Ingrid E Scheffer Mbbs

List of Publications by Year
in descending order

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560	65,313	906 116	1072 233
papers	citations	h-index	g-index

586	586	586	38672
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	ILAE Official Report: A practical clinical definition of epilepsy. Epilepsia, 2014, 55, 475-482.	5.1	3,770
2	Revised terminology and concepts for organization of seizures and epilepsies: Report of the ILAE Commission on Classification and Terminology, 2005â€“2009. Epilepsia, 2010, 51, 676-685.	5.1	3,612
3	<scp>ILAE</scp> classification of the epilepsies: Position paper of the <scp>ILAE</scp> Commission for Classification and Terminology. Epilepsia, 2017, 58, 512-521.	5.1	3,464
4	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.	5.1	2,191
5	A definition and classification of status epilepticus â€“ Report of the <scp>ILAE</scp> Task Force on Classification of Status Epilepticus. Epilepsia, 2015, 56, 1515-1523.	5.1	1,630
6	De novo mutations in epileptic encephalopathies. Nature, 2013, 501, 217-221.	27.8	1,351
7	Trial of Cannabidiol for Drug-Resistant Seizures in the Dravet Syndrome. New England Journal of Medicine, 2017, 376, 2011-2020.	27.0	1,148
8	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	12.6	1,085
9	A missense mutation in the neuronal nicotinic acetylcholine receptor Î±4 subunit is associated with autosomal dominant nocturnal frontal lobe epilepsy. Nature Genetics, 1995, 11, 201-203.	21.4	1,074
10	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.	28.9	1,007
11	Febrile seizures and generalized epilepsy associated with a mutation in the Na+-channel ÃŸ1 subunit gene SCN1B. Nature Genetics, 1998, 19, 366-370.	21.4	965
12	Mutations in filamin 1 Prevent Migration of Cerebral Cortical Neurons in Human Periventricular Heterotopia. Neuron, 1998, 21, 1315-1325.	8.1	811
13	Mutant GABAA receptor Î³2-subunit in childhood absence epilepsy and febrile seizures. Nature Genetics, 2001, 28, 49-52.	21.4	721
14	Instruction manual for the <scp>ILAE</scp> 2017 operational classification of seizure types. Epilepsia, 2017, 58, 531-542.	5.1	699
15	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. Nature Genetics, 2013, 45, 825-830.	21.4	589
16	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	21.4	583
17	Epilepsy. Nature Reviews Disease Primers, 2018, 4, 18024.	30.5	541
18	Autosomal dominant nocturnal frontal lobe epilepsy. Brain, 1995, 118, 61-73.	7.6	523

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19	The spectrum of SCN1A-related infantile epileptic encephalopathies. Brain, 2007, 130, 843-852.	7.6	501
20	The genetic landscape of the epileptic encephalopathies of infancy and childhood. Lancet Neurology, The, 2016, 15, 304-316.	10.2	474
21	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	21.4	443
22	KCNQ2 encephalopathy: Emerging phenotype of a neonatal epileptic encephalopathy. Annals of Neurology, 2012, 71, 15-25.	5.3	427
23	Truncation of the GABAA-Receptor Î³2 Subunit in a Family with Generalized Epilepsy with Febrile Seizures Plus. American Journal of Human Genetics, 2002, 70, 530-536.	6.2	425
24	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.	2.4	412
25	X-linked protocadherin 19 mutations cause female-limited epilepsy and cognitive impairment. Nature Genetics, 2008, 40, 776-781.	21.4	397
26	Mutations in the human ortholog of Aristaless cause X-linked mental retardation and epilepsy. Nature Genetics, 2002, 30, 441-445.	21.4	396
27	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	6.2	388
28	De novo mutations in ATP1A3 cause alternating hemiplegia of childhood. Nature Genetics, 2012, 44, 1030-1034.	21.4	345
29	Localization of a gene for autosomal dominant nocturnal frontal lobe epilepsy to chromosome 20q13.2. Nature Genetics, 1995, 10, 117-118.	21.4	337
30	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
31	Missense mutations in the sodium-gated potassium channel gene KCNT1 cause severe autosomal dominant nocturnal frontal lobe epilepsy. Nature Genetics, 2012, 44, 1188-1190.	21.4	333
32	Sodium-channel defects in benign familial neonatal-infantile seizures. Lancet, The, 2002, 360, 851-852.	13.7	332
33	Genome-wide mega-analysis identifies 16 loci and highlights diverse biological mechanisms in the common epilepsies. Nature Communications, 2018, 9, 5269.	12.8	331
34	SCN1Amutations and epilepsy. Human Mutation, 2005, 25, 535-542.	2.5	327
35	GRIN2A mutations cause epilepsy-aphasia spectrum disorders. Nature Genetics, 2013, 45, 1073-1076.	21.4	326
36	Somatic Mutations in Cerebral Cortical Malformations. New England Journal of Medicine, 2014, 371, 733-743.	27.0	326

