Masaya Kubota

List of Publications by Year
in descending order

104	3,421	147726	155592
papers	citations	31 h-index	55 g-index

108	108	108	4891
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Ketogenic Diet for KARS-Related Mitochondrial Dysfunction and Progressive Leukodystrophy. Neuropediatrics, 2022, 53, 065-068.	0.3	3
2	Aphasia and a Dual-Stream Language Model in a 4-Year-Old Female with Landauâ€“Kleffner Syndrome. Neuropediatrics, 2022, 53, 295-298.	0.3	1
3	Interventions for Shiga toxin-producing Escherichia coli gastroenteritis and risk of hemolytic uremic syndrome: A population-based matched case control study. PLoS ONE, 2022, 17, e0263349.	1.1	2
4	Differentiating early clinical features of Panayiotopoulos syndrome from acute encephalopathy. Brain and Development, 2022, 44, 386-390.	0.6	3
5	Changes in Magnetic Resonance Signal Fluctuation in Superior Sagittal Sinus: Deterioration of Arteriolar Vasomotor Function of Young Smokers. Tomography, 2022, 8, 657-666.	0.8	0
6	Growth charts in Cockayne syndrome type 1 and type 2. European Journal of Medical Genetics, 2021, 64, 104105.	0.7	4
7	Use of antipyretics for preventing febrile seizure recurrence in children: a systematic review and meta-analysis. European Journal of Pediatrics, 2021, 180, 987-997.	1.3	17
8	Neurological complications after living-donor liver transplantation in children. Brain and Development, 2021, 43, 637-643.	0.6	3
9	Recurrent acute necrotizing encephalopathy in a boy with RANBP2 mutation and thermolabile CPT2 variant: The first case of ANE1 in Japan. Brain and Development, 2021, 43, 873-878.	0.6	10
10	The effect of the guidelines for management of febrile seizures 2015 on clinical practices: Nationwide survey in Japan. Brain and Development, 2020, 42, 28-34.	0.6	6
11	Pyridoxal in the Cerebrospinal Fluid May Be a Better Indicator of Vitamin B6â€“dependent Epilepsy Than Pyridoxal 5â€²-Phosphate. Pediatric Neurology, 2020, 113, 33-41.	1.0	6
12	Neuroimaging evaluation and successful treatment by using directional deep brain stimulation and levodopa in a patient with GNAO1-associated movement disorder: A case report. Journal of the Neurological Sciences, 2020, 411, 116710.	0.3	15
13	Case-control association study of rare nonsynonymous variants of SCN1A and KCNQ2 in acute encephalopathy with biphasic seizures and late reduced diffusion. Journal of the Neurological Sciences, 2020, 414, 116808.	0.3	11
14	Everolimus for epilepsy and autism spectrum disorder in tuberous sclerosis complex: EXIST-3 substudy in Japan. Brain and Development, 2019, 41, 1-10.	0.6	60
15	Thermolabile polymorphism of carnitine palmitoyltransferase 2: A genetic risk factor of overall acute encephalopathy. Brain and Development, 2019, 41, 862-869.	0.6	10
16	Comprehensive analysis of coding variants highlights genetic complexity in developmental and epileptic encephalopathy. Nature Communications, 2019, 10, 2506.	5.8	46
17	Early administration of vitamins B1 and B6 and l-carnitine prevents a second attack of acute encephalopathy with biphasic seizures and late reduced diffusion: A case control study. Brain and Development, 2019, 41, 618-624.	0.6	21
18	Digital Amputation by Congenital Insensitivity to Pain with Anhidrosis. Journal of Pediatrics, 2019, 208, 290.	0.9	2

