

Lynette G Sadleir

List of Publications by Year in descending order

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Version: 2024-02-01

91
papers

7,196
citations

87723

38
h-index

62479

80
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96
all docs

96
docs citations

96
times ranked

8761
citing authors

#	ARTICLE	IF	CITATIONS
1	Infantile-onset myoclonic developmental and epileptic encephalopathy: A new <i>RARS2</i> phenotype. <i>Epilepsia Open</i> , 2022, 7, 170-180.	1.3	5
2	SUDEP risk and autonomic dysfunction in genetic epilepsies. <i>Autonomic Neuroscience: Basic and Clinical</i> , 2022, 237, 102907.	1.4	16
3	Sporadic hypothalamic hamartoma is a ciliopathy with somatic and bi-allelic contributions. <i>Human Molecular Genetics</i> , 2022, 31, 2307-2316.	1.4	8
4	Association of ultra-rare coding variants with genetic generalized epilepsy: A case-control whole exome sequencing study. <i>Epilepsia</i> , 2022, 63, 723-735.	2.6	8
5	<i>PIGN</i> encephalopathy: Characterizing the epileptology. <i>Epilepsia</i> , 2022, 63, 974-991.	2.6	4
6	ILAE definition of the Idiopathic Generalized Epilepsy Syndromes: Position statement by the ILAE Task Force on Nosology and Definitions. <i>Epilepsia</i> , 2022, 63, 1475-1499.	2.6	148
7	Common risk variants for epilepsy are enriched in families previously targeted for rare monogenic variant discovery. <i>EBioMedicine</i> , 2022, 81, 104079.	2.7	10
8	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. <i>Genetics in Medicine</i> , 2021, 23, 363-373.	1.1	28
9	New insights into the clinical and molecular spectrum of the novel <i>CYFIP2</i> -related neurodevelopmental disorder and impairment of the WRC-mediated actin dynamics. <i>Genetics in Medicine</i> , 2021, 23, 543-554.	1.1	32
10	Developmental and epileptic encephalopathy: Personal utility of a genetic diagnosis for families. <i>Epilepsia Open</i> , 2021, 6, 149-159.	1.3	11
11	Contribution of rare genetic variants to drug response in absence epilepsy. <i>Epilepsy Research</i> , 2021, 170, 106537.	0.8	9
12	Diverse genetic causes of polymicrogyria with epilepsy. <i>Epilepsia</i> , 2021, 62, 973-983.	2.6	12
13	Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. <i>Epilepsia</i> , 2021, 62, 1518-1527.	2.6	5
14	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. <i>American Journal of Human Genetics</i> , 2021, 108, 965-982.	2.6	35
15	Epidemiology of Treated Epilepsy in New Zealand Children. <i>Neurology</i> , 2021, 97, e1933-e1941.	1.5	3
16	Safety and Tolerability of Transdermal Cannabidiol Gel in Children With Developmental and Epileptic Encephalopathies. <i>JAMA Network Open</i> , 2021, 4, e2123930.	2.8	15
17	The phenotypic spectrum of X-linked, infantile onset <i>ALG13</i> -related developmental and epileptic encephalopathy. <i>Epilepsia</i> , 2021, 62, 325-334.	2.6	10
18	Expanding the genetic and phenotypic relevance of <i>KCNB1</i> variants in developmental and epileptic encephalopathies: 27 new patients and overview of the literature. <i>Human Mutation</i> , 2020, 41, 69-80.	1.1	33

