

Tomohiro Morio

List of Publications by Year in descending order

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338
papers

14,375
citations

30047

54
h-index

26591

107
g-index

360
all docs

360
docs citations

360
times ranked

16626
citing authors

#	ARTICLE	IF	CITATIONS
1	Human Inborn Errors of Immunity: 2019 Update on the Classification from the International Union of Immunological Societies Expert Committee. <i>Journal of Clinical Immunology</i> , 2020, 40, 24-64.	2.0	881
2	International Union of Immunological Societies: 2017 Primary Immunodeficiency Diseases Committee Report on Inborn Errors of Immunity. <i>Journal of Clinical Immunology</i> , 2018, 38, 96-128.	2.0	732
3	Human Tyrosine Kinase 2 Deficiency Reveals Its Requisite Roles in Multiple Cytokine Signals Involved in Innate and Acquired Immunity. <i>Immunity</i> , 2006, 25, 745-755.	6.6	601
4	Human Inborn Errors of Immunity: 2019 Update of the IUIS Phenotypic Classification. <i>Journal of Clinical Immunology</i> , 2020, 40, 66-81.	2.0	525
5	The 2017 IUIS Phenotypic Classification for Primary Immunodeficiencies. <i>Journal of Clinical Immunology</i> , 2018, 38, 129-143.	2.0	488
6	Heterozygous STAT1 gain-of-function mutations underlie an unexpectedly broad clinical phenotype. <i>Blood</i> , 2016, 127, 3154-3164.	0.6	465
7	Human Inborn Errors of Immunity: 2022 Update on the Classification from the International Union of Immunological Societies Expert Committee. <i>Journal of Clinical Immunology</i> , 2022, 42, 1473-1507.	2.0	389
8	IL23R regulates TH17 development by cooperating with ROR nuclear receptors. <i>Nature</i> , 2010, 464, 1381-1385.	13.7	361
9	Autoantibodies neutralizing type I IFNs are present in ~4% of uninfected individuals over 70 years old and account for ~20% of COVID-19 deaths. <i>Science Immunology</i> , 2021, 6, .	5.6	357
10	Clinical course of patients with WASP gene mutations. <i>Blood</i> , 2004, 103, 456-464.	0.6	320
11	Tyrosine Kinases Btk and Tec Regulate Osteoclast Differentiation by Linking RANK and ITAM Signals. <i>Cell</i> , 2008, 132, 794-806.	13.5	297
12	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. <i>Science Immunology</i> , 2021, 6, .	5.6	267
13	The extended phenotype of LPS-responsive beige-like anchor protein (LRBA) deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 137, 223-230.	1.5	247
14	Proposed guidelines for diagnosing chronic active Epstein-Barr virus infection. <i>American Journal of Hematology</i> , 2005, 80, 64-69.	2.0	246
15	BCG vaccination in patients with severe combined immunodeficiency: Complications, risks, and vaccination policies. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 133, 1134-1141.	1.5	212
16	Prognostic Factors for Chronic Active Epstein-Barr Virus Infection. <i>Journal of Infectious Diseases</i> , 2003, 187, 527-533.	1.9	207
17	Hematopoietic stem cell-engrafted NOD/SCID/IL2R β null mice develop human lymphoid systems and induce long-lasting HIV-1 infection with specific humoral immune responses. <i>Blood</i> , 2007, 109, 212-218.	0.6	196
18	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. <i>Cell</i> , 2020, 181, 1194-1199.	13.5	185

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19	X-linked thrombocytopenia (XLT) due to WAS mutations: clinical characteristics, long-term outcome, and treatment options. <i>Blood</i> , 2010, 115, 3231-3238.	0.6	178
20	The Ever-Increasing Array of Novel Inborn Errors of Immunity: an Interim Update by the IUIS Committee. <i>Journal of Clinical Immunology</i> , 2021, 41, 666-679.	2.0	165
21	Impaired CD4 and CD8 Effector Function and Decreased Memory T Cell Populations in ICOS-Deficient Patients. <i>Journal of Immunology</i> , 2009, 182, 5515-5527.	0.4	139
22	Use of a Comprehensive Polymerase Chain Reaction System for Diagnosis of Ocular Infectious Diseases. <i>Ophthalmology</i> , 2013, 120, 1761-1768.	2.5	130
23	Hematopoietic stem cell transplantation in patients with gain-of-function signal transducer and activator of transcription 1 mutations. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 141, 704-717.e5.	1.5	128
24	Autoimmune lymphoproliferative syndrome-like disease with somatic KRAS mutation. <i>Blood</i> , 2011, 117, 2887-2890.	0.6	123
25	Molecular and Virological Evidence of Viral Activation From Chromosomally Integrated Human Herpesvirus 6A in a Patient With X-Linked Severe Combined Immunodeficiency. <i>Clinical Infectious Diseases</i> , 2014, 59, 545-548.	2.9	121
26	Analysis of class switch recombination and somatic hypermutation in patients affected with autosomal dominant hyper-IgM syndrome type 2. <i>Clinical Immunology</i> , 2005, 115, 277-285.	1.4	111
27	Common cytological and cytogenetic features of Epstein-Barr virus (EBV)-positive natural killer (NK) cells and cell lines derived from patients with nasal T/NK-cell lymphomas, chronic active EBV infection and hydroa vacciniforme-like eruptions. <i>British Journal of Haematology</i> , 2003, 121, 805-814.	1.2	110
28	The risk of COVID-19 death is much greater and age dependent with type I IFN autoantibodies. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, e2200413119.	3.3	110
29	Identification of Severe Combined Immunodeficiency by T-Cell Receptor Excision Circles Quantification Using Neonatal Guthrie Cards. <i>Journal of Pediatrics</i> , 2009, 155, 829-833.	0.9	108
30	Chemokine receptor expression and functional effects of chemokines on B cells: implication in the pathogenesis of rheumatoid arthritis. <i>Arthritis Research and Therapy</i> , 2009, 11, R149.	1.6	102
31	The kinase Btk negatively regulates the production of reactive oxygen species and stimulation-induced apoptosis in human neutrophils. <i>Nature Immunology</i> , 2012, 13, 369-378.	7.0	100
32	Haploinsufficiency of A20 causes autoinflammatory and autoimmune disorders. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 141, 1485-1488.e11.	1.5	100
33	Abnormal hematopoiesis and autoimmunity in human subjects with germline IKZF1 mutations. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 140, 223-231.	1.5	99
34	Flow cytometry-based diagnosis of primary immunodeficiency diseases. <i>Allergology International</i> , 2018, 67, 43-54.	1.4	97
35	Oxidative Stress Induces Nuclear Loss of DNA Repair Proteins Ku70 and Ku80 and Apoptosis in Pancreatic Acinar AR42J Cells. <i>Journal of Biological Chemistry</i> , 2003, 278, 36676-36687.	1.6	93
36	Haploinsufficiency of TNFAIP3 (A20) by germline mutation is involved in autoimmune lymphoproliferative syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 139, 1914-1922.	1.5	91

