Sam Berkovic Am,, Faa, Fracp

List of Publications by Year
in descending order

508 papers	53,075 citations	1531 109 h-index	2072 211 g-index
528 all docs	528 docs citations	528 times ranked	35011 citing authors

#	Article	IF	Citations
1	Genome-wide association study of febrile seizures implicates fever response and neuronal excitability genes. Brain, 2022, 145, 555-568.	3.7	29
2	Plasma neurofilament light chain protein is not increased in treatment-resistant schizophrenia and first-degree relatives. Australian and New Zealand Journal of Psychiatry, 2022, 56, 1295-1305.	1.3	10
3	Sporadic hypothalamic hamartoma is a ciliopathy with somatic and bi-allelic contributions. Human Molecular Genetics, 2022, 31, 2307-2316.	1.4	8
4	OUP accepted manuscript. Brain, 2022, , .	3.7	1
5	Association of ultraâ€rare coding variants with genetic generalized epilepsy: A caseâ€“control whole exome sequencing study. Epilepsia, 2022, 63, 723-735.	2.6	8
6	Evidence for a Dual-Pathway, 2-Hit Genetic Model for Focal Cortical Dysplasia and Epilepsy. Neurology: Genetics, 2022, 8, e652.	0.9	14
7	Bi-allelic SMO variants in hypothalamic hamartoma: a recessive cause of Pallister-Hall syndrome. European Journal of Human Genetics, 2022, 30, 384-388.	1.4	6
8	Cerebrospinal fluid neurofilament light chain differentiates primary psychiatric disorders from rapidly progressive, Alzheimer's disease and frontotemporal disorders in clinical settings. Alzheimer's and Dementia, 2022, 18, 2218-2233.	0.4	24
9	Diagnostic delay in focal epilepsy: Association with brain pathology and age. Seizure: the Journal of the British Epilepsy Association, 2022, 96, 121-127.	0.9	2
10	Defective lipid signalling caused by mutations inPIK3C2Bunderlies focal epilepsy. Brain, 2022, 145, 2313-2331.	3.7	10
11	Machine learning approaches for imagingâ€based prognostication of the outcome of surgery for mesial temporal lobe epilepsy. Epilepsia, 2022, 63, 1081-1092.	2.6	10
12	Interictal EEG and ECG for SUDEP Risk Assessment: A Retrospective Multicenter Cohort Study. Frontiers in Neurology, 2022, 13, 858333.	1.1	2
13	A pharmacogenomic assessment of psychiatric adverse drug reactions to levetiracetam. Epilepsia, 2022, 63, 1563-1570.	2.6	11
14	Rare sudden unexpected death in epilepsy SCN5A variants cause changes in channel function implicating cardiac arrhythmia as a cause of death. Epilepsia, 2022, 63, .	2.6	8
15	ILAE Genetic Literacy Series: familial focal epilepsy syndromes. Epileptic Disorders, 2022, 24, 221-228.	0.7	3
16	Functional correlates of clinical phenotype and severity in recurrent SCN2A variants. Communications Biology, 2022, 5, .	2.0	13
17	Common risk variants for epilepsy are enriched in families previously targeted for rare monogenic variant discovery. EBioMedicine, 2022, 81, 104079.	2.7	10
18	Precision medicine for genetic epilepsy on the horizon: Recent advances, present challenges, and suggestions for continued progress. Epilepsia, 2022, 63, 2461-2475.	2.6	50

#	Article	IF	Citations
19	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	1.1	28
20	Progressive Myoclonus Epilepsy Caused by a Homozygous Splicing Variant of <scp>SLC7A6OS</scp>. Annals of Neurology, 2021, 89, 402-407.	2.8	5
21	Transcriptome analysis of a ring chromosome 20 patient cohort. Epilepsia, 2021, 62, e22-e28.	2.6	5
22	Epilepsy risk in offspring of affected parents; a cohort study of the â€œmaternal effectâ€•in epilepsy. Annals of Clinical and Translational Neurology, 2021, 8, 153-162.	1.7	6
23	The clinical utility of exome sequencing and extended bioinformatic analyses in adolescents and adults with a broad range of neurological phenotypes: an Australian perspective. Journal of the Neurological Sciences, 2021, 420, 117260.	0.3	16
24	Cerebrospinal fluid liquid biopsy for detecting somatic mosaicism in brain. Brain Communications, 2021, 3, fcaa235.	1.5	42
25	Newly diagnosed seizures assessed at two established first seizure clinics: Clinic characteristics, investigations, and findings over 11 years. Epilepsia Open, 2021, 6, 171-180.	1.3	11
26	The severe epilepsy syndromes of infancy: A populationâ€based study. Epilepsia, 2021, 62, 358-370.	2.6	31
27	Contribution of rare genetic variants to drug response in absence epilepsy. Epilepsy Research, 2021, 170, 106537.	0.8	9
28	Association of SLC32A1 Missense Variants With Genetic Epilepsy With Febrile Seizures Plus. Neurology, 2021, 96, e2251-e2260.	1.5	13
29	Cation leak underlies neuronal excitability in an HCN1 developmental and epileptic encephalopathy. Brain, 2021, 144, 2060-2073.	3.7	26
30	Progressive myoclonus epilepsiesâ€”Residual unsolved cases have marked genetic heterogeneity including dolichol-dependent protein glycosylation pathway genes. American Journal of Human Genetics, 2021, 108, 722-738.	2.6	41
31	Assessing the role of rare genetic variants in drugâ€resistant, nonâ€lesional focal epilepsy. Annals of Clinical and Translational Neurology, 2021, 8, 1376-1387.	1.7	16
32	Lossâ€ofâ€function variants in K_v11.1 cardiac channels as a biomarker for SUDEP. Annals of Clinical and Translational Neurology, 2021, 8, 1422-1432.	1.7	9
33	Integrated in silico and experimental assessment of disease relevance of PCDH19 Âmissense variants. Human Mutation, 2021, 42, 1030-1041.	1.1	1
34	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. American Journal of Human Genetics, 2021, 108, 965-982.	2.6	35
35	Pathogenic <scp>MAST3</scp> Variants in the <scp>STK</scp> Domain Are Associated with Epilepsy. Annals of Neurology, 2021, 90, 274-284.	2.8	7
36	Association Between Psychiatric Comorbidities and Mortality in Epilepsy. Neurology: Clinical Practice, 2021, 11, 429-437.	0.8	7

#	Article	IF	Citations
37	Improving Specificity of <scp>Cerebrospinal Fluid</scp> Liquid Biopsy for Genetic Testing. Annals of Neurology, 2021, 90, 693-694.	2.8	2
38	Postictal Psychosis in Epilepsy: A Clinicogenetic Study. Annals of Neurology, 2021, 90, 464-476.	2.8	11
39	Cutting edge approaches to detecting brain mosaicism associated with common focal epilepsies: implications for diagnosis and potential therapies. Expert Review of Neurotherapeutics, 2021, 21, 1309-1316.	1.4	5
40	Variants in ATP6V0A1 cause progressive myoclonus epilepsy and developmental and epileptic encephalopathy. Brain Communications, 2021, 3, fcab245.	1.5	10
41	Association of Short-term Heart Rate Variability and Sudden Unexpected Death in Epilepsy. Neurology, 2021, 97, .	1.5	25
42	State transitions through inhibitory interneurons in a cortical network model. PLoS Computational Biology, 2021, 17, e1009521.	1.5	10
43	Hypothalamic Hamartomas. Neurology, 2021, 97, 864-873.	1.5	12
44	Identification of a recurrent mosaic KRAS variant in brain tissue from an individual with nevus sebaceous syndrome. Journal of Physical Education and Sports Management, 2021, 7, a006133.	0.5	6
45	Progressive Myoclonus Epilepsies. Neurology: Genetics, 2021, 7, e641.	0.9	20
46	What is the motor vehicle crash risk for drivers with epilepsy? A systematic review. Journal of Transport and Health, 2021, 23, 101286.	1.1	2
47	Plasma neurofilament light chain and phosphorylated tau 181 in neurodegenerative and psychiatric disorders: moving closer towards a simple diagnostic test like a 'Câ€reactive protein' for the brain?. Alzheimer's and Dementia, 2021, 17, .	0.4	0
48	EXOME REPORT: Novel mutation in ATP6V1B2 segregating with autosomal dominant epilepsy, intellectual disability and mild gingival and nail abnormalities. European Journal of Medical Genetics, 2020, 63, 103799.	0.7	14
49	Epilepsy genetics: clinical impacts and biological insights. Lancet Neurology, The, 2020, 19, 93-100.	4.9	75
50	The â€œmaternal effectâ€•on epilepsy risk: Analysis of familial epilepsies and reassessment of prior evidence. Annals of Neurology, 2020, 87, 132-138.	2.8	2
51	SCN1A Variants in vaccineâ€related febrile seizures: A prospective study. Annals of Neurology, 2020, 87, 281-288.	2.8	15
52	Novel Missense CACNA1G Mutations Associated with Infantile-Onset Developmental and Epileptic Encephalopathy. International Journal of Molecular Sciences, 2020, 21, 6333.	1.8	7
53	Mortality in patients with psychogenic nonepileptic seizures. Neurology, 2020, 95, e643-e652.	1.5	75
54	Anterior temporal encephaloceles: Elusive, important, and rewarding to treat. Epilepsia, 2020, 61, 2675-2684.	2.6	16

