Shinichi Hirose

List of Publications by Year in descending order

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236 papers 7,362 citations

50276 46 h-index 79698 73 g-index

246 all docs 246 docs citations

246 times ranked 7763 citing authors

#	Article	IF	Citations
1	Invasive candidiasis in a neonatal intensive care unit in Fukuoka. Pediatrics International, 2022, 64, .	0.5	1
2	The effectiveness of intravenous benzodiazepine for status epilepticus in Dravet syndrome. Brain and Development, 2022, , .	1.1	0
3	Burden of seizures and comorbidities in patients with epilepsy: a survey based on the tertiary hospitalâ€based Epilepsy Syndrome Registry in Japan. Epileptic Disorders, 2022, 24, 82-94.	1.3	1
4	Newborn screening for Gaucher disease in Japan. Molecular Genetics and Metabolism Reports, 2022, 31, 100850.	1.1	4
5	CUX2 deficiency causes facilitation of excitatory synaptic transmission onto hippocampus and increased seizure susceptibility to kainate. Scientific Reports, 2022, 12, 6505.	3.3	8
6	Comparative characterization of PCDH19 missense and truncating variants in PCDH19-related epilepsy. Journal of Human Genetics, 2021, 66, 569-578.	2.3	16
7	Genetics and gene therapy in Dravet syndrome. Epilepsy and Behavior, 2021, , 108043.	1.7	9
8	Impaired neuronal activity and differential gene expression in <i>STXBP1</i> encephalopathy patient iPSC-derived GABAergic neurons. Human Molecular Genetics, 2021, 30, 1337-1348.	2.9	11
9	Inhibitory synaptic transmission is impaired at higher extracellular Ca2+ concentrations in Scn1a+/â^' mouse model of Dravet syndrome. Scientific Reports, 2021, 11, 10634.	3.3	9
10	Physical, cognitive, and social status of patients with urea cycle disorders in Japan. Molecular Genetics and Metabolism Reports, 2021, 27, 100724.	1.1	9
11	MicroRNAs miR-4535 and miR-1915-5p in amniotic fluid as predictive biomarkers for chorioamnionitis. Future Science OA, 2021, 7, FSO686.	1.9	2
12	Current medicoâ€psychoâ€social conditions of patients with West syndrome in Japan. Epileptic Disorders, 2021, 23, 579-589.	1.3	0
13	Genome sequencing and RNAâ€seq analyses of mitochondrial complex I deficiency revealed <i>Alu</i> insertionâ€mediated deletion in <i>NDUFV2</i> . Human Mutation, 2021, 42, 1422-1428.	2.5	4
14	Current status of newborn screening for Pompe disease in Japan. Orphanet Journal of Rare Diseases, 2021, 16, 516.	2.7	11
15	Clinical features of early myoclonic encephalopathy caused by a CDKL5 mutation. Brain and Development, 2020, 42, 73-76.	1.1	5
16	Coffinâ€Siris syndrome with bilateral macular dysplasia caused by a novel exonic deletion in ARID1B. Congenital Anomalies (discontinued), 2020, 60, 189-193.	0.6	2
17	Application of induced pluripotent stem cells in epilepsy. Molecular and Cellular Neurosciences, 2020, 108, 103535.	2.2	13
18	A recurrent <i>PJA1</i> variant in trigonocephaly and neurodevelopmental disorders. Annals of Clinical and Translational Neurology, 2020, 7, 1117-1131.	3.7	18

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19	Establishment of human induced pluripotent stem cells derived from skin cells of a patient with Dravet syndrome. Stem Cell Research, 2020, 47, 101857.	0.7	5
20	Newborn screening for Fabry disease in the western region of Japan. Molecular Genetics and Metabolism Reports, 2020, 22, 100562.	1.1	32
21	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.	0.6	11
22	Epigenetics explained: a topic "primer―for the epilepsy community by the ILAE Genetics/Epigenetics Task Force. Epileptic Disorders, 2020, 22, 127-141.	1.3	17
23	Characteristics of <i><scp>KCNQ</scp>2</i> variants causing either benign neonatal epilepsy or developmental and epileptic encephalopathy. Epilepsia, 2019, 60, 1870-1880.	5.1	66
24	Heart rate variability in a patient with alternating hemiplegia. Intractable and Rare Diseases Research, 2019, 8, 134-137.	0.9	0
25	Copy number variation analysis in 83 children with early-onset developmental and epileptic encephalopathy after targeted resequencing of a 109-epilepsy gene panel. Journal of Human Genetics, 2019, 64, 1097-1106.	2.3	8
26	Different types of suppression-burst patterns in patients with epilepsy of infancy with migrating focal seizures (EIMFS). Seizure: the Journal of the British Epilepsy Association, 2019, 65, 118-123.	2.0	5
27	PRRT2 mutations in Japanese patients with benign infantile epilepsy and paroxysmal kinesigenic dyskinesia. Seizure: the Journal of the British Epilepsy Association, 2019, 71, 1-5.	2.0	16
28	Newborn screening for Pompe disease in Japan: report and literature review of mutations in the GAA gene in Japanese and Asian patients. Journal of Human Genetics, 2019, 64, 741-755.	2.3	30
29	Efficacy and tolerability of perampanel in pediatric patients with Dravet syndrome. Epilepsy Research, 2019, 154, 34-38.	1.6	27
30	miR-124 dosage regulates prefrontal cortex function by dopaminergic modulation. Scientific Reports, 2019, 9, 3445.	3.3	32
31	Quinidine therapy and therapeutic drug monitoring in four patients with <i>KCNT1</i> mutations. Epileptic Disorders, 2019, 21, 48-54.	1.3	22
32	Two Cases of Juvenile Dermatomyositis: Case Series and Review of the Literature. Nishinihon Journal of Dermatology, 2019, 81, 14-17.	0.0	0
33	Generation of D1-1 TALEN isogenic control cell line from Dravet syndrome patient iPSCs using TALEN-mediated editing of the SCN1A gene. Stem Cell Research, 2018, 28, 100-104.	0.7	15
34	Further characterization of CAPOS/CAOS syndrome with the Glu818Lys mutation in the ATP1A3 gene: A case report. Brain and Development, 2018, 40, 576-581.	1.1	8
35	Somatic mosaic deletions involving <i>SCN1A</i> cause Dravet syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 657-662.	1.2	17
36	Long-term follow up of an adult with alternating hemiplegia of childhood and a p.Gly755Ser mutation in the ATP1A3 gene. Brain and Development, 2018, 40, 226-228.	1.1	6

