Ingrid E Scheffer Mbbs

List of Publications by Year
in descending order

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559 papers	65,313 citations	905 116 h-index	1072 233 g-index
586 all docs	586 docs citations	586 times ranked	38672 citing authors

#	Article	IF	Citations
1	Exome sequencing for patients with developmental and epileptic encephalopathies in clinical practice. Developmental Medicine and Child Neurology, 2023, 65, 50-57.	1.1	11
2	Precision Medicine Approaches for Infantile-Onset Developmental and Epileptic Encephalopathies. Annual Review of Pharmacology and Toxicology, 2022, 62, 641-662.	4.2	10
3	Genome-wide association study of febrile seizures implicates fever response and neuronal excitability genes. Brain, 2022, 145, 555-568.	3.7	29
4	Atypical development of Brocaâ€™s area in a large family with inherited stuttering. Brain, 2022, 145, 1177-1188.	3.7	6
5	A family study implicates GBE1 in the etiology of autism spectrum disorder. Human Mutation, 2022, 43, 16-29.	1.1	2
6	Infantileâ€onset myoclonic developmental and epileptic encephalopathy: A new RARS2 phenotype. Epilepsia Open, 2022, 7, 170-180.	1.3	5
7	Sporadic hypothalamic hamartoma is a ciliopathy with somatic and bi-allelic contributions. Human Molecular Genetics, 2022, 31, 2307-2316.	1.4	8
8	OUP accepted manuscript. Brain, 2022, , .	3.7	1
9	Association of ultraâ€rare coding variants with genetic generalized epilepsy: A caseâ€"control whole exome sequencing study. Epilepsia, 2022, 63, 723-735.	2.6	8
10	Evidence for a Dual-Pathway, 2-Hit Genetic Model for Focal Cortical Dysplasia and Epilepsy. Neurology: Genetics, 2022, 8, e652.	0.9	14
11	Development and Validation of a Prediction Model for Early Diagnosis of SCN1A -Related Epilepsies. Neurology, 2022, 98, .	1.5	24
12	Lightning progress in child neurology in the past 20 years. Lancet Neurology, The, 2022, 21, 111-113.	4.9	0
13	Defective lipid signalling caused by mutations inPIK3C2Bunderlies focal epilepsy. Brain, 2022, 145, 2313-2331.	3.7	10
14	PIGNencephalopathy: Characterizing the epileptology. Epilepsia, 2022, 63, 974-991.	2.6	4
15	Selfâ€reported impact of developmental stuttering across the lifespan. Developmental Medicine and Child Neurology, 2022, 64, 1297-1306.	1.1	7
16	Impaired Color Recognition in HCN1 Epilepsy: A Single Case Report. Frontiers in Neurology, 2022, 13, 834252.	1.1	5
17	Focal Epilepsy in Children With Tuberous Sclerosis Complex: Does Vigabatrin Control Focal Seizures?. Journal of Child Neurology, 2022, , 088307382110483.	0.7	1
18	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	2.6	16

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19	COVIDâ€19 vaccine in patients with Dravet syndrome: Observations and realâ€world experiences. Epilepsia, 2022, 63, 1778-1786.	2.6	13
20	A pharmacogenomic assessment of psychiatric adverse drug reactions to levetiracetam. Epilepsia, 2022, 63, 1563-1570.	2.6	11
21	Respiratory syncytial virus epidemic during the COVIDâ€19 pandemic. Journal of Paediatrics and Child Health, 2022, 58, 215-216.	0.4	4
22	A randomized, doubleâ€blind trial of triheptanoin for drugâ€resistant epilepsy in glucose transporter 1 deficiency syndrome. Epilepsia, 2022, 63, 1748-1760.	2.6	9
23	Rare sudden unexpected death in epilepsy SCN5A variants cause changes in channel function implicating cardiac arrhythmia as a cause of death. Epilepsia, 2022, 63, .	2.6	8
24	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.	4.9	63
25	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.	2.6	263
26	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.	2.6	84
27	Efficacy and Safety of Fenfluramine for the Treatment of Seizures Associated With Lennox-Gastaut Syndrome. JAMA Neurology, 2022, 79, 554.	4.5	43
28	International League Against Epilepsy classification and definition of epilepsy syndromes with onset at a variable age: position statement by the ILAE Task Force on Nosology and Definitions. Epilepsia, 2022, 63, 1443-1474.	2.6	81
29	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.	2.6	237
30	ILAE definition of the Idiopathic Generalized Epilepsy Syndromes: Position statement by the ILAE Task Force on Nosology and Definitions. Epilepsia, 2022, 63, 1475-1499.	2.6	148
31	International consensus on diagnosis and management of Dravet syndrome. Epilepsia, 2022, 63, 1761-1777.	2.6	62
32	Functional correlates of clinical phenotype and severity in recurrent SCN2A variants. Communications Biology, 2022, 5, .	2.0	13
33	Natural History Study of STXBP1-Developmental and Epileptic Encephalopathy Into Adulthood. Neurology, 2022, 99, .	1.5	23
34	Common risk variants for epilepsy are enriched in families previously targeted for rare monogenic variant discovery. EBioMedicine, 2022, 81, 104079.	2.7	10
35	The gain of function SCN1A disorder spectrum: novel epilepsy phenotypes and therapeutic implications. Brain, 2022, 145, 3816-3831.	3.7	43
36	The role of common genetic variation in presumed monogenic epilepsies. EBioMedicine, 2022, 81, 104098.	2.7	12

