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List of Publications by Year
in descending order

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104 papers	4,407 citations	126907 33 h-index	123424 61 g-index
107 all docs	107 docs citations	107 times ranked	7159 citing authors

#	Article	IF	Citations
1	Genomic analysis of â€œmicrophenotypesâ€•in epilepsy. American Journal of Medical Genetics, Part A, 2022, 188, 138-146.	1.2	3
2	Sex-specific disease modifiers in juvenile myoclonic epilepsy. Scientific Reports, 2022, 12, 2785.	3.3	19
3	COVIDâ€19 vaccine in patients with Dravet syndrome: Observations and realâ€world experiences. Epilepsia, 2022, 63, 1778-1786.	5.1	13
4	Adults with tuberous sclerosis complex: A distinct patient population. Epilepsia, 2022, 63, 663-671.	5.1	2
5	Progressive Worsening of Gait and Motor Abnormalities in Older Adults With Dravet Syndrome. Neurology, 2022, 98, .	1.1	10
6	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	2.4	28
7	Trait impulsivity in Juvenile Myoclonic Epilepsy. Annals of Clinical and Translational Neurology, 2021, 8, 138-152.	3.7	21
8	Evaluating risk to people with epilepsy during the COVID-19 pandemic: Preliminary findings from the COV-E study. Epilepsy and Behavior, 2021, 115, 107658.	1.7	37
9	Hyperammonemic Encephalopathy Associated with Perampanel: Case Report and Discussion. Canadian Journal of Neurological Sciences, 2021, 48, 438-439.	0.5	3
10	A systematic review of adults with Dravet syndrome. Seizure: the Journal of the British Epilepsy Association, 2021, 87, 39-45.	2.0	29
11	Precision medicine for epilepsies: are we there yet?. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 1032-1032.	1.9	0
12	Dravet syndrome: A quick transition guide for the adult neurologist. Epilepsy Research, 2021, 177, 106743.	1.6	11
13	Blood oxygen level-dependent (BOLD) response patterns with thalamic deep brain stimulation in patients with medically refractory epilepsy. Epilepsy and Behavior, 2021, 122, 108153.	1.7	13
14	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.	1.7	8
15	Genome sequencing identifies rare tandem repeat expansions and copy number variants in Lennoxâ€“Gastaut syndrome. Brain Communications, 2021, 3, fcab207.	3.3	4
16	Cerebral Corticoarterial Malformations. Clinical Neuroradiology, 2020, 30, 389-394.	1.9	3
17	Epilepsy gene panel yield and impact on outcomes for adults with unexplained seizures. Epilepsia, 2020, 61, 1797-1798.	5.1	1
18	Genetics of Epileptic Networks: from Focal to Generalized Genetic Epilepsies. Current Neurology and Neuroscience Reports, 2020, 20, 46.	4.2	12

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19	Starting stiripentol in adults with Dravet syndrome? Watch for ammonia and carnitine. Epilepsia, 2020, 61, 2435-2441.	5.1	13
20	Seizures and early onset dementia: D2HGA1 inborn error of metabolism in adults. Annals of Clinical and Translational Neurology, 2020, 7, 2052-2056.	3.7	0
21	Daily listening to Mozart reduces seizures in individuals with epilepsy: A randomized control study. Epilepsia Open, 2020, 5, 285-294.	2.4	12
22	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	7.6	47
23	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
24	Clinical utility of multigene panel testing in adults with epilepsy and intellectual disability. Epilepsia, 2019, 60, 1661-1669.	5.1	37
25	STXBP1 encephalopathy is associated with awake bruxism. Epilepsy and Behavior, 2019, 92, 121-124.	1.7	18
26	Schizophrenia is a laterâ€onset feature of <scp>PCDH</scp>19 Girls Clustering Epilepsy. Epilepsia, 2019, 60, 429-440.	5.1	23
27	Seizures and movement disorders: phenomenology, diagnostic challenges and therapeutic approaches. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 920-928.	1.9	22
28	Development of Criteria for Epilepsy Genetic Testing in Ontario, Canada. Canadian Journal of Neurological Sciences, 2019, 46, 7-13.	0.5	18
29	How can transition to adult care be best orchestrated for adolescents with epilepsy?. Epilepsy and Behavior, 2019, 93, 138-147.	1.7	39
30	Movement disorders phenomenology in focal motor seizures. Parkinsonism and Related Disorders, 2019, 61, 161-165.	2.2	7
31	Frontal infraslow activity marks the motor spasms of anti-LGI1 encephalitis. Clinical Neurophysiology, 2018, 129, 59-68.	1.5	26
32	Unilateral abdominal clonic seizures of parietal lobe origin: EEG findings. Epileptic Disorders, 2018, 20, 158-163.	1.3	6
33	Periventricular nodular heterotopia in 22q11.2 deletion and frontal lobe migration. Annals of Clinical and Translational Neurology, 2018, 5, 1314-1322.	3.7	11
34	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.	1.2	25
35	Global characterization of copy number variants in epilepsy patients from whole genome sequencing. PLoS Genetics, 2018, 14, e1007285.	3.5	50
36	The phenotype of bilateral hippocampal sclerosis and its management in â€œreal lifeâ€•clinical settings. Epilepsia, 2018, 59, 1410-1420.	5.1	6

