Christopher E Shaw

List of Publications by Year
in descending order

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	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
2	Mutations in FUS, an RNA Processing Protein, Cause Familial Amyotrophic Lateral Sclerosis Type 6. Science, 2009, 323, 1208-1211.	12.6	2,295
3	TDP-43 Mutations in Familial and Sporadic Amyotrophic Lateral Sclerosis. Science, 2008, 319, 1668-1672.	12.6	2,268
4	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
5	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
6	Mutations in prion-like domains in hnRNPA2B1 and hnRNPA1 cause multisystem proteinopathy and ALS. Nature, 2013, 495, 467-473.	27.8	1,249
7	Characterizing the RNA targets and position-dependent splicing regulation by TDP-43. Nature Neuroscience, 2011, 14, 452-458.	14.8	956
8	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
9	Amyotrophic lateral sclerosis. Nature Reviews Disease Primers, 2017, 3, 17071.	30.5	885
10	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
11	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. Science, 2015, 347, 1436-1441.	12.6	823
12	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
13	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
14	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
15	Axonal Transport of TDP-43 mRNA Granules Is Impaired by ALS-Causing Mutations. Neuron, 2014, 81, 536-543.	8.1	521
16	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517
17	Variants of the elongator protein 3 (ELP3) gene are associated with motor neuron degeneration. Human Molecular Genetics, 2009, 18, 472-481.	2.9	512
18	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

#	Article	IF	Citations
19	Molecular pathways of motor neuron injury in amyotrophic lateral sclerosis. Nature Reviews Neurology, 2011, 7, 616-630.	10.1	512
20	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
21	Common variants at 7p21 are associated with frontotemporal lobar degeneration with TDP-43 inclusions. Nature Genetics, 2010, 42, 234-239.	21.4	479
22	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
23	Senataxin, the ortholog of a yeast RNA helicase, is mutant in ataxia-ocular apraxia 2. Nature Genetics, 2004, 36, 225-227.	21.4	454
24	Controversies and priorities in amyotrophic lateral sclerosis. Lancet Neurology, The, 2013, 12, 310-322.	10.2	454
25	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
26	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
27	VAPB interacts with the mitochondrial protein PTPIP51 to regulate calcium homeostasis. Human Molecular Genetics, 2012, 21, 1299-1311.	2.9	423
28	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
29	Novel Mutations in TARDBP (TDP-43) in Patients with Familial Amyotrophic Lateral Sclerosis. PLoS Genetics, 2008, 4, e1000193.	3.5	393
30	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
31	Autophagy induction enhances TDP43 turnover and survival in neuronal ALS models. Nature Chemical Biology, 2014, 10, 677-685.	8.0	368
32	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
33	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
34	The role of mitochondria in amyotrophic lateral sclerosis. Neuroscience Letters, 2019, 710, 132933.	2.1	356
35	Genetic Evidence Implicates the Immune System and Cholesterol Metabolism in the Aetiology of Alzheimer's Disease. PLoS ONE, 2010, 5, e13950.	2.5	347
36	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

#	Article	IF	Citations
37	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
38	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
39	Oxidative damage to protein in sporadic motor neuron disease spinal cord. Annals of Neurology, 1995, 38, 691-695.	5.3	312
40	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
41	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	8.1	308
42	Analysis of amyotrophic lateral sclerosis as a multistep process: a population-based modelling study. Lancet Neurology, The, 2014, 13, 1108-1113.	10.2	302
43	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
44	A proposed staging system for amyotrophic lateral sclerosis. Brain, 2012, 135, 847-852.	7.6	296
45	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
46	Clinico-pathological features in amyotrophic lateral sclerosis with expansions in C9ORF72. Brain, 2012, 135, 751-764.	7.6	293
47	Detection of long repeat expansions from PCR-free whole-genome sequence data. Genome Research, 2017, 27, 1895-1903.	5.5	277
48	Sequestration of multiple RNA recognition motif-containing proteins by C9orf72 repeat expansions. Brain, 2014, 137, 2040-2051.	7.6	253
49	TDP-43 Proteinopathy and ALS: Insights into Disease Mechanisms and Therapeutic Targets. Neurotherapeutics, 2015, 12, 352-363.	4.4	246
50	Human iPSC-derived motoneurons harbouring TARDBP or C9ORF72 ALS mutations are dysfunctional despite maintaining viability. Nature Communications, 2015, 6, 5999.	12.8	241
51	Widespread binding of FUS along nascent RNA regulates alternative splicing in the brain. Scientific Reports, 2012, 2, 603.	3.3	231
52	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
53	FUS-SMN Protein Interactions Link the Motor Neuron Diseases ALS and SMA. Cell Reports, 2012, 2, 799-806.	6.4	229
54	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

