Janine Kirby

List of Publications by Year in descending order

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95 papers	8,768 citations	50276 46 h-index	45317 90 g-index
96 all docs	96 docs citations	96 times ranked	9696 citing authors

#	Article	IF	Citations
1	Multicentre appraisal of amyotrophic lateral sclerosis biofluid biomarkers shows primacy of blood neurofilament light chain. Brain Communications, 2022, 4, fcac029.	3.3	29
2	TDP43 proteinopathy is associated with aberrant DNA methylation in human amyotrophic lateral sclerosis. Neuropathology and Applied Neurobiology, 2021, 47, 61-72.	3.2	18
3	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	8.1	56
4	Value of systematic genetic screening of patients with amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 510-518.	1.9	69
5	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	9.0	46
6	Amyotrophic lateral sclerosis transcriptomics reveals immunological effects of low-dose interleukin-2. Brain Communications, 2021, 3, fcab141.	3.3	17
7	Neuropathological characterization of a novel TANK binding kinase (TBK1) gene loss of function mutation associated with amyotrophic lateral sclerosis. Neuropathology and Applied Neurobiology, 2020, 46, 279-291.	3.2	12
8	Rare Variant Burden Analysis within Enhancers Identifies CAV1 as an ALS Risk Gene. Cell Reports, 2020, 33, 108456.	6.4	24
9	Repeated 5-day cycles of low dose aldesleukin in amyotrophic lateral sclerosis (IMODALS): A phase 2a randomised, double-blind, placebo-controlled trial. EBioMedicine, 2020, 59, 102844.	6.1	41
10	Oligodendrocyte pathology exceeds axonal pathology in white matter in human amyotrophic lateral sclerosis. Journal of Pathology, 2020, 251, 262-271.	4.5	37
11	The involvement of regulatory T cells in amyotrophic lateral sclerosis and their therapeutic potential. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 435-444.	1.7	19
12	Multifaceted Genes in Amyotrophic Lateral Sclerosis-Frontotemporal Dementia. Frontiers in Neuroscience, 2020, 14, 684.	2.8	32
13	The Association between Polygenic Hazard and Markers of Alzheimerâ€™s Disease Following Stratification for APOE Genotype. Current Alzheimer Research, 2020, 17, 667-679.	1.4	2
14	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. Annals of Neurology, 2019, 85, 470-481.	5.3	118
15	Mutations in the Glycosyltransferase Domain of GLT8D1 Are Associated with Familial Amyotrophic Lateral Sclerosis. Cell Reports, 2019, 26, 2298-2306.e5.	6.4	57
16	Striking phenotypic variation in a family with the P506S UBQLN2 mutation including amyotrophic lateral sclerosis, spastic paraplegia, and frontotemporal dementia. Neurobiology of Aging, 2019, 73, 229.e5-229.e9.	3.1	16
17	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517
18	ALS-associated missense and nonsense TBK1 mutations can both cause loss of kinase function. Neurobiology of Aging, 2018, 71, 266.e1-266.e10.	3.1	59

