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	Vaccine breakthrough hypoxemic COVID-19 pneumonia in patients with auto-Abs neutralizing type I IFNs. Science Immunology, 2023, 8, .	5.6	35
2	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
3	Therapeutic options for CTLA-4 insufficiency. Journal of Allergy and Clinical Immunology, 2022, 149, 736-746.	1.5	68
4	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
5	Two families with TET3-related disorder showing neurodevelopmental delay with craniofacial dysmorphisms. Journal of Human Genetics, 2022, 67, 157-164.	1.1	16
6	Human genetic and immunological determinants of critical COVID-19 pneumonia. Nature, 2022, 603, 587-598.	13.7	216
7	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
8	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
9	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
10	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
11	Mendelian susceptibility to mycobacterial diseases: state of the art. Clinical Microbiology and Infection, 2022, 28, 1429-1434.	2.8	17
12	Studying severe long COVID to understand post-infectious disorders beyond COVID-19. Nature Medicine, 2022, 28, 879-882.	15.2	72
13	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
14	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
15	Respiratory viral infections in otherwise healthy humans with inherited IRF7 deficiency. Journal of Experimental Medicine, 2022, 219, .	4.2	21
16	Recessive inborn errors of type I IFN immunity in children with COVID-19 pneumonia. Journal of Experimental Medicine, 2022, 219, .	4.2	59
17	Neutralizing Type I Interferon Autoantibodies in Japanese Patients with Severe COVID-19. Journal of Clinical Immunology, 2022, 42, 1360-1370.	2.0	24
18	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

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19	IRAK4 Deficiency Presenting with Anti-NMDAR Encephalitis and HHV6 Reactivation. Journal of Clinical Immunology, 2021, 41, 125-135.	2.0	10
20	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
21	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
22	Successful Hematopoietic Stem Cell Transplantation for Autosomal Recessive STAT1 Complete Deficiency. Journal of Clinical Immunology, 2021, 41, 684-687.	2.0	5
23	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
24	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
25	T2-FLAIR Mismatch Sign and Response to Radiotherapy in Diffuse Intrinsic Pontine Glioma. Pediatric Neurosurgery, 2021, 56, 1-9.	0.4	6
26	Functional analysis of novel A20 variants in patients with atypical inflammatory diseases. Arthritis Research and Therapy, 2021, 23, 52.	1.6	15
27	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
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	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
30	A variant in human AIOLOS impairs adaptive immunity by interfering with IKAROS. Nature Immunology, 2021, 22, 893-903.	7.0	33
31	Novel STAT-3 gain-of-function variant with hypogammaglobulinemia and recurrent infection phenotype. Clinical and Experimental Immunology, 2021, 205, 354-362.	1.1	6
32	Genetic, Immunological, and Clinical Features of 32 Patients with Autosomal Recessive STAT1 Deficiency. Journal of Immunology, 2021, 207, 133-152.	0.4	33
33	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
34	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
35	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
36	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

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37	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
38	Inborn errors of STAT1 immunity. Current Opinion in Immunology, 2021, 72, 59-64.	2.4	33
39	Linkage-specific deubiquitylation by OTUD5 defines an embryonic pathway intolerant to genomic variation. Science Advances, 2021, 7, .	4.7	25
40	Gut microbiota and systemic immunity in health and disease. International Immunology, 2021, 33, 197-209.	1.8	34
41	The expansion of human T-bet ^{high} CD21 ^{low} B cells is T cell dependent. Science Immunology, 2021, 6, eabh0891.	5.6	82
42	NBS1 I171V variant underlies individual differences in chromosomal radiosensitivity within human populations. Scientific Reports, 2021, 11, 19661.	1.6	3
43	Heterozygous missense variant of the proteasome subunit Î²-type 9 causes neonatal-onset autoinflammation and immunodeficiency. Nature Communications, 2021, 12, 6819.	5.8	20
44	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
45	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
46	Human gain-of-function STAT1 mutation disturbs IL-17 immunity in mice. International Immunology, 2020, 32, 259-272.	1.8	20
47	Effects of perampanel add-on therapy on immunoglobulin levels in pediatric patients with epilepsy. Epilepsy Research, 2020, 167, 106447.	0.8	1
48	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
49	Impaired B-Cell Differentiation in a Patient With STAT1 Gain-of-Function Mutation. Frontiers in Immunology, 2020, 11, 557521.	2.2	9
50	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
51	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,749
52	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,983
53	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
54	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. Cell, 2020, 181, 1194-1199.	13.5	185

