dynamics and response of the microbiota after gene therapy to treat X-linked severe combined immunodeficiency. Genome Medicine, 2018, 10, 70.	3.6	28
351	Disrupted N-linked glycosylation as a disease mechanism in deficiency of ADA2. Journal of Allergy and Clinical Immunology, 2018, 142, 1363-1365.e8.	1.5	28
352	Prospective Study of a Novel, Radiation-Free, Reduced-Intensity Bone Marrow Transplantation Platform for Primary Immunodeficiency Diseases. Biology of Blood and Marrow Transplantation, 2020, 26, 94-106.	2.0	28
353	SASH3variants cause a novel form of X-linked combined immunodeficiency with immune dysregulation. Blood, 2021, 138, 1019-1033.	0.6	28
354	CD70 expression on T-cell subpopulations: study of normal individuals and patients with chronic immune activation. Immunology Letters, 1997, 55, 99-104.	1.1	27
355	Definition of primary immunodeficiency in 2011: a â€œtrialogueâ€•among friends. Annals of the New York Academy of Sciences, 2011, 1238, 1-6.	1.8	27
356	Hypomorphic Janus kinase 3 mutations result in a spectrum of immune defects, including partial maternal T-cell engraftment. Journal of Allergy and Clinical Immunology, 2013, 131, 1136-1145.	1.5	27
357	Late Onset Hypomorphic RAG2 Deficiency Presentation with Fatal Vaccine-Strain VZV Infection. Journal of Clinical Immunology, 2015, 35, 754-760.	2.0	27
358	Human CD40 ligand deficiency dysregulates the macrophage transcriptome causing functional defects that are improved by exogenous IFN-Î³. Journal of Allergy and Clinical Immunology, 2017, 139, 900-912.e7.	1.5	27
359	IgG Fc glycosylation as an axis of humoral immunity in childhood. Journal of Allergy and Clinical Immunology, 2020, 145, 710-713.e9.	1.5	27
360	Efficacy and safety of anti-CD45â€“saporin as conditioning agent for RAG deficiency. Journal of Allergy and Clinical Immunology, 2021, 147, 309-320.e6.	1.5	27

#	Article	IF	Citations
361	Gene Editing Rescues In vitro T Cell Development of RAG2-Deficient Induced Pluripotent Stem Cells in an Artificial Thymic Organoid System. Journal of Clinical Immunology, 2021, 41, 852-862.	2.0	27
362	In vitro cell death of activated lymphocytes in Omenn's syndrome. European Journal of Immunology, 1997, 27, 2765-2773.	1.6	26
363	The hyper-IgM (HIM) syndrome. Seminars in Immunopathology, 1998, 19, 383-399.	4.0	26
364	Immune reconstitution after in utero bone marrow transplantation in a fetus with severe combined immunodeficiency with natural killer cells. Transplantation Proceedings, 1999, 31, 2581.	0.3	26
365	The awareness among paediatricians of off-label prescribing in children: a survey of Italian hospitals. European Journal of Clinical Pharmacology, 2007, 63, 81-85.	0.8	26
366	Is it necessary to identify molecular defects in primary immunodeficiency disease?. Journal of Allergy and Clinical Immunology, 2008, 122, 1069-1073.	1.5	26
367	Out of breath: GM-CSFRÎ± mutations disrupt surfactant homeostasis. Journal of Experimental Medicine, 2008, 205, 2693-2697.	4.2	26
368	Temporal Dynamics of Antiâ€“Type 1 Interferon Autoantibodies in Patients With Coronavirus Disease 2019. Clinical Infectious Diseases, 2022, 75, e1192-e1194.	2.9	26
369	Outcomes following treatment for ADA-deficient severe combined immunodeficiency: a report from the PIDTC. Blood, 2022, 140, 685-705.	0.6	26
370	A peptide derived from the Wiskott-Aldrich syndrome (WAS) protein-interacting protein (WIP) restores WAS protein level and actin cytoskeleton reorganization in lymphocytes from patients with WAS mutations that disrupt WIP binding. Journal of Allergy and Clinical Immunology, 2011, 127, 998-1005.e2.	1.5	25
371	Major Histocompatibility Complex Class II Deficiency in Kuwait: Clinical Manifestations, Immunological Findings and Molecular Profile. Journal of Clinical Immunology, 2013, 33, 513-519.	2.0	25
372	Î¶ Chainâ€“associated protein of 70 kDa (ZAP70) deficiency in human subjects is associated with abnormalities of thymic stromal cells: Implications for T-cell tolerance. Journal of Allergy and Clinical Immunology, 2013, 131, 597-600.e2.	1.5	25
373	RAG1 Reversion Mosaicism in a Patient with Omenn Syndrome. Journal of Clinical Immunology, 2014, 34, 551-554.	2.0	25
374	Presence of hypogammaglobulinemia and abnormal antibody responses in GATA2 deficiency. Journal of Allergy and Clinical Immunology, 2014, 134, 223-226.	1.5	25
375	Hematopoietic Stem Cell Transplantation for X-Linked Thrombocytopenia With Mutations in the WAS gene. Journal of Clinical Immunology, 2015, 35, 15-21.	2.0	25
376	Broad spectrum of autoantibodies in patients with Wiskott-Aldrich syndrome and X-linked thrombocytopenia. Journal of Allergy and Clinical Immunology, 2015, 136, 1401-1404.e3.	1.5	25
377	Deletion of WASp and N-WASp in B cells cripples the germinal center response and results in production of IgM autoantibodies. Journal of Autoimmunity, 2015, 62, 81-92.	3.0	25
378	Human OTULIN haploinsufficiency impairs cell-intrinsic immunity to staphylococcal Î±-toxin. Science, 2022, 376, eabm6380.	6.0	25

