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List of Publications by Citations

529 papers	46,823 citations	106 h-index	205 g-index
586 ext. papers	56,500 ext. citations	8.4 avg, IF	7.35 L-index

#	Paper	IF	Citations
529	Revised terminology and concepts for organization of seizures and epilepsies: report of the ILAE Commission on Classification and Terminology, 2005-2009. *Epilepsia*, **2010**, 51, 676-85	6.4	2950
528	ILAE official report: a practical clinical definition of epilepsy. *Epilepsia*, **2014**, 55, 475-82	6.4	2408
527	ILAE classification of the epilepsies: Position paper of the ILAE Commission for Classification and Terminology. *Epilepsia*, **2017**, 58, 512-521	6.4	2084
526	Operational classification of seizure types by the International League Against Epilepsy: Position Paper of the ILAE Commission for Classification and Terminology. *Epilepsia*, **2017**, 58, 522-530	6.4	1340
525	De novo mutations in epileptic encephalopathies. *Nature*, **2013**, 501, 217-21	50.4	1081
524	A definition and classification of status epilepticus--Report of the ILAE Task Force on Classification of Status Epilepticus. *Epilepsia*, **2015**, 56, 1515-23	6.4	1036
523	A missense mutation in the neuronal nicotinic acetylcholine receptor alpha 4 subunit is associated with autosomal dominant nocturnal frontal lobe epilepsy. *Nature Genetics*, **1995**, 11, 201-3	36.3	922
522	Doublecortin, a brain-specific gene mutated in human X-linked lissencephaly and double cortex syndrome, encodes a putative signaling protein. *Cell*, **1998**, 92, 63-72	56.2	904
521	Febrile seizures and generalized epilepsy associated with a mutation in the Na+-channel beta1 subunit gene SCN1B. *Nature Genetics*, **1998**, 19, 366-70	36.3	851
520	Trial of Cannabidiol for Drug-Resistant Seizures in the Dravet Syndrome. *New England Journal of Medicine*, **2017**, 376, 2011-2020	59.2	771
519	Mutations in filamin 1 prevent migration of cerebral cortical neurons in human periventricular heterotopia. *Neuron*, **1998**, 21, 1315-25	13.9	729
518	Analysis of shared heritability in common disorders of the brain. *Science*, **2018**, 360,	33.3	666
517	Mutant GABA(A) receptor gamma2-subunit in childhood absence epilepsy and febrile seizures. *Nature Genetics*, **2001**, 28, 49-52	36.3	643
516	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. *Nature Genetics*, **2013**, 45, 825-30	36.3	500
515	Autosomal dominant nocturnal frontal lobe epilepsy. A distinctive clinical disorder. *Brain*, **1995**, 118 (Pt 1), 61-73	11.2	471
514	Refining analyses of copy number variation identifies specific genes associated with developmental delay. *Nature Genetics*, **2014**, 46, 1063-71	36.3	429
513	Instruction manual for the ILAE 2017 operational classification of seizure types. *Epilepsia*, **2017**, 58, 531-542	6.4	425

512	The spectrum of SCN1A-related infantile epileptic encephalopathies. *Brain*, **2007**, 130, 843-52	11.2	423
511	Truncation of the GABA(A)-receptor gamma2 subunit in a family with generalized epilepsy with febrile seizures plus. *American Journal of Human Genetics*, **2002**, 70, 530-6	11	373
510	Mutations in the human ortholog of Aristaless cause X-linked mental retardation and epilepsy. *Nature Genetics*, **2002**, 30, 441-5	36.3	356
509	X-linked protocadherin 19 mutations cause female-limited epilepsy and cognitive impairment. *Nature Genetics*, **2008**, 40, 776-81	36.3	328
508	KCNQ2 encephalopathy: emerging phenotype of a neonatal epileptic encephalopathy. *Annals of Neurology*, **2012**, 71, 15-25	9.4	322
507	The genetic landscape of the epileptic encephalopathies of infancy and childhood. *Lancet Neurology, The*, **2016**, 15, 304-16	24.1	317
506	De novo mutations in synaptic transmission genes including DNM1 cause epileptic encephalopathies. *American Journal of Human Genetics*, **2014**, 95, 360-70	11	299
505	Localization of a gene for autosomal dominant nocturnal frontal lobe epilepsy to chromosome 20q 13.2. *Nature Genetics*, **1995**, 10, 117-8	36.3	298
504	Sodium-channel defects in benign familial neonatal-infantile seizures. *Lancet, The*, **2002**, 360, 851-2	40	284
503	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. *Nature Genetics*, **2017**, 49, 515-526	36.3	283
502	Neuronal sodium-channel alpha1-subunit mutations in generalized epilepsy with febrile seizures plus. *American Journal of Human Genetics*, **2001**, 68, 859-65	11	283
501	SCN1A mutations and epilepsy. *Human Mutation*, **2005**, 25, 535-42	4.7	282
500	De novo mutations in ATP1A3 cause alternating hemiplegia of childhood. *Nature Genetics*, **2012**, 44, 1030-4	36.3	280
499	Epilepsy. *Nature Reviews Disease Primers*, **2018**, 4, 18024	51.1	269
498	GABRD encoding a protein for extra- or peri-synaptic GABAA receptors is a susceptibility locus for generalized epilepsies. *Human Molecular Genetics*, **2004**, 13, 1315-9	5.6	267
497	CHRNB2 is the second acetylcholine receptor subunit associated with autosomal dominant nocturnal frontal lobe epilepsy. *American Journal of Human Genetics*, **2001**, 68, 225-31	11	267
496	Somatic mutations in cerebral cortical malformations. *New England Journal of Medicine*, **2014**, 371, 733-43	59.2	265
495	Missense mutations in the sodium-gated potassium channel gene KCNT1 cause severe autosomal dominant nocturnal frontal lobe epilepsy. *Nature Genetics*, **2012**, 44, 1188-90	36.3	253

494	GRIN2A mutations cause epilepsy-aphasia spectrum disorders. *Nature Genetics*, **2013**, 45, 1073-6	36.3	249
493	De-novo mutations of the sodium channel gene SCN1A in alleged vaccine encephalopathy: a retrospective study. *Lancet Neurology, The*, **2006**, 5, 488-92	24.1	241
492	Generalized epilepsy with febrile seizures plus: a common childhood-onset genetic epilepsy syndrome. *Annals of Neurology*, **1999**, 45, 75-81	9.4	239
491	Mutations in DEPDC5 cause familial focal epilepsy with variable foci. *Nature Genetics*, **2013**, 45, 546-51	36.3	238
490	Early-onset absence epilepsy caused by mutations in the glucose transporter GLUT1. *Annals of Neurology*, **2009**, 66, 415-9	9.4	230
489	Optimal clinical management of children receiving dietary therapies for epilepsy: Updated recommendations of the International Ketogenic Diet Study Group. *Epilepsia Open*, **2018**, 3, 175-192	4	227
488	Benign familial neonatal-infantile seizures: characterization of a new sodium channelopathy. *Annals of Neurology*, **2004**, 55, 550-7	9.4	225
487	Navigating the channels and beyond: unravelling the genetics of the epilepsies. *Lancet Neurology, The*, **2008**, 7, 231-45	24.1	219
486	Phenotypic characterization of an alpha 4 neuronal nicotinic acetylcholine receptor subunit knock-out mouse. *Journal of Neuroscience*, **2000**, 20, 6431-41	6.6	217
485	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. *American Journal of Human Genetics*, **2017**, 101, 664-685	11	214
484	SRPX2 mutations in disorders of language cortex and cognition. *Human Molecular Genetics*, **2006**, 15, 1195-207	5.6	213
483	New concepts in classification of the epilepsies: entering the 21st century. *Epilepsia*, **2011**, 52, 1058-62	6.4	208
482	The phenotypic spectrum of SCN8A encephalopathy. *Neurology*, **2015**, 84, 480-9	6.5	199
481	PRRT2 mutations cause benign familial infantile epilepsy and infantile convulsions with choreoathetosis syndrome. *American Journal of Human Genetics*, **2012**, 90, 152-60	11	199
480	Autosomal dominant nocturnal frontal-lobe epilepsy: genetic heterogeneity and evidence for a second locus at 15q24. *American Journal of Human Genetics*, **1998**, 63, 1108-16	11	199
479	Genetic determinants of common epilepsies: a meta-analysis of genome-wide association studies. *Lancet Neurology, The*, **2014**, 13, 893-903	24.1	194
478	KCNT1 gain of function in 2 epilepsy phenotypes is reversed by quinidine. *Annals of Neurology*, **2014**, 75, 581-90	9.4	192
477	Familial and sporadic 15q13.3 microdeletions in idiopathic generalized epilepsy: precedent for disorders with complex inheritance. *Human Molecular Genetics*, **2009**, 18, 3626-31	5.6	190

