Ingrid E Scheffer Mbbs

List of Publications by Year in Descending Order

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	Evidence for a Dual-Pathway, 2-Hit Genetic Model for Focal Cortical Dysplasia and Epilepsy.. *Neurology: Genetics*, **2022**, 8, e652	3.8	1
528	Development and Validation of a Prediction Model for Early Diagnosis of -Related Epilepsies.. *Neurology*, **2022**,	6.5	2
527	Lightning progress in child neurology in the past 20 years.. *Lancet Neurology, The*, **2022**, 21, 111-113	24.1	
526	UNC13B and focal epilepsy.. *Brain*, **2022**,	11.2	1
525	Genome-wide association study of febrile seizures implicates fever response and neuronal excitability genes.. *Brain*, **2022**,	11.2	4
524	Impaired Color Recognition in Epilepsy: A Single Case Report.. *Frontiers in Neurology*, **2022**, 13, 834252	4.1	0
523	Focal Epilepsy in Children With Tuberous Sclerosis Complex: Does Vigabatrin Control Focal Seizures?. *Journal of Child Neurology*, **2022**, 8830738211048326	2.5	0
522	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
521	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
520	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
519	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
518	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
517	Common risk variants for epilepsy are enriched in families previously targeted for rare monogenic variant discovery. *EBioMedicine*, **2022**, 81, 104079	8.8	1
516	Using common genetic variants to find drugs for common epilepsies.. *Brain Communications*, **2021**, 3, fcab287	4.5	0
515	Natural History Studies and Clinical Trial Readiness for Genetic Developmental and Epileptic Encephalopathies. *Neurotherapeutics*, **2021**, 18, 1432-1444	6.4	4
514	Infantile-onset myoclonic developmental and epileptic encephalopathy: A new RARS2 phenotype. *Epilepsia Open*, **2021**,	4	1
513	Phenotypic Spectrum of Seizure Disorders in MBD5-Associated Neurodevelopmental Disorder. *Neurology: Genetics*, **2021**, 7, e579	3.8	2

512	Climate change and epilepsy: Insights from clinical and basic science studies. *Epilepsy and Behavior*, **2021**, 116, 107791	3.2	7
511	Seizures in Sotos syndrome: Phenotyping in 49 patients. *Epilepsia Open*, **2021**, 6, 425-430	4	4
510	Guidelines on the diagnosis, clinical assessments, treatment and management for CLN2 disease patients. *Orphanet Journal of Rare Diseases*, **2021**, 16, 185	4.2	4
509	Diverse genetic causes of polymicrogyria with epilepsy. *Epilepsia*, **2021**, 62, 973-983	6.4	3
508	Cation leak underlies neuronal excitability in an HCN1 developmental and epileptic encephalopathy. *Brain*, **2021**, 144, 2060-2073	11.2	7
507	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
506	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. *Brain*, **2021**, 144, 1435-1450	11.2	4
505	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
504	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	
503	Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. *Epilepsia*, **2021**, 62, 1518-1527	6.4	1
502	Integrated in silico and experimental assessment of disease relevance of PCDH19´missense variants. *Human Mutation*, **2021**, 42, 1030-1041	4.7	
501	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
500	Severe speech impairment is a distinguishing feature of FOXP1-related disorder. *Developmental Medicine and Child Neurology*, **2021**, 63, 1417-1426	3.3	3
499	Genetic convergence of developmental and epileptic encephalopathies and intellectual disability. *Developmental Medicine and Child Neurology*, **2021**, 63, 1441-1447	3.3	1
498	Pathogenic MAST3 Variants in the STK Domain Are Associated with Epilepsy. *Annals of Neurology*, **2021**, 90, 274-284	9.4	1
497	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. *Genetics in Medicine*, **2021**, 23, 363-373	8.1	4
496	Medullary tyrosine hydroxylase catecholaminergic neuronal populations in sudden unexpected death in epilepsy. *Brain Pathology*, **2021**, 31, 133-143	6	8
495	Genetic Contributions to Acquired Epilepsies. *Epilepsy Currents*, **2021**, 21, 5-13	1.3	2

494	FBXO28 causes developmental and epileptic encephalopathy with profound intellectual disability. *Epilepsia*, **2021**, 62, e13-e21	6.4	1
493	Transcriptome analysis of a ring chromosome 20 patient cohort. *Epilepsia*, **2021**, 62, e22-e28	6.4	1
492	Cerebrospinal fluid liquid biopsy for detecting somatic mosaicism in brain. *Brain Communications*, **2021**, 3, fcaa235	4.5	17
491	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
490	The severe epilepsy syndromes of infancy: A population-based study. *Epilepsia*, **2021**, 62, 358-370	6.4	7
489	Contribution of rare genetic variants to drug response in absence epilepsy. *Epilepsy Research*, **2021**, 170, 106537	3	5
488	The aetiologies of epilepsy. *Epileptic Disorders*, **2021**, 23, 1-16	1.9	2
487	Speech, Language, and Oromotor Skills in Patients With Polymicrogyria. *Neurology*, **2021**, 96, e1898-e1912	6.5	0
486	Association of Missense Variants With Genetic Epilepsy With Febrile Seizures Plus. *Neurology*, **2021**, 96, e2251-e2260	6.5	3
485	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
484	Improving Specificity of Cerebrospinal Fluid Liquid Biopsy for Genetic Testing. *Annals of Neurology*, **2021**, 90, 693-694	9.4	1
483	Defining Dravet syndrome: An essential pre-requisite for precision medicine trials. *Epilepsia*, **2021**, 62, 2205-2217	6.4	9
482	Dravet syndrome: A quick transition guide for the adult neurologist. *Epilepsy Research*, **2021**, 177, 106743	3	1
481	Postictal Psychosis in Epilepsy: A Clinicogenetic Study. *Annals of Neurology*, **2021**, 90, 464-476	9.4	1
480	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
479	Precision Medicine Approaches for Infantile-Onset Developmental and Epileptic Encephalopathies. *Annual Review of Pharmacology and Toxicology*, **2021**,	17.9	3
478	Solving the Molecular Basis of the Developmental and Epileptic Encephalopathies: Are We there Yet?. *Epilepsy Currents*, **2021**, 21, 430-432	1.3	1
477	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

476	The phenotypic spectrum of X-linked, infantile onset ALG13-related developmental and epileptic encephalopathy. *Epilepsia*, **2021**, 62, 325-334	6.4	4
475	Letter to the Editor. *Journal of Paediatrics and Child Health*, **2021**,	1.3	0
474	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
473	A standardized patient-centered characterization of the phenotypic spectrum of PCDH19 girls clustering epilepsy. *Translational Psychiatry*, **2020**, 10, 127	8.6	12
472	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17 458 subjects. *Brain*, **2020**, 143, 2106-2118	11.2	14
471	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
470	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
469	Author response: encephalopathy: A distinctive generalized developmental and epileptic encephalopathy. *Neurology*, **2020**, 94, 370	6.5	0
468	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
467	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
466	Antiepileptic Drug Teratogenicity and De Novo Genetic Variation Load. *Annals of Neurology*, **2020**, 87, 897-906	9.4	5
465	Inherited RORB pathogenic variants: Overlap of photosensitive genetic generalized and occipital lobe epilepsy. *Epilepsia*, **2020**, 61, e23-e29	6.4	5
464	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
463	Defining the phenotype of FHF1 developmental and epileptic encephalopathy. *Epilepsia*, **2020**, 61, e71-e78	6.4	2
462	Pathogenic Variants in CEP85L Cause Sporadic and Familial Posterior Predominant Lissencephaly. *Neuron*, **2020**, 106, 237-245.e8	13.9	10
461	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
460	Severe childhood speech disorder: Gene discovery highlights transcriptional dysregulation. *Neurology*, **2020**, 94, e2148-e2167	6.5	28
459	Neuronal ceroid lipofuscinosis type 2: an Australian case series. *Journal of Paediatrics and Child Health*, **2020**, 56, 1210-1218	1.3	13