#	Article	IF	Citations
379	Prenatal diagnosis of JAK3 deficient SCID. , 1999, 19, 653-656.		24
380	Natural killer cell deficiencies and severe varicella infection. Journal of Pediatrics, 2006, 148, 563-564.	0.9	24
381	N-WASP is required for B-cellâ€“mediated autoimmunity in Wiskott-Aldrich syndrome. Blood, 2016, 127, 216-220.	0.6	24
382	Molecular aspects of primary immunodeficiencies: lessons from cytokine and other signaling pathways. Journal of Clinical Investigation, 2002, 109, 1261-1269.	3.9	24
383	Reconstitution of T-cell compartment after in utero stem cell transplantation: analysis of T-cell repertoire and thymic output. Haematologica, 2004, 89, 450-61.	1.7	24
384	Molecular Modeling of the Jak3 Kinase Domains and Structural Basis for Severe Combined Immunodeficiency. Clinical Immunology, 2000, 96, 108-118.	1.4	23
385	Monocytes from Wiskott-Aldrich patients differentiate in functional mature dendritic cells with a defect in CD83 expression. European Journal of Immunology, 2001, 31, 3413-3421.	1.6	23
386	B cell function after haploidentical in utero bone marrow transplantation in a patient with severe combined immunodeficiency. Bone Marrow Transplantation, 2002, 29, 625-628.	1.3	22
387	Outcome of hematopoietic stem cell transplantation in hyper-IgM syndrome caused by CD40 deficiency. Journal of Pediatrics, 2003, 143, 141-142.	0.9	22
388	X-linked immunodeficiencies. Current Allergy and Asthma Reports, 2004, 4, 339-348.	2.4	22
389	Matched unrelated bone marrow transplant for severe combined immunodeficiency. Immunologic Research, 2007, 38, 191-200.	1.3	22
390	Genetically determined lymphopenia and autoimmune manifestations. Current Opinion in Immunology, 2008, 20, 318-324.	2.4	22
391	Exacerbated experimental arthritis in Wiskottâ€“Aldrich syndrome protein deficiency: Modulatory role of regulatory B cells. European Journal of Immunology, 2014, 44, 2692-2702.	1.6	22
392	Lessons in gene hunting: AÂRAG1 mutation presenting with agammaglobulinemia and absence of B cells. Journal of Allergy and Clinical Immunology, 2014, 134, 983-985.e1.	1.5	22
393	Combined immunodeficiency in the United States and Kuwait: Comparison of patients' characteristics and molecular diagnosis. Clinical Immunology, 2015, 161, 170-173.	1.4	22
394	Future of Care for Patients With Chronic Granulomatous Disease: Gene Therapy and Targeted Molecular Medicine. Journal of the Pediatric Infectious Diseases Society, 2018, 7, S40-S44.	0.6	22
395	RAG deficiencies: Recent advances in disease pathogenesis and novel therapeutic approaches. European Journal of Immunology, 2021, 51, 1028-1038.	1.6	22
396	Treatment of Relapsing HPV Diseases by Restored Function of Natural Killer Cells. New England Journal of Medicine, 2021, 385, 921-929.	13.9	22

#	Article	IF	Citations
397	3. Molecular Basis for X-Linked Immunodeficiencies. Advances in Genetics, 1997, 35, 57-115.	0.8	21
398	Rag Defects and Thymic Stroma: Lessons from Animal Models. Frontiers in Immunology, 2014, 5, 259.	2.2	21
399	Neutrophils drive type I interferon production and autoantibodies in patients with Wiskott-Aldrich syndrome. Journal of Allergy and Clinical Immunology, 2018, 142, 1605-1617.e4.	1.5	21
400	CD40 ligand deficiency causes functional defects of peripheral neutrophils that are improved by exogenous IFN-Î³. Journal of Allergy and Clinical Immunology, 2018, 142, 1571-1588.e9.	1.5	21
401	B cellâ€“intrinsic requirement for STK4 in humoral immunity in mice and human subjects. Journal of Allergy and Clinical Immunology, 2019, 143, 2302-2305.	1.5	21
402	F-BAR domain only protein 1 (FCHO1) deficiency is a novel cause of combined immune deficiency in human subjects. Journal of Allergy and Clinical Immunology, 2019, 143, 2317-2321.e12.	1.5	21
403	Respiratory viral infections in otherwise healthy humans with inherited IRF7 deficiency. Journal of Experimental Medicine, 2022, 219, .	4.2	21
404	Immunodeficiencies due to defects of class-switch recombination. Immunologic Research, 2007, 38, 68-77.	1.3	20
405	Partial defects of T-cell development associated with poor T-cell function. Journal of Allergy and Clinical Immunology, 2013, 131, 1297-1305.	1.5	20
406	Agammaglobulinemia associated to nasal polyposis due to a hypomorphic RAG1 mutation in a 12 years old boy. Clinical Immunology, 2016, 173, 121-123.	1.4	20
407	Ten Years of Newborn Screening for Severe Combined Immunodeficiency (SCID) in Massachusetts. Journal of Allergy and Clinical Immunology: in Practice, 2021, 9, 2060-2067.e2.	2.0	20
408	CRISPR-targetedMAGT1insertion restores XMEN patient hematopoietic stem cells and lymphocytes. Blood, 2021, 138, 2768-2780.	0.6	20
409	Pharmacokinetics of prednisone and its metabolite prednisolone in children with nephrotic syndrome during the active phase and in remission.. British Journal of Clinical Pharmacology, 1984, 17, 423-431.	1.1	19
410	Defective expression of HLA class I and CD1a molecules in boy with Marfan-like phenotype and deep skin ulcers. Journal of the American Academy of Dermatology, 1996, 35, 814-818.	0.6	19
411	Molecular and biochemical characterization of JAK3 deficiency in a patient with severe combined immunodeficiency over 20 years after bone marrow transplantation: implications for treatment. British Journal of Haematology, 1998, 102, 1363-1366.	1.2	19
412	Stem cell transplantation in primary immunodeficiencies. Current Opinion in Allergy and Clinical Immunology, 2006, 6, 443-448.	1.1	19
413	Cytogenetic and molecular characterization of a de-novo t(2p;7p) translocation involving TNS3 and EXOC6B genes in a boy with a complex syndromic phenotype. European Journal of Medical Genetics, 2008, 51, 292-302.	0.7	19
414	Primary immunodeficiencies: increasing market share. Current Opinion in Immunology, 2009, 21, 461-465.	2.4	19

