Satoshi Okada

List of Publications by Year
in descending order

163 papers	14,020 citations	44042 48 h-index	24232 110 g-index
180 all docs	180 docs citations	180 times ranked	18389 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	Gain-of-function human STAT1 mutations impair IL-17 immunity and underlie chronic mucocutaneous candidiasis. Journal of Experimental Medicine, 2011, 208, 1635-1648.	4.2	739
4	Heterozygous STAT1 gain-of-function mutations underlie an unexpectedly broad clinical phenotype. Blood, 2016, 127, 3154-3164.	0.6	465
5	Mycobacterial Disease and Impaired IFN-Î³ Immunity in Humans with Inherited ISG15 Deficiency. Science, 2012, 337, 1684-1688.	6.0	455
6	Human intracellular ISG15 prevents interferon-Î±/Î² over-amplification and auto-inflammation. Nature, 2015, 517, 89-93.	13.7	432
7	Life-threatening influenza and impaired interferon amplification in human IRF7 deficiency. Science, 2015, 348, 448-453.	6.0	389
8	Magnetic Ordering in Hexagonally Bonded Sheets with First-Row Elements. Physical Review Letters, 2001, 87, 146803.	2.9	369
9	Impairment of immunity to Candida and Mycobacterium in humans with bi-allelic RORC mutations. Science, 2015, 349, 606-613.	6.0	366
10	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
11	Phenotype, penetrance, and treatment of 133 cytotoxic T-lymphocyte antigen 4â€“insufficient subjects. Journal of Allergy and Clinical Immunology, 2018, 142, 1932-1946.	1.5	344
12	Human TYK2 deficiency: Mycobacterial and viral infections without hyper-IgE syndrome. Journal of Experimental Medicine, 2015, 212, 1641-1662.	4.2	293
13	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
14	Inborn errors of human STAT1: allelic heterogeneity governs the diversity of immunological and infectious phenotypes. Current Opinion in Immunology, 2012, 24, 364-378.	2.4	245
15	Human genetic and immunological determinants of critical COVID-19 pneumonia. Nature, 2022, 603, 587-598.	13.7	216
16	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. Cell, 2020, 181, 1194-1199.	13.5	185
17	Monogenic mutations differentially affect the quantity and quality of T follicular helper cells in patients with human primary immunodeficiencies. Journal of Allergy and Clinical Immunology, 2015, 136, 993-1006.e1.	1.5	181
18	DOCK8 deficiency impairs CD8 T cell survival and function in humans and mice. Journal of Experimental Medicine, 2011, 208, 2305-2320.	4.2	175

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19	Inherited DOCK2 Deficiency in Patients with Early-Onset Invasive Infections. New England Journal of Medicine, 2015, 372, 2409-2422.	13.9	169
20	Mendelian susceptibility to mycobacterial disease: 2014â€“2018 update. Immunology and Cell Biology, 2019, 97, 360-367.	1.0	163
21	Human IFN-Î³ immunity to mycobacteria is governed by both IL-12 and IL-23. Science Immunology, 2018, 3, .	5.6	152
22	Chronic mucocutaneous candidiasis disease associated with inborn errors of ILâ€17 immunity. Clinical and Translational Immunology, 2016, 5, e114.	1.7	148
23	Genetic, immunological, and clinical features of patients with bacterial and fungal infections due to inherited IL-17RA deficiency. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E8277-E8285.	3.3	137
24	Hematopoietic stem cell transplantation in patients with gain-of-function signal transducer and activator of transcription 1 mutations. Journal of Allergy and Clinical Immunology, 2018, 141, 704-717.e5.	1.5	128
25	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
26	New and recurrent gain-of-functionSTAT1mutations in patients with chronic mucocutaneous candidiasis from Eastern and Central Europe. Journal of Medical Genetics, 2013, 50, 567-578.	1.5	105
27	The potential of SLC6A4 gene methylation analysis for the diagnosis and treatment of major depression. Journal of Psychiatric Research, 2014, 53, 47-53.	1.5	100
28	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
