Johannes R Lemke

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/4519180/publications.pdf

Version: 2024-02-01

85 papers	5,852 citations	109311 35 h-index	85537 71 g-index
100 all docs	100 docs citations	100 times ranked	7214 citing authors

#	Article	IF	Citations
1	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Brain, 2022, 145, 2991-3009.	7.6	69
2	Exome first approach to reduce diagnostic costs and time â€“ retrospective analysis of 111 individuals with rare neurodevelopmental disorders. European Journal of Human Genetics, 2022, 30, 117-125.	2.8	22
3	Gain-of-function variants in GABRD reveal a novel pathway for neurodevelopmental disorders and epilepsy. Brain, 2022, 145, 1299-1309.	7.6	34
4	L-Serine Treatment is Associated with Improvements in Behavior, EEG, and Seizure Frequency in Individuals with GRIN-Related Disorders Due to Null Variants. Neurotherapeutics, 2022, 19, 334-341.	4.4	21
5	Complementing the phenotypical spectrum of TUBA1A tubulinopathy and its role in early-onset epilepsies. European Journal of Human Genetics, 2022, 30, 298-306.	2.8	9
6	Improving one-step scarless genome editing in Drosophila melanogaster by combining ovoD co-CRISPR selection with sgRNA target site masking. Biology Methods and Protocols, 2022, 7, bpac003.	2.2	1
7	Bi-allelic SMO variants in hypothalamic hamartoma: a recessive cause of Pallister-Hall syndrome. European Journal of Human Genetics, 2022, 30, 384-388.	2.8	6
8	Genomic basis for skin phenotype and cold adaptation in the extinct Stellerâ€™s sea cow. Science Advances, 2022, 8, eabl6496.	10.3	9
9	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	2.4	20
10	Refining Genotypes and Phenotypes in KCNA2-Related Neurological Disorders. International Journal of Molecular Sciences, 2021, 22, 2824.	4.1	20
11	The genetic landscape of intellectual disability and epilepsy in adults and the elderly: a systematic genetic work-up of 150 individuals. Genetics in Medicine, 2021, 23, 1492-1497.	2.4	31
12	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
13	Clinical and therapeutic significance of genetic variation in the GRIN gene family encoding NMDARs. Neuropharmacology, 2021, 199, 108805.	4.1	25
14	The Angelman Syndrome Online Registry â€“ A multilingual approach to support global research. European Journal of Medical Genetics, 2021, 64, 104349.	1.3	1
15	In cis TP53 and RAD51C pathogenic variants may predispose to sebaceous gland carcinomas. European Journal of Human Genetics, 2021, 29, 489-494.	2.8	0
16	Genotype-phenotype correlation on 45 individuals with West syndrome. European Journal of Paediatric Neurology, 2020, 25, 134-138.	1.6	23
17	Predicting functional effects of missense variants in voltage-gated sodium and calcium channels. Science Translational Medicine, 2020, 12, .	12.4	84
18	The Phenotypic Spectrum of PRRT2-Associated Paroxysmal Neurologic Disorders in Childhood. Biomedicines, 2020, 8, 456.	3.2	23

#	Article	IF	Citations
19	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	7.6	47
20	Predicting incidences of neurodevelopmental disorders. Brain, 2020, 143, 1046-1048.	7.6	9
21	The landscape of epilepsy-related GATOR1 variants. Genetics in Medicine, 2019, 21, 398-408.	2.4	137
22	Investigating the effects of additional truncating variants in DNA-repair genes on breast cancer risk in BRCA1-positive women. BMC Cancer, 2019, 19, 787.	2.6	10
23	De novoGRINvariants in NMDA receptor M2 channel poreâ€forming loop are associated with neurological diseases. Human Mutation, 2019, 40, 2393-2413.	2.5	48
24	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
25	Pathogenic WDFY3 variants cause neurodevelopmental disorders and opposing effects on brain size. Brain, 2019, 142, 2617-2630.	7.6	31
26	Novel synonymous and missense variants in FGFR1 causing Hartsfield syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2447-2453.	1.2	10
27	Biallelic inherited SCN8A variants, a rare cause of SCN8A â€related developmental and epileptic encephalopathy. Epilepsia, 2019, 60, 2277-2285.	5.1	18
28	Diagnostik genetisch bedingter Epilepsien. Medizinische Genetik, 2019, 31, 303-312.	0.2	1
29	Heterogeneous clinical and functional features of GRIN2D-related developmental and epileptic encephalopathy. Brain, 2019, 142, 3009-3027.	7.6	49
30	De novo variants in PAK1 lead to intellectual disability with macrocephaly and seizures. Brain, 2019, 142, 3351-3359.	7.6	29
31	From next-generation sequencing to targeted treatment of non-acquired epilepsies. Expert Review of Molecular Diagnostics, 2019, 19, 217-228.	3.1	38
32	A Recurrent Missense Variant in AP2M1 Impairs Clathrin-Mediated Endocytosis and Causes Developmental and Epileptic Encephalopathy. American Journal of Human Genetics, 2019, 104, 1060-1072.	6.2	78
33	Targeted gene sequencing in 6994 individuals with neurodevelopmental disorder with epilepsy. Genetics in Medicine, 2019, 21, 2496-2503.	2.4	45
34	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	5.1	61
35	Defining and expanding the phenotype of QARS-associated developmental epileptic encephalopathy. Neurology: Genetics, 2019, 5, e373.	1.9	5
36	Identification of a Loss-of-Function Mutation in the Context of Glutaminase Deficiency and Neonatal Epileptic Encephalopathy. JAMA Neurology, 2019, 76, 342.	9.0	33