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19	The "maternal effect" on epilepsy risk: Analysis of familial epilepsies and reassessment of prior evidence. <i>Annals of Neurology</i> , 2020, 87, 132-138.	2.8	2
20	Levetiracetam efficacy in PCDH19 Girls Clustering Epilepsy. <i>European Journal of Paediatric Neurology</i> , 2020, 24, 142-147.	0.7	16
21	Are Variants Causing Cardiac Arrhythmia Risk Factors in Sudden Unexpected Death in Epilepsy?. <i>Frontiers in Neurology</i> , 2020, 11, 925.	1.1	16
22	Developmental and epilepsy spectrum of <i>KCNB1</i> encephalopathy with long-term outcome. <i>Epilepsia</i> , 2020, 61, 2461-2473.	2.6	17
23	PCDH19 Pathogenic Variants in Males: Expanding the Phenotypic Spectrum. <i>Advances in Experimental Medicine and Biology</i> , 2020, 1298, 177-187.	0.8	15
24	A standardized patient-centered characterization of the phenotypic spectrum of PCDH19 girls clustering epilepsy. <i>Translational Psychiatry</i> , 2020, 10, 127.	2.4	22
25	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17,458 subjects. <i>Brain</i> , 2020, 143, 2106-2118.	3.7	47
26	Damaging de novo missense variants in <i>EEF1A2</i> lead to a developmental and degenerative epileptic dyskinetic encephalopathy. <i>Human Mutation</i> , 2020, 41, 1263-1279.	1.1	24
27	Inherited <i>RORB</i> pathogenic variants: Overlap of photosensitive genetic generalized and occipital lobe epilepsy. <i>Epilepsia</i> , 2020, 61, e23-e29.	2.6	14
28	A systematic review and meta-analysis of 271 PCDH19-variant individuals identifies psychiatric comorbidities, and association of seizure onset and disease severity. <i>Molecular Psychiatry</i> , 2019, 24, 241-251.	4.1	86
29	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. <i>American Journal of Human Genetics</i> , 2019, 105, 267-282.	2.6	237
30	Quantitative analysis of phenotypic elements augments traditional electroclinical classification of common familial epilepsies. <i>Epilepsia</i> , 2019, 60, 2194-2203.	2.6	0
31	Intronic ATTC repeat expansions in <i>STARD7</i> in familial adult myoclonic epilepsy linked to chromosome 2. <i>Nature Communications</i> , 2019, 10, 4920.	5.8	99
32	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. <i>Annals of Neurology</i> , 2019, 86, 821-831.	2.8	96
33	The epileptology of <i>GNB5</i> encephalopathy. <i>Epilepsia</i> , 2019, 60, e121-e127.	2.6	13
34	Generation of the induced human pluripotent stem cell lines CSSi009-A from a patient with a <i>GNB5</i> pathogenic variant, and CSSi010-A from a CRISPR/Cas9 engineered <i>GNB5</i> knock-out human cell line. <i>Stem Cell Research</i> , 2019, 40, 101547.	0.3	2
35	Autism and developmental disability caused by <i>KCNQ3</i> gain-of-function variants. <i>Annals of Neurology</i> , 2019, 86, 181-192.	2.8	73
36	Double somatic mosaicism in a child with Dravet syndrome. <i>Neurology: Genetics</i> , 2019, 5, e333.	0.9	7

