Pamela J Shaw

List of Publications by Year
in descending order

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322 papers	26,361 citations	6254 80 h-index	8866 145 g-index
334 all docs	334 docs citations	334 times ranked	22903 citing authors

#	Article	IF	Citations
1	Effects of non-invasive ventilation on survival and quality of life in patients with amyotrophic lateral sclerosis: a randomised controlled trial. Lancet Neurology, The, 2006, 5, 140-147.	10.2	922
2	Amyotrophic lateral sclerosis. Nature Reviews Disease Primers, 2017, 3, 17071.	30.5	885
3	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
4	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517
5	Oxidative stress in ALS: Key role in motor neuron injury and therapeutic target. Free Radical Biology and Medicine, 2010, 48, 629-641.	2.9	512
6	Molecular pathways of motor neuron injury in amyotrophic lateral sclerosis. Nature Reviews Neurology, 2011, 7, 616-630.	10.1	512
7	White Matter Lesions in an Unselected Cohort of the Elderly. Stroke, 2006, 37, 1391-1398.	2.0	495
8	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
9	Common variants at 7p21 are associated with frontotemporal lobar degeneration with TDP-43 inclusions. Nature Genetics, 2010, 42, 234-239.	21.4	479
10	Controversies and priorities in amyotrophic lateral sclerosis. Lancet Neurology, The, 2013, 12, 310-322.	10.2	454
11	Oxidative stress in ALS: A mechanism of neurodegeneration and a therapeutic target. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2006, 1762, 1051-1067.	3.8	382
12	Familial amyotrophic lateral sclerosis-linked SOD1 mutants perturb fast axonal transport to reduce axonal mitochondria content. Human Molecular Genetics, 2007, 16, 2720-2728.	2.9	365
13	The role of mitochondria in amyotrophic lateral sclerosis. Neuroscience Letters, 2019, 710, 132933.	2.1	356
14	Phase 1â€“2 Trial of Antisense Oligonucleotide Tofersen for SOD1 ALS. New England Journal of Medicine, 2020, 383, 109-119.	27.0	354
15	Prognosis for patients with amyotrophic lateral sclerosis: development and validation of a personalised prediction model. Lancet Neurology, The, 2018, 17, 423-433.	10.2	342
16	The C9orf72 protein interacts with Rab1a and the <scp>ULK</scp>1 complex to regulate initiation of autophagy. EMBO Journal, 2016, 35, 1656-1676.	7.8	327
17	Oxidative damage to protein in sporadic motor neuron disease spinal cord. Annals of Neurology, 1995, 38, 691-695.	5.3	312
18	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	8.1	308

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19	Direct conversion of patient fibroblasts demonstrates non-cell autonomous toxicity of astrocytes to motor neurons in familial and sporadic ALS. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 829-832.	7.1	296
20	Clinico-pathological features in amyotrophic lateral sclerosis with expansions in C9ORF72. Brain, 2012, 135, 751-764.	7.6	293
21	Mitochondrial enzyme activity in amyotrophic lateral sclerosis: Implications for the role of mitochondria in neuronal cell death. Annals of Neurology, 1999, 46, 787-790.	5.3	292
22	Update on the glutamatergic neurotransmitter system and the role of excitotoxicity in amyotrophic lateral sclerosis. Muscle and Nerve, 2002, 26, 438-458.	2.2	281
23	Detection of long repeat expansions from PCR-free whole-genome sequence data. Genome Research, 2017, 27, 1895-1903.	5.5	277
24	Sequestration of multiple RNA recognition motif-containing proteins by C9orf72 repeat expansions. Brain, 2014, 137, 2040-2051.	7.6	253
25	Mitochondrial dysfunction in a cell culture model of familial amyotrophic lateral sclerosis. Brain, 2002, 125, 1522-1533.	7.6	249
26	Systemic Delivery of scAAV9 Expressing SMN Prolongs Survival in a Model of Spinal Muscular Atrophy. Science Translational Medicine, 2010, 2, 35ra42.	12.4	246
27	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
28	CSF and Plasma Amino Acid Levels in Motor Neuron Disease: Elevation of CSF Glutamate in a Subset of Patients. Experimental Neurology, 1995, 4, 209-216.	1.7	221
29	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	21.4	218
30	Chromosome 9p21 in sporadic amyotrophic lateral sclerosis in the UK and seven other countries: a genome-wide association study. Lancet Neurology, The, 2010, 9, 986-994.	10.2	205
31	Mutations in CHMP2B in Lower Motor Neuron Predominant Amyotrophic Lateral Sclerosis (ALS). PLoS ONE, 2010, 5, e9872.	2.5	204
32	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
33	Oxidative Stress and Motor Neurone Disease. Brain Pathology, 1999, 9, 165-186.	4.1	191
34	Microglia as potential contributors to motor neuron injury in amyotrophic lateral sclerosis. Glia, 2005, 51, 241-253.	4.9	185
35	Gene expression profiling in human neurodegenerative disease. Nature Reviews Neurology, 2012, 8, 518-530.	10.1	183
36	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