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37	Neuronal Sodium-Channel Î±1-Subunit Mutations in Generalized Epilepsy with Febrile Seizures Plus. American Journal of Human Genetics, 2001, 68, 859-865.	6.2	316
38	Mutations in DEPDC5 cause familial focal epilepsy with variable foci. Nature Genetics, 2013, 45, 546-551.	21.4	301
39	CHRNB2 Is the Second Acetylcholine Receptor Subunit Associated with Autosomal Dominant Nocturnal Frontal Lobe Epilepsy*. American Journal of Human Genetics, 2001, 68, 225-231.	6.2	300
40	GABRD encoding a protein for extra- or peri-synaptic GABAA receptors is a susceptibility locus for generalized epilepsies. Human Molecular Genetics, 2004, 13, 1315-1319.	2.9	299
41	De-novo mutations of the sodium channel gene SCN1A in alleged vaccine encephalopathy: a retrospective study. Lancet Neurology, The, 2006, 5, 488-492.	10.2	295
42	The genetics of Dravet syndrome. Epilepsia, 2011, 52, 24-29.	5.1	287
43	Generalized epilepsy with febrile seizures plus: A common childhood-onset genetic epilepsy syndrome. Annals of Neurology, 1999, 45, 75-81.	5.3	271
44	Earlyâ€onset absence epilepsy caused by mutations in the glucose transporter GLUT1. Annals of Neurology, 2009, 66, 415-419.	5.3	266
45	Genetic determinants of common epilepsies: a meta-analysis of genome-wide association studies. Lancet Neurology, The, 2014, 13, 893-903.	10.2	264
46	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, 63, 1398-1442.	5.1	263
47	Benign familial neonatal-infantile seizures: Characterization of a new sodium channelopathy. Annals of Neurology, 2004, 55, 550-557.	5.3	250
48	Navigating the channels and beyond: unravelling the genetics of the epilepsies. Lancet Neurology, The, 2008, 7, 231-245.	10.2	249
49	KCNT1 gain of function in 2 epilepsy phenotypes is reversed by quinidine. Annals of Neurology, 2014, 75, 581-590.	5.3	249
50	SRPX2 mutations in disorders of language cortex and cognition. Human Molecular Genetics, 2006, 15, 1195-1207.	2.9	248
51	New concepts in classification of the epilepsies: Entering the 21st century. Epilepsia, 2011, 52, 1058-1062.	5.1	248
52	Title is missing!. Nature Genetics, 2001, 28, 49-52.	21.4	247
53	De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies. American Journal of Human Genetics, 2016, 99, 287-298.	6.2	247
54	The phenotypic spectrum of SCN8A encephalopathy. Neurology, 2015, 84, 480-489.	1.1	246