#	Article	IF	Citations
415	Bâ€cell intrinsic TLR7 signals promote depletion of the marginal zone in a murine model of Wiskottâ€“Aldrich syndrome. European Journal of Immunology, 2015, 45, 2773-2779.	1.6	19
416	Deciphering Human Cell-Autonomous Anti-HSV-1 Immunity in the Central Nervous System. Frontiers in Immunology, 2015, 6, 208.	2.2	19
417	Unrelated Hematopoietic Cell Transplantation in a Patient with Combined Immunodeficiency with Granulomatous Disease and Autoimmunity Secondary to RAG Deficiency. Journal of Clinical Immunology, 2016, 36, 725-732.	2.0	19
418	Defining a new immune deficiency syndrome: MAN2B2-CDG. Journal of Allergy and Clinical Immunology, 2020, 145, 1008-1011.	1.5	19
419	An appraisal of the Wilson & Jungner criteria in the context of genomic-based newborn screening for inborn errors of immunity. Journal of Allergy and Clinical Immunology, 2021, 147, 428-438.	1.5	19
420	Defective migration of monocyte-derived dendritic cells in LAD-1 immunodeficiency. Journal of Leukocyte Biology, 2002, 72, 650-6.	1.5	19
421	Lentivector cryptic splicing mediates increase in CD34+ clones expressing truncated HMGA2 in human X-linked severe combined immunodeficiency. Nature Communications, 2022, 13, .	5.8	19
422	BTK: 22 novel and 25 recurrent mutations in European patients with X-linked agammaglobulinemia. Human Mutation, 2004, 23, 286-286.	1.1	18
423	Advances in basic and clinical immunology in 2014. Journal of Allergy and Clinical Immunology, 2015, 135, 1132-1141.	1.5	18
424	Gut Microbiotaâ€“Host Interactions in Inborn Errors of Immunity. International Journal of Molecular Sciences, 2021, 22, 1416.	1.8	18
425	Donor source and postâ€transplantation cyclophosphamide influence outcome in allogeneic stem cell transplantation for GATA2 deficiency. British Journal of Haematology, 2022, 196, 169-178.	1.2	18
426	Percutaneous transhepatic alcoholization: a new therapeutic strategy in children with chronic granulomatous disease and liver abscess. Pediatric Infectious Disease Journal, 2002, 21, 1081-1083.	1.1	18
427	Primary immunodeficiencies: novel genes and unusual presentations. Hematology American Society of Hematology Education Program, 2019, 2019, 443-448.	0.9	18
428	Autoantibodies Against Proteins Previously Associated With Autoimmunity in Adult and Pediatric Patients With COVID-19 and Children With MIS-C. Frontiers in Immunology, 2022, 13, 841126.	2.2	18
429	Mapping of the X linked form of hyper IgM syndrome (HIGM1). Journal of Medical Genetics, 1993, 30, 202-205.	1.5	17
430	Ontogeny of CD40 expression by activated peripheral blood lymphocytes in humans. Immunology Letters, 1996, 49, 27-30.	1.1	17
431	Immunodeficiencies caused by genetic defects in protein kinases. Current Opinion in Immunology, 1996, 8, 448-453.	2.4	17
432	PIDs and cancer: an evolving story. Blood, 2010, 116, 1189-1190.	0.6	17

#	Article	IF	Citations
433	BTK inhibitors for severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2): A systematic review. Clinical Immunology, 2021, 230, 108816.	1.4	17
434	The Wiskottâ€“Aldrich syndrome: from genotypeâ€“phenotype correlation to treatment. Expert Review of Clinical Immunology, 2007, 3, 813-824.	1.3	16
435	Clinical and molecular features of X-linked hyper IgM syndrome â€“ An experience from North India. Clinical Immunology, 2018, 195, 59-66.	1.4	16
436	Disseminated and Congenital Toxoplasmosis in a Mother and Child With Activated PI3-Kinase Î´ Syndrome Type 2 (APDS2): Case Report and a Literature Review of Toxoplasma Infections in Primary Immunodeficiencies. Frontiers in Immunology, 2019, 10, 77.	2.2	16
437	The Clinical and Genetic Spectrum of 82 Patients With RAG Deficiency Including a c.256_257delAA Founder Variant in Slavic Countries. Frontiers in Immunology, 2020, 11, 900.	2.2	16
438	SARS-CoV-2 Spike Protein-Directed Monoclonal Antibodies May Ameliorate COVID-19 Complications in APECED Patients. Frontiers in Immunology, 2021, 12, 720205.	2.2	16
439	Nfkb2 variants reveal a p100-degradation threshold that defines autoimmune susceptibility. Journal of Experimental Medicine, 2021, 218, .	4.2	16
440	Molecular aspects of primary immunodeficiencies: lessons from cytokine and other signaling pathways. Journal of Clinical Investigation, 2002, 109, 1261-1269.	3.9	16
441	Elective bone marrow transplantation in a child with X-linked hyper-IgM syndrome presenting with acute respiratory distress syndrome. Bone Marrow Transplantation, 2002, 30, 49-52.	1.3	15
442	Mutations of the T-cell receptor constant region after in utero stem cell transplantation. Immunogenetics, 2004, 56, 214-9.	1.2	15
443	AIRE and immunological tolerance: insights from the study of autoimmune polyendocrinopathy candidiasis and ectodermal dystrophy. Current Opinion in Allergy and Clinical Immunology, 2004, 4, 491-496.	1.1	15
444	Advances in basic and clinical immunology in 2012. Journal of Allergy and Clinical Immunology, 2013, 131, 675-682.	1.5	15
445	Advances in basic and clinical immunology in 2013. Journal of Allergy and Clinical Immunology, 2014, 133, 967-976.	1.5	15
446	Targeted Therapy with Biologicals and Small Molecules in Primary Immunodeficiencies. Medical Principles and Practice, 2020, 29, 101-112.	1.1	15
447	Evidence of SARS-CoV-2-Specific T-Cell-Mediated Myocarditis in a MIS-A Case. Frontiers in Immunology, 2021, 12, 779026.	2.2	15
448	Neonatal T4+ lymphocytes: Analysis of the expression of 4B4 and 2H4 antigens. Clinical Immunology and Immunopathology, 1988, 46, 61-67.	2.1	14
449	Education and worldwide collaboration pays off. Nature Immunology, 2007, 8, 323-324.	7.0	14
450	Diagnosing XLP1 in patients with hemophagocytic lymphohistiocytosis. Journal of Allergy and Clinical Immunology, 2014, 134, 1381-1387.e7.	1.5	14

#	Article	IF	Citations
451	HSCT corrects primary immunodeficiency and immune dysregulation in patients with POMP-related autoinflammatory disease. Blood, 2021, 138, 1896-1901.	0.6	14
452	CD4+ cells from patients with Common Variable Immunodeficiency have a reduced ability of CD40 ligand membrane expression after in vitro stimulation. Pediatric Allergy and Immunology, 1996, 7, 176-179.	1.1	13
453	Prenatal diagnosis of RAG-deficient Omenn syndrome. , 2000, 20, 56-59.		13
454	Molecular genetic analysis of Hungarian patients with the hyper-immunoglobulin M syndrome. Molecular Immunology, 2008, 45, 278-282.	1.0	13
455	Single-center analysis of long-term outcome after hematopoietic cell transplantation in children with congenital severe T cell immunodeficiency. Immunologic Research, 2009, 44, 4-17.	1.3	13
456	Autoimmune lymphoproliferative syndrome caused by a homozygous FasL mutation that disrupts FasL assembly. Journal of Allergy and Clinical Immunology, 2016, 137, 324-327.e2.	1.5	13
457	Estimated disease incidence of RAG1/2 mutations: AÂcase report and querying the Exome Aggregation Consortium. Journal of Allergy and Clinical Immunology, 2017, 139, 690-692.e3.	1.5	13
458	Cutaneous barrier leakage and gut inflammation drive skin disease in Omenn syndrome. Journal of Allergy and Clinical Immunology, 2020, 146, 1165-1179.e11.	1.5	13
459	Lymphoid abnormalities in CD40 ligand transgenic mice suggest the need for tight regulation in gene therapy approaches to hyper immunoglobulin M (IgM) syndrome. Cancer Gene Therapy, 2000, 7, 1299-1306.	2.2	13
460	Case Report: Severe Rhabdomyolysis and Multiorgan Failure After ChAdOx1 nCoV-19 Vaccination. Frontiers in Immunology, 2022, 13, 845496.	2.2	13
461	Henoch-Schonlein syndrome and selective IgA deficiency.. Archives of Disease in Childhood, 1985, 60, 160-162.	1.0	12
462	Recombinase activating gene enzymes of lymphocytes. Current Opinion in Hematology, 2001, 8, 41-46.	1.2	12
463	The role of induced pluripotent stem cells in research and therapy of primary immunodeficiencies. Current Opinion in Immunology, 2012, 24, 617-624.	2.4	12
464	Classification of primary immunodeficiency disorders: One-fits-all does not help anymore. Clinical Immunology, 2012, 144, 24-25.	1.4	12
465	Natural killer cell hyporesponsiveness and impaired development in a CD247-deficient patient. Journal of Allergy and Clinical Immunology, 2016, 137, 942-945.e4.	1.5	12
466	Association of rare predicted loss-of-function variants of influenza-related type I IFN genes with critical COVID-19 pneumonia. Journal of Clinical Investigation, 2021, 131, .	3.9	12
467	RAG deficiency with ALPS features successfully treated with TCRÎ±Î²/CD19 cell depleted haploidentical stem cell transplant. Clinical Immunology, 2018, 187, 102-103.	1.4	12
468	Gene Therapy Using a Self-Inactivating Lentiviral Vector Improves Clinical and Laboratory Manifestations of Wiskott-Aldrich Syndrome. Blood, 2015, 126, 260-260.	0.6	12

