

Roger Pamphlett

List of Publications by Year in descending order

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Version: 2024-02-01

97
papers

4,554
citations

172457

29
h-index

123424

61
g-index

103
all docs

103
docs citations

103
times ranked

5852
citing authors

#	ARTICLE	IF	CITATIONS
1	Frequency of the C9orf72 hexanucleotide repeat expansion in patients with amyotrophic lateral sclerosis and frontotemporal dementia: a cross-sectional study. <i>Lancet Neurology</i> , The, 2012, 11, 323-330.	10.2	1,039
2	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018, 97, 1268-1283.e6.	8.1	517
3	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1043-1048.	21.4	494
4	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. <i>Nature Genetics</i> , 2021, 53, 1636-1648.	21.4	223
5	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2019, 85, 470-481.	5.3	118
6	Cross-ethnic meta-analysis identifies association of the GPX3-TNIP1 locus with amyotrophic lateral sclerosis. <i>Nature Communications</i> , 2017, 8, 611.	12.8	93
7	A genome-wide analysis of brain DNA methylation identifies new candidate genes for sporadic amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2009, 10, 418-429.	2.1	82
8	Microglia and monocytes in inflammatory CNS disease: integrating phenotype and function. <i>Acta Neuropathologica</i> , 2022, 143, 179-224.	7.7	82
9	Amyotrophic Lateral Sclerosis and Exposure to Environmental Toxins: An Australian Case-Control Study. <i>Neuroepidemiology</i> , 2006, 27, 130-135.	2.3	81
10	Genetic susceptibility to environmental toxicants in ALS. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2007, 144B, 885-890.	1.7	63
11	Epigenetic differences between monozygotic twins discordant for amyotrophic lateral sclerosis (ALS) provide clues to disease pathogenesis. <i>PLoS ONE</i> , 2017, 12, e0182638.	2.5	61
12	A gene-environment study of the paraoxonase 1 gene and pesticides in amyotrophic lateral sclerosis. <i>NeuroToxicology</i> , 2007, 28, 532-540.	3.0	59
13	Exome sequencing of case-unaffected-parents trios reveals recessive and de novo genetic variants in sporadic ALS. <i>Scientific Reports</i> , 2015, 5, 9124.	3.3	53
14	Uptake of environmental toxicants by the locus ceruleus: A potential trigger for neurodegenerative, demyelinating and psychiatric disorders. <i>Medical Hypotheses</i> , 2014, 82, 97-104.	1.5	51
15	Motor neuron disease: A primary disorder of corticomotoneurons?. <i>Muscle and Nerve</i> , 1995, 18, 314-318.	2.2	50
16	An epigenetic analysis of SOD1 and VEGF in ALS. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2007, 8, 83-86.	2.1	49
17	Meta-analysis of genome-wide DNA methylation identifies shared associations across neurodegenerative disorders. <i>Genome Biology</i> , 2021, 22, 90.	8.8	49
18	Evidence for polygenic and oligogenic basis of Australian sporadic amyotrophic lateral sclerosis. <i>Journal of Medical Genetics</i> , 2021, 58, 87-95.	3.2	48