29	Abnormal hematopoiesis and autoimmunity in human subjects with germline IKZF1 mutations. Journal of Allergy and Clinical Immunology, 2017, 140, 223-231.	1.5	99
30	Flow cytometry-based diagnosis of primary immunodeficiency diseases. Allergology International, 2018, 67, 43-54.	1.4	97
31	Inborn Errors of RNA Lariat Metabolism in Humans with Brainstem Viral Infection. Cell, 2018, 172, 952-965.e18.	13.5	92
32	Quantification of Îº-deleting recombination excision circles in Guthrie cards for the identification of early B-cell maturation defects. Journal of Allergy and Clinical Immunology, 2011, 128, 223-225.e2.	1.5	91
33	Phosphatase and tensin homolog (PTEN) mutation can cause activated phosphatidylinositol 3-kinase Î´ syndromeâ€“like immunodeficiency. Journal of Allergy and Clinical Immunology, 2016, 138, 1672-1680.e10.	1.5	87
34	Human STAT1 Gain-of-Function Heterozygous Mutations: Chronic Mucocutaneous Candidiasis and Type I Interferonopathy. Journal of Clinical Immunology, 2020, 40, 1065-1081.	2.0	86
35	The expansion of human T-bet ^{high} CD21 ^{low} B cells is T cell dependent. Science Immunology, 2021, 6, eabh0891.	5.6	82
36	Increased Risk for Malignancies in 131 Affected CTLA4 Mutation Carriers. Frontiers in Immunology, 2018, 9, 2012.	2.2	79

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37	Simple diagnosis ofSTAT1gain-of-function alleles in patients with chronic mucocutaneous candidiasis. Journal of Leukocyte Biology, 2013, 95, 667-676.	1.5	77
38	Unique and shared signaling pathways cooperate to regulate the differentiation of human CD4+ T cells into distinct effector subsets. Journal of Experimental Medicine, 2016, 213, 1589-1608.	4.2	77
39	The potential use of histone deacetylase inhibitors in the treatment of depression. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2016, 64, 320-324.	2.5	73
40	Studying severe long COVID to understand post-infectious disorders beyond COVID-19. Nature Medicine, 2022, 28, 879-882.	15.2	72
41	Identification of novel missense mutations (Phe310Leu and Gly439Arg) in a neonatal case of hypophosphatasia.. Journal of Clinical Endocrinology and Metabolism, 1996, 81, 4458-4461.	1.8	71
42	Dominant-negative STAT1 SH2 domain mutations in unrelated patients with mendelian susceptibility to mycobacterial disease. Human Mutation, 2012, 33, 1377-1387.	1.1	71
43	Gain-of-function IKBKB mutation causes human combined immune deficiency. Journal of Experimental Medicine, 2018, 215, 2715-2724.	4.2	69
44	Therapeutic options for CTLA-4 insufficiency. Journal of Allergy and Clinical Immunology, 2022, 149, 736-746.	1.5	68
45	Nephrotoxicity and its prevention by vitamin E in ferric nitrilotriacetate-promoted lipid. Lipids and Lipid Metabolism, 1987, 922, 28-33.	2.6	63
46	Induction of mesothelioma by intraperitoneal injections of ferric saccharate in male Wistar rats. British Journal of Cancer, 1989, 60, 708-711.	2.9	62
47	Recessive inborn errors of type I IFN immunity in children with COVID-19 pneumonia. Journal of Experimental Medicine, 2022, 219, .	4.2	59
48	Haploinsufficiency at the human IFNGR2 locus contributes to mycobacterial disease. Human Molecular Genetics, 2013, 22, 769-781.	1.4	58
49	Polycomb-group complex 1 acts as an E3 ubiquitin ligase for Geminin to sustain hematopoietic stem cell activity. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 10396-10401.	3.3	57
50	Safety and efficacy of treatment with asfotase alfa in patients with hypophosphatasia: Results from a Japanese clinical trial. Clinical Endocrinology, 2017, 87, 10-19.	1.2	55
51	Neurodevelopmental abnormalities associated with severe congenital neutropenia due to the R86X mutation in the HAX1 gene. Journal of Medical Genetics, 2008, 45, 802-807.	1.5	48
52	Heterozygosity for the Y701C STAT1 mutation in a multiplex kindred with multifocal osteomyelitis. Haematologica, 2013, 98, 1641-1649.	1.7	47