#	Article	IF	Citations
55	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
56	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
57	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	21.4	218
58	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
59	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
60	Mutations in CHMP2B in Lower Motor Neuron Predominant Amyotrophic Lateral Sclerosis (ALS). PLoS ONE, 2010, 5, e9872.	2.5	204
61	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
62	<scp>ALS</scp> / <scp>FTD</scp> â€ssociated <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
63	Analysis of alternative splicing associated with aging and neurodegeneration in the human brain. Genome Research, 2011, 21, 1572-1582.	5.5	199
64	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
65	Neurofilament heavy chain side arm phosphorylation regulates axonal transport of neurofilaments. Journal of Cell Biology, 2003, 161, 489-495.	5.2	185
66	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
67	Charcot-Marie-Tooth disease neurofilament mutations disrupt neurofilament assembly and axonal transport. Human Molecular Genetics, 2002, 11, 2837-2844.	2.9	183
68	Gene expression profiling in human neurodegenerative disease. Nature Reviews Neurology, 2012, 8, 518-530.	10.1	183
69	Amyotrophic lateral sclerosis in an urban setting. Journal of Neurology, 2006, 253, 1642-1643.	3.6	181
70	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
71	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	12.8	174
72	ALS-specific cognitive and behavior changes associated with advancing disease stage in ALS. Neurology, 2018, 91, e1370-e1380.	1.1	170

#	Article	IF	Citations
73	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
74	Novel insertion in the KSP region of the neurofilament heavy gene in amyotrophic lateral sclerosis (ALS). NeuroReport, 1998, 9, 3967-3970.	1.2	157
75	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
76	C9ORF72 repeat expansion causes vulnerability of motor neurons to Ca2+-permeable AMPA receptor-mediated excitotoxicity. Nature Communications, 2018, 9, 347.	12.8	151
77	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
78	Glutamate Slows Axonal Transport of Neurofilaments in Transfected Neurons. Journal of Cell Biology, 2000, 150, 165-176.	5.2	149
79	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
80	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
81	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
82	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
83	Mechanisms, models and biomarkers in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 19-32.	1.7	135
84	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
85	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
86	C9ORF72 interaction with cofilin modulates actin dynamics in motor neurons. Nature Neuroscience, 2016, 19, 1610-1618.	14.8	131
87	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
88	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
89	ALS-associated mutations in FUS disrupt the axonal distribution and function of SMN. Human Molecular Genetics, 2013, 22, 3690-3704.	2.9	130
90	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, .	12.4	129

#	Article	IF	Citations
91	Combination of neurofilament heavy chain and complement C3 as CSF biomarkers for ALS. Journal of Neurochemistry, 2011, 117, 528-537.	3.9	128
92	Amyotrophic Lateral Sclerosis in South-East England: A Population-Based Study. Neuroepidemiology, 2007, 29, 44-48.	2.3	127
93	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
94	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
95	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
96	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
97	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
98	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. Annals of Neurology, 2019, 85, 470-481.	5.3	118
99	Downregulation of MicroRNA-9 in iPSC-Derived Neurons of FTD/ALS Patients with TDP-43 Mutations. PLoS ONE, 2013, 8, e76055.	2.5	117
100	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
101	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
102	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
103	Novel FUS/TLS Mutations and Pathology in Familial and Sporadic Amyotrophic Lateral Sclerosis. Archives of Neurology, 2010, 67, 455-61.	4.5	113
104	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
105	Association of apolipoprotein E ∈4 allele with bulbar-onset motor neuron disease. Lancet, The, 1996, 347, 159-160.	13.7	111
106	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
107	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
108	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

