Louis J PtÃ¡Äek

List of Publications by Year
in descending order

123 papers	10,262 citations	50276 46 h-index	34986 98 g-index
154 all docs	154 docs citations	154 times ranked	9900 citing authors

#	Article	IF	Citations
1	Mutations in Kir2.1 Cause the Developmental and Episodic Electrical Phenotypes of Andersen's Syndrome. Cell, 2001, 105, 511-519.	28.9	921
2	Functional consequences of a CKIÎ´ mutation causing familial advanced sleep phase syndrome. Nature, 2005, 434, 640-644.	27.8	773
3	Functional and clinical characterization of KCNJ2 mutations associated with LQT7 (Andersen) Tj ETQq1 1 0.784314 rgBT /Overlock 10	8.2	457
4	Identification of a mutation in the gene causing hyperkalemic periodic paralysis. Cell, 1991, 67, 1021-1027.	28.9	405
5	Lamin B1 duplications cause autosomal dominant leukodystrophy. Nature Genetics, 2006, 38, 1114-1123.	21.4	365
6	De novo mutations in ATP1A3 cause alternating hemiplegia of childhood. Nature Genetics, 2012, 44, 1030-1034.	21.4	345
7	Mutations in a Gene Encoding a Novel Protein Containing a Phosphotyrosine-Binding Domain Cause Type 2 Cerebral Cavernous Malformations. American Journal of Human Genetics, 2003, 73, 1459-1464.	6.2	319
8	MiRP2 Forms Potassium Channels in Skeletal Muscle with Kv3.4 and Is Associated with Periodic Paralysis. Cell, 2001, 104, 217-231.	28.9	283
9	Mutations in an S4 segment of the adult skeletal muscle sodium channel cause paramyotonia congenita. Neuron, 1992, 8, 891-897.	8.1	252
10	Electrocardiographic Features in Andersen-Tawil Syndrome Patients With KCNJ2 Mutations. Circulation, 2005, 111, 2720-2726.	1.6	248
11	Mutations in Potassium Channel Kir2.6 Cause Susceptibility to Thyrotoxic Hypokalemic Periodic Paralysis. Cell, 2010, 140, 88-98.	28.9	245
12	Polyglutamine-Expanded Ataxin-7 Antagonizes CRX Function and Induces Cone-Rod Dystrophy in a Mouse Model of SCA7. Neuron, 2001, 31, 913-927.	8.1	244
13	Guidelines for Genome-Scale Analysis of Biological Rhythms. Journal of Biological Rhythms, 2017, 32, 380-393.	2.6	237
14	Glucose Sensor O-GlcNAcylation Coordinates with Phosphorylation to Regulate Circadian Clock. Cell Metabolism, 2013, 17, 291-302.	16.2	206
15	The gene for paroxysmal non-kinesigenic dyskinesia encodes an enzyme in a stress response pathway. Human Molecular Genetics, 2004, 13, 3161-3170.	2.9	196
16	A Novel Gene Causing a Mendelian Audiogenic Mouse Epilepsy. Neuron, 2001, 31, 537-544.	8.1	172
17	Randomized trials of dichlorphenamide in the periodic paralyses. Annals of Neurology, 2000, 47, 46-53.	5.3	156
18	Genetics and Physiology of the Myotonic Muscle Disorders. New England Journal of Medicine, 1993, 328, 482-489.	27.0	154