#	Article	IF	Citations
55	Generalized, focal, and combined epilepsies in families: New evidence for distinct genetic factors. Epilepsia, 2020, 61, 2667-2674.	2.6	4
56	Are Variants Causing Cardiac Arrhythmia Risk Factors in Sudden Unexpected Death in Epilepsy?. Frontiers in Neurology, 2020, 11, 925.	1.1	16
57	Neurofilament light chain in psychiatric and neurodegenerative disorders: A â€˜câ€reactive proteinâ€™ for the brain?. Alzheimer's and Dementia, 2020, 16, e041347.	0.4	1
58	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	3.7	47
59	Familial adult myoclonic epilepsy type 1 SAMD12 TTTCA repeat expansion arose 17,000 years ago and is present in Sri Lankan and Indian families. European Journal of Human Genetics, 2020, 28, 973-978.	1.4	23
60	Antiepileptic Drug Teratogenicity and De Novo Genetic Variation Load. Annals of Neurology, 2020, 87, 897-906.	2.8	9
61	Inherited RORB pathogenic variants: Overlap of photosensitive genetic generalized and occipital lobe epilepsy. Epilepsia, 2020, 61, e23-e29.	2.6	14
62	GABA-mediated tonic inhibition differentially modulates gain in functional subtypes of cortical interneurons. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 3192-3202.	3.3	33
63	The Genetics of Epilepsy. Annual Review of Genomics and Human Genetics, 2020, 21, 205-230.	2.5	116
64	Encephalopathies with KCNC1 variants: genotypeâ€phenotypeâ€functional correlations. Annals of Clinical and Translational Neurology, 2019, 6, 1263-1272.	1.7	33
65	Evidence for type-specific DNA methylation patterns in epilepsy: a discordant monozygotic twin approach. Epigenomics, 2019, 11, 951-968.	1.0	19
66	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. American Journal of Human Genetics, 2019, 105, 267-282.	2.6	237
67	Deciphering the role of epigenetics in self-limited epilepsy with centrotemporal spikes. Epilepsy Research, 2019, 156, 106163.	0.8	5
68	Predominantly nocturnal seizures post temporal lobectomy: Characteristics of an unusual outcome group. Epilepsy Research, 2019, 155, 106154.	0.8	0
69	Secondâ€hit DEPDC5 mutation is limited to dysmorphic neurons in cortical dysplasia type IIA. Annals of Clinical and Translational Neurology, 2019, 6, 1338-1344.	1.7	55
70	Somatic mutation: The hidden genetics of brain malformations and focal epilepsies. Epilepsy Research, 2019, 155, 106161.	0.8	45
71	Quantitative analysis of phenotypic elements augments traditional electroclinical classification of common familial epilepsies. Epilepsia, 2019, 60, 2194-2203.	2.6	0
72	Nomenclature of Genetically Determined Myoclonus Syndromes: Recommendations of the International Parkinson and Movement Disorder Society Task Force. Movement Disorders, 2019, 34, 1602-1613.	2.2	23

#	Article	IF	Citations
73	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	5.8	99
74	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. Annals of Neurology, 2019, 86, 821-831.	2.8	96
75	Epidemiology and etiology of infantile developmental and epileptic encephalopathies in Tasmania. Epilepsia Open, 2019, 4, 504-510.	1.3	11
76	Splice variant in ARX leading to loss of Câ€terminal region in a boy with intellectual disability and infantile onset developmental and epileptic encephalopathy. American Journal of Medical Genetics, Part A, 2019, 179, 1483-1490.	0.7	9
77	Epilepsy in families: Age at onset is a familial trait, independent of syndrome. Annals of Neurology, 2019, 86, 91-98.	2.8	11
78	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. Epilepsia, 2019, 60, 797-806.	2.6	52
79	No evidence for a BRD 2 promoter hypermethylation inÂblood leukocytes of Europeans with juvenile myoclonicÂepilepsy. Epilepsia, 2019, 60, e31-e36.	2.6	4
80	Human GABRG2 generalized epilepsy. Neurology: Genetics, 2019, 5, e340.	0.9	6
81	Epileptic encephalopathies of infancy: welcome advances. Lancet, The, 2019, 394, 2203-2204.	6.3	1
82	Kufs disease due to mutation ofCLN6: clinical, pathological and molecular genetic features. Brain, 2019, 142, 59-69.	3.7	28
83	SYNGAP1 encephalopathy. Neurology, 2019, 92, e96-e107.	1.5	131
84	Metabolic patterns and seizure outcomes following anterior temporal lobectomy. Annals of Neurology, 2019, 85, 241-250.	2.8	25
85	Clinical challenges and future therapeutic approaches for neuronal ceroid lipofuscinosis. Lancet Neurology, The, 2019, 18, 107-116.	4.9	128
86	A case series of lacosamide as adjunctive therapy in refractory sleepâ€related hypermotor epilepsy (previously nocturnal frontal lobe epilepsy). Journal of Sleep Research, 2018, 27, e12669.	1.7	10
87	Parental Mosaicism in â€œDe Novoâ€•Epileptic Encephalopathies. New England Journal of Medicine, 2018, 378, 1646-1648.	13.9	104
88	Can mutationâ€mediated effects occurring early in development cause longâ€term seizure susceptibility in genetic generalized epilepsies?. Epilepsia, 2018, 59, 915-922.	2.6	7
89	Consistency of Long-Term Subdural Electrocorticography in Humans. IEEE Transactions on Biomedical Engineering, 2018, 65, 344-352.	2.5	39
90	Teenage-onset progressive myoclonic epilepsy due to a familial C9orf72 repeat expansion. Neurology, 2018, 90, e658-e663.	1.5	9

#	Article	IF	Citations
91	Ion Channels in Genetic Epilepsy: From Genes and Mechanisms to Disease-Targeted Therapies. Pharmacological Reviews, 2018, 70, 142-173.	7.1	215
92	Gain-of-functionHCN2variants in genetic epilepsy. Human Mutation, 2018, 39, 202-209.	1.1	28
93	Precision therapy for epilepsy due to KCNT1 mutations. Neurology, 2018, 90, e67-e72.	1.5	108
94	Development of a rapid functional assay that predicts GLUT1 disease severity. Neurology: Genetics, 2018, 4, e297.	0.9	7
95	A Primate-Specific Isoform of PLEKHG6 Regulates Neurogenesis and Neuronal Migration. Cell Reports, 2018, 25, 2729-2741.e6.	2.9	43
96	Aberrant Inclusion of a Poison Exon Causes Dravet Syndrome and Related SCN1A-Associated Genetic Epilepsies. American Journal of Human Genetics, 2018, 103, 1022-1029.	2.6	76
97	Dynamic action potential clamp predicts functional separation in mild familial and severe de novo forms of SCN2A epilepsy. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E5516-E5525.	3.3	69
98	Evidence of linkage to chromosome 5p13.2â€q11.1 in a large inbred family with genetic generalized epilepsy. Epilepsia, 2018, 59, e125-e129.	2.6	3
99	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	6.0	1,085
100	Genetic generalized epilepsies. Epilepsia, 2018, 59, 1148-1153.	2.6	72
101	Somatic GNAQ mutation in the forme fruste of Sturge-Weber syndrome. Neurology: Genetics, 2018, 4, e236.	0.9	29
102	Genetic literacy series: genetic epilepsy with febrile seizures plus. Epileptic Disorders, 2018, 20, 232-238.	0.7	44
103	KANSL1 variation is not a major contributing factor in self-limited focal epilepsy syndromes of childhood. PLoS ONE, 2018, 13, e0191546.	1.1	3
104	De novo <scp>SCN</scp>1A pathogenic variants in the <scp>GEFS</scp>+ spectrum: Not always a familial syndrome. Epilepsia, 2017, 58, e26-e30.	2.6	31
105	Frequency of <scp>CNKSR</scp>2 mutation in the Xâ€linked epilepsyâ€aphasia spectrum. Epilepsia, 2017, 58, e40-e43.	2.6	23
106	<scp>ILAE</scp> classification of the epilepsies: Position paper of the <scp>ILAE</scp> Commission for Classification and Terminology. Epilepsia, 2017, 58, 512-521.	2.6	3,464
107	SCN1A clinical spectrum includes the self-limited focal epilepsies of childhood. Epilepsy Research, 2017, 131, 9-14.	0.8	12
108	Real-world utility of whole exome sequencing with targeted gene analysis for focal epilepsy. Epilepsy Research, 2017, 131, 1-8.	0.8	93

#	Article	IF	Citations
109	Evaluation of GLUT1 variation in non-acquired focal epilepsy. Epilepsy Research, 2017, 133, 54-57.	0.8	5
110	Familial epilepsy with anterior polymicrogyria as a presentation of COL18A1 mutations. European Journal of Medical Genetics, 2017, 60, 437-443.	0.7	10
111	Myoclonus epilepsy and ataxia due to <scp>KCNC</scp>1 mutation: Analysis of 20 cases and <scp>K</scp>⁺ channel properties. Annals of Neurology, 2017, 81, 677-689.	2.8	69
112	Exome sequencingâ€“based molecular autopsy of formalin-fixed paraffin-embedded tissue after sudden death. Genetics in Medicine, 2017, 19, 1127-1133.	1.1	26
113	Epilepsy research in 2016: new treatment directions. Lancet Neurology, The, 2017, 16, 7-9.	4.9	3
114	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
115	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. American Journal of Human Genetics, 2017, 101, 516-524.	2.6	43
116	Genetic epilepsy with febrile seizures plus. Neurology, 2017, 89, 1210-1219.	1.5	112
117	Optimizing genomic medicine in epilepsy through a gene-customized approach to missense variant interpretation. Genome Research, 2017, 27, 1715-1729.	2.4	150
118	Familial mesial temporal lobe epilepsy and the borderland of dÃ©jÃ vu. Annals of Neurology, 2017, 82, 166-176.	2.8	19
119	ExACtly zero or once. Neurology: Genetics, 2017, 3, e163.	0.9	37
120	Synaptic Zn²⁺ and febrile seizure susceptibility. British Journal of Pharmacology, 2017, 174, 119-125.	2.7	18
121	Sensitive quantitative detection of somatic mosaic mutation in â€œdouble cortexâ€•syndrome. Epileptic Disorders, 2017, 19, 450-455.	0.7	13
122	A case-control collapsing analysis identifies epilepsy genes implicated in trio sequencing studies focused on de novo mutations. PLoS Genetics, 2017, 13, e1007104.	1.5	25
123	Periventricular Nodular Heterotopia: Detection of Abnormal Microanatomic Fiber Structures with Whole-Brain Diffusion MR Imaging Tractography. Radiology, 2016, 281, 896-906.	3.6	23
124	Exomeâ€based analysis of cardiac arrhythmia, respiratory control, and epilepsy genes in sudden unexpected death in epilepsy. Annals of Neurology, 2016, 79, 522-534.	2.8	216
125	Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2â€“2q11.2. Human Genetics, 2016, 135, 1117-1125.	1.8	29
126	In silico prioritization based on coexpression can aid epileptic encephalopathy gene discovery. Neurology: Genetics, 2016, 2, e51.	0.9	19