#	Article	IF	Citations
37	GRIN2A-related disorders: genotype and functional consequence predict phenotype. Brain, 2019, 142, 80-92.	7.6	143
38	De Novo Variants in MAPK8IP3 Cause Intellectual Disability with Variable Brain Anomalies. American Journal of Human Genetics, 2019, 104, 203-212.	6.2	44
39	A new p.(Ile66Serfs*93) IGF2 variant is associated with pre- and postnatal growth retardation. European Journal of Endocrinology, 2019, 180, K1-K13.	3.7	16
40	Clinical and genetic spectrum of AMPD2-related pontocerebellar hypoplasia type 9. European Journal of Human Genetics, 2018, 26, 695-708.	2.8	22
41	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. Annals of Neurology, 2018, 84, 788-795.	5.3	44
42	NfL is a biomarker for adult-onset leukoencephalopathy with axonal spheroids and pigmented glia. Neurology, 2018, 91, 755-757.	1.1	11
43	De novo variants in neurodevelopmental disorders with epilepsy. Nature Genetics, 2018, 50, 1048-1053.	21.4	230
44	Haploinsufficiency of CUX1 Causes Nonsyndromic Global Developmental Delay With Possible Catchâ€up Development. Annals of Neurology, 2018, 84, 200-207.	5.3	23
45	De novo gainâ€ofâ€function variants in KCNT2 as a novel cause of developmental and epileptic encephalopathy. Annals of Neurology, 2018, 83, 1198-1204.	5.3	41
46	Relationship of electrophysiological dysfunction and clinical severity inSCN2A-related epilepsies. Human Mutation, 2018, 39, 1942-1956.	2.5	29
47	Novel homozygous missense variant of GRIN1 in two sibs with intellectual disability and autistic features without epilepsy. European Journal of Human Genetics, 2017, 25, 376-380.	2.8	30
48	Variants in CPLX1 in two families with autosomal-recessive severe infantile myoclonic epilepsy and ID. European Journal of Human Genetics, 2017, 25, 889-893.	2.8	30
49	Vitamin B6â€“Responsive Epilepsy due to a Novel KCNQ2 Mutation. Neuropediatrics, 2017, 48, 199-204.	0.6	12
50	Copy number variations in â€œclassicalâ€•obesity candidate genes are not frequently associated with severe early-onset obesity in children. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 507-515.	0.9	0
51	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. American Journal of Human Genetics, 2017, 100, 676-688.	6.2	54
52	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. Brain, 2017, 140, 1316-1336.	7.6	426
53	GRIN2Bencephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. Journal of Medical Genetics, 2017, 54, 460-470.	3.2	190
54	Mutations in GABRB3. Neurology, 2017, 88, 483-492.	1.1	87