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37	Reappraising newborn screening for cobalamin C disorder. Pediatrics and Neonatology, 2018, 59, 415-417.	0.9	O
38	Variant Intestinal-Cell Kinase in Juvenile Myoclonic Epilepsy. New England Journal of Medicine, 2018, 378, 1018-1028.	27.0	36
39	CACNA1A -related early-onset encephalopathy with myoclonic epilepsy: A case report. Brain and Development, 2018, 40, 130-133.	1.1	20
40	Recurrent autonomic and sensory neuropathy in a patient with anti-ganglionic acetylcholine receptor antibodies. ENeurologicalSci, 2018, 12, 36-38.	1.3	2
41	Establishment of a human induced stem cell line (FUi002-A) from Dravet syndrome patient carrying heterozygous R1525X mutation in SCN1A gene. Stem Cell Research, 2018, 31, 11-15.	0.7	6
42	A patient with a GNAO1 mutation with decreased spontaneous movements, hypotonia, and dystonic features. Brain and Development, 2018, 40, 926-930.	1.1	12
43	Genetic Background of Encephalopathy. , 2018, , 45-52.		1
44	Vagus nerve stimulation for generalized epilepsy with febrile seizures plus (GEFS+) accompanying seizures with impaired consciousness. Epilepsy & Behavior Case Reports, 2017, 7, 16-19.	1.5	6
45	A <i>de novo</i> missense mutation of <i>GABRB2</i> causes early myoclonic encephalopathy. Journal of Medical Genetics, 2017, 54, 202-211.	3.2	47
46	Progressive Brain Atrophy in Alternating Hemiplegia of Childhood. Movement Disorders Clinical Practice, 2017, 4, 406-411.	1.5	18
47	Abnormal γâ€∎minobutyric acid neurotransmission in a <i>Kcnq2</i> model of early onset epilepsy. Epilepsia, 2017, 58, 1430-1439.	5.1	19
48	Clinical implications of <i><scp>SCN</scp>1A</i> missense and truncation variants in a large Japanese cohort with Dravet syndrome. Epilepsia, 2017, 58, 282-290.	5.1	55
49	Two mild cases of Dravet syndrome with truncating mutation of SCN1A. Brain and Development, 2017, 39, 72-74.	1.1	8
50	New Genes for Epilepsy–Autism Comorbidity. Journal of Pediatric Neurology, 2017, 15, 105-114.	0.2	3
51	Integrative Approach with Electrophysiological and Theoretical Methods Reveals a New Role of S4 Positively Charged Residues in PKD2L1 Channel Voltage-Sensing. Scientific Reports, 2017, 7, 9760.	3.3	7
52	A de novo missense mutation in <i><scp>SLC12A5</scp></i> found in a compound heterozygote patient with epilepsy of infancy with migrating focal seizures. Clinical Genetics, 2017, 92, 654-658.	2.0	55
53	Rare variants of small effect size in neuronal excitability genes influence clinical outcome in Japanese cases of SCN1A truncation-positive Dravet syndrome. PLoS ONE, 2017, 12, e0180485.	2.5	18
54	<i>SCN8A</i> encephalopathy: Research progress and prospects. Epilepsia, 2016, 57, 1027-1035.	5.1	101

