Roger Pamphlett

List of Publications by Year
in descending order

97	4,554	172457	123424
papers	citations	29 h-index	61 g-index

103	103	103	5852
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Polygenic risk score analysis for amyotrophic lateral sclerosis leveraging cognitive performance, educational attainment and schizophrenia. European Journal of Human Genetics, 2022, 30, 532-539.	2.8	16
2	Mercury is present in neurons and oligodendrocytes in regions of the brain affected by Parkinsonâ€™s disease and co-localises with Lewy bodies. PLoS ONE, 2022, 17, e0262464.	2.5	15
3	Genome-wide study of DNA methylation shows alterations in metabolic, inflammatory, and cholesterol pathways in ALS. Science Translational Medicine, 2022, 14, eabj0264.	12.4	38
4	Microglia and monocytes in inflammatory CNS disease: integrating phenotype and function. Acta Neuropathologica, 2022, 143, 179-224.	7.7	82
5	NEK1 and STMN2 short tandem repeat lengths are not associated with Australian amyotrophic lateral sclerosis risk. Neurobiology of Aging, 2022, , .	3.1	0
6	Evidence for polygenic and oligogenic basis of Australian sporadic amyotrophic lateral sclerosis. Journal of Medical Genetics, 2021, 58, 87-95.	3.2	48
7	Mercury in the human thyroid gland: Potential implications for thyroid cancer, autoimmune thyroiditis, and hypothyroidism. PLoS ONE, 2021, 16, e0246748.	2.5	18
8	Mercury in the human adrenal medulla could contribute to increased plasma noradrenaline in aging. Scientific Reports, 2021, 11, 2961.	3.3	6
9	Meta-analysis of genome-wide DNA methylation identifies shared associations across neurodegenerative disorders. Genome Biology, 2021, 22, 90.	8.8	49
10	The Prevalence of Inorganic Mercury in Human Kidneys Suggests a Role for Toxic Metals in Essential Hypertension. Toxics, 2021, 9, 67.	3.7	11
11	Genetic analysis of GLT8D1 and ARPP21 in Australian familial and sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2021, 101, 297.e9-297.e11.	3.1	6
12	Genetic Analysis of Tryptophan Metabolism Genes in Sporadic Amyotrophic Lateral Sclerosis. Frontiers in Immunology, 2021, 12, 701550.	4.8	8
13	The prevalence of inorganic mercury in human cells increases during aging but decreases in the very old. Scientific Reports, 2021, 11, 16714.	3.3	7
14	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	9.0	46
15	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. Nature Genetics, 2021, 53, 1636-1648.	21.4	223
16	Genome-wide Meta-analysis Finds the ACSL5-ZDHHC6 Locus Is Associated with ALS and Links Weight Loss to the Disease Genetics. Cell Reports, 2020, 33, 108323.	6.4	41
17	Mercury in Pancreatic Cells of People with and without Pancreatic Cancer. International Journal of Environmental Research and Public Health, 2020, 17, 8990.	2.6	9
18	Concentrations of toxic metals and essential trace elements vary among individual neurons in the human locus ceruleus. PLoS ONE, 2020, 15, e0233300.	2.5	21

