Christopher E Shaw

List of Publications by Year
in descending order

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339 papers	50,410 citations	1704 104 h-index	1857 209 g-index
354 all docs	354 docs citations	354 times ranked	36280 citing authors

#	Article	IF	Citations
1	Elucidating the Role of Cerebellar Synaptic Dysfunction in C9orf72-ALS/FTD â€" a Systematic Review and Meta-Analysis. Cerebellum, 2022, 21, 681-714.	2.5	3
2	Genome-wide identification of the genetic basis of amyotrophic lateral sclerosis. Neuron, 2022, 110, 992-1008.e11.	8.1	51
3	Structural variation analysis of 6,500 whole genome sequences in amyotrophic lateral sclerosis. Npj Genomic Medicine, 2022, 7, 8.	3.8	23
4	Disruption of ERâ€•mitochondria tethering and signalling in C9orf72â€•associated amyotrophic lateral sclerosis and frontotemporal dementia. Aging Cell, 2022, 21, e13549.	6.7	30
5	Common Genetic Variants Contribute to Risk of Transposition of the Great Arteries. Circulation Research, 2022, 130, 166-180.	4.5	15
6	Multicentre appraisal of amyotrophic lateral sclerosis biofluid biomarkers shows primacy of blood neurofilament light chain. Brain Communications, 2022, 4, fcac029.	3.3	29
7	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
8	Unbiased metabolome screen leads to personalized medicine strategy for amyotrophic lateral sclerosis. Brain Communications, 2022, 4, fcac069.	3.3	10
9	Whole-genome sequencing reveals that variants in the Interleukin 18 Receptor Accessory Protein 3â€²UTR protect against ALS. Nature Neuroscience, 2022, 25, 433-445.	14.8	16
10	Neurotoxic Astrocytes Directly Converted from Sporadic and Familial ALS Patient Fibroblasts Reveal Signature Diversities and miR-146a Theragnostic Potential in Specific Subtypes. Cells, 2022, 11, 1186.	4.1	11
11	Creatine kinase and prognosis in amyotrophic lateral sclerosis: a literature review and multi-centre cohort analysis. Journal of Neurology, 2022, 269, 5395-5404.	3.6	6
12	Simultaneous ALS and SCA2 associated with an intermediate-length ATXN2 CAG-repeat expansion. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, 22, 579-582.	1.7	13
13	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	8.1	56
14	The Effect of <scp>SMN</scp> Gene Dosage on <scp>ALS</scp> Risk and Disease Severity. Annals of Neurology, 2021, 89, 686-697.	5.3	10
15	The gut microbiome: a key player in the complexity of amyotrophic lateral sclerosis (ALS). BMC Medicine, 2021, 19, 13.	5.5	52
16	Value of systematic genetic screening of patients with amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 510-518.	1.9	69
17	Meta-analysis of genome-wide DNA methylation identifies shared associations across neurodegenerative disorders. Genome Biology, 2021, 22, 90.	8.8	49
18	Generation of six induced pluripotent stem cell lines from patients with amyotrophic lateral sclerosis with associated genetic mutations in either FUS or ANXA11. Stem Cell Research, 2021, 52, 102246.	0.7	3

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19	Disease Mechanisms and Therapeutic Approaches in C9orf72 ALS-FTD. Biomedicines, 2021, 9, 601.	3.2	7
20	ALS-linked FUS mutants affect the localization of U7 snRNP and replication-dependent histone gene expression in human cells. Scientific Reports, 2021, 11, 11868.	3.3	7
21	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
22	Regulation of Synapse Weakening through Interactions of the Microtubule Associated Protein Tau with PACSIN1. Journal of Neuroscience, 2021, 41, 7162-7170.	3.6	12
23	Innovating Clinical Trials for Amyotrophic Lateral Sclerosis. Neurology, 2021, 97, 528-536.	1.1	19
24	Cytoplasmic TDP-43 is involved in cell fate during stress recovery. Human Molecular Genetics, 2021, 31, 166-175.	2.9	15
25	SRSF1-dependent inhibition of C9ORF72-repeat RNA nuclear export: genome-wide mechanisms for neuroprotection in amyotrophic lateral sclerosis. Molecular Neurodegeneration, 2021, 16, 53.	10.8	13
26	Demystifying the spontaneous phenomena of motor hyperexcitability. Clinical Neurophysiology, 2021, 132, 1830-1844.	1.5	10
27	Extensive phenotypic characterisation of a human TDP-43Q331KÂtransgenic mouse model of amyotrophic lateral sclerosis (ALS). Scientific Reports, 2021, 11, 16659.	3.3	12
28	Amyotrophic lateral sclerosis alters the metabolic aging profile in patient derived fibroblasts. Neurobiology of Aging, 2021, 105, 64-77.	3.1	16
29	A recessive S174X mutation in Optineurin causes amyotrophic lateral sclerosis through a loss of function via allele-specific nonsense-mediated decay. Neurobiology of Aging, 2021, 106, 1-6.	3.1	3
30	SCFD1 expression quantitative trait loci in amyotrophic lateral sclerosis are differentially expressed. Brain Communications, 2021, 3, fcab236.	3.3	14
31	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
32	Proteinopathies as Hallmarks of Impaired Gene Expression, Proteostasis and Mitochondrial Function in Amyotrophic Lateral Sclerosis. Frontiers in Neuroscience, 2021, 15, 783624.	2.8	13
33	Disrupted glycosylation of lipids and proteins is a cause of neurodegeneration. Brain, 2020, 143, 1332-1340.	7.6	58
34	Mutant C9orf72 human iPSCâ€derived astrocytes cause nonâ€cell autonomous motor neuron pathophysiology. Glia, 2020, 68, 1046-1064.	4.9	90
35	Rare Variant Burden Analysis within Enhancers Identifies CAV1 as an ALS Risk Gene. Cell Reports, 2020, 33, 108456.	6.4	24
36	SOD1-targeting therapies for neurodegenerative diseases: a review of current findings and future potential. Expert Opinion on Orphan Drugs, 2020, 8, 379-392.	0.8	2