#	Article	IF	Citations
469	Pancreatitis in systemic lupus erythematosus. Arthritis and Rheumatism, 1983, 26, 1173-1173.	6.7	11
470	Intravenous immunoglobulin in two children with Guillain-BarrÃ© syndrome. European Journal of Pediatrics, 1993, 152, 372-374.	1.3	11
471	C to T mutation causing premature termination of CD40 ligand at amino acid 221 in a patient affected by Hyper IgM syndrome. Human Mutation, 1994, 3, 73-75.	1.1	11
472	ITGB2 mutation combined with deleted ring 21 chromosome in a child with leukocyte adhesion deficiency. Journal of Allergy and Clinical Immunology, 2009, 124, 1356-1358.	1.5	11
473	A custom 148 gene-based resequencing chip and the SNP explorer software: new tools to study antibody deficiency. Human Mutation, 2010, 31, 1080-1088.	1.1	11
474	Decreased IL-10 production by EBV-transformed B cells from patients with VODI: Implications forÂthe pathogenesis of Crohn disease. Journal of Allergy and Clinical Immunology, 2012, 129, 1678-1680.	1.5	11
475	Detection of Sp110 by Flow Cytometry and Application to Screening Patients for Veno-occlusive Disease with Immunodeficiency. Journal of Clinical Immunology, 2017, 37, 707-714.	2.0	11
476	DNA recombination defects in Kuwait: Clinical, immunologic and genetic profile. Clinical Immunology, 2018, 187, 68-75.	1.4	11
477	Rapid generation of novel models of RAG1 deficiency by CRISPR/Cas9-induced mutagenesis in murine zygotes. Oncotarget, 2016, 7, 12962-12974.	0.8	11
478	Chronic Granulomatous Disease With Inflammatory Bowel Disease: Clinical Presentation, Treatment, and Outcomes From the USIDNET Registry. Journal of Allergy and Clinical Immunology: in Practice, 2022, 10, 1325-1333.e5.	2.0	11
479	Cutaneous CD30+ Cells in Children with Atopic Dermatitis. International Archives of Allergy and Immunology, 2000, 121, 224-228.	0.9	10
480	X-linked hypogammaglobulinemia and isolated growth hormone deficiency: an update. Immunologic Research, 2007, 38, 391-399.	1.3	10
481	Novel presentation of Omenn syndrome in association with aniridia. Journal of Allergy and Clinical Immunology, 2009, 123, 966-969.	1.5	10
482	The long road to optimal management for chronic granulomatous disease. Journal of Allergy and Clinical Immunology, 2013, 132, 1164-1165.	1.5	10
483	Genetic variation in schlafen genes in a patient with a recapitulation of the murine Elektra phenotype. Journal of Allergy and Clinical Immunology, 2014, 133, 1462-1465.e5.	1.5	10
484	Paravertebral Mushroom: Identification of a Novel Species of Phellinus as a Human Pathogen in Chronic Granulomatous Disease. Journal of Clinical Microbiology, 2014, 52, 2726-2729.	1.8	10
485	Two Unique Cases of X-linked SCID: A Diagnostic Challenge in the Era of Newborn Screening. Frontiers in Pediatrics, 2019, 7, 55.	0.9	10
486	Vasculitis as a Major Morbidity Factor in Patients With Partial RAG Deficiency. Frontiers in Immunology, 2020, 11, 574738.	2.2	10

#	Article	IF	Citations
487	POLD1 Deficiency Reveals a Role for POLD1 in DNA Repair and T and B Cell Development. Journal of Clinical Immunology, 2021, 41, 270-273.	2.0	10
488	Lost in Translation: Lack of CD4 Expression due to a Novel Genetic Defect. Journal of Infectious Diseases, 2021, 223, 645-654.	1.9	10
489	Clonal hematopoiesis is not significantly associated with COVID-19 disease severity. Blood, 2022, 140, 1650-1655.	0.6	10
490	IgG subclass serum levels in juvenile chronic arthritis.. Annals of the Rheumatic Diseases, 1986, 45, 400-404.	0.5	9
491	Wiskott-Aldrich syndrome carrier detection with the hypervariable marker M27Î². Human Genetics, 1992, 89, 223-228.	1.8	9
492	Update on X-linked hypogammaglobulinemia with isolated growth hormone deficiency. Current Opinion in Allergy and Clinical Immunology, 2005, 5, 510-512.	1.1	9
493	What does it take to call it a pathogenic mutation?. Clinical Immunology, 2008, 128, 285-286.	1.4	9
494	Immunodeficiency with Autoimmunity: Beyond the Paradox. Frontiers in Immunology, 2013, 4, 77.	2.2	9
495	Hematopoietic stem cell transplantation for activated phosphoinositide 3-kinase Î´ syndrome: Who, when, and how?. Journal of Allergy and Clinical Immunology, 2019, 143, 91-93.	1.5	9
496	Asymptomatic Infant With Atypical SCID and Novel Hypomorphic RAG Variant Identified by Newborn Screening: A Diagnostic and Treatment Dilemma. Frontiers in Immunology, 2020, 11, 1954.	2.2	9
497	The Pathology of Omenn's Syndrome. American Journal of Surgical Pathology, 1996, 20, 773.	2.1	9
498	T-cell lymphoblastic leukemia/lymphoma in Nijmegen breakage syndrome. Haematologica, 2004, 89, ECR27.	1.7	9
499	The Genomic Organization of the Human Transcription Factor 3 (TFE3) Gene. Genomics, 1995, 28, 491-494.	1.3	8
500	Mutation analysis in Wiskott Aldrich syndrome on chorionic villus DNA. Lancet, The, 1995, 346, 641-642.	6.3	8
501	A novel activation-induced cytidine deaminase gene mutation in a Tunisian family with hyper IgM syndrome. European Journal of Pediatrics, 2004, 163, 704-708.	1.3	8
502	Genotyping for Guiding Drug Choice in Human Immunodeficiency Virus-Infected Children Failing Multiple Antiretroviral Treatment Regimens. Pediatric Infectious Disease Journal, 2005, 24, 747-749.	1.1	8
503	A mutation in caspase-9 decreases the expression of BAFFR and ICOS in patients with immunodeficiency and lymphoproliferation. Genes and Immunity, 2015, 16, 151-161.	2.2	8
504	Diverse Autoantibody Reactivity in Cartilage-Hair Hypoplasia. Journal of Clinical Immunology, 2017, 37, 508-510.	2.0	8

