

Sam Berkovic Am,, Faa, Fracp

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479
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528
ext. papers

47,515
ext. citations

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avg, IF

6.89
L-index

#	Paper	IF	Citations
479	Revised terminology and concepts for organization of seizures and epilepsies: report of the ILAE Commission on Classification and Terminology, 2005-2009. <i>Epilepsia</i> , 2010 , 51, 676-85	6.4	2950
478	ILAE classification of the epilepsies: Position paper of the ILAE Commission for Classification and Terminology. <i>Epilepsia</i> , 2017 , 58, 512-521	6.4	2084
477	De novo mutations in epileptic encephalopathies. <i>Nature</i> , 2013 , 501, 217-21	50.4	1081
476	A missense mutation in the neuronal nicotinic acetylcholine receptor alpha 4 subunit is associated with autosomal dominant nocturnal frontal lobe epilepsy. <i>Nature Genetics</i> , 1995 , 11, 201-3	36.3	922
475	Doublecortin, a brain-specific gene mutated in human X-linked lissencephaly and double cortex syndrome, encodes a putative signaling protein. <i>Cell</i> , 1998 , 92, 63-72	56.2	904
474	A potassium channel mutation in neonatal human epilepsy. <i>Science</i> , 1998 , 279, 403-6	33.3	884
473	Febrile seizures and generalized epilepsy associated with a mutation in the Na ⁺ -channel beta1 subunit gene SCN1B. <i>Nature Genetics</i> , 1998 , 19, 366-70	36.3	851
472	Mutations in filamin 1 prevent migration of cerebral cortical neurons in human periventricular heterotopia. <i>Neuron</i> , 1998 , 21, 1315-25	13.9	729
471	Analysis of shared heritability in common disorders of the brain. <i>Science</i> , 2018 , 360,	33.3	666
470	Mutant GABA(A) receptor gamma2-subunit in childhood absence epilepsy and febrile seizures. <i>Nature Genetics</i> , 2001 , 28, 49-52	36.3	643
469	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. <i>Nature Genetics</i> , 2013 , 45, 825-30	36.3	500
468	Prediction of seizure likelihood with a long-term, implanted seizure advisory system in patients with drug-resistant epilepsy: a first-in-man study. <i>Lancet Neurology</i> , 2013 , 12, 563-71	24.1	482
467	Autosomal dominant nocturnal frontal lobe epilepsy. A distinctive clinical disorder. <i>Brain</i> , 1995 , 118 (Pt 1), 61-73	11.2	471
466	Epileptology of the first-seizure presentation: a clinical, electroencephalographic, and magnetic resonance imaging study of 300 consecutive patients. <i>Lancet</i> , 1998 , 352, 1007-11	40	440
465	The spectrum of SCN1A-related infantile epileptic encephalopathies. <i>Brain</i> , 2007 , 130, 843-52	11.2	423
464	Truncation of the GABA(A)-receptor gamma2 subunit in a family with generalized epilepsy with febrile seizures plus. <i>American Journal of Human Genetics</i> , 2002 , 70, 530-6	11	373
463	Temporal lobectomy: long-term seizure outcome, late recurrence and risks for seizure recurrence. <i>Brain</i> , 2004 , 127, 2018-30	11.2	351

462	An insertion mutation of the CHRNA4 gene in a family with autosomal dominant nocturnal frontal lobe epilepsy. <i>Human Molecular Genetics</i> , 1997 , 6, 943-7	5.6	349
461	Strikingly different clinicopathological phenotypes determined by progranulin-mutation dosage. <i>American Journal of Human Genetics</i> , 2012 , 90, 1102-7	11	336
460	X-linked protocadherin 19 mutations cause female-limited epilepsy and cognitive impairment. <i>Nature Genetics</i> , 2008 , 40, 776-81	36.3	328
459	KCNQ2 encephalopathy: emerging phenotype of a neonatal epileptic encephalopathy. <i>Annals of Neurology</i> , 2012 , 71, 15-25	9.4	322
458	Hippocampal sclerosis in temporal lobe epilepsy demonstrated by magnetic resonance imaging. <i>Annals of Neurology</i> , 1991 , 29, 175-82	9.4	309
457	Epilepsies in twins: genetics of the major epilepsy syndromes. <i>Annals of Neurology</i> , 1998 , 43, 435-45	9.4	297
456	Paroxysmal exercise-induced dyskinesia and epilepsy is due to mutations in SLC2A1, encoding the glucose transporter GLUT1. <i>Brain</i> , 2008 , 131, 1831-44	11.2	297
455	Sodium-channel defects in benign familial neonatal-infantile seizures. <i>Lancet, The</i> , 2002 , 360, 851-2	40	284
454	Magnetic resonance imaging in temporal lobe epilepsy: pathological correlations. <i>Annals of Neurology</i> , 1987 , 22, 341-7	9.4	284
453	SCN1A mutations and epilepsy. <i>Human Mutation</i> , 2005 , 25, 535-42	4.7	282
452	GABRD encoding a protein for extra- or peri-synaptic GABAA receptors is a susceptibility locus for generalized epilepsies. <i>Human Molecular Genetics</i> , 2004 , 13, 1315-9	5.6	267
451	CHRNA2 is the second acetylcholine receptor subunit associated with autosomal dominant nocturnal frontal lobe epilepsy. <i>American Journal of Human Genetics</i> , 2001 , 68, 225-31	11	267
450	Progressive myoclonus epilepsies: specific causes and diagnosis. <i>New England Journal of Medicine</i> , 1986 , 315, 296-305	59.2	267
449	Somatic mutations in cerebral cortical malformations. <i>New England Journal of Medicine</i> , 2014 , 371, 733-43	59.2	265
448	Missense mutations in the sodium-gated potassium channel gene KCNT1 cause severe autosomal dominant nocturnal frontal lobe epilepsy. <i>Nature Genetics</i> , 2012 , 44, 1188-90	36.3	253
447	GRIN2A mutations cause epilepsy-aphasia spectrum disorders. <i>Nature Genetics</i> , 2013 , 45, 1073-6	36.3	249
446	De-novo mutations of the sodium channel gene SCN1A in alleged vaccine encephalopathy: a retrospective study. <i>Lancet Neurology, The</i> , 2006 , 5, 488-92	24.1	241
445	Generalized epilepsy with febrile seizures plus: a common childhood-onset genetic epilepsy syndrome. <i>Annals of Neurology</i> , 1999 , 45, 75-81	9.4	239

444	Mutations in DEPDC5 cause familial focal epilepsy with variable foci. <i>Nature Genetics</i> , 2013 , 45, 546-51	36.3	238
443	Early-onset absence epilepsy caused by mutations in the glucose transporter GLUT1. <i>Annals of Neurology</i> , 2009 , 66, 415-9	9.4	230
442	Benign familial neonatal-infantile seizures: characterization of a new sodium channelopathy. <i>Annals of Neurology</i> , 2004 , 55, 550-7	9.4	225
441	Navigating the channels and beyond: unravelling the genetics of the epilepsies. <i>Lancet Neurology</i> , 2008 , 7, 231-45	24.1	219
440	Phenotypic characterization of an alpha 4 neuronal nicotinic acetylcholine receptor subunit knock-out mouse. <i>Journal of Neuroscience</i> , 2000 , 20, 6431-41	6.6	217
439	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2017 , 101, 664-685	11	214
438	SRPX2 mutations in disorders of language cortex and cognition. <i>Human Molecular Genetics</i> , 2006 , 15, 1195-207	5.6	213
437	Array-based gene discovery with three unrelated subjects shows SCARB2/LIMP-2 deficiency causes myoclonus epilepsy and glomerulosclerosis. <i>American Journal of Human Genetics</i> , 2008 , 82, 673-84	11	205
436	PRRT2 mutations cause benign familial infantile epilepsy and infantile convulsions with choreoathetosis syndrome. <i>American Journal of Human Genetics</i> , 2012 , 90, 152-60	11	199
435	Placebo-controlled study of levetiracetam in idiopathic generalized epilepsy. <i>Neurology</i> , 2007 , 69, 1751-60	11.2	198
434	KCNT1 gain of function in 2 epilepsy phenotypes is reversed by quinidine. <i>Annals of Neurology</i> , 2014 , 75, 581-90	9.4	192
433	Localization of epileptic foci with postictal single photon emission computed tomography. <i>Annals of Neurology</i> , 1989 , 26, 660-8	9.4	191
432	Familial and sporadic 15q13.3 microdeletions in idiopathic generalized epilepsy: precedent for disorders with complex inheritance. <i>Human Molecular Genetics</i> , 2009 , 18, 3626-31	5.6	190
431	Temporal lobe epilepsy and GEFS+ phenotypes associated with SCN1B mutations. <i>Brain</i> , 2007 , 130, 100-9	11.2	188
430	Familial temporal lobe epilepsy: a common disorder identified in twins. <i>Annals of Neurology</i> , 1996 , 40, 227-35	9.4	187
429	. <i>Nature Genetics</i> , 2001 , 28, 49-52	36.3	184
428	Kufs' disease: a critical reappraisal. <i>Brain</i> , 1988 , 111 (Pt 1), 27-62	11.2	182
427	Definition and diagnostic criteria of sleep-related hypermotor epilepsy. <i>Neurology</i> , 2016 , 86, 1834-42	6.5	182