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37	The use of biotelemetry to explore disease progression markers in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 563-573.	1.7	12
38	ALS/FTD mutations in UBQLN2 impede autophagy by reducing autophagosome acidification through loss of function. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 15230-15241.	7.1	53
39	CYLD is a causative gene for frontotemporal dementia â€“ amyotrophic lateral sclerosis. Brain, 2020, 143, 783-799.	7.6	62
40	UK case control study of smoking and risk of amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 222-227.	1.7	10
41	Magnetic resonance spectroscopy reveals mitochondrial dysfunction in amyotrophic lateral sclerosis. Brain, 2020, 143, 3603-3618.	7.6	24
42	ATXN1 repeat expansions confer risk for amyotrophic lateral sclerosis and contribute to TDP-43 mislocalization. Brain Communications, 2020, 2, fcaa064.	3.3	33
43	Cross-reactive probes on Illumina DNA methylation arrays: a large study on ALS shows that a cautionary approach is warranted in interpreting epigenome-wide association studies. NAR Genomics and Bioinformatics, 2020, 2, lqaa105.	3.2	13
44	C9orf72 intermediate expansions of 24â€“30 repeats are associated with ALS. Acta Neuropathologica Communications, 2019, 7, 115.	5.2	75
45	RRM adjacent TARDBP mutations disrupt RNA binding and enhance TDP-43 proteinopathy. Brain, 2019, 142, 3753-3770.	7.6	71
46	Astrocyte adenosine deaminase loss increases motor neuron toxicity in amyotrophic lateral sclerosis. Brain, 2019, 142, 586-605.	7.6	84
47	Heterogeneous Nuclear Ribonucleoprotein E2 (hnRNP E2) Is a Component of TDP-43 Aggregates Specifically in the A and C Pathological Subtypes of Frontotemporal Lobar Degeneration. Frontiers in Neuroscience, 2019, 13, 551.	2.8	13
48	Relative preservation of triceps over biceps strength in upper limb-onset ALS: the â€˜split elbowâ€™. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 730-733.	1.9	34
49	Predicting the future of ALS: the impact of demographic change and potential new treatments on the prevalence of ALS in the United Kingdom, 2020â€“2116. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 264-274.	1.7	21
50	Telomere length is greater in ALS than in controls: a whole genome sequencing study. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 229-234.	1.7	18
51	Biomarkers in Motor Neuron Disease: A State of the Art Review. Frontiers in Neurology, 2019, 10, 291.	2.4	87
52	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. Annals of Neurology, 2019, 85, 470-481.	5.3	118
53	Nuclear RNA foci from C9ORF72 expansion mutation form paraspeckle-like bodies. Journal of Cell Science, 2019, 132, .	2.0	36
54	Mutations in the Glycosyltransferase Domain of GLT8D1 Are Associated with Familial Amyotrophic Lateral Sclerosis. Cell Reports, 2019, 26, 2298-2306.e5.	6.4	57

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55	Genetic meta-analysis of diagnosed Alzheimerâ€™s disease identifies new risk loci and implicates AÎ², tau, immunity and lipid processing. Nature Genetics, 2019, 51, 414-430.	21.4	1,962
56	Using telehealth in motor neuron disease to increase access to specialist multidisciplinary care: a UK-based pilot and feasibility study. BMJ Open, 2019, 9, e028525.	1.9	20
57	Process evaluation and exploration of telehealth in motor neuron disease in a UK specialist centre. BMJ Open, 2019, 9, e028526.	1.9	22
58	Critical design considerations for time-to-event endpoints in amyotrophic lateral sclerosis clinical trials. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, jnnp-2019-320998.	1.9	14
59	Human genetics and neuropathology suggest a link between miR-218 and amyotrophic lateral sclerosis pathophysiology. Science Translational Medicine, 2019, 11, .	12.4	37
60	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
61	Association of NIPA1 repeat expansions with amyotrophic lateral sclerosis in a large international cohort. Neurobiology of Aging, 2019, 74, 234.e9-234.e15.	3.1	26
62	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
63	Younger age of onset in familial amyotrophic lateral sclerosis is a result of pathogenic gene variants, rather than ascertainment bias. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 268-271.	1.9	38
64	The role of mitochondria in amyotrophic lateral sclerosis. Neuroscience Letters, 2019, 710, 132933.	2.1	356
65	Objectively Monitoring Amyotrophic Lateral Sclerosis Patient Symptoms During Clinical Trials With Sensors: Observational Study. JMIR MHealth and UHealth, 2019, 7, e13433.	3.7	32
66	Overriding FUS autoregulation in mice triggers gain-of-toxic dysfunctions in RNA metabolism and autophagy-lysosome axis. ELife, 2019, 8, .	6.0	65
67	C9ORF72 repeat expansion causes vulnerability of motor neurons to Ca2+-permeable AMPA receptor-mediated excitotoxicity. Nature Communications, 2018, 9, 347.	12.8	151
68	Mitochondrial abnormalities and disruption of the neuromuscular junction precede the clinical phenotype and motor neuron loss in hFUSWT transgenic mice. Human Molecular Genetics, 2018, 27, 463-474.	2.9	74
69	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
70	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517
71	Reconsidering the causality of TIA1 mutations in ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2018, 19, 1-3.	1.7	22
72	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