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55	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. Nature Genetics, 2015, 47, 39-46.	21.4	245
56	Definition and diagnostic criteria of sleep-related hypermotor epilepsy. Neurology, 2016, 86, 1834-1842.	1.1	245
57	Dravet syndrome as epileptic encephalopathy: evidence from long-term course and neuropathology. Brain, 2011, 134, 2982-3010.	7.6	237
58	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. American Journal of Human Genetics, 2019, 105, 267-282.	6.2	237
59	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, 63, 1349-1397.	5.1	237
60	Temporal lobe epilepsy and GEFS+ phenotypes associated with SCN1B mutations. Brain, 2006, 130, 100-109.	7.6	234
61	PRRT2 Mutations Cause Benign Familial Infantile Epilepsy and Infantile Convulsions with Choreoathetosis Syndrome. American Journal of Human Genetics, 2012, 90, 152-160.	6.2	234
62	The new definition and classification of seizures and epilepsy. Epilepsy Research, 2018, 139, 73-79.	1.6	234
63	Phenotypic Characterization of an Î±₄ Neuronal Nicotinic Acetylcholine Receptor Subunit Knock-Out Mouse. Journal of Neuroscience, 2000, 20, 6431-6441.	3.6	231
64	GABRA1 and STXBP1 : Novel genetic causes of Dravet syndrome. Neurology, 2014, 82, 1245-1253.	1.1	229
65	Early and effective treatment of <scp>KCNQ</scp>2 encephalopathy. Epilepsia, 2015, 56, 685-691.	5.1	229
66	Dominantâ€negative effects of KCNQ2 mutations are associated with epileptic encephalopathy. Annals of Neurology, 2014, 75, 382-394.	5.3	225
67	Distinguishing Sleep Disorders From Seizures. Archives of Neurology, 2006, 63, 705.	4.5	223
68	Rare copy number variants are an important cause of epileptic encephalopathies. Annals of Neurology, 2011, 70, 974-985.	5.3	222
69	Clinical whole-genome sequencing in severe early-onset epilepsy reveals new genes and improves molecular diagnosis. Human Molecular Genetics, 2014, 23, 3200-3211.	2.9	222
70	Mortality in Dravet syndrome. Epilepsy Research, 2016, 128, 43-47.	1.6	218
71	Autosomal Dominant Nocturnal Frontal-Lobe Epilepsy: Genetic Heterogeneity and Evidence for a Second Locus at 15q24. American Journal of Human Genetics, 1998, 63, 1108-1116.	6.2	216
72	Exomeâ€based analysis of cardiac arrhythmia, respiratory control, and epilepsy genes in sudden unexpected death in epilepsy. Annals of Neurology, 2016, 79, 522-534.	5.3	216

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73	Clinical and imaging heterogeneity of polymicrogyria: a study of 328 patients. Brain, 2010, 133, 1415-1427.	7.6	215
74	Ion Channels in Genetic Epilepsy: From Genes and Mechanisms to Disease-Targeted Therapies. Pharmacological Reviews, 2018, 70, 142-173.	16.0	215
75	Familial and sporadic 15q13.3 microdeletions in idiopathic generalized epilepsy: precedent for disorders with complex inheritance. Human Molecular Genetics, 2009, 18, 3626-3631.	2.9	211
76	SCN2A encephalopathy. Neurology, 2015, 85, 958-966.	1.1	211
77	Extending the KCNQ2 encephalopathy spectrum. Neurology, 2013, 81, 1697-1703.	1.1	198
78	Mutations in mammalian target of rapamycin regulator DEPDC5 cause focal epilepsy with brain malformations. Annals of Neurology, 2014, 75, 782-787.	5.3	193
79	Human epilepsies: interaction of genetic and acquired factors. Trends in Neurosciences, 2006, 29, 391-397.	8.6	190
80	Mutations in the mammalian target of rapamycin pathway regulators NPRL2 and NPRL3 cause focal epilepsy. Annals of Neurology, 2016, 79, 120-131.	5.3	190
81	Ultra-rare genetic variation in common epilepsies: a case-control sequencing study. Lancet Neurology, The, 2017, 16, 135-143.	10.2	190
82	GRIN2Bencephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. Journal of Medical Genetics, 2017, 54, 460-470.	3.2	190
83	Extended spectrum of idiopathic generalized epilepsies associated with CACNA1H functional variants. Annals of Neurology, 2007, 62, 560-568.	5.3	186
84	Quinidine in the treatment of <scp>KCNT</scp>1â€positive epilepsies. Annals of Neurology, 2015, 78, 995-999.	5.3	184
85	Characterization of mutations in the genedoublecortin in patients with double cortex syndrome. Annals of Neurology, 1999, 45, 146-153.	5.3	175
86	Multicentre search for genetic susceptibility loci in sporadic epilepsy syndrome and seizure types: a case-control study. Lancet Neurology, The, 2007, 6, 970-980.	10.2	175
87	Genetic testing in the epilepsiesâ€"Report of the ILAE Genetics Commission. Epilepsia, 2010, 51, 655-670.	5.1	175
88	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. American Journal of Human Genetics, 2015, 96, 808-815.	6.2	173
89	Epilepsy and mental retardation limited to females: an under-recognized disorder. Brain, 2008, 131, 918-927.	7.6	172
90	Dose-Ranging Effect of Adjunctive Oral Cannabidiol vs Placebo on Convulsive Seizure Frequency in Dravet Syndrome. JAMA Neurology, 2020, 77, 613.	9.0	171