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19	Polyglutamine-Expanded Ataxin-7 Promotes Non-Cell-Autonomous Purkinje Cell Degeneration and Displays Proteolytic Cleavage in Ataxic Transgenic Mice. Journal of Neuroscience, 2002, 22, 4897-4905.	3.6	149
20	Defective Potassium Channel Kir2.1 Trafficking Underlies Andersen-Tawil Syndrome. Journal of Biological Chemistry, 2003, 278, 51779-51785.	3.4	147
21	The intricate dance of post-translational modifications in the rhythm of life. Nature Structural and Molecular Biology, 2016, 23, 1053-1060.	8.2	147
22	Two new genes from the human ATP-binding cassette transporter superfamily, ABCC11 and ABCC12, tandemly duplicated on chromosome 16q12. Gene, 2001, 273, 89-96.	2.2	143
23	Retinal degeneration characterizes a spinocerebellar ataxia mapping to chromosome 3p. Nature Genetics, 1995, 10, 89-93.	21.4	136
24	A PERIOD3 variant causes a circadian phenotype and is associated with a seasonal mood trait. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E1536-44.	7.1	134
25	Channelopathies: ion channel disorders of muscle as a paradigm for paroxysmal disorders of the nervous system. Neuromuscular Disorders, 1997, 7, 250-255.	0.6	126
26	Sodium channel mutations in paramyotonia congenita and hyperkalemic periodic paralysis. Annals of Neurology, 2004, 33, 300-307.	5.3	118
27	A Cryptochrome 2 mutation yields advanced sleep phase in humans. ELife, 2016, 5, .	6.0	114
28	The familial periodic paralyses and nondystrophic myotonias. American Journal of Medicine, 1998, 105, 58-70.	1.5	113
29	MicroRNA-23a promotes myelination in the central nervous system. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 17468-17473.	7.1	95
30	Array comparative genomic hybridization in patients with congenital diaphragmatic hernia: mapping of four CDH-critical regions and sequencing of candidate genes at 15q26.1â€“15q26.2. European Journal of Human Genetics, 2006, 14, 999-1008.	2.8	91
31	Andersenâ€Tawil syndrome: a model of clinical variability, pleiotropy, and genetic heterogeneity. Annals of Medicine, 2004, 36, 92-97.	3.8	85
32	Analysis of the dynamic mutation in the SCA7 gene shows marked parental effects on CAG repeat transmission. Human Molecular Genetics, 1998, 7, 525-532.	2.9	81
33	Deletions in CCM2 Are a Common Cause of Cerebral Cavernous Malformations. American Journal of Human Genetics, 2007, 80, 69-75.	6.2	80
34	Circadian Rhythm Gene Period 3 Is an Inhibitor of the Adipocyte Cell Fate. Journal of Biological Chemistry, 2011, 286, 9063-9070.	3.4	80
35	Episodic Neurological Channelopathies. Neuron, 2010, 68, 282-292.	8.1	79
36	Activation and Inactivation of the Voltage-Gated Sodium Channel: Role of Segment S5 Revealed by a Novel Hyperkalaemic Periodic Paralysis Mutation. Journal of Neuroscience, 1999, 19, 4762-4771.	3.6	77