#	Article	IF	Citations
109	SRSF1-dependent nuclear export inhibition of C9ORF72 repeat transcripts prevents neurodegeneration and associated motor deficits. Nature Communications, 2017, 8, 16063.	12.8	106
110	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
111	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
112	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
113	Review: Glial lineages and myelination in the central nervous system. Journal of Anatomy, 1997, 190, 161-200.	1.5	96
114	Amyotrophic lateral sclerosis with sensory neuropathy: part of a multisystem disorder?. Journal of Neurology, Neurosurgery and Psychiatry, 2006, 78, 750-753.	1.9	96
115	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
116	<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
117	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
118	Mutant C9orf72 human iPSCâ€derived astrocytes cause nonâ€cell autonomous motor neuron pathophysiology. Glia, 2020, 68, 1046-1064.	4.9	90
119	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
120	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
121	Biomarkers in Motor Neuron Disease: A State of the Art Review. Frontiers in Neurology, 2019, 10, 291.	2.4	87
122	ATXN2 trinucleotide repeat length correlates with risk of ALS. Neurobiology of Aging, 2017, 51, 178.e1-178.e9.	3.1	86
123	Astrocyte adenosine deaminase loss increases motor neuron toxicity in amyotrophic lateral sclerosis. Brain, 2019, 142, 586-605.	7.6	84
124	C9ORF72 GGGGCC Expanded Repeats Produce Splicing Dysregulation which Correlates with Disease Severity in Amyotrophic Lateral Sclerosis. PLoS ONE, 2015, 10, e0127376.	2.5	83
125	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
126	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

#	Article	IF	Citations
127	Meta-analysis of pharmacogenetic interactions in amyotrophic lateral sclerosis clinical trials. Neurology, 2017, 89, 1915-1922.	1.1	82
128	Transcriptomic indices of fast and slow disease progression in two mouse models of amyotrophic lateral sclerosis. Brain, 2013, 136, 3305-3332.	7.6	81
129	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
130	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
131	Broad clinical phenotypes associated with TAR-DNA binding protein (TARDBP) mutations in amyotrophic lateral sclerosis. Neurogenetics, 2010, 11, 217-225.	1.4	79
132	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
133	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
134	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
135	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
136	A feedback loop between dipeptide-repeat protein, TDP-43 and karyopherin-Î± mediates C9orf72-related neurodegeneration. Brain, 2018, 141, 2908-2924.	7.6	75
137	C9orf72 intermediate expansions of 24â€“30 repeats are associated with ALS. Acta Neuropathologica Communications, 2019, 7, 115.	5.2	75
138	ALS2/Alsin Regulates Rac-PAK Signaling and Neurite Outgrowth. Journal of Biological Chemistry, 2005, 280, 34735-34740.	3.4	74
139	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
140	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
141	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
142	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
143	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
144	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