#	Article	IF	Citations
19	A case of Niemann-Pick disease type C with neonatal liver failure initially diagnosed as neonatal hemochromatosis. Brain and Development, 2019, 41, 460-464.	0.6	14
20	Cockayne Syndrome: Clinical Aspects. , 2019, , 115-132.		0
21	Severe anti-GAD antibody-associated encephalitis after stem cell transplantation. Brain and Development, 2019, 41, 301-304.	0.6	19
22	Clinical Features of Acute Flaccid Myelitis Temporally Associated With an Enterovirus D68 Outbreak: Results of a Nationwide Survey of Acute Flaccid Paralysis in Japan, Augustâ€“December 2015. Clinical Infectious Diseases, 2018, 66, 653-664.	2.9	110
23	Functional and clinical relevance of novel mutations in a large cohort of patients with Cockayne syndrome. Journal of Medical Genetics, 2018, 55, 329-343.	1.5	55
24	Ictal pouting: kabuki visage or chapeau de gendarme?. Practical Neurology, 2018, 18, 410-412.	0.5	9
25	Cases of prolonged mild disturbance of consciousness and acute encephalopathy. Pediatrics International, 2018, 61, 175-179.	0.2	0
26	Therapeutic regimen of l-arginine for MELAS: 9-year, prospective, multicenter, clinical research. Journal of Neurology, 2018, 265, 2861-2874.	1.8	56
27	Novel heterozygous deletion mutation c.821delC in the <scp>AAA</scp> domain of <scp>BCS</scp>1L underlies BjÃ¶rnstad syndrome. Journal of Dermatology, 2017, 44, e111-e112.	0.6	5
28	Acute myeloid leukemiaâ€associated DNMT3A p.Arg882His mutation in a patient with Tattonâ€Brownâ€“Rahman overgrowth syndrome as a constitutional mutation. American Journal of Medical Genetics, Part A, 2017, 173, 250-253.	0.7	36
29	Sequential analysis of variable markers for predicting outcomes in pediatric patients with acute liver failure. Hepatology Research, 2017, 47, 1241-1251.	1.8	10
30	Simultaneous measurement of monoamine metabolites and 5-methyltetrahydrofolate in the cerebrospinal fluid of children. Clinica Chimica Acta, 2017, 465, 5-10.	0.5	8
31	A Simplified Electroencephalogram Monitoring System in the Emergency Room. Pediatric Emergency Care, 2017, Publish Ahead of Print, 487-492.	0.5	6
32	New guidelines for management of febrile seizures in Japan. Brain and Development, 2017, 39, 2-9.	0.6	60
33	Living-Donor Liver Transplantation From a Heterozygous Parent for Infantile Refsum Disease. Pediatrics, 2016, 137, e20153102-e20153102.	1.0	15
34	Reply to the letter: â€œThe diagnostic value of MRI in pediatric chronic inflammatory demyelinating polyradiculoneuropathyâ€. Brain and Development, 2016, 38, 174.	0.6	0
35	Manifestations and characteristics of congenital adrenal hyperplasia-associated encephalopathy. Brain and Development, 2016, 38, 638-647.	0.6	17
36	Novel homozygous mutation, c.400C>T (p.Arg134*), in the <scp>PVRL</scp>1 gene underlies cleft lip/palateâ€ectodermal dysplasia syndrome in an Asian patient. Journal of Dermatology, 2015, 42, 715-719.	0.6	11

#	Article	IF	Citations
37	Characteristic MRI features of chronic inflammatory demyelinating polyradiculoneuropathy. Brain and Development, 2015, 37, 894-896.	0.6	21
38	Predictive score for early diagnosis of acute encephalopathy with biphasic seizures and late reduced diffusion (AESD). Journal of the Neurological Sciences, 2015, 358, 62-65.	0.3	37
39	Hereditary sensory and autonomic neuropathy types <scp>IV</scp> and <scp>V</scp> in <scp>J</scp>apan. Pediatrics International, 2015, 57, 30-36.	0.2	30
40	Microarray analysis of 50 patients reveals the critical chromosomal regions responsible for 1p36 deletion syndrome-related complications. Brain and Development, 2015, 37, 515-526.	0.6	43
41	Nationwide survey of Cockayne syndrome in Japan: Incidence, clinical course and prognosis. Pediatrics International, 2015, 57, 339-347.	0.2	35
42	Analysis of a child who developed abnormal neuropsychiatric symptoms after administration of oseltamivir: a case report. BMC Neurology, 2015, 15, 130.	0.8	15
43	Missense mutations in sodium channel SCN1A and SCN2A predispose children to encephalopathy with severe febrile seizures. Epilepsy Research, 2015, 117, 1-6.	0.8	31
44	Leukoencephalopathy associated with 11q24 deletion involving the gene encoding hepatic and glial cell adhesion molecule in two patients. European Journal of Medical Genetics, 2015, 58, 492-496.	0.7	18
45	Clinical and genetic features of acute encephalopathy in children taking theophylline. Brain and Development, 2015, 37, 463-470.	0.6	19
46	Changes in Cerebrospinal Fluid Biomarkers in Human Herpesvirus-6-Associated Acute Encephalopathy/Febrile Seizures. Mediators of Inflammation, 2014, 2014, 1-8.	1.4	17
47	Central pontine myelinolysis following pediatric living donor liver transplantation: A case report and review of literature. Pediatric Transplantation, 2014, 18, E120-3.	0.5	9
48	Serum and CSF biomarkers in acute pediatric neurological disorders. Brain and Development, 2014, 36, 489-495.	0.6	10
49	<scp>C</scp>anavan disease: Clinical features and recent advances in research. Pediatrics International, 2014, 56, 477-483.	0.2	61
50	Novel compound heterozygous PIGT mutations caused multiple congenital anomalies-hypotonia-seizures syndrome 3. Neurogenetics, 2014, 15, 193-200.	0.7	61
51	Early onset epileptic encephalopathy caused by de novo <scp>SCN</scp>8A mutations. Epilepsia, 2014, 55, 994-1000.	2.6	142
52	De Novo Mutations in GNAO1, Encoding a GÎ±o Subunit of Heterotrimeric G Proteins, Cause Epileptic Encephalopathy. American Journal of Human Genetics, 2013, 93, 496-505.	2.6	187
53	Clinical spectrum of early onset epileptic encephalopathies caused by <scp>KCNQ2</scp> mutation. Epilepsia, 2013, 54, 1282-1287.	2.6	195
54	ADORA2A polymorphism predisposes children to encephalopathy with febrile status epilepticus. Neurology, 2013, 80, 1571-1576.	1.5	71

