Leanne M Dibbens

List of Publications by Year
in descending order

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86 papers	9,299 citations	36303 51 h-index	54911 84 g-index
86 all docs	86 docs citations	86 times ranked	8034 citing authors

#	Article	IF	Citations
1	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.	6.2	41
2	KCNT1-related epilepsies and epileptic encephalopathies: phenotypic and mutational spectrum. Brain, 2021, 144, 3635-3650.	7.6	34
3	Mild malformations of cortical development in sleepâ€related hypermotor epilepsy due to KCNT1 mutations. Annals of Clinical and Translational Neurology, 2019, 6, 386-391.	3.7	25
4	Knockout of the epilepsy gene Depdc5 in mice causes severe embryonic dysmorphology with hyperactivity of mTORC1 signalling. Scientific Reports, 2017, 7, 12618.	3.3	39
5	Genetic epilepsy with febrile seizures plus. Neurology, 2017, 89, 1210-1219.	1.1	112
6	Novel ID gene CSNK2B : The crossover from molecular diagnosis to research continues. Human Mutation, 2017, 38, 1037-1037.	2.5	0
7	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
8	Benign infantile seizures and paroxysmal dyskinesia caused by an SCN8A mutation. Annals of Neurology, 2016, 79, 428-436.	5.3	159
9	SCARB2/LIMP2 deficiency in action myoclonus-renal failure syndrome. Epileptic Disorders, 2016, 18, 63-72.	1.3	26
10	GOSR2: a progressive myoclonus epilepsy gene. Epileptic Disorders, 2016, 18, 111-114.	1.3	32
11	BRAT1â€associated neurodegeneration: Intraâ€familial phenotypic differences in siblings. American Journal of Medical Genetics, Part A, 2016, 170, 3033-3038.	1.2	18
12	Reply. Annals of Neurology, 2016, 80, 168-169.	5.3	0
13	KCNT1mutations in seizure disorders: the phenotypic spectrum and functional effects. Journal of Medical Genetics, 2016, 53, 217-225.	3.2	94
14	Multiplex families with epilepsy. Neurology, 2016, 86, 713-722.	1.1	23
15	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
16	Epileptic spasms are a feature of DEPDC5 mTORopathy. Neurology: Genetics, 2015, 1, e17.	1.9	63
17	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
18	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

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19	Evaluation of multiple putative risk alleles within the 15q13.3 region for genetic generalized epilepsy. Epilepsy Research, 2015, 117, 70-73.	1.6	6
20	Mutations in <scp>KCNT</scp>1 cause a spectrum of focal epilepsies. Epilepsia, 2015, 56, e114-20.	5.1	117
21	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. Nature Genetics, 2015, 47, 39-46.	21.4	245
22	KCNT1 gain of function in 2 epilepsy phenotypes is reversed by quinidine. Annals of Neurology, 2014, 75, 581-590.	5.3	249
23	Atypical multifocal <scp>D</scp>ravet syndrome lacks generalized seizures and may show later cognitive decline. Developmental Medicine and Child Neurology, 2014, 56, 85-90.	2.1	16
24	Mutations in mammalian target of rapamycin regulator DEPDC5 cause focal epilepsy with brain malformations. Annals of Neurology, 2014, 75, 782-787.	5.3	193
25	Genetics of epilepsy. Neurology, 2014, 83, 1042-1048.	1.1	61
26	SCN1A variations and response to multiple antiepileptic drugs. Pharmacogenomics Journal, 2014, 14, 385-389.	2.0	20
27	GABRA1 and STXBP1 : Novel genetic causes of Dravet syndrome. Neurology, 2014, 82, 1245-1253.	1.1	229
28	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
29	Do mutations in SCN1B cause Dravet syndrome?. Epilepsy Research, 2013, 103, 97-100.	1.6	11
30	Role of the sodium channel <scp>SCN</scp>9A in genetic epilepsy with febrile seizures plus and Dravet syndrome. Epilepsia, 2013, 54, e122-6.	5.1	62
31	Abnormal Processing of Autophagosomes in Transformed B Lymphocytes from SCARB2-Deficient Subjects. BioResearch Open Access, 2013, 2, 40-46.	2.6	9
32	Copy number variants are frequent in genetic generalized epilepsy with intellectual disability. Neurology, 2013, 81, 1507-1514.	1.1	140
33	PRRT2 mutation in Japanese children with benign infantile epilepsy. Brain and Development, 2013, 35, 641-646.	1.1	31
34	Mutations in DEPDC5 cause familial focal epilepsy with variable foci. Nature Genetics, 2013, 45, 546-551.	21.4	301
35	Role ofPRRT2in common paroxysmal neurological disorders: a gene with remarkable pleiotropy. Journal of Medical Genetics, 2013, 50, 133-139.	3.2	88
36	â€˜North Seaâ€™ progressive myoclonus epilepsy: phenotype of subjects with GOSR2 mutation. Brain, 2013, 136, 1146-1154.	7.6	129