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19	A rare CACNA1H variant associated with amyotrophic lateral sclerosis causes complete loss of Cav3.2â€‰T-type channel activity. Molecular Brain, 2020, 13, 33.	2.6	14
20	Significant out-of-sample classification from methylation profile scoring for amyotrophic lateral sclerosis. Npj Genomic Medicine, 2020, 5, 10.	3.8	25
21	Elemental bioimaging shows mercury and other toxic metals in normal breast tissue and in breast cancers. PLoS ONE, 2020, 15, e0228226.	2.5	17
22	Elemental imaging shows mercury in cells of the human lateral and medial geniculate nuclei. PLoS ONE, 2020, 15, e0231870.	2.5	8
23	The distribution of toxic metals in the human retina and optic nerve head: Implications for age-related macular degeneration. PLoS ONE, 2020, 15, e0241054.	2.5	21
24	Challenges in diagnosing hydroxychloroquine myopathy during the COVID â€19 pandemic. Internal Medicine Journal, 2020, 50, 1559-1562.	0.8	2
25	Mercury Is Taken Up Selectively by Cells Involved in Joint, Bone, and Connective Tissue Disorders. Frontiers in Medicine, 2019, 6, 168.	2.6	11
26	Mercury in the retina and optic nerve following prenatal exposure to mercury vapor. PLoS ONE, 2019, 14, e0220859.	2.5	22
27	Elemental Analysis of Aging Human Pituitary Glands Implicates Mercury as a Contributor to the Somatopause. Frontiers in Endocrinology, 2019, 10, 419.	3.5	14
28	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. Annals of Neurology, 2019, 85, 470-481.	5.3	118
29	Antiâ€SRP associated necrotizing autoimmune myopathy presenting with asymptomatically elevated creatine kinase. Muscle and Nerve, 2019, 59, E17-E19.	2.2	5
30	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517
31	A Comparison of Mercury Exposure from Seafood Consumption and Dental Amalgam Fillings in People with and without Amyotrophic Lateral Sclerosis (ALS): An International Online Case-Control Study. International Journal of Environmental Research and Public Health, 2018, 15, 2874.	2.6	23
32	Is psychological stress a predisposing factor for amyotrophic lateral sclerosis (ALS)? An online international case-control study of premorbid life events, occupational stress, resilience and anxiety. PLoS ONE, 2018, 13, e0204424.	2.5	7
33	Are people with amyotrophic lateral sclerosis (ALS) particularly nice? An international online caseâ€“control study of the Big Five personality factors. Brain and Behavior, 2018, 8, e01119.	2.2	3
34	Age-related accumulation of toxic metals in the human locus ceruleus. PLoS ONE, 2018, 13, e0203627.	2.5	33
35	Inorganic mercury in human astrocytes, oligodendrocytes, corticomotoneurons and the locus ceruleus: implications for multiple sclerosis, neurodegenerative disorders and gliomas. BioMetals, 2018, 31, 807-819.	4.1	39
36	Cross-ethnic meta-analysis identifies association of the GPX3-TNIP1 locus with amyotrophic lateral sclerosis. Nature Communications, 2017, 8, 611.	12.8	93

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37	Does the index-to-ring finger length ratio (2D:4D) differ in amyotrophic lateral sclerosis (ALS)? Results from an international online caseâ€“control study. BMJ Open, 2017, 7, e016924.	1.9	5
38	Environmental insults: critical triggers for amyotrophic lateral sclerosis. Translational Neurodegeneration, 2017, 6, 15.	8.0	37
39	Epigenetic differences between monozygotic twins discordant for amyotrophic lateral sclerosis (ALS) provide clues to disease pathogenesis. PLoS ONE, 2017, 12, e0182638.	2.5	61
40	Age-Related Uptake of Heavy Metals in Human Spinal Interneurons. PLoS ONE, 2016, 11, e0162260.	2.5	16
41	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1043-1048.	21.4	494
42	CACNA1Hmissense mutations associated with amyotrophic lateral sclerosis alter Ca_v3.2 T-type calcium channel activity and reticular thalamic neuron firing. Channels, 2016, 10, 466-477.	2.8	30
43	Locus ceruleus neurons in people with autism contain no histochemically-detectable mercury. BioMetals, 2016, 29, 171-175.	4.1	12
44	Rhabdomyolysis as a late complication of bariatric surgery. Journal of the Neurological Sciences, 2016, 364, 102-104.	0.6	2
45	Different Populations of Human Locus Ceruleus Neurons Contain Heavy Metals or Hyperphosphorylated Tau: Implications for Amyloid-Î² and Tau Pathology in Alzheimer's Disease. Journal of Alzheimer's Disease, 2015, 45, 437-447.	2.6	37
46	Whole genome analyses reveal no pathogenetic single nucleotide or structural differences between monozygotic twins discordant for amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2015, 16, 385-392.	1.7	27
47	Exome sequencing of case-unaffected-parents trios reveals recessive and de novo genetic variants in sporadic ALS. Scientific Reports, 2015, 5, 9124.	3.3	53
48	Designing an Internationally Accessible Web-Based Questionnaire to Discover Risk Factors for Amyotrophic Lateral Sclerosis: A Case-Control Study. JMIR Research Protocols, 2015, 4, e96.	1.0	7
49	Uptake of environmental toxicants by the locus ceruleus: A potential trigger for neurodegenerative, demyelinating and psychiatric disorders. Medical Hypotheses, 2014, 82, 97-104.	1.5	51
50	Is the Risk of Motor Neuron Disease Increased or Decreased after Cancer? An Australian Case-Control Study. PLoS ONE, 2014, 9, e103572.	2.5	6
51	Uptake of inorganic mercury by human locus ceruleus and corticomotor neurons: implications for amyotrophic lateral sclerosis. Acta Neuropathologica Communications, 2013, 1, 13.	5.2	38
52	Heavy metals in locus ceruleus and motor neurons in motor neuron disease. Acta Neuropathologica Communications, 2013, 1, 81.	5.2	25
53	Can ALS-Associated C9orf72 Repeat Expansions Be Diagnosed on a Blood DNA Test Alone?. PLoS ONE, 2013, 8, e70007.	2.5	18
54	Different Occupations Associated with Amyotrophic Lateral Sclerosis: Is Diesel Exhaust the Link?. PLoS ONE, 2013, 8, e80993.	2.5	23