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19	Association of Variants in the <i>SPTLC1</i> Gene With Juvenile Amyotrophic Lateral Sclerosis. <i>JAMA Neurology</i> , 2021, 78, 1236.	9.0	46
20	Motor neuron uptake of low dose inorganic mercury. <i>Journal of the Neurological Sciences</i> , 1996, 135, 63-67.	0.6	42
21	Genome-wide Meta-analysis Finds the <i>ACSL5-ZDHHC6</i> Locus Is Associated with ALS and Links Weight Loss to the Disease Genetics. <i>Cell Reports</i> , 2020, 33, 108323.	6.4	41
22	Inorganic mercury in human astrocytes, oligodendrocytes, corticomotoneurons and the locus ceruleus: implications for multiple sclerosis, neurodegenerative disorders and gliomas. <i>BioMetals</i> , 2018, 31, 807-819.	4.1	39
23	Uptake of inorganic mercury by human locus ceruleus and corticomotor neurons: implications for amyotrophic lateral sclerosis. <i>Acta Neuropathologica Communications</i> , 2013, 1, 13.	5.2	38
24	Genome-wide study of DNA methylation shows alterations in metabolic, inflammatory, and cholesterol pathways in ALS. <i>Science Translational Medicine</i> , 2022, 14, eabj0264.	12.4	38
25	Shrinkage of Motor Axons following Systemic Exposure to Inorganic Mercury. <i>Journal of Neuropathology and Experimental Neurology</i> , 1998, 57, 330-36.	1.7	37
26	Uptake of bismuth in motor neurons of mice after single oral doses of bismuth compounds. <i>Neurotoxicology and Teratology</i> , 2000, 22, 559-563.	2.4	37
27	Bismuth Autometallography: Protocol, Specificity, and Differentiation. <i>Journal of Histochemistry and Cytochemistry</i> , 2000, 48, 1503-1510.	2.5	37
28	Different Populations of Human Locus Ceruleus Neurons Contain Heavy Metals or Hyperphosphorylated Tau: Implications for Amyloid- β^2 and Tau Pathology in Alzheimer's Disease. <i>Journal of Alzheimer's Disease</i> , 2015, 45, 437-447.	2.6	37
29	Environmental insults: critical triggers for amyotrophic lateral sclerosis. <i>Translational Neurodegeneration</i> , 2017, 6, 15.	8.0	37
30	Severe infantile axonal neuropathy with respiratory failure. <i>Muscle and Nerve</i> , 2001, 24, 760-768.	2.2	33
31	Age-related accumulation of toxic metals in the human locus ceruleus. <i>PLoS ONE</i> , 2018, 13, e0203627.	2.5	33
32	<i>CACNA1H</i> missense mutations associated with amyotrophic lateral sclerosis alter Ca^{2+} T-type calcium channel activity and reticular thalamic neuron firing. <i>Channels</i> , 2016, 10, 466-477.	2.8	30
33	Are metallothionein genes silenced in ALS?. <i>Toxicology Letters</i> , 2007, 168, 83-87.	0.8	29
34	Somatic mutation: a cause of sporadic neurodegenerative diseases?. <i>Medical Hypotheses</i> , 2004, 62, 679-682.	1.5	28
35	Oxidative damage to nucleic acids in motor neurons containing mercury. <i>Journal of the Neurological Sciences</i> , 1998, 159, 121-126.	0.6	27
36	Whole genome analyses reveal no pathogenetic single nucleotide or structural differences between monozygotic twins discordant for amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2015, 16, 385-392.	1.7	27

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37	Heavy metals in locus ceruleus and motor neurons in motor neuron disease. <i>Acta Neuropathologica Communications</i> , 2013, 1, 81.	5.2	25
38	Significant out-of-sample classification from methylation profile scoring for amyotrophic lateral sclerosis. <i>Npj Genomic Medicine</i> , 2020, 5, 10.	3.8	25
39	Genetic variants in the promoter of TARDBP in sporadic amyotrophic lateral sclerosis. <i>Neuromuscular Disorders</i> , 2009, 19, 696-700.	0.6	24
40	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. <i>International Journal of Environmental Research and Public Health</i> , 2018, 15, 2874.	2.6	23
41	Different Occupations Associated with Amyotrophic Lateral Sclerosis: Is Diesel Exhaust the Link?. <i>PLoS ONE</i> , 2013, 8, e80993.	2.5	23
42	A polymorphism in the poliovirus receptor gene differs in motor neuron disease. <i>NeuroReport</i> , 2004, 15, 383-386.	1.2	22
43	Using case-parent trios to look for rare de novo genetic variants in adult-onset neurodegenerative diseases. <i>Journal of Neuroscience Methods</i> , 2011, 197, 297-301.	2.5	22
44	Mercury in the retina and optic nerve following prenatal exposure to mercury vapor. <i>PLoS ONE</i> , 2019, 14, e0220859.	2.5	22
45	Concentrations of toxic metals and essential trace elements vary among individual neurons in the human locus ceruleus. <i>PLoS ONE</i> , 2020, 15, e0233300.	2.5	21
46	The distribution of toxic metals in the human retina and optic nerve head: Implications for age-related macular degeneration. <i>PLoS ONE</i> , 2020, 15, e0241054.	2.5	21
47	Mercury vapor uptake into the nervous system of developing mice. <i>Neurotoxicology and Teratology</i> , 2001, 23, 191-196.	2.4	19
48	Looking for differences in copy number between blood and brain in sporadic amyotrophic lateral sclerosis. <i>Muscle and Nerve</i> , 2011, 44, 492-498.	2.2	18
49	Can ALS-Associated C9orf72 Repeat Expansions Be Diagnosed on a Blood DNA Test Alone?. <i>PLoS ONE</i> , 2013, 8, e70007.	2.5	18
50	Mercury in the human thyroid gland: Potential implications for thyroid cancer, autoimmune thyroiditis, and hypothyroidism. <i>PLoS ONE</i> , 2021, 16, e0246748.	2.5	18
51	Elemental bioimaging shows mercury and other toxic metals in normal breast tissue and in breast cancers. <i>PLoS ONE</i> , 2020, 15, e0228226.	2.5	17
52	Seesaw nystagmus and ocular tilt reaction due to adult Leigh's disease. <i>Neuro-Ophthalmology</i> , 1992, 12, 1-9.	1.0	16
53	An analysis of the entire SOD1 gene in sporadic ALS. <i>Neuromuscular Disorders</i> , 2008, 18, 545-552.	0.6	16
54	Smoking Is Not a Risk Factor for Sporadic Amyotrophic Lateral Sclerosis in an Australian Population. <i>Neuroepidemiology</i> , 2012, 38, 106-113.	2.3	16