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37	Clinical, biochemical and genetic characteristics of MOGS-CDG: a rare congenital disorder of glycosylation. Journal of Medical Genetics, 2022, 59, 1104-1115.	1.5	2
38	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	1.1	28
39	Medullary tyrosine hydroxylase catecholaminergic neuronal populations in sudden unexpected death in epilepsy. Brain Pathology, 2021, 31, 133-143.	2.1	9
40	Genetic Contributions to Acquired Epilepsies. Epilepsy Currents, 2021, 21, 5-13.	0.4	10
41	FBXO28 causes developmental and epileptic encephalopathy with profound intellectual disability. Epilepsia, 2021, 62, e13-e21.	2.6	8
42	Transcriptome analysis of a ring chromosome 20 patient cohort. Epilepsia, 2021, 62, e22-e28.	2.6	5
43	Cerebrospinal fluid liquid biopsy for detecting somatic mosaicism in brain. Brain Communications, 2021, 3, fcaa235.	1.5	42
44	Self-limited focal epilepsy and childhood apraxia of speech with WAC pathogenic variants. European Journal of Paediatric Neurology, 2021, 30, 25-28.	0.7	7
45	The severe epilepsy syndromes of infancy: A populationâ€based study. Epilepsia, 2021, 62, 358-370.	2.6	31
46	Contribution of rare genetic variants to drug response in absence epilepsy. Epilepsy Research, 2021, 170, 106537.	0.8	9
47	The aetiologies of epilepsy. Epileptic Disorders, 2021, 23, 1-16.	0.7	35
48	Speech, Language, and Oromotor Skills in Patients With Polymicrogyria. Neurology, 2021, 96, e1898-e1912.	1.5	8
49	Association of SLC32A1 Missense Variants With Genetic Epilepsy With Febrile Seizures Plus. Neurology, 2021, 96, e2251-e2260.	1.5	13
50	Phenotypic Spectrum of Seizure Disorders in MBD5-Associated Neurodevelopmental Disorder. Neurology: Genetics, 2021, 7, e579.	0.9	8
51	Climate change and epilepsy: Insights from clinical and basic science studies. Epilepsy and Behavior, 2021, 116, 107791.	0.9	30
52	Seizures in Sotos syndrome: Phenotyping in 49 patients. Epilepsia Open, 2021, 6, 425-430.	1.3	8
53	Guidelines on the diagnosis, clinical assessments, treatment and management for CLN2 disease patients. Orphanet Journal of Rare Diseases, 2021, 16, 185.	1.2	17
54	Diverse genetic causes of polymicrogyria with epilepsy. Epilepsia, 2021, 62, 973-983.	2.6	12

