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List of Publications by Year
in descending order

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104 papers	4,407 citations	126708 33 h-index	123241 61 g-index
107 all docs	107 docs citations	107 times ranked	7159 citing authors

#	Article	IF	Citations
1	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. Nature Genetics, 2013, 45, 825-830.	9.4	589
2	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
3	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. Nature Genetics, 2015, 47, 39-46.	9.4	245
4	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
5	Practical guidelines for managing adults with 22q11.2 deletion syndrome. Genetics in Medicine, 2015, 17, 599-609.	1.1	222
6	Mutation of the CLN6 Gene in Teenage-Onset Progressive Myoclonus Epilepsy. Pediatric Neurology, 2012, 47, 205-208.	1.0	143
7	Tonic seizures: A diagnostic clue of anti-LGI1 encephalitis?. Neurology, 2011, 76, 1355-1357.	1.5	135
8	Deep brain stimulation of the anterior nucleus of the thalamus: Effects of electrical stimulation on pilocarpine-induced seizures and status epilepticus. Epilepsy Research, 2008, 78, 117-123.	0.8	113
9	DEEP BRAIN STIMULATION FOR THE TREATMENT OF EPILEPSY. International Journal of Neural Systems, 2009, 19, 213-226.	3.2	105
10	Delineating the 15q13.3 microdeletion phenotype: a case series and comprehensive review of the literature. Genetics in Medicine, 2015, 17, 149-157.	1.1	103
11	Anterior Nucleus Deep Brain Stimulation for Refractory Epilepsy. Neurosurgery, 2016, 78, 802-811.	0.6	100
12	Epilepsy: Transition from pediatric to adult care. Recommendations of the Ontario epilepsy implementation task force. Epilepsia, 2017, 58, 1502-1517.	2.6	74
13	Neonatal hypocalcemia, neonatal seizures, and intellectual disability in 22q11.2 deletion syndrome. Genetics in Medicine, 2014, 16, 40-44.	1.1	73
14	Mitochondrial Encephalopathy With Lactic Acidosis and Stroke-like Episodes (MELAS) May Respond to Adjunctive Ketogenic Diet. Pediatric Neurology, 2014, 50, 498-502.	1.0	72
15	Prevalence of Pathogenic Copy Number Variation in Adults With Pediatric-Onset Epilepsy and Intellectual Disability. JAMA Neurology, 2017, 74, 1301.	4.5	72
16	Prevalence of hypocalcaemia and its associated features in 22q11Â·2 deletion syndrome. Clinical Endocrinology, 2014, 81, 190-196.	1.2	64
17	Biallelic Truncating Mutations in FMN2, Encoding the Actin-Regulatory Protein Formin 2, Cause Nonsyndromic Autosomal-Recessive Intellectual Disability. American Journal of Human Genetics, 2014, 95, 721-728.	2.6	62
18	Response to clozapine in a clinically identifiable subtype of schizophrenia. British Journal of Psychiatry, 2015, 206, 484-491.	1.7	61