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55	Transmission of C9orf72 hexanucleotide repeat expansions in sporadic amyotrophic lateral sclerosis. <i>NeuroReport</i> , 2012, 23, 556-559.	1.2	16
56	Age-Related Uptake of Heavy Metals in Human Spinal Interneurons. <i>PLoS ONE</i> , 2016, 11, e0162260.	2.5	16
57	Polygenic risk score analysis for amyotrophic lateral sclerosis leveraging cognitive performance, educational attainment and schizophrenia. <i>European Journal of Human Genetics</i> , 2022, 30, 532-539.	2.8	16
58	Mercury is present in neurons and oligodendrocytes in regions of the brain affected by Parkinson's disease and co-localises with Lewy bodies. <i>PLoS ONE</i> , 2022, 17, e0262464.	2.5	15
59	Elemental Analysis of Aging Human Pituitary Glands Implicates Mercury as a Contributor to the Somatopause. <i>Frontiers in Endocrinology</i> , 2019, 10, 419.	3.5	14
60	A rare CACNA1H variant associated with amyotrophic lateral sclerosis causes complete loss of Cav3.2-type channel activity. <i>Molecular Brain</i> , 2020, 13, 33.	2.6	14
61	Locus ceruleus neurons in people with autism contain no histochemically-detectable mercury. <i>BioMetals</i> , 2016, 29, 171-175.	4.1	12
62	Magnesium supplementation does not delay disease onset or increase survival in a mouse model of familial ALS. <i>Journal of the Neurological Sciences</i> , 2003, 216, 95-98.	0.6	11
63	Copy number imbalances in blood and hair in monozygotic twins discordant for amyotrophic lateral sclerosis. <i>Journal of Clinical Neuroscience</i> , 2011, 18, 1231-1234.	1.5	11
64	Mercury Is Taken Up Selectively by Cells Involved in Joint, Bone, and Connective Tissue Disorders. <i>Frontiers in Medicine</i> , 2019, 6, 168.	2.6	11
65	The Prevalence of Inorganic Mercury in Human Kidneys Suggests a Role for Toxic Metals in Essential Hypertension. <i>Toxics</i> , 2021, 9, 67.	3.7	11
66	Detection of mutations in whole genome-amplified DNA from laser-microdissected neurons. <i>Journal of Neuroscience Methods</i> , 2005, 147, 65-67.	2.5	9
67	It takes only 100 true/false items to test medical students: true or false?. <i>Medical Teacher</i> , 2005, 27, 468-470.	1.8	9
68	Mercury in Pancreatic Cells of People with and without Pancreatic Cancer. <i>International Journal of Environmental Research and Public Health</i> , 2020, 17, 8990.	2.6	9
69	Lead uptake in motor axons. <i>Muscle and Nerve</i> , 1992, 15, 620-625.	2.2	8
70	Is quantitation necessary for assessment of sural nerve biopsies?. <i>Muscle and Nerve</i> , 2003, 27, 562-569.	2.2	8
71	Season and weather patterns at time of birth in amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2012, 13, 459-464.	2.1	8
72	Elemental imaging shows mercury in cells of the human lateral and medial geniculate nuclei. <i>PLoS ONE</i> , 2020, 15, e0231870.	2.5	8