426	GABRA1 and STXBP1: novel genetic causes of Dravet syndrome. <i>Neurology</i> , 2014 , 82, 1245-53	6.5	180
425	Brivaracetam as adjunctive treatment for uncontrolled partial epilepsy in adults: a phase III randomized, double-blind, placebo-controlled trial. <i>Epilepsia</i> , 2014 , 55, 57-66	6.4	178
424	Distinguishing sleep disorders from seizures: diagnosing bumps in the night. <i>Archives of Neurology</i> , 2006 , 63, 705-9		178
423	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. <i>Nature Genetics</i> , 2015 , 47, 39-46	36.3	177
422	Clinical and imaging heterogeneity of polymicrogyria: a study of 328 patients. <i>Brain</i> , 2010 , 133, 1415-27	11.2	177
421	Rare copy number variants are an important cause of epileptic encephalopathies. <i>Annals of Neurology</i> , 2011 , 70, 974-85	9.4	176
420	The hidden genetics of epilepsy-a clinically important new paradigm. <i>Nature Reviews Neurology</i> , 2014 , 10, 283-92	15	170
419	A homozygous mutation in human PRICKLE1 causes an autosomal-recessive progressive myoclonus epilepsy-ataxia syndrome. <i>American Journal of Human Genetics</i> , 2008 , 83, 572-81	11	170
418	Reduced cortical inhibition in a mouse model of familial childhood absence epilepsy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2007 , 104, 17536-41	11.5	164
417	Exome-based analysis of cardiac arrhythmia, respiratory control, and epilepsy genes in sudden unexpected death in epilepsy. <i>Annals of Neurology</i> , 2016 , 79, 522-34	9.4	162
416	Genetic association studies in epilepsy: "the truth is out there". <i>Epilepsia</i> , 2004 , 45, 1429-42	6.4	161
415	Characterization of mutations in the gene doublecortin in patients with double cortex syndrome. <i>Annals of Neurology</i> , 1999 , 45, 146-53	9.4	157
414	Mechanisms of human inherited epilepsies. <i>Progress in Neurobiology</i> , 2009 , 87, 41-57	10.9	156
413	Seizure-associated hippocampal volume loss: a longitudinal magnetic resonance study of temporal lobe epilepsy. <i>Annals of Neurology</i> , 2002 , 51, 641-4	9.4	154
412	Mutations in mammalian target of rapamycin regulator DEPDC5 cause focal epilepsy with brain malformations. <i>Annals of Neurology</i> , 2014 , 75, 782-7	9.4	153
411	NREM arousal parasomnias and their distinction from nocturnal frontal lobe epilepsy: a video EEG analysis. <i>Sleep</i> , 2009 , 32, 1637-44	1.1	152
410	Epilepsy and mental retardation limited to females: an under-recognized disorder. <i>Brain</i> , 2008 , 131, 918-27		152
409	Multicentre search for genetic susceptibility loci in sporadic epilepsy syndrome and seizure types: a case-control study. <i>Lancet Neurology</i> , 2007 , 6, 970-80	24.1	152

408	Extended spectrum of idiopathic generalized epilepsies associated with CACNA1H functional variants. <i>Annals of Neurology</i> , 2007 , 62, 560-8	9.4	150
407	Interictal spikes and epileptic seizures: their relationship and underlying rhythmicity. <i>Brain</i> , 2016 , 139, 1066-78	11.2	148
406	Human epilepsies: interaction of genetic and acquired factors. <i>Trends in Neurosciences</i> , 2006 , 29, 391-397	13.3	147
405	Quinidine in the treatment of KCNT1-positive epilepsies. <i>Annals of Neurology</i> , 2015 , 78, 995-9	9.4	145
404	Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. <i>Brain</i> , 2013 , 136, 3140-50	11.2	144
403	Channelopathies as a genetic cause of epilepsy. <i>Current Opinion in Neurology</i> , 2003 , 16, 171-176	7.1	141
402	Magnetic stimulation of the brain in generalized epilepsy: reversal of cortical hyperexcitability by anticonvulsants. <i>Annals of Neurology</i> , 1993 , 34, 351-5	9.4	141
401	Mutations in the mammalian target of rapamycin pathway regulators NPRL2 and NPRL3 cause focal epilepsy. <i>Annals of Neurology</i> , 2016 , 79, 120-31	9.4	136
400	Lateralization of verbal memory and unilateral hippocampal sclerosis: evidence of task-specific effects. <i>Neuropsychology, Development and Cognition Section A: Journal of Clinical and Experimental Neuropsychology</i> , 1993 , 15, 608-18		133
399	Paroxysmal motor disorders of sleep: the clinical spectrum and differentiation from epilepsy. <i>Epilepsia</i> , 2006 , 47, 1775-91	6.4	129
398	Hypothalamic hamartoma and seizures: a treatable epileptic encephalopathy. <i>Epilepsia</i> , 2003 , 44, 969-73	6.4	129
397	SCN1A duplications and deletions detected in Dravet syndrome: implications for molecular diagnosis. <i>Epilepsia</i> , 2009 , 50, 1670-8	6.4	127
396	Effects of angiotensin II on dopamine and serotonin turnover in the striatum of conscious rats. <i>Brain Research</i> , 1993 , 613, 221-9	3.7	127
395	Childhood absence epilepsy and febrile seizures: a family with a GABA(A) receptor mutation. <i>Brain</i> , 2003 , 126, 230-40	11.2	122
394	A variant of KCC2 from patients with febrile seizures impairs neuronal Cl ⁻ extrusion and dendritic spine formation. <i>EMBO Reports</i> , 2014 , 15, 723-9	6.5	121
393	The genetics of human epilepsy. <i>Trends in Pharmacological Sciences</i> , 2003 , 24, 428-33	13.2	120
392	Transcallosal resection of hypothalamic hamartomas, with control of seizures, in children with gelastic epilepsy. <i>Neurosurgery</i> , 2001 , 48, 108-18	3.2	120
391	Autosomal dominant rolandic epilepsy and speech dyspraxia: a new syndrome with anticipation. <i>Annals of Neurology</i> , 1995 , 38, 633-42	9.4	117

390	Copy number variants are frequent in genetic generalized epilepsy with intellectual disability. <i>Neurology</i> , 2013 , 81, 1507-14	6.5	115
389	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. <i>American Journal of Human Genetics</i> , 2015 , 96, 808-15	11	114
388	Genome-wide association analysis of genetic generalized epilepsies implicates susceptibility loci at 1q43, 2p16.1, 2q22.3 and 17q21.32. <i>Human Molecular Genetics</i> , 2012 , 21, 5359-72	5.6	114
387	Kufs disease, the major adult form of neuronal ceroid lipofuscinosis, caused by mutations in CLN6. <i>American Journal of Human Genetics</i> , 2011 , 88, 566-73	11	114
386	MR imaging and spectroscopic study of epileptogenic hypothalamic hamartomas: analysis of 72 cases. <i>American Journal of Neuroradiology</i> , 2004 , 25, 450-62	4.4	114
385	Comparison of ictal SPECT and interictal PET in the presurgical evaluation of temporal lobe epilepsy. <i>Annals of Neurology</i> , 1995 , 37, 738-45	9.4	112
384	Febrile seizures and hippocampal sclerosis: frequent and related findings in intractable temporal lobe epilepsy of childhood. <i>Pediatric Neurology</i> , 1995 , 12, 201-6	2.9	111
383	Glucose transporter 1 deficiency as a treatable cause of myoclonic astatic epilepsy. <i>Archives of Neurology</i> , 2011 , 68, 1152-5		110
382	Risk factors for sudden unexpected death in epilepsy: a controlled prospective study based on coroners cases. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2003 , 12, 456-64	3.2	110
381	Occipital epilepsies: identification of specific and newly recognized syndromes. <i>Brain</i> , 2003 , 126, 753-69	11.2	109
380	Mitochondrial dysfunction in multiple symmetrical lipomatosis. <i>Annals of Neurology</i> , 1991 , 29, 566-9	9.4	109
379	Genetic architecture of idiopathic generalized epilepsy: clinical genetic analysis of 55 multiplex families. <i>Epilepsia</i> , 2004 , 45, 467-78	6.4	106
378	Validation of a questionnaire for clinical seizure diagnosis. <i>Epilepsia</i> , 1992 , 33, 1065-71	6.4	105
377	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. <i>American Journal of Human Genetics</i> , 2019 , 105, 267-282	11	104
376	Ion Channels in Genetic Epilepsy: From Genes and Mechanisms to Disease-Targeted Therapies. <i>Pharmacological Reviews</i> , 2018 , 70, 142-173	22.5	103
375	A focal epilepsy and intellectual disability syndrome is due to a mutation in TBC1D24. <i>American Journal of Human Genetics</i> , 2010 , 87, 371-5	11	100
374	Glucose transporter 1 deficiency in the idiopathic generalized epilepsies. <i>Annals of Neurology</i> , 2012 , 72, 807-15	9.4	98
373	Familial partial epilepsy with variable foci: a new partial epilepsy syndrome with suggestion of linkage to chromosome 2. <i>Annals of Neurology</i> , 1998 , 44, 890-9	9.4	97

372	Effects of vaccination on onset and outcome of Dravet syndrome: a retrospective study. <i>Lancet Neurology, The</i> , 2010 , 9, 592-8	24.1	95
371	Focal cortical myoclonus and rolandic cortical dysplasia: clarification by magnetic resonance imaging. <i>Annals of Neurology</i> , 1988 , 23, 317-25	9.4	94
370	Genetic variation of CACNA1H in idiopathic generalized epilepsy. <i>Annals of Neurology</i> , 2004 , 55, 595-6	9.4	92
369	Optimizing genomic medicine in epilepsy through a gene-customized approach to missense variant interpretation. <i>Genome Research</i> , 2017 , 27, 1715-1729	9.7	91
368	Cathepsin F mutations cause Type B Kufs disease, an adult-onset neuronal ceroid lipofuscinosis. <i>Human Molecular Genetics</i> , 2013 , 22, 1417-23	5.6	90
367	SCN2A mutations and benign familial neonatal-infantile seizures: the phenotypic spectrum. <i>Epilepsia</i> , 2007 , 48, 1138-42	6.4	90
366	Altered kinetics and benzodiazepine sensitivity of a GABAA receptor subunit mutation [gamma 2(R43Q)] found in human epilepsy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2002 , 99, 15170-5	11.5	90
365	The peri-ictal state: cortical excitability changes within 24 h of a seizure. <i>Brain</i> , 2009 , 132, 1013-21	11.2	89
364	Changes in cortical excitability differentiate generalized and focal epilepsy. <i>Annals of Neurology</i> , 2007 , 61, 324-31	9.4	89
363	Genetic and neuroradiological heterogeneity of double cortex syndrome. <i>Annals of Neurology</i> , 2000 , 47, 265-269	9.4	89
362	Clinical applications: MRI, SPECT, and PET. <i>Magnetic Resonance Imaging</i> , 1995 , 13, 1119-24	3.3	87
361	Limbic P3 potentials, seizure localization, and surgical pathology in temporal lobe epilepsy. <i>Annals of Neurology</i> , 1989 , 26, 377-85	9.4	87
360	Channelopathies in idiopathic epilepsy. <i>Neurotherapeutics</i> , 2007 , 4, 295-304	6.4	86
359	Clinical challenges and future therapeutic approaches for neuronal ceroid lipofuscinosis. <i>Lancet Neurology, The</i> , 2019 , 18, 107-116	24.1	86
358	A mutation in the Golgi Qb-SNARE gene GOSR2 causes progressive myoclonus epilepsy with early ataxia. <i>American Journal of Human Genetics</i> , 2011 , 88, 657-63	11	85
357	SCARB2 mutations in progressive myoclonus epilepsy (PME) without renal failure. <i>Annals of Neurology</i> , 2009 , 66, 532-6	9.4	84
356	Precision therapy for epilepsy due to mutations: A randomized trial of oral quinidine. <i>Neurology</i> , 2018 , 90, e67-e72	6.5	84
355	Sodium channels and the neurobiology of epilepsy. <i>Epilepsia</i> , 2012 , 53, 1849-59	6.4	83

