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List of Publications by Year
in descending order

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53 papers	5,182 citations	136950 32 h-index	182427 51 g-index
53 all docs	53 docs citations	53 times ranked	4856 citing authors

#	Article	IF	Citations
1	Association of SLC32A1 Missense Variants With Genetic Epilepsy With Febrile Seizures Plus. Neurology, 2021, 96, e2251-e2260.	1.1	13
2	Integrated in silico and experimental assessment of disease relevance of PCDH19 Âmissense variants. Human Mutation, 2021, 42, 1030-1041.	2.5	1
3	Two novel intragenic variants in the FMR1 gene in patients with suspect clinical diagnosis of Fragile X syndrome and no CGG repeat expansion. European Journal of Medical Genetics, 2020, 63, 104010.	1.3	4
4	Benign infantile seizures and paroxysmal dyskinesia caused by an SCN8A mutation. Annals of Neurology, 2016, 79, 428-436.	5.3	159
5	BRAT1â€associated neurodegeneration: Intraâ€familial phenotypic differences in siblings. American Journal of Medical Genetics, Part A, 2016, 170, 3033-3038.	1.2	18
6	Reply. Annals of Neurology, 2016, 80, 168-169.	5.3	0
7	KCNT1mutations in seizure disorders: the phenotypic spectrum and functional effects. Journal of Medical Genetics, 2016, 53, 217-225.	3.2	94
8	Multiplex families with epilepsy. Neurology, 2016, 86, 713-722.	1.1	23
9	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
10	Familial neonatal seizures in 36 families: Clinical and genetic features correlate with outcome. Epilepsia, 2015, 56, 1071-1080.	5.1	94
11	Single Nucleotide Variations in CLCN6 Identified in Patients with Benign Partial Epilepsies in Infancy and/or Febrile Seizures. PLoS ONE, 2015, 10, e0118946.	2.5	13
12	Mutations in <scp>KCNT</scp>1 cause a spectrum of focal epilepsies. Epilepsia, 2015, 56, e114-20.	5.1	117
13	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. Nature Genetics, 2015, 47, 39-46.	21.4	245
14	KCNT1 gain of function in 2 epilepsy phenotypes is reversed by quinidine. Annals of Neurology, 2014, 75, 581-590.	5.3	249
15	Mutations in mammalian target of rapamycin regulator DEPDC5 cause focal epilepsy with brain malformations. Annals of Neurology, 2014, 75, 782-787.	5.3	193
16	Genetics of epilepsy. Neurology, 2014, 83, 1042-1048.	1.1	61
17	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
18	Mutations in DEPDC5 cause familial focal epilepsy with variable foci. Nature Genetics, 2013, 45, 546-551.	21.4	301

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19	Role ofPRRT2in common paroxysmal neurological disorders: a gene with remarkable pleiotropy. Journal of Medical Genetics, 2013, 50, 133-139.	3.2	88
20	Autosomal dominant vasovagal syncope. Neurology, 2013, 80, 1485-1493.	1.1	20
21	Mutations in <scp>PRRT</scp>2 are not a common cause of infantile epileptic encephalopathies. Epilepsia, 2013, 54, e86-9.	5.1	12
22	Benign Neonatal Sleep Myoclonus. Journal of Child Neurology, 2012, 27, 1260-1263.	1.4	10
23	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
24	Familial focal epilepsy with variable foci mapped to chromosome 22q12: Expansion of the phenotypic spectrum. Epilepsia, 2012, 53, e151-5.	5.1	24
25	PRRT2 phenotypic spectrum includes sporadic and fever-related infantile seizures. Neurology, 2012, 79, 2104-2108.	1.1	75
26	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
27	KCNQ2 encephalopathy: Emerging phenotype of a neonatal epileptic encephalopathy. Annals of Neurology, 2012, 71, 15-25.	5.3	427
28	The Role of Seizure-RelatedSEZ6as a Susceptibility Gene in Febrile Seizures. Neurology Research International, 2011, 2011, 1-4.	1.3	20
29	Proposed genetic classification of the â€œbenignâ€•familial neonatal and infantile epilepsies. Epilepsia, 2011, 52, 649-650.	5.1	9
30	â€œBlinders, phenotype, and fashionable genetic analysisâ€: Setting the record straight for epilepsy!. Epilepsia, 2011, 52, 1757-1758.	5.1	2
31	Neonatal seizures and long QT Syndrome: A cardiocerebral channelopathy?. Epilepsia, 2010, 51, 293-296.	5.1	61
32	Familial neonatal seizures with intellectual disability caused by a microduplication of chromosome 2q24.3. Epilepsia, 2010, 51, 1865-1869.	5.1	30
33	De novo SCN1A mutations in Dravet syndrome and related epileptic encephalopathies are largely of paternal origin. Journal of Medical Genetics, 2010, 47, 137-141.	3.2	44
34	Novel Mutation in KCNQ2 Causing Benign Familial Neonatal Seizures. Pediatric Neurology, 2009, 41, 367-370.	2.1	10
35	Severe autosomal dominant nocturnal frontal lobe epilepsy associated with psychiatric disorders and intellectual disability. Epilepsia, 2008, 49, 2125-2129.	5.1	49
36	Deletions or duplications in KCNQ2 can cause benign familial neonatal seizures. Journal of Medical Genetics, 2007, 44, 791-796.	3.2	70