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73	A feedback loop between dipeptide-repeat protein, TDP-43 and karyopherin-Î± mediates C9orf72-related neurodegeneration. Brain, 2018, 141, 2908-2924.	7.6	75
74	ALS/FTD-Linked Mutation in FUS Suppresses Intra-axonal Protein Synthesis and Drives Disease Without Nuclear Loss-of-Function of FUS. Neuron, 2018, 100, 816-830.e7.	8.1	185
75	ALS-specific cognitive and behavior changes associated with advancing disease stage in ALS. Neurology, 2018, 91, e1370-e1380.	1.1	170
76	TDP-43 induces p53-mediated cell death of cortical progenitors and immature neurons. Scientific Reports, 2018, 8, 8097.	3.3	38
77	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
78	TDP-43 causes neurotoxicity and cytoskeletal dysfunction in primary cortical neurons. PLoS ONE, 2018, 13, e0196528.	2.5	27
79	Amyotrophic Lateral Sclerosis and Other TDP-43 Proteinopathies. , 2018, , 99-115.		0
80	ATXN2 trinucleotide repeat length correlates with risk of ALS. Neurobiology of Aging, 2017, 51, 178.e1-178.e9.	3.1	86
81	C9orf72 expansion differentially affects males with spinal onset amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 281.1-281.	1.9	33
82	Comparison of the Kingâ€™s and MiToS staging systems for ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2017, 18, 227-232.	1.7	58
83	Non-nuclear Pool of Splicing Factor SFPQ Regulates Axonal Transcripts Required for Normal Motor Development. Neuron, 2017, 94, 322-336.e5.	8.1	61
84	A comprehensive analysis of rare genetic variation in amyotrophic lateral sclerosis in the UK. Brain, 2017, 140, 1611-1618.	7.6	71
85	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, .	12.4	129
86	Amyotrophic lateral sclerosis-like superoxide dismutase 1 proteinopathy is associated with neuronal loss in Parkinsonâ€™s disease brain. Acta Neuropathologica, 2017, 134, 113-127.	7.7	78
87	Viral delivery of C9ORF72 hexanucleotide repeat expansions in mice lead to repeat length dependent neuropathology and behavioral deficits.. DMM Disease Models and Mechanisms, 2017, 10, 859-868.	2.4	25
88	C9orf72 poly GA RAN-translated protein plays a key role in amyotrophic lateral sclerosis via aggregation and toxicity. Human Molecular Genetics, 2017, 26, 4765-4777.	2.9	64
89	Amyotrophic lateral sclerosis. Nature Reviews Disease Primers, 2017, 3, 17071.	30.5	885
90	Meta-analysis of pharmacogenetic interactions in amyotrophic lateral sclerosis clinical trials. Neurology, 2017, 89, 1915-1922.	1.1	82

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91	Can Astrocytes Be a Target for Precision Medicine?. Advances in Experimental Medicine and Biology, 2017, 1007, 111-128.	1.6	7
92	Detection of long repeat expansions from PCR-free whole-genome sequence data. Genome Research, 2017, 27, 1895-1903.	5.5	277
93	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. Nature Genetics, 2017, 49, 1373-1384.	21.4	783
94	The benefit of evolving multidisciplinary care in ALS: a diagnostic cohort survival comparison. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2017, 18, 569-575.	1.7	38
95	SRSF1-dependent nuclear export inhibition of C9ORF72 repeat transcripts prevents neurodegeneration and associated motor deficits. Nature Communications, 2017, 8, 16063.	12.8	106
96	C9ORF72 and UBQLN2 mutations are causes of amyotrophic lateral sclerosis in New Zealand: a genetic and pathologic study using banked human brain tissue. Neurobiology of Aging, 2017, 49, 214.e1-214.e5.	3.1	18
97	RNA Misprocessing in C9orf72-Linked Neurodegeneration. Frontiers in Cellular Neuroscience, 2017, 11, 195.	3.7	32
98	Protein Homeostasis in Amyotrophic Lateral Sclerosis: Therapeutic Opportunities?. Frontiers in Molecular Neuroscience, 2017, 10, 123.	2.9	62
99	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
100	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
101	Advances, challenges and future directions for stem cell therapy in amyotrophic lateral sclerosis. Molecular Neurodegeneration, 2017, 12, 85.	10.8	51
102	MicroNeurotrophins Improve Survival in Motor Neuron-Astrocyte Co-Cultures but Do Not Improve Disease Phenotypes in a Mutant SOD1 Mouse Model of Amyotrophic Lateral Sclerosis. PLoS ONE, 2016, 11, e0164103.	2.5	18
103	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
104	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
105	A clinical tool for predicting survival in ALS. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 1361-1367.	1.9	57
106	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
107	<scp>ALS</scp> / <scp>FTD</scp> â€associated <scp>FUS</scp> activates <scp>GSK</scp> â€3Î² to disrupt the <scp>VAPB</scp> â€“ <scp>PTPIP</scp> 51 interaction and <scp>ER</scp> â€“mitochondria associations. EMBO Reports, 2016, 17, 1326-1342.	4.5	201
108	Gain of Toxicity from ALS/FTD-Linked Repeat Expansions in C9ORF72 Is Alleviated by Antisense Oligonucleotides Targeting GGGGCC-Containing RNAs. Neuron, 2016, 90, 535-550.	8.1	437

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109	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
110	C9ORF72 interaction with cofilin modulates actin dynamics in motor neurons. Nature Neuroscience, 2016, 19, 1610-1618.	14.8	131
111	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
112	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
113	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	21.4	218
114	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	12.8	174
115	Reply: The role ofDNAJB2in amyotrophic lateral sclerosis. Brain, 2016, 139, e58-e58.	7.6	0
116	Association of a Locus in theCAMTA1Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. JAMA Neurology, 2016, 73, 812.	9.0	57
117	The heat shock response plays an important role in TDP-43 clearance: evidence for dysfunction in amyotrophic lateral sclerosis. Brain, 2016, 139, 1417-1432.	7.6	131
118	Maturation and electrophysiological properties of human pluripotent stem cell-derived oligodendrocytes. Stem Cells, 2016, 34, 1040-1053.	3.2	65
119	Lack of association between TDP-43 pathology and tau mis-splicing in Alzheimer's disease. Neurobiology of Aging, 2016, 37, 45-46.	3.1	8
120	Retention of hexanucleotide repeat-containing intron in C9orf72 mRNA: implications for the pathogenesis of ALS/FTD. Acta Neuropathologica Communications, 2016, 4, 18.	5.2	46
121	ALS-FUS pathology revisited: singleton FUS mutations and an unusual case with both a FUS and TARDBP mutation. Acta Neuropathologica Communications, 2015, 3, 62.	5.2	22
122	C9ORF72 GGGGCC Expanded Repeats Produce Splicing Dysregulation which Correlates with Disease Severity in Amyotrophic Lateral Sclerosis. PLoS ONE, 2015, 10, e0127376.	2.5	83
123	U1 snRNP is mislocalized in ALS patient fibroblasts bearing NLS mutations in FUS and is required for motor neuron outgrowth in zebrafish. Nucleic Acids Research, 2015, 43, 3208-3218.	14.5	71
124	TDP-43 Proteinopathy and ALS: Insights into Disease Mechanisms and Therapeutic Targets. Neurotherapeutics, 2015, 12, 352-363.	4.4	246
125	VCP mutations are not a major cause of familial amyotrophic lateral sclerosis in the UK. Journal of the Neurological Sciences, 2015, 349, 209-213.	0.6	9
126	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. Science, 2015, 347, 1436-1441.	12.6	823