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37	Macrophage Activation Syndrome in Patients with Systemic Juvenile Idiopathic Arthritis under Treatment with Tocilizumab. <i>Journal of Rheumatology</i> , 2015, 42, 712-722.	1.0	90
38	Outcome in patients with Wiskott?Aldrich syndrome following stem cell transplantation: an analysis of 57 patients in Japan. <i>British Journal of Haematology</i> , 2006, 135, 362-366.	1.2	89
39	Tocilizumab in systemic juvenile idiopathic arthritis in a real-world clinical setting: results from 1 year of postmarketing surveillance follow-up of 417 patients in Japan. <i>Annals of the Rheumatic Diseases</i> , 2016, 75, 1654-1660.	0.5	89
40	Phosphatase and tensin homolog (PTEN) mutation can cause activated phosphatidylinositol 3-kinase Î syndrome-like immunodeficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 1672-1680.e10.	1.5	87
41	A significant association of viral loads with corneal endothelial cell damage in cytomegalovirus anterior uveitis. <i>British Journal of Ophthalmology</i> , 2010, 94, 336-340.	2.1	83
42	In vitro generation of functional murine heart organoids via FGF4 and extracellular matrix. <i>Nature Communications</i> , 2020, 11, 4283.	5.8	80
43	Nationwide Survey of Patients with Primary Immunodeficiency Diseases in Japan. <i>Journal of Clinical Immunology</i> , 2011, 31, 968-976.	2.0	77
44	Simple diagnosis of STAT1 gain-of-function alleles in patients with chronic mucocutaneous candidiasis. <i>Journal of Leukocyte Biology</i> , 2013, 95, 667-676.	1.5	77
45	Ataxia-telangiectasia: Immunodeficiency and survival. <i>Clinical Immunology</i> , 2017, 178, 45-55.	1.4	72
46	Current research on chronic active Epstein-Barr virus infection in Japan. <i>Pediatrics International</i> , 2014, 56, 159-166.	0.2	71
47	Gain-of-function IKKB mutation causes human combined immune deficiency. <i>Journal of Experimental Medicine</i> , 2018, 215, 2715-2724.	4.2	69
48	Therapeutic options for CTLA-4 insufficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2022, 149, 736-746.	1.5	68
49	Immunologically silent cancer clone transmission from mother to offspring. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009, 106, 17882-17885.	3.3	65
50	NADPH oxidase and apoptosis in cerulein-stimulated pancreatic acinar AR42J cells. <i>Free Radical Biology and Medicine</i> , 2005, 39, 590-602.	1.3	64
51	Hematopoietic Stem Cell Transplantation for XIAP Deficiency in Japan. <i>Journal of Clinical Immunology</i> , 2017, 37, 85-91.	2.0	63
52	Ku in the Cytoplasm Associates with CD40 in Human B Cells and Translocates into the Nucleus following Incubation with IL-4 and Anti-CD40 mAb. <i>Immunity</i> , 1999, 11, 339-348.	6.6	61
53	Recessive inborn errors of type I IFN immunity in children with COVID-19 pneumonia. <i>Journal of Experimental Medicine</i> , 2022, 219, .	4.2	59
54	Engagement of MHC class II molecules by staphylococcal superantigens activates src-type protein tyrosine kinases. <i>European Journal of Immunology</i> , 1994, 24, 651-658.	1.6	58