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19	Children's experiences following a CBT intervention to reduce dental anxiety: one year on. British Dental Journal, 2018, 225, 247-251.	0.6	12
20	Serum miRNAs miR-206, 143-3p and 374b-5p as potential biomarkers for amyotrophic lateral sclerosis (ALS). Neurobiology of Aging, 2017, 55, 123-131.	3.1	117
21	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, .	12.4	129
22	A data-driven approach links microglia to pathology and prognosis in amyotrophic lateral sclerosis. Acta Neuropathologica Communications, 2017, 5, 23.	5.2	63
23	SRSF1-dependent nuclear export inhibition of C9ORF72 repeat transcripts prevents neurodegeneration and associated motor deficits. Nature Communications, 2017, 8, 16063.	12.8	106
24	Development and Testing of a Cognitive Behavioral Therapy Resource for Childrenâ€™s Dental Anxiety. JDR Clinical and Translational Research, 2017, 2, 23-37.	1.9	34
25	Targeted Genetic Screen in Amyotrophic Lateral Sclerosis Reveals Novel Genetic Variants with Synergistic Effect on Clinical Phenotype. Frontiers in Molecular Neuroscience, 2017, 10, 370.	2.9	24
26	C9ORF72 hexanucleotide repeat exerts toxicity in a stable, inducible motor neuronal cell model, which is rescued by partial depletion of Pten. Human Molecular Genetics, 2017, 26, 1133-1145.	2.9	23
27	Small RNA Sequencing of Sporadic Amyotrophic Lateral Sclerosis Cerebrospinal Fluid Reveals Differentially Expressed miRNAs Related to Neural and Glial Activity. Frontiers in Neuroscience, 2017, 11, 731.	2.8	83
28	The genetics of amyotrophic lateral sclerosis: current insights. Degenerative Neurological and Neuromuscular Disease, 2016, 6, 49.	1.3	65
29	Oligogenic inheritance of optineurin (OPTN) and C9ORF72 mutations in ALS highlights localisation of OPTN in the TDPâ€43â€negative inclusions of C9ORF72â€ALS. Neuropathology, 2016, 36, 125-134.	1.2	35
30	Motor neurone disease/amyotrophic lateral sclerosis associated with intermediateâ€length <scp>CAG</scp> repeat expansions in <scp>Ataxinâ€2</scp> does not have 1<scp>C</scp>2â€positive polyglutamine inclusions. Neuropathology and Applied Neurobiology, 2016, 42, 377-389.	3.2	7
31	Multicenter validation of CSF neurofilaments as diagnostic biomarkers for ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2016, 17, 404-413.	1.7	84
32	Rare genetic variation in UNC13A may modify survival in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2016, 17, 593-599.	1.7	22
33	Common Themes in the Pathogenesis of Neurodegeneration. , 2016, , 1-12.		4
34	Lysosomal and phagocytic activity is increased in astrocytes during disease progression in the SOD1 G93A mouse model of amyotrophic lateral sclerosis. Frontiers in Cellular Neuroscience, 2015, 9, 410.	3.7	36
35	C9ORF72 GGGGCC Expanded Repeats Produce Splicing Dysregulation which Correlates with Disease Severity in Amyotrophic Lateral Sclerosis. PLoS ONE, 2015, 10, e0127376.	2.5	83
36	The Spectrum of C9orf72-mediated Neurodegeneration and Amyotrophic Lateral Sclerosis. Neurotherapeutics, 2015, 12, 326-339.	4.4	46

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37	Intermediate length C9orf72 expansion in an ALS patient without classical C9orf72 neuropathology. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2015, 16, 249-251.	1.7	8
38	Stratified gene expression analysis identifies major amyotrophic lateral sclerosis genes. Neurobiology of Aging, 2015, 36, 2006.e1-2006.e9.	3.1	22
39	Antisense RNA foci in the motor neurons of C9ORF72-ALS patients are associated with TDP-43 proteinopathy. Acta Neuropathologica, 2015, 130, 63-75.	7.7	149
40	The CHCHD10 P34S variant is not associated with ALS in a UK cohort of familial and sporadic patients. Neurobiology of Aging, 2015, 36, 2908.e17-2908.e18.	3.1	19
41	Gene expression signatures in motor neurone disease fibroblasts reveal dysregulation of metabolism, hypoxiaâ€response and <scp>RNA</scp> processing functions. Neuropathology and Applied Neurobiology, 2015, 41, 201-226.	3.2	73
42	Invited Review: Decoding the pathophysiological mechanisms that underlie <scp>RNA</scp> dysregulation in neurodegenerative disorders: a review of the current state of the art. Neuropathology and Applied Neurobiology, 2015, 41, 109-134.	3.2	47
43	Sequestration of multiple RNA recognition motif-containing proteins by C9orf72 repeat expansions. Brain, 2014, 137, 2040-2051.	7.6	253
44	AMYOTROPHIC LATERAL SCLEROSIS ASSOCIATED WITH AN INTERMEDIATE LENGTH GGGGCC REPEAT EXPANSION HAS DISTINCT NEUROPATHOLOGY COMPARED TO PATIENTS WITH LARGER EXPANSIONS. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, e4.130-e4.	1.9	0
45	The widening spectrum of C9ORF72-related disease; genotype/phenotype correlations and potential modifiers of clinical phenotype. Acta Neuropathologica, 2014, 127, 333-345.	7.7	150
46	TMEM106B is a genetic modifier of frontotemporal lobar degeneration with C9orf72 hexanucleotide repeat expansions. Acta Neuropathologica, 2014, 127, 407-418.	7.7	123
47	Loss of nuclear <scp>TDP</scp>â€43 in amyotrophic lateral sclerosis (<scp>ALS</scp>) causes altered expression of splicing machinery and widespread dysregulation of <scp>RNA</scp> splicing in motor neurones. Neuropathology and Applied Neurobiology, 2014, 40, 670-685.	3.2	98
48	Multicentre quality control evaluation of different biomarker candidates for amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2014, 15, 344-350.	1.7	62
49	Comparison of Blood RNA Extraction Methods Used for Gene Expression Profiling in Amyotrophic Lateral Sclerosis. PLoS ONE, 2014, 9, e87508.	2.5	25
50	Simultaneous and independent detection of C9ORF72 alleles with low and high number of GGGGCC repeats using an optimised protocol of Southern blot hybridisation. Molecular Neurodegeneration, 2013, 8, 12.	10.8	52
51	Neurodegeneration caused by intronic expansions of C9ORF72 is a clinically heterogeneous but pathologically distinct disease. Lancet, The, 2013, 381, S32.	13.7	1
52	Unravelling the enigma of selective vulnerability in neurodegeneration: motor neurons resistant to degeneration in ALS show distinct gene expression characteristics and decreased susceptibility to excitotoxicity. Acta Neuropathologica, 2013, 125, 95-109.	7.7	133
53	S[+] Apomorphine is a CNS penetrating activator of the Nrf2-ARE pathway with activity in mouse and patient fibroblast models of amyotrophic lateral sclerosis. Free Radical Biology and Medicine, 2013, 61, 438-452.	2.9	54
54	C9ORF72 transcription in a frontotemporal dementia case with two expanded alleles. Neurology, 2013, 81, 1719-1721.	1.1	25