354	Transcallosal Resection of Hypothalamic Hamartomas, with Control of Seizures, in Children with Gelastic Epilepsy. <i>Neurosurgery</i> , 2001 , 48, 108-118	3.2	83
353	Recent advances in the molecular genetics of epilepsy. <i>Journal of Medical Genetics</i> , 2013 , 50, 271-9	5.8	82
352	Chromosomal abnormalities and epilepsy: a review for clinicians and gene hunters. <i>Epilepsia</i> , 2002 , 43, 127-40	6.4	82
351	CHD2 variants are a risk factor for photosensitivity in epilepsy. <i>Brain</i> , 2015 , 138, 1198-207	11.2	81
350	Susceptibility genes for complex epilepsy. <i>Human Molecular Genetics</i> , 2005 , 14 Spec No. 2, R243-9	5.6	80
349	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. <i>Human Molecular Genetics</i> , 2015 , 24, 5250-9	5.6	78
348	Timing of de novo mutagenesis--a twin study of sodium-channel mutations. <i>New England Journal of Medicine</i> , 2010 , 363, 1335-40	59.2	78
347	Mapping of a gene determining familial partial epilepsy with variable foci to chromosome 22q11-q12. <i>American Journal of Human Genetics</i> , 1999 , 65, 1698-710	11	78
346	Long-term follow-up of febrile infection-related epilepsy syndrome. <i>Epilepsia</i> , 2012 , 53, 101-10	6.4	76
345	TBC1D24 genotype-phenotype correlation: Epilepsies and other neurologic features. <i>Neurology</i> , 2016 , 87, 77-85	6.5	75
344	Early onset absence epilepsy: 1 in 10 cases is caused by GLUT1 deficiency. <i>Epilepsia</i> , 2012 , 53, e204-7	6.4	75
343	Predicting seizure control: cortical excitability and antiepileptic medication. <i>Annals of Neurology</i> , 2010 , 67, 64-73	9.4	75
342	Augmented currents of an HCN2 variant in patients with febrile seizure syndromes. <i>Annals of Neurology</i> , 2010 , 67, 542-6	9.4	75
341	Ictal 99mTc-HMPAO single photon emission computed tomography in children with temporal lobe epilepsy. <i>Epilepsia</i> , 1993 , 34, 869-77	6.4	75
340	Familial cortical dysplasia type IIA caused by a germline mutation in DEPDC5. <i>Annals of Clinical and Translational Neurology</i> , 2015 , 2, 575-80	5.3	73
339	Genetics of the epilepsies. <i>Epilepsia</i> , 2001 , 42 Suppl 5, 16-23	6.4	73
338	Action myoclonus-renal failure syndrome: characterization of a unique cerebro-renal disorder. <i>Brain</i> , 2004 , 127, 2173-82	11.2	72
337	Long-term seizure outcome and risk factors for recurrence after extratemporal epilepsy surgery. <i>Epilepsia</i> , 2012 , 53, 970-8	6.4	70

336	Axon initial segment dysfunction in epilepsy. <i>Journal of Physiology</i> , 2010 , 588, 1829-40	3.9	70
335	The borderland of epilepsy: clinical and molecular features of phenomena that mimic epileptic seizures. <i>Lancet Neurology</i> , 2009 , 8, 370-81	24.1	69
334	Genetic epilepsy with febrile seizures plus: Refining the spectrum. <i>Neurology</i> , 2017 , 89, 1210-1219	6.5	68
333	Familial neonatal seizures in 36 families: Clinical and genetic features correlate with outcome. <i>Epilepsia</i> , 2015 , 56, 1071-80	6.4	68
332	Parental Mosaicism in "De Novo" Epileptic Encephalopathies. <i>New England Journal of Medicine</i> , 2018 , 378, 1646-1648	59.2	67
331	Nicotine-induced dystonic arousal complex in a mouse line harboring a human autosomal-dominant nocturnal frontal lobe epilepsy mutation. <i>Journal of Neuroscience</i> , 2007 , 27, 10128-42	6.6	67
330	Idiopathic generalized epilepsy with generalized and other seizures in adolescence. <i>Epilepsia</i> , 2001 , 42, 317-20	6.4	67
329	PRRT2 phenotypic spectrum includes sporadic and fever-related infantile seizures. <i>Neurology</i> , 2012 , 79, 2104-8	6.5	65
328	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. <i>Brain</i> , 2013 , 136, 3378-94	11.2	65
327	Comparison of antiepileptic drug levels in sudden unexpected deaths in epilepsy with deaths from other causes. <i>Epilepsia</i> , 1999 , 40, 1795-8	6.4	65
326	Axon initial segment dysfunction in a mouse model of genetic epilepsy with febrile seizures plus. <i>Journal of Clinical Investigation</i> , 2010 , 120, 2661-71	15.9	65
325	Epilepsy and mental retardation limited to females with PCDH19 mutations can present de novo or in single generation families. <i>Journal of Medical Genetics</i> , 2010 , 47, 211-6	5.8	64
324	Temporal lobe epilepsy subtypes: differential patterns of cerebral perfusion on ictal SPECT. <i>Epilepsia</i> , 1996 , 37, 788-95	6.4	64
323	Real-world utility of whole exome sequencing with targeted gene analysis for focal epilepsy. <i>Epilepsy Research</i> , 2017 , 131, 1-8	3	63
322	Benign mesial temporal lobe epilepsy. <i>Nature Reviews Neurology</i> , 2011 , 7, 237-40	15	63
321	Hemicranial volume deficits in patients with temporal lobe epilepsy with and without hippocampal sclerosis. <i>Epilepsia</i> , 1998 , 39, 1174-81	6.4	63
320	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. <i>EBioMedicine</i> , 2015 , 2, 1063-70	8.8	61
319	Progressive gait deterioration in adolescents with Dravet syndrome. <i>Archives of Neurology</i> , 2012 , 69, 873-8		61

318	Benign occipital epilepsies of childhood: clinical features and genetics. <i>Brain</i> , 2008 , 131, 2287-94	11.2	60
317	Familial mesial temporal lobe epilepsy: a benign epilepsy syndrome showing complex inheritance. <i>Brain</i> , 2010 , 133, 3221-31	11.2	59
316	Update on pharmacogenetics in epilepsy: a brief review. <i>Lancet Neurology, The</i> , 2006 , 5, 189-96	24.1	59
315	A childhood epilepsy mutation reveals a role for developmentally regulated splicing of a sodium channel. <i>Molecular and Cellular Neurosciences</i> , 2007 , 35, 292-301	4.8	59
314	Analyzing the etiology of benign rolandic epilepsy: a multicenter twin collaboration. <i>Epilepsia</i> , 2006 , 47, 550-5	6.4	59
313	Multiple molecular mechanisms for a single GABAA mutation in epilepsy. <i>Neurology</i> , 2013 , 80, 1003-8	6.5	58
312	CHD2 myoclonic encephalopathy is frequently associated with self-induced seizures. <i>Neurology</i> , 2015 , 84, 951-8	6.5	57
311	Phenotypic comparison of two Scottish families with mutations in different genes causing autosomal dominant nocturnal frontal lobe epilepsy. <i>Epilepsia</i> , 2003 , 44, 613-7	6.4	57
310	A multicenter study of BRD2 as a risk factor for juvenile myoclonic epilepsy. <i>Epilepsia</i> , 2007 , 48, 706-12	6.4	56
309	The psychological impact of a newly diagnosed seizure: losing and restoring perceived control. <i>Epilepsy and Behavior</i> , 2007 , 10, 223-33	3.2	56
308	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. <i>Annals of Neurology</i> , 2019 , 86, 821-831	9.4	55
307	Neonatal epilepsy syndromes and generalized epilepsy with febrile seizures plus (GEFS+). <i>Epilepsia</i> , 2005 , 46 Suppl 10, 41-7	6.4	55
306	encephalopathy: A distinctive generalized developmental and epileptic encephalopathy. <i>Neurology</i> , 2019 , 92, e96-e107	6.5	55
305	SCN1A testing for epilepsy: application in clinical practice. <i>Epilepsia</i> , 2013 , 54, 946-52	6.4	54
304	Intracortical hyperexcitability in humans with a GABAA receptor mutation. <i>Cerebral Cortex</i> , 2008 , 18, 664-9	5.1	54
303	Juvenile myoclonic epilepsy and idiopathic photosensitive occipital lobe epilepsy: is there overlap?. <i>Brain</i> , 2004 , 127, 1878-86	11.2	54
302	Networks underlying paroxysmal fast activity and slow spike and wave in Lennox-Gastaut syndrome. <i>Neurology</i> , 2013 , 81, 665-73	6.5	53
301	Cortical excitability and refractory epilepsy: a three-year longitudinal transcranial magnetic stimulation study. <i>International Journal of Neural Systems</i> , 2013 , 23, 1250030	6.2	52