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55	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
56	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
57	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
58	Autosomal recessive complete STAT1 deficiency caused by compound heterozygous intronic mutations. International Immunology, 2020, 32, 663-671.	1.8	26
59	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
60	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
61	APRIL-dependent lifelong plasmacyte maintenance and immunoglobulin production in humans. Journal of Allergy and Clinical Immunology, 2020, 146, 1109-1120.e4.	1.5	33
62	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
63	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
64	A Novel Homozygous Mutation Destabilizes IKKÎ² and Leads to Human Combined Immunodeficiency. Frontiers in Immunology, 2020, 11, 517544.	2.2	8
65	Gain-of-Function STAT1 Mutation With Familial Lymphadenopathy and Hodgkin Lymphoma. Frontiers in Pediatrics, 2019, 7, 160.	0.9	9
66	Germline-Activating RRAS2 Mutations Cause Noonan Syndrome. American Journal of Human Genetics, 2019, 104, 1233-1240.	2.6	35
67	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
68	Dr. Maeshima, et al, reply. Journal of Rheumatology, 2019, 46, 655-656.	1.0	0
69	Mendelian susceptibility to mycobacterial disease: 2014â€“2018 update. Immunology and Cell Biology, 2019, 97, 360-367.	1.0	163
70	Inborn Errors of RNA Lariat Metabolism in Humans with Brainstem Viral Infection. Cell, 2018, 172, 952-965.e18.	13.5	92
71	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
72	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

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73	Flow cytometry-based diagnosis of primary immunodeficiency diseases. Allergology International, 2018, 67, 43-54.	1.4	97
74	Human IFN-Î³ immunity to mycobacteria is governed by both IL-12 and IL-23. Science Immunology, 2018, 3, .	5.6	152
75	Increased Risk for Malignancies in 131 Affected CTLA4 Mutation Carriers. Frontiers in Immunology, 2018, 9, 2012.	2.2	79
76	Gain-of-function IKBKB mutation causes human combined immune deficiency. Journal of Experimental Medicine, 2018, 215, 2715-2724.	4.2	69
77	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
78	Enhanced AKT Phosphorylation of Circulating B Cells in Patients With Activated PI3KÎ´ Syndrome. Frontiers in Immunology, 2018, 9, 568.	2.2	15
79	Rescue of recurrent deep intronic mutation underlying cell typeâ€“dependent quantitative NEMO deficiency. Journal of Clinical Investigation, 2018, 129, 583-597.	3.9	38
80	Adipsic hypernatremia without hypothalamic lesions accompanied by autoantibodies to subfornical organ. Brain Pathology, 2017, 27, 323-331.	2.1	29
81	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
82	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
83	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
84	Abnormal hematopoiesis and autoimmunity in human subjects with germline IKZF1 mutations. Journal of Allergy and Clinical Immunology, 2017, 140, 223-231.	1.5	99
85	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
86	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
87	Significant augmentation of regulatory T cell numbers occurs during the early neonatal period. Clinical and Experimental Immunology, 2017, 190, 268-279.	1.1	35
88	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
89	A Case with Spondyloenchondrodysplasia Treated with Growth Hormone. Frontiers in Endocrinology, 2017, 8, 157.	1.5	7
90	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

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91	Clinical characteristics of septo-optic dysplasia accompanied by congenital central hypothyroidism in Japan. Clinical Pediatric Endocrinology, 2017, 26, 207-213.	0.4	11
92	Characteristic clinical features of adipsic hypernatremia patients with subfornical organ-targeting antibody. Clinical Pediatric Endocrinology, 2017, 26, 197-205.	0.4	13
93	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
94	Chronic mucocutaneous candidiasis disease associated with inborn errors of ILâ€17 immunity. Clinical and Translational Immunology, 2016, 5, e114.	1.7	148
95	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
96	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
97	Heterozygous STAT1 gain-of-function mutations underlie an unexpectedly broad clinical phenotype. Blood, 2016, 127, 3154-3164.	0.6	465
98	Screening of MCAD deficiency in Japan: 16years' experience of enzymatic and genetic evaluation. Molecular Genetics and Metabolism, 2016, 119, 322-328.	0.5	21
99	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
100	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
101	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
102	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
103	Significance of ACADM mutations identified through newborn screening of MCAD deficiency in Japan. Molecular Genetics and Metabolism, 2016, 118, 9-14.	0.5	12
104	A Patient with CTLA-4 Haploinsufficiency Presenting Gastric Cancer. Journal of Clinical Immunology, 2016, 36, 28-32.	2.0	29
105	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
106	Successful Hematopoietic Stem Cell Transplantation Using an Immunosuppressive Conditioning Regimen in Ten Patients with Severe Congenital Neutropenia: A Single-Institute Experience. Blood, 2016, 128, 3688-3688.	0.6	0
107	Electrically induced ambipolar spin vanishments in carbon nanotubes. Scientific Reports, 2015, 5, 11859.	1.6	10
108	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