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91	Infantile spasms, dystonia, and other X-linked phenotypes caused by mutations in Aristaless related homeobox gene, ARX. Brain and Development, 2002, 24, 266-268.	1.1	170
92	Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. Brain, 2013, 136, 3140-3150.	7.6	168
93	A variant of <scp>KCC</scp> 2 from patients with febrile seizures impairs neuronal Cl ^{â˜} extrusion and dendritic spine formation. EMBO Reports, 2014, 15, 723-729.	4.5	163
94	Mutations in SLC12A5 in epilepsy of infancy with migrating focal seizures. Nature Communications, 2015, 6, 8038.	12.8	160
95	A roadmap for precision medicine in the epilepsies. Lancet Neurology, The, 2015, 14, 1219-1228.	10.2	160
96	Randomized, phase III study results of clobazam in Lennox-Gastaut syndrome. Neurology, 2011, 77, 1473-1481.	1.1	159
97	Autosomal dominant rolandic epilepsy and speech dyspraxia: A new syndrome with anticipation. Annals of Neurology, 1995, 38, 633-642.	5.3	156
98	Channelopathies as a genetic cause of epilepsy. Current Opinion in Neurology, 2003, 16, 171-176.	3.6	153
99	Severe myoclonic epilepsy of infancy (Dravet syndrome): Recognition and diagnosis in adults. Neurology, 2006, 67, 2224-2226.	1.1	153
100	Dravet syndrome or genetic (generalized) epilepsy with febrile seizures plus?. Brain and Development, 2009, 31, 394-400.	1.1	152
101	SCN1A duplications and deletions detected in Dravet syndrome: Implications for molecular diagnosis. Epilepsia, 2009, 50, 1670-1678.	5.1	152
102	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
103	Electroclinical features of absence seizures in childhood absence epilepsy. Neurology, 2006, 67, 413-418.	1.1	149
104	Childhood absence epilepsy and febrile seizures: a family with a GABAA receptor mutation. Brain, 2003, 126, 230-240.	7.6	148
105	Absence epilepsies with widely variable onset are a key feature of familial GLUT1 deficiency. Neurology, 2010, 75, 432-440.	1.1	148
106	ILAE definition of the Idiopathic Generalized Epilepsy Syndromes: Position statement by the ILAE Task Force on Nosology and Definitions. Epilepsia, 2022, 63, 1475-1499.	5.1	148
107	Migrating partial seizures of infancy: expansion of the electroclinical, radiological and pathological disease spectrum. Brain, 2013, 136, 1578-1591.	7.6	144
108	Occipital epilepsies: identification of specific and newly recognized syndromes. Brain, 2003, 126, 753-769.	7.6	142

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109	Disruptive de novo mutations of DYRK1A lead to a syndromic form of autism and ID. Molecular Psychiatry, 2016, 21, 126-132.	7.9	142
110	Glut1 Deficiency Syndrome (Glut1DS): State of the art in 2020 and recommendations of the international Glut1DS study group. Epilepsia Open, 2020, 5, 354-365.	2.4	142
111	Copy number variants are frequent in genetic generalized epilepsy with intellectual disability. Neurology, 2013, 81, 1507-1514.	1.1	140
112	The management of epilepsy in children and adults. Medical Journal of Australia, 2018, 208, 226-233.	1.7	136
113	Analyzing the Etiology of Benign Rolandic Epilepsy: A Multicenter Twin Collaboration. Epilepsia, 2006, 47, 550-555.	5.1	135
114	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, 5359-5372.	2.9	134
115	The genetics of human epilepsy. Trends in Pharmacological Sciences, 2003, 24, 428-433.	8.7	131
116	SYNGAP1 encephalopathy. Neurology, 2019, 92, e96-e107.	1.1	131
117	The core network in absence epilepsy. Neurology, 2010, 75, 904-911.	1.1	129
118	Genetic Architecture of Idiopathic Generalized Epilepsy: Clinical Genetic Analysis of 55 Multiplex Families. Epilepsia, 2004, 45, 467-478.	5.1	128
119	Glucose transporter 1 deficiency in the idiopathic generalized epilepsies. Annals of Neurology, 2012, 72, 807-815.	5.3	123
120	Glucose Transporter 1 Deficiency as a Treatable Cause of Myoclonic Astatic Epilepsy. Archives of Neurology, 2011, 68, 1152.	4.5	121
121	Independent Occurrence of the CHRNA4 Ser248Phe Mutation in a Norwegian Family with Nocturnal Frontal Lobe Epilepsy. Epilepsia, 2000, 41, 529-535.	5.1	119
122	Effects of vaccination on onset and outcome of Dravet syndrome: a retrospective study. Lancet Neurology, The, 2010, 9, 592-598.	10.2	119
123	Febrile seizures. BMJ: British Medical Journal, 2007, 334, 307-311.	2.3	118
124	Clinical profile of patients with ATP1A3 mutations in Alternating Hemiplegia of Childhoodâ€"a study of 155 patients. Orphanet Journal of Rare Diseases, 2015, 10, 123.	2.7	117
125	Mutations in <scp>KCNT</scp>1 cause a spectrum of focal epilepsies. Epilepsia, 2015, 56, e114-20.	5.1	117
126	Keeping people with epilepsy safe during the COVID-19 pandemic. Neurology, 2020, 94, 1032-1037.	1.1	116