53	Clinical characteristics and treatment of 50 cases of Blau syndrome in Japan confirmed by genetic analysis of the NOD2 mutation. Annals of the Rheumatic Diseases, 2020, 79, 1492-1499.	0.5	47
54	Age-Dependent Association between Pulmonary Tuberculosis and Common TOX Variants in the 8q12â€“13 Linkage Region. American Journal of Human Genetics, 2013, 92, 407-414.	2.6	46

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55	Alanine-scanning mutagenesis of human signal transducer and activator of transcription 1 to estimate loss- or gain-of-function variants. Journal of Allergy and Clinical Immunology, 2017, 140, 232-241.	1.5	43
56	A global effort to dissect the human genetic basis of resistance to SARS-CoV-2 infection. Nature Immunology, 2022, 23, 159-164.	7.0	41
57	Rescue of recurrent deep intronic mutation underlying cell typeâ€“dependent quantitative NEMO deficiency. Journal of Clinical Investigation, 2018, 129, 583-597.	3.9	38
58	Significant augmentation of regulatory T cell numbers occurs during the early neonatal period. Clinical and Experimental Immunology, 2017, 190, 268-279.	1.1	35
59	Germline-Activating RRAS2 Mutations Cause Noonan Syndrome. American Journal of Human Genetics, 2019, 104, 1233-1240.	2.6	35
60	Vaccine breakthrough hypoxemic COVID-19 pneumonia in patients with auto-Abs neutralizing type I IFNs. Science Immunology, 2023, 8, .	5.6	35
61	Partial IFN-Î³R2 deficiency is due to protein misfolding and can be rescued by inhibitors of glycosylation. Blood, 2013, 122, 2390-2401.	0.6	34
62	Gut microbiota and systemic immunity in health and disease. International Immunology, 2021, 33, 197-209.	1.8	34
63	APRIL-dependent lifelong plasmacyte maintenance and immunoglobulin production in humans. Journal of Allergy and Clinical Immunology, 2020, 146, 1109-1120.e4.	1.5	33
64	A variant in human AIOLOS impairs adaptive immunity by interfering with IKAROS. Nature Immunology, 2021, 22, 893-903.	7.0	33
65	Genetic, Immunological, and Clinical Features of 32 Patients with Autosomal Recessive STAT1 Deficiency. Journal of Immunology, 2021, 207, 133-152.	0.4	33
66	Inborn errors of STAT1 immunity. Current Opinion in Immunology, 2021, 72, 59-64.	2.4	33
67	A Novel Homozygous p.R1105X Mutation of the AP4E1 Gene in Twins with Hereditary Spastic Paraplegia and Mycobacterial Disease. PLoS ONE, 2013, 8, e58286.	1.1	31
68	Frequent somatic mosaicism of NEMO in T cells of patients with X-linked anhidrotic ectodermal dysplasia with immunodeficiency. Blood, 2012, 119, 5458-5466.	0.6	30
69	The novel IFNGR1 mutation 774del4 produces a truncated form of interferon-Â receptor 1 and has a dominant-negative effect on interferon-Â signal transduction. Journal of Medical Genetics, 2007, 44, 485-491.	1.5	29
70	Severe developmental delay and epilepsy in a Japanese patient with severe congenital neutropenia due to HAX1 deficiency. Haematologica, 2007, 92, e123-e125.	1.7	29
71	A Patient with CTLA-4 Haploinsufficiency Presenting Gastric Cancer. Journal of Clinical Immunology, 2016, 36, 28-32.	2.0	29
72	Adipsic hypernatremia without hypothalamic lesions accompanied by autoantibodies to subfornical organ. Brain Pathology, 2017, 27, 323-331.	2.1	29

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73	Molecular pathogenesis of a novel mutation, G108D, in short-chain acyl-CoA dehydrogenase identified in subjects with short-chain acyl-CoA dehydrogenase deficiency. Human Genetics, 2010, 127, 619-628.	1.8	26
74	Hoxb4 transduction down-regulates Geminin protein, providing hematopoietic stem and progenitor cells with proliferation potential. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 21529-21534.	3.3	26
75	Newborn screening for carnitine palmitoyltransferase II deficiency using (C16 + C18:1)/C2: Evaluation of additional indices for adequate sensitivity and lower false-positivity. Molecular Genetics and Metabolism, 2017, 122, 67-75.	0.5	26
76	Autosomal recessive complete STAT1 deficiency caused by compound heterozygous intronic mutations. International Immunology, 2020, 32, 663-671.	1.8	26