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19	Dravet syndrome: Seizure control and gait in adults with different SCN1A mutations. Epilepsia, 2012, 53, 1421-1428.	2.6	58
20	Genetic basis in epilepsies caused by malformations of cortical development and in those with structurally normal brain. Human Genetics, 2009, 126, 173-193.	1.8	56
21	Antecollis and levodopa-responsive parkinsonism are late features of Dravet syndrome. Neurology, 2014, 82, 2250-2251.	1.5	56
22	Laforin is a cell membrane and endoplasmic reticulum-associated protein tyrosine phosphatase. Annals of Neurology, 2001, 49, 271-275.	2.8	52
23	Global characterization of copy number variants in epilepsy patients from whole genome sequencing. PLoS Genetics, 2018, 14, e1007285.	1.5	50
24	Movement disorders and other motor abnormalities in adults with 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 639-645.	0.7	49
25	Epilepsy transition: Challenges of caring for adults with childhoodâ€onset seizures. Epilepsia, 2014, 55, 1659-1666.	2.6	48
26	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	3.7	47
27	Socioeconomic status influences time to surgery and surgical outcome in pediatric epilepsy surgery. Epilepsy and Behavior, 2016, 55, 133-138.	0.9	45
28	Whole-Genome Sequencing Suggests Schizophrenia Risk Mechanisms in Humans with 22q11.2 Deletion Syndrome. G3: Genes, Genomes, Genetics, 2015, 5, 2453-2461.	0.8	43
29	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. American Journal of Human Genetics, 2017, 101, 516-524.	2.6	43
30	Two definite cases of sudden unexpected death in epilepsy in a family with a DEPDC5 mutation. Neurology: Genetics, 2015, 1, e28.	0.9	42
31	Phenotypic variability in hyperphosphatasia with seizures and neurologic deficit (Mabry syndrome). American Journal of Medical Genetics, Part A, 2012, 158A, 553-558.	0.7	40
32	How can transition to adult care be best orchestrated for adolescents with epilepsy?. Epilepsy and Behavior, 2019, 93, 138-147.	0.9	39
33	Neurocognitive and Seizure Outcomes of Selective Amygdalohippocampectomy versus Anterior Temporal Lobectomy for Mesial Temporal Lobe Epilepsy. Epilepsy Research & Treatment, 2014, 2014, 1-8.	1.4	37
34	Clinical utility of multigene panel testing in adults with epilepsy and intellectual disability. Epilepsia, 2019, 60, 1661-1669.	2.6	37
35	Evaluating risk to people with epilepsy during the COVID-19 pandemic: Preliminary findings from the COV-E study. Epilepsy and Behavior, 2021, 115, 107658.	0.9	37
36	Hippocampal Malrotation is Associated with Chromosome 22q11.2 Microdeletion. Canadian Journal of Neurological Sciences, 2013, 40, 652-656.	0.3	35

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37	A pilot double-blind trial using verapamil as adjuvant therapy for refractory seizures. Epilepsy Research, 2014, 108, 1642-1651.	0.8	33
38	Treatment issues for children with epilepsy transitioning to adult care. Epilepsy and Behavior, 2017, 69, 153-160.	0.9	33
39	Adult motor phenotype differentiates Dravet syndrome from Lennoxâ€Gastaut syndrome and links <scp>SCN</scp>1A to early onset parkinsonian features. Epilepsia, 2017, 58, e44-e48.	2.6	32
40	22q11.2 deletion syndrome lowers seizure threshold in adult patients without epilepsy. Epilepsia, 2017, 58, 1095-1101.	2.6	31
41	Which patients with epilepsy are at risk for psychogenic nonepileptic seizures (PNES)? A multicenter caseâ€“control study. Epilepsy and Behavior, 2016, 61, 180-184.	0.9	29
42	A systematic review of adults with Dravet syndrome. Seizure: the Journal of the British Epilepsy Association, 2021, 87, 39-45.	0.9	29
43	Myoclonus epilepsy and ataxia due to potassium channel mutation (MEAK) is caused by heterozygous KCNC1 mutations. Epileptic Disorders, 2016, 18, 135-138.	0.7	28
44	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	1.1	28
45	Source localization of small sharp spikes: Low resolution electromagnetic tomography (LORETA) reveals two distinct cortical sources. Clinical Neurophysiology, 2006, 117, 1380-1387.	0.7	27
46	Frontal infraslow activity marks the motor spasms of anti-LGI1 encephalitis. Clinical Neurophysiology, 2018, 129, 59-68.	0.7	26
47	Neuropsychiatric expression and catatonia in 22q11.2 deletion syndrome: An overview and case series. American Journal of Medical Genetics, Part A, 2018, 176, 2146-2159.	0.7	25
48	Dravet syndrome and deep brain stimulation: Seizure control after 10â€ƒyears of treatment. Epilepsia, 2010, 51, 1314-1316.	2.6	24
49	Schizophrenia is a laterâ€onset feature of <scp>PCDH</scp>19 Girls Clustering Epilepsy. Epilepsia, 2019, 60, 429-440.	2.6	23
50	Seizures and movement disorders: phenomenology, diagnostic challenges and therapeutic approaches. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 920-928.	0.9	22
51	Parietal lobe source localization and sensitivity to hyperventilation in a patient with subclinical rhythmic electrographic discharges of adults (SREDA). Clinical Neurophysiology, 2006, 117, 2257-2263.	0.7	21
52	Trait impulsivity in Juvenile Myoclonic Epilepsy. Annals of Clinical and Translational Neurology, 2021, 8, 138-152.	1.7	21
53	Genetics of epilepsies. Expert Review of Neurotherapeutics, 2007, 7, 727-734.	1.4	20
54	Sex-specific disease modifiers in juvenile myoclonic epilepsy. Scientific Reports, 2022, 12, 2785.	1.6	19