#	Article	IF	Citations
145	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
146	A comprehensive analysis of rare genetic variation in amyotrophic lateral sclerosis in the UK. Brain, 2017, 140, 1611-1618.	7.6	71
147	RRM adjacent TARDBP mutations disrupt RNA binding and enhance TDP-43 proteinopathy. Brain, 2019, 142, 3753-3770.	7.6	71
148	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
149	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
150	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
151	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
152	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
153	Value of systematic genetic screening of patients with amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 510-518.	1.9	69
154	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
155	Maturation and electrophysiological properties of human pluripotent stem cell-derived oligodendrocytes. Stem Cells, 2016, 34, 1040-1053.	3.2	65
156	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
157	Overriding FUS autoregulation in mice triggers gain-of-toxic dysfunctions in RNA metabolism and autophagy-lysosome axis. ELife, 2019, 8, .	6.0	65
158	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
159	pNfH is a promising biomarker for ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 146-149.	1.7	63
160	Protein Homeostasis in Amyotrophic Lateral Sclerosis: Therapeutic Opportunities?. Frontiers in Molecular Neuroscience, 2017, 10, 123.	2.9	62
161	CYLD is a causative gene for frontotemporal dementia â€“ amyotrophic lateral sclerosis. Brain, 2020, 143, 783-799.	7.6	62
162	Non-nuclear Pool of Splicing Factor SFPQ Regulates Axonal Transcripts Required for Normal Motor Development. Neuron, 2017, 94, 322-336.e5.	8.1	61

#	Article	IF	Citations
163	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
164	Latent Cluster Analysis of ALS Phenotypes Identifies Prognostically Differing Groups. PLoS ONE, 2009, 4, e7107.	2.5	59
165	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
166	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
167	Comparison of the Kingâ€™s and MiToS staging systems for ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2017, 18, 227-232.	1.7	58
168	Disrupted glycosylation of lipids and proteins is a cause of neurodegeneration. Brain, 2020, 143, 1332-1340.	7.6	58
169	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
170	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
171	A clinical tool for predicting survival in ALS. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 1361-1367.	1.9	57
172	Association of a Locus in theCAMTA1Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. JAMA Neurology, 2016, 73, 812.	9.0	57
173	Mutations in the Glycosyltransferase Domain of GLT8D1 Are Associated with Familial Amyotrophic Lateral Sclerosis. Cell Reports, 2019, 26, 2298-2306.e5.	6.4	57
174	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	8.1	56
175	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
176	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
177	Progress in the pathogenesis of amyotrophic lateral sclerosis. Current Neurology and Neuroscience Reports, 2001, 1, 69-76.	4.2	54
178	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
179	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
180	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

#	Article	IF	Citations
181	Proteomic analyses reveal that loss of TDP-43 affects RNA processing and intracellular transport. Neuroscience, 2015, 293, 157-170.	2.3	52
182	The gut microbiome: a key player in the complexity of amyotrophic lateral sclerosis (ALS). BMC Medicine, 2021, 19, 13.	5.5	52
183	Advances, challenges and future directions for stem cell therapy in amyotrophic lateral sclerosis. Molecular Neurodegeneration, 2017, 12, 85.	10.8	51
184	Genome-wide identification of the genetic basis of amyotrophic lateral sclerosis. Neuron, 2022, 110, 992-1008.e11.	8.1	51
185	Motor neurone disease: a practical update on diagnosis and management. Clinical Medicine, 2010, 10, 252-258.	1.9	50
186	The risk to relatives of patients with sporadic amyotrophic lateral sclerosis. Brain, 2011, 134, 3454-3457.	7.6	50
187	Mapping of Gene Expression Reveals CYP27A1 as a Susceptibility Gene for Sporadic ALS. PLoS ONE, 2012, 7, e35333.	2.5	50
188	Meta-analysis of genome-wide DNA methylation identifies shared associations across neurodegenerative disorders. Genome Biology, 2021, 22, 90.	8.8	49
189	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
190	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
191	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
192	Neurofilament subunit (NFL) head domain phosphorylation regulates axonal transport of neurofilaments. European Journal of Cell Biology, 2009, 88, 193-202.	3.6	46
193	The Spectrum of C9orf72-mediated Neurodegeneration and Amyotrophic Lateral Sclerosis. Neurotherapeutics, 2015, 12, 326-339.	4.4	46
194	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
195	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
196	TUBA4A gene analysis in sporadic amyotrophic lateral sclerosis: identification of novel mutations. Journal of Neurology, 2015, 262, 1376-1378.	3.6	44
197	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
198	Modelling C9ORF72 hexanucleotide repeat expansion in amyotrophic lateral sclerosis and frontotemporal dementia. Acta Neuropathologica, 2014, 127, 377-389.	7.7	43

