Lynette G Sadleir

List of Publications by Year
in descending order

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91	7,196	87888 38	62596 80
papers	citations	h-index	g-index

96	96	96	8761
all docs	docs citations	times ranked	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.	21.4	589
2	The spectrum of SCN1A-related infantile epileptic encephalopathies. Brain, 2007, 130, 843-852.	7.6	501
3	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	6.2	388
4	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
5	GRIN2A mutations cause epilepsy-aphasia spectrum disorders. Nature Genetics, 2013, 45, 1073-1076.	21.4	326
6	De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies. American Journal of Human Genetics, 2016, 99, 287-298.	6.2	247
7	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
8	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
9	GABRA1 and STXBP1 : Novel genetic causes of Dravet syndrome. Neurology, 2014, 82, 1245-1253.	1.1	229
10	Rare copy number variants are an important cause of epileptic encephalopathies. Annals of Neurology, 2011, 70, 974-985.	5.3	222
11	Mortality in Dravet syndrome. Epilepsy Research, 2016, 128, 43-47.	1.6	218
12	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
13	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
14	Ultra-rare genetic variation in common epilepsies: a case-control sequencing study. Lancet Neurology, The, 2017, 16, 135-143.	10.2	190
15	GRIN2Bencephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. Journal of Medical Genetics, 2017, 54, 460-470.	3.2	190
16	ILAE definition of the Idiopathic Generalized Epilepsy Syndromes: Position statement by the ILAE Task Force on Nosology and Definitions. Epilepsia, 2022, 63, 1475-1499.	5.1	148
17	Febrile seizures. BMJ: British Medical Journal, 2007, 334, 307-311.	2.3	118
18	Vigabatrin with hormonal treatment versus hormonal treatment alone (ICISS) for infantile spasms: 18-month outcomes of an open-label, randomised controlled trial. The Lancet Child and Adolescent Health, 2018, 2, 715-725.	5.6	114

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19	CHD2variants are a risk factor for photosensitivity in epilepsy. Brain, 2015, 138, 1198-1208.	7.6	112
20	Heart rate variability in epilepsy: A potential biomarker of <scp>sudden unexpected death in epilepsy</scp> risk. Epilepsia, 2018, 59, 1372-1380.	5.1	105
21	Parental Mosaicism in â€œDe Novoâ€•Epileptic Encephalopathies. New England Journal of Medicine, 2018, 378, 1646-1648.	27.0	104
22	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	12.8	99
23	Early onset absence epilepsy: 1 in 10 cases is caused by GLUT1 deficiency. Epilepsia, 2012, 53, e204-7.	5.1	97
24	Not all SCN1A epileptic encephalopathies are Dravet syndrome. Neurology, 2017, 89, 1035-1042.	1.1	97
25	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. Annals of Neurology, 2019, 86, 821-831.	5.3	96
26	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. Human Molecular Genetics, 2015, 24, 5250-5259.	2.9	93
27	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. American Journal of Human Genetics, 2018, 103, 666-678.	6.2	87
28	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.	7.9	86
29	EEG features of absence seizures in idiopathic generalized epilepsy: Impact of syndrome, age, and state. Epilepsia, 2009, 50, 1572-1578.	5.1	81
30	PRRT2 phenotypic spectrum includes sporadic and fever-related infantile seizures. Neurology, 2012, 79, 2104-2108.	1.1	75
31	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. EBioMedicine, 2015, 2, 1063-1070.	6.1	74
32	Autism and developmental disability caused by KCNQ3 gainâ€ofâ€function variants. Annals of Neurology, 2019, 86, 181-192.	5.3	73
33	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
34	Mutations of the Sonic Hedgehog Pathway Underlie Hypothalamic Hamartoma with Gelastic Epilepsy. American Journal of Human Genetics, 2016, 99, 423-429.	6.2	59
35	Clinical genetic studies in benign childhood epilepsy with centrotemporal spikes. Epilepsia, 2012, 53, 319-324.	5.1	49
36	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 103, 305-316.	6.2	48