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37	Autosomal dominant vasovagal syncope. Neurology, 2013, 80, 1485-1493.	1.1	20
38	Mutations in <scp>PRRT</scp>2 are not a common cause of infantile epileptic encephalopathies. Epilepsia, 2013, 54, e86-9.	5.1	12
39	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
40	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
41	PRRT2 phenotypic spectrum includes sporadic and fever-related infantile seizures. Neurology, 2012, 79, 2104-2108.	1.1	75
42	Rare protein sequence variation in SV2A gene does not affect response to levetiracetam. Epilepsy Research, 2012, 101, 277-279.	1.6	11
43	A case of severe hearing loss in action myoclonus renal failure syndrome resulting from mutation in SCARB2. Movement Disorders, 2012, 27, 1200-1201.	3.9	17
44	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
45	Febrile infection-related epilepsy syndrome is not caused by SCN1A mutations. Epilepsy Research, 2012, 100, 194-198.	1.6	9
46	Genetic variations and associated pathophysiology in the management of epilepsy. The Application of Clinical Genetics, 2011, 4, 113.	3.0	4
47	The Role of Seizure-RelatedSEZ6as a Susceptibility Gene in Febrile Seizures. Neurology Research International, 2011, 2011, 1-4.	1.3	20
48	Proposed genetic classification of the â€œbenignâ€•familial neonatal and infantile epilepsies. Epilepsia, 2011, 52, 649-650.	5.1	9
49	â€œBlinders, phenotype, and fashionable genetic analysisâ€• Setting the record straight for epilepsy!. Epilepsia, 2011, 52, 1757-1758.	5.1	2
50	Investigation of the 15q13.3 CNV as a genetic modifier for familial epilepsies with variable phenotypes. Epilepsia, 2011, 52, e139-e142.	5.1	9
51	Clinical and neurophysiologic features of progressive myoclonus epilepsy without renal failure caused bySCARB2mutations. Epilepsia, 2011, 52, 2356-2363.	5.1	63
52	A Mutation in the Golgi Qb-SNARE Gene GOSR2 Causes Progressive Myoclonus Epilepsy with Early Ataxia. American Journal of Human Genetics, 2011, 88, 657-663.	6.2	166
53	Mutation of SCARB2 in a Patient With Progressive Myoclonus Epilepsy and Demyelinating Peripheral Neuropathy. Archives of Neurology, 2011, 68, 812-3.	4.5	28
54	A Focal Epilepsy and Intellectual Disability Syndrome Is Due to a Mutation in TBC1D24. American Journal of Human Genetics, 2010, 87, 371-375.	6.2	111