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55	Characteristic phasic evolution of convulsive seizure in <i>PCDH19</i> â€related epilepsy. Epileptic Disorders, 2016, 18, 26-33.	1.3	12
56	A case of succinic semialdehyde dehydrogenase deficiency with status epilepticus and rapid regression. Brain and Development, 2016, 38, 866-870.	1.1	9
57	Treatment with Oral ATP decreases alternating hemiplegia of childhood with de novo ATP1A3 Mutation. Orphanet Journal of Rare Diseases, 2016, 11, 55.	2.7	17
58	Surgical versus medical treatment for children with epileptic encephalopathy in infancy and early childhood: Results of an international multicenter cohort study in Far-East Asia (the FACE study). Brain and Development, 2016, 38, 449-460.	1.1	25
59	Efficacy of antiepileptic drugs for the treatment of Dravet syndrome with different genotypes. Brain and Development, 2016, 38, 40-46.	1.1	48
60	Retigabine, a Kv7.2/Kv7.3-Channel Opener, Attenuates Drug-Induced Seizures in Knock-In Mice Harboring Kcnq2 Mutations. PLoS ONE, 2016, 11, e0150095.	2.5	61
61	Tissue Remodeling in Vascular Wall in Kawasaki Disease-Related Vasculitis Model Mice. , 2016, , 241-242.		0
62	Development of a mouse model of infantile spasms induced by N -methyl- d -aspartate. Epilepsy Research, 2015, 118, 29-33.	1.6	11
63	Phenotypes of children with 20q13.3 microdeletion affecting <i>KCNQ2</i> and <i>CHRNA4</i> Epileptic Disorders, 2015, 17, 165-171.	1.3	10
64	Single Nucleotide Variations in CLCN6 Identified in Patients with Benign Partial Epilepsies in Infancy and/or Febrile Seizures. PLoS ONE, 2015, 10, e0118946.	2.5	13
65	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.	0.6	37
66	JNK is critical for the development of Candida albicans-induced vascular lesions in a mouse model of Kawasaki Disease. Cardiovascular Pathology, 2015, 24, 33-40.	1.6	14
67	A case of recurrent encephalopathy with SCN2A missense mutation. Brain and Development, 2015, 37, 631-634.	1.1	20
68	Hereditary 1,25-dihydroxyvitamin D-resistant rickets (HVDRR) caused by a VDR mutation: A novel mechanism of dominant inheritance. Bone Reports, 2015, 2, 68-73.	0.4	8
69	Spontaneous epileptic seizures in transgenic rats harboring a human ADNFLE missense mutation in the \hat{l}^2 2-subunit of the nicotinic acetylcholine receptor. Neuroscience Research, 2015, 100, 46-54.	1.9	17
70	Immediate suppression of seizure clusters by corticosteroids in PCDH19 female epilepsy. Seizure: the Journal of the British Epilepsy Association, 2015, 27, 1-5.	2.0	47
71	Missense mutations in sodium channel SCN1A and SCN2A predispose children to encephalopathy with severe febrile seizures. Epilepsy Research, 2015, 117, 1-6.	1.6	31
72	Trans-Golgi protein p230/golgin-245 is involved in phagophore formation. Biochemical and Biophysical Research Communications, 2015, 456, 275-281.	2.1	19

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73	Clinical and genetic features of acute encephalopathy in children taking theophylline. Brain and Development, 2015, 37, 463-470.	1.1	19
74	Effect of CYP2C19 polymorphisms on stiripentol administration in Japanese cases of Dravet syndrome. Brain and Development, 2015, 37, 243-249.	1.1	13
75	Characterizing PCDH19 in human induced pluripotent stem cells (iPSCs) and iPSC-derived developing neurons: emerging role of a protein involved in controlling polarity during neurogenesis. Oncotarget, 2015, 6, 26804-26813.	1.8	30
76	Clinical Utility of Neuronal Cells Directly Converted from Fibroblasts of Patients for Neuropsychiatric Disorders: Studies of Lysosomal Storage Diseases and Channelopathy. Current Molecular Medicine, 2015, 15, 138-145.	1.3	14
77	Mutant GABAA receptor subunits in genetic (idiopathic) epilepsy. Progress in Brain Research, 2014, 213, 55-85.	1.4	133
78	Genotype–phenotype correlations in alternating hemiplegia of childhood. Neurology, 2014, 82, 482-490.	1.1	93
79	Intermediate form between alternating hemiplegia of childhood and rapidâ€onset dystonia–parkinsonism. Movement Disorders, 2014, 29, 153-154.	3.9	21
80	Obesity attenuates D ₂ autoreceptor-mediated inhibition of putative ventral tegmental area dopaminergic neurons. Physiological Reports, 2014, 2, e12004.	1.7	14
81	Case of <scp>D</scp> esbuquois dysplasia type 1: Potentially lethal skeletal dysplasia. Pediatrics International, 2014, 56, e26-9.	0.5	15
82	A novel SCN1A mutation in a cytoplasmic loop in intractable juvenile myoclonic epilepsy without febrile seizures. Epileptic Disorders, 2014, 16, 227-231.	1.3	4
83	Association of nonsense mutation in GABRG2 with abnormal trafficking of GABAA receptors in severe epilepsy. Epilepsy Research, 2014, 108, 420-432.	1.6	38
84	Elfn1 recruits presynaptic mGluR7 in trans and its loss results in seizures. Nature Communications, 2014, 5, 4501.	12.8	83
85	Exacerbation of Benign Familial Neonatal Epilepsy Induced by Massive Doses of Phenobarbital and Midazolam. Pediatric Neurology, 2014, 51, 259-261.	2.1	8
86	Early onset and focal spike discharges as indicators of poor prognosis for myoclonic-astatic epilepsy. Brain and Development, 2014, 36, 613-619.	1.1	7
87	The Kick-In System: A Novel Rapid Knock-In Strategy. PLoS ONE, 2014, 9, e88549.	2.5	14
88	An Update on Genetic Research for Epilepsy. Journal of the Japan Epilepsy Society, 2014, 32, 51-54.	0.2	0
89	Low Dose Single Treatment Carbamazepine Therapy for Convulsions Associated with Mild Gastroenteritis. Journal of the Japan Epilepsy Society, 2014, 31, 506-510.	0.2	0
90	A human Dravet syndrome model from patient induced pluripotent stem cells. Molecular Brain, 2013, 6, 19.	2.6	111