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109	Inherited DOCK2 Deficiency in Patients with Early-Onset Invasive Infections. New England Journal of Medicine, 2015, 372, 2409-2422.	13.9	169
110	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
111	Impairment of immunity to Candida and Mycobacterium in humans with bi-allelic RORC mutations. Science, 2015, 349, 606-613.	6.0	366
112	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
113	Life-threatening influenza and impaired interferon amplification in human IRF7 deficiency. Science, 2015, 348, 448-453.	6.0	389
114	Human TYK2 deficiency: Mycobacterial and viral infections without hyper-IgE syndrome. Journal of Experimental Medicine, 2015, 212, 1641-1662.	4.2	293
115	Human intracellular ISG15 prevents interferon-Î±/Î² over-amplification and auto-inflammation. Nature, 2015, 517, 89-93.	13.7	432
116	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
117	MSMD Patients with IFN-g-STAT1 Signaling Defect Present Enhanced Osteoclastogenesis and Bone Resorption. Blood, 2015, 126, 3591-3591.	0.6	3
118	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
119	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
120	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
121	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
122	Haploinsufficiency at the human IFNGR2 locus contributes to mycobacterial disease. Human Molecular Genetics, 2013, 22, 769-781.	1.4	58
123	Management of advancedâ€stage neuroblastoma in a patient with 21â€hydroxalase deficiency. Pediatrics International, 2013, 55, e96-9.	0.2	0
124	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
125	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
126	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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127	Development of rotating magnetic field system for the Î²-NMR method. Hyperfine Interactions, 2013, 220, 65-69.	0.2	3
128	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
129	<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
130	Heterozygosity for the Y701C STAT1 mutation in a multiplex kindred with multifocal osteomyelitis. Haematologica, 2013, 98, 1641-1649.	1.7	47
131	Simple diagnosis ofSTAT1gain-of-function alleles in patients with chronic mucocutaneous candidiasis. Journal of Leukocyte Biology, 2013, 95, 667-676.	1.5	77
132	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
133	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
134	Mycobacterial Disease and Impaired IFN-Î³ Immunity in Humans with Inherited ISG15 Deficiency. Science, 2012, 337, 1684-1688.	6.0	455
135	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
136	Dominant-negative STAT1 SH2 domain mutations in unrelated patients with mendelian susceptibility to mycobacterial disease. Human Mutation, 2012, 33, 1377-1387.	1.1	71
137	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
138	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
139	Clinical and Host Genetic Characteristics of Mendelian Susceptibility to Mycobacterial Diseases in Japan. Journal of Clinical Immunology, 2011, 31, 309-314.	2.0	24
140	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
141	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
142	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
143	DOCK8 deficiency impairs CD8 T cell survival and function in humans and mice. Journal of Experimental Medicine, 2011, 208, 2305-2320.	4.2	175
144	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

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145	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
146	Clinical Characteristics of Perinatal Lethal Hypophosphatasia: A Report of 6 Cases. Clinical Pediatric Endocrinology, 2010, 19, 7-13.	0.4	24
147	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
148	Cardiac infiltration in early-onset sarcoidosis associated with a novel heterozygous mutation, G481D, in CARD15. Rheumatology, 2009, 48, 706-707.	0.9	20
149	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
150	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
151	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
152	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
153	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
154	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
155	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
156	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
157	Abnormal Localization of Neutrophil Elastase in Patients with Severe Congenital Neutropenia.. Blood, 2006, 108, 1279-1279.	0.6	0
158	A comparison of the defective granulopoiesis in childhood cyclic neutropenia and in severe congenital neutropenia. Haematologica, 2005, 90, 1032-41.	1.7	22
159	Magnetic Ordering in Hexagonally Bonded Sheets with First-Row Elements. Physical Review Letters, 2001, 87, 146803.	2.9	369
160	Title is missing!. Japanese Journal of Food Microbiology, 2000, 17, 143-147.	0.3	2
161	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
162	Induction of mesothelioma by intraperitoneal injections of ferric saccharate in male Wistar rats. British Journal of Cancer, 1989, 60, 708-711.	2.9	62

#	Article	IF	Citations
163	Nephrotoxicity and its prevention by vitamin E in ferric nitrilotriacetate-promoted lipid. Lipids and Lipid Metabolism, 1987, 922, 28-33.	2.6	63