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55	Two Novel Gain-of-Function Mutations of <i>STAT1</i> Responsible for Chronic Mucocutaneous Candidiasis Disease: Impaired Production of IL-17A and IL-22, and the Presence of Anti-IL-17F Autoantibody. <i>Journal of Immunology</i> , 2014, 193, 4880-4887.	0.4	58
56	14 Years after Discovery: Clinical Follow-up on 15 Patients with Inducible Co-Stimulator Deficiency. <i>Frontiers in Immunology</i> , 2017, 8, 964.	2.2	57
57	Immunosuppressive effects of tautomycetin in vivo and in vitro via T cell-specific apoptosis induction. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2002, 99, 10617-10622.	3.3	56
58	Outcome of unrelated umbilical cord blood transplantation in 88 patients with primary immunodeficiency in Japan. <i>British Journal of Haematology</i> , 2011, 154, 363-372.	1.2	56
59	Two Brothers with Ataxia-Telangiectasia-like Disorder with Lung Adenocarcinoma. <i>Journal of Pediatrics</i> , 2009, 155, 435-438.	0.9	55
60	Lycopene inhibits <i>Helicobacter pylori</i> -induced ATM/ATR-dependent DNA damage response in gastric epithelial AGS cells. <i>Free Radical Biology and Medicine</i> , 2012, 52, 607-615.	1.3	52
61	Common variable immunodeficiency classification by quantifying T-cell receptor and immunoglobulin δ -deleting recombination excision circles. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 131, 1437-1440.e5.	1.5	52
62	Extensive gene deletions in Japanese patients with Diamond-Blackfan anemia. <i>Blood</i> , 2012, 119, 2376-2384.	0.6	49
63	Hematopoietic stem cell transplantation for progressive combined immunodeficiency and lymphoproliferation in patients with activated phosphatidylinositol-3-OH kinase δ syndrome type 1. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 266-275.	1.5	49
64	Clinical features and hematopoietic stem cell transplantations for CD40 ligand deficiency in Japan. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 136, 1018-1024.	1.5	48
65	Novel Mouse Xenograft Models Reveal a Critical Role of CD4+ T Cells in the Proliferation of EBV-Infected T and NK Cells. <i>PLoS Pathogens</i> , 2011, 7, e1002326.	2.1	46
66	Defects in autophagosome-lysosome fusion underlie Vici syndrome, a neurodevelopmental disorder with multisystem involvement. <i>Scientific Reports</i> , 2017, 7, 3552.	1.6	46
67	Ataxia-telangiectasia-mutated dependent phosphorylation of Artemis in response to DNA damage. <i>Cancer Science</i> , 2005, 96, 134-141.	1.7	45
68	RAG1 Deficiency May Present Clinically as Selective IgA Deficiency. <i>Journal of Clinical Immunology</i> , 2015, 35, 280-288.	2.0	45
69	FOXL2 transcriptionally represses <i>Sf1</i> expression by antagonizing WT1 during ovarian development in mice. <i>FASEB Journal</i> , 2014, 28, 2020-2028.	0.2	44
70	Preclinical evaluation of NUDT15-guided thiopurine therapy and its effects on toxicity and antileukemic efficacy. <i>Blood</i> , 2018, 131, 2466-2474.	0.6	43
71	Wiskott-Aldrich syndrome presenting with a clinical picture mimicking juvenile myelomonocytic leukaemia. <i>Pediatric Blood and Cancer</i> , 2013, 60, 836-841.	0.8	42
72	Multicolor Flow Cytometry for the Diagnosis of Primary Immunodeficiency Diseases. <i>Journal of Clinical Immunology</i> , 2017, 37, 486-495.	2.0	42

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73	Allogeneic hematopoietic stem cell transplantation for seven children with X-linked hyper-IgM syndrome: A single center experience. <i>American Journal of Hematology</i> , 2004, 76, 33-39.	2.0	41
74	Functional characterization and targeted correction of ATM mutations identified in Japanese patients with ataxia-telangiectasia. <i>Human Mutation</i> , 2012, 33, 198-208.	1.1	39
75	Newborn screening for congenital adrenal hyperplasia in Tokyo, Japan from 1989 to 2013: a retrospective population-based study. <i>BMC Pediatrics</i> , 2015, 15, 209.	0.7	39
76	TALEN-Mediated Gene Disruption on Y Chromosome Reveals Critical Role of EIF2S3Y in Mouse Spermatogenesis. <i>Stem Cells and Development</i> , 2015, 24, 1164-1170.	1.1	39
77	Extracellular ADP augments microglial inflammasome and NF- κ B activation via the P2Y12 receptor. <i>European Journal of Immunology</i> , 2020, 50, 205-219.	1.6	38
78	The Potential and Limits of Hematopoietic Stem Cell Transplantation for the Treatment of Autosomal Dominant Hyper-IgE Syndrome. <i>Journal of Clinical Immunology</i> , 2016, 36, 511-516.	2.0	37
79	Heterozygous <i>OAS1</i> gain-of-function variants cause an autoinflammatory immunodeficiency. <i>Science Immunology</i> , 2021, 6, .	5.6	36
80	Vaccine breakthrough hypoxemic COVID-19 pneumonia in patients with auto-Abs neutralizing type I IFNs. <i>Science Immunology</i> , 2023, 8, .	5.6	35
81	WASP is involved in proliferation and differentiation of human haemopoietic progenitors in vitro. <i>British Journal of Haematology</i> , 1999, 107, 254-262.	1.2	33
82	Omenn Syndrome—Review of Several Phenotypes of Omenn Syndrome and RAG1/RAG2 Mutations in Japan. <i>Allergology International</i> , 2006, 55, 115-119.	1.4	33
83	ROR γ -specific transcriptional interactomic inhibition suppresses autoimmunity associated with T _H 17 cells. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 18673-18678.	3.3	33
84	APRIL-dependent lifelong plasmacyte maintenance and immunoglobulin production in humans. <i>Journal of Allergy and Clinical Immunology</i> , 2020, 146, 1109-1120.e4.	1.5	33
85	A variant in human AIOLOS impairs adaptive immunity by interfering with IKAROS. <i>Nature Immunology</i> , 2021, 22, 893-903.	7.0	33
86	Novel adopted immunotherapy for mixed chimerism after unrelated cord blood transplantation in Omenn syndrome. <i>European Journal of Haematology</i> , 2005, 75, 441-444.	1.1	32
87	Identification of Human Herpesvirus 6 in a Patient With Severe Unilateral Panuveitis. <i>JAMA Ophthalmology</i> , 2007, 125, 1426.	2.6	32
88	Anti-nuclear matrix protein 2 antibody-positive inflammatory myopathies represent extensive myositis without dermatomyositis-specific rash. <i>Rheumatology</i> , 2022, 61, 1222-1227.	0.9	32
89	Quantitative PCR for the detection of genomic DNA of Epstein-Barr virus in ocular fluids of patients with uveitis. <i>Japanese Journal of Ophthalmology</i> , 2008, 52, 463-467.	0.9	31
90	Phenotypic variations between affected siblings with ataxia-telangiectasia: ataxia-telangiectasia in Japan. <i>International Journal of Hematology</i> , 2009, 90, 455-462.	0.7	31