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55	Cation leak underlies neuronal excitability in an HCN1 developmental and epileptic encephalopathy. Brain, 2021, 144, 2060-2073.	3.7	26
56	Progressive myoclonus epilepsiesâ€"Residual unsolved cases have marked genetic heterogeneity including dolichol-dependent protein glycosylation pathway genes. American Journal of Human Genetics, 2021, 108, 722-738.	2.6	41
57	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. Brain, 2021, 144, 1435-1450.	3.7	35
58	Lossâ€ofâ€function variants in K_v11.1 cardiac channels as a biomarker for SUDEP. Annals of Clinical and Translational Neurology, 2021, 8, 1422-1432.	1.7	9
59	The Australian Academy of Health and Medical Sciences: an authoritative, independent voice in the Australian landscape. Medical Journal of Australia, 2021, 214, 502.	0.8	0
60	Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. Epilepsia, 2021, 62, 1518-1527.	2.6	5
61	Integrated in silico and experimental assessment of disease relevance of PCDH19 Âmissense variants. Human Mutation, 2021, 42, 1030-1041.	1.1	1
62	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.	2.6	35
63	Severe speech impairment is a distinguishing feature of FOXP1â€related disorder. Developmental Medicine and Child Neurology, 2021, 63, 1417-1426.	1.1	24
64	Genetic convergence of developmental and epileptic encephalopathies and intellectual disability. Developmental Medicine and Child Neurology, 2021, 63, 1441-1447.	1.1	4
65	Pathogenic <scp>MAST3</scp> Variants in the <scp>STK</scp> Domain Are Associated with Epilepsy. Annals of Neurology, 2021, 90, 274-284.	2.8	7
66	Addâ€on cannabidiol in patients with Dravet syndrome: Results of a longâ€term openâ€label extension trial. Epilepsia, 2021, 62, 2505-2517.	2.6	45
67	Improving Specificity of <scp>Cerebrospinal Fluid</scp> Liquid Biopsy for Genetic Testing. Annals of Neurology, 2021, 90, 693-694.	2.8	2
68	Defining Dravet syndrome: An essential preâ€requisite for precision medicine trials. Epilepsia, 2021, 62, 2205-2217.	2.6	50
69	Dravet syndrome: A quick transition guide for the adult neurologist. Epilepsy Research, 2021, 177, 106743.	0.8	11
70	Postictal Psychosis in Epilepsy: A Clinicogenetic Study. Annals of Neurology, 2021, 90, 464-476.	2.8	11
71	Epidemiology of Treated Epilepsy in New Zealand Children. Neurology, 2021, 97, e1933-e1941.	1.5	3
72	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.	1.4	5

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73	Solving the Molecular Basis of the Developmental and Epileptic Encephalopathies: Are We there Yet?. Epilepsy Currents, 2021, 21, 153575972110381.	0.4	3
74	Safety and Tolerability of Transdermal Cannabidiol Gel in Children With Developmental and Epileptic Encephalopathies. JAMA Network Open, 2021, 4, e2123930.	2.8	15
75	The phenotypic spectrum of Xâ€linked, infantile onset ALG13â€related developmental and epileptic encephalopathy. Epilepsia, 2021, 62, 325-334.	2.6	10
76	Identification of a recurrent mosaic KRAS variant in brain tissue from an individual with nevus sebaceous syndrome. Journal of Physical Education and Sports Management, 2021, 7, a006133.	0.5	6
77	Natural History Studies and Clinical Trial Readiness for Genetic Developmental and Epileptic Encephalopathies. Neurotherapeutics, 2021, 18, 1432-1444.	2.1	22
78	Using common genetic variants to find drugs for common epilepsies. Brain Communications, 2021, 3, fcab287.	1.5	9
79	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.	0.7	14
80	Cognitive processes predicting advanced theory of mind in the broader autism phenotype. Autism Research, 2020, 13, 921-934.	2.1	10
81	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.	1.1	33
82	Deciphering the concepts behind â€œEpileptic encephalopathyâ€•and â€œDevelopmental and epileptic encephalopathyâ€• European Journal of Paediatric Neurology, 2020, 24, 11-14.	0.7	97
83	Levetiracetam efficacy in PCDH19 Girls Clustering Epilepsy. European Journal of Paediatric Neurology, 2020, 24, 142-147.	0.7	16
84	SCN1A Variants in vaccineâ€related febrile seizures: A prospective study. Annals of Neurology, 2020, 87, 281-288.	2.8	15
85	<scp>BRAT</scp>1 encephalopathy: a recessive cause of epilepsy of infancy with migrating focal seizures. Developmental Medicine and Child Neurology, 2020, 62, 1096-1099.	1.1	18
86	Germline and Mosaic Variants in PRKACA and PRKACB Cause a Multiple Congenital Malformation Syndrome. American Journal of Human Genetics, 2020, 107, 977-988.	2.6	33
87	Protocol for a single patient therapy plan: A randomised, doubleâ€blind, placeboâ€controlled Nâ€ofâ€1 trial to assess the efficacy of cannabidiol in patients with intractable epilepsy. Journal of Paediatrics and Child Health, 2020, 56, 1918-1923.	0.4	1
88	Cognitive, behavioral, and social functioning in children and adults with Dravet syndrome. Epilepsy and Behavior, 2020, 112, 107319.	0.9	21
89	Fenfluramine HCl (Fintepla^{Â®}) provides longâ€term clinically meaningful reduction in seizure frequency: Analysis of an ongoing openâ€label extension study. Epilepsia, 2020, 61, 2396-2404.	2.6	49
90	Predominant and novel de novo variants in 29 individuals with <scp>ALG13</scp> deficiency: Clinical description, biomarker status, biochemical analysis, and treatment suggestions. Journal of Inherited Metabolic Disease, 2020, 43, 1333-1348.	1.7	24

