Shinichi Hirose

List of Publications by Year
in descending order

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236	7,362	50276	79698
papers	citations	46 h-index	73 g-index

246	246	246	7763
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A missense mutation of the Na ⁺ channel Î± _{II} subunit gene Na _v 1.2 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
2	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
3	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
4	Genetic testing in the epilepsiesâ€"Report of the ILAE Genetics Commission. Epilepsia, 2010, 51, 655-670.	5.1	175
5	Molecular Genetics of Human Familial Epilepsyâ€ƒSyndromes. Epilepsia, 2002, 43, 21-25.	5.1	152
6	Mutant GABAA receptor subunits in genetic (idiopathic) epilepsy. Progress in Brain Research, 2014, 213, 55-85.	1.4	133
7	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
8	Missense mutation of the sodium channel gene SCN2A causes Dravet syndrome. Brain and Development, 2009, 31, 758-762.	1.1	123
9	Single-Photon Generation in the 1.55-Âµm Optical-Fiber Band from an InAs/InP Quantum Dot. Japanese Journal of Applied Physics, 2005, 44, L620-L622.	1.5	120
10	Genetics of Idiopathic Epilepsies. Epilepsia, 2005, 46, 38-43.	5.1	117
11	A human Dravet syndrome model from patient induced pluripotent stem cells. Molecular Brain, 2013, 6, 19.	2.6	111
12	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
13	A novel mutation ofKCNQ3 (c.925T?C) in a Japanese family with benign familial neonatal convulsions. Annals of Neurology, 2000, 47, 822-826.	5.3	104
14	Clinical spectrum of SCN2A mutations. Brain and Development, 2012, 34, 541-545.	1.1	104
15	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
16	SCN8A encephalopathy: Research progress and prospects. Epilepsia, 2016, 57, 1027-1035.	5.1	101
17	An optical horn structure for single-photon source using quantum dots at telecommunication wavelength. Journal of Applied Physics, 2007, 101, 081720.	2.5	93
18	IgG subclasses and complement pathway in segmental and global membranous nephropathy. Pediatric Nephrology, 2010, 25, 1091-1099.	1.7	93

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19	Genotypeâ€“phenotype correlations in alternating hemiplegia of childhood. Neurology, 2014, 82, 482-490.	1.1	93
20	Elfn1 recruits presynaptic mGluR7 in trans and its loss results in seizures. Nature Communications, 2014, 5, 4501.	12.8	83
21	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
22	A new paradigm of channelopathy in epilepsy syndromes: Intracellular trafficking abnormality of channel molecules. Epilepsy Research, 2006, 70, 206-217.	1.6	80
23	A recurrent KCNT1 mutation in two sporadic cases with malignant migrating partial seizures in infancy. Gene, 2013, 531, 467-471.	2.2	80
24	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
25	Autosomal dominant epilepsy with febrile seizures plus with missense mutations of the (Na+)-channel Î±1 subunit gene, SCN1A. Epilepsy Research, 2002, 48, 15-23.	1.6	74
26	Are some idiopathic epilepsies disorders of ion channels?: A working hypothesis. Epilepsy Research, 2000, 41, 191-204.	1.6	71
27	Non-classical Photon Emission from a Single InAs/InP Quantum Dot in the 1.3-Âµm Optical-Fiber Band. Japanese Journal of Applied Physics, 2004, 43, L993-L995.	1.5	71
28	Interaction of Golgin-84 with the COG Complex Mediates the Intra-Golgi Retrograde Transport. Traffic, 2010, 11, 1552-1566.	2.7	71
29	ADORA2A polymorphism predisposes children to encephalopathy with febrile status epilepticus. Neurology, 2013, 80, 1571-1576.	1.1	71
30	Microchromosomal deletions involving SCN1A and adjacent genes in severe myoclonic epilepsy in infancy. Epilepsia, 2008, 49, 1528-1534.	5.1	69
31	Age-dependent modulation of hippocampal excitability by KCNQ-channels. Epilepsy Research, 2003, 53, 81-94.	1.6	67
32	SCN1A testing for epilepsy: Application in clinical practice. Epilepsia, 2013, 54, 946-952.	5.1	67
33	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
34	Characteristics of <scp>KCNQ</scp>2 variants causing either benign neonatal epilepsy or developmental and epileptic encephalopathy. Epilepsia, 2019, 60, 1870-1880.	5.1	66
35	Electroclinical Picture of Autosomal Dominant Nocturnal Frontal Lobe Epilepsy in a Japanese Family. Epilepsia, 2000, 41, 52-58.	5.1	64
36	Retrospective multiinstitutional study of the prevalence of early death in Dravet syndrome. Epilepsia, 2011, 52, 1144-1149.	5.1	64