#	Article	IF	Citations
55	Epidemiology of hereditary sensory and autonomic neuropathy type IV and V in Japan. American Journal of Medical Genetics, Part A, 2013, 161, 871-874.	0.7	28
56	MLL2 and KDM6A mutations in patients with Kabuki syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 2234-2243.	0.7	148
57	A Case of Osteogenesis Imperfecta Type II Caused by a Novel COL1A2 Gene Mutation: Endoscopic Third Ventriculostomy to Prevent Hydrocephalus. Neuropediatrics, 2012, 43, 225-228.	0.3	2
58	Langerhans cell histiocytosis with disequilibrium. Auris Nasus Larynx, 2012, 39, 627-630.	0.5	6
59	Thermolabile CPT II variants and low blood ATP levels are closely related to severity of acute encephalopathy in Japanese children. Brain and Development, 2012, 34, 20-27.	0.6	43
60	Oxidative stress in patients with clinically mild encephalitis/encephalopathy with a reversible splenial lesion (MERS). Brain and Development, 2012, 34, 124-127.	0.6	60
61	Epidemiology of acute encephalopathy in Japan, with emphasis on the association of viruses and syndromes. Brain and Development, 2012, 34, 337-343.	0.6	328
62	Mutations of the SCN1A gene in acute encephalopathy. Epilepsia, 2012, 53, 558-564.	2.6	47
63	Brain vascular changes in Cockayne syndrome. Neuropathology, 2012, 32, 113-117.	0.7	14
64	Polymicrogyria and infantile spasms in a patient with 1p36 deletion syndrome. Brain and Development, 2011, 33, 437-441.	0.6	14
65	Carnitine palmitoyl transferase II polymorphism is associated with multiple syndromes of acute encephalopathy with various infectious diseases. Brain and Development, 2011, 33, 512-517.	0.6	67
66	Critical illness polyneuropathy after septic peritonitis in a boy with nephrotic syndrome. Pediatric Nephrology, 2010, 25, 1771-1772.	0.9	0
67	Comprehensive genetic analyses of PLP1 in patients with Pelizaeusâ€“Merzbacher disease applied by array-CGH and fiber-FISH analyses identified new mutations and variable sizes of duplications. Brain and Development, 2010, 32, 171-179.	0.6	28
68	The axonal damage marker tau protein in the cerebrospinal fluid is increased in patients with acute encephalopathy with biphasic seizures and late reduced diffusion. Brain and Development, 2010, 32, 435-439.	0.6	40
69	STXBP1 mutations in early infantile epileptic encephalopathy with suppressionâ€burst pattern. Epilepsia, 2010, 51, 2397-2405.	2.6	133
70	Highâ€dose lorazepam for convulsive status epilepticus in an infant with holoprosencephaly. Pediatrics International, 2010, 52, 664-667.	0.2	1
71	Maternal Uniparental Disomy 14 Syndrome Demonstrates Prader-Willi Syndrome-Like Phenotype. Journal of Pediatrics, 2009, 155, 900-903.e1.	0.9	46
72	Serum and cerebrospinal fluid levels of cytokines in acute encephalopathy associated with human herpesvirus-6 infection. Brain and Development, 2009, 31, 731-738.	0.6	86