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37	Genetics of the human circadian clock and sleep homeostat. Neuropsychopharmacology, 2020, 45, 45-54.	5.4	71
38	Proteolytic Cleavage of Ataxin-7 by Caspase-7 Modulates Cellular Toxicity and Transcriptional Dysregulation. Journal of Biological Chemistry, 2007, 282, 30150-30160.	3.4	69
39	Sodium channel inactivation defects are associated with acetazolamide-exacerbated hypokalemic periodic paralysis. Annals of Neurology, 2001, 50, 417-420.	5.3	68
40	Genomic context drives SCA7 CAG repeat instability, while expressed SCA7 cDNAs are intergenerationally and somatically stable in transgenic mice. Human Molecular Genetics, 2003, 12, 41-50.	2.9	68
41	Lamin B1 mediates cell-autonomous neuropathology in a leukodystrophy mouse model. Journal of Clinical Investigation, 2013, 123, 2719-2729.	8.2	68
42	FAD Regulates CRYPTOCHROME Protein Stability and Circadian Clock in Mice. Cell Reports, 2017, 19, 255-266.	6.4	64
43	A family with an unusual myotonic and myopathic phenotype and no CTG expansion (proximal myotonic) Tj ETQq1 1 0.784314 rgBT /O 143-150.	0.6	57
44	COL25A1 triggers and promotes Alzheimerâ€™s disease-like pathology in vivo. Neurogenetics, 2010, 11, 41-52.	1.4	56
45	A Rare Mutation of Î²1-Adrenergic Receptor Affects Sleep/Wake Behaviors. Neuron, 2019, 103, 1044-1055.e7.	8.1	54
46	Alternating Hemiplegia of Childhood: Retrospective Genetic Study and Genotype-Phenotype Correlations in 187 Subjects from the US AHCF Registry. PLoS ONE, 2015, 10, e0127045.	2.5	53
47	Mutations in PNKD causing paroxysmal dyskinesia alters protein cleavage and stability. Human Molecular Genetics, 2011, 20, 2322-2332.	2.9	52
48	DEC2 modulates orexin expression and regulates sleep. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 3434-3439.	7.1	51
49	TIMELESS mutation alters phase responsiveness and causes advanced sleep phase. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 12045-12053.	7.1	50
50	Human circadian variations. Journal of Clinical Investigation, 2021, 131, .	8.2	50
51	Dopamine dysregulation in a mouse model of paroxysmal nonkinesigenic dyskinesia. Journal of Clinical Investigation, 2012, 122, 507-518.	8.2	49
52	Episodic movement disorders as channelopathies. Movement Disorders, 2000, 15, 429-433.	3.9	47
53	Protein mutated in paroxysmal dyskinesia interacts with the active zone protein RIM and suppresses synaptic vesicle exocytosis. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 2935-2941.	7.1	47
54	Very large G protein-coupled receptor 1 regulates myelin-associated glycoprotein via G _{Î±s} /G _{Î±q} -mediated protein kinases A/C. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 19101-19106.	7.1	45

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55	Dual roles of FBXL3 in the mammalian circadian feedback loops are important for period determination and robustness of the clock. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 4750-4755.	7.1	44
56	Diversity of Human Clock Genotypes and Consequences. Progress in Molecular Biology and Translational Science, 2013, 119, 51-81.	1.7	43
57	Mutant neuropeptide S receptor reduces sleep duration with preserved memory consolidation. Science Translational Medicine, 2019, 11, .	12.4	43
58	Characterization of a new sodium channel mutation at arginine 1448 associated with moderate paramyotonia congenita in humans. Journal of Physiology, 1999, 518, 337-344.	2.9	41
59	Flecainide Suppresses Bidirectional Ventricular Tachycardia and Reverses Tachycardia-Induced Cardiomyopathy in Andersen-Tawil Syndrome. Journal of Cardiovascular Electrophysiology, 2007, 19, 070727020814005-???.	1.7	40
60	p75 Neurotrophin Receptor Is a Clock Gene That Regulates Oscillatory Components of Circadian and Metabolic Networks. Journal of Neuroscience, 2013, 33, 10221-10234.	3.6	38
61	Localization of the giant axonal neuropathy gene to chromosome 16q24. Annals of Neurology, 1998, 43, 143-148.	5.3	37
62	Ataxin-7 expression analysis in controls and spinocerebellar ataxia type 7 patients. Neurogenetics, 2001, 3, 83-90.	1.4	37
63	Kir2.6 Regulates the Surface Expression of Kir2.x Inward Rectifier Potassium Channels. Journal of Biological Chemistry, 2011, 286, 9526-9541.	3.4	35
64	The place of migraine as a channelopathy. Current Opinion in Neurology, 1998, 11, 217-226.	3.6	35
65	Genetic Mapping of a Locus (mass1) Causing Audiogenic Seizures in Mice. Genomics, 1998, 49, 188-192.	2.9	34
66	A double mutation in families with periodic paralysis defines new aspects of sodium channel slow inactivation. Journal of Clinical Investigation, 2000, 106, 431-438.	8.2	34
67	c-Fos immunohistochemical mapping of the audiogenic seizure network and tonotopic neuronal hyperexcitability in the inferior colliculus of the Frings mouse. Epilepsy Research, 2004, 62, 13-25.	1.6	33
68	Microfluidic droplet enrichment for targeted sequencing. Nucleic Acids Research, 2015, 43, e86-e86.	14.5	32
69	Linkage Analysis Narrows the Critical Region for Oculodentodigital Dysplasia to Chromosome 6q22â€“q23. Genomics, 1999, 58, 34-40.	2.9	31
70	Extreme morning chronotypes are often familial and not exceedingly rare: the estimated prevalence of advanced sleep phase, familial advanced sleep phase, and advanced sleepâ€“wake phase disorder in a sleep clinic population. Sleep, 2019, 42, .	1.1	31
71	Nuclear envelope protein MAN1 regulates clock through BMAL1. ELife, 2014, 3, e02981.	6.0	31
72	Genetic insights on sleep schedules: this time, it's PERsonal. Trends in Genetics, 2012, 28, 598-605.	6.7	28