#	Article	IF	Citations
505	The immunologic features of patients with early-onset and polyautoimmunity. Clinical Immunology, 2020, 211, 108326.	1.4	8
506	Nodular regenerative hyperplasia in X-linked agammaglobulinemia: An underestimated and severe complication. Journal of Allergy and Clinical Immunology, 2022, 149, 400-409.e3.	1.5	8
507	Thymic Epithelial Cell Alterations and Defective Thymopoiesis Lead to Central and Peripheral Tolerance Perturbation in MHCII Deficiency. Frontiers in Immunology, 2021, 12, 669943.	2.2	8
508	Poor T-cell receptor Î² repertoire diversity early posttransplant for severe combined immunodeficiency predicts failure of immune reconstitution. Journal of Allergy and Clinical Immunology, 2022, 149, 1113-1119.	1.5	8
509	A new immunoperoxidase assay for Lolium perenne-specltic IgE in serum based on the biotin/avidin system (BAS). Clinical and Experimental Allergy, 1984, 14, 373-378.	1.4	7
510	Western blot technique in the serological evaluation of three LAV/HTLV III-infected Italian families. Infection, 1986, 14, 60-63.	2.3	7
511	Cellular and molecular pathogenesis of X-linked lymphoproliferative disease. Current Opinion in Allergy and Clinical Immunology, 2001, 1, 513-517.	1.1	7
512	Primary immunodeficiency modeling with induced pluripotent stem cells. Current Opinion in Allergy and Clinical Immunology, 2011, 11, 505-511.	1.1	7
513	Hematopoietic stem cell transplantation for hyperâ€‹scp›I‹/scp›g‹scp›M‹/scp› syndromes. Pediatric Transplantation, 2013, 17, 1-2.	0.5	7
514	Novel INHAT Repressor (NIR) is required for early lymphocyte development. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 13930-13935.	3.3	7
515	Immune Dysregulation, Polyendocrinopathy, Enteropathy, Xâ€Linked Syndrome Associated with Neonatal Epidermolysis Bullosa Acquisita. Pediatric Dermatology, 2015, 32, e74-7.	0.5	7
516	Severe combined immune deficiency. , 2020, , 153-205.		7
517	Enhanced Transduction Lentivector Gene Therapy for Treatment of Older Patients with X-Linked Severe Combined Immunodeficiency. Blood, 2019, 134, 608-608.	0.6	7
518	The Use of Induced Pluripotent Stem Cells to Study the Effects of Adenosine Deaminase Deficiency on Human Neutrophil Development. Frontiers in Immunology, 2021, 12, 748519.	2.2	7
519	SARS-CoV-2 infection in dialysis and kidney transplant patients: immunological and serological response. Journal of Nephrology, 2022, , 1.	0.9	7
520	Effect of Interferon-Î± Therapy in a Patient with Common Variable Immunodeficiency and Chronic Epstein-Barr Virus Infection. Pediatric Hematology and Oncology, 1995, 12, 489-493.	0.3	6
521	Indications for Hemopoietic Stem Cell Transplantation. Immunology and Allergy Clinics of North America, 2010, 30, 261-262.	0.7	6
522	Intronic SH2D1A mutation with impaired SAP expression and agammaglobulinemia. Clinical Immunology, 2013, 146, 84-89.	1.4	6

#	Article	IF	Citations
523	Expanding the spectrum of skeletal dysplasia with immunodeficiency: a commentary on identification of biallelic EXTL3 mutations in a novel type of spondylo-epi-metaphyseal dysplasia. Journal of Human Genetics, 2017, 62, 737-738.	1.1	6
524	Increased proportions of Î³Î´ T lymphocytes in atypical SCID associate with disease manifestations. Clinical Immunology, 2019, 201, 30-34.	1.4	6
525	Lentiviral Hematopoietic Stem Cell Gene Therapy for Older Patients with X-Linked Severe Combined Immunodeficiency. Blood, 2015, 126, 261-261.	0.6	6
526	JAK3-DEFICIENT SEVERE COMBINED IMMUNODEFICIENCY. Immunology and Allergy Clinics of North America, 2000, 20, 97-111.	0.7	5
527	Toll-like receptor-4 genotype in children with respiratory infections. Allergy: European Journal of Allergy and Clinical Immunology, 2004, 59, 1018-1019.	2.7	5
528	Survey on retransplantation criteria for patients with severe combined immunodeficiency. Journal of Allergy and Clinical Immunology, 2014, 133, 597-599.	1.5	5
529	Advances in clinical immunology in 2015. Journal of Allergy and Clinical Immunology, 2016, 138, 1531-1540.	1.5	5
530	Response to Comments on â€œAberrant type 1 immunity drives susceptibility to mucosal fungal infectionsâ€• Science, 2021, 373, eabi8835.	6.0	5
531	Presentation of Wiskott Aldrich syndrome as isolated thrombocytopenia. Blood, 1991, 77, 1125-6.	0.6	5
532	Severe combined immunodeficiencies of the common gamma-chain/JAK3 signaling pathway. Israel Medical Association Journal, 2002, 4, 131-5.	0.1	5
533	Rubella Virusâ€“Associated Granulomas in Immunocompetent Adultsâ€”Possible Implications. JAMA Dermatology, 2022, , .	2.0	5
534	Application of Molecular Analysis to Genetic Counseling in the Wiskottâ€“Aldrich Syndrome (WAS). DNA and Cell Biology, 1993, 12, 645-649.	0.9	4
535	Natural Killer Lymphoma/Leukemia: An Uncommon Pediatric Case with Indolent Course. Leukemia and Lymphoma, 2004, 45, 1687-1689.	0.6	4
536	Treatment of immunodeficiency: Long-term outcome and quality of life. Journal of Allergy and Clinical Immunology, 2008, 122, 1065-1068.	1.5	4
537	Defective and Excessive Immunities in Pediatric Diseases. Current Pharmaceutical Design, 2012, 18, 5729-5734.	0.9	4
538	The Natural History of Children with Severe Combined Immunodeficiency Disease (SCID): The First Fifty Patients of the Primary Immune Deficiency Treatment Consortium (PIDTC) Prospective Study 6901. Biology of Blood and Marrow Transplantation, 2013, 19, S161-S162.	2.0	4
539	Genotype, Phenotype and T Cell Counts at One Year Predict Survival and Long Term Immune Reconstitution after Transplantation in Severe Combined Immune Deficiency (SCID)â€”The Primary Immune Deficiency Treatment Consortium (PIDTC). Biology of Blood and Marrow Transplantation, 2017, 23, S133-S134.	2.0	4
540	HPV: CIB1 is for EVER and EVER. Journal of Experimental Medicine, 2018, 215, 2229-2231.	4.2	4