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37	Schizophrenia is a later-onset feature of <i>PCDH19</i> Girls Clustering Epilepsy. <i>Epilepsia</i> , 2019, 60, 429-440.	2.6	23
38	Fragile Females: Case Series of Epilepsy in Girls With <i>FMR1</i> Disruption. <i>Pediatrics</i> , 2019, 144, .	1.0	5
39	Efficacy of cannabinoids in paediatric epilepsy. <i>Developmental Medicine and Child Neurology</i> , 2019, 61, 13-18.	1.1	30
40	Parental Mosaicism in De Novo Epileptic Encephalopathies. <i>New England Journal of Medicine</i> , 2018, 378, 1646-1648.	13.9	104
41	Do neurologists around the world agree when diagnosing epilepsy? Results of an international EpiNet study. <i>Epilepsy Research</i> , 2018, 139, 43-50.	0.8	3
42	Vigabatrin with hormonal treatment versus hormonal treatment alone (ICISS) for infantile spasms: 18-month outcomes of an open-label, randomised controlled trial. <i>The Lancet Child and Adolescent Health</i> , 2018, 2, 715-725.	2.7	114
43	De Novo Pathogenic Variants in <i>CACNA1E</i> Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. <i>American Journal of Human Genetics</i> , 2018, 103, 666-678.	2.6	87
44	De Novo Variants in the F-Box Protein <i>FBXO11</i> in 20 Individuals with a Variable Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2018, 103, 305-316.	2.6	48
45	Heart rate variability in epilepsy: A potential biomarker of sudden unexpected death in epilepsy risk. <i>Epilepsia</i> , 2018, 59, 1372-1380.	2.6	105
46	Ultra-rare genetic variation in common epilepsies: a case-control sequencing study. <i>Lancet Neurology</i> , The, 2017, 16, 135-143.	4.9	190
47	<i>GRIN2B</i> encephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. <i>Journal of Medical Genetics</i> , 2017, 54, 460-470.	1.5	190
48	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2017, 101, 664-685.	2.6	337
49	De Novo Mutations in <i>PPP3CA</i> Cause Severe Neurodevelopmental Disease with Seizures. <i>American Journal of Human Genetics</i> , 2017, 101, 516-524.	2.6	43
50	EpiNet as a way of involving more physicians and patients in epilepsy research: Validation study and accreditation process. <i>Epilepsia Open</i> , 2017, 2, 20-31.	1.3	4
51	Not all <i>SCN1A</i> epileptic encephalopathies are Dravet syndrome. <i>Neurology</i> , 2017, 89, 1035-1042.	1.5	97
52	Exome-based analysis of cardiac arrhythmia, respiratory control, and epilepsy genes in sudden unexpected death in epilepsy. <i>Annals of Neurology</i> , 2016, 79, 522-534.	2.8	216
53	Evaluation of non-coding variation in <i>GLUT1</i> deficiency. <i>Developmental Medicine and Child Neurology</i> , 2016, 58, 1295-1302.	1.1	20
54	De Novo Mutations in <i>SLC1A2</i> and <i>CACNA1A</i> Are Important Causes of Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2016, 99, 287-298.	2.6	247

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55	Mutations of the Sonic Hedgehog Pathway Underlie Hypothalamic Hamartoma with Gelastic Epilepsy. American Journal of Human Genetics, 2016, 99, 423-429.	2.6	59
56	Mortality in Dravet syndrome. Epilepsy Research, 2016, 128, 43-47.	0.8	218
57	A mutation in <i>COL4A2</i> causes autosomal dominant porencephaly with cataracts. American Journal of Medical Genetics, Part A, 2016, 170, 1059-1063.	0.7	17
58	Mutations in the mammalian target of rapamycin pathway regulators <i>NPRL2</i> and <i>NPRL3</i> cause focal epilepsy. Annals of Neurology, 2016, 79, 120-131.	2.8	190
59	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. EBioMedicine, 2015, 2, 1063-1070.	2.7	74
60	<i>CHD2</i> variants are a risk factor for photosensitivity in epilepsy. Brain, 2015, 138, 1198-1208.	3.7	112
61	Myoclonic occipital photosensitive epilepsy with dystonia (MOPED): A familial epilepsy syndrome. Epilepsy Research, 2015, 114, 98-105.	0.8	7
62	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. Human Molecular Genetics, 2015, 24, 5250-5259.	1.4	93
63	Response to asymmetric dimethylarginine and vascular risk in patients treated with antiepileptic drugs. Epilepsy Research, 2014, 108, 1962.	0.8	0
64	De Novo Mutations in Synaptic Transmission Genes Including DNMT1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	2.6	388
65	<i>GABRA1</i> and <i>STXBP1</i> : Novel genetic causes of Dravet syndrome. Neurology, 2014, 82, 1245-1253.	1.5	229
66	GRIN2A mutations cause epilepsy-aphasia spectrum disorders. Nature Genetics, 2013, 45, 1073-1076.	9.4	326
67	Role of the sodium channel <i>SCN9A</i> in genetic epilepsy with febrile seizures plus and Dravet syndrome. Epilepsia, 2013, 54, e122-6.	2.6	62
68	Seizure semiology in autosomal dominant epilepsy with auditory features, due to novel LGI1 mutations. Epilepsy Research, 2013, 107, 311-317.	0.8	15
69	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. Nature Genetics, 2013, 45, 825-830.	9.4	589
70	Clinical genetic study of the epilepsy-aphasia spectrum. Epilepsia, 2013, 54, 280-287.	2.6	44
71	The Epilepsy Phenome/Genome Project. Clinical Trials, 2013, 10, 568-586.	0.7	40
72	Familial Adult Myoclonic Epilepsy. Archives of Neurology, 2012, 69, 474.	4.9	36