#	Article	IF	Citations
199	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
200	Cortical involvement in four cases of primary lateral sclerosis using [11C]-flumazenil PET. Journal of Neurology, 2007, 254, 1033-1036.	3.6	42
201	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
202	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
203	Distribution of AMPA-selective glutamate receptor subunits in the human hippocampus and cerebellum. Molecular Brain Research, 1995, 31, 17-32.	2.3	41
204	Geographical Clustering of Amyotrophic Lateral Sclerosis in South-East England: A Population Study. Neuroepidemiology, 2009, 32, 81-88.	2.3	40
205	Parkinsonism in motor neuron disease: case report and literature review. Acta Neuropathologica, 1995, 89, 275-283.	7.7	39
206	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
207	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
208	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
209	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
210	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
211	TDP-43 induces p53-mediated cell death of cortical progenitors and immature neurons. Scientific Reports, 2018, 8, 8097.	3.3	38
212	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
213	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
214	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
215	Human genetics and neuropathology suggest a link between miR-218 and amyotrophic lateral sclerosis pathophysiology. Science Translational Medicine, 2019, 11, .	12.4	37
216	Deregulation of PKN1 activity disrupts neurofilament organisation and axonal transport. FEBS Letters, 2008, 582, 2303-2308.	2.8	36

#	Article	IF	Citations
217	Executive dysfunction predicts social cognition impairment in amyotrophic lateral sclerosis. Journal of Neurology, 2015, 262, 1681-1690.	3.6	36
218	Nuclear RNA foci from C9ORF72 expansion mutation form paraspeckle-like bodies. Journal of Cell Science, 2019, 132, .	2.0	36
219	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
220	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
221	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
222	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
223	ATXN1 repeat expansions confer risk for amyotrophic lateral sclerosis and contribute to TDP-43 mislocalization. Brain Communications, 2020, 2, fcaa064.	3.3	33
224	Large-scale pathways-based association study in amyotrophic lateral sclerosis. Brain, 2007, 130, 2292-2301.	7.6	32
225	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
226	RNA Misprocessing in C9orf72-Linked Neurodegeneration. Frontiers in Cellular Neuroscience, 2017, 11, 195.	3.7	32
227	Objectively Monitoring Amyotrophic Lateral Sclerosis Patient Symptoms During Clinical Trials With Sensors: Observational Study. JMIR MHealth and UHealth, 2019, 7, e13433.	3.7	32
228	Current developments in gene therapy for amyotrophic lateral sclerosis. Expert Opinion on Biological Therapy, 2015, 15, 935-947.	3.1	30
229	Disruption of ERâ€mitochondria tethering and signalling in C9orf72â€associated amyotrophic lateral sclerosis and frontotemporal dementia. Aging Cell, 2022, 21, e13549.	6.7	30
230	Multicentre appraisal of amyotrophic lateral sclerosis biofluid biomarkers shows primacy of blood neurofilament light chain. Brain Communications, 2022, 4, fcac029.	3.3	29
231	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
232	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
233	Phosphorylation of C-terminal tyrosine 526 in FUS impairs its nuclear import. Journal of Cell Science, 2015, 128, 4151-9.	2.0	27
234	TDP-43 causes neurotoxicity and cytoskeletal dysfunction in primary cortical neurons. PLoS ONE, 2018, 13, e0196528.	2.5	27