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91	Altered automatic face processing in individuals with high-functioning autism spectrum disorders: Evidence from visual evoked potentials. Research in Autism Spectrum Disorders, 2013, 7, 710-720.	1.5	15
92	A novel prophylactic effect of furosemide treatment on autosomal dominant nocturnal frontal lobe epilepsy (ADNFLE). Epilepsy Research, 2013, 107, 127-137.	1.6	20
93	Newborn screening for Fabry disease in Japan: prevalence and genotypes of Fabry disease in a pilot study. Journal of Human Genetics, 2013, 58, 548-552.	2.3	102
94	Genetic analysis of PRRT2 for benign infantile epilepsy, infantile convulsions with choreoathetosis syndrome, and benign convulsions with mild gastroenteritis. Brain and Development, 2013, 35, 524-530.	1.1	27
95	A recurrent KCNT1 mutation in two sporadic cases with malignant migrating partial seizures in infancy. Gene, 2013, 531, 467-471.	2.2	80
96	Clinical analysis of catastrophic epilepsy in infancy and early childhood: Results of the Far-East Asia Catastrophic Epilepsy (FACE) study group. Brain and Development, 2013, 35, 786-792.	1.1	12
97	Oral mexiletine for lidocaine-responsive neonatal epilepsy. Brain and Development, 2013, 35, 667-669.	1.1	12
98	A case of severe progressive early-onset epileptic encephalopathy: Unique GABAergic interneuron distribution and imaging. Journal of the Neurological Sciences, 2013, 327, 65-72.	0.6	2
99	<i>SCN1A</i> testing for epilepsy: Application in clinical practice. Epilepsia, 2013, 54, 946-952.	5.1	67
100	A case of long QT syndrome with triple gene abnormalities: Digenic mutations in KCNH2 and SCN5A and gene variant in KCNE1. Heart Rhythm, 2013, 10, 600-603.	0.7	11
101	PCDH19-related female-limited epilepsy: Further details regarding early clinical features and therapeutic efficacy. Epilepsy Research, 2013, 106, 191-199.	1.6	52
102	<i>ADORA2A</i> polymorphism predisposes children to encephalopathy with febrile status epilepticus. Neurology, 2013, 80, 1571-1576.	1.1	71
103	Obesity decreases excitability of putative ventral tegmental area GABAergic neurons. Physiological Reports, 2013, 1, e00126.	1.7	10
104	Properties of a Novel GABAA Receptor ^ ^gamma; 2 Subunit Mutation Associated With Seizures. Journal of Pharmacological Sciences, 2013, 121, 84-87.	2.5	12
105	The effect of <i>SCN1A</i> mutations on patient-derived GABAergic neurons: what are the implications for future Dravet syndrome therapeutics?. Future Neurology, 2013, 8, 487-489.	0.5	2
106	Identification of ATP1A3 Mutations by Exome Sequencing as the Cause of Alternating Hemiplegia of Childhood in Japanese Patients. PLoS ONE, 2013, 8, e56120.	2.5	79
107	Novel HCN2 Mutation Contributes to Febrile Seizures by Shifting the Channel's Kinetics in a Temperature-Dependent Manner. PLoS ONE, 2013, 8, e80376.	2.5	49
108	A Pancreatic Solid Pseudo-Papillary Tumor Detected After Abdominal Injury. Gastroenterology Research, 2013, 6, 67-70.	1.3	1

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109	Pleomorphic Adenoma of the Submandibular Gland in Children. Journal of Pediatric Hematology/Oncology, 2012, 34, e39-e41.	0.6	5
110	A Novel Mutation of Human Liver Alanine:Glyoxylate Aminotransferase Causes Primary Hyperoxaluria Type 1: Immunohistochemical Quantification and Subcellular Distribution. Acta Histochemica Et Cytochemica, 2012, 45, 121-129.	1.6	5
111	KCNQ2 abnormality in BECTS: Benign childhood epilepsy with centrotemporal spikes following benign neonatal seizures resulting from a mutation of KCNQ2. Epilepsy Research, 2012, 102, 122-125.	1.6	14
112	On the likelihood of SCN1A microdeletions or duplications in Dravet syndrome with missense mutation. Brain and Development, 2012, 34, 617-619.	1,1	2
113	Genetics of temporal lobe epilepsy. Brain and Development, 2012, 34, 609-616.	1.1	23
114	Four-year experience with prenatal diagnosis of congenital heart defects at a single referral center in Japan with focus on inaccurately diagnosed cases. Journal of Medical Ultrasonics (2001), 2012, 39, 235-240.	1.3	4
115	Prevalence of SCN1A mutations in children with suspected Dravet syndrome and intractable childhood epilepsy. Epilepsy Research, 2012, 102, 195-200.	1.6	39
116	Mutations in PRRT2 responsible for paroxysmal kinesigenic dyskinesias also cause benign familial infantile convulsions. Journal of Human Genetics, 2012, 57, 338-341.	2.3	82
117	Reduced PLP1 expression in induced pluripotent stem cells derived from a Pelizaeus–Merzbacher disease patient with a partial PLP1 duplication. Journal of Human Genetics, 2012, 57, 580-586.	2.3	19
118	PCDH19 mutation in Japanese females with epilepsy. Epilepsy Research, 2012, 99, 28-37.	1.6	45
119	A boy with a severe phenotype of succinic semialdehyde dehydrogenase deficiency. Brain and Development, 2012, 34, 107-112.	1.1	25
120	Epidemiology of acute encephalopathy in Japan, with emphasis on the association of viruses and syndromes. Brain and Development, 2012, 34, 337-343.	1.1	328
121	Clinical spectrum of SCN2A mutations. Brain and Development, 2012, 34, 541-545.	1.1	104
122	Mutations of the <i>SCN1A</i> gene in acute encephalopathy. Epilepsia, 2012, 53, 558-564.	5.1	47
123	Compromised function in the Nav1.2 Dravet syndrome mutation R1312T. Neurobiology of Disease, 2012, 47, 378-384.	4.4	25
124	Persistent cutaneous neonatal herpes caused by Herpes simplex virusâ€2. Journal of Dermatology, 2012, 39, 671-672.	1.2	1
125	Parvocellular pathway impairment in autism spectrum disorder: Evidence from visual evoked potentials. Research in Autism Spectrum Disorders, 2011, 5, 277-285.	1.5	22
126	Diagnosing nocturnal frontal lobe epilepsy: A case study of two children. Seizure: the Journal of the British Epilepsy Association, 2011, 20, 583-585.	2.0	2