#	Article	IF	Citations
127	Evaluation of nonâ€coding variation in <scp>GLUT</scp>1 deficiency. Developmental Medicine and Child Neurology, 2016, 58, 1295-1302.	1.1	20
128	Seizures as presenting and prominent symptom in choreaâ€acanthocytosis with c.2343del <scp>VPS</scp>13A gene mutation. Epilepsia, 2016, 57, 549-556.	2.6	16
129	Is FGF13 a major contributor to genetic epilepsy with febrile seizures plus?. Epilepsy Research, 2016, 128, 48-51.	0.8	7
130	A targeted resequencing gene panel for focal epilepsy. Neurology, 2016, 86, 1605-1612.	1.5	48
131	Definition and diagnostic criteria of sleep-related hypermotor epilepsy. Neurology, 2016, 86, 1834-1842.	1.5	245
132	Dominant KCNA2 mutation causes episodic ataxia and pharmacoresponsive epilepsy. Neurology, 2016, 87, 1975-1984.	1.5	71
133	Mutations of the Sonic Hedgehog Pathway Underlie Hypothalamic Hamartoma with Gelastic Epilepsy. American Journal of Human Genetics, 2016, 99, 423-429.	2.6	59
134	Progressive myoclonus epilepsy associated with SACS gene mutations. Neurology: Genetics, 2016, 2, e83.	0.9	14
135	Hippocampal malrotation is an anatomic variant and has no clinical significance in <scp>MRI</scp>â€negative temporal lobe epilepsy. Epilepsia, 2016, 57, 1719-1728.	2.6	36
136	Diagnosis and misdiagnosis of adult neuronal ceroid lipofuscinosis (Kufs disease). Neurology, 2016, 87, 579-584.	1.5	28
137	TBC1D24 genotypeâ€“phenotype correlation. Neurology, 2016, 87, 77-85.	1.5	97
138	Multiplex families with epilepsy. Neurology, 2016, 86, 713-722.	1.5	23
139	Mutations in the mammalian target of rapamycin pathway regulators NPRL2 and NPRL3 cause focal epilepsy. Annals of Neurology, 2016, 79, 120-131.	2.8	190
140	Interictal spikes and epileptic seizures: their relationship and underlying rhythmicity. Brain, 2016, 139, 1066-1078.	3.7	250
141	Corrigendum to â€œClinical and genetic analysis of a family with two rare reflex epilepsiesâ€•[Seizure â€“ Eur. J. Epilepsy 29 (2015) 90â€“96]. Seizure: the Journal of the British Epilepsy Association, 2015, 33, 104.	0.9	0
142	PRIMA1 mutation: a new cause of nocturnal frontal lobe epilepsy. Annals of Clinical and Translational Neurology, 2015, 2, 821-830.	1.7	21
143	Loss of synaptic Zn2+ transporter function increases risk of febrile seizures. Scientific Reports, 2015, 5, 17816.	1.6	33
144	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. EBioMedicine, 2015, 2, 1063-1070.	2.7	74

#	Article	IF	Citations
145	Mind the gap: Multiple events and lengthy delays before presentation with a â€œfirst seizureâ€. Epilepsia, 2015, 56, 1534-1541.	2.6	65
146	Familial neonatal seizures in 36 families: Clinical and genetic features correlate with outcome. Epilepsia, 2015, 56, 1071-1080.	2.6	94
147	Epileptic spasms are a feature of DEPDC5 mTORopathy. Neurology: Genetics, 2015, 1, e17.	0.9	63
148	Quinidine in the treatment of <scp>KCNT</scp>1â€positive epilepsies. Annals of Neurology, 2015, 78, 995-999.	2.8	184
149	Genetics of Epilepsy in Clinical Practice. Epilepsy Currents, 2015, 15, 192-196.	0.4	39
150	Mutation of the nuclear lamin geneLMNB2in progressive myoclonus epilepsy with early ataxia. Human Molecular Genetics, 2015, 24, 4483-4490.	1.4	41
151	Familial cortical dysplasia type <scp>IIA</scp> caused by a germline mutation in <scp>DEPDC</scp>5. Annals of Clinical and Translational Neurology, 2015, 2, 575-580.	1.7	95
152	Lysosomal integral membrane protein type-2 (LIMP-2/SCARB2) is a substrate of cathepsin-F, a cysteine protease mutated in type-B-Kufs-disease. Biochemical and Biophysical Research Communications, 2015, 457, 334-340.	1.0	13
153	CHD2variants are a risk factor for photosensitivity in epilepsy. Brain, 2015, 138, 1198-1208.	3.7	112
154	Myoclonic occipital photosensitive epilepsy with dystonia (MOPED): A familial epilepsy syndrome. Epilepsy Research, 2015, 114, 98-105.	0.8	7
155	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. Human Molecular Genetics, 2015, 24, 5250-5259.	1.4	93
156	CHD2 myoclonic encephalopathy is frequently associated with self-induced seizures. Neurology, 2015, 84, 951-958.	1.5	79
157	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. American Journal of Human Genetics, 2015, 96, 808-815.	2.6	173
158	Cortical microarchitecture changes in genetic epilepsy. Neurology, 2015, 84, 1308-1316.	1.5	16
159	Evaluation of multiple putative risk alleles within the 15q13.3 region for genetic generalized epilepsy. Epilepsy Research, 2015, 117, 70-73.	0.8	6
160	Electroclinical spectrum of the neuronal ceroid lipofuscinoses associated with CLN6 mutations. Neurology, 2015, 85, 316-324.	1.5	40
161	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. Nature Genetics, 2015, 47, 39-46.	9.4	245
162	Harnessing Gene Expression Networks to Prioritize Candidate Epileptic Encephalopathy Genes. PLoS ONE, 2014, 9, e102079.	1.1	25

#	Article	IF	Citations
163	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. Human Molecular Genetics, 2014, 23, 6069-6080.	1.4	61
164	Genetic analysis of PHOX2B in sudden unexpected death in epilepsy cases. Neurology, 2014, 83, 1018-1021.	1.5	19
165	The genetic basis of music ability. Frontiers in Psychology, 2014, 5, 658.	1.1	60
166	KCNT1 gain of function in 2 epilepsy phenotypes is reversed by quinidine. Annals of Neurology, 2014, 75, 581-590.	2.8	249
167	Atypical multifocal <scp>D</scp>ravet syndrome lacks generalized seizures and may show later cognitive decline. Developmental Medicine and Child Neurology, 2014, 56, 85-90.	1.1	16
168	Is focal cortical dysplasia sporadic? Family evidence for genetic susceptibility. Epilepsia, 2014, 55, e22-6.	2.6	23
169	Glucose metabolism transporters and epilepsy: Only <scp>GLUT</scp>1 has an established role. Epilepsia, 2014, 55, e18-21.	2.6	29
170	Mutations in mammalian target of rapamycin regulator DEPDC5 cause focal epilepsy with brain malformations. Annals of Neurology, 2014, 75, 782-787.	2.8	193
171	Genetics of epilepsy. Neurology, 2014, 83, 1042-1048.	1.5	61
172	Genetics of vasovagal syncope. Autonomic Neuroscience: Basic and Clinical, 2014, 184, 60-65.	1.4	19
173	Does variation in NIPA2 contribute to genetic generalized epilepsy?. Human Genetics, 2014, 133, 673-674.	1.8	7
174	A genome-wide association study and biological pathway analysis of epilepsy prognosis in a prospective cohort of newly treated epilepsy. Human Molecular Genetics, 2014, 23, 247-258.	1.4	33
175	Reduced dendritic arborization and hyperexcitability of pyramidal neurons in a Scn1b-based model of Dravet syndrome. Brain, 2014, 137, 1701-1715.	3.7	49
176	The hidden genetics of epilepsyâ€"a clinically important new paradigm. Nature Reviews Neurology, 2014, 10, 283-292.	4.9	232
177	Rasmussen encephalitis and comorbid autoimmune diseases. Neurology, 2014, 83, 1049-1055.	1.5	22
178	Recurrent generalized seizures, visual loss, and palinopsia as phenotypic features of neuronal ceroid lipofuscinosis due to progranulin gene mutation. Epilepsia, 2014, 55, e56-9.	2.6	50
179	Weight and fat distribution in patients taking valproate: A valproateâ€discordant genderâ€matched twin and sibling pair study. Epilepsia, 2014, 55, 1551-1557.	2.6	11
180	Brivaracetam as adjunctive treatment for uncontrolled partial epilepsy in adults: A phase <scp>III</scp> randomized, doubleâ€blind, placeboâ€controlled trial. Epilepsia, 2014, 55, 57-66.	2.6	217

#	Article	IF	Citations
181	â€˜Idiopathicâ€™ no more! Abnormal interaction of large-scale brain networks in generalized epilepsy. Brain, 2014, 137, 2400-2402.	3.7	9
182	GABRA1 and STXBP1 : Novel genetic causes of Dravet syndrome. Neurology, 2014, 82, 1245-1253.	1.5	229
183	Somatic Mutations in Cerebral Cortical Malformations. New England Journal of Medicine, 2014, 371, 733-743.	13.9	326
184	Phenotypeâ€“genotype complexities: opening DOORS. Lancet Neurology, The, 2014, 13, 24-25.	4.9	0
185	Concepts and controversies of juvenile myoclonic epilepsy: still an enigmatic epilepsy. Expert Review of Neurotherapeutics, 2014, 14, 819-831.	1.4	30
186	A variant of <scp>KCC</scp> 2 from patients with febrile seizures impairs neuronal Cl ^{â^’} extrusion and dendritic spine formation. EMBO Reports, 2014, 15, 723-729.	2.0	163
187	De novo mutations in epileptic encephalopathies. Nature, 2013, 501, 217-221.	13.7	1,351
188	GRIN2A mutations cause epilepsy-aphasia spectrum disorders. Nature Genetics, 2013, 45, 1073-1076.	9.4	326
189	Recent advances in the molecular genetics of epilepsy. Journal of Medical Genetics, 2013, 50, 271-279.	1.5	111
190	Do mutations in SCN1B cause Dravet syndrome?. Epilepsy Research, 2013, 103, 97-100.	0.8	11
191	Role of the sodium channel <scp>SCN</scp>9A in genetic epilepsy with febrile seizures plus and Dravet syndrome. Epilepsia, 2013, 54, e122-6.	2.6	62
192	Abnormal Processing of Autophagosomes in Transformed B Lymphocytes from SCARB2-Deficient Subjects. BioResearch Open Access, 2013, 2, 40-46.	2.6	9
193	Copy number variants are frequent in genetic generalized epilepsy with intellectual disability. Neurology, 2013, 81, 1507-1514.	1.5	140
194	Seizure semiology in autosomal dominant epilepsy with auditory features, due to novel LGI1 mutations. Epilepsy Research, 2013, 107, 311-317.	0.8	15
195	TBC1D24 mutation associated with focal epilepsy, cognitive impairment and a distinctive cerebro-cerebellar malformation. Epilepsy Research, 2013, 105, 240-244.	0.8	28
196	Mutations in DEPDC5 cause familial focal epilepsy with variable foci. Nature Genetics, 2013, 45, 546-551.	9.4	301
197	SCN1A testing for epilepsy: Application in clinical practice. Epilepsia, 2013, 54, 946-952.	2.6	67
198	CORTICAL EXCITABILITY AND REFRACTORY EPILEPSY: A THREE-YEAR LONGITUDINAL TRANSCRANIAL MAGNETIC STIMULATION STUDY. International Journal of Neural Systems, 2013, 23, 1250030.	3.2	63