#	Article	IF	Citations
73	Serum and CSF levels of cytokines in acute encephalopathy following prolonged febrile seizures. Brain and Development, 2008, 30, 47-52.	0.6	95
74	Early hemispherotomy in a patient with multilobar cortical dysplasia with intractable seizure: Clinicalâ€“neurophysiological study. European Journal of Paediatric Neurology, 2008, 12, 516-520.	0.7	4
75	Roles of matrix metalloproteinase-9 and tissue inhibitors of metalloproteinases 1 in acute encephalopathy following prolonged febrile seizures. Journal of the Neurological Sciences, 2008, 266, 126-130.	0.3	63
76	Steroid-Responsive Chronic Cerebellitis With Positive Glutamate Receptor Î´2 Antibody. Journal of Child Neurology, 2008, 23, 228-230.	0.7	20
77	A case of Panayiotopoulos syndrome showing an atypical course. Seizure: the Journal of the British Epilepsy Association, 2006, 15, 643-648.	0.9	6
78	Increased right auditory cortex activity in absolute pitch possessors. NeuroReport, 2005, 16, 1775-1779.	0.6	4
79	Severe gastrointestinal dysmotility in a patient with congenital myopathy: causal relationship to decrease of interstitial cells of Cajal. Brain and Development, 2005, 27, 447-450.	0.6	10
80	A magnetoencephalographic study of negative myoclonus in a patient with atypical benign partial epilepsy. Seizure: the Journal of the British Epilepsy Association, 2005, 14, 28-32.	0.9	14
81	Patients with benign rolandic epilepsy have a longer duration of somatosensory evoked high-frequency oscillations. Pediatrics International, 2004, 46, 631-634.	0.2	10
82	Beneficial effect of l-arginine for stroke-like episode in MELAS. Brain and Development, 2004, 26, 481-483.	0.6	64
83	A magneto- encephalographic study of astatic seizure in myoclonic astatic epilepsy. Pediatric Neurology, 2004, 31, 207-210.	1.0	4
84	Children are sensitive to averted eyes at the earliest stage of gaze processing. NeuroReport, 2004, 15, 1345-1348.	0.6	14
85	N100m in adults possessing absolute pitch. NeuroReport, 2004, 15, 1383-1386.	0.6	9
86	Magnetoencephalographic Analysis of Rolandic Discharges in a Patient With Rolandic Epilepsy Associated With Oromotor Deficits. Journal of Child Neurology, 2004, 19, 456-459.	0.7	5
87	N100m in children possessing absolute pitch. NeuroReport, 2003, 14, 899-903.	0.6	7
88	People with absolute pitch process tones with producing P300. Neuroscience Letters, 2002, 330, 247-250.	1.0	25
89	Varicella-associated acute necrotizing encephalopathy with a good prognosis. Brain and Development, 2001, 23, 54-57.	0.6	20
90	The gene copy ratios of SMN1/SMN2 in Japanese carriers with type I spinal muscular atrophy. Brain and Development, 2001, 23, 321-326.	0.6	2

#	Article	IF	Citations
91	Long-term ventilator support in patients with Werdnig-Hoffmann disease. Pediatrics International, 2000, 42, 359-363.	0.2	34
92	Magnetoencephalographic Study of Giant Somatosensory Evoked Responses in Patients With Rolandic Epilepsy. Journal of Child Neurology, 2000, 15, 370-379.	0.7	22
93	Zonisamide - induced urinary lithiasis in patients with intractable epilepsy. Brain and Development, 2000, 22, 230-233.	0.6	61
94	Control of a methicillin-resistant Staphylococcus aureus outbreak in a neonatal intensive care unit by unselective use of nasal mupirocin ointment. Journal of Hospital Infection, 2000, 46, 123-129.	1.4	67
95	New ocular movement detector system as a communication tool in ventilator-assisted Werdnigâ€“Hoffmann disease. Developmental Medicine and Child Neurology, 2000, 42, 61.	1.1	33
96	Preferential involvement of U-fibers in human herpesvirus 6-associated acute encephalopathy. Annals of Neurology, 1999, 45, 684-684.	2.8	2
97	A case of parietal lobe epilepsy with distinctive clinical and neuroradiological features. Brain and Development, 1998, 20, 179-182.	0.6	9
98	Olivopontocerebellar atrophy of neonatal onset with muscle hypertonia in two siblings. Neuropathology, 1997, 17, 225-229.	0.7	2
99	Temporary improvement of neurological symptoms with gammaglobulin therapy in a boy with adrenoleukodystrophy. Brain and Development, 1996, 18, 119-121.	0.6	8
100	Periventricular leukomalacia associated with hypocarbia. Pediatrics International, 1996, 38, 57-60.	0.2	11
101	Reduction of seizure frequency with clomipramine in patients with complex partial seizures. Brain and Development, 1995, 17, 291-293.	0.6	17
102	Erythromycin improves gastrointestinal motility in extremely low birthweight infants. Pediatrics International, 1994, 36, 198-201.	0.2	30
103	Severe Failure to Thrive in an Infant Born to a Mother with Albright Hereditary Osteodysplasia (AHO). Clinical Pediatric Endocrinology, 1994, 3, 232-233.	0.4	0
104	Adrenoleukodystrophy Associated with Cerebral Arteriovenous Malformation. Pediatrics International, 1990, 32, 543-547.	0.2	3