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127	Parageusia in an epileptic child treated with lamotrigine. Pediatrics International, 2011, 53, 1106-1107.	0.5	6
128	Impact and issues of detecting fetal congenital heart defects in Kyushu, Japan. Journal of Obstetrics and Gynaecology Research, 2011, 37, 775-781.	1.3	2
129	Mortality in Dravet syndrome: Search for risk factors in Japanese patients. Epilepsia, 2011, 52, 50-54.	5.1	57
130	Retrospective multiinstitutional study of the prevalence of early death in Dravet syndrome. Epilepsia, 2011, 52, 1144-1149.	5.1	64
131	Successful treatment of an infected wound in infants by a combination of negative pressure wound therapy and arginine supplementation. Nutrition, 2011, 27, 1141-1145.	2.4	13
132	Early-onset absence epilepsy at eight months of age. Epileptic Disorders, 2011, 13, 417-421.	1.3	1
133	International Symposium on Epilepsy in Neurometabolic Diseases (ISENMD). Brain and Development, 2011, 33, 183-187.	1.1	0
134	The developmental changes of Nav1.1 and Nav1.2 expression in the human hippocampus and temporal lobe. Brain Research, 2011, 1389, 61-70.	2.2	36
135	A case of cystic biliary atresia with an antenatally detected cyst: the possibility of changing from a correctable type with a cystic lesion (I cyst) to an uncorrectable one (IIId). Pediatric Surgery International, 2011, 27, 99-102.	1.4	15
136	Determinants of surgical repair of patent ductus arteriosus in low-birth-weight infants. Journal of Medical Ultrasonics (2001), 2011, 38, 151-155.	1.3	0
137	Autosomal dominant nocturnal frontal lobe epilepsy: a genotypic comparative study of Japanese and Korean families carrying the CHRNA4 Ser284Leu mutation. Journal of Human Genetics, 2011, 56, 609-612.	2.3	13
138	Si microchannel cooler integrated with high power amplifiers for base station of mobile communication systems. , 2011 , , .		15
139	A Case of SLE in a Young Boy. Nishinihon Journal of Dermatology, 2011, 73, 350-353.	0.0	O
140	IgG subclasses and complement pathway in segmental and global membranous nephropathy. Pediatric Nephrology, 2010, 25, 1091-1099.	1.7	93
141	Positive association between benign familial infantile convulsions and LGI4. Brain and Development, 2010, 32, 538-543.	1.1	10
142	Genetic testing in the epilepsies—Report of the ILAE Genetics Commission. Epilepsia, 2010, 51, 655-670.	5.1	175
143	Nicotinic acetylcholine receptor mutations. Epilepsia, 2010, 51, 65-65.	5.1	4
144	Interaction of Golgin-84 with the COG Complex Mediates the Intra-Golgi Retrograde Transport. Traffic, 2010, 11, 1552-1566.	2.7	71