#	Article	IF	Citations
235	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
236	Health utility decreases with increasing clinical stage in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2014, 15, 285-291.	1.7	26
237	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
238	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
239	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
240	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
241	Rare Variant Burden Analysis within Enhancers Identifies CAV1 as an ALS Risk Gene. Cell Reports, 2020, 33, 108456.	6.4	24
242	Magnetic resonance spectroscopy reveals mitochondrial dysfunction in amyotrophic lateral sclerosis. Brain, 2020, 143, 3603-3618.	7.6	24
243	CuZn-Superoxide Dismutase in D90A Heterozygotes from Recessive and Dominant ALS Pedigrees. Neurobiology of Disease, 2002, 10, 327-333.	4.4	23
244	Capturing VCP: Another Molecular Piece in the ALS Jigsaw Puzzle. Neuron, 2010, 68, 812-814.	8.1	23
245	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
246	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
247	Structural variation analysis of 6,500 whole genome sequences in amyotrophic lateral sclerosis. Npj Genomic Medicine, 2022, 7, 8.	3.8	23
248	Alexander disease with hypothermia, microcoria, and psychiatric and endocrine disturbances. Neurology, 2007, 68, 1322-1323.	1.1	22
249	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
250	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
251	Stratified gene expression analysis identifies major amyotrophic lateral sclerosis genes. Neurobiology of Aging, 2015, 36, 2006.e1-2006.e9.	3.1	22
252	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

#	Article	IF	Citations
253	Reconsidering the causality of TIA1 mutations in ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2018, 19, 1-3.	1.7	22
254	Process evaluation and exploration of telehealth in motor neuron disease in a UK specialist centre. BMJ Open, 2019, 9, e028526.	1.9	22
255	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
256	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
257	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
258	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
259	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
260	H63D polymorphism in HFE is not associated with amyotrophic lateral sclerosis. Neurobiology of Aging, 2013, 34, 1517.e5-1517.e7.	3.1	19
261	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
262	Innovating Clinical Trials for Amyotrophic Lateral Sclerosis. Neurology, 2021, 97, 528-536.	1.1	19
263	Calcium, glutamate, and amyotrophic lateral sclerosis: More evidence but no certainties. Annals of Neurology, 1999, 46, 803-805.	5.3	18
264	Variants in the ALS2 gene are not associated with sporadic amyotrophic lateral sclerosis. Neurogenetics, 2003, 4, 221-222.	1.4	18
265	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
266	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
267	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
268	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
269	Clinical and Molecular Aspects of Motor Neuron Disease. Colloquium Series on Genomic and Molecular Medicine, 2013, 2, 1-60.	0.2	18
270	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

#	Article	IF	Citations
271	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
272	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
273	Interaction between PON1 and population density in amyotrophic lateral sclerosis. NeuroReport, 2009, 20, 186-190.	1.2	17
274	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
275	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
276	Evidence of an environmental effect on survival in ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2014, 15, 528-533.	1.7	16
277	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
278	Amyotrophic lateral sclerosis alters the metabolic aging profile in patient derived fibroblasts. Neurobiology of Aging, 2021, 105, 64-77.	3.1	16
279	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
280	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
281	Screening of the regulatory and coding regions of vascular endothelial growth factor in amyotrophic lateral sclerosis. Neurogenetics, 2005, 6, 101-104.	1.4	15
282	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
283	Cytoplasmic TDP-43 is involved in cell fate during stress recovery. Human Molecular Genetics, 2021, 31, 166-175.	2.9	15
284	Common Genetic Variants Contribute to Risk of Transposition of the Great Arteries. Circulation Research, 2022, 130, 166-180.	4.5	15
285	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
286	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
287	SCFD1 expression quantitative trait loci in amyotrophic lateral sclerosis are differentially expressed. Brain Communications, 2021, 3, fcab236.	3.3	14
288	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

