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List of Publications by Year in Descending Order

479 papers	41,365 citations	103 h-index	191 g-index
528 ext. papers	47,515 ext. citations	8.6 avg, IF	6.89 L-index

#	Paper	IF	Citations
479	Evidence for a Dual-Pathway, 2-Hit Genetic Model for Focal Cortical Dysplasia and Epilepsy.. *Neurology: Genetics*, **2022**, 8, e652	3.8	1
478	Bi-allelic SMO variants in hypothalamic hamartoma: a recessive cause of Pallister-Hall syndrome.. *European Journal of Human Genetics*, **2022**,	5.3	1
477	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**,	1.2	2
476	Diagnostic delay in focal epilepsy: Association with brain pathology and age.. *Seizure: the Journal of the British Epilepsy Association*, **2022**, 96, 121-127	3.2	0
475	UNC13B and focal epilepsy.. *Brain*, **2022**,	11.2	1
474	Genome-wide association study of febrile seizures implicates fever response and neuronal excitability genes.. *Brain*, **2022**,	11.2	4
473	Interictal EEG and ECG for SUDEP Risk Assessment: A Retrospective Multicenter Cohort Study.. *Frontiers in Neurology*, **2022**, 13, 858333	4.1	0
472	Common risk variants for epilepsy are enriched in families previously targeted for rare monogenic variant discovery. *EBioMedicine*, **2022**, 81, 104079	8.8	1
471	Plasma neurofilament light chain protein is not increased in treatment-resistant schizophrenia and first-degree relatives.. *Australian and New Zealand Journal of Psychiatry*, **2021**, 48674211058684	2.6	1
470	Progressive Myoclonus Epilepsies: Diagnostic Yield With Next-Generation Sequencing in Previously Unsolved Cases. *Neurology: Genetics*, **2021**, 7, e641	3.8	2
469	What is the motor vehicle crash risk for drivers with epilepsy? A systematic review. *Journal of Transport and Health*, **2021**, 23, 101286	3	0
468	Variants in cause progressive myoclonus epilepsy and developmental and epileptic encephalopathy.. *Brain Communications*, **2021**, 3, fcab245	4.5	0
467	Association of Short-term Heart Rate Variability and Sudden Unexpected Death in Epilepsy. *Neurology*, **2021**,	6.5	4
466	State transitions through inhibitory interneurons in a cortical network model. *PLoS Computational Biology*, **2021**, 17, e1009521	5	0
465	Hypothalamic Hamartomas: Evolving Understanding and Management. *Neurology*, **2021**, 97, 864-873	6.5	0
464	Cation leak underlies neuronal excitability in an HCN1 developmental and epileptic encephalopathy. *Brain*, **2021**, 144, 2060-2073	11.2	7
463	Progressive myoclonus epilepsies-Residual unsolved cases have marked genetic heterogeneity including dolichol-dependent protein glycosylation pathway genes. *American Journal of Human Genetics*, **2021**, 108, 722-738	11	10

462	Assessing the role of rare genetic variants in drug-resistant, non-lesional focal epilepsy. *Annals of Clinical and Translational Neurology*, **2021**, 8, 1376-1387	5.3	3
461	Loss-of-function variants in K 11.1 cardiac channels as a biomarker for SUDEP. *Annals of Clinical and Translational Neurology*, **2021**, 8, 1422-1432	5.3	0
460	Integrated in silico and experimental assessment of disease relevance of PCDH19´missense variants. *Human Mutation*, **2021**, 42, 1030-1041	4.7	
459	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	11	6
458	Pathogenic MAST3 Variants in the STK Domain Are Associated with Epilepsy. *Annals of Neurology*, **2021**, 90, 274-284	9.4	1
457	Association Between Psychiatric Comorbidities and Mortality in Epilepsy. *Neurology: Clinical Practice*, **2021**, 11, 429-437	1.7	1