#	Article	IF	Citations
199	Prediction of seizure likelihood with a long-term, implanted seizure advisory system in patients with drug-resistant epilepsy: a first-in-man study. Lancet Neurology, The, 2013, 12, 563-571.	4.9	674
200	Deletions of 16p11.2 and 19p13.2 in a family with intellectual disability and generalized epilepsy. American Journal of Medical Genetics, Part A, 2013, 161, 1722-1725.	0.7	18
201	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. Nature Genetics, 2013, 45, 825-830.	9.4	589
202	Genetics of febrile seizure subtypes and syndromes: A twin study. Epilepsy Research, 2013, 105, 103-109.	0.8	36
203	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. Brain, 2013, 136, 3378-3394.	3.7	85
204	â€˜North Seaâ€™ progressive myoclonus epilepsy: phenotype of subjects with GOSR2 mutation. Brain, 2013, 136, 1146-1154.	3.7	129
205	Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. Brain, 2013, 136, 3140-3150.	3.7	168
206	Autosomal dominant vasovagal syncope. Neurology, 2013, 80, 1485-1493.	1.5	20
207	Etiology of hippocampal sclerosis: Evidence for a predisposing familial morphologic anomaly. Neurology, 2013, 81, 144-149.	1.5	51
208	Cathepsin F mutations cause Type B Kufs disease, an adult-onset neuronal ceroid lipofuscinosis. Human Molecular Genetics, 2013, 22, 1417-1423.	1.4	105
209	Networks underlying paroxysmal fast activity and slow spike and wave in Lennox-Gastaut syndrome. Neurology, 2013, 81, 665-673.	1.5	65
210	NOCTURNAL SEIZURES ONLY POST TEMPORAL LOBECTOMY: CHARACTERISTICS OF AN UNUSUAL OUTCOME GROUP. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, e2.5-e2.	0.9	0
211	Genetics of epilepsy syndromes in families with photosensitivity. Neurology, 2013, 80, 1322-1329.	1.5	40
212	Are myotonia and epilepsy linked by a chloride channel?. Neurology, 2013, 80, 1074-1075.	1.5	3
213	New Hyperekplexia Mutations Provide Insight into Glycine Receptor Assembly, Trafficking, and Activation Mechanisms. Journal of Biological Chemistry, 2013, 288, 33745-33759.	1.6	35
214	Multiple molecular mechanisms for a single GABA _A mutation in epilepsy. Neurology, 2013, 80, 1003-1008.	1.5	67
215	Clinical genetic study of the epilepsyâ€aphasia spectrum. Epilepsia, 2013, 54, 280-287.	2.6	44
216	Siblings with refractory occipital epilepsy showing localized network activity on <scp>EEG</scp>â€f<scp>MRI</scp>. Epilepsia, 2013, 54, e28-32.	2.6	4

#	Article	IF	Citations
217	Mutations in TNK2 in severe autosomal recessive infantile onset epilepsy. Annals of Neurology, 2013, 74, 496-501.	2.8	22
218	Mutations in <scp>PRRT</scp>2 are not a common cause of infantile epileptic encephalopathies. Epilepsia, 2013, 54, e86-9.	2.6	12
219	Progressive Gait Deterioration in Adolescents With Dravet Syndrome. Archives of Neurology, 2012, 69, 873-8.	4.9	95
220	Evidence for genetic factors in vasovagal syncope. Neurology, 2012, 79, 561-565.	1.5	20
221	Benign Neonatal Sleep Myoclonus. Journal of Child Neurology, 2012, 27, 1260-1263.	0.7	10
222	Familial Adult Myoclonic Epilepsy. Archives of Neurology, 2012, 69, 474.	4.9	36
223	Missense mutations in the sodium-gated potassium channel gene KCNT1 cause severe autosomal dominant nocturnal frontal lobe epilepsy. Nature Genetics, 2012, 44, 1188-1190.	9.4	333
224	Familial focal epilepsy with variable foci mapped to chromosome 22q12: Expansion of the phenotypic spectrum. Epilepsia, 2012, 53, e151-5.	2.6	24
225	Psychological trajectories in the year after a newly diagnosed seizure. Epilepsia, 2012, 53, 1774-1781.	2.6	41
226	Genome-wide association analysis of genetic generalized epilepsies implicates susceptibility loci at 1q43, 2p16.1, 2q22.3 and 17q21.32. Human Molecular Genetics, 2012, 21, 5359-5372.	1.4	134
227	In vivo loss of slow potassium channel activity in individuals with benign familial neonatal epilepsy in remission. Brain, 2012, 135, 3144-3152.	3.7	18
228	Early onset absence epilepsy: 1 in 10 cases is caused by GLUT1 deficiency. Epilepsia, 2012, 53, e204-7.	2.6	97
229	Glucose transporter 1 deficiency in the idiopathic generalized epilepsies. Annals of Neurology, 2012, 72, 807-815.	2.8	123
230	Significance of post-operative auras after temporal lobectomy: A surprising methodological trap. Epilepsy and Behavior, 2012, 23, 348-352.	0.9	1
231	Peritrigonal and temporo-occipital heterotopia with corpus callosum and cerebellar dysgenesis. Neurology, 2012, 79, 1244-1251.	1.5	31
232	PRRT2 phenotypic spectrum includes sporadic and fever-related infantile seizures. Neurology, 2012, 79, 2104-2108.	1.5	75
233	Sodium channels and the neurobiology of epilepsy. Epilepsia, 2012, 53, 1849-1859.	2.6	105
234	Rare protein sequence variation in SV2A gene does not affect response to levetiracetam. Epilepsy Research, 2012, 101, 277-279.	0.8	11

#	Article	IF	Citations
235	Overcoming Barriers to Successful Epilepsy Management. Epilepsy Currents, 2012, 12, 158-160.	0.4	8
236	A case of severe hearing loss in action myoclonus renal failure syndrome resulting from mutation in SCARB2. Movement Disorders, 2012, 27, 1200-1201.	2.2	17
237	PRRT2 Mutations Cause Benign Familial Infantile Epilepsy and Infantile Convulsions with Choreoathetosis Syndrome. American Journal of Human Genetics, 2012, 90, 152-160.	2.6	234
238	Strikingly Different Clinicopathological Phenotypes Determined by Progranulin-Mutation Dosage. American Journal of Human Genetics, 2012, 90, 1102-1107.	2.6	414
239	Longâ€term followâ€up of febrile infectionâ€"related epilepsy syndrome. Epilepsia, 2012, 53, 101-110.	2.6	100
240	Clinical genetic studies in benign childhood epilepsy with centrotemporal spikes. Epilepsia, 2012, 53, 319-324.	2.6	49
241	Genomeâ€wide linkage metaâ€analysis identifies susceptibility loci at 2q34 and 13q31.3 for genetic generalized epilepsies. Epilepsia, 2012, 53, 308-318.	2.6	32
242	Inter-session repeatability of cortical excitability measurements in patients with epilepsy. Epilepsy Research, 2012, 98, 182-186.	0.8	16
243	Febrile infection-related epilepsy syndrome is not caused by SCN1A mutations. Epilepsy Research, 2012, 100, 194-198.	0.8	9
244	Longâ€term seizure outcome and risk factors for recurrence after extratemporal epilepsy surgery. Epilepsia, 2012, 53, 970-978.	2.6	91
245	KCNQ2 encephalopathy: Emerging phenotype of a neonatal epileptic encephalopathy. Annals of Neurology, 2012, 71, 15-25.	2.8	427
246	Benign mesial temporal lobe epilepsy. Nature Reviews Neurology, 2011, 7, 237-240.	4.9	76
247	The Role of Seizure-RelatedSEZ6as a Susceptibility Gene in Febrile Seizures. Neurology Research International, 2011, 2011, 1-4.	0.5	20
248	Low blood glucose precipitates spike-and-wave activity in genetically predisposed animals. Epilepsia, 2011, 52, 115-120.	2.6	24
249	A retrospective populationâ€based study on seizures related to childhood vaccination. Epilepsia, 2011, 52, 1506-1512.	2.6	17
250	Investigation of the 15q13.3 CNV as a genetic modifier for familial epilepsies with variable phenotypes. Epilepsia, 2011, 52, e139-e142.	2.6	9
251	Clinical and neurophysiologic features of progressive myoclonus epilepsy without renal failure caused bySCARB2mutations. Epilepsia, 2011, 52, 2356-2363.	2.6	63
252	Kufs Disease, the Major Adult Form of Neuronal Ceroid Lipofuscinosis, Caused by Mutations in CLN6. American Journal of Human Genetics, 2011, 88, 566-573.	2.6	253