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37	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.	5.1	32
38	Treatment issues for children with epilepsy transitioning to adult care. Epilepsy and Behavior, 2017, 69, 153-160.	1.7	33
39	Genetic generalized epilepsy in three siblings with 8q21.13-q22.2 duplication. Seizure: the Journal of the British Epilepsy Association, 2017, 48, 57-61.	2.0	3
40	22q11.2 deletion syndrome lowers seizure threshold in adult patients without epilepsy. Epilepsia, 2017, 58, 1095-1101.	5.1	31
41	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
42	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. American Journal of Human Genetics, 2017, 101, 516-524.	6.2	43
43	Prevalence of Pathogenic Copy Number Variation in Adults With Pediatric-Onset Epilepsy and Intellectual Disability. JAMA Neurology, 2017, 74, 1301.	9.0	72
44	Epilepsy: Transition from pediatric to adult care. Recommendations of the Ontario epilepsy implementation task force. Epilepsia, 2017, 58, 1502-1517.	5.1	74
45	Periventricular nodular heterotopia and bilateral intraventricular xanthogranulomas in 22q11.2 deletion syndrome. Human Pathology: Case Reports, 2017, 9, 55-57.	0.2	2
46	Anterior Nucleus Deep Brain Stimulation for Refractory Epilepsy. Neurosurgery, 2016, 78, 802-811.	1.1	100
47	Which patients with epilepsy are at risk for psychogenic nonepileptic seizures (PNES)? A multicenter caseâ€“control study. Epilepsy and Behavior, 2016, 61, 180-184.	1.7	29
48	Myoclonus epilepsy and ataxia due to potassium channel mutation (MEAK) is caused by heterozygous KCNC1 mutations. Epileptic Disorders, 2016, 18, 135-138.	1.3	28
49	Progressive myoclonus epilepsy associated with SACS gene mutations. Neurology: Genetics, 2016, 2, e83.	1.9	14
50	Multimedia teaching material. Epileptic Disorders, 2016, 18, 216-216.	1.3	1
51	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.	1.1	16
52	Epilepsy Transition: Let's start planting the seed. European Journal of Paediatric Neurology, 2016, 20, 684-685.	1.6	3
53	Socioeconomic status influences time to surgery and surgical outcome in pediatric epilepsy surgery. Epilepsy and Behavior, 2016, 55, 133-138.	1.7	45
54	Valproic Acid and Pregnancy: Failed Other Medications. , 2016, , 63-71.		0

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55	Two definite cases of sudden unexpected death in epilepsy in a family with a DEPDC5 mutation. Neurology: Genetics, 2015, 1, e28.	1.9	42
56	GRIN1 polymorphisms do not affect susceptibility or phenotype in NMDA receptor encephalitis. Neurology: Neuroimmunology and NeuroInflammation, 2015, 2, e153.	6.0	5
57	Whole-Genome Sequencing Suggests Schizophrenia Risk Mechanisms in Humans with 22q11.2 Deletion Syndrome. G3: Genes, Genomes, Genetics, 2015, 5, 2453-2461.	1.8	43
58	Movement disorders and other motor abnormalities in adults with 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 639-645.	1.2	49
59	Practical guidelines for managing adults with 22q11.2 deletion syndrome. Genetics in Medicine, 2015, 17, 599-609.	2.4	222
60	Response to clozapine in a clinically identifiable subtype of schizophrenia. British Journal of Psychiatry, 2015, 206, 484-491.	2.8	61
61	Dravet syndrome, lamotrigine, and personalized medicine. Developmental Medicine and Child Neurology, 2015, 57, 118-119.	2.1	6
62	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. Nature Genetics, 2015, 47, 39-46.	21.4	245
63	Delineating the 15q13.3 microdeletion phenotype: a case series and comprehensive review of the literature. Genetics in Medicine, 2015, 17, 149-157.	2.4	103
64	Epilepsy transition: Challenges of caring for adults with childhoodâ€onset seizures. Epilepsia, 2014, 55, 1659-1666.	5.1	48
65	Antecollis and levodopa-responsive parkinsonism are late features of Dravet syndrome. Neurology, 2014, 82, 2250-2251.	1.1	56
66	Neonatal hypocalcemia, neonatal seizures, and intellectual disability in 22q11.2 deletion syndrome. Genetics in Medicine, 2014, 16, 40-44.	2.4	73
67	Hemimegalencephaly: what happens when children get older?. Developmental Medicine and Child Neurology, 2014, 56, 905-909.	2.1	10
68	Prevalence of hypocalcaemia and its associated features in 22q11Â·2 deletion syndrome. Clinical Endocrinology, 2014, 81, 190-196.	2.4	64
69	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.	6.2	62
70	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
71	Mitochondrial Encephalopathy With Lactic Acidosis and Stroke-like Episodes (MELAS) May Respond to Adjunctive Ketogenic Diet. Pediatric Neurology, 2014, 50, 498-502.	2.1	72
72	The multiple faces of Dravet syndrome. Developmental Medicine and Child Neurology, 2014, 56, 10-11.	2.1	0