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145	Transmission Experiment of Quantum Keys over 50 km Using High-Performance Quantum-Dot Single-Photon Source at 1.5 ŵm Wavelength. Applied Physics Express, 2010, 3, 092802.	2.4	58
146	Benign Convulsions With Mild Gastroenteritis: Is It Associated With Sodium Channel Gene SCN1A Mutation?. Journal of Child Neurology, 2010, 25, 1521-1524.	1.4	20
147	Mutational analysis of GABRG2 in a Japanese cohort with childhood epilepsies. Journal of Human Genetics, 2010, 55, 375-378.	2.3	26
148	Clinical features of Wilson disease: Analysis of 10 cases. Hepatology Research, 2010, 40, 1204-1211.	3.4	3
149	Tunneling-Injection Single-Photon Emitter Using Charged Exciton State. Japanese Journal of Applied Physics, 2009, 48, 06FF01.	1.5	0
150	Physicochemical property changes of amino acid residues that accompany missense mutations in SCN1A affect epilepsy phenotype severity. Journal of Medical Genetics, 2009, 46, 671-679.	3.2	19
151	Hepatocellular apoptosis associated with cytotoxic T/natural killerâ€eell infiltration in chronic active EBV infection. Pathology International, 2009, 59, 438-442.	1.3	2
152	Phenotype for activated tissue macrophages in histiocytic necrotizing lymphadenitis. Pathology International, 2009, 59, 631-635.	1.3	29
153	A de novo KCNQ2 mutation detected in non-familial benign neonatal convulsions. Brain and Development, 2009, 31, 27-33.	1.1	30
154	Novel de novo splice-site mutation of SCN1A in a patient with partial epilepsy with febrile seizures plus. Brain and Development, 2009, 31, 179-182.	1.1	9
155	Missense mutation of the sodium channel gene SCN2A causes Dravet syndrome. Brain and Development, 2009, 31, 758-762.	1.1	123
156	Survival and late effects on development of patients with infantile brain tumor. Pediatrics International, 2009, 51, 337-341.	0.5	13
157	Hemiconvulsion–hemiplegia syndrome in a patient with severe myoclonic epilepsy in infancy. Epilepsia, 2009, 50, 2158-2162.	5.1	36
158	Lack of potassium current in W309R mutant KCNQ3 channel causing benign familial neonatal convulsions (BFNC). Epilepsy Research, 2009, 84, 82-85.	1.6	15
159	Verification of embedded system's specification using collaborative simulation of SysML and simulink models. , 2009, , .		41
160	Neuronal Compensation and Plasticity in the Brains of Children with Remote Periventricular Leukomalacia. Rigakuryoho Kagaku, 2009, 24, 733-736.	0.1	2
161	Altered KCNQ3 Potassium Channel Function Caused by the W309R Pore-Helix Mutation Found in Human Epilepsy. Journal of Membrane Biology, 2008, 222, 55-63.	2.1	15
162	Microchromosomal deletions involving <i>SCN1A</i> and adjacent genes in severe myoclonic epilepsy in infancy. Epilepsia, 2008, 49, 1528-1534.	5.1	69

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163	Highâ€grade mature Bâ€cell lymphoma with Burkittâ€like morphology: Results of a clinicopathological study of 72 Japanese patients. Cancer Science, 2008, 99, 246-252.	3.9	18
164	Estimation of the relationship between caspaseâ€3 expression and clinical outcome of Burkitt's and Burkittâ€ike lymphoma. Cancer Science, 2008, 99, 1564-1569.	3.9	14
165	Estimation of apoptosis and cell proliferation in histiocytic necrotizing lymphadenitis using immunohistochemical double staining. Pathology International, 2008, 58, 98-103.	1.3	6
166	Histological characteristics of 21 Papua New Guinean children with highâ€grade B ell lymphoma, which is frequently associated with EBV infection. Pathology International, 2008, 58, 695-700.	1.3	9
167	Developmental changes in KCNQ2 and KCNQ3 expression in human brain: Possible contribution to the age-dependent etiology of benign familial neonatal convulsions. Brain and Development, 2008, 30, 362-369.	1.1	66
168	InP-Based Mach–Zehnder Modulator With Capacitively Loaded Traveling-Wave Electrodes. Journal of Lightwave Technology, 2008, 26, 608-615.	4.6	20
169	Rats Harboring S284L <i>Chrna4</i> Mutation Show Attenuation of Synaptic and Extrasynaptic GABAergic Transmission and Exhibit the Nocturnal Frontal Lobe Epilepsy Phenotype. Journal of Neuroscience, 2008, 28, 12465-12476.	3.6	62
170	Nuclear and excitonic spin polarization formed using cross-linearly polarized pulse pair via half-localized state in a single self-assembled quantum dot. Journal of Applied Physics, 2008, 103, 103530.	2.5	1
171	Electric field modulation of exciton recombination in InAs/GaAs quantum dots emitting at 1.3μm. Journal of Applied Physics, 2008, 104, 013504.	2.5	6
172	Genome-wide identification of febrile seizure and related epilepsy phenotype loci. Epilepsy and Seizure, 2008, 1, 30-39.	0.2	2
173	Focal Epilepsy Resulting from a de novo SCN1A Mutation. Neuropediatrics, 2007, 38, 253-256.	0.6	8
174	An optical horn structure for single-photon source using quantum dots at telecommunication wavelength. Journal of Applied Physics, 2007, 101, 081720.	2.5	93
175	Mutation screening of AP3M2 in Japanese epilepsy patients. Brain and Development, 2007, 29, 462-467.	1.1	4
176	Neuropathology of Methylmalonic Acidemia in a Child. Pediatric Neurology, 2006, 34, 156-159.	2.1	26
177	An Infant With a Mitochondrial A3243G Mutation Demonstrating the MELAS Phenotype. Pediatric Neurology, 2006, 34, 235-238.	2.1	16
178	Development and Aging Expression of Cystathionine-Beta Synthase in the Temporal Lobe and Cerebellum of Down Syndrome Patients. Neuroembryology and Aging, 2006, 4, 202-207.	0.1	10
179	Late-onset ornithine transcarbamylase deficiency in male patients: Prognostic factors and characteristics of plasma amino acid profile. Pediatrics International, 2006, 48, 105-111.	0.5	26
180	A new paradigm of channelopathy in epilepsy syndromes: Intracellular trafficking abnormality of channel molecules. Epilepsy Research, 2006, 70, 206-217.	1.6	80