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73	PKCÎ³ participates in food entrainment by regulating BMAL1. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 20679-20684.	7.1	27
74	Adult-Onset Autosomal Dominant Leukodystrophy: Linking Nuclear Envelope to Myelin. Journal of Neuroscience, 2011, 31, 1163-1166.	3.6	26
75	Mutations in Metabotropic Glutamate Receptor 1 Contribute to Natural Short Sleep Trait. Current Biology, 2021, 31, 13-24.e4.	3.9	25
76	Genetics of Human Sleep Behavioral Phenotypes. Methods in Enzymology, 2015, 552, 309-324.	1.0	24
77	Familial cortical myoclonus with a mutation in NOL3. Annals of Neurology, 2012, 72, 175-183.	5.3	23
78	Human genetics and sleep behavior. Current Opinion in Neurobiology, 2017, 44, 43-49.	4.2	23
79	Casein Kinase 1 Proteomics Reveal Prohibitin 2 Function in Molecular Clock. PLoS ONE, 2012, 7, e31987.	2.5	23
80	Episodic Disorders: Channelopathies and Beyond. Annual Review of Physiology, 2015, 77, 475-479.	13.1	21
81	The Genetics of the Human Circadian Clock. Advances in Genetics, 2011, 74, 231-247.	1.8	19
82	Ion channel Shake-down. Nature Genetics, 1994, 8, 111-112.	21.4	18
83	Anesthetic Management of Familial Hypokalemic Periodic Paralysis During Parturition. Anesthesia and Analgesia, 1999, 88, 1081-1082.	2.2	18
84	Channels and Disease. Archives of Neurology, 2004, 61, 1665.	4.5	18
85	Andersenâ€“Tawil syndrome: Report of 3 novel mutations and high risk of symptomatic cardiac involvement. Muscle and Nerve, 2015, 51, 192-196.	2.2	16
86	Channelopathies: Episodic Disorders of the Nervous System. Epilepsia, 2001, 42, 35-43.	5.1	15
87	A Novel Central Nervous Systemâ€“Enriched Spinocerebellar Ataxia Type 7 Gene Product. Archives of Neurology, 2003, 60, 97.	4.5	15
88	Enrichment of HapMap recombination hotspot predictions around human nervous system genes: evidence for positive selection ?. European Journal of Human Genetics, 2007, 15, 1071-1078.	2.8	14
89	Understanding the Role of Dicer in Astrocyte Development. PLoS ONE, 2015, 10, e0126667.	2.5	13
90	Auditory Deficits Associated with the Frings Mgr1 (Mass1) Mutation in Mice. Developmental Neuroscience, 2005, 27, 321-332.	2.0	12