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91	Are Variants Causing Cardiac Arrhythmia Risk Factors in Sudden Unexpected Death in Epilepsy?. Frontiers in Neurology, 2020, 11, 925.	1.1	16
92	Developmental and epilepsy spectrum of KCNB1 encephalopathy with longâ€term outcome. Epilepsia, 2020, 61, 2461-2473.	2.6	17
93	PCDH19 Pathogenic Variants in Males: Expanding the Phenotypic Spectrum. Advances in Experimental Medicine and Biology, 2020, 1298, 177-187.	0.8	15
94	Cardiac phenotype in ATP1A3 -related syndromes. Neurology, 2020, 95, e2866-e2879.	1.5	19
95	Tracing Autism Traits in Large Multiplex Families to Identify Endophenotypes of the Broader Autism Phenotype. International Journal of Molecular Sciences, 2020, 21, 7965.	1.8	3
96	A standardized patient-centered characterization of the phenotypic spectrum of PCDH19 girls clustering epilepsy. Translational Psychiatry, 2020, 10, 127.	2.4	22
97	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	3.7	47
98	Focal epilepsy in SCN1Aâ€mutation carrying patients: is there a role for epilepsy surgery?. Developmental Medicine and Child Neurology, 2020, 62, 1331-1335.	1.1	20
99	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.	1.4	23
100	Author response: SYNGAP1 encephalopathy: A distinctive generalized developmental and epileptic encephalopathy. Neurology, 2020, 94, 370-370.	1.5	3
101	Damaging de novo missense variants inEEF1A2lead to a developmental and degenerative epilepticâ€dyskinetic encephalopathy. Human Mutation, 2020, 41, 1263-1279.	1.1	24
102	Bi-allelic LoF NRROS Variants Impairing Active TGF-Î²1 Delivery Cause a Severe Infantile-Onset Neurodegenerative Condition with Intracranial Calcification. American Journal of Human Genetics, 2020, 106, 559-569.	2.6	18
103	Antiepileptic Drug Teratogenicity and De Novo Genetic Variation Load. Annals of Neurology, 2020, 87, 897-906.	2.8	9
104	Inherited RORB pathogenic variants: Overlap of photosensitive genetic generalized and occipital lobe epilepsy. Epilepsia, 2020, 61, e23-e29.	2.6	14
105	Glut1 Deficiency Syndrome (Glut1DS): State of the art in 2020 and recommendations of the international Glut1DS study group. Epilepsia Open, 2020, 5, 354-365.	1.3	142
106	Defining the phenotype of FHF1 developmental and epileptic encephalopathy. Epilepsia, 2020, 61, e71-e78.	2.6	11
107	Pathogenic Variants in CEP85L Cause Sporadic and Familial Posterior Predominant Lissencephaly. Neuron, 2020, 106, 237-245.e8.	3.8	21
108	Dose-Ranging Effect of Adjunctive Oral Cannabidiol vs Placebo on Convulsive Seizure Frequency in Dravet Syndrome. JAMA Neurology, 2020, 77, 613.	4.5	171