300	Neonatal seizures and long QT syndrome: a cardiocerebral channelopathy?. <i>Epilepsia</i> , 2010 , 51, 293-6	6.4	52
299	Human nocturnal frontal lobe epilepsy: pharmacogenomic profiles of pathogenic nicotinic acetylcholine receptor beta-subunit mutations outside the ion channel pore. <i>Molecular Pharmacology</i> , 2008 , 74, 379-91	4.3	51
298	Mutation in the Na ⁺ channel subunit SCN1B produces paradoxical changes in peripheral nerve excitability. <i>Brain</i> , 2005 , 128, 1841-6	11.2	51
297	Does cardiac conduction pathology contribute to sudden unexpected death in epilepsy?. <i>Epilepsy Research</i> , 2000 , 40, 17-24	3	51
296	Channelopathies as a genetic cause of epilepsy. <i>Current Opinion in Neurology</i> , 2003 , 16, 171-6	7.1	51
295	GENETICS. The Human Variome Project. <i>Science</i> , 2008 , 322, 861-2	33.3	50
294	Dominant KCNA2 mutation causes episodic ataxia and pharmaco-responsive epilepsy. <i>Neurology</i> , 2016 , 87, 1975-1984	6.5	50
293	'North Sea' progressive myoclonus epilepsy: phenotype of subjects with GOSR2 mutation. <i>Brain</i> , 2013 , 136, 1146-54	11.2	49
292	Clinical and molecular genetics of myoclonic-astatic epilepsy and severe myoclonic epilepsy in infancy (Dravet syndrome). <i>Brain and Development</i> , 2001 , 23, 732-5	2.2	49
291	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. <i>Nature Communications</i> , 2019 , 10, 4920	17.4	48
290	Molecular analysis of ring chromosome 20 syndrome reveals two distinct groups of patients. <i>Journal of Medical Genetics</i> , 2011 , 48, 1-9	5.8	48
289	Small temporal pole encephalocoles: a treatable cause of "lesion negative" temporal lobe epilepsy. <i>Epilepsia</i> , 2010 , 51, 2199-202	6.4	48
288	Developmental impact of a familial GABAA receptor epilepsy mutation. <i>Annals of Neurology</i> , 2008 , 64, 284-93	9.4	48
287	Familial partial epilepsy with variable foci: clinical features and linkage to chromosome 22q12. <i>Epilepsia</i> , 2004 , 45, 1054-60	6.4	48
286	Role of the sodium channel SCN9A in genetic epilepsy with febrile seizures plus and Dravet syndrome. <i>Epilepsia</i> , 2013 , 54, e122-6	6.4	46
285	Genetics of epilepsy: The testimony of twins in the molecular era. <i>Neurology</i> , 2014 , 83, 1042-8	6.5	46
284	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. <i>Human Molecular Genetics</i> , 2014 , 23, 6069-80	5.6	45
283	Etiology of hippocampal sclerosis: evidence for a predisposing familial morphologic anomaly. <i>Neurology</i> , 2013 , 81, 144-9	6.5	45

282	Epileptic spasms are a feature of DEPDC5 mTORopathy. <i>Neurology: Genetics</i> , 2015 , 1, e17	3.8	44
281	Verbal memory in left temporal lobe epilepsy: evidence for task-related localization. <i>Annals of Neurology</i> , 2002 , 51, 442-7	9.4	44
280	Assessment of the role of FDG PET in the diagnosis and management of children with refractory epilepsy. <i>European Journal of Nuclear Medicine and Molecular Imaging</i> , 2005 , 32, 1311-6	8.8	43
279	Clinical and neurophysiologic features of progressive myoclonus epilepsy without renal failure caused by SCARB2 mutations. <i>Epilepsia</i> , 2011 , 52, 2356-63	6.4	42
278	Exploration of the genetic architecture of idiopathic generalized epilepsies. <i>Epilepsia</i> , 2006 , 47, 1682-90	6.4	42
277	Is benign rolandic epilepsy genetically determined?. <i>Annals of Neurology</i> , 2004 , 56, 129-32	9.4	42
276	Febrile seizures: genetics and relationship to other epilepsy syndromes. <i>Current Opinion in Neurology</i> , 1998 , 11, 129-34	7.1	42
275	Mind the gap: Multiple events and lengthy delays before presentation with a "first seizure". <i>Epilepsia</i> , 2015 , 56, 1534-41	6.4	41
274	Severe autosomal dominant nocturnal frontal lobe epilepsy associated with psychiatric disorders and intellectual disability. <i>Epilepsia</i> , 2008 , 49, 2125-9	6.4	40
273	Increased serotonin receptor availability in human sleep: evidence from an [18F]MPPF PET study in narcolepsy. <i>NeuroImage</i> , 2006 , 30, 341-8	7.9	40
272	Concussive convulsions. Incidence in sport and treatment recommendations. <i>Sports Medicine</i> , 1998 , 25, 131-6	10.6	40
271	Epilepsy genetics: clinical impacts and biological insights. <i>Lancet Neurology</i> , 2020 , 19, 93-100	24.1	40
270	Aberrant Inclusion of a Poison Exon Causes Dravet Syndrome and Related SCN1A-Associated Genetic Epilepsies. <i>American Journal of Human Genetics</i> , 2018 , 103, 1022-1029	11	40
269	Dynamic action potential clamp predicts functional separation in mild familial and severe de novo forms of epilepsy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018 , 115, E5516-E5525	11.5	40
268	Myoclonus epilepsy and ataxia due to KCNC1 mutation: Analysis of 20 cases and K channel properties. <i>Annals of Neurology</i> , 2017 , 81, 677-689	9.4	39
267	Genetic generalized epilepsies. <i>Epilepsia</i> , 2018 , 59, 1148-1153	6.4	39
266	A new clinical and molecular form of Unverricht-Lundborg disease localized by homozygosity mapping. <i>Brain</i> , 2005 , 128, 652-8	11.2	39
265	Clinical genetic studies in benign childhood epilepsy with centrotemporal spikes. <i>Epilepsia</i> , 2012 , 53, 319-24	6.4	38

264	Multidrug-resistant genotype (ABCB1) and seizure recurrence in newly treated epilepsy: data from international pharmacogenetic cohorts. <i>Epilepsia</i> , 2009 , 50, 1689-96	6.4	38
263	Progressive myoclonus epilepsies: clinical and genetic aspects. <i>Epilepsia</i> , 1993 , 34 Suppl 3, S19-30	6.4	38
262	EEG in adult-onset idiopathic generalized epilepsy. <i>Epilepsia</i> , 2003 , 44, 252-6	6.4	38
261	The genetic basis of music ability. <i>Frontiers in Psychology</i> , 2014 , 5, 658	3.4	37
260	Epilepsies with single gene inheritance. <i>Brain and Development</i> , 1997 , 19, 13-8	2.2	37
259	A targeted resequencing gene panel for focal epilepsy. <i>Neurology</i> , 2016 , 86, 1605-12	6.5	37
258	Mutations of the Sonic Hedgehog Pathway Underlie Hypothalamic Hamartoma with Gelastic Epilepsy. <i>American Journal of Human Genetics</i> , 2016 , 99, 423-9	11	37
257	Psychological trajectories in the year after a newly diagnosed seizure. <i>Epilepsia</i> , 2012 , 53, 1774-81	6.4	36
256	Aggravation of generalized epilepsies. <i>Epilepsia</i> , 1998 , 39 Suppl 3, S11-4	6.4	36
255	Mortality in patients with psychogenic nonepileptic seizures. <i>Neurology</i> , 2020 , 95, e643-e652	6.5	36
254	Recurrent generalized seizures, visual loss, and palinopsia as phenotypic features of neuronal ceroid lipofuscinosis due to progranulin gene mutation. <i>Epilepsia</i> , 2014 , 55, e56-9	6.4	35
253	Balance impairment in chronic antiepileptic drug users: a twin and sibling study. <i>Epilepsia</i> , 2010 , 51, 280-8	6.4	35
252	Causes of epilepsies: insights from discordant monozygous twins. <i>Annals of Neurology</i> , 2001 , 49, 45-52	9.4	35
251	Reduced dendritic arborization and hyperexcitability of pyramidal neurons in a Scn1b-based model of Dravet syndrome. <i>Brain</i> , 2014 , 137, 1701-15	11.2	34
250	Genetics of epilepsy syndromes in families with photosensitivity. <i>Neurology</i> , 2013 , 80, 1322-9	6.5	34
249	Clinical genetic study of the epilepsy-aphasia spectrum. <i>Epilepsia</i> , 2013 , 54, 280-7	6.4	34
248	Prediction by modeling that epilepsy may be caused by very small functional changes in ion channels. <i>Archives of Neurology</i> , 2009 , 66, 1225-32		34
247	Cognitive complaints after a first seizure in adulthood: Influence of psychological adjustment. <i>Epilepsia</i> , 2009 , 50, 1012-21	6.4	34