476	Temporal lobe epilepsy and GEFS+ phenotypes associated with SCN1B mutations. *Brain*, **2007**, 130, 100-9	11.2	188
475	. *Nature Genetics*, **2001**, 28, 49-52	36.3	184
474	Definition and diagnostic criteria of sleep-related hypermotor epilepsy. *Neurology*, **2016**, 86, 1834-42	6.5	182
473	GABRA1 and STXBP1: novel genetic causes of Dravet syndrome. *Neurology*, **2014**, 82, 1245-53	6.5	180
472	De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies. *American Journal of Human Genetics*, **2016**, 99, 287-98	11	180
471	Clinical whole-genome sequencing in severe early-onset epilepsy reveals new genes and improves molecular diagnosis. *Human Molecular Genetics*, **2014**, 23, 3200-11	5.6	179
470	Distinguishing sleep disorders from seizures: diagnosing bumps in the night. *Archives of Neurology*, **2006**, 63, 705-9		178
469	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. *Nature Genetics*, **2015**, 47, 39-46	36.3	177
468	Clinical and imaging heterogeneity of polymicrogyria: a study of 328 patients. *Brain*, **2010**, 133, 1415-27	11.2	177
467	Dominant-negative effects of KCNQ2 mutations are associated with epileptic encephalopathy. *Annals of Neurology*, **2014**, 75, 382-94	9.4	176
466	Rare copy number variants are an important cause of epileptic encephalopathies. *Annals of Neurology*, **2011**, 70, 974-85	9.4	176
465	The genetics of Dravet syndrome. *Epilepsia*, **2011**, 52 Suppl 2, 24-9	6.4	170
464	Genome-wide mega-analysis identifies 16 loci and highlights diverse biological mechanisms in the common epilepsies. *Nature Communications*, **2018**, 9, 5269	17.4	169
463	SCN2A encephalopathy: A major cause of epilepsy of infancy with migrating focal seizures. *Neurology*, **2015**, 85, 958-66	6.5	163
462	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
461	Early and effective treatment of KCNQ2 encephalopathy. *Epilepsia*, **2015**, 56, 685-91	6.4	160
460	Characterization of mutations in the gene doublecortin in patients with double cortex syndrome. *Annals of Neurology*, **1999**, 45, 146-53	9.4	157
459	Mutations in mammalian target of rapamycin regulator DEPDC5 cause focal epilepsy with brain malformations. *Annals of Neurology*, **2014**, 75, 782-7	9.4	153

458	Mortality in Dravet syndrome. *Epilepsy Research*, **2016**, 128, 43-47	3	152
457	Epilepsy and mental retardation limited to females: an under-recognized disorder. *Brain*, **2008**, 131, 918-27	27.2	152
456	Multicentre search for genetic susceptibility loci in sporadic epilepsy syndrome and seizure types: a case-control study. *Lancet Neurology, The*, **2007**, 6, 970-80	24.1	152
455	Extending the KCNQ2 encephalopathy spectrum: clinical and neuroimaging findings in 17 patients. *Neurology*, **2013**, 81, 1697-703	6.5	151
454	Extended spectrum of idiopathic generalized epilepsies associated with CACNA1H functional variants. *Annals of Neurology*, **2007**, 62, 560-8	9.4	150
453	Genetic testing in the epilepsies--report of the ILAE Genetics Commission. *Epilepsia*, **2010**, 51, 655-70	6.4	147
452	Human epilepsies: interaction of genetic and acquired factors. *Trends in Neurosciences*, **2006**, 29, 391-397	13.3	147
451	Quinidine in the treatment of KCNT1-positive epilepsies. *Annals of Neurology*, **2015**, 78, 995-9	9.4	145
450	Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. *Brain*, **2013**, 136, 3140-50	11.2	144
449	Dravet syndrome as epileptic encephalopathy: evidence from long-term course and neuropathology. *Brain*, **2011**, 134, 2982-3010	11.2	143
448	Channelopathies as a genetic cause of epilepsy. *Current Opinion in Neurology*, **2003**, 16, 171-176	7.1	141
447	Infantile spasms, dystonia, and other X-linked phenotypes caused by mutations in Aristaless related homeobox gene, ARX. *Brain and Development*, **2002**, 24, 266-8	2.2	137
446	The new definition and classification of seizures and epilepsy. *Epilepsy Research*, **2018**, 139, 73-79	3	137
445	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
444	Ultra-rare genetic variation in common epilepsies: a case-control sequencing study. *Lancet Neurology, The*, **2017**, 16, 135-143	24.1	133
443	Absence epilepsies with widely variable onset are a key feature of familial GLUT1 deficiency. *Neurology*, **2010**, 75, 432-40	6.5	133
442	Randomized, phase III study results of clobazam in Lennox-Gastaut syndrome. *Neurology*, **2011**, 77, 1473-81	8.5	133
441	Dravet syndrome or genetic (generalized) epilepsy with febrile seizures plus?. *Brain and Development*, **2009**, 31, 394-400	2.2	129

440	SCN1A duplications and deletions detected in Dravet syndrome: implications for molecular diagnosis. *Epilepsia*, **2009**, 50, 1670-8	6.4	127
439	Severe myoclonic epilepsy of infancy (Dravet syndrome): recognition and diagnosis in adults. *Neurology*, **2006**, 67, 2224-6	6.5	126
438	Childhood absence epilepsy and febrile seizures: a family with a GABA(A) receptor mutation. *Brain*, **2003**, 126, 230-40	11.2	122
437	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
436	Migrating partial seizures of infancy: expansion of the electroclinical, radiological and pathological disease spectrum. *Brain*, **2013**, 136, 1578-91	11.2	120
435	The genetics of human epilepsy. *Trends in Pharmacological Sciences*, **2003**, 24, 428-33	13.2	120
434	A roadmap for precision medicine in the epilepsies. *Lancet Neurology, The*, **2015**, 14, 1219-28	24.1	119
433	Autosomal dominant rolandic epilepsy and speech dyspraxia: a new syndrome with anticipation. *Annals of Neurology*, **1995**, 38, 633-42	9.4	117
432	Copy number variants are frequent in genetic generalized epilepsy with intellectual disability. *Neurology*, **2013**, 81, 1507-14	6.5	115
431	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. *American Journal of Human Genetics*, **2015**, 96, 808-15	11	114
430	Genome-wide association analysis of genetic generalized epilepsies implicates susceptibility loci at 1q43, 2p16.1, 2q22.3 and 17q21.32. *Human Molecular Genetics*, **2012**, 21, 5359-72	5.6	114
429	Electroclinical features of absence seizures in childhood absence epilepsy. *Neurology*, **2006**, 67, 413-8	6.5	113
428	Independent occurrence of the CHRNA4 Ser248Phe mutation in a Norwegian family with nocturnal frontal lobe epilepsy. *Epilepsia*, **2000**, 41, 529-35	6.4	112
427	Glucose transporter 1 deficiency as a treatable cause of myoclonic astatic epilepsy. *Archives of Neurology*, **2011**, 68, 1152-5		110
426	encephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. *Journal of Medical Genetics*, **2017**, 54, 460-470	5.8	109
425	The core network in absence epilepsy. Differences in cortical and thalamic BOLD response. *Neurology*, **2010**, 75, 904-11	6.5	109
424	Occipital epilepsies: identification of specific and newly recognized syndromes. *Brain*, **2003**, 126, 753-69	11.2	109
423	Genetic architecture of idiopathic generalized epilepsy: clinical genetic analysis of 55 multiplex families. *Epilepsia*, **2004**, 45, 467-78	6.4	106

422	Mutations in SLC12A5 in epilepsy of infancy with migrating focal seizures. *Nature Communications*, **2015**, 6, 8038	17.4	104
421	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
420	Ion Channels in Genetic Epilepsy: From Genes and Mechanisms to Disease-Targeted Therapies. *Pharmacological Reviews*, **2018**, 70, 142-173	22.5	103
419	Glucose transporter 1 deficiency in the idiopathic generalized epilepsies. *Annals of Neurology*, **2012**, 72, 807-15	9.4	98
418	Periventricular heterotopia, mental retardation, and epilepsy associated with 5q14.3-q15 deletion. *Neurology*, **2009**, 72, 784-92	6.5	98
417	Familial partial epilepsy with variable foci: a new partial epilepsy syndrome with suggestion of linkage to chromosome 2. *Annals of Neurology*, **1998**, 44, 890-9	9.4	97
416	Febrile seizures. *BMJ, The*, **2007**, 334, 307-11	5.9	96
415	Effects of vaccination on onset and outcome of Dravet syndrome: a retrospective study. *Lancet Neurology, The*, **2010**, 9, 592-8	24.1	95
414	Homozygous PLCB1 deletion associated with malignant migrating partial seizures in infancy. *Epilepsia*, **2012**, 53, e146-50	6.4	93
413	Genetic variation of CACNA1H in idiopathic generalized epilepsy. *Annals of Neurology*, **2004**, 55, 595-6	9.4	92
412	Keeping people with epilepsy safe during the COVID-19 pandemic. *Neurology*, **2020**, 94, 1032-1037	6.5	91
411	Disruptive de novo mutations of DYRK1A lead to a syndromic form of autism and ID. *Molecular Psychiatry*, **2016**, 21, 126-32	15.1	90
410	SCN2A mutations and benign familial neonatal-infantile seizures: the phenotypic spectrum. *Epilepsia*, **2007**, 48, 1138-42	6.4	90
409	SLC25A22 is a novel gene for migrating partial seizures in infancy. *Annals of Neurology*, **2013**, 74, 873-82	9.4	89
408	Neuropsychological and functional MRI studies provide converging evidence of anterior language dysfunction in BECTS. *Epilepsia*, **2009**, 50, 2276-84	6.4	89
407	Genetic and neuroradiological heterogeneity of double cortex syndrome. *Annals of Neurology*, **2000**, 47, 265-269	9.4	89
406	Thalamic atrophy in childhood absence epilepsy. *Epilepsia*, **2006**, 47, 399-405	6.4	88
405	Channelopathies in idiopathic epilepsy. *Neurotherapeutics*, **2007**, 4, 295-304	6.4	86

