Dennis Lal

List of Publications by Year
in descending order

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93 papers	4,454 citations	116194 36 h-index	156644 58 g-index
95 all docs	95 docs citations	95 times ranked	6920 citing authors

#	Article	IF	Citations
1	Development and Validation of a Prediction Model for Early Diagnosis of SCN1A -Related Epilepsies. Neurology, 2022, 98, .	1.5	24
2	Incidence and prevalence of major epilepsy-associated brain lesions. Epilepsy and Behavior Reports, 2022, 18, 100527.	0.5	2
3	Structural mapping of GABRB3 variants reveals genotypeâ€“phenotype correlations. Genetics in Medicine, 2022, 24, 681-693.	1.1	10
4	Analysing an allelic series of rare missense variants of CACNA1I in a Swedish schizophrenia cohort. Brain, 2022, 145, 1839-1853.	3.7	18
5	DDGun: an untrained predictor of protein stability changes upon amino acid variants. Nucleic Acids Research, 2022, 50, W222-W227.	6.5	28
6	The gain of function SCN1A disorder spectrum: novel epilepsy phenotypes and therapeutic implications. Brain, 2022, 145, 3816-3831.	3.7	43
7	The <scp>ILAE</scp> consensus classification of focal cortical dysplasia: An update proposed by an ad hoc task force of the <scp>ILAE</scp> diagnostic methods commission. Epilepsia, 2022, 63, 1899-1919.	2.6	88
8	The role of common genetic variation in presumed monogenic epilepsies. EBioMedicine, 2022, 81, 104098.	2.7	12
9	Genomeâ€Wide Analysis of Copy Number Variation in Latin American Parkinson's Disease Patients. Movement Disorders, 2021, 36, 434-441.	2.2	12
10	Clinical sequencing yield in epilepsy, autism spectrum disorder, and intellectual disability: A systematic review and metaâ€analysis. Epilepsia, 2021, 62, 143-151.	2.6	77
11	Characterization of the <scp>GABRB2</scp>â€Associated Neurodevelopmental Disorders. Annals of Neurology, 2021, 89, 573-586.	2.8	14
12	Regulation of purine metabolism connects KCTD13 to a metabolic disorder with autistic features. IScience, 2021, 24, 101935.	1.9	7
13	ATP1A3-Related Disorders: An Ever-Expanding Clinical Spectrum. Frontiers in Neurology, 2021, 12, 637890.	1.1	28
14	Toward a better definition of focal cortical dysplasia: An iterative histopathological and genetic agreement trial. Epilepsia, 2021, 62, 1416-1428.	2.6	54
15	Common molecular mechanisms ofSLC6A1variant-mediated neurodevelopmental disorders in astrocytes and neurons. Brain, 2021, 144, 2499-2512.	3.7	35
16	Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. Epilepsia, 2021, 62, 1518-1527.	2.6	5
17	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.	2.6	35
18	Application of single cell genomics to focal epilepsies: A call to action. Brain Pathology, 2021, 31, e12958.	2.1	8

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19	Neocortical development and epilepsy: insights from focal cortical dysplasia and brain tumours. Lancet Neurology, The, 2021, 20, 943-955.	4.9	47
20	Using common genetic variants to find drugs for common epilepsies. Brain Communications, 2021, 3, fcab287.	1.5	9
21	Identification and quantification of oligogenic loss-of-function disorders. Genetics in Medicine, 2021, , .	1.1	1
22	Copy number variants in lipid metabolism genes are associated with gallstones disease in men. European Journal of Human Genetics, 2020, 28, 264-273.	1.4	6
23	Differential excitatory vs inhibitory SCN expression at single cell level regulates brain sodium channel function in neurodevelopmental disorders. European Journal of Paediatric Neurology, 2020, 24, 129-133.	0.7	18
24	Time to move beyond genetics towards biomedical data-driven translational genomic research in severe paediatric epilepsies. European Journal of Paediatric Neurology, 2020, 24, 35-39.	0.7	2
25	SCN1A variants from bench to bedsideâ€"improved clinical prediction from functional characterization. Human Mutation, 2020, 41, 363-374.	1.1	37
26	Identification of pathogenic variant enriched regions across genes and gene families. Genome Research, 2020, 30, 62-71.	2.4	47
27	Neurological disorder-associated genetic variants in individuals with psychogenic nonepileptic seizures. Scientific Reports, 2020, 10, 15205.	1.6	12
28	Current knowledge of SLC6A1-related neurodevelopmental disorders. Brain Communications, 2020, 2, fcaa170.	1.5	44
29	Predicting functional effects of missense variants in voltage-gated sodium and calcium channels. Science Translational Medicine, 2020, 12, .	5.8	84
30	Comprehensive characterization of amino acid positions in protein structures reveals molecular effect of missense variants. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 28201-28211.	3.3	68
31	Polygenic risk heterogeneity among focal epilepsies. Epilepsia, 2020, 61, e179-e185.	2.6	3
32	MISCAST: MIssense variant to protein StruCture Analysis web SuiTe. Nucleic Acids Research, 2020, 48, W132-W139.	6.5	14
33	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	2.6	32
34	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	3.7	47
35	An Altered Relationship between Soluble TREM2 and Inflammatory Markers in Young Adults with Down Syndrome: A Preliminary Report. Journal of Immunology, 2020, 204, 1111-1118.	0.4	17
36	Sodium channel epilepsies and neurodevelopmental disorders: from disease mechanisms to clinical application. Developmental Medicine and Child Neurology, 2020, 62, 784-792.	1.1	23

