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	Infantileâ€onset myoclonic developmental and epileptic encephalopathy: A new RARS2 phenotype. Epilepsia Open, 2022, 7, 170-180.	2.4	5
2	SUDEP risk and autonomic dysfunction in genetic epilepsies. Autonomic Neuroscience: Basic and Clinical, 2022, 237, 102907.	2.8	16
3	Sporadic hypothalamic hamartoma is a ciliopathy with somatic and bi-allelic contributions. Human Molecular Genetics, 2022, 31, 2307-2316.	2.9	8
4	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
5	PIGNencephalopathy: Characterizing the epileptology. Epilepsia, 2022, 63, 974-991.	5.1	4
6	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
7	Common risk variants for epilepsy are enriched in families previously targeted for rare monogenic variant discovery. EBioMedicine, 2022, 81, 104079.	6.1	10
8	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	2.4	28
9	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
10	Developmental and epileptic encephalopathy: Personal utility of a genetic diagnosis for families. Epilepsia Open, 2021, 6, 149-159.	2.4	11
11	Contribution of rare genetic variants to drug response in absence epilepsy. Epilepsy Research, 2021, 170, 106537.	1.6	9
12	Diverse genetic causes of polymicrogyria with epilepsy. Epilepsia, 2021, 62, 973-983.	5.1	12
13	Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. Epilepsia, 2021, 62, 1518-1527.	5.1	5
14	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
15	Epidemiology of Treated Epilepsy in New Zealand Children. Neurology, 2021, 97, e1933-e1941.	1.1	3
16	Safety and Tolerability of Transdermal Cannabidiol Gel in Children With Developmental and Epileptic Encephalopathies. JAMA Network Open, 2021, 4, e2123930.	5.9	15
17	The phenotypic spectrum of Xâ€linked, infantile onset ALG13â€related developmental and epileptic encephalopathy. Epilepsia, 2021, 62, 325-334.	5.1	10
18	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

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19	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
20	Levetiracetam efficacy in PCDH19 Girls Clustering Epilepsy. European Journal of Paediatric Neurology, 2020, 24, 142-147.	1.6	16
21	Are Variants Causing Cardiac Arrhythmia Risk Factors in Sudden Unexpected Death in Epilepsy?. Frontiers in Neurology, 2020, 11, 925.	2.4	16
22	Developmental and epilepsy spectrum of KCNB1 encephalopathy with longâ€term outcome. Epilepsia, 2020, 61, 2461-2473.	5.1	17
23	PCDH19 Pathogenic Variants in Males: Expanding the Phenotypic Spectrum. Advances in Experimental Medicine and Biology, 2020, 1298, 177-187.	1.6	15
24	A standardized patient-centered characterization of the phenotypic spectrum of PCDH19 girls clustering epilepsy. Translational Psychiatry, 2020, 10, 127.	4.8	22
25	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	7.6	47
26	Damaging de novo missense variants inEEF1A2lead to a developmental and degenerative epilepticâ€dyskinetic encephalopathy. Human Mutation, 2020, 41, 1263-1279.	2.5	24
27	Inherited RORB pathogenic variants: Overlap of photosensitive genetic generalized and occipital lobe epilepsy. Epilepsia, 2020, 61, e23-e29.	5.1	14
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	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
30	Quantitative analysis of phenotypic elements augments traditional electroclinical classification of common familial epilepsies. Epilepsia, 2019, 60, 2194-2203.	5.1	0
31	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	12.8	99
32	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. Annals of Neurology, 2019, 86, 821-831.	5.3	96
33	The epileptology of GNB5 encephalopathy. Epilepsia, 2019, 60, e121-e127.	5.1	13
34	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
35	Autism and developmental disability caused by KCNQ3 gainâ€ofâ€function variants. Annals of Neurology, 2019, 86, 181-192.	5.3	73
36	Double somatic mosaicism in a child with Dravet syndrome. Neurology: Genetics, 2019, 5, e333.	1.9	7

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37	Schizophrenia is a laterâ€onset feature of <scp>PCDH</scp>19 Girls Clustering Epilepsy. Epilepsia, 2019, 60, 429-440.	5.1	23
38	Fragile Females: Case Series of Epilepsy in Girls With FMR1 Disruption. Pediatrics, 2019, 144, .	2.1	5
39	Efficacy of cannabinoids in paediatric epilepsy. Developmental Medicine and Child Neurology, 2019, 61, 13-18.	2.1	30
40	Parental Mosaicism in â€œDe Novoâ€•Epileptic Encephalopathies. New England Journal of Medicine, 2018, 378, 1646-1648.	27.0	104
41	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
42	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
43	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
44	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
45	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
46	Ultra-rare genetic variation in common epilepsies: a case-control sequencing study. Lancet Neurology, The, 2017, 16, 135-143.	10.2	190
47	GRIN2Bencephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. Journal of Medical Genetics, 2017, 54, 460-470.	3.2	190
48	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
49	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. American Journal of Human Genetics, 2017, 101, 516-524.	6.2	43
50	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
51	Not all SCN1A epileptic encephalopathies are Dravet syndrome. Neurology, 2017, 89, 1035-1042.	1.1	97
52	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
53	Evaluation of nonâ€coding variation in <scp>GLUT</scp>1 deficiency. Developmental Medicine and Child Neurology, 2016, 58, 1295-1302.	2.1	20
54	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