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91	The Pleckstrin Homology Domain of the Wiskott-Aldrich Syndrome Protein Is Involved in the Organization of Actin Cytoskeleton. <i>Clinical Immunology</i> , 1999, 92, 128-137.	1.4	30
92	Successful allogeneic hematopoietic stem cell transplantation for chronic granulomatous disease with inflammatory complications and severe infection. <i>International Journal of Hematology</i> , 2011, 94, 479-482.	0.7	30
93	Detailed analysis of Japanese patients with adenosine deaminase 2 deficiency reveals characteristic elevation of type II interferon signature and STAT1 hyperactivation. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 148, 550-562.	1.5	30
94	Ku, Artemis, and ataxia-telangiectasia-mutated: Signalling networks in DNA damage. <i>International Journal of Biochemistry and Cell Biology</i> , 2008, 40, 598-603.	1.2	29
95	Alleviation of rheumatoid arthritis by cell-transducible methotrexate upon transcutaneous delivery. <i>Biomaterials</i> , 2012, 33, 1563-1572.	5.7	29
96	Risks and prevention of severe RS virus infection among children with immunodeficiency and Down's syndrome. <i>Journal of Infection and Chemotherapy</i> , 2014, 20, 455-459.	0.8	29
97	Novel compound heterozygous DNA ligase IV mutations in an adolescent with a slowly-progressing radiosensitive-severe combined immunodeficiency. <i>Clinical Immunology</i> , 2015, 160, 255-260.	1.4	29
98	A Patient with CTLA-4 Haploinsufficiency Presenting Gastric Cancer. <i>Journal of Clinical Immunology</i> , 2016, 36, 28-32.	2.0	29
99	Neuroprotective effects of human umbilical cord-derived mesenchymal stem cells on periventricular leukomalacia-like brain injury in neonatal rats. <i>Inflammation and Regeneration</i> , 2017, 37, 1.	1.5	29
100	Clinical and Immunological Characterization of ICF Syndrome in Japan. <i>Journal of Clinical Immunology</i> , 2018, 38, 927-937.	2.0	29
101	The increase of non-MHC-restricted cytotoxic cells (T cells or NK cells) and the abnormal differentiation of B cells in Wiskott-Aldrich syndrome. <i>Clinical Immunology and Immunopathology</i> , 1989, 52, 279-290.	2.1	28
102	Preferential Expansion of V α 39-J β 3P/V α 2-J β 3 T Cells in Nasal T-Cell Lymphoma and Chronic Active Epstein-Barr Virus Infection. <i>American Journal of Pathology</i> , 2003, 162, 1629-1638.	1.9	28
103	Successful cord blood transplantation for a CHARGE syndrome with CHD7 mutation showing DiGeorge sequence including hypoparathyroidism. <i>European Journal of Pediatrics</i> , 2010, 169, 839-844.	1.3	28
104	Robust and highly efficient hiPSC generation from patient non-mobilized peripheral blood-derived CD34+ cells using the auto-erasable Sendai virus vector. <i>Stem Cell Research and Therapy</i> , 2019, 10, 185.	2.4	28
105	Autopsy study of cerebellar degeneration in siblings with ataxia-telangiectasia-like disorder. <i>Acta Neuropathologica</i> , 2010, 119, 513-520.	3.9	27
106	Analysis of mutations and recombination activity in RAG-deficient patients. <i>Clinical Immunology</i> , 2011, 138, 172-177.	1.4	27
107	Allogeneic stem cell transplantation for X-linked agammaglobulinemia using reduced intensity conditioning as a model of the reconstitution of humoral immunity. <i>Journal of Hematology and Oncology</i> , 2016, 9, 9.	6.9	27
108	Status of KRAS in iPSCs Impacts upon Self-Renewal and Differentiation Propensity. <i>Stem Cell Reports</i> , 2018, 11, 380-394.	2.3	27

