

Ryutaro Kira

List of Publications by Year in descending order

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Version: 2024-02-01

105
papers

2,344
citations

201674

27
h-index

243625

44
g-index

107
all docs

107
docs citations

107
times ranked

3258
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations of Neuronal Voltage-gated Na ⁺ Channel alpha1 Subunit Gene SCN1A in Core Severe Myoclonic Epilepsy in Infancy (SMEI) and in Borderline SMEI (SMEB). <i>Epilepsia</i> , 2004, 45, 140-148.	5.1	180
2	Altered white matter fractional anisotropy and social impairment in children with autism spectrum disorder. <i>Brain Research</i> , 2010, 1362, 141-149.	2.2	157
3	Clinical Features of Acute Flaccid Myelitis Temporally Associated With an Enterovirus D68 Outbreak: Results of a Nationwide Survey of Acute Flaccid Paralysis in Japan, August–December 2015. <i>Clinical Infectious Diseases</i> , 2018, 66, 653-664.	5.8	110
4	Genotype–phenotype correlations in alternating hemiplegia of childhood. <i>Neurology</i> , 2014, 82, 482-490.	1.1	93
5	Clinical study of childhood acute disseminated encephalomyelitis, multiple sclerosis, and acute transverse myelitis in Fukuoka Prefecture, Japan. <i>Brain and Development</i> , 2010, 32, 454-462.	1.1	92
6	Safety and efficacy of once-daily risdiplam in type 2 and non-ambulant type 3 spinal muscular atrophy (SUNFISH part 2): a phase 3, double-blind, randomised, placebo-controlled trial. <i>Lancet Neurology</i> , The, 2022, 21, 42-52.	10.2	89
7	Acute flaccid myelitis: cause, diagnosis, and management. <i>Lancet</i> , The, 2021, 397, 334-346.	13.7	88
8	Ineffective quinidine therapy in early onset epileptic encephalopathy with <i>KCNT1</i> mutation. <i>Annals of Neurology</i> , 2016, 79, 502-503.	5.3	68
9	CSF cytokine and chemokine profiles in acute disseminated encephalomyelitis. <i>Journal of Neuroimmunology</i> , 2006, 175, 52-58.	2.3	64
10	Genetic susceptibility to simple febrile seizures: Interleukin-1 β promoter polymorphisms are associated with sporadic cases. <i>Neuroscience Letters</i> , 2005, 384, 239-244.	2.1	59
11	A nationwide survey of pediatric acquired demyelinating syndromes in Japan. <i>Neurology</i> , 2016, 87, 2006-2015.	1.1	56
12	Rhombencephalitis and Coxsackievirus A16. <i>Emerging Infectious Diseases</i> , 2009, 15, 1689-1691.	4.3	54
13	Functional <i>MxA1</i> promoter polymorphism associated with subacute sclerosing panencephalitis. <i>Neurology</i> , 2004, 62, 457-460.	1.1	47
14	Comprehensive analysis of coding variants highlights genetic complexity in developmental and epileptic encephalopathy. <i>Nature Communications</i> , 2019, 10, 2506.	12.8	46
15	Interleukin-10 is associated with resistance to febrile seizures: Genetic association and experimental animal studies. <i>Epilepsia</i> , 2009, 50, 761-767.	5.1	45
16	PD1 as a common candidate susceptibility gene of subacute sclerosing panencephalitis. <i>Human Genetics</i> , 2010, 127, 411-419.	3.8	45
17	Association of toll-like receptor 3 gene polymorphism with subacute sclerosing panencephalitis. <i>Journal of NeuroVirology</i> , 2008, 14, 486-491.	2.1	44
18	Contribution of the Interleukin 4 Gene to Susceptibility to Subacute Sclerosing Panencephalitis. <i>Archives of Neurology</i> , 2002, 59, 822-7.	4.5	42