#	Article	IF	Citations
289	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
290	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
291	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
292	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
293	Proteinopathies as Hallmarks of Impaired Gene Expression, Proteostasis and Mitochondrial Function in Amyotrophic Lateral Sclerosis. Frontiers in Neuroscience, 2021, 15, 783624.	2.8	13
294	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
295	Homozygosity analysis in amyotrophic lateral sclerosis. European Journal of Human Genetics, 2013, 21, 1429-1435.	2.8	12
296	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
297	Regulation of Synapse Weakening through Interactions of the Microtubule Associated Protein Tau with PACSIN1. Journal of Neuroscience, 2021, 41, 7162-7170.	3.6	12
298	Extensive phenotypic characterisation of a human TDP-43Q331KÂtransgenic mouse model of amyotrophic lateral sclerosis (ALS). Scientific Reports, 2021, 11, 16659.	3.3	12
299	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
300	Susceptibility genes in sporadic ALS: Separating the wheat from the chaff by international collaboration. Neurology, 2006, 67, 738-739.	1.1	10
301	Genetic analysis of amyotrophic lateral sclerosis in the Slovenian population. Neurobiology of Aging, 2015, 36, 1601.e17-1601.e20.	3.1	10
302	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
303	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
304	Demystifying the spontaneous phenomena of motor hyperexcitability. Clinical Neurophysiology, 2021, 132, 1830-1844.	1.5	10
305	Unbiased metabolome screen leads to personalized medicine strategy for amyotrophic lateral sclerosis. Brain Communications, 2022, 4, fcac069.	3.3	10
306	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

#	Article	IF	Citations
307	Polymorphisms in the GluR2 gene are not associated with amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 418-420.	3.1	8
308	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
309	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
310	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
311	Birth order and the genetics of amyotrophic lateral sclerosis. Journal of Neurology, 2008, 255, 99-102.	3.6	7
312	Comment on â€œDrug Screening for ALS Using Patient-Specific Induced Pluripotent Stem Cellsâ€. Science Translational Medicine, 2013, 5, 188le2.	12.4	7
313	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
314	Can Astrocytes Be a Target for Precision Medicine?. Advances in Experimental Medicine and Biology, 2017, 1007, 111-128.	1.6	7
315	Disease Mechanisms and Therapeutic Approaches in C9orf72 ALS-FTD. Biomedicines, 2021, 9, 601.	3.2	7
316	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
317	Reports. Journal of Interprofessional Care, 2002, 16, 289-291.	1.7	6
318	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
319	Clinical grand round: A rapidly progressive pyramidal and extrapyramidal syndrome with a supranuclear gaze palsy. Movement Disorders, 2005, 20, 826-831.	3.9	5
320	Amyotrophic Lateral Sclerosis/Motor Neuron Disease. , 2006, , 1-18.		5
321	Riluzole and Motor Neurone Disease. Practical Neurology, 2003, 3, 160-169.	1.1	3
322	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
323	Clinical aspects of motor neurone disease. Medicine, 2012, 40, 540-545.	0.4	3
324	Regionality of disease progression predicts prognosis in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2015, 16, 442-447.	1.7	3

#	Article	IF	Citations
325	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
326	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
327	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
328	Neuromuscular disease: new insights and avenues for therapy. Lancet Neurology, The, 2013, 12, 13-15.	10.2	2
329	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
330	Review: Glial lineages and myelination in the central nervous system. , 0, .		2
331	Unpicking neurodegeneration in a dish with human pluripotent stem cells. Cell Cycle, 2013, 12, 2339-2340.	2.6	1
332	Genome-Wide Identification of the Genetic Basis of Amyotrophic Lateral Sclerosis. SSRN Electronic Journal, 0, , .	0.4	1
333	A Novel Locus for Motor Neurone Disease on Chromosome 16q12-13. Clinical Science, 2003, 104, 40P-40P.	0.0	0
334	Cloning in Research and Treatment of Human Genetic Disease. , 2010, , 875-883.		0
335	Amyotrophic lateral sclerosis and other disorders of the lower motor neuron. , 0, , 136-147.		0
336	Analysis of TDP-43 and its binding partners in neurodegenerative diseases. Molecular Neurodegeneration, 2013, 8, .	10.8	0
337	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
338	Reply: The role ofDNAJB2in amyotrophic lateral sclerosis. Brain, 2016, 139, e58-e58.	7.6	0
339	Amyotrophic Lateral Sclerosis and Other TDP-43 Proteinopathies. , 2018, , 99-115.		0