456	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. *Genetics in Medicine*, **2021**, 23, 363-373	8.1	4
455	Progressive Myoclonus Epilepsy Caused by a Homozygous Splicing Variant of SLC7A6OS. *Annals of Neurology*, **2021**, 89, 402-407	9.4	1
454	Transcriptome analysis of a ring chromosome 20 patient cohort. *Epilepsia*, **2021**, 62, e22-e28	6.4	1
453	Epilepsy risk in offspring of affected parents; a cohort study of the "maternal effect" in epilepsy. *Annals of Clinical and Translational Neurology*, **2021**, 8, 153-162	5.3	0
452	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	3.2	4
451	Cerebrospinal fluid liquid biopsy for detecting somatic mosaicism in brain. *Brain Communications*, **2021**, 3, fcaa235	4.5	17
450	Newly diagnosed seizures assessed at two established first seizure clinics: Clinic characteristics, investigations, and findings over 11 years. *Epilepsia Open*, **2021**, 6, 171-180	4	4
449	The severe epilepsy syndromes of infancy: A population-based study. *Epilepsia*, **2021**, 62, 358-370	6.4	7
448	Contribution of rare genetic variants to drug response in absence epilepsy. *Epilepsy Research*, **2021**, 170, 106537	3	5
447	Association of Missense Variants With Genetic Epilepsy With Febrile Seizures Plus. *Neurology*, **2021**, 96, e2251-e2260	6.5	3
446	Improving Specificity of Cerebrospinal Fluid Liquid Biopsy for Genetic Testing. *Annals of Neurology*, **2021**, 90, 693-694	9.4	1
445	Postictal Psychosis in Epilepsy: A Clinicogenetic Study. *Annals of Neurology*, **2021**, 90, 464-476	9.4	1

444	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	4.3	0
443	Neurofilament light chain in psychiatric and neurodegenerative disorders: A □-reactive protein□for the brain?. *Alzheimers and Dementia*, **2020**, 16, e041347	1.2	
442	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17 458 subjects. *Brain*, **2020**, 143, 2106-2118	11.2	14
441	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	5.3	9
440	Antiepileptic Drug Teratogenicity and De Novo Genetic Variation Load. *Annals of Neurology*, **2020**, 87, 897-906	9.4	5
439	Inherited RORB pathogenic variants: Overlap of photosensitive genetic generalized and occipital lobe epilepsy. *Epilepsia*, **2020**, 61, e23-e29	6.4	5
438	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	11.5	15
437	The Genetics of Epilepsy. *Annual Review of Genomics and Human Genetics*, **2020**, 21, 205-230	9.7	30
436	The "maternal effect" on epilepsy risk: Analysis of familial epilepsies and reassessment of prior evidence. *Annals of Neurology*, **2020**, 87, 132-138	9.4	2
435	SCN1A Variants in vaccine-related febrile seizures: A prospective study. *Annals of Neurology*, **2020**, 87, 281-288	9.4	7
434	Novel Missense Mutations Associated with Infantile-Onset Developmental and Epileptic Encephalopathy. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	0
433	Mortality in patients with psychogenic nonepileptic seizures. *Neurology*, **2020**, 95, e643-e652	6.5	36
432	Anterior temporal encephaloceles: Elusive, important, and rewarding to treat. *Epilepsia*, **2020**, 61, 2675-2684	6.4	5
431	Generalized, focal, and combined epilepsies in families: New evidence for distinct genetic factors. *Epilepsia*, **2020**, 61, 2667-2674	6.4	1
430	Are Variants Causing Cardiac Arrhythmia Risk Factors in Sudden Unexpected Death in Epilepsy?. *Frontiers in Neurology*, **2020**, 11, 925	4.1	3
429	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	2.6	8