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37	Dysregulation of astrocyteâ€“motoneuron cross-talk in mutant superoxide dismutase 1-related amyotrophic lateral sclerosis. Brain, 2011, 134, 2627-2641.	7.6	176
38	Mitochondrial involvement in amyotrophic lateral sclerosis. Neurochemistry International, 2002, 40, 543-551.	3.8	175
39	Mutant SOD1 alters the motor neuronal transcriptome: implications for familial ALS. Brain, 2005, 128, 1686-1706.	7.6	170
40	Diagnosis and management of motor neurone disease. BMJ: British Medical Journal, 2008, 336, 658-662.	2.3	167
41	Neuronal dark matter: the emerging role of microRNAs in neurodegeneration. Frontiers in Cellular Neuroscience, 2013, 7, 178.	3.7	167
42	Microarray analysis of the astrocyte transcriptome in the aging brain: relationship to Alzheimer's pathology and APOE genotype. Neurobiology of Aging, 2011, 32, 1795-1807.	3.1	166
43	Novel insertion in the KSP region of the neurofilament heavy gene in amyotrophic lateral sclerosis (ALS). NeuroReport, 1998, 9, 3967-3970.	1.2	157
44	Astrocyte function and role in motor neuron disease: A future therapeutic target?. Glia, 2009, 57, 1251-1264.	4.9	156
45	The role of TREM2 R47H as a risk factor for Alzheimer's disease, frontotemporal lobar degeneration, amyotrophic lateral sclerosis, and Parkinson's disease. Alzheimer's and Dementia, 2015, 11, 1407-1416.	0.8	152
46	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
47	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
48	Oligodendrocytes contribute to motor neuron death in ALS via SOD1-dependent mechanism. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E6496-E6505.	7.1	139
49	The microtubule-severing protein Spastin is essential for axon outgrowth in the zebrafish embryo. Human Molecular Genetics, 2006, 15, 2763-2771.	2.9	138
50	C9orf72 expansion disrupts ATM-mediated chromosomal break repair. Nature Neuroscience, 2017, 20, 1225-1235.	14.8	138
51	Direct evidence for axonal transport defects in a novel mouse model of mutant spastinâ€induced hereditary spastic paraplegia (HSP) and human HSP patients. Journal of Neurochemistry, 2009, 110, 34-44.	3.9	135
52	Mechanisms, models and biomarkers in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 19-32.	1.7	135
53	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
54	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