458	Keeping people with epilepsy safe during the COVID-19 pandemic. *Neurology*, **2020**, 94, 1032-1037	6.5	91
457	Parental health spillover effects of paediatric rare genetic conditions. *Quality of Life Research*, **2020**, 29, 2445-2454	3.7	6
456	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. *Nature Communications*, **2020**, 11, 4932	17.4	25
455	Cognitive processes predicting advanced theory of mind in the broader autism phenotype. *Autism Research*, **2020**, 13, 921-934	5.1	2
454	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
453	Deciphering the concepts behind "Epileptic encephalopathy" and "Developmental and epileptic encephalopathy". *European Journal of Paediatric Neurology*, **2020**, 24, 11-14	3.8	35
452	Levetiracetam efficacy in PCDH19 Girls Clustering Epilepsy. *European Journal of Paediatric Neurology*, **2020**, 24, 142-147	3.8	9
451	SCN1A Variants in vaccine-related febrile seizures: A prospective study. *Annals of Neurology*, **2020**, 87, 281-288	9.4	7
450	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
449	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
448	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
447	Cognitive, behavioral, and social functioning in children and adults with Dravet syndrome. *Epilepsy and Behavior*, **2020**, 112, 107319	3.2	10
446	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
445	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
444	Are Variants Causing Cardiac Arrhythmia Risk Factors in Sudden Unexpected Death in Epilepsy?. *Frontiers in Neurology*, **2020**, 11, 925	4.1	3
443	Developmental and epilepsy spectrum of KCNB1 encephalopathy with long-term outcome. *Epilepsia*, **2020**, 61, 2461-2473	6.4	4
442	PCDH19 Pathogenic Variants in Males: Expanding the Phenotypic Spectrum. *Advances in Experimental Medicine and Biology*, **2020**, 1298, 177-187	3.6	3
441	Cardiac phenotype in -related syndromes: A multicenter cohort study. *Neurology*, **2020**, 95, e2866-e2879	6.5	6

440	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
439	The epileptology of GNB5 encephalopathy. *Epilepsia*, **2019**, 60, e121-e127	6.4	9
438	Epidemiology and etiology of infantile developmental and epileptic encephalopathies in Tasmania. *Epilepsia Open*, **2019**, 4, 504-510	4	4
437	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
436	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
435	Efficacy and tolerability of adjunctive lacosamide in pediatric patients with focal seizures. *Neurology*, **2019**, 93, e1212-e1226	6.5	17
434	Spectrum of neurodevelopmental disease associated with the GNAO1 guanosine triphosphate-binding region. *Epilepsia*, **2019**, 60, 406-418	6.4	26
433	Speech and language in bilateral perisylvian polymicrogyria: a systematic review. *Developmental Medicine and Child Neurology*, **2019**, 61, 1145-1152	3.3	6
432	Autism and developmental disability caused by KCNQ3 gain-of-function variants. *Annals of Neurology*, **2019**, 86, 181-192	9.4	30
431	Double somatic mosaicism in a child with Dravet syndrome. *Neurology: Genetics*, **2019**, 5, e333	3.8	5
430	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
429	2017 International League Against Epilepsy classifications of seizures and epilepsy are steps in the right direction. *Epilepsia*, **2019**, 60, 1040-1044	6.4	9
428	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
427	Intestinal-Cell Kinase and Juvenile Myoclonic Epilepsy. *New England Journal of Medicine*, **2019**, 380, e24	59.2	4
426	Schizophrenia is a later-onset feature of PCDH19 Girls Clustering Epilepsy. *Epilepsia*, **2019**, 60, 429-440	6.4	11
425	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
424	Why should a neurologist worry about climate change?. *Lancet Neurology, The*, **2019**, 18, 335-336	24.1	2
423	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. *American Journal of Human Genetics*, **2019**, 104, 948-956	11	17

422	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. *Epilepsia*, **2019**, 60, 797-806	6.4	24
421	Dorsal language stream anomalies in an inherited speech disorder. *Brain*, **2019**, 142, 966-977	11.2	9
420	No evidence for a BRD2 promoter hypermethylation in blood leukocytes of Europeans with juvenile myoclonic epilepsy. *Epilepsia*, **2019**, 60, e31-e36	6.4	4
419	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
418	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
417	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
416	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. *Nature Communications*, **2019**, 10, 3094	17.4	76
415	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
414	Classification as autonomic versus sensory seizures. *Epilepsia*, **2019**, 60, 2003-2005	6.4	3
413	Somatic mutation: The hidden genetics of brain malformations and focal epilepsies. *Epilepsy Research*, **2019**, 155, 106161	3	24
412	Quantitative analysis of phenotypic elements augments traditional electroclinical classification of common familial epilepsies. *Epilepsia*, **2019**, 60, 2194-2203	6.4	
411	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. *Nature Communications*, **2019**, 10, 4679	17.4	21
410	When Monogenic Isn't Monogenic-Unravelling the Oligogenic Architecture of the Developmental and Epileptic Encephalopathies. *Epilepsy Currents*, **2019**, 19, 417-419	1.3	4
409	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. *Nature Communications*, **2019**, 10, 4920	17.4	48
408	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. *Annals of Neurology*, **2019**, 86, 821-831	9.4	55
407	Molecular epidemiology of monogenic epilepsies answers key clinical questions. *Brain*, **2019**, 142, 2173-2175	11.2	
406	SLC35A2-CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. *Human Mutation*, **2019**, 40, 908-925	4.7	23
405	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	

404	Human generalized epilepsy: Increased somatosensory and striatothalamic connectivity. *Neurology: Genetics*, **2019**, 5, e340	3.8	3
403	Fragile Females: Case Series of Epilepsy in Girls With Disruption. *Pediatrics*, **2019**, 144,	7.4	4
402	Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3. *Nature Communications*, **2019**, 10, 4919	17.4	58
401	SCN1A-related phenotypes: Epilepsy and beyond. *Epilepsia*, **2019**, 60 Suppl 3, S17-S24	6.4	45
400	Reanalysis and optimisation of bioinformatic pipelines is critical for mutation detection. *Human Mutation*, **2019**, 40, 374-379	4.7	6
399	encephalopathy: A distinctive generalized developmental and epileptic encephalopathy. *Neurology*, **2019**, 92, e96-e107	6.5	55
398	Efficacy of cannabinoids in paediatric epilepsy. *Developmental Medicine and Child Neurology*, **2019**, 61, 13-18	3.3	22
397	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
396	Stiripentol efficacy and safety in Dravet syndrome: a 12-year observational study. *Developmental Medicine and Child Neurology*, **2018**, 60, 574-578	3.3	38
395	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
394	Childhood-onset generalized epilepsy in Bainbridge-Ropers syndrome. *Epilepsy Research*, **2018**, 140, 166-170	3	7
393	Early mortality in SCN8A-related epilepsies. *Epilepsy Research*, **2018**, 143, 79-81	3	38
392	Parental Mosaicism in "De Novo" Epileptic Encephalopathies. *New England Journal of Medicine*, **2018**, 378, 1646-1648	59.2	67
391	The ventrolateral medulla and medullary raphe in sudden unexpected death in epilepsy. *Brain*, **2018**, 141, 1719-1733	11.2	56
390	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. *Annals of Neurology*, **2018**, 83, 926-934	9.4	11
389	Hemiconvulsion-hemiplegia-epilepsy evolving to contralateral hemi-Lennox-Gastaut-like phenotype. *Brain and Development*, **2018**, 40, 425-428	2.2	3
388	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
387	Ion Channels in Genetic Epilepsy: From Genes and Mechanisms to Disease-Targeted Therapies. *Pharmacological Reviews*, **2018**, 70, 142-173	22.5	103