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109	Effects of Interleukin-6 and Granulocyte Colony-Stimulating Factor on the Proliferation of Leukemic Blast Progenitors from Acute Myeloblastic Leukemia Patients. <i>Japanese Journal of Cancer Research</i> , 1990, 81, 979-986.	1.7	26
110	Analysis of serum soluble CD40 ligand (sCD40L) in the patients undergoing allogeneic stem cell transplantation: platelet is a major source of serum sCD40L. <i>European Journal of Haematology</i> , 2005, 74, 54-60.	1.1	26
111	Apoptosis of macrophages induced by <i>Trichomonas vaginalis</i> through the phosphorylation of p38 mitogen-activated protein kinase that locates at downstream of mitochondria-dependent caspase activation. <i>International Journal of Biochemistry and Cell Biology</i> , 2006, 38, 638-647.	1.2	26
112	Analysis of serum granulysin in patients with hematopoietic stem-cell transplantation: Its usefulness as a marker of graft-versus-host reaction. <i>American Journal of Hematology</i> , 2006, 81, 340-348.	2.0	26
113	Process for immune defect and chromosomal translocation during early thymocyte development lacking ATM. <i>Blood</i> , 2012, 120, 789-799.	0.6	26
114	Autosomal recessive complete STAT1 deficiency caused by compound heterozygous intronic mutations. <i>International Immunology</i> , 2020, 32, 663-671.	1.8	26
115	Granulocyte Colony-stimulating Factor-dependent Growth of an Acute Myeloblastic Leukemia Cell Line. <i>Japanese Journal of Cancer Research</i> , 1990, 81, 625-631.	1.7	25
116	Am80, a retinoic acid receptor agonist, ameliorates murine vasculitis through the suppression of neutrophil migration and activation. <i>Arthritis and Rheumatism</i> , 2013, 65, 503-512.	6.7	25
117	Hematopoietic Stem Cell Transplantation for X-Linked Thrombocytopenia With Mutations in the WAS gene. <i>Journal of Clinical Immunology</i> , 2015, 35, 15-21.	2.0	25
118	CD40-CD40 ligand (CD40L) interactions and X-linked hyperIgM syndrome (HIGMX-1). <i>Clinical Immunology and Immunopathology</i> , 1995, 76, S208-S213.	2.1	23
119	Knockdown of XAB2 Enhances All-Trans Retinoic Acid-Induced Cellular Differentiation in All-Trans Retinoic Acid-Sensitive and -Resistant Cancer Cells. <i>Cancer Research</i> , 2007, 67, 1019-1029.	0.4	23
120	Qualitatively differential regulation of T cell activation and apoptosis by T cell receptor ζ chain ITAMs and their tyrosine residues. <i>International Immunology</i> , 2004, 16, 1225-1236.	1.8	22
121	Dysregulation of Epstein-Barr Virus Infection in Hypomorphic ZAP70 Mutation. <i>Journal of Infectious Diseases</i> , 2018, 218, 825-834.	1.9	22
122	Mechanisms of genotype-phenotype correlation in autosomal dominant anhidrotic ectodermal dysplasia with immune deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 141, 1060-1073.e3.	1.5	22
123	Pathogenic mutations identified by a multimodality approach in 117 Japanese Fanconi anemia patients. <i>Haematologica</i> , 2019, 104, 1962-1973.	1.7	22
124	Atypical SIFD with novel TRNT1 mutations: a case study on the pathogenesis of B-cell deficiency. <i>International Journal of Hematology</i> , 2019, 109, 382-389.	0.7	22
125	Failure of mefloquine therapy in progressive multifocal leukoencephalopathy: Report of two Japanese patients without human immunodeficiency virus infection. <i>Journal of the Neurological Sciences</i> , 2013, 324, 190-194.	0.3	20
126	Transcription activator-like effector nuclease-mediated transduction of exogenous gene into IL2RG locus. <i>Scientific Reports</i> , 2015, 4, 5043.	1.6	20

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127	Genetic analysis of undiagnosed ataxia-telangiectasia-like disorders. <i>Brain and Development</i> , 2019, 41, 150-157.	0.6	20
128	Whole-Exome Sequencing-Based Approach for Germline Mutations in Patients with Inborn Errors of Immunity. <i>Journal of Clinical Immunology</i> , 2020, 40, 729-740.	2.0	20
129	Successful Nonmyeloablative Cord Blood Transplantation for an Infant With Malignant Infantile Osteopetrosis. <i>Journal of Pediatric Hematology/Oncology</i> , 2005, 27, 495-498.	0.3	19
130	Vesical varices and telangiectasias in a patient with ataxia telangiectasia. <i>Pediatric Nephrology</i> , 2008, 23, 1005-1008.	0.9	19
131	Rapid Detection of Intracellular p47phox and p67phox by Flow Cytometry; Useful Screening Tests for Chronic Granulomatous Disease. <i>Journal of Clinical Immunology</i> , 2013, 33, 857-864.	2.0	19
132	Mutations in Bruton's tyrosine kinase impair IgA responses. <i>International Journal of Hematology</i> , 2015, 101, 305-313.	0.7	19
133	Decreased $\alpha\beta$ Heterodimer among CD8 Molecules of Peripheral Blood T Cells in Wiskott-Aldrich Syndrome. <i>Clinical Immunology and Immunopathology</i> , 1996, 81, 129-135.	2.1	18
134	Female hyper IgM syndrome type 1 with a chromosomal translocation disrupting CD40LG. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2006, 1762, 335-340.	1.8	18
135	Common Variable Immunodeficiency Caused by FANC Mutations. <i>Journal of Clinical Immunology</i> , 2017, 37, 434-444.	2.0	18
136	Genetic heterogeneity of uncharacterized childhood autoimmune diseases with lymphoproliferation. <i>Pediatric Blood and Cancer</i> , 2018, 65, e26831.	0.8	18
137	High frequencies of asymptomatic Epstein-Barr virus viremia in affected and unaffected individuals with CTLA4 mutations. <i>Clinical Immunology</i> , 2018, 195, 45-48.	1.4	18
138	WDR11 is another causative gene for coloboma, cardiac anomaly and growth retardation in 10q26 deletion syndrome. <i>European Journal of Medical Genetics</i> , 2020, 63, 103626.	0.7	18
139	Inborn errors of immunity—recent advances in research on the pathogenesis. <i>Inflammation and Regeneration</i> , 2021, 41, 9.	1.5	18
140	T and B cell abnormalities, pneumocystis pneumonia, and chronic lymphocytic leukemia associated with an AIOLOS defect in patients. <i>Journal of Experimental Medicine</i> , 2021, 218, .	4.2	18
141	Richter syndrome with two B cell clones possessing different surface immunoglobulins and immunoglobulin gene rearrangements. <i>American Journal of Hematology</i> , 1990, 35, 32-36.	2.0	17
142	Early Activation Events Induced by the Staphylococcal Superantigen Toxic Shock Syndrome Toxin-1 in Human Peripheral Blood Monocytes. <i>Clinical Immunology and Immunopathology</i> , 1994, 70, 137-144.	2.1	17
143	EWSR 1/ ELF 5 induces acute myeloid leukemia by inhibiting p53/p21 pathway. <i>Cancer Science</i> , 2016, 107, 1745-1754.	1.7	17
144	Hematopoietic Cell Transplantation for Severe Combined Immunodeficiency Patients: a Japanese Retrospective Study. <i>Journal of Clinical Immunology</i> , 2021, 41, 1865-1877.	2.0	17