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19	Challenges in detecting genomic copy number aberrations using next-generation sequencing data and the eXome Hidden Markov Model: a clinical exome-first diagnostic approach. <i>Human Genome Variation</i> , 2016, 3, 16025.	0.7	38
20	Subependymal giant cell astrocytoma: clinical and neuroimaging features of four cases. <i>Journal of Clinical Neuroscience</i> , 2001, 8, 31-34.	1.5	36
21	Genomic backgrounds of Japanese patients with undiagnosed neurodevelopmental disorders. <i>Brain and Development</i> , 2019, 41, 776-782.	1.1	36
22	Diagnostic usefulness of diffusion-weighted magnetic resonance imaging in influenza-associated acute encephalopathy or encephalitis. <i>Brain and Development</i> , 2000, 22, 451-453.	1.1	35
23	Moyamoya Syndrome in a Splenectomized Patient With β^2 -Thalassemia Intermedia. <i>Journal of Child Neurology</i> , 2006, 21, 75-77.	1.4	34
24	Serial MRI findings of acute flaccid myelitis during an outbreak of enterovirus D68 infection in Japan. <i>Brain and Development</i> , 2019, 41, 443-451.	1.1	31
25	Nonsense mutation in exon 4 of human complement C9 gene is the major cause of Japanese complement C9 deficiency. <i>Human Genetics</i> , 1998, 102, 605-610.	3.8	29
26	Genetic susceptibility to febrile seizures: Case-control association studies. <i>Brain and Development</i> , 2010, 32, 57-63.	1.1	28
27	Neurotrophin-3 Levels in Cerebrospinal Fluid From Children With Bacterial Meningitis, Viral Meningitis, or Encephalitis. <i>Journal of Child Neurology</i> , 2000, 15, 19-21.	1.4	27
28	Magnetic resonance studies of brain lesions in patients with Kawasaki disease. <i>Brain and Development</i> , 2006, 28, 30-33.	1.1	27
29	Reversible Posterior Leukoencephalopathy Syndrome in Children With Cancers. <i>Journal of Pediatric Hematology/Oncology</i> , 2003, 25, 236-239.	0.6	23
30	Interleukin- 1β enhances susceptibility to hyperthermia-induced seizures in developing rats. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2009, 18, 211-214.	2.0	23
31	De novo variants in CUL3 are associated with global developmental delays with or without infantile spasms. <i>Journal of Human Genetics</i> , 2020, 65, 727-734.	2.3	23
32	Benign convulsion with mild gastroenteritis and benign familial infantile seizure. <i>Epilepsy Research</i> , 2006, 68, 269-271.	1.6	22
33	Founder effect of the C9 R95X mutation in Orientals. <i>Human Genetics</i> , 2003, 112, 244-248.	3.8	21
34	Molecular epidemiology of C9 deficiency heterozygotes with an Arg95Stop mutation of the C9 gene in Japan. <i>Journal of Human Genetics</i> , 1999, 44, 109-111.	2.3	20
35	Age-related changes of the MR appearance of CNS involvement in neurocutaneous melanosis complex. <i>Pediatric Radiology</i> , 2000, 30, 866-868.	2.0	20
36	A magnetoencephalographic study on development of the somatosensory cortex in infants. <i>NeuroReport</i> , 2001, 12, 3227-3231.	1.2	20