386	Myoclonic absence seizures with complex gestural automatisms. *European Journal of Paediatric Neurology*, **2018**, 22, 532-535	3.8	3
385	ADGRV1 is implicated in myoclonic epilepsy. *Epilepsia*, **2018**, 59, 381-388	6.4	23
384	Abnormal Cell Sorting Underlies the Unique X-Linked Inheritance of PCDH19 Epilepsy. *Neuron*, **2018**, 97, 59-66.e5	13.9	58
383	Epilepsy. *Nature Reviews Disease Primers*, **2018**, 4, 18024	51.1	269
382	Gain-of-function HCN2 variants in genetic epilepsy. *Human Mutation*, **2018**, 39, 202-209	4.7	19
381	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
380	Analysis of shared heritability in common disorders of the brain. *Science*, **2018**, 360,	33.3	666
379	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
378	Infantile Spasms of Unknown Cause: Predictors of Outcome and Genotype-Phenotype Correlation. *Pediatric Neurology*, **2018**, 87, 48-56	2.9	22
377	A population-based cost-effectiveness study of early genetic testing in severe epilepsies of infancy. *Epilepsia*, **2018**, 59, 1177-1187	6.4	46
376	Somatic mutation in the of Sturge-Weber syndrome. *Neurology: Genetics*, **2018**, 4, e236	3.8	18
375	Genetic literacy series: genetic epilepsy with febrile seizures plus. *Epileptic Disorders*, **2018**, 20, 232-238	1.9	30
374	Therapeutic use of medicinal cannabis in difficult to manage epilepsy. *British Journal of Clinical Pharmacology*, **2018**, 84, 2488-2490	3.8	5
373	Heart rate variability in epilepsy: A potential biomarker of sudden unexpected death in epilepsy risk. *Epilepsia*, **2018**, 59, 1372-1380	6.4	68
372	KANSL1 variation is not a major contributing factor in self-limited focal epilepsy syndromes of childhood. *PLoS ONE*, **2018**, 13, e0191546	3.7	3
371	The management of epilepsy in children and adults. *Medical Journal of Australia*, **2018**, 208, 226-233	4	58
370	Severe infantile onset developmental and epileptic encephalopathy caused by mutations in autophagy gene WDR45. *Epilepsia*, **2018**, 59, e5-e13	6.4	34
369	A new classification and class 1 evidence transform clinical practice in epilepsy. *Lancet Neurology, The*, **2018**, 17, 7-8	24.1	4

368	Clinical and molecular characterization of -related severe early-onset epilepsy. *Neurology*, **2018**, 90, e55-e66	6.5	53
367	Precision therapy for epilepsy due to mutations: A randomized trial of oral quinidine. *Neurology*, **2018**, 90, e67-e72	6.5	84
366	The new definition and classification of seizures and epilepsy. *Epilepsy Research*, **2018**, 139, 73-79	3	137
365	Sleep problems in Dravet syndrome: a modifiable comorbidity. *Developmental Medicine and Child Neurology*, **2018**, 60, 192-198	3.3	28
364	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
363	De novo variants in the alternative exon 5 of SCN8A cause epileptic encephalopathy. *Genetics in Medicine*, **2018**, 20, 275-281	8.1	13
362	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
361	Anleitung ([Instruction manual]) zur Anwendung der operationalen Klassifikation von Anfallsformen der ILAE 2017. *Zeitschrift Fur Epileptologie*, **2018**, 31, 282-295	0.1	10
360	The Key to FAME: Intronic Repeat Expansions Cause Human Epilepsies. *Epilepsy Currents*, **2018**, 18, 238-239	2.3	0
359	ILAE-Klassifikation der Epilepsien: Positionspapier der ILAE-Kommission für Klassifikation und Terminologie. *Zeitschrift Fur Epileptologie*, **2018**, 31, 296-306	0.1	17
358	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
357	Development of a rapid functional assay that predicts GLUT1 disease severity. *Neurology: Genetics*, **2018**, 4, e297	3.8	5
356	A Primate-Specific Isoform of PLEKHG6 Regulates Neurogenesis and Neuronal Migration. *Cell Reports*, **2018**, 25, 2729-2741.e6	10.6	27
355	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
354	Genome-wide mega-analysis identifies 16 loci and highlights diverse biological mechanisms in the common epilepsies. *Nature Communications*, **2018**, 9, 5269	17.4	169
353	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
352	The phenotype of developmental and epileptic encephalopathy. *Neurology*, **2018**, 91, e1112-e1124	6.5	80
351	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

350	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
349	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
348	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
347	Dysarthria and broader motor speech deficits in Dravet syndrome. *Neurology*, **2017**, 88, 743-749	6.5	19
346	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
345	Ultra-rare genetic variation in common epilepsies: a case-control sequencing study. *Lancet Neurology, The*, **2017**, 16, 135-143	24.1	133
344	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
343	De novo SCN1A pathogenic variants in the GEFS+ spectrum: Not always a familial syndrome. *Epilepsia*, **2017**, 58, e26-e30	6.4	21
342	Frequency of CNKSR2 mutation in the X-linked epilepsy-aphasia spectrum. *Epilepsia*, **2017**, 58, e40-e43	6.4	15
341	Myoclonic Absence Seizures in Dravet Syndrome. *Pediatric Neurology*, **2017**, 70, 67-69	2.9	9
340	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
339	Instruction manual for the ILAE 2017 operational classification of seizure types. *Epilepsia*, **2017**, 58, 531-542	6.4	425
338	ILAE classification of the epilepsies: Position paper of the ILAE Commission for Classification and Terminology. *Epilepsia*, **2017**, 58, 512-521	6.4	2084
337	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. *Nature Genetics*, **2017**, 49, 515-526	36.3	283
336	Real-world utility of whole exome sequencing with targeted gene analysis for focal epilepsy. *Epilepsy Research*, **2017**, 131, 1-8	3	63
335	Evaluation of GLUT1 variation in non-acquired focal epilepsy. *Epilepsy Research*, **2017**, 133, 54-57	3	4
334	DEPDC5 as a potential therapeutic target for epilepsy. *Expert Opinion on Therapeutic Targets*, **2017**, 21, 591-600	6.4	23
333	The epileptology of Koolen-de Vries syndrome: Electro-clinico-radiologic findings in 31 patients. *Epilepsia*, **2017**, 58, 1085-1094	6.4	8

332	Familial epilepsy with anterior polymicrogyria as a presentation of COL18A1 mutations. *European Journal of Medical Genetics*, **2017**, 60, 437-443	2.6	9
331	Trial of Cannabidiol for Drug-Resistant Seizures in the Dravet Syndrome. *New England Journal of Medicine*, **2017**, 376, 2011-2020	59.2	771
330	Fatal Cerebral Edema With Status Epilepticus in Children With Dravet Syndrome: Report of 5 Cases. *Pediatrics*, **2017**, 139,	7.4	21
329	Is a microRNA-328 binding site in associated with Rolandic epilepsy?. *Annals of Clinical and Translational Neurology*, **2017**, 4, 276-277	5.3	1
328	encephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. *Journal of Medical Genetics*, **2017**, 54, 460-470	5.8	109
327	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
326	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. *American Journal of Human Genetics*, **2017**, 101, 664-685	11	214
325	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. *American Journal of Human Genetics*, **2017**, 101, 516-524	11	29
324	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
323	Genetic epilepsy with febrile seizures plus: Refining the spectrum. *Neurology*, **2017**, 89, 1210-1219	6.5	68
322	Familial mesial temporal lobe epilepsy and the borderland of dJ□vu. *Annals of Neurology*, **2017**, 82, 166-176	9.4	12
321	Response to the numbering of seizure types. *Epilepsia*, **2017**, 58, 1300-1301	6.4	
320	The effect of the ketogenic diet on the developing skeleton. *Epilepsy Research*, **2017**, 136, 62-66	3	30
319	Reply. *Annals of Neurology*, **2017**, 81, 328-329	9.4	
318	Not all epileptic encephalopathies are Dravet syndrome: Early profound Thr226Met phenotype. *Neurology*, **2017**, 89, 1035-1042	6.5	62
317	How long for epilepsy remission in the ILAE definition?. *Epilepsia*, **2017**, 58, 1486-1487	6.4	3
316	encephalopathy: A new disease of vesicle fission. *Neurology*, **2017**, 89, 385-394	6.5	46
315	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