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127	The phenotype of SCN8A developmental and epileptic encephalopathy. Neurology, 2018, 91, e1112-e1124.	1.1	114
128	CHD2variants are a risk factor for photosensitivity in epilepsy. Brain, 2015, 138, 1198-1208.	7.6	112
129	Genetic epilepsy with febrile seizures plus. Neurology, 2017, 89, 1210-1219.	1.1	112
130	Familial partial epilepsy with variable foci: A new partial epilepsy syndrome with suggestion of linkage to chromosome 2. Annals of Neurology, 1998, 44, 890-899.	5.3	111
131	Recent advances in the molecular genetics of epilepsy. Journal of Medical Genetics, 2013, 50, 271-279.	3.2	111
132	Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3. Nature Communications, 2019, 10, 4919.	12.8	111
133	Periventricular heterotopia, mental retardation, and epilepsy associated with 5q14.3-q15 deletion. Neurology, 2009, 72, 784-792.	1.1	110
134	Mirror neuron system involvement in empathy: A critical look at the evidence. Social Neuroscience, 2011, 6, 327-335.	1.3	110
135	Precision therapy for epilepsy due to KCNT1 mutations. Neurology, 2018, 90, e67-e72.	1.1	108
136	A set of regulatory genes co-expressed in embryonic human brain is implicated in disrupted speech development. Molecular Psychiatry, 2019, 24, 1065-1078.	7.9	106
137	Subcortical band heterotopia (SBH) in males: clinical, imaging and genetic findings in comparison with females. Brain, 2002, 125, 2507-2522.	7.6	105
138	Heart rate variability in epilepsy: A potential biomarker of <scp>sudden unexpected death in epilepsy</scp> risk. Epilepsia, 2018, 59, 1372-1380.	5.1	105
139	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	12.8	105
140	Neuropsychological and functional MRI studies provide converging evidence of anterior language dysfunction in BECTS. Epilepsia, 2009, 50, 2276-2284.	5.1	104
141	Homozygous PLCB1 deletion associated with malignant migrating partial seizures in infancy. Epilepsia, 2012, 53, e146-50.	5.1	104
142	Parental Mosaicism in â€œDe Novoâ€•Epileptic Encephalopathies. New England Journal of Medicine, 2018, 378, 1646-1648.	27.0	104
143	SCN1Aâ€related phenotypes: Epilepsy and beyond. Epilepsia, 2019, 60, S17-S24.	5.1	103
144	Genetic variation ofCACNA1H in idiopathic generalized epilepsy. Annals of Neurology, 2004, 55, 595-596.	5.3	102

