Christopher E Shaw

List of Publications by Year
in descending order

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339 papers	50,410 citations	1981 104 h-index	2142 209 g-index
354 all docs	354 docs citations	354 times ranked	39783 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.	1.4	3
2	Genome-wide identification of the genetic basis of amyotrophic lateral sclerosis. Neuron, 2022, 110, 992-1008.e11.	3.8	51
3	Structural variation analysis of 6,500 whole genome sequences in amyotrophic lateral sclerosis. Npj Genomic Medicine, 2022, 7, 8.	1.7	23
4	Disruption of ERâ€•mitochondria tethering and signalling in C9orf72â€•associated amyotrophic lateral sclerosis and frontotemporal dementia. Aging Cell, 2022, 21, e13549.	3.0	30
5	Common Genetic Variants Contribute to Risk of Transposition of the Great Arteries. Circulation Research, 2022, 130, 166-180.	2.0	15
6	Multicentre appraisal of amyotrophic lateral sclerosis biofluid biomarkers shows primacy of blood neurofilament light chain. Brain Communications, 2022, 4, fcac029.	1.5	29
7	Genome-wide study of DNA methylation shows alterations in metabolic, inflammatory, and cholesterol pathways in ALS. Science Translational Medicine, 2022, 14, eabj0264.	5.8	38
8	Unbiased metabolome screen leads to personalized medicine strategy for amyotrophic lateral sclerosis. Brain Communications, 2022, 4, fcac069.	1.5	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.	7.1	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.	1.8	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.	1.8	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.	1.1	13
13	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	3.8	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.	2.8	10
15	The gut microbiome: a key player in the complexity of amyotrophic lateral sclerosis (ALS). BMC Medicine, 2021, 19, 13.	2.3	52
16	Value of systematic genetic screening of patients with amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 510-518.	0.9	69
17	Meta-analysis of genome-wide DNA methylation identifies shared associations across neurodegenerative disorders. Genome Biology, 2021, 22, 90.	3.8	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.	0.3	3