314	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
313	Mortality in Dravet syndrome. *Epilepsy Research*, **2016**, 128, 43-47	3	152
312	TBC1D24 genotype-phenotype correlation: Epilepsies and other neurologic features. *Neurology*, **2016**, 87, 77-85	6.5	75
311	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
310	A mutation in COL4A2 causes autosomal dominant porencephaly with cataracts. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 1059-63	2.5	13
309	Disruptive de novo mutations of DYRK1A lead to a syndromic form of autism and ID. *Molecular Psychiatry*, **2016**, 21, 126-32	15.1	90
308	Multiplex families with epilepsy: Success of clinical and molecular genetic characterization. *Neurology*, **2016**, 86, 713-22	6.5	22
307	The genetic landscape of the epileptic encephalopathies of infancy and childhood. *Lancet Neurology, The*, **2016**, 15, 304-16	24.1	317
306	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
305	SCN8A encephalopathy: Research progress and prospects. *Epilepsia*, **2016**, 57, 1027-35	6.4	82
304	Periventricular Nodular Heterotopia: Detection of Abnormal Microanatomic Fiber Structures with Whole-Brain Diffusion MR Imaging Tractography. *Radiology*, **2016**, 281, 896-906	20.5	13
303	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
302	In silico prioritization based on coexpression can aid epileptic encephalopathy gene discovery. *Neurology: Genetics*, **2016**, 2, e51	3.8	15
301	Evaluation of non-coding variation in GLUT1 deficiency. *Developmental Medicine and Child Neurology*, **2016**, 58, 1295-1302	3.3	19
300	Is FGF13 a major contributor to genetic epilepsy with febrile seizures plus?. *Epilepsy Research*, **2016**, 128, 48-51	3	6
299	Epilepsy in KCNH1-related syndromes. *Epileptic Disorders*, **2016**, 18, 123-36	1.9	21
298	Early neuroimaging markers of FOXP2 intragenic deletion. *Scientific Reports*, **2016**, 6, 35192	4.9	15
297	A targeted resequencing gene panel for focal epilepsy. *Neurology*, **2016**, 86, 1605-12	6.5	37

296	Definition and diagnostic criteria of sleep-related hypermotor epilepsy. *Neurology*, **2016**, 86, 1834-42	6.5	182
295	Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hypotonia, and Seizures. *American Journal of Human Genetics*, **2016**, 98, 1001-1010	11	70
294	Dominant KCNA2 mutation causes episodic ataxia and pharmacoresponsive epilepsy. *Neurology*, **2016**, 87, 1975-1984	6.5	50
293	De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies. *American Journal of Human Genetics*, **2016**, 99, 287-98	11	180
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	Pitfalls in genetic testing: the story of missed SCN1A mutations. *Molecular Genetics & Genomic Medicine*, **2016**, 4, 457-64	2.3	50
290	Lamotrigine can be beneficial in patients with Dravet syndrome. *Developmental Medicine and Child Neurology*, **2015**, 57, 200-2	3.3	37
289	CHD2 variants are a risk factor for photosensitivity in epilepsy. *Brain*, **2015**, 138, 1198-207	11.2	81
288	Myoclonic occipital photosensitive epilepsy with dystonia (MOPED): A familial epilepsy syndrome. *Epilepsy Research*, **2015**, 114, 98-105	3	7
287	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. *Human Molecular Genetics*, **2015**, 24, 5250-9	5.6	78
286	Seizures are regulated by ubiquitin-specific peptidase 9 X-linked (USP9X), a de-ubiquitinase. *PLoS Genetics*, **2015**, 11, e1005022	6	49
285	Early and effective treatment of KCNQ2 encephalopathy. *Epilepsia*, **2015**, 56, 685-91	6.4	160
284	CHD2 myoclonic encephalopathy is frequently associated with self-induced seizures. *Neurology*, **2015**, 84, 951-8	6.5	57
283	New genes for focal epilepsies with speech and language disorders. *Current Neurology and Neuroscience Reports*, **2015**, 15, 35	6.6	43
282	"It's good to know": experiences of gene identification and result disclosure in familial epilepsies. *Epilepsy Research*, **2015**, 112, 64-71	3	9
281	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
280	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. *American Journal of Human Genetics*, **2015**, 96, 808-15	11	114
279	Mutations in SLC12A5 in epilepsy of infancy with migrating focal seizures. *Nature Communications*, **2015**, 6, 8038	17.4	104

278	Evaluation of multiple putative risk alleles within the 15q13.3 region for genetic generalized epilepsy. *Epilepsy Research*, **2015**, 117, 70-3	3	3
277	A roadmap for precision medicine in the epilepsies. *Lancet Neurology, The*, **2015**, 14, 1219-28	24.1	119
276	PIK-ing the right gene for polymicrogyria. *Lancet Neurology, The*, **2015**, 14, 1147-8	24.1	
275	Mutations in KCNT1 cause a spectrum of focal epilepsies. *Epilepsia*, **2015**, 56, e114-20	6.4	83
274	SCN2A encephalopathy: A major cause of epilepsy of infancy with migrating focal seizures. *Neurology*, **2015**, 85, 958-66	6.5	163
273	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. *Nature Genetics*, **2015**, 47, 39-46	36.3	177
272	PRIMA1 mutation: a new cause of nocturnal frontal lobe epilepsy. *Annals of Clinical and Translational Neurology*, **2015**, 2, 821-30	5.3	19
271	Copy number variant analysis from exome data in 349 patients with epileptic encephalopathy. *Annals of Neurology*, **2015**, 78, 323-8	9.4	44
270	Loss of synaptic Zn2+ transporter function increases risk of febrile seizures. *Scientific Reports*, **2015**, 5, 17816	4.9	29
269	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. *EBioMedicine*, **2015**, 2, 1063-70	8.8	61
268	Epilepsy with auditory features: A heterogeneous clinico-molecular disease. *Neurology: Genetics*, **2015**, 1, e5	3.8	38
267	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
266	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
265	Familial neonatal seizures in 36 families: Clinical and genetic features correlate with outcome. *Epilepsia*, **2015**, 56, 1071-80	6.4	68
264	Epileptic spasms are a feature of DEPDC5 mTORopathy. *Neurology: Genetics*, **2015**, 1, e17	3.8	44
263	Quinidine in the treatment of KCNT1-positive epilepsies. *Annals of Neurology*, **2015**, 78, 995-9	9.4	145
262	Vaccination Triggers, Rather Than Causes, Seizures. *Epilepsy Currents*, **2015**, 15, 335-7	1.3	9
261	Mutation of the nuclear lamin gene LMNB2 in progressive myoclonus epilepsy with early ataxia. *Human Molecular Genetics*, **2015**, 24, 4483-90	5.6	33