404	A new molecular mechanism for severe myoclonic epilepsy of infancy: exonic deletions in SCN1A. *Neurology*, **2006**, 67, 1094-5	6.5	85
403	Subcortical band heterotopia (SBH) in males: clinical, imaging and genetic findings in comparison with females. *Brain*, **2002**, 125, 2507-22	11.2	85
402	Precision therapy for epilepsy due to mutations: A randomized trial of oral quinidine. *Neurology*, **2018**, 90, e67-e72	6.5	84
401	Mutations in KCNT1 cause a spectrum of focal epilepsies. *Epilepsia*, **2015**, 56, e114-20	6.4	83
400	Dose-Ranging Effect of Adjunctive Oral Cannabidiol vs Placebo on Convulsive Seizure Frequency in Dravet Syndrome: A Randomized Clinical Trial. *JAMA Neurology*, **2020**, 77, 613-621	17.2	83
399	Clinical profile of patients with ATP1A3 mutations in Alternating Hemiplegia of Childhood-a study of 155 patients. *Orphanet Journal of Rare Diseases*, **2015**, 10, 123	4.2	83
398	Recent advances in the molecular genetics of epilepsy. *Journal of Medical Genetics*, **2013**, 50, 271-9	5.8	82
397	Chromosomal abnormalities and epilepsy: a review for clinicians and gene hunters. *Epilepsia*, **2002**, 43, 127-40	6.4	82
396	SCN8A encephalopathy: Research progress and prospects. *Epilepsia*, **2016**, 57, 1027-35	6.4	82
395	CHD2 variants are a risk factor for photosensitivity in epilepsy. *Brain*, **2015**, 138, 1198-207	11.2	81
394	Phospholipase C beta 1 deficiency is associated with early-onset epileptic encephalopathy. *Brain*, **2010**, 133, 2964-70	11.2	81
393	Susceptibility genes for complex epilepsy. *Human Molecular Genetics*, **2005**, 14 Spec No. 2, R243-9	5.6	80
392	The phenotype of developmental and epileptic encephalopathy. *Neurology*, **2018**, 91, e1112-e1124	6.5	80
391	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. *Human Molecular Genetics*, **2015**, 24, 5250-9	5.6	78
390	Timing of de novo mutagenesis--a twin study of sodium-channel mutations. *New England Journal of Medicine*, **2010**, 363, 1335-40	59.2	78
389	Mapping of a gene determining familial partial epilepsy with variable foci to chromosome 22q11-q12. *American Journal of Human Genetics*, **1999**, 65, 1698-710	11	78
388	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. *Nature Communications*, **2019**, 10, 3094	17.4	76
387	Long-term follow-up of febrile infection-related epilepsy syndrome. *Epilepsia*, **2012**, 53, 101-10	6.4	76

386	Mirror neuron system involvement in empathy: a critical look at the evidence. *Social Neuroscience*, **2011**, 6, 327-35	2	76
385	TBC1D24 genotype-phenotype correlation: Epilepsies and other neurologic features. *Neurology*, **2016**, 87, 77-85	6.5	75
384	Early onset absence epilepsy: 1 in 10 cases is caused by GLUT1 deficiency. *Epilepsia*, **2012**, 53, e204-7	6.4	75
383	Augmented currents of an HCN2 variant in patients with febrile seizure syndromes. *Annals of Neurology*, **2010**, 67, 542-6	9.4	75
382	X-linked myoclonic epilepsy with spasticity and intellectual disability: mutation in the homeobox gene ARX. *Neurology*, **2002**, 59, 348-56	6.5	75
381	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
380	Focal epileptiform spikes do not show a canonical BOLD response in patients with benign rolandic epilepsy (BECTS). *NeuroImage*, **2010**, 51, 252-60	7.9	73
379	Genetics of the epilepsies. *Epilepsia*, **2001**, 42 Suppl 5, 16-23	6.4	73
378	Autoantibodies and epilepsy. *Epilepsia*, **2011**, 52 Suppl 3, 18-22	6.4	71
377	Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hypotonia, and Seizures. *American Journal of Human Genetics*, **2016**, 98, 1001-1010	11	70
376	Classification of the epilepsies: New concepts for discussion and debate-Special report of the ILAE Classification Task Force of the Commission for Classification and Terminology. *Epilepsia Open*, **2016**, 1, 37-44	4	68
375	Heart rate variability in epilepsy: A potential biomarker of sudden unexpected death in epilepsy risk. *Epilepsia*, **2018**, 59, 1372-1380	6.4	68
374	Genetic epilepsy with febrile seizures plus: Refining the spectrum. *Neurology*, **2017**, 89, 1210-1219	6.5	68
373	Familial neonatal seizures in 36 families: Clinical and genetic features correlate with outcome. *Epilepsia*, **2015**, 56, 1071-80	6.4	68
372	Parental Mosaicism in "De Novo" Epileptic Encephalopathies. *New England Journal of Medicine*, **2018**, 378, 1646-1648	59.2	67
371	PRRT2 phenotypic spectrum includes sporadic and fever-related infantile seizures. *Neurology*, **2012**, 79, 2104-8	6.5	65
370	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. *Brain*, **2013** , 136, 3378-94	11.2	65
369	Epilepsy and mental retardation limited to females with PCDH19 mutations can present de novo or in single generation families. *Journal of Medical Genetics*, **2010**, 47, 211-6	5.8	64

368	Real-world utility of whole exome sequencing with targeted gene analysis for focal epilepsy. *Epilepsy Research*, **2017**, 131, 1-8	3	63
367	Epi4K: gene discovery in 4,000 genomes. *Epilepsia*, **2012**, 53, 1457-67	6.4	63
366	Not all epileptic encephalopathies are Dravet syndrome: Early profound Thr226Met phenotype. *Neurology*, **2017**, 89, 1035-1042	6.5	62
365	A set of regulatory genes co-expressed in embryonic human brain is implicated in disrupted speech development. *Molecular Psychiatry*, **2019**, 24, 1065-1078	15.1	62
364	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. *EBioMedicine*, **2015**, 2, 1063-70	8.8	61
363	EEG features of absence seizures in idiopathic generalized epilepsy: impact of syndrome, age, and state. *Epilepsia*, **2009**, 50, 1572-8	6.4	61
362	Progressive gait deterioration in adolescents with Dravet syndrome. *Archives of Neurology*, **2012**, 69, 873-8		61
361	Is the ketogenic diet effective in specific epilepsy syndromes?. *Epilepsy Research*, **2012**, 100, 252-7	3	60
360	Benign occipital epilepsies of childhood: clinical features and genetics. *Brain*, **2008**, 131, 2287-94	11.2	60
359	Familial mesial temporal lobe epilepsy: a benign epilepsy syndrome showing complex inheritance. *Brain*, **2010**, 133, 3221-31	11.2	59
358	A childhood epilepsy mutation reveals a role for developmentally regulated splicing of a sodium channel. *Molecular and Cellular Neurosciences*, **2007**, 35, 292-301	4.8	59
357	Analyzing the etiology of benign rolandic epilepsy: a multicenter twin collaboration. *Epilepsia*, **2006**, 47, 550-5	6.4	59
356	Abnormal Cell Sorting Underlies the Unique X-Linked Inheritance of PCDH19 Epilepsy. *Neuron*, **2018** , 97, 59-66.e5	13.9	58
355	The management of epilepsy in children and adults. *Medical Journal of Australia*, **2018**, 208, 226-233	4	58
354	A new locus for generalized epilepsy with febrile seizures plus maps to chromosome 2. *American Journal of Human Genetics*, **2000**, 66, 698-701	11	58
353	Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3. *Nature Communications*, **2019**, 10, 4919	17.4	58
352	CHD2 myoclonic encephalopathy is frequently associated with self-induced seizures. *Neurology*, **2015**, 84, 951-8	6.5	57
351	Phenotypic comparison of two Scottish families with mutations in different genes causing autosomal dominant nocturnal frontal lobe epilepsy. *Epilepsia*, **2003**, 44, 613-7	6.4	57

350	The ventrolateral medulla and medullary raphe in sudden unexpected death in epilepsy. *Brain*, **2018**, 141, 1719-1733	11.2	56
349	Efficacy of the ketogenic diet: which epilepsies respond?. *Epilepsia*, **2012**, 53, e55-9	6.4	56
348	A multicenter study of BRD2 as a risk factor for juvenile myoclonic epilepsy. *Epilepsia*, **2007**, 48, 706-12	6.4	56
347	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. *Annals of Neurology*, **2019**, 86, 821-831	9.4	55
346	Neonatal epilepsy syndromes and generalized epilepsy with febrile seizures plus (GEFS+). *Epilepsia*, **2005**, 46 Suppl 10, 41-7	6.4	55
345	encephalopathy: A distinctive generalized developmental and epileptic encephalopathy. *Neurology*, **2019**, 92, e96-e107	6.5	55
344	Small intragenic deletion in FOXP2 associated with childhood apraxia of speech and dysarthria. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2321-6	2.5	54
343	SCN1A testing for epilepsy: application in clinical practice. *Epilepsia*, **2013**, 54, 946-52	6.4	54
342	Juvenile myoclonic epilepsy and idiopathic photosensitive occipital lobe epilepsy: is there overlap?. *Brain*, **2004**, 127, 1878-86	11.2	54
341	Clinical and molecular characterization of -related severe early-onset epilepsy. *Neurology*, **2018**, 90, e55-e66	6.5	53
340	4.45 Mb microduplication in chromosome band 14q12 including FOXG1 in a girl with refractory epilepsy and intellectual impairment. *European Journal of Medical Genetics*, **2009**, 52, 440-2	2.6	52
339	Neonatal seizures and long QT syndrome: a cardiocerebral channelopathy?. *Epilepsia*, **2010**, 51, 293-6	6.4	52
338	Human nocturnal frontal lobe epilepsy: pharmocogenomic profiles of pathogenic nicotinic acetylcholine receptor beta-subunit mutations outside the ion channel pore. *Molecular Pharmacology*, **2008**, 74, 379-91	4.3	51
337	Channelopathies as a genetic cause of epilepsy. *Current Opinion in Neurology*, **2003**, 16, 171-6	7.1	51
336	The clinical utility of an SCN1A genetic diagnosis in infantile-onset epilepsy. *Developmental Medicine and Child Neurology*, **2013**, 55, 154-61	3.3	50
335	Dominant KCNA2 mutation causes episodic ataxia and pharmacoresponsive epilepsy. *Neurology*, **2016**, 87, 1975-1984	6.5	50
334	Pitfalls in genetic testing: the story of missed SCN1A mutations. *Molecular Genetics & Genomic Medicine*, **2016**, 4, 457-64	2.3	50
333	Seizures are regulated by ubiquitin-specific peptidase 9 X-linked (USP9X), a de-ubiquitinase. *PLoS Genetics*, **2015**, 11, e1005022	6	49