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55	Smoking Is Not a Risk Factor for Sporadic Amyotrophic Lateral Sclerosis in an Australian Population. Neuroepidemiology, 2012, 38, 106-113.	2.3	16
56	Transmission of C9orf72 hexanucleotide repeat expansions in sporadic amyotrophic lateral sclerosis. NeuroReport, 2012, 23, 556-559.	1.2	16
57	Season and weather patterns at time of birth in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 459-464.	2.1	8
58	An approach to finding brain-situated mutations in sporadic Parkinson's disease. Parkinsonism and Related Disorders, 2012, 18, 82-85.	2.2	7
59	Frequency of the C9orf72 hexanucleotide repeat expansion in patients with amyotrophic lateral sclerosis and frontotemporal dementia: a cross-sectional study. Lancet Neurology, The, 2012, 11, 323-330.	10.2	1,039
60	The â€œsomatic-spreadâ€•hypothesis for sporadic neurodegenerative diseases. Medical Hypotheses, 2011, 77, 544-547.	1.5	6
61	Copy number imbalances in blood and hair in monozygotic twins discordant for amyotrophic lateral sclerosis. Journal of Clinical Neuroscience, 2011, 18, 1231-1234.	1.5	11
62	Inorganic mercury within motor neurons does not cause the TDP-43 changes seen in sporadic ALS. Toxicology Letters, 2011, 201, 58-61.	0.8	7
63	Looking for differences in copy number between blood and brain in sporadic amyotrophic lateral sclerosis. Muscle and Nerve, 2011, 44, 492-498.	2.2	18
64	Using case-parent trios to look for rare de novo genetic variants in adult-onset neurodegenerative diseases. Journal of Neuroscience Methods, 2011, 197, 297-301.	2.5	22
65	STUDY OF 962 PATIENTS INDICATES PROGRESSIVE MUSCULAR ATROPHY IS A FORM OF ALS. Neurology, 2010, 74, 1926-1927.	1.1	6
66	DHPLC can be used to detect low-level mutations in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2010, 11, 76-82.	2.1	6
67	Genetic variants in the promoter of TARDBP in sporadic amyotrophic lateral sclerosis. Neuromuscular Disorders, 2009, 19, 696-700.	0.6	24
68	A genome-wide analysis of brain DNA methylation identifies new candidate genes for sporadic amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2009, 10, 418-429.	2.1	82
69	A comparison of the lengths of androgen receptor triplet repeats in brain and blood in motor neuron diseases. Journal of the Neurological Sciences, 2008, 267, 125-128.	0.6	6
70	An analysis of the entire SOD1 gene in sporadic ALS. Neuromuscular Disorders, 2008, 18, 545-552.	0.6	16
71	An epigenetic analysis of SOD1 and VEGF in ALS. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2007, 8, 83-86.	2.1	49
72	A geneâ€“environment study of the paraoxonase 1 gene and pesticides in amyotrophic lateral sclerosis. NeuroToxicology, 2007, 28, 532-540.	3.0	59