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145	Characterization of Epstein-Barr Virus (EBV)-Positive NK Cells Isolated from Hydroa Vacciniforme-Like Eruptions. <i>Microbiology and Immunology</i> , 2003, 47, 543-552.	0.7	16
146	Amelioration of neurodegenerative diseases by cell death-induced cytoplasmic delivery of humanin. <i>Journal of Controlled Release</i> , 2013, 166, 307-315.	4.8	16
147	Artemisinin-dependent <i>scp</i> DNA double-strand break formation at stalled replication forks. <i>Cancer Science</i> , 2013, 104, 703-710.	1.7	16
148	Recent advances in the study of immunodeficiency and DNA damage response. <i>International Journal of Hematology</i> , 2017, 106, 357-365.	0.7	16
149	Transplantation of Human Autologous Synovial Mesenchymal Stem Cells with Trisomy 7 into the Knee Joint and 5-Years of Follow-up. <i>Stem Cells Translational Medicine</i> , 2021, 10, 1530-1543.	1.6	16
150	Ataxia-Telangiectasia-Mutated-Dependent Activation of Ku in Human Fibroblasts Exposed to Hydrogen Peroxide. <i>Annals of the New York Academy of Sciences</i> , 2006, 1091, 76-82.	1.8	15
151	Two novel <i>scp</i> HSD3B2 missense mutations with diverse residual enzymatic activities for Δ^5 -steroids. <i>Clinical Endocrinology</i> , 2014, 80, 782-789.	1.2	15
152	Palivizumab Use In Japanese Infants And Children With Immunocompromised Conditions. <i>Pediatric Infectious Disease Journal</i> , 2014, 33, 1183-1185.	1.1	15
153	Wiskott-Aldrich Syndrome in a Girl Caused by Heterozygous <i>WASP</i> Mutation and Extremely Skewed X-Chromosome Inactivation: A Novel Association with Maternal Uniparental Isodisomy 6. <i>Neonatology</i> , 2015, 107, 185-190.	0.9	15
154	Enhanced AKT Phosphorylation of Circulating B Cells in Patients With Activated PI3K γ Syndrome. <i>Frontiers in Immunology</i> , 2018, 9, 568.	2.2	15
155	Inherited CARD9 Deficiency in a Child with Invasive Disease Due to <i>Exophiala dermatitidis</i> and Two Older but Asymptomatic Siblings. <i>Journal of Clinical Immunology</i> , 2021, 41, 975-986.	2.0	15
156	Hematopoietic Cell Transplantation Rescues Inflammatory Bowel Disease and Dysbiosis of Gut Microbiota in XIAP Deficiency. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2021, 9, 3767-3780.	2.0	15
157	Acute Cerebellitis and Concurrent Encephalitis Associated with Parvovirus B19 Infection. <i>Pediatric Infectious Disease Journal</i> , 2012, 31, 427.	1.1	14
158	Aberrant glycosylation of IgA in Wiskott-Aldrich syndrome and X-linked thrombocytopenia. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 131, 587-590.e3.	1.5	14
159	Amelioration of intractable epilepsy by adjunct vagus nerve stimulation therapy in a girl with a CDKL5 mutation. <i>Brain and Development</i> , 2017, 39, 341-344.	0.6	14
160	Droplet Digital PCR-Based Chimerism Analysis for Primary Immunodeficiency Diseases. <i>Journal of Clinical Immunology</i> , 2018, 38, 300-306.	2.0	14
161	Long-term outcome and chimerism in patients with Wiskott-Aldrich syndrome treated by hematopoietic cell transplantation: a retrospective nationwide survey. <i>International Journal of Hematology</i> , 2019, 110, 364-369.	0.7	14
162	A postzygotic KRAS mutation in a patient with Schimmelpenning syndrome presenting with lipomatosis, renovascular hypertension, and diabetes mellitus. <i>Journal of Human Genetics</i> , 2019, 64, 177-181.	1.1	14

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164	Population Pharmacokinetics of Intravenous Busulfan in Japanese Pediatric Patients With Primary Immunodeficiency Diseases. <i>Journal of Clinical Pharmacology</i> , 2018, 58, 327-331.	1.0	13
165	Hematopoietic Cell Transplantation for Chronic Granulomatous Disease in Japan. <i>Frontiers in Immunology</i> , 2020, 11, 1617.	2.2	13
166	Inborn errors of IKAROS and AIOLOS. <i>Current Opinion in Immunology</i> , 2021, 72, 239-248.	2.4	13
167	Characterization of the Î³c chain among 27 unrelated Japanese patients with X-linked severe combined immunodeficiency (X-SCID). <i>Human Genetics</i> , 2000, 107, 406-408.	1.8	12
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172	Whole-exome sequence analysis of ataxia telangiectasia-like phenotype. <i>Journal of the Neurological Sciences</i> , 2014, 340, 86-90.	0.3	12
173	Clinical characteristics of adolescent cases with Type A insulin resistance syndrome caused by heterozygous mutations in the Î² subunit of the insulin receptor (INSR) gene. <i>Journal of Diabetes</i> , 2019, 11, 46-54.	0.8	12
174	Epstein-Barr Virus-Associated Î³Î´ T-Cell Lymphoproliferative Disorder Associated With Hypomorphic IL2RG Mutation. <i>Frontiers in Pediatrics</i> , 2019, 7, 15.	0.9	12
175	Infliximab treatment for refractory COVID-19-associated multisystem inflammatory syndrome in a Japanese child. <i>Journal of Infection and Chemotherapy</i> , 2022, 28, 814-818.	0.8	12
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177	Myhre syndrome: Age-dependent progressive phenotype. <i>Pediatrics International</i> , 2017, 59, 1205-1206.	0.2	11
178	Survey of the awareness of adult rheumatologists regarding transitional care for patients with juvenile idiopathic arthritis in Japan. <i>Modern Rheumatology</i> , 2018, 28, 981-985.	0.9	11
179	Noncoding RNA transcription at enhancers and genome folding in cancer. <i>Cancer Science</i> , 2019, 110, 2328-2336.	1.7	11
180	A synonymous splice site mutation in IL2RG gene causes late-onset combined immunodeficiency. <i>International Journal of Hematology</i> , 2019, 109, 603-611.	0.7	11