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73	An international pilot study of an internet-based platform to facilitate clinical research in epilepsy: The EpiNet project. <i>Epilepsia</i> , 2012, 53, 1829-1835.	2.6	12
74	Early onset absence epilepsy: 1 in 10 cases is caused by GLUT1 deficiency. <i>Epilepsia</i> , 2012, 53, e204-7.	2.6	97
75	<i>PRRT2</i> phenotypic spectrum includes sporadic and fever-related infantile seizures. <i>Neurology</i> , 2012, 79, 2104-2108.	1.5	75
76	Family studies of individuals with eyelid myoclonia with absences. <i>Epilepsia</i> , 2012, 53, 2141-2148.	2.6	32
77	<i>PRRT2</i> Mutations Cause Benign Familial Infantile Epilepsy and Infantile Convulsions with Choreoathetosis Syndrome. <i>American Journal of Human Genetics</i> , 2012, 90, 152-160.	2.6	234
78	Clinical genetic studies in benign childhood epilepsy with centrotemporal spikes. <i>Epilepsia</i> , 2012, 53, 319-324.	2.6	49
79	Rare copy number variants are an important cause of epileptic encephalopathies. <i>Annals of Neurology</i> , 2011, 70, 974-985.	2.8	222
80	Electroclinical features of absence seizures in sleep. <i>Epilepsy Research</i> , 2011, 93, 216-220.	0.8	16
81	Optimizing Electroencephalographic Studies for Epilepsy Diagnosis in Children With New-Onset Seizures. <i>Archives of Neurology</i> , 2010, 67, 1345-9.	4.9	8
82	Using the Internet to recruit patients for epilepsy trials: Results of a New Zealand pilot study. <i>Epilepsia</i> , 2010, 51, 868-873.	2.6	7
83	Automatisms in Absence Seizures in Children With Idiopathic Generalized Epilepsy. <i>Archives of Neurology</i> , 2009, 66, 729-34.	4.9	26
84	EEG features of absence seizures in idiopathic generalized epilepsy: Impact of syndrome, age, and state. <i>Epilepsia</i> , 2009, 50, 1572-1578.	2.6	81
85	Factors influencing clinical features of absence seizures. <i>Epilepsia</i> , 2008, 49, 2100-2107.	2.6	36
86	Juvenile Alpers Disease. <i>Archives of Neurology</i> , 2008, 65, 121-4.	4.9	39
87	The spectrum of <i>SCN1A</i> -related infantile epileptic encephalopathies. <i>Brain</i> , 2007, 130, 843-852.	3.7	501
88	Febrile seizures. <i>BMJ: British Medical Journal</i> , 2007, 334, 307-311.	2.4	118
89	Spasms in children with definite and probable mitochondrial disease*. <i>European Journal of Neurology</i> , 2004, 11, 103-110.	1.7	15
90	Acute Flaccid Paralysis from Echovirus Type 33 Infection. <i>Journal of Clinical Microbiology</i> , 2003, 41, 2230-2232.	1.8	26

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91	Acquired brachial plexus neuropathy in the neonate: a rare presentation of late-onset group B streptococcal osteomyelitis. <i>Developmental Medicine and Child Neurology</i> , 1998, 40, 496-499.	1.1	26