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127	The Spectrum of C9orf72-mediated Neurodegeneration and Amyotrophic Lateral Sclerosis. Neurotherapeutics, 2015, 12, 326-339.	4.4	46
128	Impact of disease, cognitive and behavioural factors on caregiver outcome in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2015, 16, 316-323.	1.7	33
129	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
130	Regionality of disease progression predicts prognosis in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2015, 16, 442-447.	1.7	3
131	Genetic analysis of amyotrophic lateral sclerosis in the Slovenian population. Neurobiology of Aging, 2015, 36, 1601.e17-1601.e20.	3.1	10
132	Stratified gene expression analysis identifies major amyotrophic lateral sclerosis genes. Neurobiology of Aging, 2015, 36, 2006.e1-2006.e9.	3.1	22
133	TUBA4A gene analysis in sporadic amyotrophic lateral sclerosis: identification of novel mutations. Journal of Neurology, 2015, 262, 1376-1378.	3.6	44
134	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
135	Current developments in gene therapy for amyotrophic lateral sclerosis. Expert Opinion on Biological Therapy, 2015, 15, 935-947.	3.1	30
136	Proteomic analyses reveal that loss of TDP-43 affects RNA processing and intracellular transport. Neuroscience, 2015, 293, 157-170.	2.3	52
137	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
138	Phosphorylation of C-terminal tyrosine 526 in FUS impairs its nuclear import. Journal of Cell Science, 2015, 128, 4151-9.	2.0	27
139	Wild type human TDP-43 potentiates ALS-linked mutant TDP-43 driven progressive motor and cortical neuron degeneration with pathological features of ALS. Acta Neuropathologica Communications, 2015, 3, 36.	5.2	73
140	Human iPSC-derived motoneurons harbouring TARDBP or C9ORF72 ALS mutations are dysfunctional despite maintaining viability. Nature Communications, 2015, 6, 5999.	12.8	241
141	Executive dysfunction predicts social cognition impairment in amyotrophic lateral sclerosis. Journal of Neurology, 2015, 262, 1681-1690.	3.6	36
142	Altered age-related changes in bioenergetic properties and mitochondrial morphology in fibroblasts from sporadic amyotrophic lateral sclerosis patients. Neurobiology of Aging, 2015, 36, 2893-2903.	3.1	38
143	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
144	Dipeptide repeat protein inclusions are rare in the spinal cord and almost absent from motor neurons in C9ORF72 mutant amyotrophic lateral sclerosis and are unlikely to cause their degeneration. Acta Neuropathologica Communications, 2015, 3, 38.	5.2	80

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145	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
146	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
147	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
148	Novel mutations support a role for Profilin 1 in the pathogenesis of ALS. Neurobiology of Aging, 2015, 36, 1602.e17-1602.e27.	3.1	87
149	An Evaluation of a SVA Retrotransposon in the FUS Promoter as a Transcriptional Regulator and Its Association to ALS. PLoS ONE, 2014, 9, e90833.	2.5	32
150	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
151	ERâ€“mitochondria associations are regulated by the VAPBâ€“PTPIP51 interaction and are disrupted by ALS/FTD-associated TDP-43. Nature Communications, 2014, 5, 3996.	12.8	463
152	A serum microRNA signature for amyotrophic lateral sclersosis reveals convergent RNA processing defects and identifies presymptomatic mutation carriers. Brain, 2014, 137, 2875-2876.	7.6	0
153	Psychological as well as illness factors influence acceptance of non-invasive ventilation (NIV) and gastrostomy in amyotrophic lateral sclerosis (ALS): A prospective population study. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2014, 15, 376-387.	1.7	42
154	Differential roles of the ubiquitin proteasome system (UPS) and autophagy in the clearance of soluble and aggregated TDP-43 species. Journal of Cell Science, 2014, 127, 1263-78.	2.0	216
155	Sequestration of multiple RNA recognition motif-containing proteins by C9orf72 repeat expansions. Brain, 2014, 137, 2040-2051.	7.6	253
156	Autosomal dominant inheritance of rapidly progressive amyotrophic lateral sclerosis due to a truncation mutation in the fused in sarcoma (FUS) gene. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2014, 15, 557-562.	1.7	15
157	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
158	Modelling C9ORF72 hexanucleotide repeat expansion in amyotrophic lateral sclerosis and frontotemporal dementia. Acta Neuropathologica, 2014, 127, 377-389.	7.7	43
159	A blinded international study on the reliability of genetic testing for GGGGCC-repeat expansions inC9orf72reveals marked differences in results among 14 laboratories. Journal of Medical Genetics, 2014, 51, 419-424.	3.2	118
160	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
161	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
162	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

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163	Analysis of amyotrophic lateral sclerosis as a multistep process: a population-based modelling study. Lancet Neurology, The, 2014, 13, 1108-1113.	10.2	302
164	The evaluation of pain in amyotrophic lateral sclerosis: A case controlled observational study. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2014, 15, 520-527.	1.7	55
165	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	8.1	308
166	No evidence for shared genetic basis of common variants in multiple sclerosis and amyotrophic lateral sclerosis. Human Molecular Genetics, 2014, 23, 1916-1922.	2.9	23
167	Autophagy induction enhances TDP43 turnover and survival in neuronal ALS models. Nature Chemical Biology, 2014, 10, 677-685.	8.0	368
168	<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
169	Evidence of an environmental effect on survival in ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2014, 15, 528-533.	1.7	16
170	Estimating clinical stage of amyotrophic lateral sclerosis from the ALS Functional Rating Scale. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2014, 15, 279-284.	1.7	111
171	Health utility decreases with increasing clinical stage in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2014, 15, 285-291.	1.7	26
172	Tar DNA-binding protein-43 (TDP-43) regulates axon growth in vitro and in vivo. Neurobiology of Disease, 2014, 65, 25-34.	4.4	26
173	Axonal Transport of TDP-43 mRNA Granules Is Impaired by ALS-Causing Mutations. Neuron, 2014, 81, 536-543.	8.1	521
174	Allele-Specific Knockdown of ALS-Associated Mutant TDP-43 in Neural Stem Cells Derived from Induced Pluripotent Stem Cells. PLoS ONE, 2014, 9, e91269.	2.5	39
175	Early Detection of Motor Dysfunction in the SOD1G93A Mouse Model of Amyotrophic Lateral Sclerosis (ALS) Using Home Cage Running Wheels. PLoS ONE, 2014, 9, e107918.	2.5	16
176	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
177	Hexanucleotide Repeats in ALS/FTD Form Length-Dependent RNA Foci, Sequester RNA Binding Proteins, and Are Neurotoxic. Cell Reports, 2013, 5, 1178-1186.	6.4	419
178	Residual association at C9orf72 suggests an alternative amyotrophic lateral sclerosis-causing hexanucleotide repeat. Neurobiology of Aging, 2013, 34, 2234.e1-2234.e7.	3.1	22
179	Mechanisms, models and biomarkers in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 19-32.	1.7	135
180	Analysis of TDP-43 and its binding partners in neurodegenerative diseases. Molecular Neurodegeneration, 2013, 8, .	10.8	0