428	Epilepsy genetics: clinical impacts and biological insights. *Lancet Neurology, The*, **2020**, 19, 93-100	24.1	40
427	Epidemiology and etiology of infantile developmental and epileptic encephalopathies in Tasmania. *Epilepsia Open*, **2019**, 4, 504-510	4	4

426	Splice variant in ARX leading to loss of C-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	2.5	5
425	Epilepsy in families: Age at onset is a familial trait, independent of syndrome. *Annals of Neurology*, **2019**, 86, 91-98	9.4	5
424	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. *Epilepsia*, **2019**, 60, 797-806	6.4	24
423	No evidence for a BRD2 promoter hypermethylation in blood leukocytes of Europeans with juvenile myoclonic epilepsy. *Epilepsia*, **2019**, 60, e31-e36	6.4	4
422	Encephalopathies with KCNC1 variants: genotype-phenotype-functional correlations. *Annals of Clinical and Translational Neurology*, **2019**, 6, 1263-1272	5.3	12
421	Evidence for type-specific DNA methylation patterns in epilepsy: a discordant monozygotic twin approach. *Epigenomics*, **2019**, 11, 951-968	4.4	11
420	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	11	104
419	Deciphering the role of epigenetics in self-limited epilepsy with centrotemporal spikes. *Epilepsy Research*, **2019**, 156, 106163	3	2
418	Predominantly nocturnal seizures post temporal lobectomy: Characteristics of an unusual outcome group. *Epilepsy Research*, **2019**, 155, 106154	3	
417	Second-hit DEPDC5 mutation is limited to dysmorphic neurons in cortical dysplasia type IIA. *Annals of Clinical and Translational Neurology*, **2019**, 6, 1338-1344	5.3	26
416	Somatic mutation: The hidden genetics of brain malformations and focal epilepsies. *Epilepsy Research*, **2019**, 155, 106161	3	24
415	Quantitative analysis of phenotypic elements augments traditional electroclinical classification of common familial epilepsies. *Epilepsia*, **2019**, 60, 2194-2203	6.4	
414	Nomenclature of Genetically Determined Myoclonus Syndromes: Recommendations of the International Parkinson and Movement Disorder Society Task Force. *Movement Disorders*, **2019**, 34, 1602-1613	7	12
413	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. *Nature Communications*, **2019**, 10, 4920	17.4	48
412	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. *Annals of Neurology*, **2019**, 86, 821-831	9.4	55
411	Human generalized epilepsy: Increased somatosensory and striatothalamic connectivity. *Neurology: Genetics*, **2019**, 5, e340	3.8	3
410	Epileptic encephalopathies of infancy: welcome advances. *Lancet, The*, **2019**, 394, 2203-2204	40	1
409	Kufs disease due to mutation of CLN6: clinical, pathological and molecular genetic features. *Brain*, **2019**, 142, 59-69	11.2	14

408	encephalopathy: A distinctive generalized developmental and epileptic encephalopathy. *Neurology*, **2019**, 92, e96-e107	6.5	55
407	Metabolic patterns and seizure outcomes following anterior temporal lobectomy. *Annals of Neurology*, **2019**, 85, 241-250	9.4	16
406	Clinical challenges and future therapeutic approaches for neuronal ceroid lipofuscinosis. *Lancet Neurology, The*, **2019**, 18, 107-116	24.1	86
405	A case series of lacosamide as adjunctive therapy in refractory sleep-related hypermotor epilepsy (previously nocturnal frontal lobe epilepsy). *Journal of Sleep Research*, **2018**, 27, e12669	5.8	8
404	Parental Mosaicism in "De Novo" Epileptic Encephalopathies. *New England Journal of Medicine*, **2018**, 378, 1646-1648	59.2	67