332	Etiological heterogeneity of familial periventricular heterotopia and hydrocephalus. *Brain and Development*, **2004**, 26, 326-34	2.2	49
331	Clinical and molecular genetics of myoclonic-astatic epilepsy and severe myoclonic epilepsy in infancy (Dravet syndrome). *Brain and Development*, **2001**, 23, 732-5	2.2	49
330	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. *Nature Communications*, **2019**, 10, 4920	17.4	48
329	Molecular analysis of ring chromosome 20 syndrome reveals two distinct groups of patients. *Journal of Medical Genetics*, **2011**, 48, 1-9	5.8	48
328	Familial partial epilepsy with variable foci: clinical features and linkage to chromosome 22q12. *Epilepsia*, **2004**, 45, 1054-60	6.4	48
327	A systematic review and meta-analysis of 271 PCDH19-variant individuals identifies psychiatric comorbidities, and association of seizure onset and disease severity. *Molecular Psychiatry*, **2019**, 24, 241-251	15.1	47
326	A population-based cost-effectiveness study of early genetic testing in severe epilepsies of infancy. *Epilepsia*, **2018**, 59, 1177-1187	6.4	46
325	Role of the sodium channel SCN9A in genetic epilepsy with febrile seizures plus and Dravet syndrome. *Epilepsia*, **2013**, 54, e122-6	6.4	46
324	encephalopathy: A new disease of vesicle fission. *Neurology*, **2017**, 89, 385-394	6.5	46
323	Genetics of epilepsy: The testimony of twins in the molecular era. *Neurology*, **2014**, 83, 1042-8	6.5	46
322	Translational research in epilepsy genetics: sodium channels in man to interneuronopathy in mouse. *Archives of Neurology*, **2009**, 66, 21-6		46
321	Glut1 Deficiency Syndrome (Glut1DS): State of the art in 2020 and recommendations of the international Glut1DS study group. *Epilepsia Open*, **2020**, 5, 354-365	4	45
320	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. *Human Molecular Genetics*, **2014**, 23, 6069-80	5.6	45
319	Etiology of hippocampal sclerosis: evidence for a predisposing familial morphologic anomaly. *Neurology*, **2013**, 81, 144-9	6.5	45
318	SCN1A-related phenotypes: Epilepsy and beyond. *Epilepsia*, **2019**, 60 Suppl 3, S17-S24	6.4	45
317	Neonatal nonepileptic myoclonus is a prominent clinical feature of KCNQ2 gain-of-function variants R201C and R201H. *Epilepsia*, **2017**, 58, 436-445	6.4	44
316	Copy number variant analysis from exome data in 349 patients with epileptic encephalopathy. *Annals of Neurology*, **2015**, 78, 323-8	9.4	44
315	Epileptic spasms are a feature of DEPDC5 mTORopathy. *Neurology: Genetics*, **2015**, 1, e17	3.8	44

314	GRIN2A: an aptly named gene for speech dysfunction. *Neurology*, **2015**, 84, 586-93	6.5	44
313	Diagnosis and long-term course of Dravet syndrome. *European Journal of Paediatric Neurology*, **2012**, 16 Suppl 1, S5-8	3.8	44
312	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. *American Journal of Human Genetics*, **2018**, 103, 666-678	11	44
311	New genes for focal epilepsies with speech and language disorders. *Current Neurology and Neuroscience Reports*, **2015**, 15, 35	6.6	43
310	Expanding the molecular basis and phenotypic spectrum of X-linked Joubert syndrome associated with OFD1 mutations. *European Journal of Human Genetics*, **2012**, 20, 806-9	5.3	43
309	Assessment of the role of FDG PET in the diagnosis and management of children with refractory epilepsy. *European Journal of Nuclear Medicine and Molecular Imaging*, **2005**, 32, 1311-6	8.8	43
308	Reduction of seizure frequency after epilepsy surgery in a patient with STXBP1 encephalopathy and clinical description of six novel mutation carriers. *Epilepsia*, **2013**, 54, e74-80	6.4	42
307	Exploration of the genetic architecture of idiopathic generalized epilepsies. *Epilepsia*, **2006**, 47, 1682-90	6.4	42
306	Is benign rolandic epilepsy genetically determined?. *Annals of Neurology*, **2004**, 56, 129-32	9.4	42
305	Febrile seizures: genetics and relationship to other epilepsy syndromes. *Current Opinion in Neurology*, **1998**, 11, 129-34	7.1	42
304	Whole-genome linkage scan for epilepsy-related photosensitivity: a mega-analysis. *Epilepsy Research*, **2010**, 89, 286-94	3	40
303	Severe autosomal dominant nocturnal frontal lobe epilepsy associated with psychiatric disorders and intellectual disability. *Epilepsia*, **2008**, 49, 2125-9	6.4	40
302	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
301	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
300	Stiripentol efficacy and safety in Dravet syndrome: a 12-year observational study. *Developmental Medicine and Child Neurology*, **2018**, 60, 574-578	3.3	38
299	Early mortality in SCN8A-related epilepsies. *Epilepsy Research*, **2018**, 143, 79-81	3	38
298	Epilepsy with auditory features: A heterogeneous clinico-molecular disease. *Neurology: Genetics*, **2015**, 1, e5	3.8	38
297	Epilepsy genetics revolutionizes clinical practice. *Neuropediatrics*, **2014**, 45, 70-4	1.6	38

296	Clinical genetic studies in benign childhood epilepsy with centrotemporal spikes. *Epilepsia*, **2012**, 53, 319-24	6.4	38
295	Lamotrigine can be beneficial in patients with Dravet syndrome. *Developmental Medicine and Child Neurology*, **2015**, 57, 200-2	3.3	37
294	Epilepsies with single gene inheritance. *Brain and Development*, **1997**, 19, 13-8	2.2	37
293	A targeted resequencing gene panel for focal epilepsy. *Neurology*, **2016**, 86, 1605-12	6.5	37
292	Mutations of the Sonic Hedgehog Pathway Underlie Hypothalamic Hamartoma with Gelastic Epilepsy. *American Journal of Human Genetics*, **2016**, 99, 423-9	11	37
291	Deciphering the concepts behind "Epileptic encephalopathy" and "Developmental and epileptic encephalopathy". *European Journal of Paediatric Neurology*, **2020**, 24, 11-14	3.8	35
290	Reduced dendritic arborization and hyperexcitability of pyramidal neurons in a Scn1b-based model of Dravet syndrome. *Brain*, **2014**, 137, 1701-15	11.2	34
289	Genetics of epilepsy syndromes in families with photosensitivity. *Neurology*, **2013**, 80, 1322-9	6.5	34
288	Clinical genetic study of the epilepsy-aphasia spectrum. *Epilepsia*, **2013**, 54, 280-7	6.4	34
287	The epilepsy phenome/genome project. *Clinical Trials*, **2013**, 10, 568-86	2.2	34
286	Severe infantile onset developmental and epileptic encephalopathy caused by mutations in autophagy gene WDR45. *Epilepsia*, **2018**, 59, e5-e13	6.4	34
285	The 2017 ILAE classification of seizure types and the epilepsies: what do people with epilepsy and their caregivers need to know?. *Epileptic Disorders*, **2018**, 20, 77-87	1.9	34
284	Mutation of the nuclear lamin gene LMNB2 in progressive myoclonus epilepsy with early ataxia. *Human Molecular Genetics*, **2015**, 24, 4483-90	5.6	33
283	De novo SCN1A mutations in Dravet syndrome and related epileptic encephalopathies are largely of paternal origin. *Journal of Medical Genetics*, **2010**, 47, 137-41	5.8	33
282	Vaccination, seizures and 'vaccine damage'. *Current Opinion in Neurology*, **2007**, 20, 181-7	7.1	33
281	Long-term safety and efficacy of clobazam for Lennox-Gastaut syndrome: interim results of an open-label extension study. *Epilepsy and Behavior*, **2012**, 25, 687-94	3.2	32
280	Ectopic posterior pituitary lobe and periventricular heterotopia: cerebral malformations with the same underlying mechanism?. *American Journal of Neuroradiology*, **2002**, 23, 1475-81	4.4	32
279	Genetics of febrile seizure subtypes and syndromes: a twin study. *Epilepsy Research*, **2013**, 105, 103-9	3	31

278	Autism and developmental disability caused by KCNQ3 gain-of-function variants. *Annals of Neurology*, **2019**, 86, 181-192	9.4	30
277	Genetic literacy series: genetic epilepsy with febrile seizures plus. *Epileptic Disorders*, **2018**, 20, 232-238	1.9	30
276	The effect of the ketogenic diet on the developing skeleton. *Epilepsy Research*, **2017**, 136, 62-66	3	30
275	Genetic testing in epilepsy: what should you be doing?. *Epilepsy Currents*, **2011**, 11, 107-11	1.3	30
274	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. *American Journal of Human Genetics*, **2017**, 101, 516-524	11	29
273	Loss of synaptic Zn2+ transporter function increases risk of febrile seizures. *Scientific Reports*, **2015**, 5, 17816	4.9	29
272	Cyclin-dependent kinase-like 5 (CDKL5) mutation screening in Rett syndrome and related disorders. *Twin Research and Human Genetics*, **2010**, 13, 168-78	2.2	29
271	Factors influencing clinical features of absence seizures. *Epilepsia*, **2008**, 49, 2100-7	6.4	29
270	Severe childhood speech disorder: Gene discovery highlights transcriptional dysregulation. *Neurology*, **2020**, 94, e2148-e2167	6.5	28
269	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
268	Genome-wide linkage meta-analysis identifies susceptibility loci at 2q34 and 13q31.3 for genetic generalized epilepsies. *Epilepsia*, **2012**, 53, 308-18	6.4	28
267	Sleep problems in Dravet syndrome: a modifiable comorbidity. *Developmental Medicine and Child Neurology*, **2018**, 60, 192-198	3.3	28
266	A Primate-Specific Isoform of PLEKHG6 Regulates Neurogenesis and Neuronal Migration. *Cell Reports*, **2018**, 25, 2729-2741.e6	10.6	27
265	Spectrum of neurodevelopmental disease associated with the GNAO1 guanosine triphosphate-binding region. *Epilepsia*, **2019**, 60, 406-418	6.4	26
264	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
263	Faulty cardiac repolarization reserve in alternating hemiplegia of childhood broadens the phenotype. *Brain*, **2015**, 138, 2859-74	11.2	26
262	Genetics of human partial epilepsy. *Current Opinion in Neurology*, **1997**, 10, 110-4	7.1	26
261	Fenfluramine HCl (Fintepla) provides long-term clinically meaningful reduction in seizure frequency: Analysis of an ongoing open-label extension study. *Epilepsia*, **2020**, 61, 2396-2404	6.4	26