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55	ALS-associated mutations in FUS disrupt the axonal distribution and function of SMN. Human Molecular Genetics, 2013, 22, 3690-3704.	2.9	130
56	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, .	12.4	129
57	Amyotrophic Lateral Sclerosis Associated with Genetic Abnormalities in the Gene Encoding Cu/Zn Superoxide Dismutase: Molecular Pathology of Five New Cases, and Comparison with Previous Reports and 73 Sporadic Cases of ALS. Journal of Neuropathology and Experimental Neurology, 1998, 57, 895-904.	1.7	124
58	TMEM106B is a genetic modifier of frontotemporal lobar degeneration with C9orf72 hexanucleotide repeat expansions. Acta Neuropathologica, 2014, 127, 407-418.	7.7	123
59	A genome-wide association meta-analysis identifies a novel locus at 17q11.2 associated with sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2014, 23, 2220-2231.	2.9	123
60	The RNA of the glutamate transporter EAAT2 is variably spliced in amyotrophic lateral sclerosis and normal individuals. Journal of the Neurological Sciences, 1999, 170, 45-50.	0.6	121
61	Familial amyotrophic lateral sclerosis with a mutation in exon 4 of the Cu/Zn superoxide dismutase gene: pathological and immunocytochemical changes. Acta Neuropathologica, 1996, 92, 395-403.	7.7	120
62	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. Annals of Neurology, 2019, 85, 470-481.	5.3	118
63	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
64	The chromosome 9 ALS and FTD locus is probably derived from a single founder. Neurobiology of Aging, 2012, 33, 209.e3-209.e8.	3.1	115
65	Hereditary spastic paraparesis: Disrupted intracellular transport associated with spastin mutation. Annals of Neurology, 2003, 54, 748-759.	5.3	114
66	Novel FUS/TLS Mutations and Pathology in Familial and Sporadic Amyotrophic Lateral Sclerosis. Archives of Neurology, 2010, 67, 455-61.	4.5	113
67	The expression of the glial glutamate transporter protein EAAT2 in motor neuron disease: an immunohistochemical study. European Journal of Neuroscience, 1998, 10, 2481-2489.	2.6	111
68	Expression of the glial glutamate transporter EAAT2 in the human CNS: an immunohistochemical study. Molecular Brain Research, 1997, 52, 17-31.	2.3	110
69	SRSF1-dependent nuclear export inhibition of C9ORF72 repeat transcripts prevents neurodegeneration and associated motor deficits. Nature Communications, 2017, 8, 16063.	12.8	106
70	Production of monocyte chemoattractant proteinâ€1 in amyotrophic lateral sclerosis. Muscle and Nerve, 2005, 32, 541-544.	2.2	104
71	Exome sequencing in amyotrophic lateral sclerosis implicates a novel gene, DNAJC7, encoding a heat-shock protein. Nature Neuroscience, 2019, 22, 1966-1974.	14.8	101
72	The expression of neuronal voltage-dependent calcium channels in human cerebellum. Molecular Brain Research, 1995, 34, 271-282.	2.3	100

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73	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
74	Development and Characterisation of a Glutamate-Sensitive Motor Neurone Cell Line. Journal of Neurochemistry, 2008, 74, 1895-1902.	3.9	97
75	Rasch analysis of the hospital anxiety and depression scale (hads) for use in motor neurone disease. Health and Quality of Life Outcomes, 2011, 9, 82.	2.4	96
76	Peroxynitrite and Hydrogen Peroxide Induced Cell Death in the NSC34 Neuroblastoma Ã— Spinal Cord Cell Line: Role of Poly(ADPâ€Ribose) Polymerase. Journal of Neurochemistry, 1998, 70, 501-508.	3.9	91
77	The Cellular and Molecular Pathology of the Motor System in Hereditary Spastic Paraparesis due to Mutation of the Spastin Gene. Journal of Neuropathology and Experimental Neurology, 2003, 62, 1166-1177.	1.7	91
78	<scp>C9orf72</scp> and <scp>UNC13A</scp> are shared risk loci for amyotrophic lateral sclerosis and frontotemporal dementia: A genomeâ€wide metaâ€analysis. Annals of Neurology, 2014, 76, 120-133.	5.3	91
79	Alterations in the blood brain barrier in ageing cerebral cortex in relationship to Alzheimer-type pathology: A study in the MRC-CFAS population neuropathology cohort. Neuroscience Letters, 2011, 505, 25-30.	2.1	90
80	Expression of Vascular Endothelial Growth Factor and Its Receptors in the Central Nervous System in Amyotrophic Lateral Sclerosis. Journal of Neuropathology and Experimental Neurology, 2006, 65, 26-36.	1.7	87
81	Biomarkers in Motor Neuron Disease: A State of the Art Review. Frontiers in Neurology, 2019, 10, 291.	2.4	87
82	Excitotoxicity and motor neurone disease: A review of the evidence. Journal of the Neurological Sciences, 1994, 124, 6-13.	0.6	86
83	An in vitro screening cascade to identify neuroprotective antioxidants in ALS. Free Radical Biology and Medicine, 2009, 46, 1127-1138.	2.9	86
84	Astrocyte adenosine deaminase loss increases motor neuron toxicity in amyotrophic lateral sclerosis. Brain, 2019, 142, 586-605.	7.6	84
85	C9ORF72 GGGGCC Expanded Repeats Produce Splicing Dysregulation which Correlates with Disease Severity in Amyotrophic Lateral Sclerosis. PLoS ONE, 2015, 10, e0127376.	2.5	83
86	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
87	Physical activity as an exogenous risk factor in motor neuron disease (MND): A review of the evidence. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2009, 10, 191-204.	2.1	82
88	Early interneuron dysfunction in ALS: Insights from a mutant sod1 zebrafish model. Annals of Neurology, 2013, 73, 246-258.	5.3	82
89	Meta-analysis of pharmacogenetic interactions in amyotrophic lateral sclerosis clinical trials. Neurology, 2017, 89, 1915-1922.	1.1	82
90	Transcriptomic indices of fast and slow disease progression in two mouse models of amyotrophic lateral sclerosis. Brain, 2013, 136, 3305-3332.	7.6	81