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182	Ex vivo expanded donor CD4 ⁺ lymphocyte infusion against relapsing neuroblastoma: A transient graft-versus-tumor effect. <i>Pediatric Blood and Cancer</i> , 2009, 52, 895-897.	0.8	10
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184	Hematopoietic stem cell transplantation for pulmonary alveolar proteinosis associated with primary immunodeficiency disease. <i>International Journal of Hematology</i> , 2018, 107, 610-614.	0.7	10
185	The environmental risk assessment of cell-processing facilities for cell therapy in a Japanese academic institution. <i>PLoS ONE</i> , 2020, 15, e0236600.	1.1	10
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190	Early hypoperfusion on arterial spin labeling may be a diagnostic marker for acute encephalopathy with biphasic seizures and late reduced diffusion. <i>Brain and Development</i> , 2017, 39, 722.	0.6	9
191	Maternal T and B cell engraftment in two cases of X-linked severe combined immunodeficiency with IgG1 gammopathy. <i>Clinical Immunology</i> , 2017, 183, 112-120.	1.4	9
192	A Case of MECP2 Duplication Syndrome with Gonadotropin-Dependent Precocious Puberty. <i>Hormone Research in Paediatrics</i> , 2017, 87, 271-276.	0.8	9
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195	Perinatal factors affecting growth and development at age 3 years in extremely low birth weight infants born small for gestational age. <i>Clinical Pediatric Endocrinology</i> , 2018, 27, 31-38.	0.4	9
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200	Effect of reduced-intensity conditioning and the risk of late-onset non-infectious pulmonary complications in pediatric patients. <i>European Journal of Haematology</i> , 2017, 99, 525-531.	1.1	8
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203	The progression of salt-wasting and the body weight change during the first 2 weeks of life in classical 21-hydroxylase deficiency patients. <i>Clinical Endocrinology</i> , 2021, 94, 229-236.	1.2	8
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205	Transducible form of p47phox and p67phox compensate for defective NADPH oxidase activity in neutrophils of patients with chronic granulomatous disease. <i>Biochemical and Biophysical Research Communications</i> , 2012, 417, 162-168.	1.0	7
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214	Vaccination for Patients with Inborn Errors of Immunity: a Nationwide Survey in Japan. <i>Journal of Clinical Immunology</i> , 2022, 42, 183-194.	2.0	7
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219	Comprehensive molecular diagnosis of Epstein-Barr virus-associated lymphoproliferative diseases using next-generation sequencing. <i>International Journal of Hematology</i> , 2018, 108, 319-328.	0.7	6
220	Complications of <i>Listeria</i> meningitis in two immunocompetent children. <i>Pediatrics International</i> , 2018, 60, 491-492.	0.2	6
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222	Inherited chromosomally integrated human herpesvirus-6 in a patient with XIAP deficiency. <i>Transplant Infectious Disease</i> , 2020, 22, e13331.	0.7	6
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224	Importance of pediatric rheumatologists and transitional care for juvenile idiopathic arthritis-associated uveitis: a retrospective series of 9 cases. <i>Pediatric Rheumatology</i> , 2020, 18, 26.	0.9	6
225	Utility of novel T-cell-specific extracellular vesicles in monitoring and evaluation of acute GVHD. <i>International Journal of Hematology</i> , 2021, 113, 910-920.	0.7	6
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236	Leucine-rich alpha-2-glycoprotein 1 and angiotensinogen as diagnostic biomarkers for Kawasaki disease. <i>PLoS ONE</i> , 2021, 16, e0257138.	1.1	5
237	T-Cell Development Failure At β 2-Selection Checkpoint and TCR α/β Locus Break Formation Associated with Chromosome 14 Translocation in Ataxia-Telangiectasia Mutated Deficient Mice. <i>Blood</i> , 2011, 118, 184-184.	0.6	5
238	AIOLOS Variants Causing Immunodeficiency in Human and Mice. <i>Frontiers in Immunology</i> , 2022, 13, 866582.	2.2	5
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243	Recurrent mitral valve regurgitation with neutrophil infiltration in a patient with multiple aseptic abscesses. <i>Modern Rheumatology</i> , 2014, 24, 537-539.	0.9	4
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246	A novel COL11A1 missense mutation in siblings with non-ocular Stickler syndrome. <i>Human Genome Variation</i> , 2016, 3, 16003.	0.4	4
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248	Total body irradiation for hematopoietic stem cell transplantation during early childhood is associated with the risk for diabetes mellitus. <i>Endocrine</i> , 2018, 61, 76-82.	1.1	4
249	Gonadal failure among female patients after hematopoietic stem cell transplantation for non-malignant diseases. <i>Clinical Pediatric Endocrinology</i> , 2019, 28, 105-112.	0.4	4
250	Intravenous immunoglobulin (IVIg) efficiency in women with common variable immunodeficiency (CVID) decreases significantly during pregnancy. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2019, 32, 3092-3096.	0.7	4
251	DNA Ligase IV Deficiency Identified by Chance Following Vaccine-Derived Rubella Virus Infection. <i>Journal of Clinical Immunology</i> , 2020, 40, 1187-1190.	2.0	4
252	Cytomegalovirus Laryngitis in Primary Combined Immunodeficiency Diseases. <i>Journal of Clinical Immunology</i> , 2021, 41, 243-247.	2.0	4