#	Article	IF	Citations
253	A Mutation in the Golgi Qb-SNARE Gene GOSR2 Causes Progressive Myoclonus Epilepsy with Early Ataxia. American Journal of Human Genetics, 2011, 88, 657-663.	2.6	166
254	Rare copy number variants are an important cause of epileptic encephalopathies. Annals of Neurology, 2011, 70, 974-985.	2.8	222
255	Molecular analysis of ring chromosome 20 syndrome reveals two distinct groups of patients. Journal of Medical Genetics, 2011, 48, 1-9.	1.5	61
256	Glucose Transporter 1 Deficiency as a Treatable Cause of Myoclonic Astatic Epilepsy. Archives of Neurology, 2011, 68, 1152.	4.9	121
257	Mutation of SCARB2 in a Patient With Progressive Myoclonus Epilepsy and Demyelinating Peripheral Neuropathy. Archives of Neurology, 2011, 68, 812-3.	4.9	28
258	Profiles of psychosocial outcome after epilepsy surgery: The role of personality. Epilepsia, 2010, 51, 1133-1138.	2.6	36
259	A Focal Epilepsy and Intellectual Disability Syndrome Is Due to a Mutation in TBC1D24. American Journal of Human Genetics, 2010, 87, 371-375.	2.6	111
260	Epilepsy: insights into causes and treatment dilemmas. Lancet Neurology, The, 2010, 9, 9-11.	4.9	0
261	Effects of vaccination on onset and outcome of Dravet syndrome: a retrospective study. Lancet Neurology, The, 2010, 9, 592-598.	4.9	119
262	Vaccination and Dravet syndrome â€“ Authors' reply. Lancet Neurology, The, 2010, 9, 1148-1149.	4.9	1
263	New therapeutic opportunities in epilepsy: A genetic perspective. , 2010, 128, 274-280.		11
264	Predicting seizure control: Cortical excitability and antiepileptic medication. Annals of Neurology, 2010, 67, 64-73.	2.8	84
265	Augmented currents of an HCN2 variant in patients with febrile seizure syndromes. Annals of Neurology, 2010, 67, 542-546.	2.8	96
266	Balance impairment in chronic antiepileptic drug users: A twin and sibling study. Epilepsia, 2010, 51, 280-288.	2.6	42
267	Neonatal seizures and long QT Syndrome: A cardiocerebral channelopathy?. Epilepsia, 2010, 51, 293-296.	2.6	61
268	The Epilepsy Genetic Association Database (epiGAD): Analysis of 165 genetic association studies, 1996â€“2008. Epilepsia, 2010, 51, 686-689.	2.6	43
269	The borderland of epilepsy: A clinical and molecular view, 100â€ƒyears on. Epilepsia, 2010, 51, 3-4.	2.6	12
270	Revised terminology and concepts for organization of seizures and epilepsies: Report of the ILAE Commission on Classification and Terminology, 2005â€“2009. Epilepsia, 2010, 51, 676-685.	2.6	3,612

#	Article	IF	Citations
271	Clinical features of seizures associated with parahippocampal/inferior temporal lesions compared to those with hippocampal sclerosis. Epilepsia, 2010, 51, 1906-1909.	2.6	7
272	Can changes in cortical excitability distinguish progressive from juvenile myoclonic epilepsy?. Epilepsia, 2010, 51, 2084-2088.	2.6	32
273	Familial neonatal seizures with intellectual disability caused by a microduplication of chromosome 2q24.3. Epilepsia, 2010, 51, 1865-1869.	2.6	30
274	Small temporal pole encephaloceles: A treatable cause of â€œlesion negativeâ€•temporal lobe epilepsy. Epilepsia, 2010, 51, 2199-2202.	2.6	62
275	Familial Lennoxâ€Gastaut syndrome in male siblings with a novel DCX mutation and anterior pachygyria. Epilepsia, 2010, 51, 1902-1905.	2.6	9
276	A duplication in 1q21.3 in a family with early onset and childhood absence epilepsy. Epilepsia, 2010, 51, 2453-2456.	2.6	12
277	Mild adolescent/adult onset epilepsy and paroxysmal exerciseâ€•induced dyskinesia due to GLUT1 deficiency. Epilepsia, 2010, 51, 2466-2469.	2.6	22
278	Axon initial segment dysfunction in epilepsy. Journal of Physiology, 2010, 588, 1829-1840.	1.3	80
279	Copy number variants--an unexpected risk factor for the idiopathic generalized epilepsies. Brain, 2010, 133, 7-8.	3.7	20
280	Timing of De Novo Mutagenesis â€” A Twin Study of Sodium-Channel Mutations. New England Journal of Medicine, 2010, 363, 1335-1340.	13.9	100
281	Epilepsy and mental retardation limited to females with PCDH19 mutations can present de novo or in single generation families. Journal of Medical Genetics, 2010, 47, 211-216.	1.5	74
282	Clinical and imaging heterogeneity of polymicrogyria: a study of 328 patients. Brain, 2010, 133, 1415-1427.	3.7	215
283	Familial mesial temporal lobe epilepsy: a benign epilepsy syndrome showing complex inheritance. Brain, 2010, 133, 3221-3231.	3.7	74
284	Key epilepsy gene gets further phenotypic delineation. Neurology, 2010, 75, 18-19.	1.5	0
285	De novo SCN1A mutations in Dravet syndrome and related epileptic encephalopathies are largely of paternal origin. Journal of Medical Genetics, 2010, 47, 137-141.	1.5	44
286	Detection of microchromosomal aberrations in refractory epilepsy: a pilot study. Epileptic Disorders, 2010, 12, 192-198.	0.7	14
287	35. Peripheral nerve excitability testing shows distinctive ion channel dysfunction in patients with KCNQ2 mutations and epilepsy. Journal of Clinical Neuroscience, 2010, 17, 1621.	0.8	0
288	Neuropsychological function in patients with a single gene mutation associated with autosomal dominant nocturnal frontal lobe epilepsy. Epilepsy and Behavior, 2010, 17, 531-535.	0.9	14

#	Article	IF	Citations
289	Reduced variance in monozygous twins for multiple MR parameters: Implications for disease studies and the genetic basis of brain structure. NeuroImage, 2010, 49, 1536-1544.	2.1	7
290	Axon initial segment dysfunction in a mouse model of genetic epilepsy with febrile seizures plus. Journal of Clinical Investigation, 2010, 120, 2661-2671.	3.9	77
291	Familial and sporadic 15q13.3 microdeletions in idiopathic generalized epilepsy: precedent for disorders with complex inheritance. Human Molecular Genetics, 2009, 18, 3626-3631.	1.4	211
292	The peri-ictal state: cortical excitability changes within 24 h of a seizure. Brain, 2009, 132, 1013-1021.	3.7	108
293	Prediction by Modeling That Epilepsy May Be Caused by Very Small Functional Changes in Ion Channels. Archives of Neurology, 2009, 66, 1225-32.	4.9	44
294	The borderland of epilepsy: clinical and molecular features of phenomena that mimic epileptic seizures. Lancet Neurology, The, 2009, 8, 370-381.	4.9	88
295	Earlyâ€onset absence epilepsy caused by mutations in the glucose transporter GLUT1. Annals of Neurology, 2009, 66, 415-419.	2.8	266
296	SCARB2 mutations in progressive myoclonus epilepsy (PME) without renal failure. Annals of Neurology, 2009, 66, 532-536.	2.8	90
297	Cognitive complaints after a first seizure in adulthood: Influence of psychological adjustment. Epilepsia, 2009, 50, 1012-1021.	2.6	38
298	SCN1A duplications and deletions detected in Dravet syndrome: Implications for molecular diagnosis. Epilepsia, 2009, 50, 1670-1678.	2.6	152
299	Does a SCN1A gene mutation confer earlier age of onset of febrile seizures in GEFS+?. Epilepsia, 2009, 50, 953-956.	2.6	22
300	Multidrugâ€resistant genotype (ABCB1) and seizure recurrence in newly treated epilepsy: Data from international pharmacogenetic cohorts. Epilepsia, 2009, 50, 1689-1696.	2.6	39
301	Mechanisms of human inherited epilepsies. Progress in Neurobiology, 2009, 87, 41-57.	2.8	185
302	102. Epilepsy, AEDs and Balance Function - A Twin and Sibling Study. Journal of Clinical Neuroscience, 2009, 16, 462.	0.8	0
303	Bone Health and Age of Commencement of Anti-epileptic Medication: An AED-Discordant Twin and Sibling Pair Study. Journal of Clinical Neuroscience, 2009, 16, 1522.	0.8	0
304	Graft-Versus-Host Disease. , 2009, , 746-746.		0
305	Personality Development in the Context of Intractable Epilepsy. Archives of Neurology, 2009, 66, 68-72.	4.9	34
306	NREM Arousal Parasomnias and Their Distinction from Nocturnal Frontal Lobe Epilepsy: A Video EEG Analysis. Sleep, 2009, 32, 1637-1644.	0.6	195