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55	STXBP1 encephalopathy is associated with awake bruxism. Epilepsy and Behavior, 2019, 92, 121-124.	0.9	18
56	Development of Criteria for Epilepsy Genetic Testing in Ontario, Canada. Canadian Journal of Neurological Sciences, 2019, 46, 7-13.	0.3	18
57	Microinjection of GABAergic agents into the anterior nucleus of the thalamus modulates pilocarpine-induced seizures and status epilepticus. Seizure: the Journal of the British Epilepsy Association, 2010, 19, 242-246.	0.9	16
58	Identification of a homozygous missense mutation in LRP2 and a hemizygous missense mutation in TSPYL2 in a family with mild intellectual disability. Psychiatric Genetics, 2016, 26, 66-73.	0.6	16
59	Chromosome 1p36 in migraine with aura. NeuroReport, 2012, 23, 45-48.	0.6	14
60	Progressive myoclonus epilepsy associated with SACS gene mutations. Neurology: Genetics, 2016, 2, e83.	0.9	14
61	Starting stiripentol in adults with Dravet syndrome? Watch for ammonia and carnitine. Epilepsia, 2020, 61, 2435-2441.	2.6	13
62	Blood oxygen level-dependent (BOLD) response patterns with thalamic deep brain stimulation in patients with medically refractory epilepsy. Epilepsy and Behavior, 2021, 122, 108153.	0.9	13
63	COVIDâ€19 vaccine in patients with Dravet syndrome: Observations and realâ€world experiences. Epilepsia, 2022, 63, 1778-1786.	2.6	13
64	Genetics of Epileptic Networks: from Focal to Generalized Genetic Epilepsies. Current Neurology and Neuroscience Reports, 2020, 20, 46.	2.0	12
65	Daily listening to Mozart reduces seizures in individuals with epilepsy: A randomized control study. Epilepsia Open, 2020, 5, 285-294.	1.3	12
66	Atypical absences and recurrent absence status in an adult with Angelman syndrome due to the UBE3A mutation. Epileptic Disorders, 2005, 7, 227-30.	0.7	12
67	Periventricular nodular heterotopia in 22q11.2 deletion and frontal lobe migration. Annals of Clinical and Translational Neurology, 2018, 5, 1314-1322.	1.7	11
68	Dravet syndrome: A quick transition guide for the adult neurologist. Epilepsy Research, 2021, 177, 106743.	0.8	11
69	Hemimegalencephaly: what happens when children get older?. Developmental Medicine and Child Neurology, 2014, 56, 905-909.	1.1	10
70	Progressive Worsening of Gait and Motor Abnormalities in Older Adults With Dravet Syndrome. Neurology, 2022, 98, .	1.5	10
71	Deep brain stimulation for the management of seizures in MECP2 duplication syndrome. Seizure: the Journal of the British Epilepsy Association, 2014, 23, 405-407.	0.9	9
72	Treatment options for epileptic myoclonus and epilepsy syndromes associated with myoclonus. Expert Opinion on Pharmacotherapy, 2009, 10, 1549-1560.	0.9	8