#	Article	IF	Citations
181	Developmental changes in the expression of GABAA receptor alpha 1 and gamma 2 subunits in human temporal lobe, hippocampus and basal ganglia: An implication for consideration on age-related epilepsy. Epilepsy Research, 2006, 71, 47-53.	1.6	29
182	Triggered single-photon emission and cross-correlation properties in InAlAs quantum dot. Physica E: Low-Dimensional Systems and Nanostructures, 2006, 32, 144-147.	2.7	9
183	Involvement of Ca2+-induced Ca2+ releasing system in interleukin- $1\hat{l}^2$ -associated adenosine release. European Journal of Pharmacology, 2006, 532, 246-252.	3.5	11
184	Development of Electrically Driven Single-Quantum-Dot Device at Optical Fiber Bands. Japanese Journal of Applied Physics, 2006, 45, 3621-3624.	1.5	13
185	Single InAs/InP quantum dot spectroscopy in 1.3–1.55μm telecommunication band. Physica E: Low-Dimensional Systems and Nanostructures, 2005, 26, 185-189.	2.7	11
186	Observation of Overhauser shift in a self-assembled InAlAs quantum dot. Physica E: Low-Dimensional Systems and Nanostructures, 2005, 29, 510-514.	2.7	7
187	Screening method for organic aciduria by spectrofluorometric measurement of total dicarboxylic acids in human urine based on intramolecular excimer-forming fluorescence derivatization. Analytica Chimica Acta, 2005, 534, 177-183.	5.4	16
188	Genetics of Idiopathic Epilepsies. Epilepsia, 2005, 46, 38-43.	5.1	117
189	Carnitine palmitoyltransferase II deficiency due to a novel gene variant in a patient with rhabdomyolysis and ARF. American Journal of Kidney Diseases, 2005, 45, 596-602.	1.9	21
190	Mutations in the NHLRC1 gene are the common cause for Lafora disease in the Japanese population. Journal of Human Genetics, 2005, 50, 347-352.	2.3	32
191	Photon Antibunching Observed from an InAlAs Single Quantum Dot. Japanese Journal of Applied Physics, 2005, 44, L793-L796.	1.5	24
192	Ultranarrow Photoluminescence Line In 1.3–1.55 μm Of Single InAs/InP Quantum Dots. AIP Conference Proceedings, 2005, , .	0.4	0
193	Biphasic actions of topiramate on monoamine exocytosis associated with both soluble N-ethylmaleimide-sensitive factor attachment protein receptors and Ca2+-induced Ca2+-releasing systems. Neuroscience, 2005, 134, 233-246.	2.3	51
194	Single-Photon Generation in the 1.55-µm Optical-Fiber Band from an InAs/InP Quantum Dot. Japanese Journal of Applied Physics, 2005, 44, L620-L622.	1.5	120
195	Mechanisms of Calcium-Associated Exocytosis of Striatal Dopamine and DOPA Release Studied by In Vivo Microdialysis., 2005,, 89-99.		0
196	Non-classical Photon Emission from a Single InAs/InP Quantum Dot in the 1.3-µm Optical-Fiber Band. Japanese Journal of Applied Physics, 2004, 43, L993-L995.	1.5	71
197	Observation of Exciton Transition in 1.3-1.55 µm Band from Single InAs/InP Quantum Dots in Mesa Structure. Japanese Journal of Applied Physics, 2004, 43, L349-L351.	1.5	47
198	Mutations of Neuronal Voltage-gated Na+ Channel alpha1 Subunit Gene SCN1A in Core Severe Myoclonic Epilepsy in Infancy (SMEI) and in Borderline SMEI (SMEB). Epilepsia, 2004, 45, 140-148.	5.1	180

#	Article	IF	Citations
199	Seizure phenotypes of a family with missense mutations in SCN2A. Pediatric Neurology, 2004, 31, 150-152.	2.1	28
200	Protein kinase associated with gating and closing transmission mechanisms in temporoammonic pathway. Neuropharmacology, 2004, 47, 485-504.	4.1	33
201	Both 3,4-dihydroxyphenylalanine and dopamine releases are regulated by Ca2+-induced Ca2+ releasing system in rat striatum. Neuroscience Letters, 2004, 362, 244-248.	2.1	11
202	Pharmacological discrimination of protein kinase associated exocytosis mechanisms between dopamine and 3,4-dihydroxyphenylalanine in rat striatum using in vivo microdialysis. Neuroscience Letters, 2004, 363, 120-124.	2.1	11
203	Determination of exocytosis mechanisms of DOPA in rat striatum using in vivo microdialysis. Neuroscience Letters, 2004, 367, 241-245.	2.1	6
204	Epilepsy Associated with Abnormalities of GABAA Receptor and K+ Channel. Journal of the Japan Epilepsy Society, 2004, 22, 160-170.	0.2	0
205	X-Linked mental retardation and epilepsy: pathogenetic significance of ARX mutations. Brain and Development, 2003, 25, 161-165.	1.1	25
206	The genetics of febrile seizures and related epilepsy syndromes. Brain and Development, 2003, 25, 304-312.	1.1	43
207	Age-dependent modulation of hippocampal excitability by KCNQ-channels. Epilepsy Research, 2003, 53, 81-94.	1.6	67
208	Effect of valproic acid on the urinary metabolic profile of a patient with succinic semialdehyde dehydrogenase deficiency. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2003, 792, 99-106.	2.3	27
209	Essential Roles of Perforin in Antigen-Specific Cytotoxicity Mediated by Human CD4+T Lymphocytes: Analysis Using the Combination of Hereditary Perforin-Deficient Effector Cells and Fas-Deficient Target Cells. Journal of Immunology, 2003, 170, 2205-2213.	0.8	33
210	Effect of Growth Hormone on High Plasma Levels of Glucagon-Like Peptide-1 (GLP-1) in Hypophysectomized Rats. Experimental and Clinical Endocrinology and Diabetes, 2002, 110, 361-363.	1.2	4
211	Identification and Characterization of Temperature-Sensitive Mild Mutations in Three Japanese Patients with Nonsevere Forms of Very-Long-Chain Acyl-CoA Dehydrogenase Deficiency. Molecular Genetics and Metabolism, 2002, 75, 227-234.	1.1	17
212	Exocytosis mechanism as a new targeting site for mechanisms of action of antiepileptic drugs. Life Sciences, 2002, 72, 465-473.	4.3	44
213	Genetics of epilepsy: current status and perspectives. Neuroscience Research, 2002, 44, 11-30.	1.9	45
214	Genetic abnormalities underlying familial epilepsy syndromes. Brain and Development, 2002, 24, 211-222.	1.1	43
215	Rapid and sensitive detection of urinary 4-hydroxybutyric acid and its related compounds by gas chromatography–mass spectrometry in a patient with succinic semialdehyde dehydrogenase deficiency. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2002, 776, 57-63.	2.3	35
216	Molecular Genetics of Human Familial Epilepsyâ€∫Syndromes. Epilepsia, 2002, 43, 21-25.	5.1	152