260	Peritrigonal and temporo-occipital heterotopia with corpus callosum and cerebellar dysgenesis. *Neurology*, **2012**, 79, 1244-51	6.5	25
259	Bilateral posterior periventricular nodular heterotopia: a recognizable cortical malformation with a spectrum of associated brain abnormalities. *American Journal of Neuroradiology*, **2013**, 34, 432-8	4.4	25
258	Familial adult myoclonic epilepsy: recognition of mild phenotypes and refinement of the 2q locus. *Archives of Neurology*, **2012**, 69, 474-81		25
257	Familial neonatal seizures with intellectual disability caused by a microduplication of chromosome 2q24.3. *Epilepsia*, **2010**, 51, 1865-9	6.4	25
256	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. *Nature Communications*, **2020**, 11, 4932	17.4	25
255	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. *Epilepsia*, **2019**, 60, 797-806	6.4	24
254	Somatic mutation: The hidden genetics of brain malformations and focal epilepsies. *Epilepsy Research*, **2019**, 155, 106161	3	24
253	Absence of mutations in the LGI1 receptor ADAM22 gene in autosomal dominant lateral temporal epilepsy. *Epilepsy Research*, **2007**, 76, 41-8	3	24
252	DEPDC5 as a potential therapeutic target for epilepsy. *Expert Opinion on Therapeutic Targets*, **2017**, 21, 591-600	6.4	23
251	ADGRV1 is implicated in myoclonic epilepsy. *Epilepsia*, **2018**, 59, 381-388	6.4	23
250	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
249	Loss of function of the retinoid-related nuclear receptor (RORB) gene and epilepsy. *European Journal of Human Genetics*, **2016**, 24, 1761-1770	5.3	23
248	Glucose metabolism transporters and epilepsy: only GLUT1 has an established role. *Epilepsia*, **2014**, 55, e18-21	6.4	23
247	Genetic dissection of the common epilepsies. *Current Opinion in Neurology*, **2006**, 19, 157-63	7.1	23
246	Autosomal dominant nocturnal frontal lobe epilepsy. *Epilepsia*, **2000**, 41, 1059-60	6.4	23
245	SLC35A2-CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. *Human Mutation*, **2019**, 40, 908-925	4.7	23
244	Multiplex families with epilepsy: Success of clinical and molecular genetic characterization. *Neurology*, **2016**, 86, 713-22	6.5	22
243	Infantile Spasms of Unknown Cause: Predictors of Outcome and Genotype-Phenotype Correlation. *Pediatric Neurology*, **2018**, 87, 48-56	2.9	22

242	Harnessing gene expression networks to prioritize candidate epileptic encephalopathy genes. *PLoS ONE*, **2014**, 9, e102079	3.7	22
241	Is focal cortical dysplasia sporadic? Family evidence for genetic susceptibility. *Epilepsia*, **2014**, 55, e22-6	6.4	22
240	Automatisms in absence seizures in children with idiopathic generalized epilepsy. *Archives of Neurology*, **2009**, 66, 729-34		22
239	Reflex seizures in patients with malformations of cortical development and refractory epilepsy. *Epilepsia*, **2005**, 46, 1224-34	6.4	22
238	Efficacy of cannabinoids in paediatric epilepsy. *Developmental Medicine and Child Neurology*, **2019**, 61, 13-18	3.3	22
237	De novo SCN1A pathogenic variants in the GEFS+ spectrum: Not always a familial syndrome. *Epilepsia*, **2017**, 58, e26-e30	6.4	21
236	Fatal Cerebral Edema With Status Epilepticus in Children With Dravet Syndrome: Report of 5 Cases. *Pediatrics*, **2017**, 139,	7.4	21
235	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. *American Journal of Human Genetics*, **2018**, 103, 305-316	11	21
234	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. *Nature Communications*, **2019**, 10, 4679	17.4	21
233	Gene expression analysis in absence epilepsy using a monozygotic twin design. *Epilepsia*, **2008**, 49, 1546-54	5.4	21
232	Epilepsy in KCNH1-related syndromes. *Epileptic Disorders*, **2016**, 18, 123-36	1.9	21
231	Epilepsy classification: a cycle of evolution and revolution. *Current Opinion in Neurology*, **2013**, 26, 163-7	7.1	20
230	Dysarthria and broader motor speech deficits in Dravet syndrome. *Neurology*, **2017**, 88, 743-749	6.5	19
229	Gain-of-function HCN2 variants in genetic epilepsy. *Human Mutation*, **2018**, 39, 202-209	4.7	19
228	PRIMA1 mutation: a new cause of nocturnal frontal lobe epilepsy. *Annals of Clinical and Translational Neurology*, **2015**, 2, 821-30	5.3	19
227	Family studies of individuals with eyelid myoclonia with absences. *Epilepsia*, **2012**, 53, 2141-8	6.4	19
226	Severe infantile epilepsies: molecular genetics challenge clinical classification. *Brain*, **2003**, 126, 513-4	11.2	19
225	Evaluation of non-coding variation in GLUT1 deficiency. *Developmental Medicine and Child Neurology*, **2016**, 58, 1295-1302	3.3	19

224	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
223	Somatic mutation in the of Sturge-Weber syndrome. *Neurology: Genetics*, **2018**, 4, e236	3.8	18
222	Familial focal epilepsy with variable foci mapped to chromosome 22q12: expansion of the phenotypic spectrum. *Epilepsia*, **2012**, 53, e151-5	6.4	18
221	Mutations in TNK2 in severe autosomal recessive infantile onset epilepsy. *Annals of Neurology*, **2013**, 74, 496-501	9.4	18
220	Efficacy and tolerability of adjunctive lacosamide in pediatric patients with focal seizures. *Neurology*, **2019**, 93, e1212-e1226	6.5	17
219	Perception of impact of Dravet syndrome on children and caregivers in multiple countries: looking beyond seizures. *Developmental Medicine and Child Neurology*, **2019**, 61, 1229-1236	3.3	17
218	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. *American Journal of Human Genetics*, **2019**, 104, 948-956	11	17
217	Does a SCN1A gene mutation confer earlier age of onset of febrile seizures in GEFS+?. *Epilepsia*, **2009**, 50, 953-6	6.4	17
216	Early seizures: causal events or predisposition to adult epilepsy?. *Lancet Neurology, The*, **2007**, 6, 643-51	24.1	17
215	Cerebrospinal fluid liquid biopsy for detecting somatic mosaicism in brain. *Brain Communications*, **2021**, 3, fcaa235	4.5	17
214	ILAE-Klassifikation der Epilepsien: Positionspapier der ILAE-Kommission f□ Klassifikation und Terminologie. *Zeitschrift Fur Epileptologie*, **2018**, 31, 296-306	0.1	17
213	Features of the broader autism phenotype in people with epilepsy support shared mechanisms between epilepsy and autism spectrum disorder. *Neuroscience and Biobehavioral Reviews*, **2017**, 75, 203-233	9	16
212	Inhibition of Upf2-Dependent Nonsense-Mediated Decay Leads to Behavioral and Neurophysiological Abnormalities by Activating the Immune Response. *Neuron*, **2019**, 104, 665-679.e8	13.9	16
211	Genetic analysis of PHOX2B in sudden unexpected death in epilepsy cases. *Neurology*, **2014**, 83, 1018-21	6.5	16
210	Deletions of 16p11.2 and 19p13.2 in a family with intellectual disability and generalized epilepsy. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1722-5	2.5	16
209	Copy number variants--an unexpected risk factor for the idiopathic generalized epilepsies. *Brain*, **2010**, 133, 7-8	11.2	16
208	Genetics of the epilepsies. *Current Opinion in Pediatrics*, **2000**, 12, 536-42	3.2	16
207	A twin study of genetic influences on epilepsy outcome. *Twin Research and Human Genetics*, **2003**, 6, 140-6		16

206	Frequency of CNKSR2 mutation in the X-linked epilepsy-aphasia spectrum. *Epilepsia*, **2017**, 58, e40-e43	6.4	15
205	Seizure semiology in autosomal dominant epilepsy with auditory features, due to novel LGI1 mutations. *Epilepsy Research*, **2013**, 107, 311-7	3	15
204	Autosomal dominant vasovagal syncope: clinical features and linkage to chromosome 15q26. *Neurology*, **2013**, 80, 1485-93	6.5	15
203	Lennox-Gastaut syndrome of unknown cause: phenotypic characteristics of patients in the Epilepsy Phenome/Genome Project. *Epilepsia*, **2013**, 54, 1898-904	6.4	15
202	The Role of Seizure-Related SEZ6 as a Susceptibility Gene in Febrile Seizures. *Neurology Research International*, **2011**, 2011, 917565	1.7	15
201	Genetics of the epilepsies. *Current Opinion in Neurology*, **1999**, 12, 177-82	7.1	15
200	In silico prioritization based on coexpression can aid epileptic encephalopathy gene discovery. *Neurology: Genetics*, **2016**, 2, e51	3.8	15
199	Early neuroimaging markers of FOXP2 intragenic deletion. *Scientific Reports*, **2016**, 6, 35192	4.9	15
198	ILAE classification and definition of epilepsy syndromes with onset in neonates and infants: Position statement by the ILAE Task Force on Nosology and Definitions.. *Epilepsia*, **2022**,	6.4	15
197	Gonadal mosaicism of a novel IQSEC2 variant causing female limited intellectual disability and epilepsy. *European Journal of Human Genetics*, **2017**, 25, 763-767	5.3	14
196	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17 458 subjects. *Brain*, **2020**, 143, 2106-2118	11.2	14
195	In vivo loss of slow potassium channel activity in individuals with benign familial neonatal epilepsy in remission. *Brain*, **2012**, 135, 3144-52	11.2	14
194	A retrospective population-based study on seizures related to childhood vaccination. *Epilepsia*, **2011**, 52, 1506-12	6.4	14
193	Mosaicism of a missense SCN1A mutation and Dravet syndrome in a Roma/Gypsy family. *Epileptic Disorders*, **2010**, 12, 117-24	1.9	14
192	Expanding the genetic and phenotypic relevance of KCNB1 variants in developmental and epileptic encephalopathies: 27 new patients and overview of the literature. *Human Mutation*, **2020**, 41, 69-80	4.7	14
191	Neuronal ceroid lipofuscinosis type 2: an Australian case series. *Journal of Paediatrics and Child Health*, **2020**, 56, 1210-1218	1.3	13
190	A mutation in COL4A2 causes autosomal dominant porencephaly with cataracts. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 1059-63	2.5	13
189	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