260	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
259	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
258	Faulty cardiac repolarization reserve in alternating hemiplegia of childhood broadens the phenotype. *Brain*, **2015**, 138, 2859-74	11.2	26
257	Variants in KCNJ11 and BAD do not predict response to ketogenic dietary therapies for epilepsy. *Epilepsy Research*, **2015**, 118, 22-8	3	6
256	GRIN2A: an aptly named gene for speech dysfunction. *Neurology*, **2015**, 84, 586-93	6.5	44
255	The phenotypic spectrum of SCN8A encephalopathy. *Neurology*, **2015**, 84, 480-9	6.5	199
254	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
253	Reduced dendritic arborization and hyperexcitability of pyramidal neurons in a Scn1b-based model of Dravet syndrome. *Brain*, **2014**, 137, 1701-15	11.2	34
252	ILAE official report: a practical clinical definition of epilepsy. *Epilepsia*, **2014**, 55, 475-82	6.4	2408
251	Refining analyses of copy number variation identifies specific genes associated with developmental delay. *Nature Genetics*, **2014**, 46, 1063-71	36.3	429
250	Epilepsy: Beyond the single nucleotide variant in epilepsy genetics. *Nature Reviews Neurology*, **2014** , 10, 490-1	15	10
249	De novo mutations in synaptic transmission genes including DNM1 cause epileptic encephalopathies. *American Journal of Human Genetics*, **2014**, 95, 360-70	11	299
248	GABRA1 and STXBP1: novel genetic causes of Dravet syndrome. *Neurology*, **2014**, 82, 1245-53	6.5	180
247	Somatic mutations in cerebral cortical malformations. *New England Journal of Medicine*, **2014**, 371, 733-43	59.2	265
246	Genetic determinants of common epilepsies: a meta-analysis of genome-wide association studies. *Lancet Neurology, The*, **2014**, 13, 893-903	24.1	194
245	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
244	Harnessing gene expression networks to prioritize candidate epileptic encephalopathy genes. *PLoS ONE*, **2014**, 9, e102079	3.7	22
243	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. *Human Molecular Genetics*, **2014**, 23, 6069-80	5.6	45

242	Genetic analysis of PHOX2B in sudden unexpected death in epilepsy cases. *Neurology*, **2014**, 83, 1018-21	6.5	16
241	Epilepsy genetics revolutionizes clinical practice. *Neuropediatrics*, **2014**, 45, 70-4	1.6	38
240	Dominant-negative effects of KCNQ2 mutations are associated with epileptic encephalopathy. *Annals of Neurology*, **2014**, 75, 382-94	9.4	176
239	KCNT1 gain of function in 2 epilepsy phenotypes is reversed by quinidine. *Annals of Neurology*, **2014**, 75, 581-90	9.4	192
238	Transition to adult life in the monogenic epilepsies. *Epilepsia*, **2014**, 55 Suppl 3, 12-5	6.4	6
237	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
236	Is focal cortical dysplasia sporadic? Family evidence for genetic susceptibility. *Epilepsia*, **2014**, 55, e22-6	6.4	22
235	Glucose metabolism transporters and epilepsy: only GLUT1 has an established role. *Epilepsia*, **2014**, 55, e18-21	6.4	23
234	Mutations in mammalian target of rapamycin regulator DEPDC5 cause focal epilepsy with brain malformations. *Annals of Neurology*, **2014**, 75, 782-7	9.4	153
233	Genetics of epilepsy: The testimony of twins in the molecular era. *Neurology*, **2014**, 83, 1042-8	6.5	46
232	Does variation in NIPA2 contribute to genetic generalized epilepsy?. *Human Genetics*, **2014**, 133, 673-4	6.3	6
231	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
230	De novo mutations in epileptic encephalopathies. *Nature*, **2013**, 501, 217-21	50.4	1081
229	GRIN2A mutations cause epilepsy-aphasia spectrum disorders. *Nature Genetics*, **2013**, 45, 1073-6	36.3	249
228	Recent advances in the molecular genetics of epilepsy. *Journal of Medical Genetics*, **2013**, 50, 271-9	5.8	82
227	Do mutations in SCN1B cause Dravet syndrome?. *Epilepsy Research*, **2013**, 103, 97-100	3	9
226	Role of the sodium channel SCN9A in genetic epilepsy with febrile seizures plus and Dravet syndrome. *Epilepsia*, **2013**, 54, e122-6	6.4	46
225	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

224	Epilepsy in 2012: Advances in epilepsy shed light on key questions. *Nature Reviews Neurology*, **2013**, 9, 66-8	15	2
223	Copy number variants are frequent in genetic generalized epilepsy with intellectual disability. *Neurology*, **2013**, 81, 1507-14	6.5	115
222	Seizure semiology in autosomal dominant epilepsy with auditory features, due to novel LGI1 mutations. *Epilepsy Research*, **2013**, 107, 311-7	3	15
221	Racial and ethnic differences in epilepsy classification among probands in the Epilepsy Phenome/Genome Project (EPGP). *Epilepsy Research*, **2013**, 107, 306-10	3	6
220	The clinical utility of an SCN1A genetic diagnosis in infantile-onset epilepsy. *Developmental Medicine and Child Neurology*, **2013**, 55, 154-61	3.3	50
219	Mutations in DEPDC5 cause familial focal epilepsy with variable foci. *Nature Genetics*, **2013**, 45, 546-51	36.3	238
218	SCN1A testing for epilepsy: application in clinical practice. *Epilepsia*, **2013**, 54, 946-52	6.4	54
217	Genetics of idiopathic epilepsies. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2013**, 111, 567-78	3	12
216	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
215	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. *Nature Genetics*, **2013**, 45, 825-30	36.3	500
214	Genetics of febrile seizure subtypes and syndromes: a twin study. *Epilepsy Research*, **2013**, 105, 103-9	3	31
213	SLC25A22 is a novel gene for migrating partial seizures in infancy. *Annals of Neurology*, **2013**, 74, 873-82	9.4	89
212	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. *Brain*, **2013**, 136, 3378-94	11.2	65
211	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
210	Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. *Brain*, **2013**, 136, 3140-50	11.2	144
209	Autosomal dominant vasovagal syncope: clinical features and linkage to chromosome 15q26. *Neurology*, **2013**, 80, 1485-93	6.5	15
208	Etiology of hippocampal sclerosis: evidence for a predisposing familial morphologic anomaly. *Neurology*, **2013**, 81, 144-9	6.5	45
207	Epilepsy classification: a cycle of evolution and revolution. *Current Opinion in Neurology*, **2013**, 26, 163-7	7.1	20

206	Migrating partial seizures of infancy: expansion of the electroclinical, radiological and pathological disease spectrum. *Brain*, **2013**, 136, 1578-91	11.2	120
205	Genetics of epilepsy syndromes in families with photosensitivity. *Neurology*, **2013**, 80, 1322-9	6.5	34
204	Clinical genetic study of the epilepsy-aphasia spectrum. *Epilepsia*, **2013**, 54, 280-7	6.4	34
203	Siblings with refractory occipital epilepsy showing localized network activity on EEG-fMRI. *Epilepsia*, **2013**, 54, e28-32	6.4	4
202	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
201	Lennox-Gastaut syndrome of unknown cause: phenotypic characteristics of patients in the Epilepsy Phenome/Genome Project. *Epilepsia*, **2013**, 54, 1898-904	6.4	15
200	Mutations in TNK2 in severe autosomal recessive infantile onset epilepsy. *Annals of Neurology*, **2013**, 74, 496-501	9.4	18
199	Mutations in PRRT2 are not a common cause of infantile epileptic encephalopathies. *Epilepsia*, **2013** , 54, e86-9	6.4	11
198	Head stereotypies in STXBP1 encephalopathy. *Developmental Medicine and Child Neurology*, **2013**, 55, 769-72	3.3	11
197	Extending the KCNQ2 encephalopathy spectrum: clinical and neuroimaging findings in 17 patients. *Neurology*, **2013**, 81, 1697-703	6.5	151
196	The epilepsy phenome/genome project. *Clinical Trials*, **2013**, 10, 568-86	2.2	34
195	Febrile infection-related epilepsy syndrome is not caused by SCN1A mutations. *Epilepsy Research*, **2012**, 100, 194-8	3	8
194	Efficacy of the ketogenic diet: which epilepsies respond?. *Epilepsia*, **2012**, 53, e55-9	6.4	56
193	Epilepsy: a classification for all seasons?. *Epilepsia*, **2012**, 53 Suppl 2, 6-9	6.4	10
192	KCNQ2 encephalopathy: emerging phenotype of a neonatal epileptic encephalopathy. *Annals of Neurology*, **2012**, 71, 15-25	9.4	322
191	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
190	Epi4K: gene discovery in 4,000 genomes. *Epilepsia*, **2012**, 53, 1457-67	6.4	63
189	Homozygous PLCB1 deletion associated with malignant migrating partial seizures in infancy. *Epilepsia*, **2012**, 53, e146-50	6.4	93