#	Article	IF	Citations
307	Generalized (Genetic) Epilepsy with Febrile Seizures Plus, Severe Myoclonic Epilepsy of Infancy. , 2009, , 693-695.		0
308	Developmental impact of a familial GABA_A receptor epilepsy mutation. Annals of Neurology, 2008, 64, 284-293.	2.8	55
309	Array-Based Gene Discovery with Three Unrelated Subjects Shows SCARB2/LIMP-2 Deficiency Causes Myoclonus Epilepsy and Glomerulosclerosis. American Journal of Human Genetics, 2008, 82, 673-684.	2.6	230
310	A Homozygous Mutation in Human PRICKLE1 Causes an Autosomal-Recessive Progressive Myoclonus Epilepsy-Ataxia Syndrome. American Journal of Human Genetics, 2008, 83, 572-581.	2.6	199
311	X-linked protocadherin 19 mutations cause female-limited epilepsy and cognitive impairment. Nature Genetics, 2008, 40, 776-781.	9.4	397
312	Gene expression analysis in absence epilepsy using a monozygotic twin design. Epilepsia, 2008, 49, 1546-1554.	2.6	24
313	Severe autosomal dominant nocturnal frontal lobe epilepsy associated with psychiatric disorders and intellectual disability. Epilepsia, 2008, 49, 2125-2129.	2.6	49
314	Navigating the channels and beyond: unravelling the genetics of the epilepsies. Lancet Neurology, The, 2008, 7, 231-245.	4.9	249
315	452: Chronic anti-epileptic drug treatment is associated with lower balance function scores â€“ a twin and matched sibling AED-discordant pair study. Journal of Clinical Neuroscience, 2008, 15, 360-361.	0.8	0
316	Obstetric Events as a Risk Factor for Febrile Seizures: A Community-Based Twin Study. Twin Research and Human Genetics, 2008, 11, 634-640.	0.3	2
317	Intracortical Hyperexcitability in Humans with a GABAA Receptor Mutation. Cerebral Cortex, 2008, 18, 664-669.	1.6	59
318	Paroxysmal exercise-induced dyskinesia and epilepsy is due to mutations in SLC2A1, encoding the glucose transporter GLUT1. Brain, 2008, 131, 1831-1844.	3.7	340
319	Benign occipital epilepsies of childhood: clinical features and genetics. Brain, 2008, 131, 2287-2294.	3.7	63
320	The Human Variome Project. Science, 2008, 322, 861-862.	6.0	63
321	Human Nocturnal Frontal Lobe Epilepsy: Pharmocogenomic Profiles of Pathogenic Nicotinic Acetylcholine Receptor Î²-Subunit Mutations outside the Ion Channel Pore. Molecular Pharmacology, 2008, 74, 379-391.	1.0	58
322	Association of a Nicotinic Receptor Mutation with Reduced Height and Blunted Physostigmine-Stimulated Growth Hormone Release. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 634-637.	1.8	8
323	Epilepsy and mental retardation limited to females: an under-recognized disorder. Brain, 2008, 131, 918-927.	3.7	172
324	Multifocal epilepsy: the role of palliative resection - intractable frontal and occipital lobe epilepsy secondary to radiotherapy for acute lymphoblastic leukaemia. Epileptic Disorders, 2008, 10, 362-70.	0.7	6

#	Article	IF	Citations
325	Reduced cortical inhibition in a mouse model of familial childhood absence epilepsy. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 17536-17541.	3.3	192
326	Nicotine-Induced Dystonic Arousal Complex in a Mouse Line Harboring a Human Autosomal-Dominant Nocturnal Frontal Lobe Epilepsy Mutation. Journal of Neuroscience, 2007, 27, 10128-10142.	1.7	72
327	Impaired verbal associative learning after resection of left perirhinal cortex. Brain, 2007, 130, 1423-1431.	3.7	27
328	The spectrum of SCN1A-related infantile epileptic encephalopathies. Brain, 2007, 130, 843-852.	3.7	501
329	Scale for Distinguishing Sleep Disorders From Seizuresâ€"Reply. Archives of Neurology, 2007, 64, 1206.	4.9	0
330	Placebo-controlled study of levetiracetam in idiopathic generalized epilepsy. Neurology, 2007, 69, 1751-1760.	1.5	246
331	Vaccination, seizures and â€˜vaccine damageâ€™. Current Opinion in Neurology, 2007, 20, 181-187.	1.8	38
332	The psychological impact of a newly diagnosed seizure: Losing and restoring perceived control. Epilepsy and Behavior, 2007, 10, 223-233.	0.9	72
333	Adjunctive therapy of uncontrolled partial seizures with levetiracetam in Australian patients. Epilepsy and Behavior, 2007, 11, 338-342.	0.9	15
334	646: Chronic anti-epileptic drug treatment is associated with clinically-significant impairment in balance function â€“ a twin and matched sibling pair pilot study. Journal of Clinical Neuroscience, 2007, 14, 1034.	0.8	0
335	A childhood epilepsy mutation reveals a role for developmentally regulated splicing of a sodium channel. Molecular and Cellular Neurosciences, 2007, 35, 292-301.	1.0	68
336	627: Diagnosing bumps in the night: Distinguishing parasomnias from NFLE using video EEG monitoring. Journal of Clinical Neuroscience, 2007, 14, 1027.	0.8	0
337	Genetic Epilepsies. , 2007, , 371-383.		1
338	Changes in cortical excitability differentiate generalized and focal epilepsy. Annals of Neurology, 2007, 61, 324-331.	2.8	114
339	Extended spectrum of idiopathic generalized epilepsies associated with CACNA1H functional variants. Annals of Neurology, 2007, 62, 560-568.	2.8	186
340	Channelopathies in idiopathic epilepsy. Neurotherapeutics, 2007, 4, 295-304.	2.1	101
341	Association studies and functional validation or functional validation alone?. Epilepsy Research, 2007, 74, 237-238.	0.8	3
342	Absence of mutations in the LGI1 receptor ADAM22 gene in autosomal dominant lateral temporal epilepsy. Epilepsy Research, 2007, 76, 41-48.	0.8	24

#	Article	IF	Citations
343	Hippocampal Sclerosis: MR Prediction of Seizure Intractability. Epilepsia, 2007, 48, 315-323.	2.6	23
344	A Multicenter Study of BRD2 as a Risk Factor for Juvenile Myoclonic Epilepsy. Epilepsia, 2007, 48, 706-712.	2.6	76
345	Founder Effect with Variable Age at Onset in Arab Families with Lafora Disease and EPM2A Mutation. Epilepsia, 2007, 48, 1011-1014.	2.6	22
346	Response to Tinuper et al.. Epilepsia, 2007, 48, 1034-1034.	2.6	0
347	SCN2A Mutations and Benign Familial Neonatal-Infantile Seizures: The Phenotypic Spectrum. Epilepsia, 2007, 48, 1138-1142.	2.6	102
348	Is Photosensitive Epilepsy Less Common in Males Due to Variation in X Chromosome Photopigment Genes?. Epilepsia, 2007, 48, 1807-1809.	2.6	10
349	Multicentre search for genetic susceptibility loci in sporadic epilepsy syndrome and seizure types: a case-control study. Lancet Neurology, The, 2007, 6, 970-980.	4.9	175
350	Contributor's List. , 2007, , vii-ix.		0
351	GEFS ⁺ where focal seizures evolve from generalized spike wave: videoâ€EEG study of two children. Epileptic Disorders, 2007, 9, 307-314.	0.7	10
352	Increased serotonin receptor availability in human sleep: Evidence from an [18F]MPPF PET study in narcolepsy. NeuroImage, 2006, 30, 341-348.	2.1	47
353	A GABAA receptor mutation causing generalized epilepsy reduces benzodiazepine receptor binding. NeuroImage, 2006, 32, 995-1000.	2.1	36
354	Febrile seizures: traffic slows in the heat. Trends in Molecular Medicine, 2006, 12, 343-344.	3.5	6
355	Human epilepsies: interaction of genetic and acquired factors. Trends in Neurosciences, 2006, 29, 391-397.	4.2	190
356	Analyzing the Etiology of Benign Rolandic Epilepsy: A Multicenter Twin Collaboration. Epilepsia, 2006, 47, 550-555.	2.6	135
357	Paroxysmal Motor Disorders of Sleep: The Clinical Spectrum and Differentiation from Epilepsy. Epilepsia, 2006, 47, 1775-1791.	2.6	149
358	Exploration of the Genetic Architecture of Idiopathic Generalized Epilepsies. Epilepsia, 2006, 47, 1682-1690.	2.6	45
359	Comment. Epilepsia, 2006, 47, 1751-1752.	2.6	0
360	Update on pharmacogenetics in epilepsy: a brief review. Lancet Neurology, The, 2006, 5, 189-196.	4.9	70

#	Article	IF	Citations
361	De-novo mutations of the sodium channel gene SCN1A in alleged vaccine encephalopathy: a retrospective study. Lancet Neurology, The, 2006, 5, 488-492.	4.9	295
362	Prediction of drug resistance in epilepsy: not as easy as ABC. Lancet Neurology, The, 2006, 5, 641-642.	4.9	5
363	What happens now? Ongoing outcome after post-temporal lobectomy seizure recurrence. Neurology, 2006, 67, 1671-1673.	1.5	10
364	Distinguishing Sleep Disorders From Seizures. Archives of Neurology, 2006, 63, 705.	4.9	223
365	SRPX2 mutations in disorders of language cortex and cognition. Human Molecular Genetics, 2006, 15, 1195-1207.	1.4	248
366	Temporal lobe epilepsy and GEFS+ phenotypes associated with SCN1B mutations. Brain, 2006, 130, 100-109.	3.7	234
367	Efficacy and safety of levetiracetam 1000â€“3000mg/day in patients with refractory partial-onset seizures: a multicenter, open-label single-arm study. Epilepsy Research, 2005, 63, 1-9.	0.8	39
368	Neonatal Epilepsy Syndromes and Generalized Epilepsy with Febrile Seizures Plus (GEFS+). Epilepsia, 2005, 46, 41-47.	2.6	63
369	Is Variation in the GABA(B) Receptor 1 Gene Associated with Temporal Lobe Epilepsy?. Epilepsia, 2005, 46, 778-780.	2.6	15
370	SCN1Amutations and epilepsy. Human Mutation, 2005, 25, 535-542.	1.1	327
371	Early seizures after temporal lobectomy predict subsequent seizure recurrence. Annals of Neurology, 2005, 57, 283-288.	2.8	38
372	Assessment of the role of FDG PET in the diagnosis and management of children with refractory epilepsy. European Journal of Nuclear Medicine and Molecular Imaging, 2005, 32, 1311-1316.	3.3	49
373	Tramadol and newâ€onset seizures. Medical Journal of Australia, 2005, 182, 42-43.	0.8	27
374	Mutation in the Na+ channel subunit SCN1B produces paradoxical changes in peripheral nerve excitability. Brain, 2005, 128, 1841-1846.	3.7	54
375	A new clinical and molecular form of Unverricht-Lundborg disease localized by homozygosity mapping. Brain, 2005, 128, 652-658.	3.7	45
376	Treatment of new-onset epilepsy: seizures beget discussion. Lancet, The, 2005, 365, 1985-1986.	6.3	12
377	Susceptibility genes for complex epilepsy. Human Molecular Genetics, 2005, 14, R243-R249.	1.4	92
378	Introduction to Epilepsy. , 2005, , 1-16.		3