188	Is variation in the GABA(B) receptor 1 gene associated with temporal lobe epilepsy?. *Epilepsia*, **2005**, 46, 778-80	6.4	13
187	Periventricular Nodular Heterotopia: Detection of Abnormal Microanatomic Fiber Structures with Whole-Brain Diffusion MR Imaging Tractography. *Radiology*, **2016**, 281, 896-906	20.5	13
186	De novo variants in the alternative exon 5 of SCN8A cause epileptic encephalopathy. *Genetics in Medicine*, **2018**, 20, 275-281	8.1	13
185	A standardized patient-centered characterization of the phenotypic spectrum of PCDH19 girls clustering epilepsy. *Translational Psychiatry*, **2020**, 10, 127	8.6	12
184	Familial mesial temporal lobe epilepsy and the borderland of dJ□vu. *Annals of Neurology*, **2017**, 82, 166-176	9.4	12
183	Genetics of idiopathic epilepsies. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2013**, 111, 567-78	3	12
182	Milder phenotypes of glucose transporter type 1 deficiency syndrome. *Developmental Medicine and Child Neurology*, **2011**, 53, 664-8	3.3	12
181	Electroclinical features of absence seizures in sleep. *Epilepsy Research*, **2011**, 93, 216-20	3	12
180	Detection of microchromosomal aberrations in refractory epilepsy: a pilot study. *Epileptic Disorders*, **2010**, 12, 192-8	1.9	12
179	Neuropsychological function in patients with a single gene mutation associated with autosomal dominant nocturnal frontal lobe epilepsy. *Epilepsy and Behavior*, **2010**, 17, 531-5	3.2	12
178	A duplication in 1q21.3 in a family with early onset and childhood absence epilepsy. *Epilepsia*, **2010**, 51, 2453-6	6.4	12
177	International League Against Epilepsy classification and definition of epilepsy syndromes with onset in childhood: Position paper by the ILAE Task Force on Nosology and Definitions.. *Epilepsia*, **2022**,	6.4	12
176	Schizophrenia is a later-onset feature of PCDH19 Girls Clustering Epilepsy. *Epilepsia*, **2019**, 60, 429-440	6.4	11
175	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. *Annals of Neurology*, **2018**, 83, 926-934	9.4	11
174	Mutations in PRRT2 are not a common cause of infantile epileptic encephalopathies. *Epilepsia*, **2013**, 54, e86-9	6.4	11
173	Head stereotypies in STXBP1 encephalopathy. *Developmental Medicine and Child Neurology*, **2013**, 55, 769-72	3.3	11
172	Electroencephalographic abnormalities during sleep in children with developmental speech-language disorders: a case-control study. *Developmental Medicine and Child Neurology*, **2009**, 51, 228-34	3.3	11
171	Damaging de novo missense variants in EEF1A2 lead to a developmental and degenerative epileptic-dyskinetic encephalopathy. *Human Mutation*, **2020**, 41, 1263-1279	4.7	10

170	Pathogenic Variants in CEP85L Cause Sporadic and Familial Posterior Predominant Lissencephaly. *Neuron*, **2020**, 106, 237-245.e8	13.9	10
169	Epilepsy: Beyond the single nucleotide variant in epilepsy genetics. *Nature Reviews Neurology*, **2014**, 10, 490-1	15	10
168	Epilepsy: a classification for all seasons?. *Epilepsia*, **2012**, 53 Suppl 2, 6-9	6.4	10
167	Rare protein sequence variation in SV2A gene does not affect response to levetiracetam. *Epilepsy Research*, **2012**, 101, 277-9	3	10
166	Revidierte Terminologie und Konzepte zur Einteilung von epileptischen Anfällen und Epilepsien: Bericht der Klassifikations- und Terminologiekommission der Internationalen Liga gegen Epilepsie, 2005–2009. *Aktuelle Neurologie*, **2010**, 37, 120-130		10
165	Does genotype determine phenotype? Sodium channel mutations in Dravet syndrome and GEFS+. *Neurology*, **2011**, 76, 588-9	6.5	10
164	Locus for febrile seizures. *Annals of Neurology*, **2000**, 47, 840-1	9.4	10
163	Cognitive, behavioral, and social functioning in children and adults with Dravet syndrome. *Epilepsy and Behavior*, **2020**, 112, 107319	3.2	10
162	Predominant and novel de novo variants in 29 individuals with ALG13 deficiency: Clinical description, biomarker status, biochemical analysis, and treatment suggestions. *Journal of Inherited Metabolic Disease*, **2020**, 43, 1333-1348	5.4	10
161	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
160	Anleitung (Instruction manual) zur Anwendung der operationalen Klassifikation von Anfallsformen der ILAE 2017. *Zeitschrift Fur Epileptologie*, **2018**, 31, 282-295	0.1	10
159	Add-on cannabidiol in patients with Dravet syndrome: Results of a long-term open-label extension trial. *Epilepsia*, **2021**, 62, 2505-2517	6.4	10
158	Myoclonic Absence Seizures in Dravet Syndrome. *Pediatric Neurology*, **2017**, 70, 67-69	2.9	9
157	Familial epilepsy with anterior polymicrogyria as a presentation of COL18A1 mutations. *European Journal of Medical Genetics*, **2017**, 60, 437-443	2.6	9
156	The epileptology of GNB5 encephalopathy. *Epilepsia*, **2019**, 60, e121-e127	6.4	9
155	2017 International League Against Epilepsy classifications of seizures and epilepsy are steps in the right direction. *Epilepsia*, **2019**, 60, 1040-1044	6.4	9
154	Dorsal language stream anomalies in an inherited speech disorder. *Brain*, **2019**, 142, 966-977	11.2	9
153	"It's good to know": experiences of gene identification and result disclosure in familial epilepsies. *Epilepsy Research*, **2015**, 112, 64-71	3	9

152	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
151	Do mutations in SCN1B cause Dravet syndrome?. *Epilepsy Research*, **2013**, 103, 97-100	3	9
150	Vaccination Triggers, Rather Than Causes, Seizures. *Epilepsy Currents*, **2015**, 15, 335-7	1.3	9
149	What is at stake in a classification?. *Epilepsia*, **2011**, 52, 1205-1208	6.4	9
148	Benign neonatal sleep myoclonus: an autosomal dominant form not allelic to KCNQ2 or KCNQ3. *Journal of Child Neurology*, **2012**, 27, 1260-3	2.5	9
147	Febrile convulsions and genetic susceptibility: role of the neuronal nicotinic acetylcholine receptor alpha 4 subunit. *Epilepsia*, **2004**, 45, 561; author reply 561-2	6.4	9
146	How long should a routine EEG be?. *Journal of Clinical Neuroscience*, **1999**, 6, 492-3	2.2	9
145	Levetiracetam efficacy in PCDH19 Girls Clustering Epilepsy. *European Journal of Paediatric Neurology*, **2020**, 24, 142-147	3.8	9
144	Germline and Mosaic Variants in PRKACA and PRKACB Cause a Multiple Congenital Malformation Syndrome. *American Journal of Human Genetics*, **2020**, 107, 977-988	11	9
143	Randomized Controlled Trial of Melatonin for Sleep Disturbance in Dravet Syndrome: The DREAMS Study. *Journal of Clinical Sleep Medicine*, **2018**, 14, 1697-1704	3.1	9
142	Operationale Klassifikation der Anfallsformen durch die Internationale Liga gegen Epilepsie: Positionspapier der ILAE-Klassifikations- und Terminologiekommission. *Zeitschrift Fur Epileptologie*, **2018**, 31, 272-281	0.1	9
141	Defining Dravet syndrome: An essential pre-requisite for precision medicine trials. *Epilepsia*, **2021**, 62, 2205-2217	6.4	9
140	GEFS+ where focal seizures evolve from generalized spike wave: video-EEG study of two children. *Epileptic Disorders*, **2007**, 9, 307-14	1.9	9
139	The epileptology of Koolen-de Vries syndrome: Electro-clinico-radiologic findings in 31 patients. *Epilepsia*, **2017**, 58, 1085-1094	6.4	8
138	Recessive variants in ZNF142 cause a complex neurodevelopmental disorder with intellectual disability, speech impairment, seizures, and dystonia. *Genetics in Medicine*, **2019**, 21, 2532-2542	8.1	8
137	Focal epilepsy in SCN1A-mutation carrying patients: is there a role for epilepsy surgery?. *Developmental Medicine and Child Neurology*, **2020**, 62, 1331-1335	3.3	8
136	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
135	Febrile infection-related epilepsy syndrome is not caused by SCN1A mutations. *Epilepsy Research*, **2012**, 100, 194-8	3	8

