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List of Publications by Year
in descending order

508 papers	53,075 citations	1301 109 h-index	1799 211 g-index
528 all docs	528 docs citations	528 times ranked	32214 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.	7.6	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.	2.3	10
3	Sporadic hypothalamic hamartoma is a ciliopathy with somatic and bi-allelic contributions. Human Molecular Genetics, 2022, 31, 2307-2316.	2.9	8
4	OUP accepted manuscript. Brain, 2022, , .	7.6	1
5	Association of ultraâ€rare coding variants with genetic generalized epilepsy: A caseâ€"control whole exome sequencing study. Epilepsia, 2022, 63, 723-735.	5.1	8
6	Evidence for a Dual-Pathway, 2-Hit Genetic Model for Focal Cortical Dysplasia and Epilepsy. Neurology: Genetics, 2022, 8, e652.	1.9	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.	2.8	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.	0.8	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.	2.0	2
10	Defective lipid signalling caused by mutations inPIK3C2Bunderlies focal epilepsy. Brain, 2022, 145, 2313-2331.	7.6	10
11	Machine learning approaches for imagingâ€based prognostication of the outcome of surgery for mesial temporal lobe epilepsy. Epilepsia, 2022, 63, 1081-1092.	5.1	10
12	Interictal EEG and ECG for SUDEP Risk Assessment: A Retrospective Multicenter Cohort Study. Frontiers in Neurology, 2022, 13, 858333.	2.4	2
13	A pharmacogenomic assessment of psychiatric adverse drug reactions to levetiracetam. Epilepsia, 2022, 63, 1563-1570.	5.1	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, .	5.1	8
15	ILAE Genetic Literacy Series: familial focal epilepsy syndromes. Epileptic Disorders, 2022, 24, 221-228.	1.3	3
16	Functional correlates of clinical phenotype and severity in recurrent SCN2A variants. Communications Biology, 2022, 5, .	4.4	13
17	Common risk variants for epilepsy are enriched in families previously targeted for rare monogenic variant discovery. EBioMedicine, 2022, 81, 104079.	6.1	10
18	Precision medicine for genetic epilepsy on the horizon: Recent advances, present challenges, and suggestions for continued progress. Epilepsia, 2022, 63, 2461-2475.	5.1	50

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19	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	2.4	28
20	Progressive Myoclonus Epilepsy Caused by a Homozygous Splicing Variant of <scp>SLC7A6OS</scp>. Annals of Neurology, 2021, 89, 402-407.	5.3	5
21	Transcriptome analysis of a ring chromosome 20 patient cohort. Epilepsia, 2021, 62, e22-e28.	5.1	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.	3.7	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.	0.6	16
24	Cerebrospinal fluid liquid biopsy for detecting somatic mosaicism in brain. Brain Communications, 2021, 3, fcaa235.	3.3	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.	2.4	11
26	The severe epilepsy syndromes of infancy: A populationâ€based study. Epilepsia, 2021, 62, 358-370.	5.1	31
27	Contribution of rare genetic variants to drug response in absence epilepsy. Epilepsy Research, 2021, 170, 106537.	1.6	9
28	Association of SLC32A1 Missense Variants With Genetic Epilepsy With Febrile Seizures Plus. Neurology, 2021, 96, e2251-e2260.	1.1	13
29	Cation leak underlies neuronal excitability in an HCN1 developmental and epileptic encephalopathy. Brain, 2021, 144, 2060-2073.	7.6	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.	6.2	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.	3.7	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.	3.7	9
33	Integrated in silico and experimental assessment of disease relevance of PCDH19 Âmissense variants. Human Mutation, 2021, 42, 1030-1041.	2.5	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.	6.2	35
35	Pathogenic <scp>MAST3</scp> Variants in the <scp>STK</scp> Domain Are Associated with Epilepsy. Annals of Neurology, 2021, 90, 274-284.	5.3	7
36	Association Between Psychiatric Comorbidities and Mortality in Epilepsy. Neurology: Clinical Practice, 2021, 11, 429-437.	1.6	7