#	Article	IF	Citations
379	Treatment with anti-epileptic drugs. Australian Family Physician, 2005, 34, 1017-20.	0.5	10
380	GABRD encoding a protein for extra- or peri-synaptic GABAA receptors is a susceptibility locus for generalized epilepsies. Human Molecular Genetics, 2004, 13, 1315-1319.	1.4	299
381	Action myoclonus-renal failure syndrome: characterization of a unique cerebro-renal disorder. Brain, 2004, 127, 2173-2182.	3.7	89
382	Juvenile myoclonic epilepsy and idiopathic photosensitive occipital lobe epilepsy: is there overlap?. Brain, 2004, 127, 1878-1886.	3.7	72
383	Genetic Association Studies in Epilepsy: "The Truth Is Out There". Epilepsia, 2004, 45, 1429-1442.	2.6	179
384	Familial Partial Epilepsy with Variable Foci: Clinical Features and Linkage to Chromosome 22q12. Epilepsia, 2004, 45, 1054-1060.	2.6	71
385	Genetic Architecture of Idiopathic Generalized Epilepsy: Clinical Genetic Analysis of 55 Multiplex Families. Epilepsia, 2004, 45, 467-478.	2.6	128
386	Subtle Microscopic Abnormalities in Hippocampal Sclerosis Do Not Predict Clinical Features of Temporal Lobe Epilepsy. Epilepsia, 2004, 45, 940-947.	2.6	26
387	Genetic variation ofCACNA1H in idiopathic generalized epilepsy. Annals of Neurology, 2004, 55, 595-596.	2.8	102
388	Benign familial neonatal-infantile seizures: Characterization of a new sodium channelopathy. Annals of Neurology, 2004, 55, 550-557.	2.8	250
389	Is benign rolandic epilepsy genetically determined?. Annals of Neurology, 2004, 56, 129-132.	2.8	52
390	Temporal lobectomy: long-term seizure outcome, late recurrence and risks for seizure recurrence. Brain, 2004, 127, 2018-2030.	3.7	510
391	Glioneuronal tumours in neurofibromatosis type 1: MRI-pathological study. Journal of Clinical Neuroscience, 2004, 11, 745-747.	0.8	24
392	Chapter 42 The idiopathic generalized epilepsies across life. Supplements To Clinical Neurophysiology, 2004, 57, 408-414.	2.1	1
393	MR imaging and spectroscopic study of epileptogenic hypothalamic hamartomas: analysis of 72 cases. American Journal of Neuroradiology, 2004, 25, 450-62.	1.2	134
394	Epilepsy in Offspring of Whom Both Parents Have Idiopathic Generalized Epilepsy: Biparental Inheritance. Epilepsia, 2003, 44, 1250-1254.	2.6	6
395	Phenotypic Comparison of Two Scottish Families with Mutations in Different Genes Causing Autosomal Dominant Nocturnal Frontal Lobe Epilepsy. Epilepsia, 2003, 44, 613-617.	2.6	67
396	EEG in Adultâ€onset Idiopathic Generalizedâ€ƒEpilepsy. Epilepsia, 2003, 44, 252-256.	2.6	41

#	Article	IF	Citations
397	Hypothalamic Hamartoma and Seizures: A Treatable Epileptic Encephalopathy. Epilepsia, 2003, 44, 969-973.	2.6	153
398	Occipital epilepsies: identification of specific and newly recognized syndromes. Brain, 2003, 126, 753-769.	3.7	142
399	The genetics of human epilepsy. Trends in Pharmacological Sciences, 2003, 24, 428-433.	4.0	131
400	Risk factors for sudden unexpected death in epilepsy: a controlled prospective study based on coroners cases. Seizure: the Journal of the British Epilepsy Association, 2003, 12, 456-464.	0.9	140
401	Childhood absence epilepsy and febrile seizures: a family with a GABAA receptor mutation. Brain, 2003, 126, 230-240.	3.7	148
402	Channelopathies as a genetic cause of epilepsy. Current Opinion in Neurology, 2003, 16, 171-176.	1.8	153
403	A Twin Study of Genetic Influences on Epilepsy Outcome. Twin Research and Human Genetics, 2003, 6, 140-146.	1.5	17
404	Generics - equal or not?. Australian Prescriber, 2003, 26, 124-125.	0.5	1
405	Channelopathies as a genetic cause of epilepsy. Current Opinion in Neurology, 2003, 16, 171-176.	1.8	82
406	Altered kinetics and benzodiazepine sensitivity of a GABAA receptor subunit mutation [Â2(R43Q)] found in human epilepsy. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 15170-15175.	3.3	104
407	Sleep Neurology - A Wakeup Call for Neurologists. Practical Neurology, 2002, 2, 2-3.	0.5	0
408	Transcranial Magnetic Stimulation and Epilepsy. Journal of Clinical Neurophysiology, 2002, 19, 294-306.	0.9	26
409	Sodium-channel defects in benign familial neonatal-infantile seizures. Lancet, The, 2002, 360, 851-852.	6.3	332
410	Proconvulsant-induced seizures in Î±4 nicotinic acetylcholine receptor subunit knockout mice. Neuropharmacology, 2002, 43, 55-64.	2.0	20
411	Truncation of the GABAA-Receptor Î³2 Subunit in a Family with Generalized Epilepsy with Febrile Seizures Plus. American Journal of Human Genetics, 2002, 70, 530-536.	2.6	425
412	Direct and indirect measures of verbal relational memory following anterior temporal lobectomy. Neuropsychologia, 2002, 40, 302-316.	0.7	34
413	Verbal memory in left temporal lobe epilepsy: Evidence for task-related localization. Annals of Neurology, 2002, 51, 442-447.	2.8	54
414	Seizure-associated hippocampal volume loss: A longitudinal magnetic resonance study of temporal lobe epilepsy. Annals of Neurology, 2002, 51, 641-644.	2.8	172

#	Article	IF	Citations
415	Idiopathic Generalized Epilepsies: Do Sporadic and Familial Cases Differ?. Epilepsia, 2002, 42, 1399-1402.	2.6	12
416	Idiopathic Generalized Epilepsy with Generalized and Other Seizures inâ€ƒAdolescence. Epilepsia, 2002, 42, 317-320.	2.6	73
417	Chromosomal Abnormalities and Epilepsy: A Review for Clinicians and Gene Hunters. Epilepsia, 2002, 43, 127-140.	2.6	98
418	Treatment of an unprovoked tonic-clonic seizure. Journal of Clinical Neuroscience, 2001, 8, 189.	0.8	0
419	CHRNB2 Is the Second Acetylcholine Receptor Subunit Associated with Autosomal Dominant Nocturnal Frontal Lobe Epilepsy*. American Journal of Human Genetics, 2001, 68, 225-231.	2.6	300
420	Clinical and molecular genetics of myoclonicâ€“astatic epilepsy and severe myoclonic epilepsy in infancy (Dravet syndrome). Brain and Development, 2001, 23, 732-735.	0.6	57
421	Nocturnal frontal lobe epilepsy. , 2001, , 97-110.		0
422	Transcallosal Resection of Hypothalamic Hamartomas, with Control of Seizures, in Children with Gelastic Epilepsy. Neurosurgery, 2001, 48, 108-118.	0.6	150
423	Transcallosal Resection of Hypothalamic Hamartomas, with Control of Seizures, in Children with Gelastic Epilepsy. Neurosurgery, 2001, 48, 108-118.	0.6	97
424	AUStralian Study of Titration to Effect Profile of Safety (AUSâ€STEPS): Highâ€Dose Gabapentin (Neurontin) in Partial Seizures. Epilepsia, 2001, 42, 1335-1339.	2.6	19
425	Causes of epilepsies: Insights from discordant monozygous twins. Annals of Neurology, 2001, 49, 45-52.	2.8	41
426	Mutant GABAA receptor Î³2-subunit in childhood absence epilepsy and febrile seizures. Nature Genetics, 2001, 28, 49-52.	9.4	721
427	Genetics of the Epilepsies. Epilepsia, 2001, 42, 16-23.	2.6	12
428	Genetics of the Epilepsies. Epilepsia, 2001, 42, 16-23.	2.6	91
429	Title is missing!. Nature Genetics, 2001, 28, 49-52.	9.4	247
430	Genetics of the epilepsies. Current Opinion in Pediatrics, 2000, 12, 536-542.	1.0	28
431	Components of verbal learning and hippocampal damage assessed by T2 relaxometry. Journal of the International Neuropsychological Society, 2000, 6, 529-538.	1.2	29
432	Genetic and neuroradiological heterogeneity of double cortex syndrome. Annals of Neurology, 2000, 47, 265-269.	2.8	94

#	Article	IF	Citations
433	The hippocampal sclerosis whodunit: Enter the genes. Annals of Neurology, 2000, 47, 557-558.	2.8	41
434	Locus for febrile seizures. Annals of Neurology, 2000, 47, 840-841.	2.8	12
435	Does cardiac conduction pathology contribute to sudden unexpected death in epilepsy?. Epilepsy Research, 2000, 40, 17-24.	0.8	63
436	Ictal SPECT and Interictal PET in the Localization of Occipital Lobe Epilepsy. Epilepsia, 2000, 41, 463-466.	2.6	32
437	Prolactin Levels in Sudden Unexpected Death in Epilepsy. Epilepsia, 2000, 41, 48-51.	2.6	11
438	Phenotypic Characterization of an Î±₄ Neuronal Nicotinic Acetylcholine Receptor Subunit Knock-Out Mouse. Journal of Neuroscience, 2000, 20, 6431-6441.	1.7	231
439	Deaths due to brain injury among footballers in Victoria, 1968â€1999. Medical Journal of Australia, 2000, 172, 217-219.	0.8	28
440	Neurological disorders. Medical Journal of Australia, 2000, 172, 393-393.	0.8	0
441	The hippocampal sclerosis whodunit: Enter the genes. , 2000, 47, 557.		3
442	Benign Partial Seizures of Adolescence. Epilepsia, 1999, 40, 1244-1247.	2.6	15
443	Comparison of Antiepileptic Drug Levels in Sudden Unexpected Deaths in Epilepsy with Deaths from Other Causes. Epilepsia, 1999, 40, 1795-1798.	2.6	78
444	Occurrence of Hippocampal Sclerosis: Is One Hemisphere or Gender More Vulnerable?. Epilepsia, 1999, 40, 1816-1820.	2.6	26
445	Generalized epilepsy with febrile seizures plus: A common childhood-onset genetic epilepsy syndrome. Annals of Neurology, 1999, 45, 75-81.	2.8	271
446	Characterization of mutations in the genedoublecortin in patients with double cortex syndrome. Annals of Neurology, 1999, 45, 146-153.	2.8	175
447	Mapping of a Gene Determining Familial Partial Epilepsy with Variable Foci to Chromosome 22q11-q12. American Journal of Human Genetics, 1999, 65, 1698-1710.	2.6	89
448	Reorganization of verbal memory and language: A case of dissociation. Journal of the International Neuropsychological Society, 1999, 5, 69-74.	1.2	17
449	Genetics of the epilepsies. Current Opinion in Neurology, 1999, 12, 177-182.	1.8	26
450	Febrile seizures and generalized epilepsy associated with a mutation in the Na+-channel ÃŸ1 subunit gene SCN1B. Nature Genetics, 1998, 19, 366-370.	9.4	965