#	Article	IF	Citations
541	Lack of specific T- and B-cell clonal expansions in multiple sclerosis patients with progressive multifocal leukoencephalopathy. Scientific Reports, 2019, 9, 16605.	1.6	4
542	Updates on new monogenic inborn errors of immunity. Pediatric Allergy and Immunology, 2020, 31, 57-59.	1.1	4
543	International Retrospective Study of Allogeneic Hematopoietic Cell Transplantation (HCT) for Activated Phosphoinositide 3-Kinase Delta (PI3K) Syndrome. Biology of Blood and Marrow Transplantation, 2020, 26, S14-S15.	2.0	4
544	Skewed TCR Alpha, but not Beta, Gene Rearrangements and Lymphoma Associated with a Pathogenic TRAC Variant. Journal of Clinical Immunology, 2021, 41, 1395-1399.	2.0	4
545	Carrier detection in X-linked adrenoleukodystrophy by determination of very long chain fatty acid levels and by linkage analysis. European Journal of Pediatrics, 1992, 151, 761-763.	1.3	3
546	An exemplum of XLA. Clinical Immunology, 2008, 126, 137-139.	1.4	3
547	Into the wildâ€"The use and abuse of stem cells in clinical practice. American Journal of Hematology, 2013, 88, 447-448.	2.0	3
548	In Wiskott-Aldrich syndrome, platelet count matters. Blood, 2013, 121, 1484-1485.	0.6	3
549	Poor T Cell Reconstitution at 100 Days after T Cell-Replete Hematopoietic Cell Transplantation (HCT) for SCID Is Associated with Later Risk of Death or Need for 2nd Transplant in the 6901 Prospective Study of the Pidtc. Biology of Blood and Marrow Transplantation, 2016, 22, S101-S102.	2.0	3
550	Cadherin 17 mutation associated with leaky severe combined immune deficiency is corrected by HSCT. Blood Advances, 2017, 1, 2083-2087.	2.5	3
551	Adult-Onset Myopathy in a Patient with Hypomorphic RAG2 Mutations and Combined Immune Deficiency. Journal of Clinical Immunology, 2018, 38, 642-645.	2.0	3
552	Phosphate Transporter Profiles in Murine and Human Thymi Identify Thymocytes at Distinct Stages of Differentiation. Frontiers in Immunology, 2020, 11, 1562.	2.2	3
553	Transplantation Outcomes for Children with Severe Combined Immune Deficiency (SCID) Have Improved over Time: A 36-Year Summary Report By the Primary Immune Deficiency Treatment Consortium (PIDTC). Biology of Blood and Marrow Transplantation, 2020, 26, S18-S19.	2.0	3
554	Reduction in the rate and improvement in the prognosis of COVID-19 in haematological patients over time. Leukemia, 2021, 35, 632-634.	3.3	3
555	IFNÎ³R1 deficiency presenting with visceral leishmaniasis and Mycobacterium Avium infections mimicking HLH. Pediatric Allergy and Immunology, 2022, 33, .	1.1	3
556	X-linked lymphoproliferative disease: the dark side of 2b4 function. Advances in Experimental Medicine and Biology, 2001, 495, 63-67.	0.8	3
557	Somatic Gene Therapy for X-Linked Severe Combined Immunodeficiency Using a Self-Inactivating Modified Gammaretroviral Vector Results in An Improved Preclinical Safety Profile and Early Clinical Efficacy in a Human Patient. Blood, 2011, 118, 164-164.	0.6	3
558	In-Utero Transplantation of Parental CD34 Haematopoietic Progenitor Cells in a Patient With X-Linked Severe Combined Immunodeficiency (SCIDXI). Obstetrical and Gynecological Survey, 1997, 52, 404-405.	0.2	3

#	Article	IF	Citations
559	Combined T- and B-Cell Immunodeficiencies. , 2017, , 83-182.		3
560	Expansion of large granular lymphocyte subsets in Wiskott-Aldrich syndrome. Haematologica, 1995, 80, 521-5.	1.7	3
561	Granulocyte Transfusions in Patients with Chronic Granulomatous Disease Undergoing Hematopoietic Cell Transplantation or Gene Therapy. Journal of Clinical Immunology, 2022, 42, 1026-1035.	2.0	3
562	Purine nucleoside phosphorylase deficiency induces p53-mediated intrinsic apoptosis in human induced pluripotent stem cell-derived neurons. Scientific Reports, 2022, 12, .	1.6	3
563	A Girl with Diabetes and Severe Combined Immunodeficiency from Adenosine Deaminase Deficiency. Journal of Pediatric Endocrinology and Metabolism, 1997, 10, 425-8.	0.4	2
564	Prenatal diagnosis of X-linked hyper-IGM syndrome by direct detection of mutation Q220X in the CD40L gene using PCR-mediated site directed mutagenesis. Prenatal Diagnosis, 2000, 20, 822-823.	1.1	2
565	Correcting CGD safely, iPSo facto. Blood, 2011, 117, 5554-5556.	0.6	2
566	Early Hematopoietic Cell Transplant (HCT) Outcomes of Children with Severe Combined Immunodeficiency Disease (SCID): The First Seventy Four Patients of the Primary Immune Deficiency Treatment Consortium (PIDTC) Prospective Study 6901. Biology of Blood and Marrow Transplantation, 2015, 21, S289-S291.	2.0	2
567	Friendly fire: anti-cytokine antibodies elicited by microbes. Nature Medicine, 2016, 22, 973-975.	15.2	2
568	Transplant for NEMO: this and much, much more. Blood, 2017, 130, 1391-1393.	0.6	2
569	Immunodeficiencies. , 2017, , 705-722.e2.		2
570	Congenital Disorders of Lymphocyte Function. , 2018, , 710-723.e3.		2
571	From Natural Killer Cell Receptor Discovery to Characterization of Natural Killer Cell Defects in Primary Immunodeficiencies. Frontiers in Immunology, 2019, 10, 1757.	2.2	2
572	Spatiotemporal Gradient of Cortical Neuron Death Contributes to Microcephaly in Knock-In Mouse Model of Ligase 4 Syndrome. American Journal of Pathology, 2019, 189, 2440-2449.	1.9	2
573	Invasive and Allergic Complications Due to Aspergillus fumigatus in Allogeneic Hematopoietic Cell Transplantation (HCT) Primary Immunodeficiency (PID) Patients. Biology of Blood and Marrow Transplantation, 2019, 25, S356.	2.0	2
574	Opinion and Special Articles: Cerebellar ataxia and liver failure complicating IPEX syndrome. Neurology, 2020, 96, 10.1212/WNL.0000000000011195.	1.5	2
575	Targeted pharmacologic immunomodulation for inborn errors of immunity. British Journal of Clinical Pharmacology, 2020, , .	1.1	2
576	Novel Compound Heterozygous Mutations in ZAP70 Leading to a SCID Phenotype with Normal Downstream In vitro Signaling. Journal of Clinical Immunology, 2021, 41, 470-472.	2.0	2