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37	A catalogue of new incidence estimates of monogenic neurodevelopmental disorders caused by de novo variants. Brain, 2020, 143, 1099-1105.	3.7	64
38	Paternal-Age-Related de Novo Mutations and Risk for Five Disorders. Obstetrical and Gynecological Survey, 2020, 75, 104-105.	0.2	0
39	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. Epilepsia, 2020, 61, 387-399.	2.6	65
40	Vasospasm Following Hemispherectomy: A Case Report of a Novel Complication. World Neurosurgery, 2020, 137, 357-361.	0.7	1
41	Copy Number Variation and Clinical Outcomes in Patients With GermlinePTENMutations. JAMA Network Open, 2020, 3, e1920415.	2.8	19
42	Pleiotropy of polygenic factors associated with focal and generalized epilepsy in the general population. PLoS ONE, 2020, 15, e0232292.	1.1	14
43	The role of sodium channels in sudden unexpected death in pediatrics. Molecular Genetics & Genomic Medicine, 2020, 8, e1309.	0.6	14
44	Title is missing!. , 2020, 15, e0232292.		0
45	Title is missing!. , 2020, 15, e0232292.		0
46	Title is missing!. , 2020, 15, e0232292.		0
47	Title is missing!. , 2020, 15, e0232292.		0
48	Rare De Novo Missense Variants in RNA Helicase DDX6 Cause Intellectual Disability and Dysmorphic Features and Lead to P-Body Defects and RNA Dysregulation. American Journal of Human Genetics, 2019, 105, 509-525.	2.6	50
49	CDKL5 deficiency disorder: Relationship between genotype, epilepsy, cortical visual impairment, and development. Epilepsia, 2019, 60, 1733-1742.	2.6	97
50	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.	2.6	237
51	Assessment of genetic variant burden in epilepsy-associated brain lesions. European Journal of Human Genetics, 2019, 27, 1738-1744.	1.4	12
52	Polygenic burden in focal and generalized epilepsies. Brain, 2019, 142, 3473-3481.	3.7	90
53	Spectrum of neurodevelopmental disease associated with the GNAO1 guanosine triphosphateâ€“binding region. Epilepsia, 2019, 60, 406-418.	2.6	53
54	Contribution of rare and common variants to intellectual disability in a sub-isolate of Northern Finland. Nature Communications, 2019, 10, 410.	5.8	32

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55	Simple ClinVar: an interactive web server to explore and retrieve gene and disease variants aggregated in ClinVar database. Nucleic Acids Research, 2019, 47, W99-W105.	6.5	51
56	Targeted gene sequencing in 6994 individuals with neurodevelopmental disorder with epilepsy. Genetics in Medicine, 2019, 21, 2496-2503.	1.1	45
57	Variant Score Rankerâ€"a web application for intuitive missense variant prioritization. Bioinformatics, 2019, 35, 4478-4479.	1.8	5
58	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	2.6	61
59	The spectrum of intermediate <scp>SCN</scp>8Aâ€related epilepsy. Epilepsia, 2019, 60, 830-844.	2.6	70
60	Cyclin-Dependent Kinase-Like 5 Deficiency Disorder: Clinical Review. Pediatric Neurology, 2019, 97, 18-25.	1.0	145
61	Functional Interpretation of Single Amino Acid Substitutions in 1,330 Disease-Associated Genes. Biophysical Journal, 2019, 116, 420a-421a.	0.2	0
62	Spectrum of GABAA receptor variants in epilepsy. Current Opinion in Neurology, 2019, 32, 183-190.	1.8	59
63	Duplications at 19q13.33 in patients with neurodevelopmental disorders. Neurology: Genetics, 2018, 4, e210.	0.9	4
64	The contribution of CACNA1A, ATP1A2 and SCN1A mutations in hemiplegic migraine: A clinical and genetic study in Finnish migraine families. Cephalalgia, 2018, 38, 1849-1863.	1.8	38
65	Progress in Understanding and Treating SCN2A-Mediated Disorders. Trends in Neurosciences, 2018, 41, 442-456.	4.2	210
66	Exome-wide analysis of mutational burden in patients with typical and atypical Rolandic epilepsy. European Journal of Human Genetics, 2018, 26, 258-264.	1.4	22
67	Common Variant Burden Contributes to the Familial Aggregation of Migraine in 1,589 Families. Neuron, 2018, 98, 743-753.e4.	3.8	63
68	Guidelineâ€based and bioinformatic reassessment of lesionâ€associated gene and variant pathogenicity in focal human epilepsies. Epilepsia, 2018, 59, 2145-2152.	2.6	8
69	Rare gene deletions in genetic generalized and Rolandic epilepsies. PLoS ONE, 2018, 13, e0202022.	1.1	6
70	De novo variants in neurodevelopmental disorders with epilepsy. Nature Genetics, 2018, 50, 1048-1053.	9.4	230
71	Genetics Sheds New Light on Congenital Hydrocephalus Biology. Neuron, 2018, 99, 246-247.	3.8	2
72	Rare coding variants in genes encoding GABAA receptors in genetic generalised epilepsies: an exome-based case-control study. Lancet Neurology, The, 2018, 17, 699-708.	4.9	67