#	Article	IF	Citations
181	Management of sialorrhoea in motor neuron disease: A survey of current UK practice. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 521-527.	1.7	28
182	Controversies and priorities in amyotrophic lateral sclerosis. Lancet Neurology, The, 2013, 12, 310-322.	10.2	454
183	Age of onset of amyotrophic lateral sclerosis is modulated by a locus on 1p34.1. Neurobiology of Aging, 2013, 34, 357.e7-357.e19.	3.1	69
184	Neuromuscular disease: new insights and avenues for therapy. Lancet Neurology, The, 2013, 12, 13-15.	10.2	2
185	Mixed tau, TDP-43 and p62 pathology in FTLD associated with a C9ORF72 repeat expansion and p.Ala239Thr MAPT (tau) variant. Acta Neuropathologica, 2013, 125, 303-310.	7.7	73
186	Overexpression of human wild-type FUS causes progressive motor neuron degeneration in an age- and dose-dependent fashion. Acta Neuropathologica, 2013, 125, 273-288.	7.7	225
187	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
188	Association studies indicate that protein disulfide isomerase is a risk factor in amyotrophic lateral sclerosis. Free Radical Biology and Medicine, 2013, 58, 81-86.	2.9	42
189	H63D polymorphism in HFE is not associated with amyotrophic lateral sclerosis. Neurobiology of Aging, 2013, 34, 1517.e5-1517.e7.	3.1	19
190	pNfH is a promising biomarker for ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 146-149.	1.7	63
191	ALS-linked TDP-43 mutations produce aberrant RNA splicing and adult-onset motor neuron disease without aggregation or loss of nuclear TDP-43. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, E736-45.	7.1	370
192	Mutations in prion-like domains in hnRNPA2B1 and hnRNPA1 cause multisystem proteinopathy and ALS. Nature, 2013, 495, 467-473.	27.8	1,249
193	Clinical and Molecular Aspects of Motor Neuron Disease. Colloquium Series on Genomic and Molecular Medicine, 2013, 2, 1-60.	0.2	18
194	Transcriptomic indices of fast and slow disease progression in two mouse models of amyotrophic lateral sclerosis. Brain, 2013, 136, 3305-3332.	7.6	81
195	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
196	Homozygosity analysis in amyotrophic lateral sclerosis. European Journal of Human Genetics, 2013, 21, 1429-1435.	2.8	12
197	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
198	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

#	Article	IF	Citations
199	A prospective pilot study measuring muscle volumetric change in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 414-423.	1.7	14
200	Comment on â€œDrug Screening for ALS Using Patient-Specific Induced Pluripotent Stem Cellsâ€• Science Translational Medicine, 2013, 5, 188le2.	12.4	7
201	Loss and gain of Drosophila TDP-43 impair synaptic efficacy and motor control leading to age-related neurodegeneration by loss-of-function phenotypes. Human Molecular Genetics, 2013, 22, 1539-1557.	2.9	115
202	Is language impairment more common than executive dysfunction in amyotrophic lateral sclerosis?. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 494-498.	1.9	130
203	ALS-associated mutations in FUS disrupt the axonal distribution and function of SMN. Human Molecular Genetics, 2013, 22, 3690-3704.	2.9	130
204	ALS mutant FUS disrupts nuclear localization and sequesters wild-type FUS within cytoplasmic stress granules. Human Molecular Genetics, 2013, 22, 2676-2688.	2.9	199
205	Drosophila TDP-43 dysfunction in glia and muscle cells cause cytological and behavioural phenotypes that characterize ALS and FTLD. Human Molecular Genetics, 2013, 22, 3883-3893.	2.9	72
206	Astrocyte pathology and the absence of non-cell autonomy in an induced pluripotent stem cell model of TDP-43 proteinopathy. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 4697-4702.	7.1	301
207	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
208	Unpicking neurodegeneration in a dish with human pluripotent stem cells. Cell Cycle, 2013, 12, 2339-2340.	2.6	1
209	Downregulation of MicroRNA-9 in iPSC-Derived Neurons of FTD/ALS Patients with TDP-43 Mutations. PLoS ONE, 2013, 8, e76055.	2.5	117
210	Amyotrophic lateral sclerosis-associated mutant VAPBP56S perturbs calcium homeostasis to disrupt axonal transport of mitochondria. Human Molecular Genetics, 2012, 21, 1979-1988.	2.9	112
211	VAPB interacts with the mitochondrial protein PTPIP51 to regulate calcium homeostasis. Human Molecular Genetics, 2012, 21, 1299-1311.	2.9	423
212	No evidence for a large difference in ALS frequency in populations of African and European origin: A population based study in inner city London. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 66-68.	2.1	13
213	Clinico-pathological features in amyotrophic lateral sclerosis with expansions in C9ORF72. Brain, 2012, 135, 751-764.	7.6	293
214	The Role of Variation at Aî²PP, PSEN1, PSEN2, and MAPT in Late Onset Alzheimer's Disease. Journal of Alzheimer's Disease, 2012, 28, 377-387.	2.6	53
215	FUS-SMN Protein Interactions Link the Motor Neuron Diseases ALS and SMA. Cell Reports, 2012, 2, 799-806.	6.4	229
216	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