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55	The C9ORF72 expansion mutation is a common cause of ALS+/â”FTD in Europe and has a single founder. European Journal of Human Genetics, 2013, 21, 102-108.	2.8	201
56	Concurrence of multiple sclerosis and amyotrophic lateral sclerosis in patients with hexanucleotide repeat expansions ofC9ORF72. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 79-87.	1.9	57
57	ALS-associated mutations in FUS disrupt the axonal distribution and function of SMN. Human Molecular Genetics, 2013, 22, 3690-3704.	2.9	130
58	C9ORF72 expansions, parkinsonism, and Parkinson disease. Neurology, 2013, 81, 808-811.	1.1	57
59	Lack of unique neuropathology in amyotrophic lateral sclerosis associated with <scp>p.K54E</scp> angiogenin (<scp>ANG</scp>) mutation. Neuropathology and Applied Neurobiology, 2013, 39, 562-571.	3.2	14
60	Investigating cell death mechanisms in amyotrophic lateral sclerosis using transcriptomics. Frontiers in Cellular Neuroscience, 2013, 7, 259.	3.7	23
61	Neuronal dark matter: the emerging role of microRNAs in neurodegeneration. Frontiers in Cellular Neuroscience, 2013, 7, 178.	3.7	167
62	159â€…Dysregulation of the cross-talk with astrocytes as a contributory factor to motor neuron injury in motor neuron disease. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, e1.115-e1.	1.9	0
63	Clinico-pathological features in amyotrophic lateral sclerosis with expansions in C9ORF72. Brain, 2012, 135, 751-764.	7.6	293
64	Gene expression profiling in human neurodegenerative disease. Nature Reviews Neurology, 2012, 8, 518-530.	10.1	183
65	Genetics of Familial Amyotrophic Lateral Sclerosis. , 2012, , .		0
66	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
67	HFEH63D, C282Y andAGTR1A1166C Polymorphisms and Brain White Matter Lesions in the Aging Brain. Journal of Neurogenetics, 2011, 25, 7-14.	1.4	10
68	Molecular pathways of motor neuron injury in amyotrophic lateral sclerosis. Nature Reviews Neurology, 2011, 7, 616-630.	10.1	512
69	Molecular pathology and genetic advances in amyotrophic lateral sclerosis: an emerging molecular pathway and the significance of glial pathology. Acta Neuropathologica, 2011, 122, 657-671.	7.7	134
70	Brain Iron Dysregulation and the Risk of Ageing White Matter Lesions. NeuroMolecular Medicine, 2011, 13, 289-299.	3.4	18
71	Phosphatase and tensin homologue/protein kinase B pathway linked to motor neuron survival in human superoxide dismutase 1-related amyotrophic lateral sclerosis. Brain, 2011, 134, 506-517.	7.6	71
72	Dysregulation of astrocyteâ€“motoneuron cross-talk in mutant superoxide dismutase 1-related amyotrophic lateral sclerosis. Brain, 2011, 134, 2627-2641.	7.6	176