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37	Sjogren's syndrome-associated meningoencephalomyelitis: Cerebrospinal fluid cytokine levels and therapeutic utility of tacrolimus. <i>Journal of the Neurological Sciences</i> , 2008, 267, 182-186.	0.6	20
38	Involuntary movements and coma as the prognostic marker for acute encephalopathy with biphasic seizures and late reduced diffusion. <i>Journal of the Neurological Sciences</i> , 2016, 370, 39-43.	0.6	20
39	Surgical histopathology of limited dorsal myeloschisis with flat skin lesion. <i>Child's Nervous System</i> , 2019, 35, 119-128.	1.1	20
40	Epstein-Barr Virus-associated Meningoencephalomyelitis: Intrathecal Reactivation of the Virus in an Immunocompetent Child. <i>Journal of Child Neurology</i> , 2008, 23, 1072-1077.	1.4	17
41	Clinical and MRI characteristics of acute encephalopathy in congenital adrenal hyperplasia. <i>Journal of the Neurological Sciences</i> , 2011, 306, 91-93.	0.6	17
42	Liposteroid against refractory pulmonary haemorrhage in idiopathic pulmonary haemosiderosis. <i>European Journal of Pediatrics</i> , 1994, 153, 687-690.	2.7	16
43	Leucine-rich alpha-2 glycoprotein in the cerebrospinal fluid is a potential inflammatory biomarker for meningitis. <i>Journal of the Neurological Sciences</i> , 2018, 392, 51-55.	0.6	16
44	Reorganization of the primary somatosensory area in epilepsy associated with focal cortical dysplasia. <i>Developmental Medicine and Child Neurology</i> , 2000, 42, 839.	2.1	15
45	Neurotrophin-4 and glial cell line-derived neurotrophic factor in cerebrospinal fluid from meningitis/encephalitis patients. <i>Pediatric Neurology</i> , 2002, 27, 102-105.	2.1	14
46	Serum levels of matrix metalloproteinase-9 and tissue inhibitors of metalloproteinases 1 in subacute sclerosing panencephalitis. <i>Journal of the Neurological Sciences</i> , 2007, 252, 45-48.	0.6	13
47	Early onset of moyamoya syndrome in a Down syndrome patient with the genetic variant RNF213 p.R4810K. <i>Brain and Development</i> , 2015, 37, 822-824.	1.1	13
48	A case of pontine tegmental cap dysplasia with comorbidity of oculoauriculovertebral spectrum. <i>Brain and Development</i> , 2015, 37, 171-174.	1.1	13
49	High-intensity basal ganglia lesions on T1-weighted images in two toddlers with elevated blood manganese with portosystemic shunts. <i>Neuroradiology</i> , 1999, 41, 195-198.	2.2	12
50	MR choroid plexus sign of iron overload. <i>Neurology</i> , 2000, 55, 1340-1340.	1.1	12
51	Analysis of MxA, IL-4, and IRF-1 Genes in Filipino Patients with Subacute Sclerosing Panencephalitis. <i>Neuropediatrics</i> , 2006, 37, 222-228.	0.6	12
52	Strategy in short-term memory for pictures in childhood: A near-infrared spectroscopy study. <i>NeuroImage</i> , 2011, 54, 2394-2400.	4.2	11
53	Parental age and child growth and development: Child health check-up data. <i>Pediatrics International</i> , 2011, 53, 709-714.	0.5	11
54	Deletions of SCN2A and SCN3A genes in a patient with West syndrome and autistic spectrum disorder. Seizure: the Journal of the British Epilepsy Association, 2018, 60, 91-93.	2.0	10