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91	Microarray RNA Expression Analysis of Cerebral White Matter Lesions Reveals Changes in Multiple Functional Pathways. Stroke, 2009, 40, 369-375.	2.0	80
92	Optimised and Rapid Pre-clinical Screening in the SOD1G93A Transgenic Mouse Model of Amyotrophic Lateral Sclerosis (ALS). PLoS ONE, 2011, 6, e23244.	2.5	80
93	Broad clinical phenotypes associated with TAR-DNA binding protein (TARDBP) mutations in amyotrophic lateral sclerosis. Neurogenetics, 2010, 11, 217-225.	1.4	79
94	Differential Localization of Voltage-Dependent Calcium Channel Î±₁Subunits at the Human and Rat Neuromuscular Junction. Journal of Neuroscience, 1997, 17, 6226-6235.	3.6	78
95	Apoptosis in amyotrophic lateral sclerosisâ€"what is the evidence?. Lancet Neurology, The, 2005, 4, 500-509.	10.2	78
96	Superoxide dismutase 1 mutation in a cellular model of amyotrophic lateral sclerosis shifts energy generation from oxidative phosphorylation to glycolysis. Neurobiology of Aging, 2014, 35, 1499-1509.	3.1	77
97	The quantitative autoradiographic distribution of [3H]MK-801 binding sites in the normal human spinal cord. Brain Research, 1991, 539, 164-168.	2.2	76
98	Use of clinical staging in amyotrophic lateral sclerosis for phase 3 clinical trials. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 45-49.	1.9	75
99	C9orf72 intermediate expansions of 24â€"30 repeats are associated with ALS. Acta Neuropathologica Communications, 2019, 7, 115.	5.2	75
100	Non-invasive ventilation in motor neuron disease: an update of current UK practice. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 371-376.	1.9	74
101	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
102	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
103	A comprehensive analysis of rare genetic variation in amyotrophic lateral sclerosis in the UK. Brain, 2017, 140, 1611-1618.	7.6	71
104	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
105	The natural history of motor neuron disease: Assessing the impact of specialist care. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 13-19.	1.7	70
106	Value of systematic genetic screening of patients with amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 510-518.	1.9	69
107	Alterations of the bloodâ€"brain barrier in cerebral white matter lesions in the ageing brain. Neuroscience Letters, 2010, 486, 246-251.	2.1	68
108	[3H]d-aspartate binding sites in the normal human spinal cord and changes in motor neuron disease: a quantitative autoradiographic study. Brain Research, 1994, 655, 195-201.	2.2	66