403	Can mutation-mediated effects occurring early in development cause long-term seizure susceptibility in genetic generalized epilepsies?. *Epilepsia*, **2018**, 59, 915-922	6.4	6
402	Consistency of Long-Term Subdural Electrocorticography in Humans. *IEEE Transactions on Biomedical Engineering*, **2018**, 65, 344-352	5	27
401	Teenage-onset progressive myoclonic epilepsy due to a familial repeat expansion. *Neurology*, **2018**, 90, e658-e663	6.5	7
400	Ion Channels in Genetic Epilepsy: From Genes and Mechanisms to Disease-Targeted Therapies. *Pharmacological Reviews*, **2018**, 70, 142-173	22.5	103
399	Gain-of-function HCN2 variants in genetic epilepsy. *Human Mutation*, **2018**, 39, 202-209	4.7	19
398	Analysis of shared heritability in common disorders of the brain. *Science*, **2018**, 360,	33.3	666
397	Genetic generalized epilepsies. *Epilepsia*, **2018**, 59, 1148-1153	6.4	39
396	Somatic mutation in the of Sturge-Weber syndrome. *Neurology: Genetics*, **2018**, 4, e236	3.8	18
395	Genetic literacy series: genetic epilepsy with febrile seizures plus. *Epileptic Disorders*, **2018**, 20, 232-238	1.9	30
394	KANSL1 variation is not a major contributing factor in self-limited focal epilepsy syndromes of childhood. *PLoS ONE*, **2018**, 13, e0191546	3.7	3
393	Precision therapy for epilepsy due to mutations: A randomized trial of oral quinidine. *Neurology*, **2018**, 90, e67-e72	6.5	84
392	ILAE-Klassifikation der Epilepsien: Positionspapier der ILAE-Kommission f□ Klassifikation und Terminologie. *Zeitschrift Fur Epileptologie*, **2018**, 31, 296-306	0.1	17
391	Development of a rapid functional assay that predicts GLUT1 disease severity. *Neurology: Genetics*, **2018**, 4, e297	3.8	5

390	A Primate-Specific Isoform of PLEKHG6 Regulates Neurogenesis and Neuronal Migration. *Cell Reports*, **2018**, 25, 2729-2741.e6	10.6	27
389	Aberrant Inclusion of a Poison Exon Causes Dravet Syndrome and Related SCN1A-Associated Genetic Epilepsies. *American Journal of Human Genetics*, **2018**, 103, 1022-1029	11	40
388	Dynamic action potential clamp predicts functional separation in mild familial and severe de novo forms of epilepsy. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, E5516-E5525	11.5	40
387	Evidence of linkage to chromosome 5p13.2-q11.1 in a large inbred family with genetic generalized epilepsy. *Epilepsia*, **2018**, 59, e125-e129	6.4	2
386	De novo SCN1A pathogenic variants in the GEFS+ spectrum: Not always a familial syndrome. *Epilepsia*, **2017**, 58, e26-e30	6.4	21
385	Frequency of CNKSR2 mutation in the X-linked epilepsy-aphasia spectrum. *Epilepsia*, **2017**, 58, e40-e43	6.4	15
384	ILAE classification of the epilepsies: Position paper of the ILAE Commission for Classification and Terminology. *Epilepsia*, **2017**, 58, 512-521	6.4	2084
383	SCN1A clinical spectrum includes the self-limited focal epilepsies of childhood. *Epilepsy Research*, **2017**, 131, 9-14	3	4
382	Real-world utility of whole exome sequencing with targeted gene analysis for focal epilepsy. *Epilepsy Research*, **2017**, 131, 1-8	3	63
381	Evaluation of GLUT1 variation in non-acquired focal epilepsy. *Epilepsy Research*, **2017**, 133, 54-57	3	4
380	Familial epilepsy with anterior polymicrogyria as a presentation of COL18A1 mutations. *European Journal of Medical Genetics*, **2017**, 60, 437-443	2.6	9
379	Myoclonus epilepsy and ataxia due to KCNC1 mutation: Analysis of 20 cases and K channel properties. *Annals of Neurology*, **2017**, 81, 677-689	9.4	39