#	Article	IF	CITATIONS
217	Mutation (Ser284Leu) of neuronal nicotinic acetylcholine receptor α4 subunit associated with frontal lobe epilepsy causes faster desensitization of the rat receptor expressed in oocytes. Epilepsy Research, 2002, 48, 181-186.	1.6	49
218	Autosomal dominant epilepsy with febrile seizures plus with missense mutations of the (Na+)-channel $\hat{l}\pm 1$ subunit gene, SCN1A. Epilepsy Research, 2002, 48, 15-23.	1.6	74
219	Mutation screening for Japanese Lafora's disease patients: identification of novel sequence variants in the coding and upstream regulatory regions of EPM2A gene. Molecular and Cellular Probes, 2001, 15, 281-289.	2.1	16
220	Effects of <i>L</i> -Carnitine Supplementation on Renal Anemia in Poor Responders to Erythropoietin. Blood Purification, 2001, 19, 24-32.	1.8	51
221	A missense mutation of the Na $<$ sup $>+sup> channel \hat{l}\pm<sub>IIsub> subunit gene <i>Nai><sub>vsub><i>1.2i>in a patient with febrile and afebrile seizures causes channel dysfunction. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 6384-6389.$	7.1	360
222	A novel mutation of KCNQ3 (c.925T?C) in a Japanese family with benign familial neonatal convulsions. Annals of Neurology, 2000, 47, 822-826.	5.3	104
223	A novel SSCP variant (c.828G>A) within the M2 domain of the human neuronal nicotinic acetylcholine receptor ?4 subunit gene,CHRNA4. Human Mutation, 2000, 16, 450-450.	2.5	0
224	Are some idiopathic epilepsies disorders of ion channels?: A working hypothesis. Epilepsy Research, 2000, 41, 191-204.	1.6	71
225	Electroclinical Picture of Autosomal Dominant Nocturnal Frontal Lobe Epilepsy in a Japanese Family. Epilepsia, 2000, 41, 52-58.	5.1	64
226	A novel mutation of KCNQ3 (c.925Tâ†'C) in a Japanese family with benign familial neonatal convulsions. Annals of Neurology, 2000, 47, 822-826.	5.3	7
227	A G to A transition at the last nucleotide of exon 6 of the \hat{I}^3 c gene (868Gâ†'A) may result in either a splice or missense mutation in patients with X-linked severe combined immunodeficiency. Human Genetics, 1999, 104, 36-42.	3.8	19
228	A novel intragenetic PVUII marker in the human neuronal nicotinic acetylcholine receptor a4 subunit gene (CHRNA4)., 1999, 14, 93-93.		3
229	Carnitine depletion during total parenteral nutrition despite oral Lâ€carnitine supplementation. Pediatrics International, 1997, 39, 194-200.	0.5	8
230	The R40H mutation in a late onset type of human ornithine transcarbamylase deficiency in male patients. Human Genetics, 1997, 99, 171-176.	3.8	19
231	Japanese Cases of Type 1 Thanatophoric Dysplasia Exclusively Carry a C to T Transition at Nucleotide 742 of the Fibroblast Growth Factor Receptor 3 Gene. Biochemical and Biophysical Research Communications, 1996, 227, 236-239.	2.1	12
232	[43] Mammalian glycosylphosphatidylinositol-anchored proteins and intracellular precursors. Methods in Enzymology, 1995, 250, 582-614.	1.0	19
233	Primary structure of human placental 5'-nucleotidase and identification of the glycolipid anchor in the mature form. FEBS Journal, 1990, 191, 563-569.	0.2	129
234	Relationship between Maturation of the Skin and Electrical Skin Resistance. Pediatric Research, 1987, 21, 21-24.	2.3	8

#	Article	lF	CITATIONS
235	Brefeldin A arrests the intracellular transport of a precursor of complement C3 before its conversion site in rat hepatocytes. FEBS Letters, 1987, 214, 135-138.	2.8	104
236	Immunoblotting Analysis of Plasma Protein Processing in the Secretory Pathway of Rat liver: Identification of Proteolytic Conversion Sites of Complement Pro-C3 and Prohaptoglobin1. Journal of Biochemistry, 1986, 100, 1669-1675.	1.7	19