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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.	6.2	59
56	Mortality in Dravet syndrome. Epilepsy Research, 2016, 128, 43-47.	1.6	218
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	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
59	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. EBioMedicine, 2015, 2, 1063-1070.	6.1	74
60	CHD2variants are a risk factor for photosensitivity in epilepsy. Brain, 2015, 138, 1198-1208.	7.6	112
61	Myoclonic occipital photosensitive epilepsy with dystonia (MOPED): A familial epilepsy syndrome. Epilepsy Research, 2015, 114, 98-105.	1.6	7
62	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. Human Molecular Genetics, 2015, 24, 5250-5259.	2.9	93
63	Response to â€œAsymmetric dimethylarginine and vascular risk in patients treated with antiepileptic drugsâ€• Epilepsy Research, 2014, 108, 1962.	1.6	0
64	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	6.2	388
65	GABRA1 and STXBP1 : Novel genetic causes of Dravet syndrome. Neurology, 2014, 82, 1245-1253.	1.1	229
66	GRIN2A mutations cause epilepsy-aphasia spectrum disorders. Nature Genetics, 2013, 45, 1073-1076.	21.4	326
67	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
68	Seizure semiology in autosomal dominant epilepsy with auditory features, due to novel LGI1 mutations. Epilepsy Research, 2013, 107, 311-317.	1.6	15
69	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. Nature Genetics, 2013, 45, 825-830.	21.4	589
70	Clinical genetic study of the epilepsyâ€aphasia spectrum. Epilepsia, 2013, 54, 280-287.	5.1	44
71	The Epilepsy Phenome/Genome Project. Clinical Trials, 2013, 10, 568-586.	1.6	40
72	Familial Adult Myoclonic Epilepsy. Archives of Neurology, 2012, 69, 474.	4.5	36

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73	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
74	Early onset absence epilepsy: 1 in 10 cases is caused by GLUT1 deficiency. Epilepsia, 2012, 53, e204-7.	5.1	97
75	PRRT2 phenotypic spectrum includes sporadic and fever-related infantile seizures. Neurology, 2012, 79, 2104-2108.	1.1	75
76	Family studies of individuals with eyelid myoclonia with absences. Epilepsia, 2012, 53, 2141-2148.	5.1	32
77	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
78	Clinical genetic studies in benign childhood epilepsy with centrotemporal spikes. Epilepsia, 2012, 53, 319-324.	5.1	49
79	Rare copy number variants are an important cause of epileptic encephalopathies. Annals of Neurology, 2011, 70, 974-985.	5.3	222
80	Electroclinical features of absence seizures in sleep. Epilepsy Research, 2011, 93, 216-220.	1.6	16
81	Optimizing Electroencephalographic Studies for Epilepsy Diagnosis in Children With New-Onset Seizures. Archives of Neurology, 2010, 67, 1345-9.	4.5	8
82	Using the Internet to recruit patients for epilepsy trials: Results of a New Zealand pilot study. Epilepsia, 2010, 51, 868-873.	5.1	7
83	Automatisms in Absence Seizures in Children With Idiopathic Generalized Epilepsy. Archives of Neurology, 2009, 66, 729-34.	4.5	26
84	EEG features of absence seizures in idiopathic generalized epilepsy: Impact of syndrome, age, and state. Epilepsia, 2009, 50, 1572-1578.	5.1	81
85	Factors influencing clinical features of absence seizures. Epilepsia, 2008, 49, 2100-2107.	5.1	36
86	Juvenile Alpers Disease. Archives of Neurology, 2008, 65, 121-4.	4.5	39
87	The spectrum of SCN1A-related infantile epileptic encephalopathies. Brain, 2007, 130, 843-852.	7.6	501
88	Febrile seizures. BMJ: British Medical Journal, 2007, 334, 307-311.	2.3	118
89	Spasms in children with definite and probable mitochondrial disease*. European Journal of Neurology, 2004, 11, 103-110.	3.3	15
90	Acute Flaccid Paralysis from Echovirus Type 33 Infection. Journal of Clinical Microbiology, 2003, 41, 2230-2232.	3.9	26

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91	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