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73	KANSL1 variation is not a major contributing factor in self-limited focal epilepsy syndromes of childhood. PLoS ONE, 2018, 13, e0191546.	1.1	3
74	X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in AIFM1. Neurogenetics, 2017, 18, 185-194.	0.7	38
75	Heterogeneous contribution of microdeletions in the development of common generalised and focal epilepsies. Journal of Medical Genetics, 2017, 54, 598-606.	1.5	22
76	Idiopathic focal epilepsies: the â€œlost tribeâ€• Epileptic Disorders, 2016, 18, 252-288.	0.7	65
77	Increased Probability of Co-Occurrence of Two Rare Diseases in Consanguineous Families and Resolution of a Complex Phenotype by Next Generation Sequencing. PLoS ONE, 2016, 11, e0146040.	1.1	32
78	Evaluation of Presumably Disease Causing SCN1A Variants in a Cohort of Common Epilepsy Syndromes. PLoS ONE, 2016, 11, e0150426.	1.1	22
79	Simultaneous impairment of neuronal and metabolic function of mutated gephyrin in a patient with epileptic encephalopathy. EMBO Molecular Medicine, 2015, 7, 1580-1594.	3.3	39
80	Extending the phenotypic spectrum of <scp>RBFOX</scp>1 deletions: Sporadic focal epilepsy. Epilepsia, 2015, 56, e129-33.	2.6	38
81	Rare variants in Î³â€aminobutyric acid type <scp>A</scp> receptor genes in rolandic epilepsy and related syndromes. Annals of Neurology, 2015, 77, 972-986.	2.8	51
82	Investigation of GRIN2A in common epilepsy phenotypes. Epilepsy Research, 2015, 115, 95-99.	0.8	44
83	Burden Analysis of Rare Microdeletions Suggests a Strong Impact of Neurodevelopmental Genes in Genetic Generalised Epilepsies. PLoS Genetics, 2015, 11, e1005226.	1.5	91
84	Recessive mutations inSLC13A5result in a loss of citrate transport and cause neonatal epilepsy, developmental delay and teeth hypoplasia. Brain, 2015, 138, 3238-3250.	3.7	96
85	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. Human Molecular Genetics, 2014, 23, 6069-6080.	1.4	61
86	DEPDC5 mutations in genetic focal epilepsies of childhood. Annals of Neurology, 2014, 75, 788-792.	2.8	105
87	Analysis of ELP4, SRPX2, and interacting genes in typical and atypical rolandic epilepsy. Epilepsia, 2014, 55, e89-93.	2.6	50
88	Exonic microdeletions of the gephyrin gene impair GABAergic synaptic inhibition in patients with idiopathic generalized epilepsy. Neurobiology of Disease, 2014, 67, 88-96.	2.1	51
89	Mutations in GRIN2A cause idiopathic focal epilepsy with rolandic spikes. Nature Genetics, 2013, 45, 1067-1072.	9.4	391
90	Homozygous missense mutation of NDUFV1 as the cause of infantile bilateral striatal necrosis. Neurogenetics, 2013, 14, 85-87.	0.7	140

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91	Rare exonic deletions of the <scp>RBFOX1</scp> gene increase risk of idiopathic generalized epilepsy. Epilepsia, 2013, 54, 265-271.	2.6	59
92	RBFOX1 and RBFOX3 Mutations in Rolandic Epilepsy. PLoS ONE, 2013, 8, e73323.	1.1	94
93	ATP11C is critical for the internalization of phosphatidylserine and differentiation of B lymphocytes. Nature Immunology, 2011, 12, 441-449.	7.0	117