#	Article	IF	Citations
217	Polymorphisms in the GluR2 gene are not associated with amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 418-420.	3.1	8
218	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
219	Mutation analysis of VCP in British familial and sporadic amyotrophic lateral sclerosis patients. Neurobiology of Aging, 2012, 33, 2721.e1-2721.e2.	3.1	16
220	Screening for OPTN mutations in a cohort of British amyotrophic lateral sclerosis patients. Neurobiology of Aging, 2012, 33, 2948.e15-2948.e17.	3.1	18
221	Gene expression profiling in human neurodegenerative disease. Nature Reviews Neurology, 2012, 8, 518-530.	10.1	183
222	Clinical aspects of motor neurone disease. Medicine, 2012, 40, 540-545.	0.4	3
223	Mutant induced pluripotent stem cell lines recapitulate aspects of TDP-43 proteinopathies and reveal cell-specific vulnerability. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 5803-5808.	7.1	308
224	A proposed staging system for amyotrophic lateral sclerosis. Brain, 2012, 135, 847-852.	7.6	296
225	Widespread binding of FUS along nascent RNA regulates alternative splicing in the brain. Scientific Reports, 2012, 2, 603.	3.3	231
226	Eating-derived pleasure in amyotrophic lateral sclerosis as a predictor of non-oral feeding. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 555-559.	2.1	18
227	Mapping of Gene Expression Reveals CYP27A1 as a Susceptibility Gene for Sporadic ALS. PLoS ONE, 2012, 7, e35333.	2.5	50
228	An MND/ALS phenotype associated with C9orf72 repeat expansion: Abundant p62â€positive, TDPâ€43â€negative inclusions in cerebral cortex, hippocampus and cerebellum but without associated cognitive decline. Neuropathology, 2012, 32, 505-514.	1.2	110
229	Molecular pathways of motor neuron injury in amyotrophic lateral sclerosis. Nature Reviews Neurology, 2011, 7, 616-630.	10.1	512
230	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
231	Combination of neurofilament heavy chain and complement C3 as CSF biomarkers for ALS. Journal of Neurochemistry, 2011, 117, 528-537.	3.9	128
232	Common variants at ABCA7, MS4A6A/MS4A4E, EPHA1, CD33 and CD2AP are associated with Alzheimer's disease. Nature Genetics, 2011, 43, 429-435.	21.4	1,708
233	Characterizing the RNA targets and position-dependent splicing regulation by TDP-43. Nature Neuroscience, 2011, 14, 452-458.	14.8	956
234	Endosomal accumulation of APP in wobbler motor neurons reflects impaired vesicle trafficking: Implications for human motor neuron disease. BMC Neuroscience, 2011, 12, 24.	1.9	38

#	Article	IF	Citations
235	Optineurin inclusions occur in a minority of TDP-43 positive ALS and FTLD-TDP cases and are rarely observed in other neurodegenerative disorders. Acta Neuropathologica, 2011, 121, 519-527.	7.7	70
236	p62 positive, TDP-43 negative, neuronal cytoplasmic and intranuclear inclusions in the cerebellum and hippocampus define the pathology of C9orf72-linked FTLD and MND/ALS. Acta Neuropathologica, 2011, 122, 691-702.	7.7	432
237	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
238	Protocol for a double-blind randomised placebo-controlled trial of lithium carbonate in patients with amyotrophic Lateral Sclerosis (LiCALS) [Eudract number: 2008-006891-31]. BMC Neurology, 2011, 11, 111.	1.8	16
239	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
240	Mutational analysis reveals the FUS homolog TAF15 as a candidate gene for familial amyotrophic lateral sclerosis. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2011, 156, 285-290.	1.7	148
241	The risk to relatives of patients with sporadic amyotrophic lateral sclerosis. Brain, 2011, 134, 3454-3457.	7.6	50
242	Low index-to-ring finger length ratio in sporadic ALS supports prenatally defined motor neuronal vulnerability. Journal of Neurology, Neurosurgery and Psychiatry, 2011, 82, 635-7.	1.9	48
243	Analysis of alternative splicing associated with aging and neurodegeneration in the human brain. Genome Research, 2011, 21, 1572-1582.	5.5	199
244	Broad clinical phenotypes associated with TAR-DNA binding protein (TARDBP) mutations in amyotrophic lateral sclerosis. Neurogenetics, 2010, 11, 217-225.	1.4	79
245	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
246	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
247	Common variants at 7p21 are associated with frontotemporal lobar degeneration with TDP-43 inclusions. Nature Genetics, 2010, 42, 234-239.	21.4	479
248	Genetic Evidence Implicates the Immune System and Cholesterol Metabolism in the Aetiology of Alzheimer's Disease. PLoS ONE, 2010, 5, e13950.	2.5	347
249	Familial amyotrophic lateral sclerosis is associated with a mutation in D-amino acid oxidase. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 7556-7561.	7.1	229
250	Motor neurone disease: a practical update on diagnosis and management. Clinical Medicine, 2010, 10, 252-258.	1.9	50
251	Nuclear import impairment causes cytoplasmic trans-activation response DNA-binding protein accumulation and is associated with frontotemporal lobar degeneration. Brain, 2010, 133, 1763-1771.	7.6	165
252	Dominant mutations in the cation channel gene transient receptor potential vanilloid 4 cause an unusual spectrum of neuropathies. Brain, 2010, 133, 1798-1809.	7.6	113