378	Exome sequencing-based molecular autopsy of formalin-fixed paraffin-embedded tissue after sudden death. *Genetics in Medicine*, **2017**, 19, 1127-1133	8.1	18
377	Epilepsy research in 2016: new treatment directions. *Lancet Neurology, The*, **2017**, 16, 7-9	24.1	3
376	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. *American Journal of Human Genetics*, **2017**, 101, 664-685	11	214
375	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. *American Journal of Human Genetics*, **2017**, 101, 516-524	11	29
374	A case-control collapsing analysis identifies epilepsy genes implicated in trio sequencing studies focused on de novo mutations. *PLoS Genetics*, **2017**, 13, e1007104	6	18
373	Genetic epilepsy with febrile seizures plus: Refining the spectrum. *Neurology*, **2017**, 89, 1210-1219	6.5	68

372	Optimizing genomic medicine in epilepsy through a gene-customized approach to missense variant interpretation. *Genome Research*, **2017**, 27, 1715-1729	9.7	91
371	Familial mesial temporal lobe epilepsy and the borderland of dJ□vu. *Annals of Neurology*, **2017**, 82, 166-176	9.4	12
370	ExACtly zero or once: A clinically helpful guide to assessing genetic variants in mild epilepsies. *Neurology: Genetics*, **2017**, 3, e163	3.8	24
369	Synaptic Zn and febrile seizure susceptibility. *British Journal of Pharmacology*, **2017**, 174, 119-125	8.6	15
368	Sensitive quantitative detection of somatic mosaic mutation in "double cortex" syndrome. *Epileptic Disorders*, **2017**, 19, 450-455	1.9	6
367	Hippocampal malrotation is an anatomic variant and has no clinical significance in MRI-negative temporal lobe epilepsy. *Epilepsia*, **2016**, 57, 1719-1728	6.4	23
366	Diagnosis and misdiagnosis of adult neuronal ceroid lipofuscinosis (Kufs disease). *Neurology*, **2016**, 87, 579-84	6.5	19
365	TBC1D24 genotype-phenotype correlation: Epilepsies and other neurologic features. *Neurology*, **2016**, 87, 77-85	6.5	75
364	Multiplex families with epilepsy: Success of clinical and molecular genetic characterization. *Neurology*, **2016**, 86, 713-22	6.5	22
363	Mutations in the mammalian target of rapamycin pathway regulators NPRL2 and NPRL3 cause focal epilepsy. *Annals of Neurology*, **2016**, 79, 120-31	9.4	136
362	Interictal spikes and epileptic seizures: their relationship and underlying rhythmicity. *Brain*, **2016**, 139, 1066-78	11.2	148
361	Periventricular Nodular Heterotopia: Detection of Abnormal Microanatomic Fiber Structures with Whole-Brain Diffusion MR Imaging Tractography. *Radiology*, **2016**, 281, 896-906	20.5	13
360	Exome-based analysis of cardiac arrhythmia, respiratory control, and epilepsy genes in sudden unexpected death in epilepsy. *Annals of Neurology*, **2016**, 79, 522-34	9.4	162
359	Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2-2q11.2. *Human Genetics*, **2016**, 135, 1117-25	6.3	16
358	In silico prioritization based on coexpression can aid epileptic encephalopathy gene discovery. *Neurology: Genetics*, **2016**, 2, e51	3.8	15
357	Evaluation of non-coding variation in GLUT1 deficiency. *Developmental Medicine and Child Neurology*, **2016**, 58, 1295-1302	3.3	19
356	Seizures as presenting and prominent symptom in chorea-acanthocytosis with c.2343del VPS13A gene mutation. *Epilepsia*, **2016**, 57, 549-56	6.4	13
355	Is FGF13 a major contributor to genetic epilepsy with febrile seizures plus?. *Epilepsy Research*, **2016**, 128, 48-51	3	6

354	A targeted resequencing gene panel for focal epilepsy. *Neurology*, **2016**, 86, 1605-12	6.5	37