77	Development of a New Enzymatic Diagnosis Method for Very-long-chain Acyl-CoA Dehydrogenase Deficiency by Detecting 2-Hexadecenoyl-CoA Production and its Application in Tandem Mass Spectrometry-based Selective Screening and Newborn Screening in Japan. Pediatric Research, 2008, 64, 667-672.	1.1	25
78	Linkage-specific deubiquitylation by OTUD5 defines an embryonic pathway intolerant to genomic variation. Science Advances, 2021, 7, .	4.7	25
79	Clinical Characteristics of Perinatal Lethal Hypophosphatasia: A Report of 6 Cases. Clinical Pediatric Endocrinology, 2010, 19, 7-13.	0.4	24
80	Clinical and Host Genetic Characteristics of Mendelian Susceptibility to Mycobacterial Diseases in Japan. Journal of Clinical Immunology, 2011, 31, 309-314.	2.0	24
81	Neutralizing Type I Interferon Autoantibodies in Japanese Patients with Severe COVID-19. Journal of Clinical Immunology, 2022, 42, 1360-1370.	2.0	24
82	The C-Terminal Region of Serotonin Transporter Is Important for Its Trafficking and Glycosylation. Journal of Pharmacological Sciences, 2009, 111, 392-404.	1.1	22
83	Atypical SIFD with novel TRNT1 mutations: a case study on the pathogenesis of B-cell deficiency. International Journal of Hematology, 2019, 109, 382-389.	0.7	22
84	A comparison of the defective granulopoiesis in childhood cyclic neutropenia and in severe congenital neutropenia. Haematologica, 2005, 90, 1032-41.	1.7	22
85	IL-17 T Cellsâ€™ Defective Differentiation In Vitro Despite Normal Range Ex Vivo in Chronic Mucocutaneous Candidiasis Due to STAT1 Mutation. Journal of Investigative Dermatology, 2014, 134, 1155-1157.	0.3	21
86	Screening of MCAD deficiency in Japan: 16years' experience of enzymatic and genetic evaluation. Molecular Genetics and Metabolism, 2016, 119, 322-328.	0.5	21
87	Molecular mechanism and structural basis of gain-of-function of STAT1 caused by pathogenic R274Q mutation. Journal of Biological Chemistry, 2017, 292, 6240-6254.	1.6	21
88	From Your Nose to Your Toes: A Review of Severe Acute Respiratory Syndrome Coronavirus 2 Pandemicâ€'Associated Pernio. Journal of Investigative Dermatology, 2021, 141, 2791-2796.	0.3	21
89	Identification of discriminative gene-level and protein-level features associated with pathogenic gain-of-function and loss-of-function variants. American Journal of Human Genetics, 2021, 108, 2301-2318.	2.6	21
90	Respiratory viral infections in otherwise healthy humans with inherited IRF7 deficiency. Journal of Experimental Medicine, 2022, 219, .	4.2	21

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91	Cardiac infiltration in early-onset sarcoidosis associated with a novel heterozygous mutation, G481D, in CARD15. Rheumatology, 2009, 48, 706-707.	0.9	20
92	Human gain-of-function STAT1 mutation disturbs IL-17 immunity in mice. International Immunology, 2020, 32, 259-272.	1.8	20
93	Whole-Exome Sequencing-Based Approach for Germline Mutations in Patients with Inborn Errors of Immunity. Journal of Clinical Immunology, 2020, 40, 729-740.	2.0	20
94	Ruxolitinib treatment of a patient with steroid-dependent severe autoimmunity due to STAT1 gain-of-function mutation. International Journal of Hematology, 2020, 112, 258-262.	0.7	20
95	Heterozygous missense variant of the proteasome subunit Î²-type 9 causes neonatal-onset autoinflammation and immunodeficiency. Nature Communications, 2021, 12, 6819.	5.8	20
96	Disseminated BCG Infection Mimicking Metastatic Nasopharyngeal Carcinoma in an Immunodeficient Child with a Novel Hypomorphic NEMO Mutation. Journal of Clinical Immunology, 2011, 31, 802-810.	2.0	18
97	A gain-of-function mutation of STAT1: A novel genetic factor contributing to chronic mucocutaneous candidiasis. Acta Microbiologica Et Immunologica Hungarica, 2017, 64, 191-201.	0.4	18
98	Individual Clinically Diagnosed with CHARGE Syndrome but with a Mutation in KMT2D, a Gene Associated with Kabuki Syndrome: A Case Report. Frontiers in Genetics, 2017, 8, 210.	1.1	18