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55	Effects of vaccination on onset and outcome of Dravet syndrome: a retrospective study. Lancet Neurology, The, 2010, 9, 592-598.	10.2	119
56	Augmented currents of an HCN2 variant in patients with febrile seizure syndromes. Annals of Neurology, 2010, 67, 542-546.	5.3	96
57	Genetics of the epilepsies: Genetic twists in the channels and other tales. Epilepsia, 2010, 51, 33-36.	5.1	8
58	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
59	Epilepsy and mental retardation limited to females with PCDH19 mutations can present de novo or in single generation families. Journal of Medical Genetics, 2010, 47, 211-216.	3.2	74
60	Detection of microchromosomal aberrations in refractory epilepsy: a pilot study. Epileptic Disorders, 2010, 12, 192-198.	1.3	14
61	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
62	Dravet syndrome or genetic (generalized) epilepsy with febrile seizures plus?. Brain and Development, 2009, 31, 394-400.	1.1	152
63	SCARB2 mutations in progressive myoclonus epilepsy (PME) without renal failure. Annals of Neurology, 2009, 66, 532-536.	5.3	90
64	SCN1A duplications and deletions detected in Dravet syndrome: Implications for molecular diagnosis. Epilepsia, 2009, 50, 1670-1678.	5.1	152
65	Does a SCN1A gene mutation confer earlier age of onset of febrile seizures in GEFS+?. Epilepsia, 2009, 50, 953-956.	5.1	22
66	Chipping away at the common epilepsies with complex genetics: the 15q13.3 microdeletion shows the way. Genome Medicine, 2009, 1, 33.	8.2	17
67	The role of neuronal GABAA receptor subunit mutations in idiopathic generalized epilepsies. Neuroscience Letters, 2009, 453, 162-165.	2.1	37
68	Array-Based Gene Discovery with Three Unrelated Subjects Shows SCARB2/LIMP-2 Deficiency Causes Myoclonus Epilepsy and Glomerulosclerosis. American Journal of Human Genetics, 2008, 82, 673-684.	6.2	230
69	X-linked protocadherin 19 mutations cause female-limited epilepsy and cognitive impairment. Nature Genetics, 2008, 40, 776-781.	21.4	397
70	Gene expression analysis in absence epilepsy using a monozygotic twin design. Epilepsia, 2008, 49, 1546-1554.	5.1	24
71	Epilepsy and mental retardation limited to females: an under-recognized disorder. Brain, 2008, 131, 918-927.	7.6	172
72	Reduced cortical inhibition in a mouse model of familial childhood absence epilepsy. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 17536-17541.	7.1	192

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73	The spectrum of SCN1A-related infantile epileptic encephalopathies. Brain, 2007, 130, 843-852.	7.6	501
74	Channelopathies in idiopathic epilepsy. Neurotherapeutics, 2007, 4, 295-304.	4.4	101
75	Is Photosensitive Epilepsy Less Common in Males Due to Variation in X Chromosome Photopigment Genes?. Epilepsia, 2007, 48, 1807-1809.	5.1	10
76	NEDD4-2as a potential candidate susceptibility gene for epileptic photosensitivity. Genes, Brain and Behavior, 2007, 6, 750-755.	2.2	56
77	A polygenic heterogeneity model for common epilepsies with complex genetics. Genes, Brain and Behavior, 2007, 6, 593-597.	2.2	52
78	Â Subunit Susceptibility Variants E177A and R220H Associated with Complex Epilepsy Alter Channel Gating and Surface Expression of Â4beta2Â GABAA Receptors. Journal of Neuroscience, 2006, 26, 1499-1506.	3.6	81
79	Temporal lobe epilepsy and GEFS+ phenotypes associated with SCN1B mutations. Brain, 2006, 130, 100-109.	7.6	234
80	Neonatal Epilepsy Syndromes and Generalized Epilepsy with Febrile Seizures Plus (GEFS+). Epilepsia, 2005, 46, 41-47.	5.1	63
81	SCN1Amutations and epilepsy. Human Mutation, 2005, 25, 535-542.	2.5	327
82	Susceptibility genes for complex epilepsy. Human Molecular Genetics, 2005, 14, R243-R249.	2.9	92
83	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
84	Genetic Architecture of Idiopathic Generalized Epilepsy: Clinical Genetic Analysis of 55 Multiplex Families. Epilepsia, 2004, 45, 467-478.	5.1	128
85	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
86	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