#	Article	IF	Citations
451	Hemicranial Volume Deficits in Patients with Temporal Lobe Epilepsy With and Without Hippocampal Sclerosis. Epilepsia, 1998, 39, 1174-1181.	2.6	72
452	Aggravation of Generalized Epilepsies. Epilepsia, 1998, 39, S11-4.	2.6	43
453	Epilepsies in twins: Genetics of the major epilepsy syndromes. Annals of Neurology, 1998, 43, 435-445.	2.8	365
454	Epileptology of the first-seizure presentation. Lancet, The, 1998, 352, 1856.	6.3	0
455	Epileptology of the first-seizure presentation: a clinical, electroencephalographic, and magnetic resonance imaging study of 300 consecutive patients. Lancet, The, 1998, 352, 1007-1011.	6.3	532
456	A Potassium Channel Mutation in Neonatal Human Epilepsy. Science, 1998, 279, 403-406.	6.0	1,013
457	Familial partial epilepsy with variable foci: A new partial epilepsy syndrome with suggestion of linkage to chromosome 2. Annals of Neurology, 1998, 44, 890-899.	2.8	111
458	doublecortin, a Brain-Specific Gene Mutated in Human X-Linked Lissencephaly and Double Cortex Syndrome, Encodes a Putative Signaling Protein. Cell, 1998, 92, 63-72.	13.5	1,007
459	Mutations in filamin 1 Prevent Migration of Cerebral Cortical Neurons in Human Periventricular Heterotopia. Neuron, 1998, 21, 1315-1325.	3.8	811
460	Concussive Convulsions. Sports Medicine, 1998, 25, 131-136.	3.1	54
461	Febrile seizures: genetics and relationship to other epilepsy syndromes. Current Opinion in Neurology, 1998, 11, 129-134.	1.8	53
462	Genetics of human partial epilepsy. Current Opinion in Neurology, 1997, 10, 110-114.	1.8	32
463	An Insertion Mutation of the CHRNA4 Gene in a Family With Autosomal Dominant Nocturnal Frontal Lobe Epilepsy. Human Molecular Genetics, 1997, 6, 943-947.	1.4	381
464	Developmental genetics of deleted mtDNA in mitochondrial oculomyopathy. Journal of the Neurological Sciences, 1997, 145, 155-162.	0.3	13
465	Epilepsy rounds. Journal of Epilepsy, 1997, 10, 49-51.	0.4	1
466	Poppy tea and the baker's first seizure. Lancet, The, 1997, 350, 716.	6.3	30
467	Epilepsies with single gene inheritance. Brain and Development, 1997, 19, 13-18.	0.6	39
468	Human Epileptogenesis and Hypothalamic Hamartomas: New Lessons from an Experiment of Nature. Epilepsia, 1997, 38, 1-3.	2.6	108

#	Article	IF	Citations
469	Adults with Epilepsy: Is Monotherapy the Only Answer?. Epilepsia, 1997, 38, S9.	2.6	0
470	Epilepsy Genes and the Genetics of Epilepsy Syndromes: The Promise of New Therapies Based on Genetic Knowledge. Epilepsia, 1997, 38, S32-6.	2.6	36
471	Epilepsy: progress in solving mysteries and dispelling myths. Medical Journal of Australia, 1996, 165, 245-246.	0.8	0
472	Temporal Lobe Epilepsy Subtypes: Differential Patterns of Cerebral Perfusion on Ictal SPECT. Epilepsia, 1996, 37, 788-795.	2.6	70
473	Familial temporal lobe epilepsy: A common disorder identified in twins. Annals of Neurology, 1996, 40, 227-235.	2.8	211
474	Does Naming Contribute to Memory Self-Report in Temporal Lobe Epilepsy?. Journal of Clinical and Experimental Neuropsychology, 1996, 18, 98-109.	0.8	31
475	Dementia and myoclonus: Differential diagnosis of early-onset alzheimer's disease. Annals of Neurology, 1995, 37, 412-412.	2.8	13
476	Comparison of ictal SPECT and interictal PET in the presurgical evaluation of temporal lobe epilepsy. Annals of Neurology, 1995, 37, 738-745.	2.8	140
477	Autosomal dominant rolandic epilepsy and speech dyspraxia: A new syndrome with anticipation. Annals of Neurology, 1995, 38, 633-642.	2.8	156
478	Clinical applications: MRI, SPECT, and PET. Magnetic Resonance Imaging, 1995, 13, 1119-1124.	1.0	99
479	Febrile seizures and hippocampal sclerosis: Frequent and related findings in intractable temporal lobe epilepsy of childhood. Pediatric Neurology, 1995, 12, 201-206.	1.0	122
480	A missense mutation in the neuronal nicotinic acetylcholine receptor Î±4 subunit is associated with autosomal dominant nocturnal frontal lobe epilepsy. Nature Genetics, 1995, 11, 201-203.	9.4	1,074
481	Autosomal dominant nocturnal frontal lobe epilepsy. Brain, 1995, 118, 61-73.	3.7	523
482	Positron emission tomography ii. Neurology. Australian Prescriber, 1995, 18, 15-18.	0.5	0
483	New autosomal-dominant partial epilepsy syndrome. Pediatric Neurology, 1994, 11, 95.	1.0	6
484	Prospective study of recent-onset temporal lobe epilepsy in childhood. Pediatric Neurology, 1994, 11, 144.	1.0	0
485	Rey figure distortions reflect nonverbal recall differences between right and left foci in unilateral temporal lobe epilepsy. Archives of Clinical Neuropsychology, 1994, 9, 451-460.	0.3	6
486	P3 latency jitter assessed using 2 techniques. I. Simulated data and surface recordings in normal subjects. Electroencephalography and Clinical Neurophysiology - Evoked Potentials, 1994, 92, 352-364.	2.0	19

#	Article	IF	Citations
487	The influence of changes in the intensity of magnetic stimulation on coil output. Muscle and Nerve, 1993, 16, 1338-1341.	1.0	12
488	Magnetic stimulation of the brain in generalized epilepsy: Reversal of cortical hyperexcitability by anticonvulsants. Annals of Neurology, 1993, 34, 351-355.	2.8	174
489	Effects of angiotensin II on dopamine and serotonin turnover in the striatum of conscious rats. Brain Research, 1993, 613, 221-229.	1.1	137
490	Lateralization of verbal memory and unilateral hippocampal sclerosis: Evidence of task-specific effects. Neuropsychology, Development and Cognition Section A: Journal of Clinical and Experimental Neuropsychology, 1993, 15, 608-618.	1.4	159
491	Ictal 99mTc-HMPAO Single Photon Emission Computed Tomography in Children with Temporal Lobe Epilepsy. Epilepsia, 1993, 34, 869-877.	2.6	85
492	Progressive Myoclonus Epilepsies: Clinical and Genetic Aspects. Epilepsia, 1993, 34, S19-30.	2.6	52
493	Validation of a Questionnaire for Clinical Seizure Diagnosis. Epilepsia, 1992, 33, 1065-1071.	2.6	110
494	HLA-DR2 negative narcolepsy in Australian caucasians: Clinical features, serology and sequence specific oligonucleotide typing. Journal of the Neurological Sciences, 1992, 113, 26-30.	0.3	0
495	Chronic encephalitis (rasmussen's syndrome) and ipsilateral uveitis. Annals of Neurology, 1992, 32, 826-829.	2.8	24
496	Functional respiratory chain studies in subjects with chronic progressive external ophthalmoplegia and large heteroplasmic mitochondrial DNA deletions. Journal of the Neurological Sciences, 1991, 102, 92-99.	0.3	17
497	Hippocampal sclerosis in temporal lobe epilepsy demonstrated by magnetic resonance imaging. Annals of Neurology, 1991, 29, 175-182.	2.8	354
498	Mitochondrial dysfunction in multiple symmetrical lipomatosis. Annals of Neurology, 1991, 29, 566-569.	2.8	123
499	The Ramsay Hunt syndrome is no longer a useful diagnostic category. Movement Disorders, 1989, 4, 13-17.	2.2	23
500	Limbic P3 potentials, seizure localization, and surgical pathology in temporal lobe epilepsy. Annals of Neurology, 1989, 26, 377-385.	2.8	94
501	Localization of epileptic foci with postictal single photon emission computed tomography. Annals of Neurology, 1989, 26, 660-668.	2.8	212
502	Kufs disease: Clinical features and forms. American Journal of Medical Genetics Part A, 1988, 31, 105-109.	2.4	26
503	The Newfoundland aggregate of neuronal ceroid-lipofuscinosis. American Journal of Medical Genetics Part A, 1988, 31, 111-116.	2.4	23
504	Focal cortical myoclonus and rolandic cortical dysplasia: Clarification by magnetic resonance imaging. Annals of Neurology, 1988, 23, 317-325.	2.8	104

#	Article	IF	Citations
505	KUFS' DISEASE: A CRITICAL REAPPRAISAL. Brain, 1988, 111, 27-62.	3.7	210
506	Magnetic resonance imaging in temporal lobe epilepsy: Pathological correlations. Annals of Neurology, 1987, 22, 341-347.	2.8	324
507	Progressive Myoclonus Epilepsies: Specific Causes and Diagnosis. New England Journal of Medicine, 1986, 315, 296-305.	13.9	301
508	Acetylation of histones in isolated avian erythroid nuclei. Nucleic Acids and Protein Synthesis, 1977, 475, 160-167.	1.7	1