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109	Oxidative Damage and Motor Neurone Disease Difficulties in the Measurement of Protein Carbonyls in Human Brain Tissue. Free Radical Research, 1996, 24, 397-406.	3.3	65
110	Physical exercise is a risk factor for amyotrophic lateral sclerosis: Convergent evidence from Mendelian randomisation, transcriptomics and risk genotypes. EBioMedicine, 2021, 68, 103397.	6.1	65
111	Differences in protein quality control correlate with phenotype variability in 2 mouse models of familial amyotrophic lateral sclerosis. Neurobiology of Aging, 2015, 36, 492-504.	3.1	63
112	Immune response in peripheral axons delays disease progression in SOD1G93A mice. Journal of Neuroinflammation, 2016, 13, 261.	7.2	63
113	A data-driven approach links microglia to pathology and prognosis in amyotrophic lateral sclerosis. Acta Neuropathologica Communications, 2017, 5, 23.	5.2	63
114	Ursodeoxycholic Acid Improves Mitochondrial Function and Redistributes Drp1 in Fibroblasts from Patients with Either Sporadic or Familial Alzheimer's Disease. Journal of Molecular Biology, 2018, 430, 3942-3953.	4.2	63
115	Protein Homeostasis in Amyotrophic Lateral Sclerosis: Therapeutic Opportunities?. Frontiers in Molecular Neuroscience, 2017, 10, 123.	2.9	62
116	Mitochondrial Dysfunction in Alzheimerâ€™s Disease: A Biomarker of the Future?. Biomedicines, 2021, 9, 63.	3.2	62
117	Glial Proliferation and Metabotropic Glutamate Receptor Expression in Amyotrophic Lateral Sclerosis. Journal of Neuropathology and Experimental Neurology, 2004, 63, 831-840.	1.7	60
118	C9orf72 expansion within astrocytes reduces metabolic flexibility in amyotrophic lateral sclerosis. Brain, 2019, 142, 3771-3790.	7.6	59
119	Pattern of spread and prognosis in lower limb-onset ALS. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2010, 11, 369-373.	2.1	58
120	The nuclear retention of transcription factor FOXO3a correlates with a DNA damage response and increased glutamine synthetase expression by astrocytes suggesting a neuroprotective role in the ageing brain. Neuroscience Letters, 2015, 609, 11-17.	2.1	58
121	Comparison of the Kingâ€™s and MiToS staging systems for ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2017, 18, 227-232.	1.7	58
122	Disrupted glycosylation of lipids and proteins is a cause of neurodegeneration. Brain, 2020, 143, 1332-1340.	7.6	58
123	Impairment of mitochondrial anti-oxidant defence in SOD1-related motor neuron injury and amelioration by ebselen. Brain, 2006, 129, 1693-1709.	7.6	57
124	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
125	C9ORF72 expansions, parkinsonism, and Parkinson disease. Neurology, 2013, 81, 808-811.	1.1	57
126	Association of a Locus in theCAMTA1Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. JAMA Neurology, 2016, 73, 812.	9.0	57

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127	Mutations in the Glycosyltransferase Domain of GLT8D1 Are Associated with Familial Amyotrophic Lateral Sclerosis. Cell Reports, 2019, 26, 2298-2306.e5.	6.4	57
128	Gastrostomy use in motor neurone disease (MND): A review, meta-analysis and survey of current practice. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 96-104.	1.7	55
129	Motor neuron disease in a patient with a mitochondrial tRNAIlemutation. Annals of Neurology, 2006, 59, 570-574.	5.3	54
130	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
131	A new zebrafish model produced by TILLING of SOD1-related amyotrophic lateral sclerosis replicates key features of the disease and represents a tool for in vivo therapeutic screening. DMM Disease Models and Mechanisms, 2014, 7, 73-81.	2.4	53
132	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
133	The gut microbiome: a key player in the complexity of amyotrophic lateral sclerosis (ALS). BMC Medicine, 2021, 19, 13.	5.5	52
134	Low expression of GluR2 AMPA receptor subunit protein by human motor neurons. NeuroReport, 1999, 10, 261-265.	1.2	51
135	Advances, challenges and future directions for stem cell therapy in amyotrophic lateral sclerosis. Molecular Neurodegeneration, 2017, 12, 85.	10.8	51
136	Genome-wide identification of the genetic basis of amyotrophic lateral sclerosis. Neuron, 2022, 110, 992-1008.e11.	8.1	51
137	Motor neurone disease: a practical update on diagnosis and management. Clinical Medicine, 2010, 10, 252-258.	1.9	50
138	Meta-analysis of genome-wide DNA methylation identifies shared associations across neurodegenerative disorders. Genome Biology, 2021, 22, 90.	8.8	49
139	Cu/Zn superoxide dismutase (SOD1) mutations associated with familial amyotrophic lateral sclerosis (ALS) affect cellular free radical release in the presence of oxidative stress. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders: Official Publication of the World Federation of Neurology, Research Group on Motor Neuron Diseases, 2002, 3, 75-85.	1.2	48
140	The use of non-invasive ventilation at end of life in patients with motor neurone disease: A qualitative exploration of family carer and health professional experiences. Palliative Medicine, 2013, 27, 516-523.	3.1	47
141	PTEN Depletion Decreases Disease Severity and Modestly Prolongs Survival in a Mouse Model of Spinal Muscular Atrophy. Molecular Therapy, 2015, 23, 270-277.	8.2	47
142	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
143	Stable transgenic C9orf72 zebrafish model key aspects of the ALS/FTD phenotype and reveal novel pathological features. Acta Neuropathologica Communications, 2018, 6, 125.	5.2	47
144	The Spectrum of C9orf72-mediated Neurodegeneration and Amyotrophic Lateral Sclerosis. Neurotherapeutics, 2015, 12, 326-339.	4.4	46