134	Investigation of the 15q13.3 CNV as a genetic modifier for familial epilepsies with variable phenotypes. *Epilepsia*, **2011**, 52, e139-42	6.4	8
133	Is photosensitive epilepsy less common in males due to variation in X chromosome photopigment genes?. *Epilepsia*, **2007**, 48, 1807-1809	6.4	8
132	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
131	Medullary tyrosine hydroxylase catecholaminergic neuronal populations in sudden unexpected death in epilepsy. *Brain Pathology*, **2021**, 31, 133-143	6	8
130	Myoclonic occipital photosensitive epilepsy with dystonia (MOPED): A familial epilepsy syndrome. *Epilepsy Research*, **2015**, 114, 98-105	3	7
129	Bi-allelic LoF NRROS Variants Impairing Active TGF-β Delivery Cause a Severe Infantile-Onset Neurodegenerative Condition with Intracranial Calcification. *American Journal of Human Genetics*, **2020**, 106, 559-569	11	7
128	Childhood-onset generalized epilepsy in Bainbridge-Ropers syndrome. *Epilepsy Research*, **2018**, 140, 166-170	3	7
127	Favourable response to ketogenic dietary therapies: undiagnosed glucose 1 transporter deficiency syndrome is only one factor. *Developmental Medicine and Child Neurology*, **2015**, 57, 969-76	3.3	7
126	SCN1A Variants in vaccine-related febrile seizures: A prospective study. *Annals of Neurology*, **2020**, 87, 281-288	9.4	7
125	BRAT1 encephalopathy: a recessive cause of epilepsy of infancy with migrating focal seizures. *Developmental Medicine and Child Neurology*, **2020**, 62, 1096-1099	3.3	7
124	Climate change and epilepsy: Insights from clinical and basic science studies. *Epilepsy and Behavior*, **2021**, 116, 107791	3.2	7
123	Cation leak underlies neuronal excitability in an HCN1 developmental and epileptic encephalopathy. *Brain*, **2021**, 144, 2060-2073	11.2	7
122	The severe epilepsy syndromes of infancy: A population-based study. *Epilepsia*, **2021**, 62, 358-370	6.4	7
121	Speech and language in bilateral perisylvian polymicrogyria: a systematic review. *Developmental Medicine and Child Neurology*, **2019**, 61, 1145-1152	3.3	6
120	Clinical and genetic analysis of a family with two rare reflex epilepsies. *Seizure: the Journal of the British Epilepsy Association*, **2015**, 29, 90-6	3.2	6
119	Parental health spillover effects of paediatric rare genetic conditions. *Quality of Life Research*, **2020**, 29, 2445-2454	3.7	6
118	The ketogenic diet is effective for refractory epilepsy associated with acquired structural epileptic encephalopathy. *Developmental Medicine and Child Neurology*, **2018**, 60, 718-723	3.3	6
117	Genome-wide association study: Exploring the genetic basis for responsiveness to ketogenic dietary therapies for drug-resistant epilepsy. *Epilepsia*, **2018**, 59, 1557-1566	6.4	6

116	Racial and ethnic differences in epilepsy classification among probands in the Epilepsy Phenome/Genome Project (EPGP). *Epilepsy Research*, **2013**, 107, 306-10	3	6
115	Variants in KCNJ11 and BAD do not predict response to ketogenic dietary therapies for epilepsy. *Epilepsy Research*, **2015**, 118, 22-8	3	6
114	Transition to adult life in the monogenic epilepsies. *Epilepsia*, **2014**, 55 Suppl 3, 12-5	6.4	6
113	Does variation in NIPA2 contribute to genetic generalized epilepsy?. *Human Genetics*, **2014**, 133, 673-4	6.3	6
112	Optimizing electroencephalographic studies for epilepsy diagnosis in children with new-onset seizures. *Archives of Neurology*, **2010**, 67, 1345-9		6
111	Association of a nicotinic receptor mutation with reduced height and blunted physostigmine-stimulated growth hormone release. *Journal of Clinical Endocrinology and Metabolism*, **2008**, 93, 634-7	5.6	6
110	New autosomal-dominant partial epilepsy syndrome. *Pediatric Neurology*, **1994**, 11, 95	2.9	6
109	Cardiac phenotype in -related syndromes: A multicenter cohort study. *Neurology*, **2020**, 95, e2866-e2879	6.5	6
108	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
107	Is FGF13 a major contributor to genetic epilepsy with febrile seizures plus?. *Epilepsy Research*, **2016**, 128, 48-51	3	6
106	Reanalysis and optimisation of bioinformatic pipelines is critical for mutation detection. *Human Mutation*, **2019**, 40, 374-379	4.7	6
105	Double somatic mosaicism in a child with Dravet syndrome. *Neurology: Genetics*, **2019**, 5, e333	3.8	5
104	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
103	Antiepileptic Drug Teratogenicity and De Novo Genetic Variation Load. *Annals of Neurology*, **2020**, 87, 897-906	9.4	5
102	Inherited RORB pathogenic variants: Overlap of photosensitive genetic generalized and occipital lobe epilepsy. *Epilepsia*, **2020**, 61, e23-e29	6.4	5
101	Therapeutic use of medicinal cannabis in difficult to manage epilepsy. *British Journal of Clinical Pharmacology*, **2018**, 84, 2488-2490	3.8	5
100	A novel GEFS+ locus on 12p13.33 in a large Roma family. *Epilepsy Research*, **2011**, 97, 198-207	3	5
99	Genetics of the epilepsies: genetic twists in the channels and other tales. *Epilepsia*, **2010**, 51 Suppl 1, 33-6	6.4	5

98	The role of genetics and ethnicity in epilepsy management. *Acta Neurologica Scandinavica*, **2005**, 181, 47-51	3.8	5
97	Genetics of the Epilepsies. *Epilepsia*, **2001**, 42, 16-23	6.4	5
96	Contribution of rare genetic variants to drug response in absence epilepsy. *Epilepsy Research*, **2021**, 170, 106537	3	5
95	Development of a rapid functional assay that predicts GLUT1 disease severity. *Neurology: Genetics*, **2018**, 4, e297	3.8	5
94	Multifocal epilepsy: the role of palliative resection - intractable frontal and occipital lobe epilepsy secondary to radiotherapy for acute lymphoblastic leukaemia. *Epileptic Disorders*, **2008**, 10, 362-70	1.9	5
93	Evaluation of GLUT1 variation in non-acquired focal epilepsy. *Epilepsy Research*, **2017**, 133, 54-57	3	4
92	Epidemiology and etiology of infantile developmental and epileptic encephalopathies in Tasmania. *Epilepsia Open*, **2019**, 4, 504-510	4	4
91	Intestinal-Cell Kinase and Juvenile Myoclonic Epilepsy. *New England Journal of Medicine*, **2019**, 380, e24	59.2	4
90	No evidence for a BRD2 promoter hypermethylation in blood leukocytes of Europeans with juvenile myoclonic epilepsy. *Epilepsia*, **2019**, 60, e31-e36	6.4	4
89	Generation of seven iPSC lines from peripheral blood mononuclear cells suitable to investigate Autism Spectrum Disorder. *Stem Cell Research*, **2019**, 39, 101516	1.6	4
88	When Monogenic Isn't Monogenic-Unravelling the Oligogenic Architecture of the Developmental and Epileptic Encephalopathies. *Epilepsy Currents*, **2019**, 19, 417-419	1.3	4
87	Siblings with refractory occipital epilepsy showing localized network activity on EEG-fMRI. *Epilepsia*, **2013**, 54, e28-32	6.4	4
86	Classification and clinical features of absence epilepsies: how evidence leads to changing concepts. *Epilepsia*, **2008**, 49, 2140-1	6.4	4
85	Natural History Studies and Clinical Trial Readiness for Genetic Developmental and Epileptic Encephalopathies. *Neurotherapeutics*, **2021**, 18, 1432-1444	6.4	4
84	Developmental and epilepsy spectrum of KCNB1 encephalopathy with long-term outcome. *Epilepsia*, **2020**, 61, 2461-2473	6.4	4
83	Seizures in Sotos syndrome: Phenotyping in 49 patients. *Epilepsia Open*, **2021**, 6, 425-430	4	4
82	Guidelines on the diagnosis, clinical assessments, treatment and management for CLN2 disease patients. *Orphanet Journal of Rare Diseases*, **2021**, 16, 185	4.2	4
81	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. *Brain*, **2021**, 144, 1435-1450	11.2	4

80	Fragile Females: Case Series of Epilepsy in Girls With Disruption. *Pediatrics*, **2019**, 144,	7.4	4
79	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. *Genetics in Medicine*, **2021**, 23, 363-373	8.1	4
78	Self-limited focal epilepsy and childhood apraxia of speech with WAC pathogenic variants. *European Journal of Paediatric Neurology*, **2021**, 30, 25-28	3.8	4
77	A new classification and class 1 evidence transform clinical practice in epilepsy. *Lancet Neurology, The*, **2018**, 17, 7-8	24.1	4
76	The phenotypic spectrum of X-linked, infantile onset ALG13-related developmental and epileptic encephalopathy. *Epilepsia*, **2021**, 62, 325-334	6.4	4
75	Genome-wide association study of febrile seizures implicates fever response and neuronal excitability genes.. *Brain*, **2022**,	11.2	4
74	Methodology for classification and definition of epilepsy syndromes with list of syndromes: Report of the ILAE Task Force on Nosology and Definitions.. *Epilepsia*, **2022**,	6.4	4
73	Evaluation of multiple putative risk alleles within the 15q13.3 region for genetic generalized epilepsy. *Epilepsy Research*, **2015**, 117, 70-3	3	3
72	Hemiconvulsion-hemiplegia-epilepsy evolving to contralateral hemi-Lennox-Gastaut-like phenotype. *Brain and Development*, **2018**, 40, 425-428	2.2	3
71	Myoclonic absence seizures with complex gestural automatisms. *European Journal of Paediatric Neurology*, **2018**, 22, 532-535	3.8	3
70	KANSL1 variation is not a major contributing factor in self-limited focal epilepsy syndromes of childhood. *PLoS ONE*, **2018**, 13, e0191546	3.7	3
69	Classification as autonomic versus sensory seizures. *Epilepsia*, **2019**, 60, 2003-2005	6.4	3
68	How long for epilepsy remission in the ILAE definition?. *Epilepsia*, **2017**, 58, 1486-1487	6.4	3
67	Genetics of the epilepsies: channelopathies and beyond. *Epilepsia*, **2011**, 52, 192-3; discussion 193-6	6.4	3
66	Association studies and functional validation or functional validation alone?. *Epilepsy Research*, **2007**, 74, 237-8	3	3
65	Extreme virilization in patients with congenital adrenal hyperplasia fails to induce descent of the ovary. *Pediatric Surgery International*, **1988**, 3-3, 165	2.1	3
64	Genetic (Generalized) Epilepsy with Febrile Seizures Plus **2008**, 29-33		3
63	Are Variants Causing Cardiac Arrhythmia Risk Factors in Sudden Unexpected Death in Epilepsy?. *Frontiers in Neurology*, **2020**, 11, 925	4.1	3