353	Definition and diagnostic criteria of sleep-related hypermotor epilepsy. *Neurology*, **2016**, 86, 1834-42	6.5	182
352	Dominant KCNA2 mutation causes episodic ataxia and pharmacoresponsive epilepsy. *Neurology*, **2016**, 87, 1975-1984	6.5	50
351	Mutations of the Sonic Hedgehog Pathway Underlie Hypothalamic Hamartoma with Gelastic Epilepsy. *American Journal of Human Genetics*, **2016**, 99, 423-9	11	37
350	Progressive myoclonus epilepsy associated with SACS gene mutations. *Neurology: Genetics*, **2016**, 2, e83	3.8	7
349	CHD2 variants are a risk factor for photosensitivity in epilepsy. *Brain*, **2015**, 138, 1198-207	11.2	81
348	Myoclonic occipital photosensitive epilepsy with dystonia (MOPED): A familial epilepsy syndrome. *Epilepsy Research*, **2015**, 114, 98-105	3	7
347	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. *Human Molecular Genetics*, **2015**, 24, 5250-9	5.6	78
346	CHD2 myoclonic encephalopathy is frequently associated with self-induced seizures. *Neurology*, **2015**, 84, 951-8	6.5	57
345	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. *American Journal of Human Genetics*, **2015**, 96, 808-15	11	114
344	Cortical microarchitecture changes in genetic epilepsy. *Neurology*, **2015**, 84, 1308-16	6.5	14
343	Evaluation of multiple putative risk alleles within the 15q13.3 region for genetic generalized epilepsy. *Epilepsy Research*, **2015**, 117, 70-3	3	3
342	Electroclinical spectrum of the neuronal ceroid lipofuscinoses associated with CLN6 mutations. *Neurology*, **2015**, 85, 316-24	6.5	27
341	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. *Nature Genetics*, **2015**, 47, 39-46	36.3	177
340	PRIMA1 mutation: a new cause of nocturnal frontal lobe epilepsy. *Annals of Clinical and Translational Neurology*, **2015**, 2, 821-30	5.3	19
339	Loss of synaptic Zn2+ transporter function increases risk of febrile seizures. *Scientific Reports*, **2015**, 5, 17816	4.9	29
338	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. *EBioMedicine*, **2015**, 2, 1063-70	8.8	61
337	Mind the gap: Multiple events and lengthy delays before presentation with a "first seizure". *Epilepsia*, **2015**, 56, 1534-41	6.4	41

336	Familial neonatal seizures in 36 families: Clinical and genetic features correlate with outcome. *Epilepsia*, **2015**, 56, 1071-80	6.4	68
335	Epileptic spasms are a feature of DEPDC5 mTORopathy. *Neurology: Genetics*, **2015**, 1, e17	3.8	44
334	Quinidine in the treatment of KCNT1-positive epilepsies. *Annals of Neurology*, **2015**, 78, 995-9	9.4	145
333	Genetics of Epilepsy in Clinical Practice. *Epilepsy Currents*, **2015**, 15, 192-6	1.3	31
332	Mutation of the nuclear lamin gene LMNB2 in progressive myoclonus epilepsy with early ataxia. *Human Molecular Genetics*, **2015**, 24, 4483-90	5.6	33
331	Familial cortical dysplasia type IIA caused by a germline mutation in DEPDC5. *Annals of Clinical and Translational Neurology*, **2015**, 2, 575-80	5.3	73
330	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, 334-40	3.4	11
329	Reduced dendritic arborization and hyperexcitability of pyramidal neurons in a Scn1b-based model of Dravet syndrome. *Brain*, **2014**, 137, 1701-15	11.2	34
328	The hidden genetics of epilepsy-a clinically important new paradigm. *Nature Reviews Neurology*, **2014**, 10, 283-92	15	170
327	Rasmussen encephalitis and comorbid autoimmune diseases: A window into disease mechanism?. *Neurology*, **2014**, 83, 1049-55	6.5	13