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37	Improving Specificity of <scp>Cerebrospinal Fluid</scp> Liquid Biopsy for Genetic Testing. Annals of Neurology, 2021, 90, 693-694.	5.3	2
38	Postictal Psychosis in Epilepsy: A Clinicogenetic Study. Annals of Neurology, 2021, 90, 464-476.	5.3	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.	2.8	5
40	Variants in ATP6V0A1 cause progressive myoclonus epilepsy and developmental and epileptic encephalopathy. Brain Communications, 2021, 3, fcab245.	3.3	10
41	Association of Short-term Heart Rate Variability and Sudden Unexpected Death in Epilepsy. Neurology, 2021, 97, .	1.1	25
42	State transitions through inhibitory interneurons in a cortical network model. PLoS Computational Biology, 2021, 17, e1009521.	3.2	10
43	Hypothalamic Hamartomas. Neurology, 2021, 97, 864-873.	1.1	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.	1.2	6
45	Progressive Myoclonus Epilepsies. Neurology: Genetics, 2021, 7, e641.	1.9	20
46	What is the motor vehicle crash risk for drivers with epilepsy? A systematic review. Journal of Transport and Health, 2021, 23, 101286.	2.2	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, .	0.8	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.	1.3	14
49	Epilepsy genetics: clinical impacts and biological insights. Lancet Neurology, The, 2020, 19, 93-100.	10.2	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.	5.3	2
51	SCN1A Variants in vaccineâ€related febrile seizures: A prospective study. Annals of Neurology, 2020, 87, 281-288.	5.3	15
52	Novel Missense CACNA1G Mutations Associated with Infantile-Onset Developmental and Epileptic Encephalopathy. International Journal of Molecular Sciences, 2020, 21, 6333.	4.1	7
53	Mortality in patients with psychogenic nonepileptic seizures. Neurology, 2020, 95, e643-e652.	1.1	75
54	Anterior temporal encephaloceles: Elusive, important, and rewarding to treat. Epilepsia, 2020, 61, 2675-2684.	5.1	16

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55	Generalized, focal, and combined epilepsies in families: New evidence for distinct genetic factors. Epilepsia, 2020, 61, 2667-2674.	5.1	4
56	Are Variants Causing Cardiac Arrhythmia Risk Factors in Sudden Unexpected Death in Epilepsy?. Frontiers in Neurology, 2020, 11, 925.	2.4	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.	0.8	1
58	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	7.6	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.	2.8	23
60	Antiepileptic Drug Teratogenicity and De Novo Genetic Variation Load. Annals of Neurology, 2020, 87, 897-906.	5.3	9
61	Inherited RORB pathogenic variants: Overlap of photosensitive genetic generalized and occipital lobe epilepsy. Epilepsia, 2020, 61, e23-e29.	5.1	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.	7.1	33
63	The Genetics of Epilepsy. Annual Review of Genomics and Human Genetics, 2020, 21, 205-230.	6.2	116
64	Encephalopathies with KCNC1 variants: genotypeâ€phenotypeâ€functional correlations. Annals of Clinical and Translational Neurology, 2019, 6, 1263-1272.	3.7	33
65	Evidence for type-specific DNA methylation patterns in epilepsy: a discordant monozygotic twin approach. Epigenomics, 2019, 11, 951-968.	2.1	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.	6.2	237
67	Deciphering the role of epigenetics in self-limited epilepsy with centrotemporal spikes. Epilepsy Research, 2019, 156, 106163.	1.6	5
68	Predominantly nocturnal seizures post temporal lobectomy: Characteristics of an unusual outcome group. Epilepsy Research, 2019, 155, 106154.	1.6	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.	3.7	55
70	Somatic mutation: The hidden genetics of brain malformations and focal epilepsies. Epilepsy Research, 2019, 155, 106161.	1.6	45
71	Quantitative analysis of phenotypic elements augments traditional electroclinical classification of common familial epilepsies. Epilepsia, 2019, 60, 2194-2203.	5.1	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.	3.9	23