#	Article	IF	Citations
577	Infectious Complications Predict Premature CD8+ T-cell Senescence in CD40 Ligand-Deficient Patients. Journal of Clinical Immunology, 2021, 41, 795-806.	2.0	2
578	Clinical Manifestations, Mutational Analysis, and Immunological Phenotype in Patients with RAG1/2 Mutations: First Cases Series from Mexico and Description of Two Novel Mutations. Journal of Clinical Immunology, 2021, 41, 1291-1302.	2.0	2
579	Successful Therapy of a Patient with a Novel STAT1 Gain of Function Mutation and Life-Threatening Cytopenias with Janus Kinase Inhibitor Ruxolitinib. Blood, 2015, 126, 3434-3434.	0.6	2
580	Congenital and acquired defects of immunity: An everâ€evolving story. Pediatric Allergy and Immunology, 2022, 33, 61-64.	1.1	2
581	Primary Immunodeficiencies: Milestones in the History of Pediatric Immunology. Pediatric Hematology and Oncology, 1991, 8, 203-214.	0.3	1
582	Ureteral obstruction in a patient with chronic granulomatous disease, receiving combined prophylaxis with IFN-Î³ and antibiotics. European Journal of Pediatrics, 1998, 157, 352-353.	1.3	1
583	Mechanisms of primary immunodeficiencies: from bed-side to bench and back. Drug Discovery Today Disease Mechanisms, 2004, 1, 383-390.	0.8	1
584	Gene therapy sculpts the bone. Blood, 2007, 109, 5067-5068.	0.6	1
585	Reply. Journal of Allergy and Clinical Immunology, 2009, 123, 1419-1420.	1.5	1
586	Severe Combined Immunodeficiencies. , 2014, , 87-141.		1
587	From Immunodeficiency to Autoimmunity. , 2014, , 41-49.		1
588	PRKDC mutations associated with immunodeficiency, granuloma and aire-dependent autoimmunity. Pediatric Rheumatology, 2014, 12, .	0.9	1
589	Retrospective Study of 240 Patients with Severe Combined Immunodeficiency Transplanted from 2000-2009: A Report from the Primary Immune Deficiency Treatment Consortium of North America. Biology of Blood and Marrow Transplantation, 2014, 20, S24-S25.	2.0	1
590	A119: Deep Sequencing Analysis of the T Regulatory and T Effector Repertoire in Juvenile Idiopathic Arthritis. Arthritis and Rheumatology, 2014, 66, S156-S156.	2.9	1
591	Successful Hematopoietic Stem Cell Transplant For CD40 Deficiency Manifesting As Hyper-IgM Syndrome With Absent CD40 Expression and Marked Lymphocytosis. Journal of Allergy and Clinical Immunology, 2014, 133, AB95.	1.5	1
592	A Case of Leaky SCID with Variable Presentation in Two Siblings Identified By Newborn Screening. Journal of Allergy and Clinical Immunology, 2015, 135, AB15.	1.5	1
593	Human Genetic Defects Resulting in Increased Susceptibility to Viral Infections. , 2016, , 375-388.		1
594	O-008â€ƒAberrant Anti-inflammatory Macrophage Function and Differentiation in Wiskott-Aldrich Syndrome Protein-Deficient Mice and Humans. Inflammatory Bowel Diseases, 2016, 22, S3.	0.9	1

#	Article	IF	Citations
595	T Cell Immunodeficiencies. , 2016, , 80-89.e4.		1
596	Low T-Cell Receptor Î² ( ) Repertoire Diversity Early Post-Transplant for Severe Combined Immunodeficiency (SCID) Predicts Subsequent Failure of Immune Reconstitution. Biology of Blood and Marrow Transplantation, 2020, 26, S306.	2.0	1
597	Complete Absence of CD3Î³ Protein Expression Is Responsible for Combined Immunodeficiency with Autoimmunity Rather than SCID. Journal of Clinical Immunology, 2021, 41, 482-485.	2.0	1
598	Development of Autologous, Oligoclonal, Poorly Functioning T Lymphocytes in a Patient With Autosomal Recessive Severe Combined Immunodeficiency Caused by Defects of the Jak3 Tyrosine Kinase. Blood, 1998, 91, 949-955.	0.6	1
599	Primary immune deficiencies unravel the molecular basis of immune response. Reviews in Clinical and Experimental Hematology, 2003, 7, 84-111.	0.1	1
600	Polymorphonuclear function and respiratory syncytial virus infections in children. Infection, 1983, 11, 232-233.	2.3	0
601	Combined immunodeficiency phenotype associated with inappropriate spontaneous and activation-induced apoptosis. Clinical and Experimental Immunology, 1997, 108, 484-489.	1.1	0
602	Report of the ESID collaborative study on clinical features and molecular analysis in X-linked hyper-IgM syndrome. Molecular Immunology, 1998, 35, 665.	1.0	0
603	OR.1. Hypomorphic Rag1 and Lig4 Mutants are a Model for Human Leaky SCID. Clinical Immunology, 2008, 127, S4.	1.4	0
604	OR.2. Adoptive Transfer of Hypomorphic Rag2 Mutant Fetal Liver Cells in Reconstituted Thymic Architecture Can Prevent Peripheral Immunopathology. Clinical Immunology, 2008, 127, S4-S5.	1.4	0
605	F.65. The Wiskott-Aldrich Syndrome Protein is Essential for Development and Correct Function of the Marginal Zone. Clinical Immunology, 2008, 127, S64.	1.4	0
606	OR.88. Identification of a Subset of AIRE Expressing Dendritic Cell in Human Peripheral Lymphoid Tissues: Evidences for a Role in the Induction of Peripheral Tolerance. Clinical Immunology, 2009, 131, S36-S37.	1.4	0
607	F.121. B Cell-mediated Autoimmunity in Hypomorphic rag1 and lig4 Mouse Mutants as Models for Human Leaky SCID. Clinical Immunology, 2009, 131, S126.	1.4	0
608	Josiah F. Wedgwood (1950-2009). Journal of Allergy and Clinical Immunology, 2010, 125, 506.	1.5	0
609	T Cell Immunodeficiencies. , 2010, , 98-109.		0
610	Regulatory T-like cells: Another variation on the theme of hypomorphic mutations?. Clinical Immunology, 2011, 141, 242-243.	1.4	0
611	Immunodeficiency Disorders. , 2013, , 1-30.		0
612	Immunodeficiency Disorders. , 2014, , .		0