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19	Disease Mechanisms and Therapeutic Approaches in C9orf72 ALS-FTD. Biomedicines, 2021, 9, 601.	1.4	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.	1.6	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.	2.7	65
22	Regulation of Synapse Weakening through Interactions of the Microtubule Associated Protein Tau with PACSIN1. Journal of Neuroscience, 2021, 41, 7162-7170.	1.7	12
23	Innovating Clinical Trials for Amyotrophic Lateral Sclerosis. Neurology, 2021, 97, 528-536.	1.5	19
24	Cytoplasmic TDP-43 is involved in cell fate during stress recovery. Human Molecular Genetics, 2021, 31, 166-175.	1.4	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.	4.4	13
26	Demystifying the spontaneous phenomena of motor hyperexcitability. Clinical Neurophysiology, 2021, 132, 1830-1844.	0.7	10
27	Extensive phenotypic characterisation of a human TDP-43Q331KÂtransgenic mouse model of amyotrophic lateral sclerosis (ALS). Scientific Reports, 2021, 11, 16659.	1.6	12
28	Amyotrophic lateral sclerosis alters the metabolic aging profile in patient derived fibroblasts. Neurobiology of Aging, 2021, 105, 64-77.	1.5	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.	1.5	3
30	SCFD1 expression quantitative trait loci in amyotrophic lateral sclerosis are differentially expressed. Brain Communications, 2021, 3, fcab236.	1.5	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.	9.4	223
32	Proteinopathies as Hallmarks of Impaired Gene Expression, Proteostasis and Mitochondrial Function in Amyotrophic Lateral Sclerosis. Frontiers in Neuroscience, 2021, 15, 783624.	1.4	13
33	Disrupted glycosylation of lipids and proteins is a cause of neurodegeneration. Brain, 2020, 143, 1332-1340.	3.7	58
34	Mutant C9orf72 human iPSCâ€derived astrocytes cause nonâ€cell autonomous motor neuron pathophysiology. Glia, 2020, 68, 1046-1064.	2.5	90
35	Rare Variant Burden Analysis within Enhancers Identifies CAV1 as an ALS Risk Gene. Cell Reports, 2020, 33, 108456.	2.9	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.	0.5	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.	1.1	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.	3.3	53
39	CYLD is a causative gene for frontotemporal dementia â€“ amyotrophic lateral sclerosis. Brain, 2020, 143, 783-799.	3.7	62
40	UK case control study of smoking and risk of amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 222-227.	1.1	10
41	Magnetic resonance spectroscopy reveals mitochondrial dysfunction in amyotrophic lateral sclerosis. Brain, 2020, 143, 3603-3618.	3.7	24
42	ATXN1 repeat expansions confer risk for amyotrophic lateral sclerosis and contribute to TDP-43 mislocalization. Brain Communications, 2020, 2, fcaa064.	1.5	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.	1.5	13
44	C9orf72 intermediate expansions of 24â€“30 repeats are associated with ALS. Acta Neuropathologica Communications, 2019, 7, 115.	2.4	75
45	RRM adjacent TARDBP mutations disrupt RNA binding and enhance TDP-43 proteinopathy. Brain, 2019, 142, 3753-3770.	3.7	71
46	Astrocyte adenosine deaminase loss increases motor neuron toxicity in amyotrophic lateral sclerosis. Brain, 2019, 142, 586-605.	3.7	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.	1.4	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.	0.9	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.	1.1	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.	1.1	18
51	Biomarkers in Motor Neuron Disease: A State of the Art Review. Frontiers in Neurology, 2019, 10, 291.	1.1	87
52	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. Annals of Neurology, 2019, 85, 470-481.	2.8	118
53	Nuclear RNA foci from C9ORF72 expansion mutation form paraspeckle-like bodies. Journal of Cell Science, 2019, 132, .	1.2	36
54	Mutations in the Glycosyltransferase Domain of GLT8D1 Are Associated with Familial Amyotrophic Lateral Sclerosis. Cell Reports, 2019, 26, 2298-2306.e5.	2.9	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.	9.4	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.	0.8	20
57	Process evaluation and exploration of telehealth in motor neuron disease in a UK specialist centre. BMJ Open, 2019, 9, e028526.	0.8	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.	0.9	14
59	Human genetics and neuropathology suggest a link between miR-218 and amyotrophic lateral sclerosis pathophysiology. Science Translational Medicine, 2019, 11, .	5.8	37
60	Exome sequencing in amyotrophic lateral sclerosis implicates a novel gene, DNAJC7, encoding a heat-shock protein. Nature Neuroscience, 2019, 22, 1966-1974.	7.1	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.	1.5	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.	1.5	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.	0.9	38
64	The role of mitochondria in amyotrophic lateral sclerosis. Neuroscience Letters, 2019, 710, 132933.	1.0	356
65	Objectively Monitoring Amyotrophic Lateral Sclerosis Patient Symptoms During Clinical Trials With Sensors: Observational Study. JMIR MHealth and UHealth, 2019, 7, e13433.	1.8	32
66	Overriding FUS autoregulation in mice triggers gain-of-toxic dysfunctions in RNA metabolism and autophagy-lysosome axis. ELife, 2019, 8, .	2.8	65
67	C9ORF72 repeat expansion causes vulnerability of motor neurons to Ca2+-permeable AMPA receptor-mediated excitotoxicity. Nature Communications, 2018, 9, 347.	5.8	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.	1.4	74
69	Prognosis for patients with amyotrophic lateral sclerosis: development and validation of a personalised prediction model. Lancet Neurology, The, 2018, 17, 423-433.	4.9	342
70	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	3.8	517
71	Reconsidering the causality of TIA1 mutations in ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2018, 19, 1-3.	1.1	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.	2.4	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.	3.7	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.	3.8	185
75	ALS-specific cognitive and behavior changes associated with advancing disease stage in ALS. Neurology, 2018, 91, e1370-e1380.	1.5	170
76	TDP-43 induces p53-mediated cell death of cortical progenitors and immature neurons. Scientific Reports, 2018, 8, 8097.	1.6	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.	1.5	59
78	TDP-43 causes neurotoxicity and cytoskeletal dysfunction in primary cortical neurons. PLoS ONE, 2018, 13, e0196528.	1.1	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.	1.5	86
81	C9orf72 expansion differentially affects males with spinal onset amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 281.1-281.	0.9	33
82	Comparison of the Kingâ€™s and MiToS staging systems for ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2017, 18, 227-232.	1.1	58
83	Non-nuclear Pool of Splicing Factor SFPQ Regulates Axonal Transcripts Required for Normal Motor Development. Neuron, 2017, 94, 322-336.e5.	3.8	61
84	A comprehensive analysis of rare genetic variation in amyotrophic lateral sclerosis in the UK. Brain, 2017, 140, 1611-1618.	3.7	71
85	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, .	5.8	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.	3.9	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.	1.2	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.	1.4	64
89	Amyotrophic lateral sclerosis. Nature Reviews Disease Primers, 2017, 3, 17071.	18.1	885
90	Meta-analysis of pharmacogenetic interactions in amyotrophic lateral sclerosis clinical trials. Neurology, 2017, 89, 1915-1922.	1.5	82