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73	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	12.8	99
74	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. Annals of Neurology, 2019, 86, 821-831.	5.3	96
75	Epidemiology and etiology of infantile developmental and epileptic encephalopathies in Tasmania. Epilepsia Open, 2019, 4, 504-510.	2.4	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.	1.2	9
77	Epilepsy in families: Age at onset is a familial trait, independent of syndrome. Annals of Neurology, 2019, 86, 91-98.	5.3	11
78	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. Epilepsia, 2019, 60, 797-806.	5.1	52
79	No evidence for a BRD 2 promoter hypermethylation inÂblood leukocytes of Europeans with juvenile myoclonicÂepilepsy. Epilepsia, 2019, 60, e31-e36.	5.1	4
80	Human GABRG2 generalized epilepsy. Neurology: Genetics, 2019, 5, e340.	1.9	6
81	Epileptic encephalopathies of infancy: welcome advances. Lancet, The, 2019, 394, 2203-2204.	13.7	1
82	Kufs disease due to mutation ofCLN6: clinical, pathological and molecular genetic features. Brain, 2019, 142, 59-69.	7.6	28
83	SYNGAP1 encephalopathy. Neurology, 2019, 92, e96-e107.	1.1	131
84	Metabolic patterns and seizure outcomes following anterior temporal lobectomy. Annals of Neurology, 2019, 85, 241-250.	5.3	25
85	Clinical challenges and future therapeutic approaches for neuronal ceroid lipofuscinosis. Lancet Neurology, The, 2019, 18, 107-116.	10.2	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.	3.2	10
87	Parental Mosaicism in â€œDe Novoâ€•Epileptic Encephalopathies. New England Journal of Medicine, 2018, 378, 1646-1648.	27.0	104
88	Can mutationâ€mediated effects occurring early in development cause longâ€term seizure susceptibility in genetic generalized epilepsies?. Epilepsia, 2018, 59, 915-922.	5.1	7
89	Consistency of Long-Term Subdural Electrocorticography in Humans. IEEE Transactions on Biomedical Engineering, 2018, 65, 344-352.	4.2	39
90	Teenage-onset progressive myoclonic epilepsy due to a familial C9orf72 repeat expansion. Neurology, 2018, 90, e658-e663.	1.1	9

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91	Ion Channels in Genetic Epilepsy: From Genes and Mechanisms to Disease-Targeted Therapies. Pharmacological Reviews, 2018, 70, 142-173.	16.0	215
92	Gain-of-functionHCN2variants in genetic epilepsy. Human Mutation, 2018, 39, 202-209.	2.5	28
93	Precision therapy for epilepsy due to KCNT1 mutations. Neurology, 2018, 90, e67-e72.	1.1	108
94	Development of a rapid functional assay that predicts GLUT1 disease severity. Neurology: Genetics, 2018, 4, e297.	1.9	7
95	A Primate-Specific Isoform of PLEKHG6 Regulates Neurogenesis and Neuronal Migration. Cell Reports, 2018, 25, 2729-2741.e6.	6.4	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.	6.2	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.	7.1	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.	5.1	3
99	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	12.6	1,085
100	Genetic generalized epilepsies. Epilepsia, 2018, 59, 1148-1153.	5.1	72
101	Somatic GNAQ mutation in the forme fruste of Sturge-Weber syndrome. Neurology: Genetics, 2018, 4, e236.	1.9	29
102	Genetic literacy series: genetic epilepsy with febrile seizures plus. Epileptic Disorders, 2018, 20, 232-238.	1.3	44
103	KANSL1 variation is not a major contributing factor in self-limited focal epilepsy syndromes of childhood. PLoS ONE, 2018, 13, e0191546.	2.5	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.	5.1	31
105	Frequency of <scp>CNKSR</scp>2 mutation in the Xâ€linked epilepsyâ€aphasia spectrum. Epilepsia, 2017, 58, e40-e43.	5.1	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.	5.1	3,464
107	SCN1A clinical spectrum includes the self-limited focal epilepsies of childhood. Epilepsy Research, 2017, 131, 9-14.	1.6	12
108	Real-world utility of whole exome sequencing with targeted gene analysis for focal epilepsy. Epilepsy Research, 2017, 131, 1-8.	1.6	93