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73	Are metallothionein genes silenced in ALS?. Toxicology Letters, 2007, 168, 83-87.	0.8	29
74	Genetic susceptibility to environmental toxicants in ALS. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 885-890.	1.7	63
75	Amyotrophic Lateral Sclerosis and Exposure to Environmental Toxins: An Australian Case-Control Study. Neuroepidemiology, 2006, 27, 130-135.	2.3	81
76	Screening the metallothionein III gene in sporadic amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2005, 6, 115-117.	2.1	3
77	Detection of mutations in whole genome-amplified DNA from laser-microdissected neurons. Journal of Neuroscience Methods, 2005, 147, 65-67.	2.5	9
78	Does selenium deficiency unmask mercury toxicity in motor neurons?. Neurotoxicology and Teratology, 2005, 27, 241-244.	2.4	6
79	Flaviviruses in motor neuron disease. Muscle and Nerve, 2005, 32, 108-109.	2.2	3
80	It takes only 100 trueâ€“false items to test medical students: true or false?. Medical Teacher, 2005, 27, 468-470.	1.8	9
81	Somatic mutation: a cause of sporadic neurodegenerative diseases?. Medical Hypotheses, 2004, 62, 679-682.	1.5	28
82	A polymorphism in the poliovirus receptor gene differs in motor neuron disease. NeuroReport, 2004, 15, 383-386.	1.2	22
83	Is quantitation necessary for assessment of sural nerve biopsies?. Muscle and Nerve, 2003, 27, 562-569.	2.2	8
84	Magnesium supplementation does not delay disease onset or increase survival in a mouse model of familial ALS. Journal of the Neurological Sciences, 2003, 216, 95-98.	0.6	11
85	Zinc in the spinal cord of a mutant SOD1 mouse model of ALS. NeuroReport, 2003, 14, 547-549.	1.2	0
86	Severe infantile axonal neuropathy with respiratory failure. Muscle and Nerve, 2001, 24, 760-768.	2.2	33
87	Mercury vapor uptake into the nervous system of developing mice. Neurotoxicology and Teratology, 2001, 23, 191-196.	2.4	19
88	Uptake of bismuth in motor neurons of mice after single oral doses of bismuth compounds. Neurotoxicology and Teratology, 2000, 22, 559-563.	2.4	37
89	Bismuth Autometallography: Protocol, Specificity, and Differentiation. Journal of Histochemistry and Cytochemistry, 2000, 48, 1503-1510.	2.5	37
90	Oxidative damage to nucleic acids in motor neurons containing mercury. Journal of the Neurological Sciences, 1998, 159, 121-126.	0.6	27

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91	Shrinkage of Motor Axons following Systemic Exposure to Inorganic Mercury. Journal of Neuropathology and Experimental Neurology, 1998, 57, 330-36.	1.7	37
92	Motor neuron uptake of low dose inorganic mercury. Journal of the Neurological Sciences, 1996, 135, 63-67.	0.6	42
93	Motor neuron disease: A primary disorder of corticomotoneurons?. Muscle and Nerve, 1995, 18, 314-318.	2.2	50
94	Spinal Cord Injury After Forceps Rotation: the Role of Glioneuronal Heterotopias. Australian and New Zealand Journal of Obstetrics and Gynaecology, 1993, 33, 91-93.	1.0	4
95	Seesaw nystagmus and ocular tilt reaction due to adult Leigh's disease. Neuro-Ophthalmology, 1992, 12, 1-9.	1.0	16
96	The effect of nerve crush and botulinum toxin on lead uptake in motor axons. Acta Neuropathologica, 1992, 84, 89-93.	7.7	5
97	Lead uptake in motor axons. Muscle and Nerve, 1992, 15, 620-625.	2.2	8