188	Familial focal epilepsy with variable foci mapped to chromosome 22q12: expansion of the phenotypic spectrum. *Epilepsia*, **2012**, 53, e151-5	6.4	18
187	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
186	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
185	Early onset absence epilepsy: 1 in 10 cases is caused by GLUT1 deficiency. *Epilepsia*, **2012**, 53, e204-7	6.4	75
184	Glucose transporter 1 deficiency in the idiopathic generalized epilepsies. *Annals of Neurology*, **2012**, 72, 807-15	9.4	98
183	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
182	Peritrigonal and temporo-occipital heterotopia with corpus callosum and cerebellar dysgenesis. *Neurology*, **2012**, 79, 1244-51	6.5	25
181	Is the ketogenic diet effective in specific epilepsy syndromes?. *Epilepsy Research*, **2012**, 100, 252-7	3	60
180	PRRT2 phenotypic spectrum includes sporadic and fever-related infantile seizures. *Neurology*, **2012**, 79, 2104-8	6.5	65
179	Family studies of individuals with eyelid myoclonia with absences. *Epilepsia*, **2012**, 53, 2141-8	6.4	19
178	Rare protein sequence variation in SV2A gene does not affect response to levetiracetam. *Epilepsy Research*, **2012**, 101, 277-9	3	10
177	Diagnosis and long-term course of Dravet syndrome. *European Journal of Paediatric Neurology*, **2012**, 16 Suppl 1, S5-8	3.8	44
176	De novo mutations in ATP1A3 cause alternating hemiplegia of childhood. *Nature Genetics*, **2012**, 44, 1030[illegible]	36.3	280
175	PRRT2 mutations cause benign familial infantile epilepsy and infantile convulsions with choreoathetosis syndrome. *American Journal of Human Genetics*, **2012**, 90, 152-60	11	199
174	Long-term follow-up of febrile infection-related epilepsy syndrome. *Epilepsia*, **2012**, 53, 101-10	6.4	76
173	Clinical genetic studies in benign childhood epilepsy with centrotemporal spikes. *Epilepsia*, **2012**, 53, 319-24	6.4	38
172	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
171	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

170	Progressive gait deterioration in adolescents with Dravet syndrome. *Archives of Neurology*, **2012**, 69, 873-8		61
169	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
168	Familial adult myoclonic epilepsy: recognition of mild phenotypes and refinement of the 2q locus. *Archives of Neurology*, **2012**, 69, 474-81		25
167	Mirror neuron system involvement in empathy: a critical look at the evidence. *Social Neuroscience*, **2011**, 6, 327-35	2	76
166	Genetic testing in epilepsy: what should you be doing?. *Epilepsy Currents*, **2011**, 11, 107-11	1.3	30
165	The Role of Seizure-Related SEZ6 as a Susceptibility Gene in Febrile Seizures. *Neurology Research International*, **2011**, 2011, 917565	1.7	15
164	Milder phenotypes of glucose transporter type 1 deficiency syndrome. *Developmental Medicine and Child Neurology*, **2011**, 53, 664-8	3.3	12
163	Genetics of the epilepsies: channelopathies and beyond. *Epilepsia*, **2011**, 52, 192-3; discussion 193-6	6.4	3
162	The genetics of Dravet syndrome. *Epilepsia*, **2011**, 52 Suppl 2, 24-9	6.4	170
161	Autoantibodies and epilepsy. *Epilepsia*, **2011**, 52 Suppl 3, 18-22	6.4	71
160	New concepts in classification of the epilepsies: entering the 21st century. *Epilepsia*, **2011**, 52, 1058-62	6.4	208
159	What is at stake in a classification?. *Epilepsia*, **2011**, 52, 1205-1208	6.4	9
158	A retrospective population-based study on seizures related to childhood vaccination. *Epilepsia*, **2011**, 52, 1506-12	6.4	14
157	Investigation of the 15q13.3 CNV as a genetic modifier for familial epilepsies with variable phenotypes. *Epilepsia*, **2011**, 52, e139-42	6.4	8
156	A novel GEFS+ locus on 12p13.33 in a large Roma family. *Epilepsy Research*, **2011**, 97, 198-207	3	5
155	Rare copy number variants are an important cause of epileptic encephalopathies. *Annals of Neurology*, **2011**, 70, 974-85	9.4	176
154	Electroclinical features of absence seizures in sleep. *Epilepsy Research*, **2011**, 93, 216-20	3	12
153	Randomized, phase III study results of clobazam in Lennox-Gastaut syndrome. *Neurology*, **2011**, 77, 1473-81	6.5	133

152	Dravet syndrome as epileptic encephalopathy: evidence from long-term course and neuropathology. *Brain*, **2011**, 134, 2982-3010	11.2	143
151	Does genotype determine phenotype? Sodium channel mutations in Dravet syndrome and GEFS+. *Neurology*, **2011**, 76, 588-9	6.5	10
150	Molecular analysis of ring chromosome 20 syndrome reveals two distinct groups of patients. *Journal of Medical Genetics*, **2011**, 48, 1-9	5.8	48
149	Glucose transporter 1 deficiency as a treatable cause of myoclonic astatic epilepsy. *Archives of Neurology*, **2011**, 68, 1152-5		110
148	Copy number variants--an unexpected risk factor for the idiopathic generalized epilepsies. *Brain*, **2010**, 133, 7-8	11.2	16
147	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
146	Timing of de novo mutagenesis--a twin study of sodium-channel mutations. *New England Journal of Medicine*, **2010**, 363, 1335-40	59.2	78
145	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
144	The core network in absence epilepsy. Differences in cortical and thalamic BOLD response. *Neurology*, **2010**, 75, 904-11	6.5	109
143	Absence epilepsies with widely variable onset are a key feature of familial GLUT1 deficiency. *Neurology*, **2010**, 75, 432-40	6.5	133
142	Clinical and imaging heterogeneity of polymicrogyria: a study of 328 patients. *Brain*, **2010**, 133, 1415-27	11.2	177
141	Familial mesial temporal lobe epilepsy: a benign epilepsy syndrome showing complex inheritance. *Brain*, **2010**, 133, 3221-31	11.2	59
140	Phospholipase C beta 1 deficiency is associated with early-onset epileptic encephalopathy. *Brain*, **2010**, 133, 2964-70	11.2	81
139	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
138	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
137	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
136	Detection of microchromosomal aberrations in refractory epilepsy: a pilot study. *Epileptic Disorders*, **2010**, 12, 192-8	1.9	12
135	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