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37	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	7.6	47
38	Clinical genetic study of the epilepsyâ€aphasia spectrum. Epilepsia, 2013, 54, 280-287.	5.1	44
39	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. American Journal of Human Genetics, 2017, 101, 516-524.	6.2	43
40	The Epilepsy Phenome/Genome Project. Clinical Trials, 2013, 10, 568-586.	1.6	40
41	Juvenile Alpers Disease. Archives of Neurology, 2008, 65, 121-4.	4.5	39
42	Factors influencing clinical features of absence seizures. Epilepsia, 2008, 49, 2100-2107.	5.1	36
43	Familial Adult Myoclonic Epilepsy. Archives of Neurology, 2012, 69, 474.	4.5	36
44	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.	6.2	35
45	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.	2.5	33
46	Family studies of individuals with eyelid myoclonia with absences. Epilepsia, 2012, 53, 2141-2148.	5.1	32
47	New insights into the clinical and molecular spectrum of the novel CYFIP2-related neurodevelopmental disorder and impairment of the WRC-mediated actin dynamics. Genetics in Medicine, 2021, 23, 543-554.	2.4	32
48	Efficacy of cannabinoids in paediatric epilepsy. Developmental Medicine and Child Neurology, 2019, 61, 13-18.	2.1	30
49	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	2.4	28
50	Acute Flaccid Paralysis from Echovirus Type 33 Infection. Journal of Clinical Microbiology, 2003, 41, 2230-2232.	3.9	26
51	Acquired brachialâ€plexus neuropathy in the neonate: a rare presentation of lateâ€onset groupâ€B streptococcal osteomyelitis. Developmental Medicine and Child Neurology, 1998, 40, 496-499.	2.1	26
52	Automatisms in Absence Seizures in Children With Idiopathic Generalized Epilepsy. Archives of Neurology, 2009, 66, 729-34.	4.5	26
53	Damaging de novo missense variants inEEF1A2lead to a developmental and degenerative epilepticâ€dyskinetic encephalopathy. Human Mutation, 2020, 41, 1263-1279.	2.5	24
54	Schizophrenia is a lateâ€onset feature of <scp>PCDH</scp>19 Girls Clustering Epilepsy. Epilepsia, 2019, 60, 429-440.	5.1	23

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55	A standardized patient-centered characterization of the phenotypic spectrum of PCDH19 girls clustering epilepsy. Translational Psychiatry, 2020, 10, 127.	4.8	22
56	Evaluation of nonâ€coding variation in <scp>GLUT</scp>1 deficiency. Developmental Medicine and Child Neurology, 2016, 58, 1295-1302.	2.1	20
57	A mutation in COL4A2 causes autosomal dominant porencephaly with cataracts. American Journal of Medical Genetics, Part A, 2016, 170, 1059-1063.	1.2	17
58	Developmental and epilepsy spectrum of KCNB1 encephalopathy with longâ€term outcome. Epilepsia, 2020, 61, 2461-2473.	5.1	17
59	Electroclinical features of absence seizures in sleep. Epilepsy Research, 2011, 93, 216-220.	1.6	16
60	Levetiracetam efficacy in PCDH19 Girls Clustering Epilepsy. European Journal of Paediatric Neurology, 2020, 24, 142-147.	1.6	16
61	Are Variants Causing Cardiac Arrhythmia Risk Factors in Sudden Unexpected Death in Epilepsy?. Frontiers in Neurology, 2020, 11, 925.	2.4	16
62	SUDEP risk and autonomic dysfunction in genetic epilepsies. Autonomic Neuroscience: Basic and Clinical, 2022, 237, 102907.	2.8	16
63	Spasms in children with definite and probable mitochondrial disease*. European Journal of Neurology, 2004, 11, 103-110.	3.3	15
64	Seizure semiology in autosomal dominant epilepsy with auditory features, due to novel LGI1 mutations. Epilepsy Research, 2013, 107, 311-317.	1.6	15
65	PCDH19 Pathogenic Variants in Males: Expanding the Phenotypic Spectrum. Advances in Experimental Medicine and Biology, 2020, 1298, 177-187.	1.6	15
66	Safety and Tolerability of Transdermal Cannabidiol Gel in Children With Developmental and Epileptic Encephalopathies. JAMA Network Open, 2021, 4, e2123930.	5.9	15
67	Inherited RORB pathogenic variants: Overlap of photosensitive genetic generalized and occipital lobe epilepsy. Epilepsia, 2020, 61, e23-e29.	5.1	14
68	The epileptology of GNB5 encephalopathy. Epilepsia, 2019, 60, e121-e127.	5.1	13
69	An international pilot study of an internetâ€based platform to facilitate clinical research in epilepsy: The EpiNet project. Epilepsia, 2012, 53, 1829-1835.	5.1	12
70	Diverse genetic causes of polymicrogyria with epilepsy. Epilepsia, 2021, 62, 973-983.	5.1	12
71	Developmental and epileptic encephalopathy: Personal utility of a genetic diagnosis for families. Epilepsia Open, 2021, 6, 149-159.	2.4	11
72	The phenotypic spectrum of Xâ€linked, infantile onset ALG13â€related developmental and epileptic encephalopathy. Epilepsia, 2021, 62, 325-334.	5.1	10