246	Early seizures after temporal lobectomy predict subsequent seizure recurrence. <i>Annals of Neurology</i> , 2005 , 57, 283-8	9.4	34
245	Mutation of the nuclear lamin gene LMNB2 in progressive myoclonus epilepsy with early ataxia. <i>Human Molecular Genetics</i> , 2015 , 24, 4483-90	5.6	33
244	De novo SCN1A mutations in Dravet syndrome and related epileptic encephalopathies are largely of paternal origin. <i>Journal of Medical Genetics</i> , 2010 , 47, 137-41	5.8	33
243	Vaccination, seizures and 'vaccine damage'. <i>Current Opinion in Neurology</i> , 2007 , 20, 181-7	7.1	33
242	Efficacy and safety of levetiracetam 1000-3000 mg/day in patients with refractory partial-onset seizures: a multicenter, open-label single-arm study. <i>Epilepsy Research</i> , 2005 , 63, 1-9	3	32
241	Genetics of Epilepsy in Clinical Practice. <i>Epilepsy Currents</i> , 2015 , 15, 192-6	1.3	31
240	Genetics of febrile seizure subtypes and syndromes: a twin study. <i>Epilepsy Research</i> , 2013 , 105, 103-9	3	31
239	Profiles of psychosocial outcome after epilepsy surgery: the role of personality. <i>Epilepsia</i> , 2010 , 51, 1133-8	6.4	31
238	The Epilepsy Genetic Association Database (epiGAD): analysis of 165 genetic association studies, 1996-2008. <i>Epilepsia</i> , 2010 , 51, 686-9	6.4	31
237	Epilepsy genes and the genetics of epilepsy syndromes: the promise of new therapies based on genetic knowledge. <i>Epilepsia</i> , 1997 , 38 Suppl 9, S32-6	6.4	31
236	Ictal SPECT and interictal PET in the localization of occipital lobe epilepsy. <i>Epilepsia</i> , 2000 , 41, 463-6	6.4	31
235	The Genetics of Epilepsy. <i>Annual Review of Genomics and Human Genetics</i> , 2020 , 21, 205-230	9.7	30
234	Genetic literacy series: genetic epilepsy with febrile seizures plus. <i>Epileptic Disorders</i> , 2018 , 20, 232-238	1.9	30
233	A GABAA receptor mutation causing generalized epilepsy reduces benzodiazepine receptor binding. <i>NeuroImage</i> , 2006 , 32, 995-1000	7.9	30
232	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. <i>American Journal of Human Genetics</i> , 2017 , 101, 516-524	11	29
231	Loss of synaptic Zn ²⁺ transporter function increases risk of febrile seizures. <i>Scientific Reports</i> , 2015 , 5, 17816	4.9	29
230	New hyperekplexia mutations provide insight into glycine receptor assembly, trafficking, and activation mechanisms. <i>Journal of Biological Chemistry</i> , 2013 , 288, 33745-33759	5.4	29
229	Can changes in cortical excitability distinguish progressive from juvenile myoclonic epilepsy?. <i>Epilepsia</i> , 2010 , 51, 2084-8	6.4	29

228	Direct and indirect measures of verbal relational memory following anterior temporal lobectomy. <i>Neuropsychologia</i> , 2002 , 40, 302-16	3.2	29
227	A genome-wide association study and biological pathway analysis of epilepsy prognosis in a prospective cohort of newly treated epilepsy. <i>Human Molecular Genetics</i> , 2014 , 23, 247-58	5.6	28
226	Genome-wide linkage meta-analysis identifies susceptibility loci at 2q34 and 13q31.3 for genetic generalized epilepsies. <i>Epilepsia</i> , 2012 , 53, 308-18	6.4	28
225	Electroclinical spectrum of the neuronal ceroid lipofuscinoses associated with CLN6 mutations. <i>Neurology</i> , 2015 , 85, 316-24	6.5	27
224	Consistency of Long-Term Subdural Electrooculography in Humans. <i>IEEE Transactions on Biomedical Engineering</i> , 2018 , 65, 344-352	5	27
223	Concepts and controversies of juvenile myoclonic epilepsy: still an enigmatic epilepsy. <i>Expert Review of Neurotherapeutics</i> , 2014 , 14, 819-31	4.3	27
222	A Primate-Specific Isoform of PLEKHG6 Regulates Neurogenesis and Neuronal Migration. <i>Cell Reports</i> , 2018 , 25, 2729-2741.e6	10.6	27
221	Second-hit DEPDC5 mutation is limited to dysmorphic neurons in cortical dysplasia type IIA. <i>Annals of Clinical and Translational Neurology</i> , 2019 , 6, 1338-1344	5.3	26
220	TBC1D24 mutation associated with focal epilepsy, cognitive impairment and a distinctive cerebro-cerebellar malformation. <i>Epilepsy Research</i> , 2013 , 105, 240-4	3	26
219	Genetics of human partial epilepsy. <i>Current Opinion in Neurology</i> , 1997 , 10, 110-4	7.1	26
218	Peritrigonal and temporo-occipital heterotopia with corpus callosum and cerebellar dysgenesis. <i>Neurology</i> , 2012 , 79, 1244-51	6.5	25
217	Mutation of SCARB2 in a patient with progressive myoclonus epilepsy and demyelinating peripheral neuropathy. <i>Archives of Neurology</i> , 2011 , 68, 812-3		25
216	Familial adult myoclonic epilepsy: recognition of mild phenotypes and refinement of the 2q locus. <i>Archives of Neurology</i> , 2012 , 69, 474-81		25
215	Personality development in the context of intractable epilepsy. <i>Archives of Neurology</i> , 2009 , 66, 68-72		25
214	Familial neonatal seizures with intellectual disability caused by a microduplication of chromosome 2q24.3. <i>Epilepsia</i> , 2010 , 51, 1865-9	6.4	25
213	Deaths due to brain injury among footballers in Victoria, 1968-1999. <i>Medical Journal of Australia</i> , 2000 , 172, 217-9	4	25
212	Occurrence of hippocampal sclerosis: is one hemisphere or gender more vulnerable?. <i>Epilepsia</i> , 1999 , 40, 1816-20	6.4	25
211	Kufs disease: clinical features and forms. <i>American Journal of Medical Genetics Part A</i> , 1988 , 5, 105-9		25

210	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. <i>Epilepsia</i> , 2019 , 60, 797-806	6.4	24
209	Somatic mutation: The hidden genetics of brain malformations and focal epilepsies. <i>Epilepsy Research</i> , 2019 , 155, 106161	3	24
208	ExACTly zero or once: A clinically helpful guide to assessing genetic variants in mild epilepsies. <i>Neurology: Genetics</i> , 2017 , 3, e163	3.8	24
207	Absence of mutations in the LGI1 receptor ADAM22 gene in autosomal dominant lateral temporal epilepsy. <i>Epilepsy Research</i> , 2007 , 76, 41-8	3	24
206	Components of verbal learning and hippocampal damage assessed by T2 relaxometry. <i>Journal of the International Neuropsychological Society</i> , 2000 , 6, 529-38	3.1	24
205	Does naming contribute to memory self-report in temporal lobe epilepsy?. <i>Journal of Clinical and Experimental Neuropsychology</i> , 1996 , 18, 98-109	2.1	24
204	Hippocampal malrotation is an anatomic variant and has no clinical significance in MRI-negative temporal lobe epilepsy. <i>Epilepsia</i> , 2016 , 57, 1719-1728	6.4	23
203	Glucose metabolism transporters and epilepsy: only GLUT1 has an established role. <i>Epilepsia</i> , 2014 , 55, e18-21	6.4	23
202	Poppy tea and the baker's first seizure. <i>Lancet, The</i> , 1997 , 350, 716	4.0	23
201	Impaired verbal associative learning after resection of left perirhinal cortex. <i>Brain</i> , 2007 , 130, 1423-31	11.2	23
200	Glioneuronal tumours in neurofibromatosis type 1: MRI-pathological study. <i>Journal of Clinical Neuroscience</i> , 2004 , 11, 745-7	2.2	23
199	Multiplex families with epilepsy: Success of clinical and molecular genetic characterization. <i>Neurology</i> , 2016 , 86, 713-22	6.5	22
198	Harnessing gene expression networks to prioritize candidate epileptic encephalopathy genes. <i>PLoS ONE</i> , 2014 , 9, e102079	3.7	22
197	Is focal cortical dysplasia sporadic? Family evidence for genetic susceptibility. <i>Epilepsia</i> , 2014 , 55, e22-6	6.4	22
196	Subtle microscopic abnormalities in hippocampal sclerosis do not predict clinical features of temporal lobe epilepsy. <i>Epilepsia</i> , 2004 , 45, 940-7	6.4	22
195	Transcranial magnetic stimulation and epilepsy. <i>Journal of Clinical Neurophysiology</i> , 2002 , 19, 294-306	2.2	22
194	The Ramsay Hunt syndrome is no longer a useful diagnostic category. <i>Movement Disorders</i> , 1989 , 4, 13-77		22
193	De novo SCN1A pathogenic variants in the GEFS+ spectrum: Not always a familial syndrome. <i>Epilepsia</i> , 2017 , 58, e26-e30	6.4	21

192	Gene expression analysis in absence epilepsy using a monozygotic twin design. <i>Epilepsia</i> , 2008 , 49, 1546-54	6.4	21
191	Hippocampal sclerosis: MR prediction of seizure intractability. <i>Epilepsia</i> , 2007 , 48, 315-23	6.4	21
190	Mild adolescent/adult onset epilepsy and paroxysmal exercise-induced dyskinesia due to GLUT1 deficiency. <i>Epilepsia</i> , 2010 , 51, 2466-9	6.4	20
189	Tramadol and new-onset seizures. <i>Medical Journal of Australia</i> , 2005 , 182, 42-3	4	20
188	Gain-of-function HCN2 variants in genetic epilepsy. <i>Human Mutation</i> , 2018 , 39, 202-209	4.7	19
187	Diagnosis and misdiagnosis of adult neuronal ceroid lipofuscinosis (Kufs disease). <i>Neurology</i> , 2016 , 87, 579-84	6.5	19
186	PRIMA1 mutation: a new cause of nocturnal frontal lobe epilepsy. <i>Annals of Clinical and Translational Neurology</i> , 2015 , 2, 821-30	5.3	19
185	Chronic encephalitis (Rasmussen's syndrome) and ipsilateral uveitis. <i>Annals of Neurology</i> , 1992 , 32, 826-9.	4	19
184	The Newfoundland aggregate of neuronal ceroid-lipofuscinosis. <i>American Journal of Medical Genetics Part A</i> , 1988 , 5, 111-6		19
183	Evaluation of non-coding variation in GLUT1 deficiency. <i>Developmental Medicine and Child Neurology</i> , 2016 , 58, 1295-1302	3.3	19
182	Exome sequencing-based molecular autopsy of formalin-fixed paraffin-embedded tissue after sudden death. <i>Genetics in Medicine</i> , 2017 , 19, 1127-1133	8.1	18
181	A case-control collapsing analysis identifies epilepsy genes implicated in trio sequencing studies focused on de novo mutations. <i>PLoS Genetics</i> , 2017 , 13, e1007104	6	18
180	Somatic mutation in the of Sturge-Weber syndrome. <i>Neurology: Genetics</i> , 2018 , 4, e236	3.8	18
179	Familial focal epilepsy with variable foci mapped to chromosome 22q12: expansion of the phenotypic spectrum. <i>Epilepsia</i> , 2012 , 53, e151-5	6.4	18
178	Mutations in TNK2 in severe autosomal recessive infantile onset epilepsy. <i>Annals of Neurology</i> , 2013 , 74, 496-501	9.4	18
177	Does a SCN1A gene mutation confer earlier age of onset of febrile seizures in GEFS+?. <i>Epilepsia</i> , 2009 , 50, 953-6	6.4	17
176	Proconvulsant-induced seizures in alpha(4) nicotinic acetylcholine receptor subunit knockout mice. <i>Neuropharmacology</i> , 2002 , 43, 55-64	5.5	17
175	Reorganization of verbal memory and language: a case of dissociation. <i>Journal of the International Neuropsychological Society</i> , 1999 , 5, 69-74	3.1	17