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37	Generalized epilepsy with febrile seizures plusâ€“associated sodium channel Î²1 subunit mutations severely reduce beta subunitâ€“mediated modulation of sodium channel function. Neuroscience, 2007, 148, 164-174.	2.3	61
38	A childhood epilepsy mutation reveals a role for developmentally regulated splicing of a sodium channel. Molecular and Cellular Neurosciences, 2007, 35, 292-301.	2.2	68
39	Extended spectrum of idiopathic generalized epilepsies associated with CACNA1H functional variants. Annals of Neurology, 2007, 62, 560-568.	5.3	186
40	Channelopathies in idiopathic epilepsy. Neurotherapeutics, 2007, 4, 295-304.	4.4	101
41	Association studies and functional validation or functional validation alone?. Epilepsy Research, 2007, 74, 237-238.	1.6	3
42	SCN2A Mutations and Benign Familial Neonatal-Infantile Seizures: The Phenotypic Spectrum. Epilepsia, 2007, 48, 1138-1142.	5.1	102
43	A polygenic heterogeneity model for common epilepsies with complex genetics. Genes, Brain and Behavior, 2007, 6, 593-597.	2.2	52
44	Is Variation in the GABA(B) Receptor 1 Gene Associated with Temporal Lobe Epilepsy?. Epilepsia, 2005, 46, 778-780.	5.1	15
45	Novel mutations in the KCNQ2 gene link epilepsy to a dysfunction of the KCNQ2-calmodulin interaction. Journal of Medical Genetics, 2004, 41, 35e-35.	3.2	62
46	Generalized Epilepsy with Febrile Seizures Plus (GEFS+): Clinical Spectrum in Seven Italian Families Unrelated to SCN1A, SCN1B, and GABRG2 Gene Mutations. Epilepsia, 2004, 45, 149-158.	5.1	67
47	Febrile Convulsions and Genetic Susceptibility: Role of the Neuronal Nicotinic Acetylcholine Receptor alpha4 Subunit. Epilepsia, 2004, 45, 561-561.	5.1	10
48	Genetic variation ofCACNA1H in idiopathic generalized epilepsy. Annals of Neurology, 2004, 55, 595-596.	5.3	102
49	Benign familial neonatal-infantile seizures: Characterization of a new sodium channelopathy. Annals of Neurology, 2004, 55, 550-557.	5.3	250
50	Sodium-channel defects in benign familial neonatal-infantile seizures. Lancet, The, 2002, 360, 851-852.	13.7	332
51	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
52	Absence of Mutations Raises Doubts about the Role of the 70-kD Peroxisomal Membrane Protein in Zellweger Syndrome. American Journal of Human Genetics, 1997, 60, 1535-1539.	6.2	15
53	Absence of Mutations Raises Doubts about the Role of the 70â€kD Peroxisomal Membrane Protein in Zellweger Syndrome. American Journal of Human Genetics, 1997, 60, 1535-1538.	6.2	0