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73	A pilot double-blind trial using verapamil as adjuvant therapy for refractory seizures. Epilepsy Research, 2014, 108, 1642-1651.	1.6	33
74	Deep brain stimulation for the management of seizures in MECP2 duplication syndrome. Seizure: the Journal of the British Epilepsy Association, 2014, 23, 405-407.	2.0	9
75	Reply From the Authors. Pediatric Neurology, 2014, 51, e5-e6.	2.1	1
76	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. Nature Genetics, 2013, 45, 825-830.	21.4	589
77	Hippocampal Malrotation is Associated with Chromosome 22q11.2 Microdeletion. Canadian Journal of Neurological Sciences, 2013, 40, 652-656.	0.5	35
78	Nonlesional Focal Epilepsy: A Challenge from Genes to Surgery. Canadian Journal of Neurological Sciences, 2013, 40, 137-138.	0.5	1
79	Chromosome 1p36 in migraine with aura. NeuroReport, 2012, 23, 45-48.	1.2	14
80	Temporal Lobe Epilepsy and Hippocampal Stimulation. Canadian Journal of Neurological Sciences, 2012, 39, 830-832.	0.5	0
81	Multistage preictal seizure analysis using Hidden Markov Model. International Journal of Biomedical Engineering and Technology, 2012, 10, 160.	0.2	3
82	Dravet syndrome: Seizure control and gait in adults with different SCN1A mutations. Epilepsia, 2012, 53, 1421-1428.	5.1	58
83	A comparison of antiepileptic drug therapy in patients with severe intellectual disability and patients with normal intellect. Epilepsy and Behavior, 2012, 25, 196-199.	1.7	4
84	Mutation of the CLN6 Gene in Teenage-Onset Progressive Myoclonus Epilepsy. Pediatric Neurology, 2012, 47, 205-208.	2.1	143
85	Phenotypic variability in hyperphosphatasia with seizures and neurologic deficit (Mabry syndrome). American Journal of Medical Genetics, Part A, 2012, 158A, 553-558.	1.2	40
86	Tonic seizures: A diagnostic clue of anti-LGI1 encephalitis?. Neurology, 2011, 76, 1355-1357.	1.1	135
87	Dravet syndrome and deep brain stimulation: Seizure control after 10â€ƒyears of treatment. Epilepsia, 2010, 51, 1314-1316.	5.1	24
88	Seizure Recurrence 29 Years After Hemispherectomy for Sturge Weber Syndrome. Canadian Journal of Neurological Sciences, 2010, 37, 141-144.	0.5	2
89	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.	2.0	16
90	DEEP BRAIN STIMULATION FOR THE TREATMENT OF EPILEPSY. International Journal of Neural Systems, 2009, 19, 213-226.	5.2	105

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91	Treatment options for epileptic myoclonus and epilepsy syndromes associated with myoclonus. Expert Opinion on Pharmacotherapy, 2009, 10, 1549-1560.	1.8	8
92	Genetic basis in epilepsies caused by malformations of cortical development and in those with structurally normal brain. Human Genetics, 2009, 126, 173-193.	3.8	56
93	Alfentanil induced electrocorticographic activation: A promising tool for presurgical evaluation of temporallobe epilepsy (TLE) patients. Canadian Journal of Anaesthesia, 2008, 55, 4736991-4736992.	1.6	0
94	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.	1.6	113
95	Novel antiseizure drug mechanisms. Future Neurology, 2007, 2, 73-86.	0.5	1
96	Genetics of epilepsies. Expert Review of Neurotherapeutics, 2007, 7, 727-734.	2.8	20
97	Source localization of small sharp spikes: Low resolution electromagnetic tomography (LORETA) reveals two distinct cortical sources. Clinical Neurophysiology, 2006, 117, 1380-1387.	1.5	27
98	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.	1.5	21
99	Protein therapy for Unverrichtâ€“Lundborg disease using cystatin B transduction by TAT-PTD. Epilepsy Research, 2006, 72, 75-79.	1.6	5
100	Clinical aspects of temporal/limbic epilepsy and their relationships to intractability. Advances in Neurology, 2006, 97, 39-44.	0.8	3
101	Atypical absences and recurrent absence status in an adult with Angelman syndrome due to the UBE3A mutation. Epileptic Disorders, 2005, 7, 227-30.	1.3	12
102	On the need for battery replacement before end of service in vagus nerve stimulation for epilepsy. Epilepsy and Behavior, 2004, 5, 612-613.	1.7	1
103	Laforin is a cell membrane and endoplasmic reticulum-associated protein tyrosine phosphatase. Annals of Neurology, 2001, 49, 271-275.	5.3	52
104	Time Is Brain: The Importance of an Accurate SCN1A Prediction Score in the Era of Precision Medicine. Epilepsy Currents, 0, , 153575972210960.	0.8	0