#	Article	IF	Citations
253	Novel FUS/TLS Mutations and Pathology in Familial and Sporadic Amyotrophic Lateral Sclerosis. Archives of Neurology, 2010, 67, 455-61.	4.5	113
254	Capturing VCP: Another Molecular Piece in the ALS Jigsaw Puzzle. Neuron, 2010, 68, 812-814.	8.1	23
255	Amyotrophic lateral sclerosis mutant vesicle-associated membrane protein-associated protein-B transgenic mice develop TAR-DNA-binding protein-43 pathology. Neuroscience, 2010, 167, 774-785.	2.3	69
256	Cloning in Research and Treatment of Human Genetic Disease. , 2010, , 875-883.		0
257	Evaluation of two different methods for per-oral gastrostomy tube placement in patients with motor neuron disease (MND): PIG versus PEG procedures. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2010, 11, 531-536.	2.1	18
258	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
259	The association between ALS and population density: A population based study. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2010, 11, 435-438.	2.1	18
260	Mutations in CHMP2B in Lower Motor Neuron Predominant Amyotrophic Lateral Sclerosis (ALS). PLoS ONE, 2010, 5, e9872.	2.5	204
261	Latent Cluster Analysis of ALS Phenotypes Identifies Prognostically Differing Groups. PLoS ONE, 2009, 4, e7107.	2.5	59
262	Variants of the elongator protein 3 (ELP3) gene are associated with motor neuron degeneration. Human Molecular Genetics, 2009, 18, 472-481.	2.9	512
263	Geographical Clustering of Amyotrophic Lateral Sclerosis in South-East England: A Population Study. Neuroepidemiology, 2009, 32, 81-88.	2.3	40
264	Reduced expression of the Kinesin-Associated Protein 3 (KIFAP3) gene increases survival in sporadic amyotrophic lateral sclerosis. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 9004-9009.	7.1	177
265	Neurofilament subunit (NFL) head domain phosphorylation regulates axonal transport of neurofilaments. European Journal of Cell Biology, 2009, 88, 193-202.	3.6	46
266	Genome-wide association study identifies variants at CLU and PICALM associated with Alzheimer's disease. Nature Genetics, 2009, 41, 1088-1093.	21.4	2,697
267	Genome-wide association study identifies 19p13.3 (UNC13A) and 9p21.2 as susceptibility loci for sporadic amyotrophic lateral sclerosis. Nature Genetics, 2009, 41, 1083-1087.	21.4	344
268	Frontotemporal lobar degeneration with ubiquitinated tauâ€negative inclusions and additional Î±â€synuclein pathology but also unusual cerebellar ubiquitinated p62â€positive, TDPâ€43â€negative inclusions. Neuropathology, 2009, 29, 466-471.	1.2	19
269	TDPâ€43 is consistently coâ€localized with ubiquitinated inclusions in sporadic and Guam amyotrophic lateral sclerosis but not in familial amyotrophic lateral sclerosis with and without SOD1 mutations. Neuropathology, 2009, 29, 672-683.	1.2	108
270	Four novel SPG3A/atlastin mutations identified in autosomal dominant hereditary spastic paraplegia kindreds with intraâ€familial variability in age of onset and complex phenotype. Clinical Genetics, 2009, 75, 485-489.	2.0	21

#	Article	IF	Citations
271	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
272	Mushroom-cage gastrostomy tube placement in patients with amyotrophic lateral sclerosis: a 5-year experience in 104 patients in a single institution. European Radiology, 2009, 19, 1763-1771.	4.5	27
273	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
274	Mutations in FUS, an RNA Processing Protein, Cause Familial Amyotrophic Lateral Sclerosis Type 6. Science, 2009, 323, 1208-1211.	12.6	2,295
275	Interaction between PON1 and population density in amyotrophic lateral sclerosis. NeuroReport, 2009, 20, 186-190.	1.2	17
276	The Role of Copy Number Variation in Susceptibility to Amyotrophic Lateral Sclerosis: Genome-Wide Association Study and Comparison with Published Loci. PLoS ONE, 2009, 4, e8175.	2.5	39
277	Birth order and the genetics of amyotrophic lateral sclerosis. Journal of Neurology, 2008, 255, 99-102.	3.6	7
278	Progressive loss of PAX6, TBR2, NEUROD and TBR1 mRNA gradients correlates with translocation of EMX2 to the cortical plate during human cortical development. European Journal of Neuroscience, 2008, 28, 1449-1456.	2.6	69
279	Deregulation of PKN1 activity disrupts neurofilament organisation and axonal transport. FEBS Letters, 2008, 582, 2303-2308.	2.8	36
280	TDP-43 Mutations in Familial and Sporadic Amyotrophic Lateral Sclerosis. Science, 2008, 319, 1668-1672.	12.6	2,268
281	A common haplotype within the PON1 promoter region is associated with sporadic ALS. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2008, 9, 306-314.	2.1	37
282	Association study on glutathione Sâ€transferase omega 1 and 2 and familial ALS. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2008, 9, 81-84.	2.1	19
283	Novel Mutations in TARDBP (TDP-43) in Patients with Familial Amyotrophic Lateral Sclerosis. PLoS Genetics, 2008, 4, e1000193.	3.5	393
284	Alexander disease with hypothermia, microcoria, and psychiatric and endocrine disturbances. Neurology, 2007, 68, 1322-1323.	1.1	22
285	Volumetric cortical loss in sporadic and familial amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2007, 8, 343-347.	2.1	45
286	Large-scale pathways-based association study in amyotrophic lateral sclerosis. Brain, 2007, 130, 2292-2301.	7.6	32
287	Amyotrophic Lateral Sclerosis in South-East England: A Population-Based Study. Neuroepidemiology, 2007, 29, 44-48.	2.3	127
288	Chapter 14 Familial amyotrophic lateral sclerosis. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2007, 82, 279-300.	1.8	3