#	Article	IF	Citations
55	Multigene panel next generation sequencing in a patient with cherry red macular spot: Identification of two novel mutations in NEU1 gene causing sialidosis type I associated with mild to unspecific biochemical and enzymatic findings. Molecular Genetics and Metabolism Reports, 2017, 10, 1-4.	1.1	9
56	Delineating SPTAN1 associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. Brain, 2017, 140, 2322-2336.	7.6	82
57	High-Throughput Sequencing as First-Tier Diagnostics in Congenital and Early-Onset Disorders. JAMA Pediatrics, 2017, 171, 833.	6.2	8
58	Reply. Annals of Neurology, 2017, 81, 328-329.	5.3	0
59	Clinical spectrum and genotypeâ€“phenotype associations of KCNA2-related encephalopathies. Brain, 2017, 140, 2337-2354.	7.6	117
60	Neurodevelopmental Disorders Caused by De Novo Variants in KCNB1 Genotypes and Phenotypes. JAMA Neurology, 2017, 74, 1228.	9.0	79
61	De Novo Variants in GRIA4 Lead to Intellectual Disability with or without Seizures and Gait Abnormalities. American Journal of Human Genetics, 2017, 101, 1013-1020.	6.2	53
62	Commentary: GATOR Complexâ€Associated Epilepsies. Epilepsia, 2017, 58, 1121-1122.	5.1	0
63	De novo GABRG2mutations associated with epileptic encephalopathies. Brain, 2017, 140, 49-67.	7.6	80
64	Benign infantile seizures and paroxysmal dyskinesia caused by an SCN8A mutation. Annals of Neurology, 2016, 79, 428-436.	5.3	159
65	Delineating the GRIN1 phenotypic spectrum. Neurology, 2016, 86, 2171-2178.	1.1	157
66	GRIN2D Recurrent De Novo Dominant Mutation Causes a Severe Epileptic Encephalopathy Treatable with NMDA Receptor Channel Blockers. American Journal of Human Genetics, 2016, 99, 802-816.	6.2	138
67	Tentative clinical diagnosis of Lujanâ€Fryns syndromeâ€”A conglomeration of different genetic entities?. American Journal of Medical Genetics, Part A, 2016, 170, 94-102.	1.2	11
68	Phenotypic spectrum of GABRA1. Neurology, 2016, 87, 1140-1151.	1.1	113
69	A recurrent mutation in KCNA2 as a novel cause of hereditary spastic paraplegia and ataxia. Annals of Neurology, 2016, 80, .	5.3	49
70	Novel KCNQ3 Mutation in a Large Family with Benign Familial Neonatal Epilepsy: A Rare Cause of Neonatal Seizures. Molecular Syndromology, 2016, 7, 189-196.	0.8	19
71	Phenotypic Variability from Benign Infantile Epilepsy to Ohtahara Syndrome Associated with a Novel Mutation in SCN2A. Molecular Syndromology, 2016, 7, 182-188.	0.8	31
72	Reply. Annals of Neurology, 2016, 80, 168-169.	5.3	0

#	Article	IF	Citations
73	STXBP1 encephalopathy. Neurology, 2016, 86, 954-962.	1.1	264
74	Evaluation of Presumably Disease Causing SCN1A Variants in a Cohort of Common Epilepsy Syndromes. PLoS ONE, 2016, 11, e0150426.	2.5	22
75	Investigation of GRIN2A in common epilepsy phenotypes. Epilepsy Research, 2015, 115, 95-99.	1.6	44
76	Galanin pathogenic mutations in temporal lobe epilepsy. Human Molecular Genetics, 2015, 24, 3082-3091.	2.9	23
77	De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy. Nature Genetics, 2015, 47, 393-399.	21.4	224
78	Epileptic Encephalopathies in Childhood: The Role of Genetic Testing. Seminars in Neurology, 2015, 35, 310-322.	1.4	7
79	Monogenic Human Skin Disorders. Dermatology, 2014, 229, 55-64.	2.1	27
80	DEPDC5 mutations in genetic focal epilepsies of childhood. Annals of Neurology, 2014, 75, 788-792.	5.3	105
81	GRIN2B mutations in west syndrome and intellectual disability with focal epilepsy. Annals of Neurology, 2014, 75, 147-154.	5.3	195
82	Mutations in STX1B, encoding a presynaptic protein, cause fever-associated epilepsy syndromes. Nature Genetics, 2014, 46, 1327-1332.	21.4	178
83	Mutations in GRIN2A cause idiopathic focal epilepsy with rolandic spikes. Nature Genetics, 2013, 45, 1067-1072.	21.4	391
84	Extending the KCNQ2 encephalopathy spectrum. Neurology, 2013, 81, 1697-1703.	1.1	198
85	Targeted next generation sequencing as a diagnostic tool in epileptic disorders. Epilepsia, 2012, 53, 1387-1398.	5.1	299