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109	How gene discovery has transformed management of people with epilepsy. European Journal of Paediatric Neurology, 2020, 24, 2.	0.7	1
110	Severe childhood speech disorder. Neurology, 2020, 94, e2148-e2167.	1.5	68
111	Neuronal ceroid lipofuscinosis type 2: an Australian case series. Journal of Paediatrics and Child Health, 2020, 56, 1210-1218.	0.4	19
112	Keeping people with epilepsy safe during the COVID-19 pandemic. Neurology, 2020, 94, 1032-1037.	1.5	116
113	Parental health spillover effects of paediatric rare genetic conditions. Quality of Life Research, 2020, 29, 2445-2454.	1.5	28
114	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	5.8	105
115	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.	4.1	86
116	Generation of seven iPSC lines from peripheral blood mononuclear cells suitable to investigate Autism Spectrum Disorder. Stem Cell Research, 2019, 39, 101516.	0.3	4
117	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.	2.6	237
118	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	5.8	150
119	Secondâ€hit‹i› DEPDC5‹/i› mutation is limited to dysmorphic neurons in cortical dysplasia type IIA. Annals of Clinical and Translational Neurology, 2019, 6, 1338-1344.	1.7	55
120	Classification as autonomic versus sensory seizures. Epilepsia, 2019, 60, 2003-2005.	2.6	4
121	Somatic mutation: The hidden genetics of brain malformations and focal epilepsies. Epilepsy Research, 2019, 155, 106161.	0.8	45
122	Quantitative analysis of phenotypic elements augments traditional electroclinical classification of common familial epilepsies. Epilepsia, 2019, 60, 2194-2203.	2.6	0
123	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	5.8	43
124	When Monogenic Isnâ€™t Monogenicâ€"Unravelling the Oligogenic Architecture of the Developmental and Epileptic Encephalopathies. Epilepsy Currents, 2019, 19, 417-419.	0.4	5
125	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	5.8	99
126	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. Annals of Neurology, 2019, 86, 821-831.	2.8	96

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127	The epileptology of GNB5 encephalopathy. Epilepsia, 2019, 60, e121-e127.	2.6	13
128	Epidemiology and etiology of infantile developmental and epileptic encephalopathies in Tasmania. Epilepsia Open, 2019, 4, 504-510.	1.3	11
129	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.	0.5	3
130	Inhibition of Upf2-Dependent Nonsense-Mediated Decay Leads to Behavioral and Neurophysiological Abnormalities by Activating the Immune Response. Neuron, 2019, 104, 665-679.e8.	3.8	43
131	Efficacy and tolerability of adjunctive lacosamide in pediatric patients with focal seizures. Neurology, 2019, 93, e1212-e1226.	1.5	40
132	Spectrum of neurodevelopmental disease associated with the GNAO1 guanosine triphosphateâ€"binding region. Epilepsia, 2019, 60, 406-418.	2.6	53
133	Speech and language in bilateral perisylvian polymicrogyria: a systematic review. Developmental Medicine and Child Neurology, 2019, 61, 1145-1152.	1.1	9
134	Autism and developmental disability caused by KCNQ3 gainâ€ofâ€function variants. Annals of Neurology, 2019, 86, 181-192.	2.8	73
135	Double somatic mosaicism in a child with Dravet syndrome. Neurology: Genetics, 2019, 5, e333.	0.9	7
136	Splice variant in ARX leading to loss of Câ€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.	0.7	9
137	2017 International League Against Epilepsy classifications of seizures and epilepsy are steps in the right direction. Epilepsia, 2019, 60, 1040-1044.	2.6	15
138	Recessive variants in ZNF142 cause a complex neurodevelopmental disorder with intellectual disability, speech impairment, seizures, and dystonia. Genetics in Medicine, 2019, 21, 2532-2542.	1.1	17
139	Intestinal-Cell Kinase and Juvenile Myoclonic Epilepsy. New England Journal of Medicine, 2019, 380, e24.	13.9	4
140	Schizophrenia is a laterâ€onset feature of <scp>PCDH</scp>19 Girls Clustering Epilepsy. Epilepsia, 2019, 60, 429-440.	2.6	23
141	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.	1.1	39
142	Why should a neurologist worry about climate change?. Lancet Neurology, The, 2019, 18, 335-336.	4.9	3
143	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	2.6	45
144	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. Epilepsia, 2019, 60, 797-806.	2.6	52