99	Six yearsâ€™ accomplishment of the Initiative on Rare and Undiagnosed Diseases: nationwide project in Japan to discover causes, mechanisms, and cures. Journal of Human Genetics, 2022, 67, 505-513.	1.1	17
100	Mendelian susceptibility to mycobacterial diseases: state of the art. Clinical Microbiology and Infection, 2022, 28, 1429-1434.	2.8	17
101	Steroid-Dependent ACTH-Produced Thymic Carcinoid: Regulation of POMC Gene Expression by Cortisol via Methylation of Its Promoter Region. Hormone Research in Paediatrics, 2007, 67, 257-262.	0.8	16
102	Two families with TET3-related disorder showing neurodevelopmental delay with craniofacial dysmorphisms. Journal of Human Genetics, 2022, 67, 157-164.	1.1	16
103	Identification of the integrin Î²3 L718P mutation in a pedigree with autosomal dominant thrombocytopenia with anisocytosis. British Journal of Haematology, 2013, 160, 521-529.	1.2	15
104	Incidence and Characteristics of Adrenal Crisis in Children Younger than 7 Years with 21-Hydroxylase Deficiency: A Nationwide Survey in Japan. Hormone Research in Paediatrics, 2018, 89, 166-171.	0.8	15
105	Enhanced AKT Phosphorylation of Circulating B Cells in Patients With Activated PI3KÎ´ Syndrome. Frontiers in Immunology, 2018, 9, 568.	2.2	15
106	Functional analysis of novel A20 variants in patients with atypical inflammatory diseases. Arthritis Research and Therapy, 2021, 23, 52.	1.6	15
107	Inherited CARD9 Deficiency in a Child with Invasive Disease Due to Exophiala dermatitidis and Two Older but Asymptomatic Siblings. Journal of Clinical Immunology, 2021, 41, 975-986.	2.0	15
108	Extrapulmonary tuberculosis mimicking Mendelian susceptibility to mycobacterial disease in a patient with signal transducer andÂactivator of transcription 1 (STAT1) gain-of-function mutation. Journal of Allergy and Clinical Immunology, 2016, 137, 619-622.e1.	1.5	14

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109	Characteristic clinical features of adipsic hypernatremia patients with subfornical organ-targeting antibody. Clinical Pediatric Endocrinology, 2017, 26, 197-205.	0.4	13
110	Acute severe encephalopathy related to human herpesvirus-6 infection in a patient with carnitine palmitoyltransferase 2 deficiency carrying thermolabile variants. Brain and Development, 2013, 35, 449-453.	0.6	12
111	Significance of ACADM mutations identified through newborn screening of MCAD deficiency in Japan. Molecular Genetics and Metabolism, 2016, 118, 9-14.	0.5	12
112	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
113	Clinical characteristics of septo-optic dysplasia accompanied by congenital central hypothyroidism in Japan. Clinical Pediatric Endocrinology, 2017, 26, 207-213.	0.4	11
114	Enhanced osteoclastogenesis in patients with MSMD due to impaired response to IFN-Î³. Journal of Allergy and Clinical Immunology, 2022, 149, 252-261.e6.	1.5	11
115	Case Report: A New Gain-of-Function Mutation of STAT1 Identified in a Patient With Chronic Mucocutaneous Candidiasis and Rosacea-Like Demodicosis: An Emerging Association. Frontiers in Immunology, 2021, 12, 760019.	2.2	11
116	Empty sella/pituitary atrophy and endocrine impairments as a consequence of radiation and chemotherapy in long-term survivors of childhood leukemia. International Journal of Hematology, 2011, 94, 399-402.	0.7	10
117	<scp>VLCAD</scp> deficiency in a patient who recovered from ventricular fibrillation, but died suddenly of a respiratory syncytial virus infection. Pediatrics International, 2013, 55, 775-778.	0.2	10
118	Electrically induced ambipolar spin vanishments in carbon nanotubes. Scientific Reports, 2015, 5, 11859.	1.6	10
119	Mosaicism of an ELANE Mutation in an Asymptomatic Mother in a Familial Case of Cyclic Neutropenia. Journal of Clinical Immunology, 2015, 35, 512-516.	2.0	10