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254	Primary cutaneous cryptococcosis associated with acute lymphoblastic leukemia.. <i>Nishinihon Journal of Dermatology</i> , 1988, 50, 862-866.	0.0	4
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256	Successful ruxolitinib administration for a patient with steroidâ€‘refractory idiopathic pneumonia syndrome following hematopoietic stem cell transplantation: A case report and literature review. <i>Clinical Case Reports (discontinued)</i> , 2021, 9, e05242.	0.2	4
257	In vivo class switch of B cells after cord blood stem cell transplantation in severe combined immune deficient (SCID) patient. <i>American Journal of Hematology</i> , 2000, 65, 176-177.	2.0	3
258	Fatal degeneration of specialized cardiac muscle associated with chronic active Epsteinâ€‘Barr virus infection. <i>Pediatrics International</i> , 2009, 51, 846-848.	0.2	3
259	Graft versus host disease-dependent renal dysfunction after hematopoietic stem cell transplantation. <i>CEN Case Reports</i> , 2014, 3, 202-205.	0.5	3
260	Transient abnormal myelopoiesis in nonâ€‘ <i>scp</i> >D</i> own syndrome neonate. <i>Pediatrics International</i> , 2015, 57, e14-7.	0.2	3
261	Steroid-responsive Status Epilepticus Caused by Human Parvovirus B19 Encephalitis. <i>Pediatric Infectious Disease Journal</i> , 2016, 35, 227-228.	1.1	3
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265	Pharmacokinetic properties of PrivigenÂ® in Japanese patients with primary immunodeficiency. <i>Immunological Medicine</i> , 2019, 42, 162-168.	1.4	3
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267	A case of generalized lipodystrophy-associated progeroid syndrome treated by leptin replacement with short and long-term monitoring of the metabolic and endocrine profiles. <i>Endocrine Journal</i> , 2020, 67, 211-218.	0.7	3
268	Another Exciting Dataâ€‘HCT Successfully Cured Patients with DADA2. <i>Journal of Clinical Immunology</i> , 2021, 41, 1443-1445.	2.0	3
269	Stem cell transplantation for pediatric patients with adrenoleukodystrophy: A nationwide retrospective analysis in Japan. <i>Pediatric Transplantation</i> , 2022, 26, e14125.	0.5	3
270	Endocrinopathies in Inborn Errors of Immunity. <i>Frontiers in Immunology</i> , 2021, 12, 786241.	2.2	3

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272	Two ovarian candidate enhancers, identified by time series enhancer RNA analyses, harbor rare genetic variations identified in ovarian insufficiency. <i>Human Molecular Genetics</i> , 2022, 31, 2223-2235.	1.4	3
273	Cartilage–air hypoplasia with T–cell dysfunction. <i>Pediatrics International</i> , 2022, 64, e15080.	0.2	3
274	Reliability of antinuclear matrix protein 2 antibody assays in idiopathic inflammatory myopathies is dependent on target protein properties. <i>Journal of Dermatology</i> , 2022, 49, 441-447.	0.6	3
275	An adult case of suspected A20 haploinsufficiency mimicking polyarteritis nodosa. <i>Rheumatology</i> , 2022, 61, e337-e340.	0.9	3
276	Generation and Function of $\gamma\delta$ T Cells after Allogeneic Bone Marrow Transplantation in Humans: Comparison in Absence or Presence of HLA-Matched or Mismatched Thymus. <i>Pediatrics International</i> , 1991, 33, 146-158.	0.2	2
277	Qualitative and quantitative differences in the intensity of Fas-mediated intracellular signals determine life and death in T cells. <i>International Journal of Hematology</i> , 2010, 92, 262-270.	0.7	2
278	Prominent eosinophilia but less eosinophil activation in a patient with Omenn syndrome. <i>Pediatrics International</i> , 2010, 52, e196-9.	0.2	2
279	Effect of eculizumab and recombinant human soluble thrombomodulin combination therapy in a 7-year-old girl with atypical hemolytic uremic syndrome due to anti-factor H autoantibodies. <i>CEN Case Reports</i> , 2014, 3, 110-117.	0.5	2
280	An infant case of severe hypereosinophilia and systemic symptoms with multiple drug hypersensitivity and reactivation of cytomegalovirus and BK virus. <i>Allergy International</i> , 2017, 66, 479-481.	1.4	2
281	Type 1 diabetes mellitus associated with activated phosphatidylinositol 3–kinase delta syndrome, type 2. <i>Journal of Diabetes</i> , 2018, 10, 421-422.	0.8	2
282	Hematopoietic cell transplantation for myeloid/NK cell precursor acute leukemia in second remission. <i>Clinical Case Reports (discontinued)</i> , 2018, 6, 1023-1028.	0.2	2
283	Hematopoietic cell transplantation for asymptomatic X-linked lymphoproliferative syndrome type 1. <i>Allergy, Asthma and Clinical Immunology</i> , 2018, 14, 82.	0.9	2
284	High-throughput analysis revealed the unique immunoglobulin gene rearrangements in plasmacytoma-like post-transplant lymphoproliferative disorder. <i>British Journal of Haematology</i> , 2020, 189, e164-e168.	1.2	2
285	Prematurity at less than 24 weeks of gestation is a risk for prolonged hyperglycemia in extremely low-birth weight infants. <i>Endocrine</i> , 2020, 70, 71-77.	1.1	2
286	Disseminated fusariosis in a child after haploidentical hematopoietic stem cell transplantation. <i>Pediatrics International</i> , 2020, 62, 419-420.	0.2	2
287	Successful treatment of joint and fascial chronic graft-versus-host disease with baricitinib. <i>Rheumatology</i> , 2021, . .	0.9	2
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