#	Article	IF	Citations
613	Evaluation of a Novel Missense Activation-Induced Deaminase AID Mutation in a Child with Hyper IgM Syndrome: Is it a Pathogenic Mutation?. Journal of Allergy and Clinical Immunology, 2014, 133, AB70.	1.5	0
614	A Novel ORAI1 Mutation Resulting In T+B+NK+ SCID With Normal Lymphocyte Proliferation. Journal of Allergy and Clinical Immunology, 2014, 133, AB94.	1.5	0
615	Hematopoietic Stem Cell Therapy. , 2016, , 152-159.e3.		0
616	An Atypical Severe Combined Immunodeficiency (SCID) Case Diagnosis Complicated by Alternative Care in the Era of Newborn Screening (NBS) for SCID. Journal of Allergy and Clinical Immunology, 2017, 139, AB18.	1.5	0
617	Generation of human induced pluripotent stem cell lines from patients with selective IgA deficiency. Stem Cell Research, 2019, 41, 101613.	0.3	0
618	Successful Allogeneic Hematopoietic Cell Transplantation (HCT) with Low Toxicity and Gvhd in a Heterogeneous Cohort of Primary Immunodeficiency (PID) Patients. Biology of Blood and Marrow Transplantation, 2019, 25, S42-S43.	2.0	0
619	Immune Reconstitution Therapy for Immunodeficiency. , 2019, , 1115-1128.e1.		0
620	Novel Missense Mutation inSP110Associated with Combined Immunodeficiency and Advanced Liver Disease Without VOD. Journal of Clinical Immunology, 2020, 40, 236-239.	2.0	0
621	Are we diagnosing too late? RAG deficiency in young adults with end organ damage. Journal of Allergy and Clinical Immunology, 2021, 147, AB69.	1.5	0
622	Jak3 and the Pathogenesis of Severe Combined Immunodeficiency. , 2003, , 623-636.		0
623	Novel Munc13-4 Mutations in Patients with Hemophagocytic Lymphohistiocytosis.. Blood, 2005, 106, 2807-2807.	0.6	0
624	MUNC13â€“4 Mutations in Patients with Hemophagocytic Lymphohistiocytosis Are Scattered over the Functional Domains of the Protein.. Blood, 2006, 108, 1249-1249.	0.6	0
625	Stem cell transplantation and immune reconstitution in immunodeficiency. , 2008, , 1237-1251.		0
626	Immunodeficiencies. , 2010, , 785-803.		0
627	T-Cell Development. , 2011, , 1596-1603.		0
628	T-Cell Immune Defects. , 2012, , 1297-1306.		0
629	Induced Pluripotent Stem Cells From a Patient with Reticular Dysgenesis Recapitulate Defective Myelopoiesis in-Vitro: A Disease Model to Enhance Our Understanding of a Rare Disease.. Blood, 2012, 120, 2142-2142.	0.6	0
630	Stem cell transplantation and immune reconstitution in immunodeficiency. , 2013, , 1007-1019.		0

#	Article	IF	Citations
631	Wiskott-Aldrich syndrome proteinâ€“mediated actin dynamics control type-I interferon production in plasmacytoid dendritic cells. Journal of Cell Biology, 2013, 200, i6-i6.	2.3	0
632	Reticular dysgenesisâ€“associated AK2 protects hematopoietic stem and progenitor cell development from oxidative stress. Journal of Cell Biology, 2015, 210, 2102OIA141.	2.3	0
633	2011 â€“ MOUSE EMBRYONIC LYMPHOCYTES CONTRIBUTE TO AUTOIMMUNITY. Experimental Hematology, 2020, 88, S31.	0.2	0
634	In Memoriamâ€”Thomas Alexander Waldmann, M.D.. Journal of Clinical Immunology, 2021, , 1.	2.0	0
635	Allogeneic Hematopoietic Stem-Cell Transplantation in Patients with GATA 2 Deficiency: Influence of Donor Stem Cell Source and Post-Transplantation Cyclophosphamide. Blood, 2020, 136, 37-38.	0.6	0
636	Impaired Sars-Cov-2 Specific Antibody Responses in Patients Treated with Anti-CD20 Antibodies. Blood, 2020, 136, 47-48.	0.6	0
637	Longitudinal Serological Response to Sars-COV-2 in Patients Affected By Hematologic Diseases. Blood, 2020, 136, 4-4.	0.6	0
638	Morbidity, Mortality, and Therapeutics in Combined Immunodeficiency: Data from the USIDNET Registry. Journal of Allergy and Clinical Immunology: in Practice, 2022, , .	2.0	0
639	Hematopoietic Cell Transplantation in 240 Patients with Chronic Granulomatous Disease: A Pidtc Report. Transplantation and Cellular Therapy, 2022, 28, S101-S103.	0.6	0
640	A Primary Immune Deficiency Treatment Consortium (PIDTC) Study of Chronic and Late Onset Medical Complications after Initial Hematopoietic Cell Transplantation (HCT) for Severe Combined Immunodeficiency Disease (SCID). Transplantation and Cellular Therapy, 2022, 28, S339-S340.	0.6	0
641	OP019: A tertiary care clinical sequencing program for patients with suspected immune defects: Results from the first 1000 families. Genetics in Medicine, 2022, 24, S350.	1.1	0
642	Sars-Cov-2 Infection and Vaccination in Recipients of Reduced-Intensity Conditioning, Posttransplantation Cyclophosphamide (PTCy)-Based Allogeneic Hematopoietic Cell Transplantation (HCT). Transplantation and Cellular Therapy, 2022, 28, S350-S351.	0.6	0
643	Outcomes after Hematopoietic Cell Transplant and Gene Therapy for Adenosine Deaminase (ADA) Severe Combined Immune Deficiency: A Combined Analysis from the Primary Immune Deficiency Treatment Consortium (PIDTC) 6901 and 6902 Studies. Transplantation and Cellular Therapy, 2022, 28, S100-S101.	0.6	0