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55	A case of childhood stiff-person syndrome with striatal lesions: A possible entity distinct from the classical adult form. <i>Brain and Development</i> , 2013, 35, 575-578.	1.1	9
56	The clinical characteristics of pediatric coronavirus disease 2019 in 2020 in Japan. <i>Pediatrics International</i> , 2022, 64, .	0.5	9
57	Fulminant sepsis/meningitis due to <i>Haemophilus influenzae</i> in a protein C-deficient heterozygote treated with activated protein C therapy. <i>European Journal of Pediatrics</i> , 2009, 168, 673-677.	2.7	8
58	Global Central Nervous System Atrophy in Spinal Muscular Atrophy Type 0. <i>Annals of Neurology</i> , 2019, 86, 801-802.	5.3	8
59	Long surviving classical Menkes disease treated with weekly intravenous copper therapy. <i>Journal of Trace Elements in Medicine and Biology</i> , 2019, 54, 172-174.	3.0	8
60	Clinical and genetic characteristics of patients with Doose syndrome. <i>Epilepsia Open</i> , 2020, 5, 442-450.	2.4	8
61	A novel R275X mutation of the SLC25A15 gene in a Japanese patient with the HHH syndrome. <i>Brain and Development</i> , 2006, 28, 332-335.	1.1	7
62	EPSTEIN-BARR VIRUS LOAD IN CEREBROSPINAL FLUID OF PATIENTS WITH CHRONIC ACTIVE EPSTEIN-BARR VIRUS INFECTION. <i>Pediatric Infectious Disease Journal</i> , 2008, 27, 1027-1030.	2.0	7
63	Venous anomaly analogous to vertical embryonic positioning of the straight sinus associated with atretic cephalocele at the suboccipital region. <i>Child's Nervous System</i> , 2017, 33, 179-182.	1.1	7
64	West Syndrome in an Infant With Vitamin B12 Deficiency Born to Autoantibodies Positive Mother. <i>Frontiers in Pediatrics</i> , 2019, 7, 531.	1.9	7
65	A nation-wide survey of Japanese pediatric MOG antibody-associated diseases. <i>Brain and Development</i> , 2021, 43, 705-713.	1.1	7
66	The relationship between retrieval success and task performance during the recognition of meaningless shapes: An event-related near-infrared spectroscopy study. <i>Neuroscience Research</i> , 2007, 59, 191-198.	1.9	6
67	Decision-making dilemmas of paediatricians: a qualitative study in Japan. <i>BMJ Open</i> , 2019, 9, e026579.	1.9	6
68	De novo p.G696S mutation in COL4A1 causes intracranial calcification and late-onset cerebral hemorrhage: A case report and review of the literature. <i>European Journal of Medical Genetics</i> , 2020, 63, 103825.	1.3	6
69	The recurrent postzygotic pathogenic variant p.Glu47Lys in RHOA causes a novel recognizable neuroectodermal phenotype. <i>Human Mutation</i> , 2020, 41, 591-599.	2.5	6
70	Isolated cranial neuritis of the oculomotor nerve: Expanding the MOG phenotype?. <i>Multiple Sclerosis and Related Disorders</i> , 2020, 41, 102040.	2.0	6
71	Liposteroid against refractory pulmonary haemorrhage in idiopathic pulmonary haemosiderosis. <i>European Journal of Pediatrics</i> , 1994, 153, 687-690.	2.7	6
72	Bilateral basal ganglial necrosis after allogeneic bone marrow transplantation in a child with Kostmann syndrome. <i>Bone Marrow Transplantation</i> , 1999, 23, 515-517.	2.4	5

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73	Acute disseminated encephalomyelitis in a female with hereditary neuropathy with susceptibility to pressure palsy. <i>Pediatric Neurology</i> , 2000, 22, 302-304.	2.1	5
74	Nasal Dermal Sinus Associated with a Dumbbell-Shaped Dermoid: A Case Report. <i>Journal of Neurological Surgery Reports</i> , 2016, 77, e94-e97.	0.6	5
75	Mutations in NSD1 and NFIX in Three Patients with Clinical Features of Sotos Syndrome and Malan Syndrome. <i>Journal of Pediatric Genetics</i> , 2017, 06, 234-237.	0.7	5
76	Acute Flaccid Myelitis With Neuroradiological Finding of Brachial Plexus Swelling. <i>Pediatric Neurology</i> , 2020, 109, 85-88.	2.1	5
77	Gene expression profiles in peripheral blood mononuclear cells from patients with subacute sclerosing panencephalitis using oligonucleotide microarrays. <i>Journal of NeuroVirology</i> , 2005, 11, 299-305.	2.1	4
78	Mulberries in the urine: a tell-tale sign of Fabry disease. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 745-746.	3.6	4
79	Sequential radiologic findings in osteopathia striata with cranial sclerosis. <i>Diagnostic and Interventional Imaging</i> , 2019, 100, 529-531.	3.2	4
80	An acute encephalopathy with reduced diffusion in BRAF-associated cardio-facio-cutaneous syndrome. <i>Brain and Development</i> , 2019, 41, 378-381.	1.1	4
81	Effect of a vaccine information statement (VIS) on immunization status and parental knowledge, attitudes, and beliefs regarding infant immunization in Japan. <i>Vaccine</i> , 2020, 38, 8049-8054.	3.8	4
82	Comprehensive genetic analysis confers high diagnostic yield in 16 Japanese patients with corpus callosum anomalies. <i>Journal of Human Genetics</i> , 2021, 66, 1061-1068.	2.3	4
83	Magnetoencephalographic analysis of hypsarrhythmia in West Syndrome. <i>Journal of Epilepsy</i> , 1997, 10, 131-138.	0.4	3
84	Complement component 9 deficiency is not a susceptibility factor for SLE. <i>Lupus</i> , 2000, 9, 456-457.	1.6	3
85	Autopsy Case of Later-Onset Pontocerebellar Hypoplasia Type 1: Pontine Atrophy and Pyramidal Tract Involvement. <i>Journal of Child Neurology</i> , 2010, 25, 1429-1434.	1.4	3
86	Periodic Epileptiform Discharges in Children With Advanced Stages of Progressive Myoclonic Epilepsy. <i>Clinical EEG and Neuroscience</i> , 2016, 47, 317-323.	1.7	3
87	Novel A178P mutation in <i>SLC16A2</i> in a patient with Allan-Herndon-Dudley syndrome. <i>Congenital Anomalies (discontinued)</i> , 2018, 58, 143-144.	0.6	3
88	Disseminated cortical and subcortical lesions in neonatal enterovirus 71 encephalitis. <i>Journal of NeuroVirology</i> , 2020, 26, 790-792.	2.1	3
89	Three-Year Longitudinal Motor Function and Disability Level of Acute Flaccid Myelitis. <i>Pediatric Neurology</i> , 2021, 116, 14-19.	2.1	3
90	Incidence and risk factors of acute encephalopathy with biphasic seizures in febrile status epilepticus. <i>Brain and Development</i> , 2022, 44, 36-43.	1.1	3