120	A nationwide questionnaire survey targeting Japanese pediatric endocrinologists regarding transitional care in childhood, adolescent, and young adult cancer survivors. Clinical Pediatric Endocrinology, 2020, 29, 55-62.	0.4	10
121	IRAK4 Deficiency Presenting with Anti-NMDAR Encephalitis and HHV6 Reactivation. Journal of Clinical Immunology, 2021, 41, 125-135.	2.0	10
122	Clinical and Immunological Heterogeneity in Japanese Patients with Gain-of-Function Variants in STAT3. Journal of Clinical Immunology, 2021, 41, 780-790.	2.0	10
123	Human T Follicular Helper Cells in Primary Immunodeficiency: Quality Just as Important as Quantity. Journal of Clinical Immunology, 2016, 36, 40-47.	2.0	9
124	Gain-of-Function STAT1 Mutation With Familial Lymphadenopathy and Hodgkin Lymphoma. Frontiers in Pediatrics, 2019, 7, 160.	0.9	9
125	Ruxolitinib Response in an Infant With Veryâ€earlyâ€onset Inflammatory Bowel Disease and Gainâ€ofâ€function STAT1 Mutation. Journal of Pediatric Gastroenterology and Nutrition, 2020, 71, e132-e133.	0.9	9
126	Impaired B-Cell Differentiation in a Patient With STAT1 Gain-of-Function Mutation. Frontiers in Immunology, 2020, 11, 557521.	2.2	9

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127	A Novel Homozygous Mutation Destabilizes IKKÎ² and Leads to Human Combined Immunodeficiency. Frontiers in Immunology, 2020, 11, 517544.	2.2	8
128	Antiâ€human neutrophil antigenâ€1a, â€1b, and â€2 antibodies in neonates and children with immune neutropenias analyzed by extracted granulocyte antigen immunofluorescence assay. Transfusion, 2017, 57, 2586-2594.	0.8	7
129	A Case with Spondyloenchondrodysplasia Treated with Growth Hormone. Frontiers in Endocrinology, 2017, 8, 157.	1.5	7
130	Decreased Expression in Nuclear Factor-ÎºB Essential Modulator Due to a Novel Splice-Site Mutation Causes X-linked Ectodermal Dysplasia with Immunodeficiency. Journal of Clinical Immunology, 2011, 31, 762-772.	2.0	6
131	Two Japanese patients with the renal form of pseudohypoaldosteronism type 1 caused by mutations of <i>NR3C2</i>. Clinical Pediatric Endocrinology, 2015, 24, 135-138.	0.4	6
132	T2-FLAIR Mismatch Sign and Response to Radiotherapy in Diffuse Intrinsic Pontine Glioma. Pediatric Neurosurgery, 2021, 56, 1-9.	0.4	6
133	Novel STAT-3 gain-of-function variant with hypogammaglobulinemia and recurrent infection phenotype. Clinical and Experimental Immunology, 2021, 205, 354-362.	1.1	6
134	A Case Report of a Japanese Boy with Morquio A Syndrome: Effects of Enzyme Replacement Therapy Initiated at the Age of 24 Months. International Journal of Molecular Sciences, 2020, 21, 989.	1.8	5
135	Downregulation of endothelial nitric oxide synthase (eNOS) and endothelin-1 (ET-1) in a co-culture system with human stimulated X-linked CGD neutrophils. PLoS ONE, 2020, 15, e0230665.	1.1	5
136	Development and operation of an electrostatic time-of-flight detector for the Rare RI storage Ring. Nuclear Instruments and Methods in Physics Research, Section A: Accelerators, Spectrometers, Detectors and Associated Equipment, 2021, 986, 164713.	0.7	5
137	Successful Hematopoietic Stem Cell Transplantation for Autosomal Recessive STAT1 Complete Deficiency. Journal of Clinical Immunology, 2021, 41, 684-687.	2.0	5
138	The frequencies of very long-chain acyl-CoA dehydrogenase deficiency genetic variants in Japan have changed since the implementation of expanded newborn screening. Molecular Genetics and Metabolism, 2022, 136, 74-79.	0.5	5
139	Bone marrow transplantation from a human leukocyte antigen-mismatched unrelated donor in a case with C1q deficiency associated with refractory systemic lupus erythematosus. International Journal of Hematology, 2021, 113, 302-307.	0.7	4
140	Possible involvement of regulatory T cell abnormalities and variational usage of TCR repertoire in children with autoimmune neutropenia. Clinical and Experimental Immunology, 2021, 204, 1-13.	1.1	4