174	P3 latency jitter assessed using 2 techniques. I. Simulated data and surface recordings in normal subjects. <i>Electroencephalography and Clinical Neurophysiology - Evoked Potentials</i> , 1994 , 92, 352-64		17
173	Functional respiratory chain studies in subjects with chronic progressive external ophthalmoplegia and large heteroplasmic mitochondrial DNA deletions. <i>Journal of the Neurological Sciences</i> , 1991 , 102, 92-9	3.2	17
172	Cerebrospinal fluid liquid biopsy for detecting somatic mosaicism in brain. <i>Brain Communications</i> , 2021 , 3, fcaa235	4.5	17
171	ILAE-Klassifikation der Epilepsien: Positionspapier der ILAE-Kommission für Klassifikation und Terminologie. <i>Zeitschrift Für Epileptologie</i> , 2018 , 31, 296-306	0.1	17
170	Genetic analysis of PHOX2B in sudden unexpected death in epilepsy cases. <i>Neurology</i> , 2014 , 83, 1018-216.5		16
169	Deletions of 16p11.2 and 19p13.2 in a family with intellectual disability and generalized epilepsy. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 1722-5	2.5	16
168	Low blood glucose precipitates spike-and-wave activity in genetically predisposed animals. <i>Epilepsia</i> , 2011 , 52, 115-20	6.4	16
167	Copy number variants--an unexpected risk factor for the idiopathic generalized epilepsies. <i>Brain</i> , 2010 , 133, 7-8	11.2	16
166	AUStralian study of titration to effect profile of safety (AUS-STEPS): high-dose gabapentin (neurontin) in partial seizures. <i>Epilepsia</i> , 2001 , 42, 1335-9	6.4	16
165	Genetics of the epilepsies. <i>Current Opinion in Pediatrics</i> , 2000 , 12, 536-42	3.2	16
164	A twin study of genetic influences on epilepsy outcome. <i>Twin Research and Human Genetics</i> , 2003 , 6, 140-6		16
163	Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2-2q11.2. <i>Human Genetics</i> , 2016 , 135, 1117-25	6.3	16
162	Metabolic patterns and seizure outcomes following anterior temporal lobectomy. <i>Annals of Neurology</i> , 2019 , 85, 241-250	9.4	16
161	Frequency of CNKSR2 mutation in the X-linked epilepsy-aphasia spectrum. <i>Epilepsia</i> , 2017 , 58, e40-e43	6.4	15
160	GABA-mediated tonic inhibition differentially modulates gain in functional subtypes of cortical interneurons. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020 , 117, 3192-3202	11.5	15
159	Seizure semiology in autosomal dominant epilepsy with auditory features, due to novel LGI1 mutations. <i>Epilepsy Research</i> , 2013 , 107, 311-7	3	15
158	Synaptic Zn and febrile seizure susceptibility. <i>British Journal of Pharmacology</i> , 2017 , 174, 119-125	8.6	15
157	Autosomal dominant vasovagal syncope: clinical features and linkage to chromosome 15q26. <i>Neurology</i> , 2013 , 80, 1485-93	6.5	15

156	The Role of Seizure-Related SEZ6 as a Susceptibility Gene in Febrile Seizures. <i>Neurology Research International</i> , 2011 , 2011, 917565	1.7	15
155	Founder effect with variable age at onset in Arab families with Lafora disease and EPM2A mutation. <i>Epilepsia</i> , 2007 , 48, 1011-4	6.4	15
154	Genetics of the epilepsies. <i>Current Opinion in Neurology</i> , 1999 , 12, 177-82	7.1	15
153	In silico prioritization based on coexpression can aid epileptic encephalopathy gene discovery. <i>Neurology: Genetics</i> , 2016 , 2, e51	3.8	15
152	Cortical microarchitecture changes in genetic epilepsy. <i>Neurology</i> , 2015 , 84, 1308-16	6.5	14
151	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17 458 subjects. <i>Brain</i> , 2020 , 143, 2106-2118	11.2	14
150	Genetics of vasovagal syncope. <i>Autonomic Neuroscience: Basic and Clinical</i> , 2014 , 184, 60-5	2.4	14
149	In vivo loss of slow potassium channel activity in individuals with benign familial neonatal epilepsy in remission. <i>Brain</i> , 2012 , 135, 3144-52	11.2	14
148	A retrospective population-based study on seizures related to childhood vaccination. <i>Epilepsia</i> , 2011 , 52, 1506-12	6.4	14
147	Kufs disease due to mutation of CLN6: clinical, pathological and molecular genetic features. <i>Brain</i> , 2019 , 142, 59-69	11.2	14
146	Rasmussen encephalitis and comorbid autoimmune diseases: A window into disease mechanism?. <i>Neurology</i> , 2014 , 83, 1049-55	6.5	13
145	Atypical multifocal Dravet syndrome lacks generalized seizures and may show later cognitive decline. <i>Developmental Medicine and Child Neurology</i> , 2014 , 56, 85-90	3.3	13
144	Evidence for genetic factors in vasovagal syncope: a twin-family study. <i>Neurology</i> , 2012 , 79, 561-5	6.5	13
143	Is variation in the GABA(B) receptor 1 gene associated with temporal lobe epilepsy?. <i>Epilepsia</i> , 2005 , 46, 778-80	6.4	13
142	Benign partial seizures of adolescence. <i>Epilepsia</i> , 1999 , 40, 1244-7	6.4	13
141	Periventricular Nodular Heterotopia: Detection of Abnormal Microanatomic Fiber Structures with Whole-Brain Diffusion MR Imaging Tractography. <i>Radiology</i> , 2016 , 281, 896-906	20.5	13
140	Seizures as presenting and prominent symptom in chorea-acanthocytosis with c.2343del VPS13A gene mutation. <i>Epilepsia</i> , 2016 , 57, 549-56	6.4	13
139	Encephalopathies with KCNC1 variants: genotype-phenotype-functional correlations. <i>Annals of Clinical and Translational Neurology</i> , 2019 , 6, 1263-1272	5.3	12

138	Nomenclature of Genetically Determined Myoclonus Syndromes: Recommendations of the International Parkinson and Movement Disorder Society Task Force. <i>Movement Disorders</i> , 2019 , 34, 1602-1613	7	12
137	Familial mesial temporal lobe epilepsy and the borderland of djuvu. <i>Annals of Neurology</i> , 2017 , 82, 166-176	3.4	12
136	A case of severe hearing loss in action myoclonus renal failure syndrome resulting from mutation in SCARB2. <i>Movement Disorders</i> , 2012 , 27, 1200-1	7	12
135	Detection of microchromosomal aberrations in refractory epilepsy: a pilot study. <i>Epileptic Disorders</i> , 2010 , 12, 192-8	1.9	12
134	Neuropsychological function in patients with a single gene mutation associated with autosomal dominant nocturnal frontal lobe epilepsy. <i>Epilepsy and Behavior</i> , 2010 , 17, 531-5	3.2	12
133	A duplication in 1q21.3 in a family with early onset and childhood absence epilepsy. <i>Epilepsia</i> , 2010 , 51, 2453-6	6.4	12
132	Developmental genetics of deleted mtDNA in mitochondrial oculomyopathy. <i>Journal of the Neurological Sciences</i> , 1997 , 145, 155-62	3.2	12
131	Dementia and myoclonus: differential diagnosis of early-onset Alzheimer's disease. <i>Annals of Neurology</i> , 1995 , 37, 412	9.4	12
130	The influence of changes in the intensity of magnetic stimulation on coil output. <i>Muscle and Nerve</i> , 1993 , 16, 1338-41	3.4	12
129	Evidence for type-specific DNA methylation patterns in epilepsy: a discordant monozygotic twin approach. <i>Epigenomics</i> , 2019 , 11, 951-968	4.4	11
128	Inter-session repeatability of cortical excitability measurements in patients with epilepsy. <i>Epilepsy Research</i> , 2012 , 98, 182-6	3	11
127	Lysosomal integral membrane protein type-2 (LIMP-2/SCARB2) is a substrate of cathepsin-F, a cysteine protease mutated in type-B-Kufs-disease. <i>Biochemical and Biophysical Research Communications</i> , 2015 , 457, 334-40	3.4	11
126	Mutations in PRRT2 are not a common cause of infantile epileptic encephalopathies. <i>Epilepsia</i> , 2013 , 54, e86-9	6.4	11
125	Adjunctive therapy of uncontrolled partial seizures with levetiracetam in Australian patients. <i>Epilepsy and Behavior</i> , 2007 , 11, 338-42	3.2	11
124	Treatment of new-onset epilepsy: seizures beget discussion. <i>Lancet, The</i> , 2005 , 365, 1985-6	4.0	11
123	Idiopathic generalized epilepsies: do sporadic and familial cases differ?. <i>Epilepsia</i> , 2001 , 42, 1399-402	6.4	11
122	Weight and fat distribution in patients taking valproate: a valproate-discordant gender-matched twin and sibling pair study. <i>Epilepsia</i> , 2014 , 55, 1551-7	6.4	10
121	Rare protein sequence variation in SV2A gene does not affect response to levetiracetam. <i>Epilepsy Research</i> , 2012 , 101, 277-9	3	10