62	PCDH19 Pathogenic Variants in Males: Expanding the Phenotypic Spectrum. *Advances in Experimental Medicine and Biology*, **2020**, 1298, 177-187	3.6	3
61	Diverse genetic causes of polymicrogyria with epilepsy. *Epilepsia*, **2021**, 62, 973-983	6.4	3
60	Severe speech impairment is a distinguishing feature of FOXP1-related disorder. *Developmental Medicine and Child Neurology*, **2021**, 63, 1417-1426	3.3	3
59	Human generalized epilepsy: Increased somatosensory and striatothalamic connectivity. *Neurology: Genetics*, **2019**, 5, e340	3.8	3
58	Association of Missense Variants With Genetic Epilepsy With Febrile Seizures Plus. *Neurology*, **2021**, 96, e2251-e2260	6.5	3
57	Precision Medicine Approaches for Infantile-Onset Developmental and Epileptic Encephalopathies. *Annual Review of Pharmacology and Toxicology*, **2021**,	17.9	3
56	Safety and efficacy of ganaxolone in patients with CDKL5 deficiency disorder: results from the double-blind phase of a randomised, placebo-controlled, phase 3 trial.. *Lancet Neurology, The*, **2022**, 21, 417-427	24.1	3
55	Why should a neurologist worry about climate change?. *Lancet Neurology, The*, **2019**, 18, 335-336	24.1	2
54	Defining the phenotype of FHF1 developmental and epileptic encephalopathy. *Epilepsia*, **2020**, 61, e71-e78	6.4	2
53	Epilepsy in 2012: Advances in epilepsy shed light on key questions. *Nature Reviews Neurology*, **2013**, 9, 66-8	15	2
52	Development and Validation of a Prediction Model for Early Diagnosis of -Related Epilepsies.. *Neurology*, **2022**,	6.5	2
51	Genome-wide association study of febrile seizures identifies seven new loci implicating fever response and neuronal excitability genes		2
50	Cognitive processes predicting advanced theory of mind in the broader autism phenotype. *Autism Research*, **2020**, 13, 921-934	5.1	2
49	Phenotypic Spectrum of Seizure Disorders in MBD5-Associated Neurodevelopmental Disorder. *Neurology: Genetics*, **2021**, 7, e579	3.8	2
48	Genetic Contributions to Acquired Epilepsies. *Epilepsy Currents*, **2021**, 21, 5-13	1.3	2
47	The aetiologies of epilepsy. *Epileptic Disorders*, **2021**, 23, 1-16	1.9	2
46	Mosaic uniparental disomy results in GM1 gangliosidosis with normal enzyme assay. *American Journal of Medical Genetics, Part A*, **2018**, 176, 230-234	2.5	2
45	Safety and Tolerability of Transdermal Cannabidiol Gel in Children With Developmental and Epileptic Encephalopathies: A Nonrandomized Controlled Trial. *JAMA Network Open*, **2021**, 4, e2123930	10.4	2

44	Is a microRNA-328 binding site in associated with Rolandic epilepsy?. *Annals of Clinical and Translational Neurology*, **2017**, 4, 276-277	5.3	1
43	Looking to the Future: Speech, Language, and Academic Outcomes in an Adolescent with Childhood Apraxia of Speech. *Folia Phoniatrica Et Logopaedica*, **2019**, 71, 203-215	1.5	1
42	Tracing Autism Traits in Large Multiplex Families to Identify Endophenotypes of the Broader Autism Phenotype. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	1
41	Vaccination and Dravet syndrome Authors' reply. *Lancet Neurology, The*, **2010**, 9, 1148-1149	24.1	1
40	Epileptiform EEG abnormalities in children with language regression. *Neurology*, **2006**, 67, 1527; author reply 1527	6.5	1
39	Evidence for a Dual-Pathway, 2-Hit Genetic Model for Focal Cortical Dysplasia and Epilepsy.. *Neurology: Genetics*, **2022**, 8, e652	3.8	1
38	UNC13B and focal epilepsy.. *Brain*, **2022**,	11.2	1
37	Infantile-onset myoclonic developmental and epileptic encephalopathy: A new RARS2 phenotype. *Epilepsia Open*, **2021**,	4	1
36	Protocol for a single patient therapy plan: A randomised, double-blind, placebo-controlled N-of-1 trial to assess the efficacy of cannabidiol in patients with intractable epilepsy. *Journal of Paediatrics and Child Health*, **2020**, 56, 1918-1923	1.3	1
35	Pathogenic MAST3 variants in the STK domain are associated with epilepsy		1
34	Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. *Epilepsia*, **2021**, 62, 1518-1527	6.4	1
33	Genetic convergence of developmental and epileptic encephalopathies and intellectual disability. *Developmental Medicine and Child Neurology*, **2021**, 63, 1441-1447	3.3	1
32	Pathogenic MAST3 Variants in the STK Domain Are Associated with Epilepsy. *Annals of Neurology*, **2021**, 90, 274-284	9.4	1
31	FBXO28 causes developmental and epileptic encephalopathy with profound intellectual disability. *Epilepsia*, **2021**, 62, e13-e21	6.4	1
30	Transcriptome analysis of a ring chromosome 20 patient cohort. *Epilepsia*, **2021**, 62, e22-e28	6.4	1
29	Improving Specificity of Cerebrospinal Fluid Liquid Biopsy for Genetic Testing. *Annals of Neurology*, **2021**, 90, 693-694	9.4	1
28	Dravet syndrome: A quick transition guide for the adult neurologist. *Epilepsy Research*, **2021**, 177, 106743	3	1
27	Postictal Psychosis in Epilepsy: A Clinicogenetic Study. *Annals of Neurology*, **2021**, 90, 464-476	9.4	1

26	Solving the Molecular Basis of the Developmental and Epileptic Encephalopathies: Are We there Yet?. *Epilepsy Currents*, **2021**, 21, 430-432	1.3	1
25	Common risk variants for epilepsy are enriched in families previously targeted for rare monogenic variant discovery. *EBioMedicine*, **2022**, 81, 104079	8.8	1
24	Author response: encephalopathy: A distinctive generalized developmental and epileptic encephalopathy. *Neurology*, **2020**, 94, 370	6.5	0
23	Genetic Epilepsies **2007**, 371-383		0
22	Using common genetic variants to find drugs for common epilepsies.. *Brain Communications*, **2021**, 3, fcab287	4.5	0
21	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
20	Speech, Language, and Oromotor Skills in Patients With Polymicrogyria. *Neurology*, **2021**, 96, e1898-e1912	6.5	0
19	The Key to FAME: Intronic Repeat Expansions Cause Human Epilepsies. *Epilepsy Currents*, **2018**, 18, 238-239	3.9	0
18	Enhanced Sensitivity to Angry Voices in People with Features of the Broader Autism Phenotype. *Journal of Autism and Developmental Disorders*, **2018**, 48, 3899-3911	4.6	0
17	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
16	Impaired Color Recognition in Epilepsy: A Single Case Report.. *Frontiers in Neurology*, **2022**, 13, 834252	4.1	0
15	Focal Epilepsy in Children With Tuberous Sclerosis Complex: Does Vigabatrin Control Focal Seizures?. *Journal of Child Neurology*, **2022**, 8830738211048326	2.5	0
14	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome.. *American Journal of Human Genetics*, **2022**, 109, 601-617	11	0
13	Letter to the Editor. *Journal of Paediatrics and Child Health*, **2021**,	1.3	0
12	PIK-ing the right gene for polymicrogyria. *Lancet Neurology, The*, **2015**, 14, 1147-8	24.1	
11	Quantitative analysis of phenotypic elements augments traditional electroclinical classification of common familial epilepsies. *Epilepsia*, **2019**, 60, 2194-2203	6.4	
10	Response to the numbering of seizure types. *Epilepsia*, **2017**, 58, 1300-1301	6.4	
9	Reply. *Annals of Neurology*, **2017**, 81, 328-329	9.4	

8	Genetic epilepsy with febrile seizures plus74-77	
7	Revidierte Terminologie und Konzepte zur Einteilung von epileptischen Anfällen und Epilepsien. *Zeitschrift Fur Epileptologie*, **2010**, 23, 227-237	0.1
6	Invited comments on the Shostak and Ottman review. *Epilepsia*, **2006**, 47, 1751-2; author reply 1755-6	6.4
5	Lightning progress in child neurology in the past 20 years.. *Lancet Neurology, The*, **2022**, 21, 111-113	24.1
4	Molecular epidemiology of monogenic epilepsies answers key clinical questions. *Brain*, **2019**, 142, 2173-2175	11.2
3	The Australian Academy of Health and Medical Sciences: an authoritative, independent voice in the Australian landscape. *Medical Journal of Australia*, **2021**, 214, 502-504.e1	4
2	Integrated in silico and experimental assessment of disease relevance of PCDH19 missense variants. *Human Mutation*, **2021**, 42, 1030-1041	4.7
1	009 Axonal excitability properties in dravet syndrome reflect effect of loss of sodium channels. *Journal of Neurology, Neurosurgery and Psychiatry*, **2019**, 90, A4.1-A4	5.5