#	Article	IF	Citations
289	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
290	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
291	Cortical involvement in four cases of primary lateral sclerosis using [11C]-flumazenil PET. Journal of Neurology, 2007, 254, 1033-1036.	3.6	42
292	Familial amyotrophic lateral sclerosis with frontotemporal dementia is linked to a locus on chromosome 9p13.2â€“21.3. Brain, 2006, 129, 868-876.	7.6	363
293	Amyotrophic lateral sclerosis in an urban setting. Journal of Neurology, 2006, 253, 1642-1643.	3.6	181
294	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
295	Susceptibility genes in sporadic ALS: Separating the wheat from the chaff by international collaboration. Neurology, 2006, 67, 738-739.	1.1	10
296	Survival of patients with ALS following institution of enteral feeding is related to preâ€procedure oximetry: A retrospective review of 98 patients in a single centre. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2006, 7, 16-21.	2.1	57
297	Amyotrophic lateral sclerosis with sensory neuropathy: part of a multisystem disorder?. Journal of Neurology, Neurosurgery and Psychiatry, 2006, 78, 750-753.	1.9	96
298	Amyotrophic Lateral Sclerosis/Motor Neuron Disease. , 2006, , 1-18.		5
299	Clinical grand round: A rapidly progressive pyramidal and extrapyramidal syndrome with a supranuclear gaze palsy. Movement Disorders, 2005, 20, 826-831.	3.9	5
300	Screening of the regulatory and coding regions of vascular endothelial growth factor in amyotrophic lateral sclerosis. Neurogenetics, 2005, 6, 101-104.	1.4	15
301	ALS2/Alsin Regulates Rac-PAK Signaling and Neurite Outgrowth. Journal of Biological Chemistry, 2005, 280, 34735-34740.	3.4	74
302	Senataxin, the ortholog of a yeast RNA helicase, is mutant in ataxia-ocular apraxia 2. Nature Genetics, 2004, 36, 225-227.	21.4	454
303	Evidence of widespread cerebral microglial activation in amyotrophic lateral sclerosis: an [11C](R)-PK11195 positron emission tomography study. Neurobiology of Disease, 2004, 15, 601-609.	4.4	630
304	p38Î± stress-activated protein kinase phosphorylates neurofilaments and is associated with neurofilament pathology in amyotrophic lateral sclerosis. Molecular and Cellular Neurosciences, 2004, 26, 354-364.	2.2	104
305	Variants in the ALS2 gene are not associated with sporadic amyotrophic lateral sclerosis. Neurogenetics, 2003, 4, 221-222.	1.4	18
306	Riluzole and Motor Neurone Disease. Practical Neurology, 2003, 3, 160-169.	1.1	3

#	Article	IF	Citations
307	VEGF is a modifier of amyotrophic lateral sclerosis in mice and humans and protects motoneurons against ischemic death. Nature Genetics, 2003, 34, 383-394.	21.4	794
308	Spastin and paraplegin gene analysis in selected cases of motor neurone disease (MND). Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders: Official Publication of the World Federation of Neurology, Research Group on Motor Neuron Diseases, 2003, 4, 96-99.	1.2	12
309	Two Families with Familial Amyotrophic Lateral Sclerosis Are Linked to a Novel Locus on Chromosome 16q. American Journal of Human Genetics, 2003, 73, 390-396.	6.2	76
310	Neurofilament heavy chain side arm phosphorylation regulates axonal transport of neurofilaments. Journal of Cell Biology, 2003, 161, 489-495.	5.2	185
311	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
312	A Novel Locus for Motor Neurone Disease on Chromosome 16q12-13. Clinical Science, 2003, 104, 40P-40P.	0.0	0
313	Identification of a Novel, Membrane-Associated Neuronal Kinase, Cyclin-Dependent Kinase 5/p35-Regulated Kinase. Journal of Neuroscience, 2003, 23, 4975-4983.	3.6	60
314	Charcot-Marie-Tooth disease neurofilament mutations disrupt neurofilament assembly and axonal transport. Human Molecular Genetics, 2002, 11, 2837-2844.	2.9	183
315	Prognostic modelling of therapeutic interventions in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders: Official Publication of the World Federation of Neurology, Research Group on Motor Neuron Diseases, 2002, 3, 15-21.	1.2	82
316	CuZn-Superoxide Dismutase in D90A Heterozygotes from Recessive and Dominant ALS Pedigrees. Neurobiology of Disease, 2002, 10, 327-333.	4.4	23
317	Reports. Journal of Interprofessional Care, 2002, 16, 289-291.	1.7	6
318	D90A-SOD1 mediated amyotrophic lateral sclerosis: A single founder for all cases with evidence for aCis-acting disease modifier in the recessive haplotype. Human Mutation, 2002, 20, 473-473.	2.5	90
319	Overexpressed human survival motor neurone isoforms, SMNÎ”exon7 and SMN+exon7, both form intranuclear gems but differ in cytoplasmic distribution. FEBS Letters, 2001, 495, 31-38.	2.8	7
320	Progress in the pathogenesis of amyotrophic lateral sclerosis. Current Neurology and Neuroscience Reports, 2001, 1, 69-76.	4.2	54
321	The enhanced antigen-specific production of cytokines induced by pertussis toxin is due to clonal expansion of T cells and not to altered effector functions of long-term memory cells. European Journal of Immunology, 2000, 30, 2422-2431.	2.9	65
322	Glutamate Slows Axonal Transport of Neurofilaments in Transfected Neurons. Journal of Cell Biology, 2000, 150, 165-176.	5.2	149
323	Phosphorylation of neurofilament heavy chain side-arms by stress activated protein kinase-1b/Jun N-terminal kinase-3. Journal of Cell Science, 2000, 113, 401-407.	2.0	90
324	Calcium, glutamate, and amyotrophic lateral sclerosis: More evidence but no certainties. Annals of Neurology, 1999, 46, 803-805.	5.3	18

#	Article	IF	Citations
325	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
326	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
327	Novel insertion in the KSP region of the neurofilament heavy gene in amyotrophic lateral sclerosis (ALS). NeuroReport, 1998, 9, 3967-3970.	1.2	157
328	Review: Glial lineages and myelination in the central nervous system. Journal of Anatomy, 1997, 190, 161-200.	1.5	96
329	Association of apolipoprotein E ∈4 allele with bulbar-onset motor neuron disease. Lancet, The, 1996, 347, 159-160.	13.7	111
330	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
331	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
332	Oxidative damage to protein in sporadic motor neuron disease spinal cord. Annals of Neurology, 1995, 38, 691-695.	5.3	312
333	Parkinsonism in motor neuron disease: case report and literature review. Acta Neuropathologica, 1995, 89, 275-283.	7.7	39
334	Distribution of AMPA-selective glutamate receptor subunits in the human hippocampus and cerebellum. Molecular Brain Research, 1995, 31, 17-32.	2.3	41
335	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
336	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
337	Amyotrophic lateral sclerosis and other disorders of the lower motor neuron. , 0, , 136-147.		0
338	Review: Glial lineages and myelination in the central nervous system. , 0, .		2
339	Genome-Wide Identification of the Genetic Basis of Amyotrophic Lateral Sclerosis. SSRN Electronic Journal, 0, , .	0.4	1