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73	Genetic Analysis of Tryptophan Metabolism Genes in Sporadic Amyotrophic Lateral Sclerosis. <i>Frontiers in Immunology</i> , 2021, 12, 701550.	4.8	8
74	Inorganic mercury within motor neurons does not cause the TDP-43 changes seen in sporadic ALS. <i>Toxicology Letters</i> , 2011, 201, 58-61.	0.8	7
75	An approach to finding brain-situated mutations in sporadic Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , 2012, 18, 82-85.	2.2	7
76	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. <i>PLoS ONE</i> , 2018, 13, e0204424.	2.5	7
77	The prevalence of inorganic mercury in human cells increases during aging but decreases in the very old. <i>Scientific Reports</i> , 2021, 11, 16714.	3.3	7
78	Designing an Internationally Accessible Web-Based Questionnaire to Discover Risk Factors for Amyotrophic Lateral Sclerosis: A Case-Control Study. <i>JMIR Research Protocols</i> , 2015, 4, e96.	1.0	7
79	Does selenium deficiency unmask mercury toxicity in motor neurons?. <i>Neurotoxicology and Teratology</i> , 2005, 27, 241-244.	2.4	6
80	A comparison of the lengths of androgen receptor triplet repeats in brain and blood in motor neuron diseases. <i>Journal of the Neurological Sciences</i> , 2008, 267, 125-128.	0.6	6
81	STUDY OF 962 PATIENTS INDICATES PROGRESSIVE MUSCULAR ATROPHY IS A FORM OF ALS. <i>Neurology</i> , 2010, 74, 1926-1927.	1.1	6
82	DHPLC can be used to detect low-level mutations in amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2010, 11, 76-82.	2.1	6
83	The "somatic-spread" hypothesis for sporadic neurodegenerative diseases. <i>Medical Hypotheses</i> , 2011, 77, 544-547.	1.5	6
84	Mercury in the human adrenal medulla could contribute to increased plasma noradrenaline in aging. <i>Scientific Reports</i> , 2021, 11, 2961.	3.3	6
85	Genetic analysis of GLT8D1 and ARPP21 in Australian familial and sporadic amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2021, 101, 297.e9-297.e11.	3.1	6
86	Is the Risk of Motor Neuron Disease Increased or Decreased after Cancer? An Australian Case-Control Study. <i>PLoS ONE</i> , 2014, 9, e103572.	2.5	6
87	The effect of nerve crush and botulinum toxin on lead uptake in motor axons. <i>Acta Neuropathologica</i> , 1992, 84, 89-93.	7.7	5
88	Does the index-to-ring finger length ratio (2D:4D) differ in amyotrophic lateral sclerosis (ALS)? Results from an international online case-control study. <i>BMJ Open</i> , 2017, 7, e016924.	1.9	5
89	Anti-SRP associated necrotizing autoimmune myopathy presenting with asymptotically elevated creatine kinase. <i>Muscle and Nerve</i> , 2019, 59, E17-E19.	2.2	5
90	Spinal Cord Injury After Forceps Rotation: the Role of Glioneuronal Heterotopias. <i>Australian and New Zealand Journal of Obstetrics and Gynaecology</i> , 1993, 33, 91-93.	1.0	4

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91	Screening the metallothionein III gene in sporadic amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2005, 6, 115-117.	2.1	3
92	Flaviviruses in motor neuron disease. <i>Muscle and Nerve</i> , 2005, 32, 108-109.	2.2	3
93	Are people with amyotrophic lateral sclerosis (ALS) particularly nice? An international online caseâ€“control study of the Big Five personality factors. <i>Brain and Behavior</i> , 2018, 8, e01119.	2.2	3
94	Rhabdomyolysis as a late complication of bariatric surgery. <i>Journal of the Neurological Sciences</i> , 2016, 364, 102-104.	0.6	2
95	Challenges in diagnosing hydroxychloroquine myopathy during the COVID â€“19 pandemic. <i>Internal Medicine Journal</i> , 2020, 50, 1559-1562.	0.8	2
96	Zinc in the spinal cord of a mutant SOD1 mouse model of ALS. <i>NeuroReport</i> , 2003, 14, 547-549.	1.2	0
97	NEK1 and STMN2 short tandem repeat lengths are not associated with Australian amyotrophic lateral sclerosis risk. <i>Neurobiology of Aging</i> , 2022, , .	3.1	0