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37	Rats Harboring S284LChrna4Mutation 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
38	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
39	Transmission Experiment of Quantum Keys over 50 km Using High-Performance Quantum-Dot Single-Photon Source at 1.5 Âµm Wavelength. Applied Physics Express, 2010, 3, 092802.	2.4	58
40	Mortality in Dravet syndrome: Search for risk factors in Japanese patients. Epilepsia, 2011, 52, 50-54.	5.1	57
41	Clinical implications of <scp>SCN</scp>1A missense and truncation variants in a large Japanese cohort with Dravet syndrome. Epilepsia, 2017, 58, 282-290.	5.1	55
42	A de novo missense mutation in <scp>SLC12A5</scp> found in a compound heterozygote patient with epilepsy of infancy with migrating focal seizures. Clinical Genetics, 2017, 92, 654-658.	2.0	55
43	PCDH19-related female-limited epilepsy: Further details regarding early clinical features and therapeutic efficacy. Epilepsy Research, 2013, 106, 191-199.	1.6	52
44	Effects of L-Carnitine Supplementation on Renal Anemia in Poor Responders to Erythropoietin. Blood Purification, 2001, 19, 24-32.	1.8	51
45	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
46	Mutation (Ser284Leu) of neuronal nicotinic acetylcholine receptor Î±4 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
47	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
48	Efficacy of antiepileptic drugs for the treatment of Dravet syndrome with different genotypes. Brain and Development, 2016, 38, 40-46.	1.1	48
49	Observation of Exciton Transition in 1.3-1.55 Âµm Band from Single InAs/InP Quantum Dots in Mesa Structure. Japanese Journal of Applied Physics, 2004, 43, L349-L351.	1.5	47
50	Mutations of the SCN1A gene in acute encephalopathy. Epilepsia, 2012, 53, 558-564.	5.1	47
51	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
52	Ade novomissense mutation ofGABRB2causes early myoclonic encephalopathy. Journal of Medical Genetics, 2017, 54, 202-211.	3.2	47
53	Genetics of epilepsy: current status and perspectives. Neuroscience Research, 2002, 44, 11-30.	1.9	45
54	PCDH19 mutation in Japanese females with epilepsy. Epilepsy Research, 2012, 99, 28-37.	1.6	45

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55	Exocytosis mechanism as a new targeting site for mechanisms of action of antiepileptic drugs. Life Sciences, 2002, 72, 465-473.	4.3	44
56	Genetic abnormalities underlying familial epilepsy syndromes. Brain and Development, 2002, 24, 211-222.	1.1	43
57	The genetics of febrile seizures and related epilepsy syndromes. Brain and Development, 2003, 25, 304-312.	1.1	43
58	Verification of embedded system's specification using collaborative simulation of SysML and simulink models. , 2009, , .		41
59	Prevalence of SCN1A mutations in children with suspected Dravet syndrome and intractable childhood epilepsy. Epilepsy Research, 2012, 102, 195-200.	1.6	39
60	Association of nonsense mutation in GABRG2 with abnormal trafficking of GABAA receptors in severe epilepsy. Epilepsy Research, 2014, 108, 420-432.	1.6	38
61	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
62	Hemiconvulsionâ€“hemiplegia syndrome in a patient with severe myoclonic epilepsy in infancy. Epilepsia, 2009, 50, 2158-2162.	5.1	36
63	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
64	Variant Intestinal-Cell Kinase in Juvenile Myoclonic Epilepsy. New England Journal of Medicine, 2018, 378, 1018-1028.	27.0	36
65	Rapid and sensitive detection of urinary 4-hydroxybutyric acid and its related compounds by gas chromatographyâ€“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
66	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
67	Protein kinase associated with gating and closing transmission mechanisms in temporoammonic pathway. Neuropharmacology, 2004, 47, 485-504.	4.1	33
68	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
69	miR-124 dosage regulates prefrontal cortex function by dopaminergic modulation. Scientific Reports, 2019, 9, 3445.	3.3	32
70	Newborn screening for Fabry disease in the western region of Japan. Molecular Genetics and Metabolism Reports, 2020, 22, 100562.	1.1	32
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	A de novo KCNQ2 mutation detected in non-familial benign neonatal convulsions. Brain and Development, 2009, 31, 27-33.	1.1	30