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145	SCN2A Mutations and Benign Familial Neonatal-Infantile Seizures: The Phenotypic Spectrum. Epilepsia, 2007, 48, 1138-1142.	5.1	102
146	SLC25A22 is a novel gene for migrating partial seizures in infancy. Annals of Neurology, 2013, 74, 873-882.	5.3	102
147	Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hypotonia, and Seizures. American Journal of Human Genetics, 2016, 98, 1001-1010.	6.2	102
148	Channelopathies in idiopathic epilepsy. Neurotherapeutics, 2007, 4, 295-304.	4.4	101
149	SCN8A encephalopathy: Research progress and prospects. Epilepsia, 2016, 57, 1027-1035.	5.1	101
150	Timing of De Novo Mutagenesis â€" A Twin Study of Sodium-Channel Mutations. New England Journal of Medicine, 2010, 363, 1335-1340.	27.0	100
151	Longâ€term followâ€up of febrile infectionâ€"related epilepsy syndrome. Epilepsia, 2012, 53, 101-110.	5.1	100
152	Abnormal Cell Sorting Underlies the Unique X-Linked Inheritance of PCDH19 Epilepsy. Neuron, 2018, 97, 59-66.e5.	8.1	100
153	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	12.8	99
154	Chromosomal Abnormalities and Epilepsy: A Review for Clinicians and Gene Hunters. Epilepsia, 2002, 43, 127-140.	5.1	98
155	Early onset absence epilepsy: 1 in 10 cases is caused by GLUT1 deficiency. Epilepsia, 2012, 53, e204-7.	5.1	97
156	TBC1D24 genotypeâ€"phenotype correlation. Neurology, 2016, 87, 77-85.	1.1	97
157	Not all SCN1A epileptic encephalopathies are Dravet syndrome. Neurology, 2017, 89, 1035-1042.	1.1	97
158	Deciphering the concepts behind â€œEpileptic encephalopathyâ€• and â€œDevelopmental and epileptic encephalopathyâ€• European Journal of Paediatric Neurology, 2020, 24, 11-14.	1.6	97
159	A new molecular mechanism for severe myoclonic epilepsy of infancy: Exonic deletions in SCN1A. Neurology, 2006, 67, 1094-1095.	1.1	96
160	Augmented currents of an HCN2 variant in patients with febrile seizure syndromes. Annals of Neurology, 2010, 67, 542-546.	5.3	96
161	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. Annals of Neurology, 2019, 86, 821-831.	5.3	96
162	Phospholipase C beta 1 deficiency is associated with early-onset epileptic encephalopathy. Brain, 2010, 133, 2964-2970.	7.6	95

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163	Progressive Gait Deterioration in Adolescents With Dravet Syndrome. Archives of Neurology, 2012, 69, 873-8.	4.5	95
164	Familial cortical dysplasia type <scp>IIA</scp> caused by a germline mutation in <scp>DEPDC</scp>5. Annals of Clinical and Translational Neurology, 2015, 2, 575-580.	3.7	95
165	Genetic and neuroradiological heterogeneity of double cortex syndrome. Annals of Neurology, 2000, 47, 265-269.	5.3	94
166	Thalamic Atrophy in Childhood Absence Epilepsy. Epilepsia, 2006, 47, 399-405.	5.1	94
167	Familial neonatal seizures in 36 families: Clinical and genetic features correlate with outcome. Epilepsia, 2015, 56, 1071-1080.	5.1	94
168	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. Human Molecular Genetics, 2015, 24, 5250-5259.	2.9	93
169	Classification of the epilepsies: New concepts for discussion and debateâ€"Special report of the ILAE Classification Task Force of the Commission for Classification and Terminology. Epilepsia Open, 2016, 1, 37-44.	2.4	93
170	Real-world utility of whole exome sequencing with targeted gene analysis for focal epilepsy. Epilepsy Research, 2017, 131, 1-8.	1.6	93
171	Susceptibility genes for complex epilepsy. Human Molecular Genetics, 2005, 14, R243-R249.	2.9	92
172	Genetics of the Epilepsies. Epilepsia, 2001, 42, 16-23.	5.1	91
173	Autoantibodies and epilepsy. Epilepsia, 2011, 52, 18-22.	5.1	90
174	Mapping of a Gene Determining Familial Partial Epilepsy with Variable Foci to Chromosome 22q11-q12. American Journal of Human Genetics, 1999, 65, 1698-1710.	6.2	89
175	Clinical and molecular characterization of KCNT1 -related severe early-onset epilepsy. Neurology, 2018, 90, e55-e66.	1.1	89
176	X-linked myoclonic epilepsy with spasticity and intellectual disability. Neurology, 2002, 59, 348-356.	1.1	88
177	DNM1 encephalopathy. Neurology, 2017, 89, 385-394.	1.1	87
178	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.	6.2	87
179	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.	7.9	86
180	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. Brain, 2013, 136, 3378-3394.	7.6	85

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181	Methodology for classification and definition of epilepsy syndromes with list of syndromes: Report of the ILAE Task Force on Nosology and Definitions. Epilepsia, 2022, 63, 1333-1348.	5.1	84
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183	Channelopathies as a genetic cause of epilepsy. Current Opinion in Neurology, 2003, 16, 171-176.	3.6	82
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