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91	Neurodevelopmental Outcomes of High-Risk Preterm Infants. <i>Neurology: Clinical Practice</i> , 2021, 11, 398-405.	1.6	3
92	Description of Restrictively Defined Acute Flaccid Myelitis. <i>JAMA Pediatrics</i> , 2019, 173, 702.	6.2	2
93	Subcortical axonal loss with glial reactions following partial status epilepticus with neuroradiological findings of reduced subcortical diffusion. <i>Neurological Sciences</i> , 2019, 40, 851-855.	1.9	2
94	Infantile spasms and early-onset progressive polycystic renal lesions associated with TSC2/PKD1 contiguous gene deletion syndrome. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2021, 86, 82-84.	2.0	2
95	Favorable outcomes of interferon- β and ribavirin treatment for a male with subacute sclerosing panencephalitis. <i>Journal of Neuroimmunology</i> , 2021, 358, 577656.	2.3	2
96	Case Report: Acute Fulminant Cerebral Edema With Perivascular Abnormalities Related to Kawasaki Disease. <i>Frontiers in Pediatrics</i> , 2021, 9, 732110.	1.9	2
97	Monogenic causes of pigmentary mosaicism. <i>Human Genetics</i> , 2022, , .	3.8	2
98	Cytotoxic lesion of the corpus callosum exclusively at the genu in a case of callosal hypogenesis. <i>Journal of Neuroradiology</i> , 2019, 46, 222-223.	1.1	1
99	Huge Adenomatous Goiter Reaching the Aortic Arch with Elevated Thyroglobulin. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 1997, 10, 641-4.	0.9	0
100	Advanced paternal age and impaired childhood cognitive development: Reply. <i>Pediatrics International</i> , 2012, 54, 582-582.	0.5	0
101	Water Immersion-Induced Skin Wrinkling Test in Complex Regional Pain Syndrome. <i>Pediatric Neurology</i> , 2015, 52, 649-650.	2.1	0
102	Influenza-associated encephalopathy with focal late reduced diffusion circumscribing a pre-existing cortical lesion. <i>Journal of Neuroradiology</i> , 2020, 47, 241-243.	1.1	0
103	Brain-sparing cord blood transplantation for the borderline stage of adrenoleukodystrophy. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 28, 100778.	1.1	0
104	Clinical and electrophysiological features of acute flaccid myelitis: A national cohort study. <i>Clinical Neurophysiology</i> , 2021, 132, 2456-2463.	1.5	0
105	Letter to the Editor. <i>Journal of Paediatrics and Child Health</i> , 2020, 56, 348-349.	0.8	0