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145	Long-term physical activity: an exogenous risk factor for sporadic amyotrophic lateral sclerosis?. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2016, 17, 377-384.	1.7	46
146	Non-NMDA receptors in motor neuron disease (MND): a quantitative autoradiographic study in spinal cord and motor cortex using [3H]CNQX and [3H]kainate. Brain Research, 1994, 655, 186-194.	2.2	45
147	Nonverbal visual attention, but not recognition memory or learning, processes are impaired in motor neurone disease. Neuropsychologia, 1996, 34, 377-385.	1.6	45
148	A Reduced Astrocyte Response to Î²-Amyloid Plaques in the Ageing Brain Associates with Cognitive Impairment. PLoS ONE, 2015, 10, e0118463.	2.5	45
149	Effect of lipid profile on prognosis in the patients with amyotrophic lateral sclerosis: Insights from the olesoxime clinical trial. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2015, 16, 478-484.	1.7	45
150	Novel genotype-phenotype and MRI correlations in a large cohort of patients with SPG7 mutations. Neurology: Genetics, 2018, 4, e279.	1.9	44
151	Identification of a novel exon 4 SOD1 mutation in a sporadic amyotrophic lateral sclerosis patient. Molecular and Cellular Probes, 1994, 8, 329-330.	2.1	43
152	Factors influencing decision-making in relation to timing of gastrostomy insertion in patients with motor neurone disease. BMJ Supportive and Palliative Care, 2014, 4, 57-63.	1.6	43
153	Quantitative Study of Synaptophysin Immunoreactivity of Cerebral Cortex and Spinal Cord in Motor Neuron Disease. Journal of Neuropathology and Experimental Neurology, 1995, 54, 673-679.	1.7	42
154	The Effect of SOD1 Mutation on Cellular Bioenergetic Profile and Viability in Response to Oxidative Stress and Influence of Mutation-Type. PLoS ONE, 2013, 8, e68256.	2.5	42
155	Distribution of AMPA-selective glutamate receptor subunits in the human hippocampus and cerebellum. Molecular Brain Research, 1995, 31, 17-32.	2.3	41
156	Imaging muscle as a potential biomarker of denervation in motor neuron disease. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 248-255.	1.9	41
157	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
158	A neuronal <scp>DNA</scp> damage response is detected at the earliest stages of <scp>A</scp>lzheimer's neuropathology and correlates with cognitive impairment in the <scp>M</scp>edical <scp>R</scp>esearch <scp>C</scp>ouncil's <scp>C</scp>ognitive <scp>F</scp>unction and <scp>A</scp>geing <scp>S</scp>tudy ageing brain cohort. Neuropathology and Applied Neurobiology, 2015, 41, 483-496.	3.2	40
159	Immunocytochemical study of the distribution of the free radical scavenging enzymes CU/ZN superoxide dismutase (SOD1); MN superoxide dismutase (MN SOD) and catalase in the normal human spinal cord and in motor neuron disease. Journal of the Neurological Sciences, 1997, 147, 115-125.	0.6	39
160	The expression of the glutamate re-uptake transporter excitatory amino acid transporter 1 (EAAT1) in the normal human CNS and in motor neurone disease: an immunohistochemical study. Neuroscience, 2002, 109, 27-44.	2.3	39
161	A preliminary randomized trial of the mechanical insufflator-exsufflator versus breath-stacking technique in patients with amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2015, 16, 448-455.	1.7	39
162	ALS: life and death in a bad neighborhood. Nature Medicine, 2006, 12, 885-887.	30.7	38

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