326	Recurrent generalized seizures, visual loss, and palinopsia as phenotypic features of neuronal ceroid lipofuscinosis due to progranulin gene mutation. *Epilepsia*, **2014**, 55, e56-9	6.4	35
325	Weight and fat distribution in patients taking valproate: a valproate-discordant gender-matched twin and sibling pair study. *Epilepsia*, **2014**, 55, 1551-7	6.4	10
324	Brivaracetam as adjunctive treatment for uncontrolled partial epilepsy in adults: a phase III randomized, double-blind, placebo-controlled trial. *Epilepsia*, **2014**, 55, 57-66	6.4	178
323	'Idiopathic' no more! Abnormal interaction of large-scale brain networks in generalized epilepsy. *Brain*, **2014**, 137, 2400-2	11.2	9
322	GABRA1 and STXBP1: novel genetic causes of Dravet syndrome. *Neurology*, **2014**, 82, 1245-53	6.5	180
321	Somatic mutations in cerebral cortical malformations. *New England Journal of Medicine*, **2014**, 371, 733-43	59.2	265
320	Phenotype-genotype complexities: opening DOORS. *Lancet Neurology, The*, **2014**, 13, 24-5	24.1	
319	Concepts and controversies of juvenile myoclonic epilepsy: still an enigmatic epilepsy. *Expert Review of Neurotherapeutics*, **2014**, 14, 819-31	4.3	27

318	A variant of KCC2 from patients with febrile seizures impairs neuronal Cl- extrusion and dendritic spine formation. *EMBO Reports*, **2014**, 15, 723-9	6.5	121
317	Harnessing gene expression networks to prioritize candidate epileptic encephalopathy genes. *PLoS ONE*, **2014**, 9, e102079	3.7	22
316	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. *Human Molecular Genetics*, **2014**, 23, 6069-80	5.6	45
315	Genetic analysis of PHOX2B in sudden unexpected death in epilepsy cases. *Neurology*, **2014**, 83, 1018-21	6.5	16
314	The genetic basis of music ability. *Frontiers in Psychology*, **2014**, 5, 658	3.4	37
313	KCNT1 gain of function in 2 epilepsy phenotypes is reversed by quinidine. *Annals of Neurology*, **2014**, 75, 581-90	9.4	192
312	Atypical multifocal Dravet syndrome lacks generalized seizures and may show later cognitive decline. *Developmental Medicine and Child Neurology*, **2014**, 56, 85-90	3.3	13
311	Is focal cortical dysplasia sporadic? Family evidence for genetic susceptibility. *Epilepsia*, **2014**, 55, e22-6	6.4	22
310	Glucose metabolism transporters and epilepsy: only GLUT1 has an established role. *Epilepsia*, **2014**, 55, e18-21	6.4	23
309	Mutations in mammalian target of rapamycin regulator DEPDC5 cause focal epilepsy with brain malformations. *Annals of Neurology*, **2014**, 75, 782-7	9.4	153
308	Genetics of epilepsy: The testimony of twins in the molecular era. *Neurology*, **2014**, 83, 1042-8	6.5	46
307	Genetics of vasovagal syncope. *Autonomic Neuroscience: Basic and Clinical*, **2014**, 184, 60-5	2.4	14
306	Does variation in NIPA2 contribute to genetic generalized epilepsy?. *Human Genetics*, **2014**, 133, 673-4	6.3	6
305	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, 247-58	5.6	28
304	De novo mutations in epileptic encephalopathies. *Nature*, **2013**, 501, 217-21	50.4	1081
303	GRIN2A mutations cause epilepsy-aphasia spectrum disorders. *Nature Genetics*, **2013**, 45, 1073-6	36.3	249
302	Recent advances in the molecular genetics of epilepsy. *Journal of Medical Genetics*, **2013**, 50, 271-9	5.8	82
301	Do mutations in SCN1B cause Dravet syndrome?. *Epilepsy Research*, **2013**, 103, 97-100	3	9

300	Role of the sodium channel SCN9A in genetic epilepsy with febrile seizures plus and Dravet syndrome. *Epilepsia*, **2013**, 54, e122-6	6.4	46
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