134	Optimizing electroencephalographic studies for epilepsy diagnosis in children with new-onset seizures. *Archives of Neurology*, **2010**, 67, 1345-9		6
133	Mosaicism of a missense SCN1A mutation and Dravet syndrome in a Roma/Gypsy family. *Epileptic Disorders*, **2010**, 12, 117-24	1.9	14
132	Revidierte Terminologie und Konzepte zur Einteilung von epileptischen Anfllen und Epilepsien. *Zeitschrift Fur Epileptologie*, **2010**, 23, 227-237	0.1	
131	Effects of vaccination on onset and outcome of Dravet syndrome: a retrospective study. *Lancet Neurology, The*, **2010**, 9, 592-8	24.1	95
130	Vaccination and Dravet syndrome Authors' reply. *Lancet Neurology, The*, **2010**, 9, 1148-1149	24.1	1
129	Augmented currents of an HCN2 variant in patients with febrile seizure syndromes. *Annals of Neurology*, **2010**, 67, 542-6	9.4	75
128	Whole-genome linkage scan for epilepsy-related photosensitivity: a mega-analysis. *Epilepsy Research*, **2010**, 89, 286-94	3	40
127	Neonatal seizures and long QT syndrome: a cardiocerebral channelopathy?. *Epilepsia*, **2010**, 51, 293-6	6.4	52
126	Genetic testing in the epilepsies--report of the ILAE Genetics Commission. *Epilepsia*, **2010**, 51, 655-70	6.4	147
125	Genetics of the epilepsies: genetic twists in the channels and other tales. *Epilepsia*, **2010**, 51 Suppl 1, 33-6	6.4	5
124	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
123	Familial neonatal seizures with intellectual disability caused by a microduplication of chromosome 2q24.3. *Epilepsia*, **2010**, 51, 1865-9	6.4	25
122	A duplication in 1q21.3 in a family with early onset and childhood absence epilepsy. *Epilepsia*, **2010**, 51, 2453-6	6.4	12
121	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
120	Automatisms in absence seizures in children with idiopathic generalized epilepsy. *Archives of Neurology*, **2009**, 66, 729-34		22
119	Dravet syndrome or genetic (generalized) epilepsy with febrile seizures plus?. *Brain and Development*, **2009**, 31, 394-400	2.2	129
118	Early-onset absence epilepsy caused by mutations in the glucose transporter GLUT1. *Annals of Neurology*, **2009**, 66, 415-9	9.4	230
117	EEG features of absence seizures in idiopathic generalized epilepsy: impact of syndrome, age, and state. *Epilepsia*, **2009**, 50, 1572-8	6.4	61

116	SCN1A duplications and deletions detected in Dravet syndrome: implications for molecular diagnosis. *Epilepsia*, **2009**, 50, 1670-8	6.4	127
115	Does a SCN1A gene mutation confer earlier age of onset of febrile seizures in GEFS+?. *Epilepsia*, **2009**, 50, 953-6	6.4	17
114	Neuropsychological and functional MRI studies provide converging evidence of anterior language dysfunction in BECTS. *Epilepsia*, **2009**, 50, 2276-84	6.4	89
113	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
112	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
111	Periventricular heterotopia, mental retardation, and epilepsy associated with 5q14.3-q15 deletion. *Neurology*, **2009**, 72, 784-92	6.5	98
110	Translational research in epilepsy genetics: sodium channels in man to interneuronopathy in mouse. *Archives of Neurology*, **2009**, 66, 21-6		46
109	X-linked protocadherin 19 mutations cause female-limited epilepsy and cognitive impairment. *Nature Genetics*, **2008**, 40, 776-81	36.3	328
108	Gene expression analysis in absence epilepsy using a monozygotic twin design. *Epilepsia*, **2008**, 49, 1546-54	6.4	21
107	Severe autosomal dominant nocturnal frontal lobe epilepsy associated with psychiatric disorders and intellectual disability. *Epilepsia*, **2008**, 49, 2125-9	6.4	40
106	Factors influencing clinical features of absence seizures. *Epilepsia*, **2008**, 49, 2100-7	6.4	29
105	Classification and clinical features of absence epilepsies: how evidence leads to changing concepts. *Epilepsia*, **2008**, 49, 2140-1	6.4	4
104	Navigating the channels and beyond: unravelling the genetics of the epilepsies. *Lancet Neurology, The*, **2008**, 7, 231-45	24.1	219
103	Benign occipital epilepsies of childhood: clinical features and genetics. *Brain*, **2008**, 131, 2287-94	11.2	60
102	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
101	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
100	Epilepsy and mental retardation limited to females: an under-recognized disorder. *Brain*, **2008**, 131, 918-27	11.2	152
99	Genetic (Generalized) Epilepsy with Febrile Seizures Plus **2008**, 29-33		3

98	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
97	Febrile seizures. *BMJ, The*, **2007**, 334, 307-11	5.9	96
96	Genetic Epilepsies **2007**, 371-383		0
95	Extended spectrum of idiopathic generalized epilepsies associated with CACNA1H functional variants. *Annals of Neurology*, **2007**, 62, 560-8	9.4	150
94	Channelopathies in idiopathic epilepsy. *Neurotherapeutics*, **2007**, 4, 295-304	6.4	86
93	Association studies and functional validation or functional validation alone?. *Epilepsy Research*, **2007**, 74, 237-8	3	3
92	Absence of mutations in the LGI1 receptor ADAM22 gene in autosomal dominant lateral temporal epilepsy. *Epilepsy Research*, **2007**, 76, 41-8	3	24
91	A multicenter study of BRD2 as a risk factor for juvenile myoclonic epilepsy. *Epilepsia*, **2007**, 48, 706-12	6.4	56
90	SCN2A mutations and benign familial neonatal-infantile seizures: the phenotypic spectrum. *Epilepsia*, **2007**, 48, 1138-42	6.4	90
89	Is photosensitive epilepsy less common in males due to variation in X chromosome photopigment genes?. *Epilepsia*, **2007**, 48, 1807-1809	6.4	8
88	Early seizures: causal events or predisposition to adult epilepsy?. *Lancet Neurology, The*, **2007**, 6, 643-51	24.1	17
87	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
86	Temporal lobe epilepsy and GEFS+ phenotypes associated with SCN1B mutations. *Brain*, **2007**, 130, 100-9	11.2	188
85	The spectrum of SCN1A-related infantile epileptic encephalopathies. *Brain*, **2007**, 130, 843-52	11.2	423
84	Vaccination, seizures and 'vaccine damage'. *Current Opinion in Neurology*, **2007**, 20, 181-7	7.1	33
83	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
82	GEFS+ where focal seizures evolve from generalized spike wave: video-EEG study of two children. *Epileptic Disorders*, **2007**, 9, 307-14	1.9	9
81	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

80	Severe myoclonic epilepsy of infancy (Dravet syndrome): recognition and diagnosis in adults. *Neurology*, **2006**, 67, 2224-6	6.5	126
79	Epileptiform EEG abnormalities in children with language regression. *Neurology*, **2006**, 67, 1527; author reply 1527	6.5	1
78	Distinguishing sleep disorders from seizures: diagnosing bumps in the night. *Archives of Neurology*, **2006**, 63, 705-9		178
77	SRPX2 mutations in disorders of language cortex and cognition. *Human Molecular Genetics*, **2006**, 15, 1195-207	5.6	213
76	A new molecular mechanism for severe myoclonic epilepsy of infancy: exonic deletions in SCN1A. *Neurology*, **2006**, 67, 1094-5	6.5	85
75	Electroclinical features of absence seizures in childhood absence epilepsy. *Neurology*, **2006**, 67, 413-8	6.5	113
74	Human epilepsies: interaction of genetic and acquired factors. *Trends in Neurosciences*, **2006**, 29, 391-397	13.3	147
73	Genetic dissection of the common epilepsies. *Current Opinion in Neurology*, **2006**, 19, 157-63	7.1	23
72	Thalamic atrophy in childhood absence epilepsy. *Epilepsia*, **2006**, 47, 399-405	6.4	88
71	Analyzing the etiology of benign rolandic epilepsy: a multicenter twin collaboration. *Epilepsia*, **2006**, 47, 550-5	6.4	59
70	Exploration of the genetic architecture of idiopathic generalized epilepsies. *Epilepsia*, **2006**, 47, 1682-90	6.4	42
69	Invited comments on the Shostak and Ottman review. *Epilepsia*, **2006**, 47, 1751-2; author reply 1755-6	6.4	
68	Susceptibility genes for complex epilepsy. *Human Molecular Genetics*, **2005**, 14 Spec No. 2, R243-9	5.6	80
67	Neonatal epilepsy syndromes and generalized epilepsy with febrile seizures plus (GEFS+). *Epilepsia*, **2005**, 46 Suppl 10, 41-7	6.4	55
66	Is variation in the GABA(B) receptor 1 gene associated with temporal lobe epilepsy?. *Epilepsia*, **2005**, 46, 778-80	6.4	13
65	Reflex seizures in patients with malformations of cortical development and refractory epilepsy. *Epilepsia*, **2005**, 46, 1224-34	6.4	22
64	The role of genetics and ethnicity in epilepsy management. *Acta Neurologica Scandinavica*, **2005**, 181, 47-51	3.8	5
63	SCN1A mutations and epilepsy. *Human Mutation*, **2005**, 25, 535-42	4.7	282