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91	Can Astrocytes Be a Target for Precision Medicine?. Advances in Experimental Medicine and Biology, 2017, 1007, 111-128.	0.8	7
92	Detection of long repeat expansions from PCR-free whole-genome sequence data. Genome Research, 2017, 27, 1895-1903.	2.4	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.	9.4	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.	1.1	38
95	SRSF1-dependent nuclear export inhibition of C9ORF72 repeat transcripts prevents neurodegeneration and associated motor deficits. Nature Communications, 2017, 8, 16063.	5.8	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.	1.5	18
97	RNA Misprocessing in C9orf72-Linked Neurodegeneration. Frontiers in Cellular Neuroscience, 2017, 11, 195.	1.8	32
98	Protein Homeostasis in Amyotrophic Lateral Sclerosis: Therapeutic Opportunities?. Frontiers in Molecular Neuroscience, 2017, 10, 123.	1.4	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.	1.4	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.	1.4	23
101	Advances, challenges and future directions for stem cell therapy in amyotrophic lateral sclerosis. Molecular Neurodegeneration, 2017, 12, 85.	4.4	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.	1.1	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.	0.7	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.	1.8	7
105	A clinical tool for predicting survival in ALS. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 1361-1367.	0.9	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.	3.5	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.	2.0	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.	3.8	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.	3.3	139
110	C9ORF72 interaction with cofilin modulates actin dynamics in motor neurons. Nature Neuroscience, 2016, 19, 1610-1618.	7.1	131
111	Rare genetic variation in UNC13A may modify survival in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2016, 17, 593-599.	1.1	22
112	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1043-1048.	9.4	494
113	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	9.4	218
114	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	5.8	174
115	Reply: The role ofDNAJB2in amyotrophic lateral sclerosis. Brain, 2016, 139, e58-e58.	3.7	0
116	Association of a Locus in theCAMTA1Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. JAMA Neurology, 2016, 73, 812.	4.5	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.	3.7	131
118	Maturation and electrophysiological properties of human pluripotent stem cell-derived oligodendrocytes. Stem Cells, 2016, 34, 1040-1053.	1.4	65
119	Lack of association between TDP-43 pathology and tau mis-splicing in Alzheimer's disease. Neurobiology of Aging, 2016, 37, 45-46.	1.5	8
120	Retention of hexanucleotide repeat-containing intron in C9orf72 mRNA: implications for the pathogenesis of ALS/FTD. Acta Neuropathologica Communications, 2016, 4, 18.	2.4	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.	2.4	22
122	C9ORF72 GGGGCC Expanded Repeats Produce Splicing Dysregulation which Correlates with Disease Severity in Amyotrophic Lateral Sclerosis. PLoS ONE, 2015, 10, e0127376.	1.1	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.	6.5	71
124	TDP-43 Proteinopathy and ALS: Insights into Disease Mechanisms and Therapeutic Targets. Neurotherapeutics, 2015, 12, 352-363.	2.1	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.	0.3	9
126	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. Science, 2015, 347, 1436-1441.	6.0	823

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127	The Spectrum of C9orf72-mediated Neurodegeneration and Amyotrophic Lateral Sclerosis. Neurotherapeutics, 2015, 12, 326-339.	2.1	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.	1.1	33
129	Intermediate length C9orf72 expansion in an ALS patient without classical C9orf72 neuropathology. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2015, 16, 249-251.	1.1	8
130	Regionality of disease progression predicts prognosis in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2015, 16, 442-447.	1.1	3
131	Genetic analysis of amyotrophic lateral sclerosis in the Slovenian population. Neurobiology of Aging, 2015, 36, 1601.e17-1601.e20.	1.5	10
132	Stratified gene expression analysis identifies major amyotrophic lateral sclerosis genes. Neurobiology of Aging, 2015, 36, 2006.e1-2006.e9.	1.5	22
133	TUBA4A gene analysis in sporadic amyotrophic lateral sclerosis: identification of novel mutations. Journal of Neurology, 2015, 262, 1376-1378.	1.8	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.	3.9	149
135	Current developments in gene therapy for amyotrophic lateral sclerosis. Expert Opinion on Biological Therapy, 2015, 15, 935-947.	1.4	30
136	Proteomic analyses reveal that loss of TDP-43 affects RNA processing and intracellular transport. Neuroscience, 2015, 293, 157-170.	1.1	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.	0.4	152
138	Phosphorylation of C-terminal tyrosine 526 in FUS impairs its nuclear import. Journal of Cell Science, 2015, 128, 4151-9.	1.2	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.	2.4	73
140	Human iPSC-derived motoneurons harbouring TARDBP or C9ORF72 ALS mutations are dysfunctional despite maintaining viability. Nature Communications, 2015, 6, 5999.	5.8	241
141	Executive dysfunction predicts social cognition impairment in amyotrophic lateral sclerosis. Journal of Neurology, 2015, 262, 1681-1690.	1.8	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.	1.5	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.	1.5	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.	2.4	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.	0.9	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.	1.8	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.	1.8	47
148	Novel mutations support a role for Profilin 1 in the pathogenesis of ALS. Neurobiology of Aging, 2015, 36, 1602.e17-1602.e27.	1.5	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.	1.1	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.	3.3	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.	5.8	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.	3.7	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.	1.1	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.	1.2	216
155	Sequestration of multiple RNA recognition motif-containing proteins by C9orf72 repeat expansions. Brain, 2014, 137, 2040-2051.	3.7	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.	1.1	15
157	The widening spectrum of C9ORF72-related disease; genotype/phenotype correlations and potential modifiers of clinical phenotype. Acta Neuropathologica, 2014, 127, 333-345.	3.9	150
158	Modelling C9ORF72 hexanucleotide repeat expansion in amyotrophic lateral sclerosis and frontotemporal dementia. Acta Neuropathologica, 2014, 127, 377-389.	3.9	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.	1.5	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.	1.5	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.	1.8	98
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