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109	Evaluation of GLUT1 variation in non-acquired focal epilepsy. Epilepsy Research, 2017, 133, 54-57.	1.6	5
110	Familial epilepsy with anterior polymicrogyria as a presentation of COL18A1 mutations. European Journal of Medical Genetics, 2017, 60, 437-443.	1.3	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.	5.3	69
112	Exome sequencingâ€"based molecular autopsy of formalin-fixed paraffin-embedded tissue after sudden death. Genetics in Medicine, 2017, 19, 1127-1133.	2.4	26
113	Epilepsy research in 2016: new treatment directions. Lancet Neurology, The, 2017, 16, 7-9.	10.2	3
114	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
115	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. American Journal of Human Genetics, 2017, 101, 516-524.	6.2	43
116	Genetic epilepsy with febrile seizures plus. Neurology, 2017, 89, 1210-1219.	1.1	112
117	Optimizing genomic medicine in epilepsy through a gene-customized approach to missense variant interpretation. Genome Research, 2017, 27, 1715-1729.	5.5	150
118	Familial mesial temporal lobe epilepsy and the borderland of dÃ©jÃ vu. Annals of Neurology, 2017, 82, 166-176.	5.3	19
119	ExACtly zero or once. Neurology: Genetics, 2017, 3, e163.	1.9	37
120	Synaptic Zn²⁺ and febrile seizure susceptibility. British Journal of Pharmacology, 2017, 174, 119-125.	5.4	18
121	Sensitive quantitative detection of somatic mosaic mutation in â€œdouble cortexâ€•syndrome. Epileptic Disorders, 2017, 19, 450-455.	1.3	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.	3.5	25
123	Periventricular Nodular Heterotopia: Detection of Abnormal Microanatomic Fiber Structures with Whole-Brain Diffusion MR Imaging Tractography. Radiology, 2016, 281, 896-906.	7.3	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.	5.3	216
125	Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2â€"2q11.2. Human Genetics, 2016, 135, 1117-1125.	3.8	29
126	In silico prioritization based on coexpression can aid epileptic encephalopathy gene discovery. Neurology: Genetics, 2016, 2, e51.	1.9	19

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127	Evaluation of nonâ€coding variation in <scp>GLUT</scp>1 deficiency. Developmental Medicine and Child Neurology, 2016, 58, 1295-1302.	2.1	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.	5.1	16
129	Is FGF13 a major contributor to genetic epilepsy with febrile seizures plus?. Epilepsy Research, 2016, 128, 48-51.	1.6	7
130	A targeted resequencing gene panel for focal epilepsy. Neurology, 2016, 86, 1605-1612.	1.1	48
131	Definition and diagnostic criteria of sleep-related hypermotor epilepsy. Neurology, 2016, 86, 1834-1842.	1.1	245
132	Dominant KCNA2 mutation causes episodic ataxia and pharmacoresponsive epilepsy. Neurology, 2016, 87, 1975-1984.	1.1	71
133	Mutations of the Sonic Hedgehog Pathway Underlie Hypothalamic Hamartoma with Gelastic Epilepsy. American Journal of Human Genetics, 2016, 99, 423-429.	6.2	59
134	Progressive myoclonus epilepsy associated with SACS gene mutations. Neurology: Genetics, 2016, 2, e83.	1.9	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.	5.1	36
136	Diagnosis and misdiagnosis of adult neuronal ceroid lipofuscinosis (Kufs disease). Neurology, 2016, 87, 579-584.	1.1	28
137	TBC1D24 genotypeâ€“phenotype correlation. Neurology, 2016, 87, 77-85.	1.1	97
138	Multiplex families with epilepsy. Neurology, 2016, 86, 713-722.	1.1	23
139	Mutations in the mammalian target of rapamycin pathway regulators NPRL2 and NPRL3 cause focal epilepsy. Annals of Neurology, 2016, 79, 120-131.	5.3	190
140	Interictal spikes and epileptic seizures: their relationship and underlying rhythmicity. Brain, 2016, 139, 1066-1078.	7.6	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.	2.0	0
142	PRIMA1 mutation: a new cause of nocturnal frontal lobe epilepsy. Annals of Clinical and Translational Neurology, 2015, 2, 821-830.	3.7	21
143	Loss of synaptic Zn2+ transporter function increases risk of febrile seizures. Scientific Reports, 2015, 5, 17816.	3.3	33
144	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. EBioMedicine, 2015, 2, 1063-1070.	6.1	74