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145	Dorsal language stream anomalies in an inherited speech disorder. Brain, 2019, 142, 966-977.	3.7	16
146	No evidence for a BRD 2 promoter hypermethylation inÂblood leukocytes of Europeans with juvenile myoclonicÂepilepsy. Epilepsia, 2019, 60, e31-e36.	2.6	4
147	SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	1.1	39
148	009â€…Axonal excitability properties in dravetâ€™s syndrome reflect effect of loss of sodium channels. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, A4.1-A4.	0.9	0
149	Human GABRG2 generalized epilepsy. Neurology: Genetics, 2019, 5, e340.	0.9	6
150	Effect of ZX008 (Fenfluramine HCl Oral Solution) on Total Seizures in Dravet Syndrome. Epilepsy and Behavior, 2019, 101, 106789.	0.9	0
151	Fragile Females: Case Series of Epilepsy in Girls With FMR1 Disruption. Pediatrics, 2019, 144, .	1.0	5
152	Long-Term Cardiovascular Safety of Fenfluramine HCl in the Treatment of Dravet Syndrome: Interim Analysis of an Open-Label Safety Extension Study. Epilepsy and Behavior, 2019, 101, 106791.	0.9	0
153	Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3. Nature Communications, 2019, 10, 4919.	5.8	111
154	SCN1Aâ€related phenotypes: Epilepsy and beyond. Epilepsia, 2019, 60, S17-S24.	2.6	103
155	Reanalysis and optimisation of bioinformatic pipelines is critical for mutation detection. Human Mutation, 2019, 40, 374-379.	1.1	7
156	SYNGAP1 encephalopathy. Neurology, 2019, 92, e96-e107.	1.5	131
157	Efficacy of cannabinoids in paediatric epilepsy. Developmental Medicine and Child Neurology, 2019, 61, 13-18.	1.1	30
158	A set of regulatory genes co-expressed in embryonic human brain is implicated in disrupted speech development. Molecular Psychiatry, 2019, 24, 1065-1078.	4.1	106
159	OUP accepted manuscript. Brain, 2019, 142, 2173-2175.	3.7	0
160	(CBD) Significantly Reduces Convulsive Seizure Frequency in Dravet Syndrome: Results of a Dose-Ranging, Multicentre, Randomised, Double-Blind, Placebo-Controlled Trial. , 2019, 50, .		0
161	Stiripentol efficacy and safety in Dravet syndrome: a 12â€year observational study. Developmental Medicine and Child Neurology, 2018, 60, 574-578.	1.1	51
162	A case series of lacosamide as adjunctive therapy in refractory sleepâ€related hypermotor epilepsy (previously nocturnal frontal lobe epilepsy). Journal of Sleep Research, 2018, 27, e12669.	1.7	10

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163	Childhood-onset generalized epilepsy in Bainbridge-Ropers syndrome. Epilepsy Research, 2018, 140, 166-170.	0.8	12
164	Early mortality in SCN8A -related epilepsies. Epilepsy Research, 2018, 143, 79-81.	0.8	48
165	Parental Mosaicism in â€œDe Novoâ€•Epileptic Encephalopathies. New England Journal of Medicine, 2018, 378, 1646-1648.	13.9	104
166	The ventrolateral medulla and medullary raphe in sudden unexpected death in epilepsy. Brain, 2018, 141, 1719-1733.	3.7	80
167	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. Annals of Neurology, 2018, 83, 926-934.	2.8	20
168	Hemiconvulsion-hemiplegia-epilepsy evolving to contralateral hemi-Lennox-Gastaut-like phenotype. Brain and Development, 2018, 40, 425-428.	0.6	3
169	The ketogenic diet is effective for refractory epilepsy associated with acquired structural epileptic encephalopathy. Developmental Medicine and Child Neurology, 2018, 60, 718-723.	1.1	8
170	Ion Channels in Genetic Epilepsy: From Genes and Mechanisms to Disease-Targeted Therapies. Pharmacological Reviews, 2018, 70, 142-173.	7.1	215
171	Myoclonic absence seizures with complex gestural automatisms. European Journal of Paediatric Neurology, 2018, 22, 532-535.	0.7	4
172	ADGRV1 is implicated in myoclonic epilepsy. Epilepsia, 2018, 59, 381-388.	2.6	31
173	Abnormal Cell Sorting Underlies the Unique X-Linked Inheritance of PCDH19 Epilepsy. Neuron, 2018, 97, 59-66.e5.	3.8	100
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