141	Current Perspectives on Neonatal Screening for Propionic Acidemia in Japan: An Unexpectedly High Incidence of Patients with Mild Disease Caused by a Common PCCB Variant. International Journal of Neonatal Screening, 2021, 7, 35.	1.2	4
142	Clinical impact of the dose and blood concentration of lacosamide in Japanese pediatric patients with epilepsy: A cohort study. Epilepsy and Behavior, 2022, 129, 108614.	0.9	4
143	A novel mutation (Gln433Glu) in exon 12 combined with the G insertion in exon 13 causes severe factor XI deficiency in Japanese patients. Blood Coagulation and Fibrinolysis, 2007, 18, 519-523.	0.5	3
144	Development of rotating magnetic field system for the Î²-NMR method. Hyperfine Interactions, 2013, 220, 65-69.	0.2	3

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145	A 2-year-old Japanese girl with TNF receptorâ€“associated periodic syndrome: A case report of the youngest diagnosed proband in Japan. Modern Rheumatology, 2016, 26, 798-801.	0.9	3
146	Clinical Significance of Serum Soluble TNF Receptor I/II Ratio for the Differential Diagnosis of Tumor Necrosis Factor Receptor-Associated Periodic Syndrome From Other Autoinflammatory Diseases. Frontiers in Immunology, 2020, 11, 576152.	2.2	3
147	Successful allogeneic bone marrow transplantation using immunosuppressive conditioning regimen for a patient with red blood cell transfusion-dependent pyruvate kinase deficiency anemia. Hematology Reports, 2020, 12, 8305.	0.3	3
148	Pneumococcal Serotype-specific Opsonophagocytic Activity in Interleukin-1 Receptor-associated Kinase 4-deficient Patients. Pediatric Infectious Disease Journal, 2021, 40, 460-463.	1.1	3
149	MSMD Patients with IFN-g-STAT1 Signaling Defect Present Enhanced Osteoclastogenesis and Bone Resorption. Blood, 2015, 126, 3591-3591.	0.6	3
150	NBS1 I171V variant underlies individual differences in chromosomal radiosensitivity within human populations. Scientific Reports, 2021, 11, 19661.	1.6	3
151	Title is missing!. Japanese Journal of Food Microbiology, 2000, 17, 143-147.	0.3	2
152	Non-androgen secreting adrenocortical carcinoma in preadolescence: a case report and literature review. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 1313-1317.	0.4	1
153	Effects of perampanel add-on therapy on immunoglobulin levels in pediatric patients with epilepsy. Epilepsy Research, 2020, 167, 106447.	0.8	1
154	Pilot Study on Neonatal Screening for Methylmalonic Acidemia Caused by Defects in the Adenosylcobalamin Synthesis Pathway and Homocystinuria Caused by Defects in Homocysteine Remethylation. International Journal of Neonatal Screening, 2021, 7, 39.	1.2	1
155	Impairment of IL-17 Immunity to Candida and IFN-g Immunity to Mycobacterium in Humans with Bi-Allelic Rorc mutations. Blood, 2015, 126, 205-205.	0.6	1
156	A novel intronic <scp>PORCN</scp> variant creating an alternative splice acceptor site in a mother and her daughter with focal dermal hypoplasia. American Journal of Medical Genetics, Part A, 2022, 188, 1612-1617.	0.7	1
157	Early diagnosis of partial interferon-Î³ receptor 1 deficiency prevents the development of Bacille de Calmette et GuÃ©rin osteomyelitis. Clinical Immunology, 2022, 235, 108933.	1.4	1
158	Management of advancedâ€stage neuroblastoma in a patient with 21â€hydroxalase deficiency. Pediatrics International, 2013, 55, e96-9.	0.2	0
159	Dr. Maeshima, et al, reply. Journal of Rheumatology, 2019, 46, 655-656.	1.0	0
160	Abnormal Localization of Neutrophil Elastase in Patients with Severe Congenital Neutropenia.. Blood, 2006, 108, 1279-1279.	0.6	0
161	Gain-of-Phosphorylation Mutations in Coiled-Coil and DNA-Binding Domain of STAT1 Identified in Japanese Patients with Chronic Mucocutaneous Candidiasis. Blood, 2012, 120, 4831-4831.	0.6	0
162	Excessive Nitric Oxide Production of CGD Neutrophils Induces the Down-Regulation of NOS3 and EDN1 Expression in Human Endothelial Cells. Blood, 2014, 124, 2726-2726.	0.6	0

#	Article	IF	Citations
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