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145	Mind the gap: Multiple events and lengthy delays before presentation with a â€œfirst seizureâ€. Epilepsia, 2015, 56, 1534-1541.	5.1	65
146	Familial neonatal seizures in 36 families: Clinical and genetic features correlate with outcome. Epilepsia, 2015, 56, 1071-1080.	5.1	94
147	Epileptic spasms are a feature of DEPDC5 mTORopathy. Neurology: Genetics, 2015, 1, e17.	1.9	63
148	Quinidine in the treatment of <scp>KCNT</scp>1â€positive epilepsies. Annals of Neurology, 2015, 78, 995-999.	5.3	184
149	Genetics of Epilepsy in Clinical Practice. Epilepsy Currents, 2015, 15, 192-196.	0.8	39
150	Mutation of the nuclear lamin geneLMNB2in progressive myoclonus epilepsy with early ataxia. Human Molecular Genetics, 2015, 24, 4483-4490.	2.9	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.	3.7	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.	2.1	13
153	CHD2variants are a risk factor for photosensitivity in epilepsy. Brain, 2015, 138, 1198-1208.	7.6	112
154	Myoclonic occipital photosensitive epilepsy with dystonia (MOPED): A familial epilepsy syndrome. Epilepsy Research, 2015, 114, 98-105.	1.6	7
155	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. Human Molecular Genetics, 2015, 24, 5250-5259.	2.9	93
156	CHD2 myoclonic encephalopathy is frequently associated with self-induced seizures. Neurology, 2015, 84, 951-958.	1.1	79
157	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. American Journal of Human Genetics, 2015, 96, 808-815.	6.2	173
158	Cortical microarchitecture changes in genetic epilepsy. Neurology, 2015, 84, 1308-1316.	1.1	16
159	Evaluation of multiple putative risk alleles within the 15q13.3 region for genetic generalized epilepsy. Epilepsy Research, 2015, 117, 70-73.	1.6	6
160	Electroclinical spectrum of the neuronal ceroid lipofuscinoses associated with CLN6 mutations. Neurology, 2015, 85, 316-324.	1.1	40
161	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. Nature Genetics, 2015, 47, 39-46.	21.4	245
162	Harnessing Gene Expression Networks to Prioritize Candidate Epileptic Encephalopathy Genes. PLoS ONE, 2014, 9, e102079.	2.5	25

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163	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. Human Molecular Genetics, 2014, 23, 6069-6080.	2.9	61
164	Genetic analysis of PHOX2B in sudden unexpected death in epilepsy cases. Neurology, 2014, 83, 1018-1021.	1.1	19
165	The genetic basis of music ability. Frontiers in Psychology, 2014, 5, 658.	2.1	60
166	KCNT1 gain of function in 2 epilepsy phenotypes is reversed by quinidine. Annals of Neurology, 2014, 75, 581-590.	5.3	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.	2.1	16
168	Is focal cortical dysplasia sporadic? Family evidence for genetic susceptibility. Epilepsia, 2014, 55, e22-6.	5.1	23
169	Glucose metabolism transporters and epilepsy: Only <scp>GLUT</scp>1 has an established role. Epilepsia, 2014, 55, e18-21.	5.1	29
170	Mutations in mammalian target of rapamycin regulator DEPDC5 cause focal epilepsy with brain malformations. Annals of Neurology, 2014, 75, 782-787.	5.3	193
171	Genetics of epilepsy. Neurology, 2014, 83, 1042-1048.	1.1	61
172	Genetics of vasovagal syncope. Autonomic Neuroscience: Basic and Clinical, 2014, 184, 60-65.	2.8	19
173	Does variation in NIPA2 contribute to genetic generalized epilepsy?. Human Genetics, 2014, 133, 673-674.	3.8	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.	2.9	33
175	Reduced dendritic arborization and hyperexcitability of pyramidal neurons in a Scn1b-based model of Dravet syndrome. Brain, 2014, 137, 1701-1715.	7.6	49
176	The hidden genetics of epilepsyâ€"a clinically important new paradigm. Nature Reviews Neurology, 2014, 10, 283-292.	10.1	232
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