120	New therapeutic opportunities in epilepsy: a genetic perspective. <i>Pharmacology & Therapeutics</i> , 2010 , 128, 274-80	13.9	10
119	Locus for febrile seizures. <i>Annals of Neurology</i> , 2000 , 47, 840-1	9.4	10
118	Progressive myoclonus epilepsies-Residual unsolved cases have marked genetic heterogeneity including dolichol-dependent protein glycosylation pathway genes. <i>American Journal of Human Genetics</i> , 2021 , 108, 722-738	11	10
117	Treatment with anti-epileptic drugs. <i>Australian Family Physician</i> , 2005 , 34, 1017-20		10
116	Familial epilepsy with anterior polymicrogyria as a presentation of COL18A1 mutations. <i>European Journal of Medical Genetics</i> , 2017 , 60, 437-443	2.6	9
115	Familial adult myoclonic epilepsy type 1 SAMD12 TTTCA repeat expansion arose 17,000 years ago and is present in Sri Lankan and Indian families. <i>European Journal of Human Genetics</i> , 2020 , 28, 973-978	5.3	9
114	'Idiopathic' no more! Abnormal interaction of large-scale brain networks in generalized epilepsy. <i>Brain</i> , 2014 , 137, 2400-2	11.2	9
113	Do mutations in SCN1B cause Dravet syndrome?. <i>Epilepsy Research</i> , 2013 , 103, 97-100	3	9
112	Benign neonatal sleep myoclonus: an autosomal dominant form not allelic to KCNQ2 or KCNQ3. <i>Journal of Child Neurology</i> , 2012 , 27, 1260-3	2.5	9
111	The borderland of epilepsy: a clinical and molecular view, 100 years on. <i>Epilepsia</i> , 2010 , 51 Suppl 1, 3-4	6.4	9
110	What happens now? Ongoing outcome after post-temporal lobectomy seizure recurrence. <i>Neurology</i> , 2006 , 67, 1671-3	6.5	9
109	GEFS+ where focal seizures evolve from generalized spike wave: video-EEG study of two children. <i>Epileptic Disorders</i> , 2007 , 9, 307-14	1.9	9
108	A case series of lacosamide as adjunctive therapy in refractory sleep-related hypermotor epilepsy (previously nocturnal frontal lobe epilepsy). <i>Journal of Sleep Research</i> , 2018 , 27, e12669	5.8	8
107	Febrile infection-related epilepsy syndrome is not caused by SCN1A mutations. <i>Epilepsy Research</i> , 2012 , 100, 194-8	3	8
106	Investigation of the 15q13.3 CNV as a genetic modifier for familial epilepsies with variable phenotypes. <i>Epilepsia</i> , 2011 , 52, e139-42	6.4	8
105	Familial Lennox-Gastaut syndrome in male siblings with a novel DCX mutation and anterior pachygyria. <i>Epilepsia</i> , 2010 , 51, 1902-5	6.4	8
104	Is photosensitive epilepsy less common in males due to variation in X chromosome photopigment genes?. <i>Epilepsia</i> , 2007 , 48, 1807-1809	6.4	8
103	Prolactin levels in sudden unexpected death in epilepsy. <i>Epilepsia</i> , 2000 , 41, 48-51	6.4	8

102	EXOME REPORT: Novel mutation in ATP6V1B2 segregating with autosomal dominant epilepsy, intellectual disability and mild gingival and nail abnormalities. <i>European Journal of Medical Genetics</i> , 2020 , 63, 103799	2.6	8
101	Myoclonic occipital photosensitive epilepsy with dystonia (MOPED): A familial epilepsy syndrome. <i>Epilepsy Research</i> , 2015 , 114, 98-105	3	7
100	Teenage-onset progressive myoclonic epilepsy due to a familial repeat expansion. <i>Neurology</i> , 2018 , 90, e658-e663	6.5	7
99	Abnormal Processing of Autophagosomes in Transformed B Lymphocytes from SCARB2-Deficient Subjects. <i>BioResearch Open Access</i> , 2013 , 2, 40-6	2.4	7
98	Overcoming barriers to successful epilepsy management. <i>Epilepsy Currents</i> , 2012 , 12, 158-60	1.3	7
97	Reduced variance in monozygous twins for multiple MR parameters: implications for disease studies and the genetic basis of brain structure. <i>NeuroImage</i> , 2010 , 49, 1536-44	7.9	7
96	SCN1A Variants in vaccine-related febrile seizures: A prospective study. <i>Annals of Neurology</i> , 2020 , 87, 281-288	9.4	7
95	Cation leak underlies neuronal excitability in an HCN1 developmental and epileptic encephalopathy. <i>Brain</i> , 2021 , 144, 2060-2073	11.2	7
94	Progressive myoclonus epilepsy associated with SACS gene mutations. <i>Neurology: Genetics</i> , 2016 , 2, e833.8	3.8	7
93	The severe epilepsy syndromes of infancy: A population-based study. <i>Epilepsia</i> , 2021 , 62, 358-370	6.4	7
92	Can mutation-mediated effects occurring early in development cause long-term seizure susceptibility in genetic generalized epilepsies?. <i>Epilepsia</i> , 2018 , 59, 915-922	6.4	6
91	Sensitive quantitative detection of somatic mosaic mutation in "double cortex" syndrome. <i>Epileptic Disorders</i> , 2017 , 19, 450-455	1.9	6
90	Does variation in NIPA2 contribute to genetic generalized epilepsy?. <i>Human Genetics</i> , 2014 , 133, 673-4	6.3	6
89	Association of a nicotinic receptor mutation with reduced height and blunted physostigmine-stimulated growth hormone release. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2008 , 93, 634-7	5.6	6
88	Febrile seizures: traffic slows in the heat. <i>Trends in Molecular Medicine</i> , 2006 , 12, 343-4	11.5	6
87	New autosomal-dominant partial epilepsy syndrome. <i>Pediatric Neurology</i> , 1994 , 11, 95	2.9	6
86	Rey figure distortions reflect nonverbal recall differences between right and left foci in unilateral temporal lobe epilepsy. <i>Archives of Clinical Neuropsychology</i> , 1994 , 9, 451-460	2.7	6
85	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. <i>American Journal of Human Genetics</i> , 2021 , 108, 965-982	11	6

84	Is FGF13 a major contributor to genetic epilepsy with febrile seizures plus?. <i>Epilepsy Research</i> , 2016 , 128, 48-51	3	6
83	Splice variant in ARX leading to loss of C-terminal region in a boy with intellectual disability and infantile onset developmental and epileptic encephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 1483-1490	2.5	5
82	Epilepsy in families: Age at onset is a familial trait, independent of syndrome. <i>Annals of Neurology</i> , 2019 , 86, 91-98	9.4	5
81	Antiepileptic Drug Teratogenicity and De Novo Genetic Variation Load. <i>Annals of Neurology</i> , 2020 , 87, 897-906	9.4	5
80	Inherited RORB pathogenic variants: Overlap of photosensitive genetic generalized and occipital lobe epilepsy. <i>Epilepsia</i> , 2020 , 61, e23-e29	6.4	5
79	Clinical features of seizures associated with parahippocampal/inferior temporal lesions compared to those with hippocampal sclerosis. <i>Epilepsia</i> , 2010 , 51, 1906-9	6.4	5
78	Prediction of drug resistance in epilepsy: not as easy as ABC. <i>Lancet Neurology, The</i> , 2006 , 5, 641-2	24.1	5
77	Genetics of the Epilepsies. <i>Epilepsia</i> , 2001 , 42, 16-23	6.4	5
76	Anterior temporal encephaloceles: Elusive, important, and rewarding to treat. <i>Epilepsia</i> , 2020 , 61, 2675-2684	6.4	5
75	Contribution of rare genetic variants to drug response in absence epilepsy. <i>Epilepsy Research</i> , 2021 , 170, 106537	3	5
74	Development of a rapid functional assay that predicts GLUT1 disease severity. <i>Neurology: Genetics</i> , 2018 , 4, e297	3.8	5
73	Multifocal epilepsy: the role of palliative resection - intractable frontal and occipital lobe epilepsy secondary to radiotherapy for acute lymphoblastic leukaemia. <i>Epileptic Disorders</i> , 2008 , 10, 362-70	1.9	5
72	SCN1A clinical spectrum includes the self-limited focal epilepsies of childhood. <i>Epilepsy Research</i> , 2017 , 131, 9-14	3	4
71	Evaluation of GLUT1 variation in non-acquired focal epilepsy. <i>Epilepsy Research</i> , 2017 , 133, 54-57	3	4
70	Epidemiology and etiology of infantile developmental and epileptic encephalopathies in Tasmania. <i>Epilepsia Open</i> , 2019 , 4, 504-510	4	4
69	No evidence for a BRD2 promoter hypermethylation in blood leukocytes of Europeans with juvenile myoclonic epilepsy. <i>Epilepsia</i> , 2019 , 60, e31-e36	6.4	4
68	Siblings with refractory occipital epilepsy showing localized network activity on EEG-fMRI. <i>Epilepsia</i> , 2013 , 54, e28-32	6.4	4
67	Epilepsy in offspring of whom both parents have idiopathic generalized epilepsy: biparental inheritance. <i>Epilepsia</i> , 2003 , 44, 1250-4	6.4	4