62	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
61	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
60	Juvenile myoclonic epilepsy and idiopathic photosensitive occipital lobe epilepsy: is there overlap?. *Brain*, **2004**, 127, 1878-86	11.2	54
59	Familial partial epilepsy with variable foci: clinical features and linkage to chromosome 22q12. *Epilepsia*, **2004**, 45, 1054-60	6.4	48
58	Genetic architecture of idiopathic generalized epilepsy: clinical genetic analysis of 55 multiplex families. *Epilepsia*, **2004**, 45, 467-78	6.4	106
57	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
56	Etiological heterogeneity of familial periventricular heterotopia and hydrocephalus. *Brain and Development*, **2004**, 26, 326-34	2.2	49
55	Genetic variation of CACNA1H in idiopathic generalized epilepsy. *Annals of Neurology*, **2004**, 55, 595-6	9.4	92
54	Benign familial neonatal-infantile seizures: characterization of a new sodium channelopathy. *Annals of Neurology*, **2004**, 55, 550-7	9.4	225
53	Is benign rolandic epilepsy genetically determined?. *Annals of Neurology*, **2004**, 56, 129-32	9.4	42
52	Childhood absence epilepsy and febrile seizures: a family with a GABA(A) receptor mutation. *Brain*, **2003**, 126, 230-40	11.2	122
51	Severe infantile epilepsies: molecular genetics challenge clinical classification. *Brain*, **2003**, 126, 513-4	11.2	19
50	Channelopathies as a genetic cause of epilepsy. *Current Opinion in Neurology*, **2003**, 16, 171-176	7.1	141
49	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
48	Occipital epilepsies: identification of specific and newly recognized syndromes. *Brain*, **2003**, 126, 753-69	11.2	109
47	The genetics of human epilepsy. *Trends in Pharmacological Sciences*, **2003**, 24, 428-33	13.2	120
46	Channelopathies as a genetic cause of epilepsy. *Current Opinion in Neurology*, **2003**, 16, 171-6	7.1	51
45	A twin study of genetic influences on epilepsy outcome. *Twin Research and Human Genetics*, **2003**, 6, 140-6		16

44	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
43	Chromosomal abnormalities and epilepsy: a review for clinicians and gene hunters. *Epilepsia*, **2002**, 43, 127-40	6.4	82
42	Mutations in the human ortholog of Aristaless cause X-linked mental retardation and epilepsy. *Nature Genetics*, **2002**, 30, 441-5	36.3	356
41	X-linked myoclonic epilepsy with spasticity and intellectual disability: mutation in the homeobox gene ARX. *Neurology*, **2002**, 59, 348-56	6.5	75
40	Subcortical band heterotopia (SBH) in males: clinical, imaging and genetic findings in comparison with females. *Brain*, **2002**, 125, 2507-22	11.2	85
39	Sodium-channel defects in benign familial neonatal-infantile seizures. *Lancet, The*, **2002**, 360, 851-2	40	284
38	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
37	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
36	Mutant GABA(A) receptor gamma2-subunit in childhood absence epilepsy and febrile seizures. *Nature Genetics*, **2001**, 28, 49-52	36.3	643
35	Genetics of the Epilepsies. *Epilepsia*, **2001**, 42, 16-23	6.4	5
34	Genetics of the epilepsies. *Epilepsia*, **2001**, 42 Suppl 5, 16-23	6.4	73
33	. *Nature Genetics*, **2001**, 28, 49-52	36.3	184
32	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
31	Neuronal sodium-channel alpha1-subunit mutations in generalized epilepsy with febrile seizures plus. *American Journal of Human Genetics*, **2001**, 68, 859-65	11	283
30	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
29	Genetics of the epilepsies. *Current Opinion in Pediatrics*, **2000**, 12, 536-42	3.2	16
28	Genetic and neuroradiological heterogeneity of double cortex syndrome. *Annals of Neurology*, **2000** , 47, 265-269	9.4	89
27	Locus for febrile seizures. *Annals of Neurology*, **2000**, 47, 840-1	9.4	10

26	Independent occurrence of the CHRNA4 Ser248Phe mutation in a Norwegian family with nocturnal frontal lobe epilepsy. *Epilepsia*, **2000**, 41, 529-35	6.4	112
25	Autosomal dominant nocturnal frontal lobe epilepsy. *Epilepsia*, **2000**, 41, 1059-60	6.4	23
24	Phenotypic characterization of an alpha 4 neuronal nicotinic acetylcholine receptor subunit knock-out mouse. *Journal of Neuroscience*, **2000**, 20, 6431-41	6.6	217
23	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
22	Generalized epilepsy with febrile seizures plus: a common childhood-onset genetic epilepsy syndrome. *Annals of Neurology*, **1999**, 45, 75-81	9.4	239
21	Characterization of mutations in the gene doublecortin in patients with double cortex syndrome. *Annals of Neurology*, **1999**, 45, 146-53	9.4	157
20	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
19	How long should a routine EEG be?. *Journal of Clinical Neuroscience*, **1999**, 6, 492-3	2.2	9
18	Genetics of the epilepsies. *Current Opinion in Neurology*, **1999**, 12, 177-82	7.1	15
17	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
16	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
15	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
14	Mutations in filamin 1 prevent migration of cerebral cortical neurons in human periventricular heterotopia. *Neuron*, **1998**, 21, 1315-25	13.9	729
13	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
12	Febrile seizures: genetics and relationship to other epilepsy syndromes. *Current Opinion in Neurology*, **1998**, 11, 129-34	7.1	42
11	Genetics of human partial epilepsy. *Current Opinion in Neurology*, **1997**, 10, 110-4	7.1	26
10	Epilepsies with single gene inheritance. *Brain and Development*, **1997**, 19, 13-8	2.2	37
9	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

8	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
7	Autosomal dominant nocturnal frontal lobe epilepsy. A distinctive clinical disorder. *Brain*, **1995**, 118 (Pt 1), 61-73	11.2	471
6	Autosomal dominant rolandic epilepsy and speech dyspraxia: a new syndrome with anticipation. *Annals of Neurology*, **1995**, 38, 633-42	9.4	117
5	New autosomal-dominant partial epilepsy syndrome. *Pediatric Neurology*, **1994**, 11, 95	2.9	6
4	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
3	Genetic epilepsy with febrile seizures plus74-77		
2	Genome-wide association study of febrile seizures identifies seven new loci implicating fever response and neuronal excitability genes		2
1	Pathogenic MAST3 variants in the STK domain are associated with epilepsy		1