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73	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
74	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
75	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
76	Phenotype for activated tissue macrophages in histiocytic necrotizing lymphadenitis. Pathology International, 2009, 59, 631-635.	1.3	29
77	Seizure phenotypes of a family with missense mutations in SCN2A. Pediatric Neurology, 2004, 31, 150-152.	2.1	28
78	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
79	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
80	Efficacy and tolerability of perampanel in pediatric patients with Dravet syndrome. Epilepsy Research, 2019, 154, 34-38.	1.6	27
81	Neuropathology of Methylmalonic Acidemia in a Child. Pediatric Neurology, 2006, 34, 156-159.	2.1	26
82	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
83	Mutational analysis of GABRG2 in a Japanese cohort with childhood epilepsies. Journal of Human Genetics, 2010, 55, 375-378.	2.3	26
84	X-Linked mental retardation and epilepsy: pathogenetic significance of ARX mutations. Brain and Development, 2003, 25, 161-165.	1.1	25
85	A boy with a severe phenotype of succinic semialdehyde dehydrogenase deficiency. Brain and Development, 2012, 34, 107-112.	1.1	25
86	Compromised function in the Nav1.2 Dravet syndrome mutation R1312T. Neurobiology of Disease, 2012, 47, 378-384.	4.4	25
87	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
88	Photon Antibunching Observed from an InAlAs Single Quantum Dot. Japanese Journal of Applied Physics, 2005, 44, L793-L796.	1.5	24
89	Genetics of temporal lobe epilepsy. Brain and Development, 2012, 34, 609-616.	1.1	23
90	Parvocellular pathway impairment in autism spectrum disorder: Evidence from visual evoked potentials. Research in Autism Spectrum Disorders, 2011, 5, 277-285.	1.5	22

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91	Quinidine therapy and therapeutic drug monitoring in four patients with KCNT1 mutations. Epileptic Disorders, 2019, 21, 48-54.	1.3	22
92	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
93	Intermediate form between alternating hemiplegia of childhood and rapidâ€onset dystoniaâ€“parkinsonism. Movement Disorders, 2014, 29, 153-154.	3.9	21
94	InP-Based Machâ€“Zehnder Modulator With Capacitively Loaded Traveling-Wave Electrodes. Journal of Lightwave Technology, 2008, 26, 608-615.	4.6	20
95	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
96	A novel prophylactic effect of furosemide treatment on autosomal dominant nocturnal frontal lobe epilepsy (ADNFLE). Epilepsy Research, 2013, 107, 127-137.	1.6	20
97	A case of recurrent encephalopathy with SCN2A missense mutation. Brain and Development, 2015, 37, 631-634.	1.1	20
98	CACNA1A -related early-onset encephalopathy with myoclonic epilepsy: A case report. Brain and Development, 2018, 40, 130-133.	1.1	20
99	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
100	[43] Mammalian glycosylphosphatidylinositol-anchored proteins and intracellular precursors. Methods in Enzymology, 1995, 250, 582-614.	1.0	19
101	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
102	A G to A transition at the last nucleotide of exon 6 of the Î³c 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
103	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
104	Reduced PLP1 expression in induced pluripotent stem cells derived from a Pelizaeusâ€“Merzbacher disease patient with a partial PLP1 duplication. Journal of Human Genetics, 2012, 57, 580-586.	2.3	19
105	Trans-Golgi protein p230/golgin-245 is involved in phagophore formation. Biochemical and Biophysical Research Communications, 2015, 456, 275-281.	2.1	19
106	Clinical and genetic features of acute encephalopathy in children taking theophylline. Brain and Development, 2015, 37, 463-470.	1.1	19
107	Abnormal Î³â€aminobutyric acid neurotransmission in a Kcnq2 model of early onset epilepsy. Epilepsia, 2017, 58, 1430-1439.	5.1	19
108	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

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109	Progressive Brain Atrophy in Alternating Hemiplegia of Childhood. Movement Disorders Clinical Practice, 2017, 4, 406-411.	1.5	18
110	A recurrent PJA1 variant in trigonocephaly and neurodevelopmental disorders. Annals of Clinical and Translational Neurology, 2020, 7, 1117-1131.	3.7	18
111	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
112	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
113	Spontaneous epileptic seizures in transgenic rats harboring a human ADNFLE missense mutation in the Î²2-subunit of the nicotinic acetylcholine receptor. Neuroscience Research, 2015, 100, 46-54.	1.9	17
114	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
115	Somatic mosaic deletions involving SCN1A cause Dravet syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 657-662.	1.2	17
116	Epigenetics explained: a topic â€œprimerâ€•for the epilepsy community by the ILAE Genetics/Epigenetics Task Force. Epileptic Disorders, 2020, 22, 127-141.	1.3	17
117	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
118	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
119	An Infant With a Mitochondrial A3243G Mutation Demonstrating the MELAS Phenotype. Pediatric Neurology, 2006, 34, 235-238.	2.1	16
120	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
121	Comparative characterization of PCDH19 missense and truncating variants in PCDH19-related epilepsy. Journal of Human Genetics, 2021, 66, 569-578.	2.3	16
122	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
123	Lack of potassium current in W309R mutant KCNQ3 channel causing benign familial neonatal convulsions (BFNC). Epilepsy Research, 2009, 84, 82-85.	1.6	15
124	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
125	Si microchannel cooler integrated with high power amplifiers for base station of mobile communication systems. , 2011, , .		15
126	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