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73	Broad clinical phenotypes associated with TAR-DNA binding protein (TARDBP) mutations in amyotrophic lateral sclerosis. Neurogenetics, 2010, 11, 217-225.	1.4	79
74	Novel FUS/TLS Mutations and Pathology in Familial and Sporadic Amyotrophic Lateral Sclerosis. Archives of Neurology, 2010, 67, 455-61.	4.5	113
75	Mutations in CHMP2B in Lower Motor Neuron Predominant Amyotrophic Lateral Sclerosis (ALS). PLoS ONE, 2010, 5, e9872.	2.5	204
76	New pedigrees and novel mutation expand the phenotype of REEP1-associated hereditary spastic paraplegia (HSP). Neurogenetics, 2009, 10, 105-110.	1.4	42
77	Transcriptional response of the neuromuscular system to exercise training and potential implications for ALS. Journal of Neurochemistry, 2009, 109, 1714-1724.	3.9	37
78	HSP60 IS A RARE CAUSE OF HEREDITARY SPASTIC PARAPARESIS, BUT MAY ACT AS A GENETIC MODIFIER. Neurology, 2008, 70, 1717-1718.	1.1	21
79	Gene Expression Assays. Advances in Clinical Chemistry, 2007, 44, 247-292.	3.7	32
80	Microarray Analysis of the Cellular Pathways Involved in the Adaptation to and Progression of Motor Neuron Injury in the SOD1 G93A Mouse Model of Familial ALS. Journal of Neuroscience, 2007, 27, 9201-9219.	3.6	179
81	MUTATIONS IN VAPB ARE NOT ASSOCIATED WITH SPORADIC ALS. Neurology, 2007, 68, 1951-1953.	1.1	10
82	Pathological TDPâ€43 distinguishes sporadic amyotrophic lateral sclerosis from amyotrophic lateral sclerosis with SOD1 mutations. Annals of Neurology, 2007, 61, 427-434.	5.3	840
83	Impairment of mitochondrial anti-oxidant defence in SOD1-related motor neuron injury and amelioration by ebselen. Brain, 2006, 129, 1693-1709.	7.6	57
84	Clinical features of hereditary spastic paraplegia due to spastin mutation. Neurology, 2006, 67, 45-51.	1.1	118
85	Screening of the regulatory and coding regions of vascular endothelial growth factor in amyotrophic lateral sclerosis. Neurogenetics, 2005, 6, 101-104.	1.4	15
86	Mutant SOD1 alters the motor neuronal transcriptome: implications for familial ALS. Brain, 2005, 128, 1686-1706.	7.6	170
87	Selective loss of neurofilament expression in Cu/Zn superoxide dismutase (SOD1) linked amyotrophic lateral sclerosis. Journal of Neurochemistry, 2004, 82, 1118-1128.	3.9	70
88	No association with common Caucasian genotypes in exons 8, 13 and 14 of the human cytoplasmic dynein heavy chain gene (DNCHC1) and familial motor neuron disorders. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders: Official Publication of the World Federation of Neurology, Research Group on Motor Neuron Diseases, 2003, 4, 150-157.	1.2	26
89	Analysis of the Cytosolic Proteome in a Cell Culture Model of Familial Amyotrophic Lateral Sclerosis Reveals Alterations to the Proteasome, Antioxidant Defenses, and Nitric Oxide Synthetic Pathways. Journal of Biological Chemistry, 2003, 278, 6371-6383.	3.4	103
90	Differential gene expression in a cell culture model of SOD1-related familial motor neurone disease. Human Molecular Genetics, 2002, 11, 2061-2075.	2.9	31

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91	Mutation screening of manganese superoxide dismutase in amyotrophic lateral sclerosis. NeuroReport, 2001, 12, 2319-2322.	1.2	32
92	Machine perfusion for kidneys: how to do it at minimal cost. Transplant International, 2001, 14, 103-107.	1.6	22
93	Prognostic Factors for Relapse and Pelvic Lymph Node Metastases in Early Stage I Adenocarcinoma of the Cervix. Gynecologic Oncology, 1999, 74, 423-427.	1.4	32
94	Insights Arising from Gene Expression Profiling in Amyotrophic Lateral Sclerosis. , 0, , .		0
95	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. SSRN Electronic Journal, 0, , .	0.4	4