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91	Novel familial cases of ICCA (infantile convulsions with paroxysmal choreoathetosis) syndrome. Epileptic Disorders, 2010, 12, 199-204.	1.3	12
92	Periodic paralyses and nondystrophic myotonias. Advances in Neurology, 2002, 88, 235-52.	0.8	12
93	Microglia are involved in the protection of memories formed during sleep deprivation. Neurobiology of Sleep and Circadian Rhythms, 2022, 12, 100073.	2.8	10
94	Paroxysmal Non-Kinesigenic Dyskinesia Caused by the Mutation of <i>MR-1</i> in a Large Polish Kindred. European Neurology, 2009, 61, 39-41.	1.4	9
95	Sick and tired: how molecular regulators of human sleep schedules and duration impact immune function. Current Opinion in Neurobiology, 2013, 23, 873-879.	4.2	9
96	Solving the mystery of human sleep schedules one mutation at a time. Critical Reviews in Biochemistry and Molecular Biology, 2013, 48, 465-475.	5.2	9
97	Episodic and Electrical Nervous System Disorders Caused by Nonchannel Genes. Annual Review of Physiology, 2015, 77, 525-541.	13.1	9
98	Disorders of sleep and circadian rhythms. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 148, 531-538.	1.8	8
99	A Mitochondrial <scp>tRNA</scp> Mutation Causes Axonal <scp>CMT</scp> in a Large Venezuelan Family. Annals of Neurology, 2020, 88, 830-842.	5.3	7
100	Clinicâ€“based study of family history of vascular risk factors and migraine. Journal of Headache and Pain, 2005, 6, 412-416.	6.0	6
101	Familial natural short sleep mutations reduce Alzheimer pathology in mice. IScience, 2022, 25, 103964.	4.1	6
102	Channel Surfing. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 4879-4880.	3.6	5
103	What's new in epilepsy genetics?. Molecular Psychiatry, 2003, 8, 463-465.	7.9	5
104	Bioinformatic analysis of human CNS-expressed ion channels as candidates for episodic nervous system disorders. Neurogenetics, 2007, 8, 159-168.	1.4	5
105	Channelopathies: Episodic Disorders of the Nervous System. Novartis Foundation Symposium, 2008, , 87-108.	1.1	5
106	Molecular biology of episodic movement disorders. Advances in Neurology, 2002, 89, 453-8.	0.8	3
107	Genetic Approaches to Human Behavior. Methods in Enzymology, 2005, 393, 239-250.	1.0	2
108	Channelopathies: episodic disorders of the nervous system. Novartis Foundation Symposium, 2002, 241, 87-104; discussion 104-8, 226-32.	1.1	2

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109	Correspondence. Medical Hypotheses, 2000, 55, 457.	1.5	1
110	No Gastrointestinal Dysmotility in Transgenic Mouse Models of Migraine. Headache, 2020, 60, 396-404.	3.9	1
111	GENETICS OF EPILEPSY. CONTINUUM Lifelong Learning in Neurology, 2005, 11, 79-94.	0.8	0
112	Approaching Inherited Disease on a Genomic Scale. Current Genomics, 2005, 6, 545-549.	1.6	0
113	Report of a Turkish girl with Andersen-Tawil syndrome. Journal of Pediatric Neurology, 2015, 04, 279-282.	0.2	0
114	Developing the field of neurogenetics. Neurogenetics, 2017, 18, 183-184.	1.4	0
115	0153 Extreme Morning Chronotypes Are Often Familial And Not Exceedingly Rare: The Estimated Prevalence Of Familial Advanced Sleep Phase (FASP) In A Sleep Clinic Population. Sleep, 2019, 42, A62-A63.	1.1	0
116	Raymond Leslie White (1943â€“2018). American Journal of Human Genetics, 2019, 104, 8-10.	6.2	0
117	The whole is greater than the sum of the parts. Journal of Clinical Investigation, 2021, 131, .	8.2	0
118	Welcoming articles on genotype-dependent clinical features and diagnostics. Neurogenetics, 2021, 22, 103-104.	1.4	0
119	Spinocerebellar ataxia type 4. , 2001, , 440-444.		0
120	Epilepsies as channelopathies. , 2001, , 1-14.		0
121	Proteolytic cleavage of ataxin-7 by caspase-7 modulates cellular toxicity and transcriptional dysregulation. VOLUME 282 (2007) PAGES 30150-30160. Journal of Biological Chemistry, 2008, 283, 16960.	3.4	0
122	Louis PtÃ¡Äek receives the 2015 ASCI/Stanley J. Korsmeyer Award. Journal of Clinical Investigation, 2015, 125, 1369-1370.	8.2	0
123	Genetic and biological factors in sleep. , 2022, , 73-95.		0