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127	Case of <scp>D</scp>esbuquois dysplasia type 1: Potentially lethal skeletal dysplasia. Pediatrics International, 2014, 56, e26-9.	0.5	15
128	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
129	Estimation of the relationship between caspaseâ€3 expression and clinical outcome of Burkitt's and Burkittâ€like lymphoma. Cancer Science, 2008, 99, 1564-1569.	3.9	14
130	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
131	Obesity attenuates D₂autoreceptor-mediated inhibition of putative ventral tegmental area dopaminergic neurons. Physiological Reports, 2014, 2, e12004.	1.7	14
132	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
133	The Kick-In System: A Novel Rapid Knock-In Strategy. PLoS ONE, 2014, 9, e88549.	2.5	14
134	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
135	Development of Electrically Driven Single-Quantum-Dot Device at Optical Fiber Bands. Japanese Journal of Applied Physics, 2006, 45, 3621-3624.	1.5	13
136	Survival and late effects on development of patients with infantile brain tumor. Pediatrics International, 2009, 51, 337-341.	0.5	13
137	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
138	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
139	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
140	Effect of CYP2C19 polymorphisms on stiripentol administration in Japanese cases of Dravet syndrome. Brain and Development, 2015, 37, 243-249.	1.1	13
141	Application of induced pluripotent stem cells in epilepsy. Molecular and Cellular Neurosciences, 2020, 108, 103535.	2.2	13
142	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
143	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
144	Oral mexiletine for lidocaine-responsive neonatal epilepsy. Brain and Development, 2013, 35, 667-669.	1.1	12

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145	Properties of a Novel GABAA Receptor ^|^gamma;2 Subunit Mutation Associated With Seizures. Journal of Pharmacological Sciences, 2013, 121, 84-87.	2.5	12
146	Characteristic phasic evolution of convulsive seizure in PCDH19â€related epilepsy. Epileptic Disorders, 2016, 18, 26-33.	1.3	12
147	A patient with a GNAO1 mutation with decreased spontaneous movements, hypotonia, and dystonic features. Brain and Development, 2018, 40, 926-930.	1.1	12
148	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
149	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
150	Single InAs/InP quantum dot spectroscopy in 1.3â€“1.55Î¼m telecommunication band. Physica E: Low-Dimensional Systems and Nanostructures, 2005, 26, 185-189.	2.7	11
151	Involvement of Ca2+-induced Ca2+ releasing system in interleukin-1Î²-associated adenosine release. European Journal of Pharmacology, 2006, 532, 246-252.	3.5	11
152	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
153	Development of a mouse model of infantile spasms induced by N -methyl- d -aspartate. Epilepsy Research, 2015, 118, 29-33.	1.6	11
154	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
155	Impaired neuronal activity and differential gene expression in STXBP1 encephalopathy patient iPSC-derived GABAergic neurons. Human Molecular Genetics, 2021, 30, 1337-1348.	2.9	11
156	Current status of newborn screening for Pompe disease in Japan. Orphanet Journal of Rare Diseases, 2021, 16, 516.	2.7	11
157	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
158	Positive association between benign familial infantile convulsions and LGI4. Brain and Development, 2010, 32, 538-543.	1.1	10
159	Obesity decreases excitability of putative ventral tegmental area GABAergic neurons. Physiological Reports, 2013, 1, e00126.	1.7	10
160	Phenotypes of children with 20q13.3 microdeletion affecting KCNQ2 and CHRNA4. Epileptic Disorders, 2015, 17, 165-171.	1.3	10
161	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
162	Histological characteristics of 21 Papua New Guinean children with highâ€grade Bâ€cell lymphoma, which is frequently associated with EBV infection. Pathology International, 2008, 58, 695-700.	1.3	9

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163	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
164	A case of succinic semialdehyde dehydrogenase deficiency with status epilepticus and rapid regression. Brain and Development, 2016, 38, 866-870.	1.1	9
165	Genetics and gene therapy in Dravet syndrome. Epilepsy and Behavior, 2021, , 108043.	1.7	9
166	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
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