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73	Common risk variants for epilepsy are enriched in families previously targeted for rare monogenic variant discovery. EBioMedicine, 2022, 81, 104079.	6.1	10
74	Contribution of rare genetic variants to drug response in absence epilepsy. Epilepsy Research, 2021, 170, 106537.	1.6	9
75	Optimizing Electroencephalographic Studies for Epilepsy Diagnosis in Children With New-Onset Seizures. Archives of Neurology, 2010, 67, 1345-9.	4.5	8
76	Sporadic hypothalamic hamartoma is a ciliopathy with somatic and bi-allelic contributions. Human Molecular Genetics, 2022, 31, 2307-2316.	2.9	8
77	Association of ultraâ€rare coding variants with genetic generalized epilepsy: A caseâ€“control whole exome sequencing study. Epilepsia, 2022, 63, 723-735.	5.1	8
78	Using the Internet to recruit patients for epilepsy trials: Results of a New Zealand pilot study. Epilepsia, 2010, 51, 868-873.	5.1	7
79	Myoclonic occipital photosensitive epilepsy with dystonia (MOPED): A familial epilepsy syndrome. Epilepsy Research, 2015, 114, 98-105.	1.6	7
80	Double somatic mosaicism in a child with Dravet syndrome. Neurology: Genetics, 2019, 5, e333.	1.9	7
81	Fragile Females: Case Series of Epilepsy in Girls With FMR1 Disruption. Pediatrics, 2019, 144, .	2.1	5
82	Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. Epilepsia, 2021, 62, 1518-1527.	5.1	5
83	Infantileâ€onset myoclonic developmental and epileptic encephalopathy: A new RARS2 phenotype. Epilepsia Open, 2022, 7, 170-180.	2.4	5
84	EpiNet as a way of involving more physicians and patients in epilepsy research: Validation study and accreditation process. Epilepsia Open, 2017, 2, 20-31.	2.4	4
85	PIGNencephalopathy: Characterizing the epileptology. Epilepsia, 2022, 63, 974-991.	5.1	4
86	Do neurologists around the world agree when diagnosing epilepsy? â€“ Results of an international EpiNet study. Epilepsy Research, 2018, 139, 43-50.	1.6	3
87	Epidemiology of Treated Epilepsy in New Zealand Children. Neurology, 2021, 97, e1933-e1941.	1.1	3
88	Generation of the induced human pluripotent stem cell lines CSSi009-A from a patient with a GNB5 pathogenic variant, and CSSi010-A from a CRISPR/Cas9 engineered GNB5 knock-out human cell line. Stem Cell Research, 2019, 40, 101547.	0.7	2
89	The â€œmaternal effectâ€•on epilepsy risk: Analysis of familial epilepsies and reassessment of prior evidence. Annals of Neurology, 2020, 87, 132-138.	5.3	2
90	Response to â€œAsymmetric dimethylarginine and vascular risk in patients treated with antiepileptic drugsâ€• Epilepsy Research, 2014, 108, 1962.	1.6	0

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91	Quantitative analysis of phenotypic elements augments traditional electroclinical classification of common familial epilepsies. Epilepsia, 2019, 60, 2194-2203.	5.1	0