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73	Impact of the COVID-19 pandemic on people with epilepsy: Findings from the Brazilian arm of the COV-E study. Epilepsy and Behavior, 2021, 123, 108261.	0.9	8
74	Movement disorders phenomenology in focal motor seizures. Parkinsonism and Related Disorders, 2019, 61, 161-165.	1.1	7
75	Dravet syndrome, lamotrigine, and personalized medicine. Developmental Medicine and Child Neurology, 2015, 57, 118-119.	1.1	6
76	Unilateral abdominal clonic seizures of parietal lobe origin: EEG findings. Epileptic Disorders, 2018, 20, 158-163.	0.7	6
77	The phenotype of bilateral hippocampal sclerosis and its management in â€œreal lifeâ€•clinical settings. Epilepsia, 2018, 59, 1410-1420.	2.6	6
78	Protein therapy for Unverrichtâ€“Lundborg disease using cystatin B transduction by TAT-PTD. Epilepsy Research, 2006, 72, 75-79.	0.8	5
79	GRIN1 polymorphisms do not affect susceptibility or phenotype in NMDA receptor encephalitis. Neurology: Neuroimmunology and NeuroInflammation, 2015, 2, e153.	3.1	5
80	A comparison of antiepileptic drug therapy in patients with severe intellectual disability and patients with normal intellect. Epilepsy and Behavior, 2012, 25, 196-199.	0.9	4
81	Genome sequencing identifies rare tandem repeat expansions and copy number variants in Lennoxâ€“Gastaut syndrome. Brain Communications, 2021, 3, fcab207.	1.5	4
82	Multistage preictal seizure analysis using Hidden Markov Model. International Journal of Biomedical Engineering and Technology, 2012, 10, 160.	0.2	3
83	Epilepsy Transition: Let's start planting the seed. European Journal of Paediatric Neurology, 2016, 20, 684-685.	0.7	3
84	Genetic generalized epilepsy in three siblings with 8q21.13-q22.2 duplication. Seizure: the Journal of the British Epilepsy Association, 2017, 48, 57-61.	0.9	3
85	Cerebral Corticoarterial Malformations. Clinical Neuroradiology, 2020, 30, 389-394.	1.0	3
86	Hyperammonemic Encephalopathy Associated with Perampanel: Case Report and Discussion. Canadian Journal of Neurological Sciences, 2021, 48, 438-439.	0.3	3
87	Genomic analysis of â€œmicrophenotypesâ€•in epilepsy. American Journal of Medical Genetics, Part A, 2022, 188, 138-146.	0.7	3
88	Clinical aspects of temporal/limbic epilepsy and their relationships to intractability. Advances in Neurology, 2006, 97, 39-44.	0.8	3
89	Seizure Recurrence 29 Years After Hemispherectomy for Sturge Weber Syndrome. Canadian Journal of Neurological Sciences, 2010, 37, 141-144.	0.3	2
90	Periventricular nodular heterotopia and bilateral intraventricular xanthogranulomas in 22q11.2 deletion syndrome. Human Pathology: Case Reports, 2017, 9, 55-57.	0.2	2

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91	Adults with tuberous sclerosis complex: A distinct patient population. Epilepsia, 2022, 63, 663-671.	2.6	2
92	On the need for battery replacement before end of service in vagus nerve stimulation for epilepsy. Epilepsy and Behavior, 2004, 5, 612-613.	0.9	1
93	Novel antiseizure drug mechanisms. Future Neurology, 2007, 2, 73-86.	0.9	1
94	Nonlesional Focal Epilepsy: A Challenge from Genes to Surgery. Canadian Journal of Neurological Sciences, 2013, 40, 137-138.	0.3	1
95	Reply From the Authors. Pediatric Neurology, 2014, 51, e5-e6.	1.0	1
96	Multimedia teaching material. Epileptic Disorders, 2016, 18, 216-216.	0.7	1
97	Epilepsy gene panel yield and impact on outcomes for adults with unexplained seizures. Epilepsia, 2020, 61, 1797-1798.	2.6	1
98	Alfentanil induced electrocorticographic activation: A promising tool for presurgical evaluation of temporallobe epilepsy (TLE) patients. Canadian Journal of Anaesthesia, 2008, 55, 4736991-4736992.	0.7	0
99	Temporal Lobe Epilepsy and Hippocampal Stimulation. Canadian Journal of Neurological Sciences, 2012, 39, 830-832.	0.3	0
100	The multiple faces of Dravet syndrome. Developmental Medicine and Child Neurology, 2014, 56, 10-11.	1.1	0
101	Seizures and early onset dementia: D2HGA1 inborn error of metabolism in adults. Annals of Clinical and Translational Neurology, 2020, 7, 2052-2056.	1.7	0
102	Precision medicine for epilepsies: are we there yet?. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 1032-1032.	0.9	0
103	Valproic Acid and Pregnancy: Failed Other Medications. , 2016, , 63-71.		0
104	Time Is Brain: The Importance of an Accurate SCN1A Prediction Score in the Era of Precision Medicine. Epilepsy Currents, 0, , 153575972210960.	0.4	0