66	Association of Short-term Heart Rate Variability and Sudden Unexpected Death in Epilepsy. <i>Neurology</i> , 2021 ,	6.5	4
65	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. <i>Genetics in Medicine</i> , 2021 , 23, 363-373	8.1	4
64	The clinical utility of exome sequencing and extended bioinformatic analyses in adolescents and adults with a broad range of neurological phenotypes: an Australian perspective. <i>Journal of the Neurological Sciences</i> , 2021 , 420, 117260	3.2	4
63	Newly diagnosed seizures assessed at two established first seizure clinics: Clinic characteristics, investigations, and findings over 11 years. <i>Epilepsia Open</i> , 2021 , 6, 171-180	4	4
62	Genome-wide association study of febrile seizures implicates fever response and neuronal excitability genes.. <i>Brain</i> , 2022 ,	11.2	4
61	Epilepsy research in 2016: new treatment directions. <i>Lancet Neurology</i> , 2017 , 16, 7-9	24.1	3
60	Evaluation of multiple putative risk alleles within the 15q13.3 region for genetic generalized epilepsy. <i>Epilepsy Research</i> , 2015 , 117, 70-3	3	3
59	KANSL1 variation is not a major contributing factor in self-limited focal epilepsy syndromes of childhood. <i>PLoS ONE</i> , 2018 , 13, e0191546	3.7	3
58	Association studies and functional validation or functional validation alone?. <i>Epilepsy Research</i> , 2007 , 74, 237-8	3	3
57	Are Variants Causing Cardiac Arrhythmia Risk Factors in Sudden Unexpected Death in Epilepsy?. <i>Frontiers in Neurology</i> , 2020 , 11, 925	4.1	3
56	Assessing the role of rare genetic variants in drug-resistant, non-lesional focal epilepsy. <i>Annals of Clinical and Translational Neurology</i> , 2021 , 8, 1376-1387	5.3	3
55	Human generalized epilepsy: Increased somatosensory and striatothalamic connectivity. <i>Neurology: Genetics</i> , 2019 , 5, e340	3.8	3
54	Association of Missense Variants With Genetic Epilepsy With Febrile Seizures Plus. <i>Neurology</i> , 2021 , 96, e2251-e2260	6.5	3
53	Deciphering the role of epigenetics in self-limited epilepsy with centrotemporal spikes. <i>Epilepsy Research</i> , 2019 , 156, 106163	3	2
52	Obstetric events as a risk factor for febrile seizures: a community-based twin study. <i>Twin Research and Human Genetics</i> , 2008 , 11, 634-40	2.2	2
51	Cerebrospinal fluid neurofilament light chain differentiates primary psychiatric disorders from rapidly progressive, Alzheimer's disease and frontotemporal disorders in clinical settings.. <i>Alzheimer's and Dementia</i> , 2022 ,	1.2	2
50	Genome-wide association study of febrile seizures identifies seven new loci implicating fever response and neuronal excitability genes		2
49	Progressive Myoclonus Epilepsies: Diagnostic Yield With Next-Generation Sequencing in Previously Unsolved Cases. <i>Neurology: Genetics</i> , 2021 , 7, e641	3.8	2

48	The "maternal effect" on epilepsy risk: Analysis of familial epilepsies and reassessment of prior evidence. <i>Annals of Neurology</i> , 2020 , 87, 132-138	9.4	2
47	Evidence of linkage to chromosome 5p13.2-q11.1 in a large inbred family with genetic generalized epilepsy. <i>Epilepsia</i> , 2018 , 59, e125-e129	6.4	2
46	Significance of post-operative auras after temporal lobectomy: a surprising methodological trap. <i>Epilepsy and Behavior</i> , 2012 , 23, 348-52	3.2	1
45	Vaccination and Dravet syndrome [Authors' reply]. <i>Lancet Neurology, The</i> , 2010 , 9, 1148-1149	24.1	1
44	The idiopathic generalized epilepsies across life. <i>Supplements To Clinical Neurophysiology</i> , 2004 , 57, 408-14		1
43	Acetylation of histones in isolated avian erythroid nuclei. <i>Nucleic Acids and Protein Synthesis</i> , 1977 , 475, 160-7		1
42	Evidence for a Dual-Pathway, 2-Hit Genetic Model for Focal Cortical Dysplasia and Epilepsy.. <i>Neurology: Genetics</i> , 2022 , 8, e652	3.8	1
41	Bi-allelic SMO variants in hypothalamic hamartoma: a recessive cause of Pallister-Hall syndrome.. <i>European Journal of Human Genetics</i> , 2022 ,	5.3	1
40	Plasma neurofilament light chain protein is not increased in treatment-resistant schizophrenia and first-degree relatives.. <i>Australian and New Zealand Journal of Psychiatry</i> , 2021 , 48674211058684	2.6	1
39	UNC13B and focal epilepsy.. <i>Brain</i> , 2022 ,	11.2	1
38	Generalized, focal, and combined epilepsies in families: New evidence for distinct genetic factors. <i>Epilepsia</i> , 2020 , 61, 2667-2674	6.4	1
37	Pathogenic MAST3 Variants in the STK Domain Are Associated with Epilepsy. <i>Annals of Neurology</i> , 2021 , 90, 274-284	9.4	1
36	Association Between Psychiatric Comorbidities and Mortality in Epilepsy. <i>Neurology: Clinical Practice</i> , 2021 , 11, 429-437	1.7	1
35	Epileptic encephalopathies of infancy: welcome advances. <i>Lancet, The</i> , 2019 , 394, 2203-2204	40	1
34	Progressive Myoclonus Epilepsy Caused by a Homozygous Splicing Variant of SLC7A6OS. <i>Annals of Neurology</i> , 2021 , 89, 402-407	9.4	1
33	Transcriptome analysis of a ring chromosome 20 patient cohort. <i>Epilepsia</i> , 2021 , 62, e22-e28	6.4	1
32	Improving Specificity of Cerebrospinal Fluid Liquid Biopsy for Genetic Testing. <i>Annals of Neurology</i> , 2021 , 90, 693-694	9.4	1
31	Postictal Psychosis in Epilepsy: A Clinicogenetic Study. <i>Annals of Neurology</i> , 2021 , 90, 464-476	9.4	1

30	Common risk variants for epilepsy are enriched in families previously targeted for rare monogenic variant discovery. <i>EBioMedicine</i> , 2022 , 81, 104079	8.8	1
29	Epilepsy rounds. <i>Journal of Epilepsy</i> , 1997 , 10, 49-51		0
28	Genetic Epilepsies 2007 , 371-383		0
27	Diagnostic delay in focal epilepsy: Association with brain pathology and age.. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2022 , 96, 121-127	3.2	0
26	What is the motor vehicle crash risk for drivers with epilepsy? A systematic review. <i>Journal of Transport and Health</i> , 2021 , 23, 101286	3	0
25	Variants in cause progressive myoclonus epilepsy and developmental and epileptic encephalopathy.. <i>Brain Communications</i> , 2021 , 3, Fcab245	4.5	0
24	State transitions through inhibitory interneurons in a cortical network model. <i>PLoS Computational Biology</i> , 2021 , 17, e1009521	5	0
23	Hypothalamic Hamartomas: Evolving Understanding and Management. <i>Neurology</i> , 2021 , 97, 864-873	6.5	0
22	Novel Missense Mutations Associated with Infantile-Onset Developmental and Epileptic Encephalopathy. <i>International Journal of Molecular Sciences</i> , 2020 , 21,	6.3	0
21	Loss-of-function variants in K 11.1 cardiac channels as a biomarker for SUDEP. <i>Annals of Clinical and Translational Neurology</i> , 2021 , 8, 1422-1432	5.3	0
20	Epilepsy risk in offspring of affected parents; a cohort study of the "maternal effect" in epilepsy. <i>Annals of Clinical and Translational Neurology</i> , 2021 , 8, 153-162	5.3	0
19	Cutting edge approaches to detecting brain mosaicism associated with common focal epilepsies: implications for diagnosis and potential therapies. <i>Expert Review of Neurotherapeutics</i> , 2021 , 21, 1309-1318	4.3	0
18	Interictal EEG and ECG for SUDEP Risk Assessment: A Retrospective Multicenter Cohort Study.. <i>Frontiers in Neurology</i> , 2022 , 13, 858333	4.1	0
17	Neurofilament light chain in psychiatric and neurodegenerative disorders: A β -reactive protein for the brain?. <i>Alzheimer's and Dementia</i> , 2020 , 16, e041347	1.2	
16	Predominantly nocturnal seizures post temporal lobectomy: Characteristics of an unusual outcome group. <i>Epilepsy Research</i> , 2019 , 155, 106154	3	
15	Quantitative analysis of phenotypic elements augments traditional electroclinical classification of common familial epilepsies. <i>Epilepsia</i> , 2019 , 60, 2194-2203	6.4	
14	Phenotype-genotype complexities: opening DOORS. <i>Lancet Neurology</i> , 2014 , 13, 24-5	24.1	
13	NOCTURNAL SEIZURES ONLY POST TEMPORAL LOBECTOMY: CHARACTERISTICS OF AN UNUSUAL OUTCOME GROUP. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2013 , 84, e2.5-e2	5.5	

12	Key epilepsy gene gets further phenotypic delineation. <i>Neurology</i> , 2010 , 75, 18-9	6.5
11	Epilepsy: insights into causes and treatment dilemmas. <i>Lancet Neurology, The</i> , 2010 , 9, 9-11	24.1
10	Adults with Epilepsy: Is Monotherapy the Only Answer?. <i>Epilepsia</i> , 1997 , 38, S9-S12	6.4
9	Epileptology of the first-seizure presentation. <i>Lancet, The</i> , 1998 , 352, 1856	40
8	Response to Tinuper et al.. <i>Epilepsia</i> , 2007 , 48, 1034-1034	6.4
7	Scale for Distinguishing Sleep Disorders From SeizuresReply. <i>Archives of Neurology</i> , 2007 , 64, 1206	
6	Invited comments on the Shostak and Ottman review. <i>Epilepsia</i> , 2006 , 47, 1751-2; author reply 1755-6	6.4
5	Sleep Neurology - A Wakeup Call for Neurologists. <i>Practical Neurology</i> , 2002 , 2, 2-3	2.4
4	Neurological disorders. <i>Medical Journal of Australia</i> , 2000 , 172, 393	4
3	Nocturnal frontal lobe epilepsy 2001 , 97-110	
2	HLA-DR2 negative narcolepsy in Australian Caucasians: clinical features, serology and sequence specific oligonucleotide typing. <i>Journal of the Neurological Sciences</i> , 1992 , 113, 26-30	3.2
1	Integrated in silico and experimental assessment of disease relevance of PCDH19 missense variants. <i>Human Mutation</i> , 2021 , 42, 1030-1041	4.7